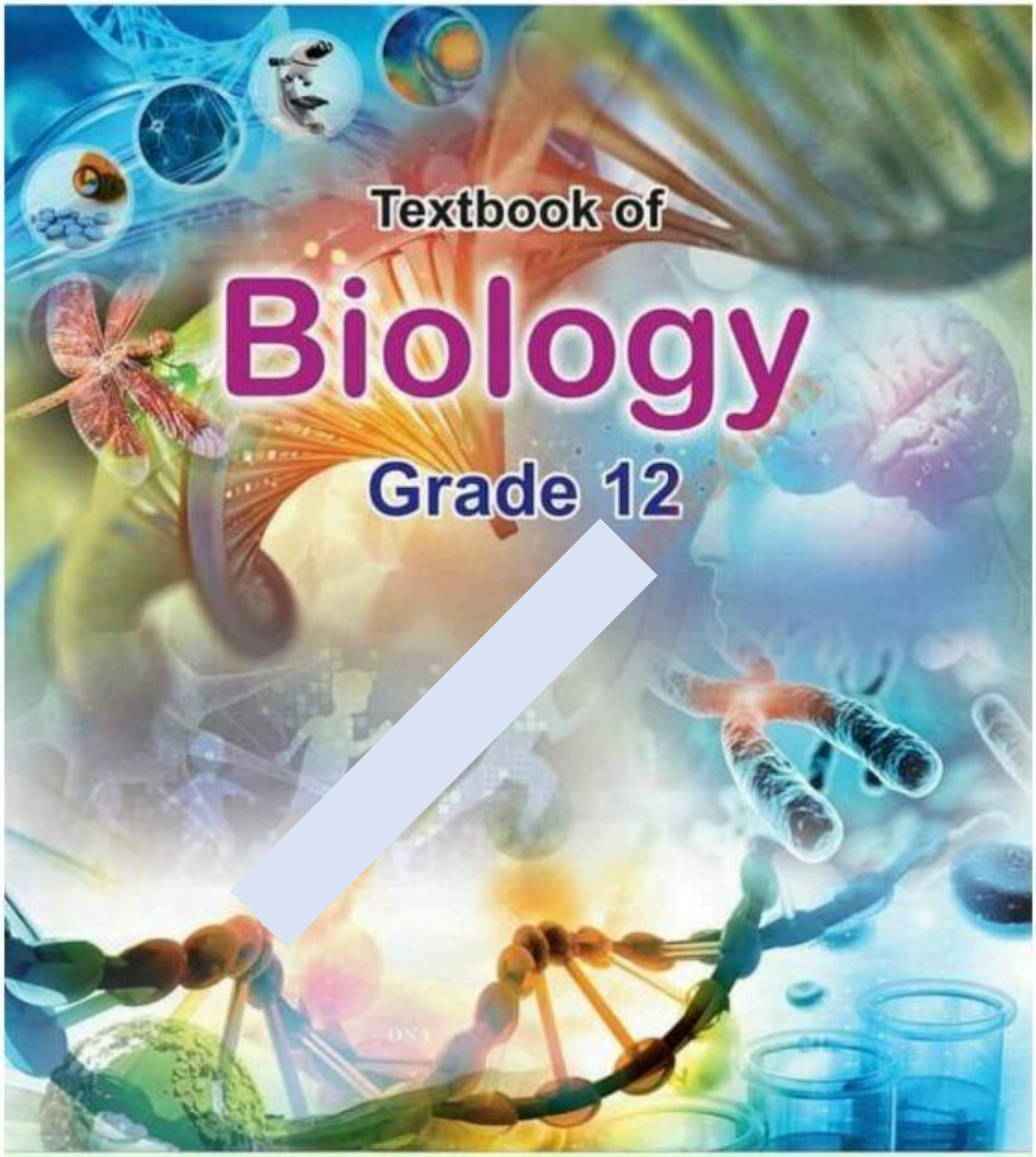

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OUR MOTTO

• Standards • Outcomes • Access • Style

Note

The material given in the box (Science titbits, Did you know, Critical thinking, STSC, Activity, Teacher's Point) and parenthesis, are not part of the textor SLO's.

Textbook of
Biology Grade - 12

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PREFACE

Biology Grade - 12 is developed according to the National Curriculum 2006 . It is being published since 2014 and now it is presented under the new management and supervision of textbook development, principles and guidelines with new design and layout.

The standard includes higher thinking, deep knowledge, problem solving substantive conversation and connections to the world beyond the class room and achieve the target set by the curriculum. The special features of the textbook are:

- Each chapter begins with a brief recalling statement i.e., introduction to the chapter. The textbook has coloured illustrations to capture the students' attention. Where necessary, concept mapping has also been incorporated.
- Necessary 'Tit Bits' and 'Critical Thinking' have been added in each chapter for motivating the students to apply their intelligence and acquire more knowledge.
- The exercises include multiple choice questions, short answer questions and extensive questions.
- At the end of the book a glossary and has been annexed.

In each chapter Science, Technology and Society connections are explained in accordance with the curriculum. These interventions will serve as a guide for evaluating the students' skills development through the chapter knowledge and their abilities to apply knowledge to the scientific and social problems. The duration or the number of periods is also allocated to complete each chapter, so that the teachers can develop their teaching strategy and plans in an effective manner accordingly.

Quality of Standards and Actualization of Style is our motto. With these elaborations, this series of new development is presented for use. However there is always room for improvement and suggestion from the teacher and the community will be highly appreciated to make the book more valuable and to make the textbook more interesting, informative and useful for the students. After educational feedback, research report and reviewed by NCC through review committees, **now this is the revised addition of the book biology for Grade-XII for the year 2020.**

May Allah Guides and helps us. (Ameen).

National Book Foundation

بِسْمِ اللّٰهِ الرَّحْمٰنِ الرَّحِيْمِ

In the Name of Allah, the Most Gracious, the Most Merciful

CONTENTS

14

RESPIRATION

After completing this lesson, you will be able to

- Define the respiratory surface and list its properties.
- Describe the main structural features and functions of the components of human respiratory system.
- Describe the ventilation mechanism in humans.
- State lung volumes.
- Explain how breathing is controlled.
- Describe the transport of oxygen and carbon dioxide through blood.
- Describe the role of respiratory pigments.
- State the causes, symptoms and treatment of upper Respiratory Tract Infections (sinusitis) and lower Respiratory Tract Infections (pulmonary tuberculosis).
- Describe the disorders of lungs (lung cancer).
- List the effects of smoking on respiratory system.

Reading

Like other life processes, the respiration process also occurs at cellular level and organismic level. The process of respiration that occurs at cellular level is also called **internal respiration** which is a catabolic process. It involves the breakdown of complex organic compounds into simpler molecules with the release of energy. On the other hand, the process of respiration that occurs at organismic level is also called **external respiration**. It involves the inhaling of oxygen and exhaling of carbon dioxide. Both the processes are interlinked as the oxygen, required for cellular respiration, is inhaled from environment while the carbon dioxide which is produced in cellular respiration, is exhaled into the environment. This chapter deals with various aspects of respiration.

14.1 PROPERTIES OF RESPIRATORY SURFACE

The area where gaseous exchange with the environment actually takes place is called the **respiratory surface**. The respiratory surface must have the following properties so that diffusion can occur effectively: (a) It must be moist and permeable so that gases can pass through. (b) It must be thin, because diffusion is only efficient over distance of 1 mm or less. (c) It should possess a large surface area so that sufficient amount of gases can be exchanged according to the organism's need. (d) It should possess a good blood supply. (e) There should be a good ventilation mechanism to maintain a steep diffusion gradient across the respiratory surface.

14.2 RESPIRATORY SYSTEM OF MAN

The body system which is responsible for the exchange of gases between body fluid and outer environment is called **respiratory system.**

The human respiratory system can be divided into two regions, upper respiratory tract and lower respiratory tract.

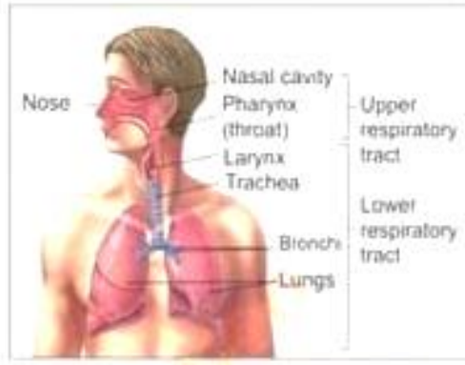


Fig. 14.1 Human respiratory tract

14.2.1 Upper Respiratory Tract

The upper respiratory tract includes nostrils, nasal cavity and pharynx.

Nose

The nose is only externally visible part of the respiratory system. Human nose is composed of bones, cartilage and fatty tissues. The external openings of nose are called **nostrils** and the inner hollow spaces are called **nasal cavities**. There are two nasal cavities which are partitioned by means of nasal septum (the part of nasal bone). The anterior parts of nasal cavities near the nostrils are called **vestibules** which contain hair. Both the nostrils and nasal cavities are lined by ciliated mucous membranes.

Nose hair, mucus and cilia serve as a defence mechanism against the harmful pathogens and particulate matter present in the air. The mucus and cilia filter the air and prevent the entry of foreign particles such as microorganisms, dust and particulate matter inside the respiratory system. The mucus also helps in moistening the air. Cilia move the trapped substances to the pharynx for their removal. Underneath the mucous membrane, there are blood capillaries that help to warm the air to about 30°C, depending upon the external temperature.

Fig. 14.2 Pharynx

Pharynx

Pharynx is cone-shaped passageway leading from the oral and nasal cavities to the oesophagus and larynx. The pharynx is part of the digestive system and also the respiratory system. The human pharynx is conventionally divided into three sections: the **nasopharynx,** the **oropharynx**, and the **laryngopharynx**

14.2.2 Lower Respiratory Tract

The lower respiratory tract includes the larynx, trachea, bronchi and lungs.

Larynx

The **larynx** is an enlargement in the airway at the top of the trachea and below the pharynx. The larynx is composed primarily of muscles and cartilages. One of the cartilages is the **epiglottis**.

This structure usually stands upright and allows air to enter the larynx. During swallowing, however **larynx** is raised and the epiglottis is pressed downward. As a result, the epiglottis partially covers the opening into the larynx and helps to prevent foods and liquids from entering the air passages. The opening of the larynx is called **glottis**. It is also lined with mucous membrane. Inside the larynx, there are two vocal cords which are responsible for vocalization.

Science Titbits

The alveoli of human lungs are lined with a **surfactant**, a film of lipoprotein that lowers the surface tension and prevents them from closing. Surfactant also speeds up the transport of oxygen and carbon dioxide between the air and liquid lining the alveolus and helps to kill any bacteria, which reach the alveoli. Surfactant is constantly being secreted and reabsorbed in a healthy lung.

Trachea

The trachea or windpipe is a membranous tube. It consists of dense regular tissue and smooth muscle reinforced with 15-20 C-shaped pieces of cartilage.

Bronchi and bronchioles

The trachea divides to form two smaller tubes called **primary bronchi**. The primary bronchi divide into **secondary bronchi** within each lung. There are two secondary bronchi in the left lung and three in the right lung. The secondary bronchi, in turn, give rise to **tertiary bronchi**. The bronchi continue to branch, finally giving rise to **bronchioles** which are less than 1mm in diameter. The bronchioles also subdivide several times to become even smaller **terminal bronchioles**. In the secondary bronchi, the C-shaped cartilages are replaced with cartilage plates but the bronchioles and their terminal branches have no cartilage structures.

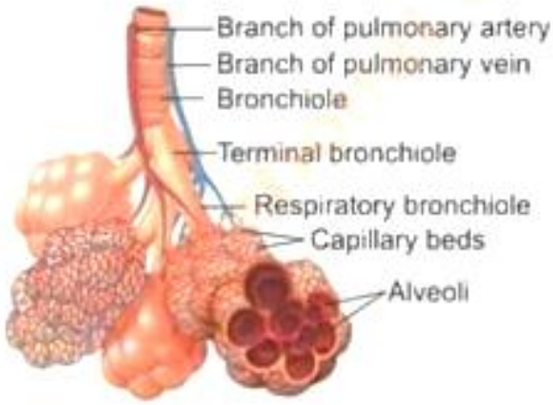


Fig. 14.3 Alveoli

Alveolar ducts and alveoli

The terminal bronchioles divide to form **alveolar ducts**. These alveolar ducts end at tiny air filled chambers called **alveoli** which are the sites of gas exchange between the air and the blood. There are over 700 million alveoli present in the lungs,. The wall of each alveolus is only 0.1 µm thick. On its outsides is a dense network of blood capillaries. Lining each alveolus is moist squamous epithelium. This consists of very thin, flattened cells, reducing the distance over which diffusion must occur. **Collagen** and **elastin** proteins are also present in their walls which allow the alveoli to expand and recoil easily during breathing.

External structure of lungs

The lungs are the principal organs of respiration. Each lung is conical in shape, with its base resting on the diaphragm and its apex extends to a point just above the clavicle. The right and left

lungs are separated medially by the heart and **mediastinum**, which is the area between the lungs.

The left lung has two lobes, **superior lobe** and **inferior lobe**. The left lung shares space with the heart. The right lung has three lobes. The **hilum** is a triangular shaped depression of both the lungs where the blood vessels and airways pass into the lungs. The lungs are spongy due to presence of alveoli.

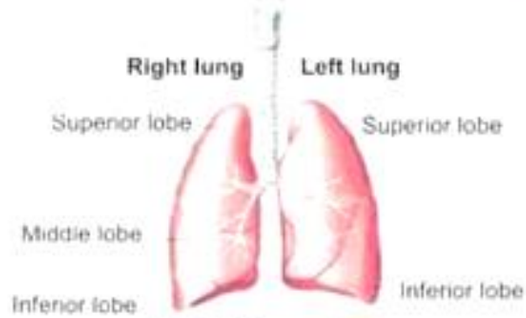


Fig. 14.4 Human lungs

14.2.3 The Mechanism of Breathing (Ventilation)

The lungs themselves neither draw in air nor push it out. The diaphragm and the intercostal muscles accomplish the expansion and contraction of the lungs. The **diaphragm** is a large dome of skeletal muscle that separates the thoracic cavity from abdominal cavity. There are two sets of **intercostal muscles** between each pair of ribs: the external intercostal and the internal intercostal. The muscle fibres run diagonally but in opposite direction in the two sets of muscles. Breathing takes place in two phases i.e., inspiration and expiration.

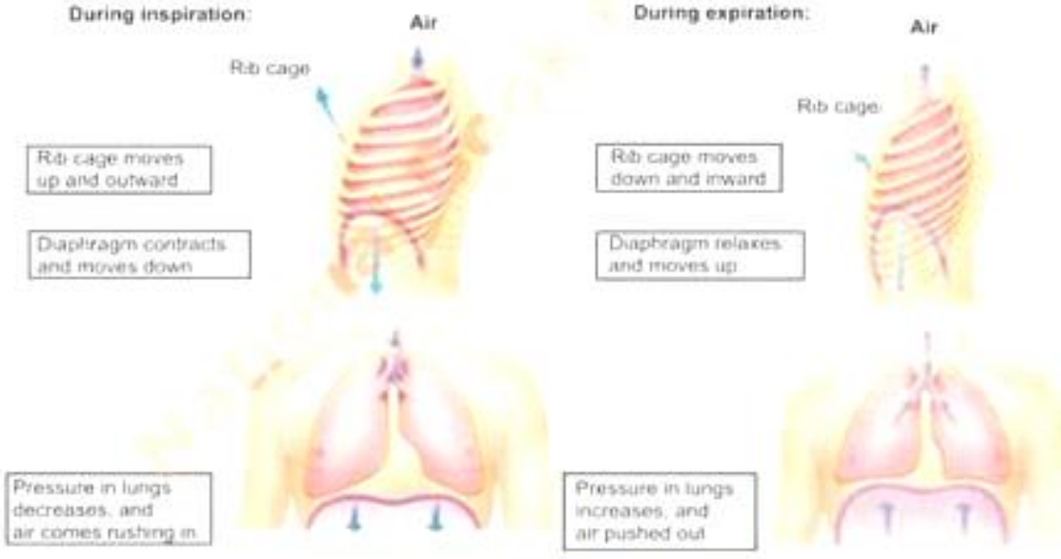


Fig. 14.5 Mechanism of breathing in human

Inspiration: It is taking in of air; it is the active phase of breathing. During inspiration contraction of the diaphragm causes its dome shape to flatten or less dome shape whereas contraction of the external intercostal and relaxation of the internal intercostal causes the rib cage to move upward and forward. Both these events result in increase of inner space of thoracic cavity. Consequently, the pressure in the thorax and hence in the lungs, is reduced to less than atmospheric pressure. Air therefore enters the lungs and alveoli become inflated.

Expiration: It is the removal of air out of the lungs; it is the passive phase of breathing. During expiration relaxation of the diaphragm causes it to become more dome shape whereas relaxation of the external intercostal and contraction of the internal intercostal cause the rib cage to move downward and backward. Both these events result in decrease of inner space of thoracic cavity. Consequently, the pressure in the thorax and hence in the lungs, is increased to more than atmospheric pressure, therefore, air is forced to expelled from the lungs.

14.2.4 Respiratory Volumes

Breathing (inspiration and expiration) occurs in a cyclical manner due to the movements of the chest wall and the lungs. The resulting changes in pressure, causes changes in lung volumes, i.e., the amount of air the lungs are capable of occupying. These volumes tend to vary, depending on the depth of respiration, gender, age and in certain respiratory diseases. Respiratory volume is the amount of air inhaled, exhaled and stored within the lungs at any given time. The amount of air which is inhaled or exhaled at rest is called **tidal volume**. The average **tidal volume** is 500ml. The amount of extra air inhaled (above tidal volume) during a deep breath is called **inspiratory reserved volume**. This can be as high as 3000ml. Total lung capacity of human is 6000ml.

Science, Technology and Society Connections

Mouth to mouth artificial respiration. (Cardiopulmonary Resuscitation (CPR)

Mouth to mouth artificial respiration is called resuscitation. It is a technique used to recover a person who has stopped breathing. In this technique, the rescuer presses his or her mouth against the mouth of the victim and allowing for passive inhalation, forces air into the lungs at intervals of several seconds.

What to do:

1. Stretch out victim on his back and kneel close to his side. Loosen any tight clothing around his neck or chest.
2. Remove foreign objects if present from victim's mouth and throat by finger sweeping.
3. Lift up chin and tilt head back as far as possible. If the head is not tilted, the tongue may block the throat.
4. Begin the resuscitation immediately. Pinch the nostrils together with the thumb and index finger of the hand that is pressing on the victim's forehead. This prevents the loss of air through the nose during resuscitation.
5. Inhale deeply.
6. Place your mouth tightly around the victim's mouth (over mouth and nose of small children) and blow into the air passage with brief intervals. Continue this activity so long as there is any pulse or heartbeat.
7. Watch the victim's chest. When you see it rise, stop blowing, raise your mouth, turn your head to the side and listen for exhalation.
8. If patient is revived, keep him warm and do not move him until the doctor arrives, or at least for half or one hour.

Science, Technology and Society Connections

Justify why birds perform much better than man at high altitudes.

The efficiency of the lungs of birds is that they can extract considerably more oxygen from a given quantity of air than our lungs. This is because birds have haemoglobin with a very high affinity for oxygen. They have a very large number of capillaries in lungs. They have one-way flow of air through the lungs so there is no dead air (residual volume) in the bird's lung. Birds have several large air sacs in addition to their lungs.

14.2.5 Control of Breathing (Ventilation)

Normally we are not conscious of our breathing because it is controlled involuntarily. A **breathing centre** located in the **medulla** of the brain carries out involuntary control of breathing. The ventral portion of the breathing centre acts to increase the rate and depth of inspiration and is called **inspiratory centre**. The dorsal and lateral portions inhibit inspiration and stimulate expiration. These regions form the **expiratory centre**.

Through the **cerebral cortex** it is possible to consciously or unconsciously increase or decrease the rate and depth of the respiratory movement. A person may also stop breathing voluntarily. Occasionally people are able to hold their breath until the blood partial pressure of oxygen declines to a level low enough that they lose consciousness. After consciousness is lost, the respiratory centre resumes its normal function in automatically controlling respiration. Emotions acting through the **limbic system** of the brain can also affect the respiratory centre.

Science, Technology and Society Connections

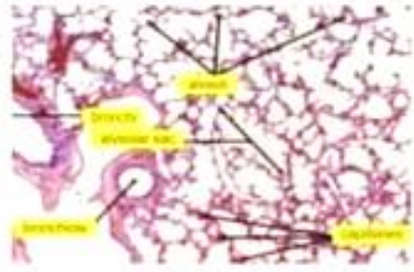
Describe the development of artificial breathing apparatus (for use under water and at high altitude and by fireman)

The word SCUBA is an acronym for Self-Contained Underwater Breathing Apparatus. It is also called aqualung.

A typical aqualung contains compressed air or a mixture called Nitrox which consists of about 35 percent oxygen and 65 percent nitrogen. This apparatus consists of a tank containing highly compressed air in which the pressure down to an ambient pressure so divers could breathe comfortably at any depth.

Skills: Analyzing, Interpreting and Communication

- Draw and label a diagram to illustrate the microscopic structure of human lung with the help of slides.
- Trace the path of air through different parts of human respiratory system.



Human lung tissue

14.3 TRANSPORT OF GASES

Like other materials, respiratory gases are also transported in various regions of the body by means of blood. The blood transports oxygen from the lungs to different tissues and carbon dioxide from tissues to the lungs.

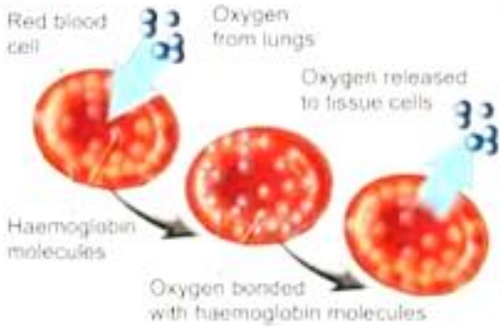


Fig. 14.6 Transport of oxygen

Science Titbits

The oxygen carrying capacity of blood is directly proportional to the partial pressure of oxygen (PO_2). Maximum oxygen carrying capacity of arterial blood is 20 ml/100 ml of blood (100% saturated) which is achieved at 100 mmHg PO_2. The 5 ml of O_2 is released to the tissues by each 100 ml blood. Oxygen carrying capacity is sensitive to a variety of environmental conditions like rise in body temperature, drop in pH of blood and partial pressures of carbon dioxide and oxygen.

Science Titbits

The amount of haemoglobin is 15 gms/100 ml of blood. Since 1gm Hb can combine with 1.34 ml of O_2, therefore 100 ml blood combines with 20 ml O_2 (100% saturated). Normally each 100 ml of arterial blood contains 19.4 ml O_2 (i.e., it is 97% saturated; PO_2 is 95 mmHg), while 100 ml of venous blood contains 14.4 ml O_2 (i.e., it is 75% saturated; PO_2 is 40 mmHg).

14.3.1 Transport of Oxygen in Blood

Approximately 97% of oxygen is carried by the red blood cells as **oxyhaemoglobin**, while 3% is transported as dissolved oxygen in the **plasma**. At its high partial pressure oxygen binds with haemoglobin. This binding is reversible that occurs in the lungs in the presence of enzyme carbonic anhydrase. Each molecule of haemoglobin can bind with four molecules of oxygen to form oxyhaemoglobin.

$$Hb + 4O_2 \xrightarrow{\text{Carbonic anhydrase}} Hb4O_2$$

The ability of haemoglobin to bind with oxygen is called **oxygen carrying capacity** of blood.

14.3.2 Transport of Carbon dioxide in Blood

Carbon dioxide is transported in the blood in three main ways: (i) In the form of bicarbonate ions. (ii) In the form of carboxyhaemoglobin.

(iii) Dissolved in plasma.

(i) As bicarbonate ions

Approximately 70% of carbon dioxide is carried in the blood as bicarbonate ions. Carbon dioxide diffuses into the blood, enters the red blood cells and combines with water to form carbonic acid in the presence of enzyme carbonic anhydrase. The chemical reaction can be depicted as follows:

$$CO_2 + H_2O \underset{}{\overset{\text{Carbonic anhydrase}}{\rightleftharpoons}} H_2CO_3$$

Carbonic acid, H_2CO_3 is an unstable compound and dissociates to form hydrogen ions and bicarbonate ions.

$$H_2CO_3 \overset{\text{Carbonic anhydrase}}{\rightleftharpoons} H^+ + HCO_3^-$$

Accumulation of H^+ ions increases acidity in the blood, i.e., it leads to the decrease in pH. This does not occur since haemoglobin buffers the hydrogen formed. The

hydrogen ion readily associates with oxyhaemoglobin ($Hb4O_2$) to form haemoglobinic acid (HHb) and oxygen is released to the tissue.

$$Hb4O_2 + H^+ \longrightarrow HHb + 4O_2$$

From inside of the erythrocytes negatively charged HCO_3^- ions diffuse to the plasma. This is balanced by the diffusion of chloride ions, (Cl^-), in the opposite direction. This is achieved by special bicarbonate-chloride carrier proteins that exist in the RBC membrane. This protein moves the two ions in opposite directions, maintaining the balance of ions on either side. This is called the **chloride shift** or **Hamburger' phenomenon.**

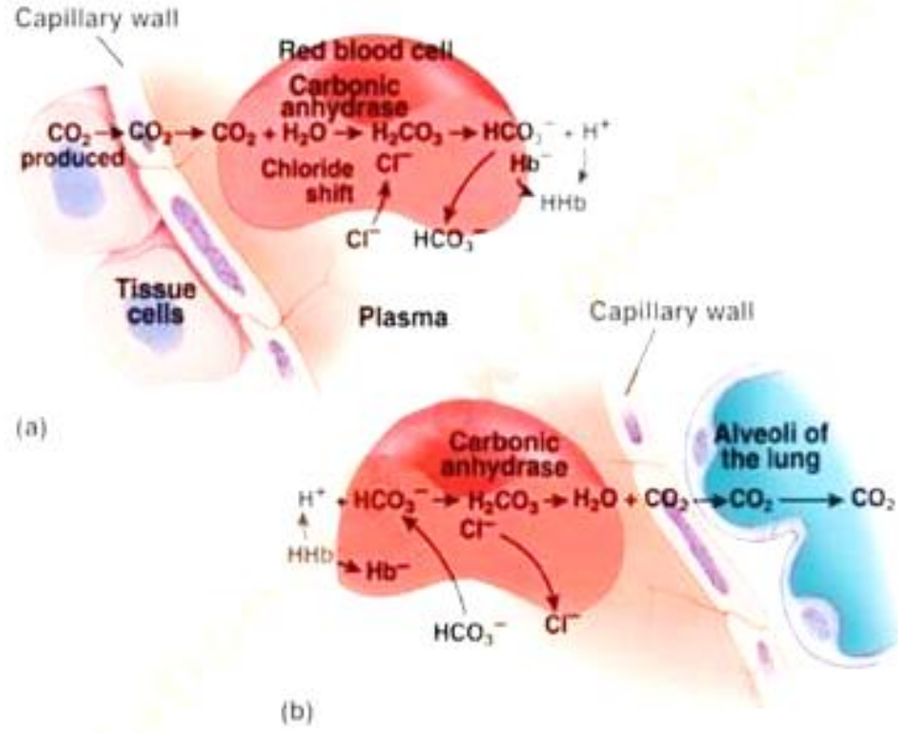


Fig. 14.7 Transport of CO_2 as bicarbonate ions (a) Transfer of CO_2 from tissues to blood (b) Transfer of CO_2 from blood to lungs

The chloride ions that enter the RBC combine with potassium (K^+) to form potassium chloride, whereas bicarbonate ions in the blood plasma combine with Na^+ to form sodium bicarbonates. The blood pH is thus maintained at approximately 7.4 by the buffer mechanism that exists in blood.

Transport of CO_2 depends on the partial pressure of CO_2. The partial pressure of CO_2 is higher in tissues than blood so it diffuses into blood here it react with water and transported to the lungs as bicarbonate ion. In lungs process reverses and bicarbonate ions combine with hydrogen ion to release carbon dioxide and water.

(ii) As carboxyhaemoglobin

About 23% of carbon dioxide is carried as carboxyhaemoglobin. CO_2 combines with the globin part of haemoglobin. The reaction depends upon the partial pressure of CO_2. When the PCO_2 is higher in the tissues than blood, formation of carboxyhaemoglobin occurs. When the PCO_2 is higher in the blood than tissues as in case of lungs, carboxyhaemoglobin releases its CO_2.

(iii) As dissolved CO_2 in plasma

Only 7% of carbon dioxide is carried this way. This is rather inefficient way to carry carbon dioxide, but it does occur.

Science, Technology and Society Connections

Describe the carbon monoxide poisoning (caused by gas heaters left on overnight in closed environments).

Gases that have undergone incomplete combustion produce CO and toxic fumes (hydrogen cyanide). In carbon monoxide poisoning caused by gas heaters, left on overnight in closed environments, CO binds to haemoglobin preventing the uptake of oxygen by haemoglobin. The symptoms of CO poisoning are nausea, vomiting, headache, mental status changes, and cherry-red lips. CO binds to haemoglobin with affinity 249 times greater than that of oxygen. CO poisoning also decreases ability of haemoglobin to release oxygen to tissue.

14.3.3 Respiratory Pigments

Respiratory pigments are coloured molecules, which act as oxygen carriers by binding reversibly to oxygen. All known respiratory pigments contain a coloured non-protein portion e.g., **haem** in the haemoglobin. The two well-known respiratory pigments are haemoglobin and myoglobin.

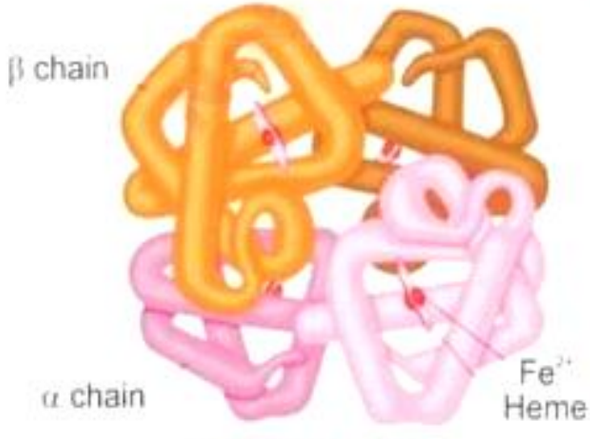


Fig. 14.8 Haemoglobin

Haemoglobin

It contains four globin protein chains, each associated with haem, an iron-containing group. Iron combines loosely with oxygen and in this way oxygen is carried in the blood. At high oxygen concentrations, the pigment combines with oxygen, whereas at low oxygen concentrations the oxygen is quickly released.

Myoglobin

It consists of one polypeptide chain. This chain is associated with an

iron containing ring structure. This iron can bind with one molecule of oxygen. It is found in skeletal muscles and is the main reason why meat appears red. It serves as an intermediate compound for the transfer of oxygen from haemoglobin to aerobic metabolic processes of the muscle cells. Myoglobin releases oxygen when the partial pressure of oxygen is below 20 mmHg. In this way it stores oxygen in resting muscle, only releasing it when supplies of oxyhaemoglobin have been exhausted.

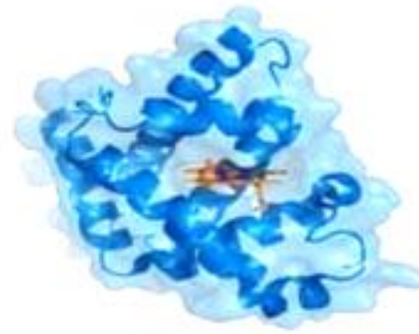


Fig. 14.9: Myoglobin

Table 14.1 Differences between haemoglobin and myoglobin

Haemoglobin	Myoglobin
(1) It consists of four polypeptide chains.	(1) It consists of one polypeptide chain.
(2) Each molecule possesses four iron containing haem groups.	(2) Each molecule possesses one iron containing haem group.
(3) Four oxygen molecules can bind to each haemoglobin molecule.	(3) Only one oxygen molecule can bind to each myoglobin molecule.
(4) It is found in RBCs.	(4) It is found in muscles.
(5) It transports oxygen.	(5) It stores oxygen.
(6) It has less affinity with oxygen.	(6) It has more affinity with oxygen.
(7) It loses oxygen at PO_2 60 mmHg.	(7) It loses oxygen at PO_2 20 mmHg.

Science, Technology and Society Connections

Relate the transportation of gases to hiccups, sneezing and snoring.

Hiccups: It is the spasmodic contraction of the diaphragm while the glottis is closed, producing a sharp respiratory sound. It is reflexive and serves no known functions.

Sneezing: Deep inspiration is followed by a closure of the glottis. The forceful expiration that results abruptly opens the glottis, sending a blast air through the nasal cavity. The eyelids close reflexively during sneeze. Sneezing is a reflexive response to irritating stimulus of the nasal mucosa. Sneezing clears the upper respiratory passages.

Snoring: It is a rough, raspy noise that can occur when a sleeping person inhales through the mouth and nose. The noise usually is made by vibration of the soft palate which may occur as a result of vocal cord vibration.

14.4 RESPIRATORY DISORDERS

Several defence mechanisms protect the delicate lungs from the harmful substances we breathe. The **hair** around the nostrils, the **mucous lining** in the nose and pharynx and the **cilia** which are mucous elevator, serve to remove foreign particles in the inspired air. Continued inhalation of harmful substances results in the respiratory disorders.

14.4.1 Upper Respiratory Tract Infection

The infections of the upper respiratory tract include sinusitis, etc.

Sinusitis

Sinusitis is an inflammation of the nasal sinuses that may be acute (symptoms last 2 - 8 weeks) or chronic (symptoms last much longer). The sinuses are holes in the skull between the facial bones.

Cause: Sinusitis is generally caused by cold and wet climate. Atmospheric pollution, smoke, dust, overcrowding, dental infections, viral infections etc., also cause sinusitis.

Symptoms: Fever, nasal obstruction, raspy voice, pus-like nasal discharge, loss of sense of smell, facial pain or headache that is sometimes aggravated by bending over.

Treatment: If a bacterial infection is present, antibiotics or sulpha drugs are usually prescribed. Beside it the physician may also prescribe nebulization which can be useful in reducing inflammation in the sinuses and nose and to accelerate recovery.

14.4.2 Lower Respiratory Tract Infection

The infections of lower respiratory tract include, pulmonary tuberculosis etc.

Pulmonary Tuberculosis

Pulmonary Tuberculosis (TB) is a highly contagious chronic bacterial infection of lungs. When people have pulmonary tuberculosis, the alveoli burst and are replaced by inelastic connective tissue. The cells of the lung tissue build a protective capsule around the bacilli and isolate them from rest of the body. This tiny capsule is called **tubercle**. The tubercles can rupture, releasing bacteria that infect other parts of the lung.

Science Titbits

About 15 percent of TB patients may develop the disease in an organ other than the lung, such as the lymph nodes, GI tract, and bones and joints.

Cause: Pulmonary tuberculosis is caused by *Mycobacterium tuberculosis*.

Symptoms: There is a low-grade intermittent fever usually in the evening, night sweats, weight loss, anorexia, depression, weakness and dry cough with sputum, dull ache in the chest due to Inflammation of the pleura of the lungs.

Treatment: Taking medicines for 9 months regularly can cure T.B disease. This is called **Daily Observed Treatment Short Course (DOTS).** This treatment is given to patients under supervision to ensure that the "medicines intake" completely cures the patient.

14.4.3 Disorders of the Lungs

There are many disorders that affect lungs. Emphysema and lung cancer are two common examples of disorders of lungs.

Lung Cancer

Cancer is a **malignant** tumour which may develop due to uncontrolled cell division.

Cause: Smoking is the main cause of lung cancer because tobacco smoke contains many carcinogens. In addition to this, asbestos, arsenic, radiations such as gamma and x-rays, the sun, and compounds in car exhaust fumes are all examples of carcinogens.

Symptoms: The first event appears to be thickening and **callusing** (over growth) of the cells lining the bronchi. Then there is a loss of cilia so that it is impossible to prevent dust and dirt from setting in the lungs. The tumour may grow until the bronchus is blocked, cutting off the supply of air to that lung.

Treatment: The only treatment that offers a possibility of cure is to remove a lobe or the lung completely before secondary growths have time to form. This operation is called **pneumonectomy**. Treatments also include chemotherapy and radiotherapy.

Science Titbits

The spread of pulmonary TB can be controlled by some preventive measures like:
(1) Living room should be well ventilated and bright.
(2) Always cover the mouth with cloth during coughing and sneezing.
(3) Avoid spitting openly.
(4) Always burry or burn the sputum of patient.
(5) The patients should spit in a utensil with lime powder to prevent the spread of disease.
(6) The use of masks and other respiratory isolation procedures to prevent spread to medical personal is also important.

14.3.4 Effects of Smoking

The effects of smoking on respiratory system are:

1) Cigarette smoking causes about 87% of lung cancer.
2) Besides lung cancer, cigarette smoking is also a major cause of cancer of the mouth, larynx and oesophagus.
3) Cigarette smoking causes other lung diseases e.g., chronic bronchitis, emphysema.
4) Cigarette smokes contain chemicals which irritate the air passages and lungs, causing early morning cough.

Fig. 14.10: Effects of smoking

5) Smokers are likely to get pneumonia because damaged or destroyed cilia cannot protect lungs from bacteria and viruses that float in the air.
6) Almost immediately, smoking can make it hard to breathe. Within a short time, it can also worsen asthma and allergies.

Activity

1. Identification of different parts of the respiratory and reproductive system of a dissected frog (dissection would be done by the teacher)
2. Examination of sheep lungs
3. Comparison and interpretation of the X-ray films of lungs of a smoker with that of a healthy man

Exercise

Review Questions

1. Select the correct answer

(i) When blood leaves the capillary bed most of the carbon dioxide is in the form of
(A) carbonate ions (B) bicarbonate ions
(C) hydrogen ions (D) hydroxyl ions

(ii) When you inhale, the diaphragm
(A) relaxes and moves upward (B) relaxes and moves downward
(C) contracts and moves upward (D) contracts and moves downward

(iii) With which other system do specialised respiratory systems most closely interface in exchanging gases between the cells and the environment?
(A) the skin (B) the excretory system
(C) the circulatory system (D) the muscular system

(iv) Which of the following is the respiratory surface in human respiratory system:
(A) larynx (B) trachea (C) bronchi (D) alveoli

(v) How is most of the oxygen transported in the blood?
(A) dissolved in plasma (B) bound to haemogloibin
(C) as bicarbonate (D) dissolved in water

(vi) The lateral walls of the chest cavity of man are composed of the:
(A) ribs (B) intercostal muscles
(C) ribs and intercostal muscles (D) ribs, intercostal muscles and diaphragm

(vii) Which of the following factors is the most effective in accelerating the rate of breathing in man?
(A) a lack of oxygen in the blood (B) a lack of oxygen in the tissues
(C) an excess of carbon dioxide in the lungs (D) an excess of carbon dioxide in the blood

(viii) Which of the following changes will increase the body's rate of carbon dioxide excretion into the alveoli?
(A) holding the breath
(B) the breakdown of alveolar tissue as a result of disease
(C) a decrease in the partial pressure of carbon dioxide in the alveolar air
(D) a decrease in the pulmonary circulation

(ix) Breathing is an example of
(A) counter current exchange (B) cellular respiration
(C) ventilation (D) diffusion

(x) Which event is not associated with the activity of expiration?
(A) contraction of diaphragm (B) more dome like shape of diaphragm
(C) backward and downward movement of rib cage
(D) relaxation of external intercostals muscles

(xi) Respiratory pigments

(A) combine reversibly with only oxygen (B) all have four haem groups

(C) attach to the alveolar wall (D) None of them

(xii) Which sequence most accurately describes the sequence of airflow in the human respiratory system?

1. pharynx 2. bronchus 3.trachea 4.larynx 5.alveolus 6.bronchiole

(A) 4, 1, 3, 2, 5, 6 (B) 1, 4, 3, 2, 5, 6

(C) 4, 1, 3, 2, 6, 5 (D) 1, 4, 3, 2, 6, 5

Short Questions

2. What is respiratory surface? Write the properties of respiratory surface.
3. What organs constitute the respiratory system?
4. How nose and nasal cavity function in filtering the incoming air?
5. What is the role of 'pharynx' in human respiration?
6. Describe the structure and function of human larynx.
7. Describe the structure and function of alveoli.
8. How the contraction and relaxation of human lungs take place?
9. What is respiratory reserved volume?
10. What is chloride shift?
11. What are the advantages of having millions of alveoli rather than a pair of simple balloon like lungs?
12. Write the differences between:

(a) Internal and external respiration (b) Upper and lower respiratory tract

(c) Bronchi and bronchioles (d) Oxyhaemoglobin and carboxyhaemoglobin

(e) Haemoglobin and myoglobin

Extensive Questions

18. Describe the human upper respiratory tract.
19. Describe the human lower respiratory tract.
20. Describe the mechanism of breathing in man.
21. How the control of breathing takes place?
22. Explain the transport of oxygen in blood.
23. Explain the transport of carbon dioxide in blood.
24. What is the role of respiratory pigments in man?
25. Describe the cause, symptoms and treatments of:

(a) Sinusitis

(b) Pulmonary tuberculosis

(c) Lung cancer

18

CHEMICAL COORDINATION

After completing this lesson, you will be able to

- State the role of hormones as chemical messengers.
- Describe the chemical nature of hormones and correlate it with important hormones.
- Trace the path of the chemical message from its release from the endocrine gland to its action at the target site.
- Explain the two modes of hormone action at the cells of target site.
- Locate the following endocrine glands in human body; pituitary, thyroid, parathyroid, pancreas, adrenal, gonads.
- Name the hormonal secretions of the above-mentioned glands.
- Outline the major functions of the hormones of above mentioned glands and also relate the problems associated with the imbalance of these hormones.
- Explain the neurosecretory role of hypothalamus.
- Describe the functions of the hormones secreted by the endocrine tissue other than the mentioned above.
- Outline the concept of Feedback mechanism of hormones.
- Describe positive feedback with reference to Oxytocin and negative feedback with reference to Insulin and Glucagon.

Reading

The cellular functions needed to be continuously regulated. The nerve fibres do not innervate all the cells of the body; a special kind of coordination system is thus required. The endocrine system serves the role to coordinate most body cells. The hormonal system is concerned with control of the different metabolic functions of the body, such as the rate of chemical reactions, the transport of substances through the cell membranes, growth, and secretions. This coordination is called **chemical coordination**.

18.1 HORMONES – THE CHEMICAL MESSENGERS

Glands are the tissues that produce and release some products called **secretions**. There are two types of glands in the body, **exocrine gland** and **endocrine glands.** An endocrine gland or ductless gland secretes chemicals called **hormones** which affect the cells in other parts of the body.

18.1.1 Hormone as a Chemical Messenger

A hormone is a small soluble organic molecule which is effective in low concentration. It is essentially a **chemical messenger** that transports a signal from one cell to another. It has its effect at a site where specific receptors are present, called the target; hence it is termed as **messenger**.

Chemical nature of hormones

All the hormones are organic substances of varying structural complexity. Chemically, they may belong to any of the following categories.

(a) Steroid hormones: The hormones secreted by the adrenal cortex, testes, ovaries and placenta are composed of steroids e.g., cortisone, aldosterone, testosterone, estrogen, progesterone.

(b) Proteinous hormones: Somatotrophic, thyrotrophic and gonadotrophic hormones are secreted by the anterior lobe of pituitary gland and insulin hormone is secreted by pancreas.

(c) Catecholamine: Adrenaline and noradrenaline are secreted by the adrenal medulla.

(d) Amino acid derivative: Thyroxine hormone is secreted by the thyroid gland.

(e) Peptide Hormones: These include melanocyte stimulating hormone, the hormones oxytocin and vasopressin, adrenocorticotrophic hormone, calcitonin and parathormone.

Science Titbits

In 1902, Bayliss and Starling, prepared an extract from the duodenum which stimulated secretion of pancreatic digestive juices when it was injected into the bloodstream. They called the product 'secretin', and coined the term 'hormone', meaning 'to excite' or 'to set in motion.'

18.1.2 Mode of Hormone Action

Protein Hormones

Protein and peptide hormones cannot pass through cells' plasma membrane because they are water soluble. These hormones (**first messenger**) bind with their receptors on the plasma membrane of target cell, starting a series of events in the cell which generates **second messenger** (e.g. cAMP). The second messenger then triggers various changes in the cell including activation of enzymes, gene activation.

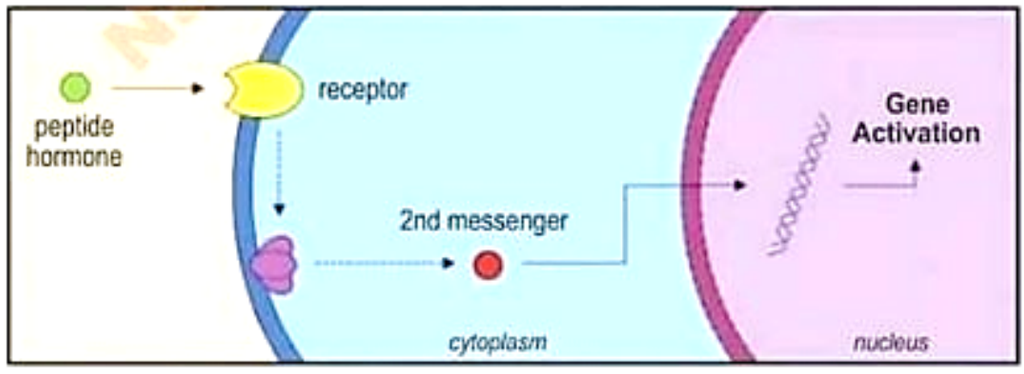


Fig. 18.1 Mode of action of protein hormone.

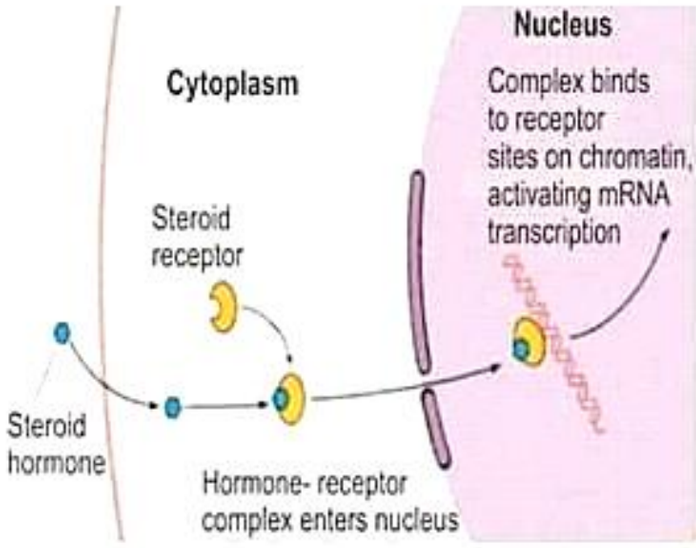


Fig. 18.2 Model of action of steroid hormone

Steroid Hormones

Steroid and thyroid hormones can pass through plasma membrane because they are lipid soluble. Receptors for these hormones are located inside target cells, in the cytoplasm or nucleus. Hormones bind with their receptors to form hormone-receptor complex. This complex then binds with promoter region of particular gene, acting as transcription factor. mRNA of that gene is formed by transcription and translated into protein. Target cell activities are modified by the altered gene expression.

18.2 ENDOCRINE SYSTEM OF MAN

Endocrine system is the type of glandular system, consists of some 20 ductless glands lying in different parts of the body. Some of the major endocrine glands, their locations and hormonal secretions are shown in the figure 18.3.

18.2.1 Neurosecretory Role of Hypothalamus

Hypothalamus is part of forebrain. It regulates a wide spectrum of physiological functions such as hunger, thirst, sleep and temperature. Hypothalamus also monitors metabolites and hormone levels in the blood. The hypothalamus is the master control centre of the endocrine system. Its endocrine signals directly control the pituitary gland. It contains special groups of **neurosecretory cells.** These cells conduct impulses and have developed secretory capacity to a high level. These cells produce regulatory hormones which regulate the synthesis and secretion of pituitary hormones. The hormones produced by the hypothalamus are either the **releasing factors** which stimulate secretions of pituitary hormones or **inhibiting factors** which inhibit secretion of pituitary hormones. These are produced in the cell bodies of the cells and packed into the granules and are transported down to the axon by cytoplasmic streaming. The axon endings of the neurosecretory cells synapse with blood capillaries and release their hormones into the blood when stimulated. The hormones from hypothalamus and their functions are given in the table 18.1.

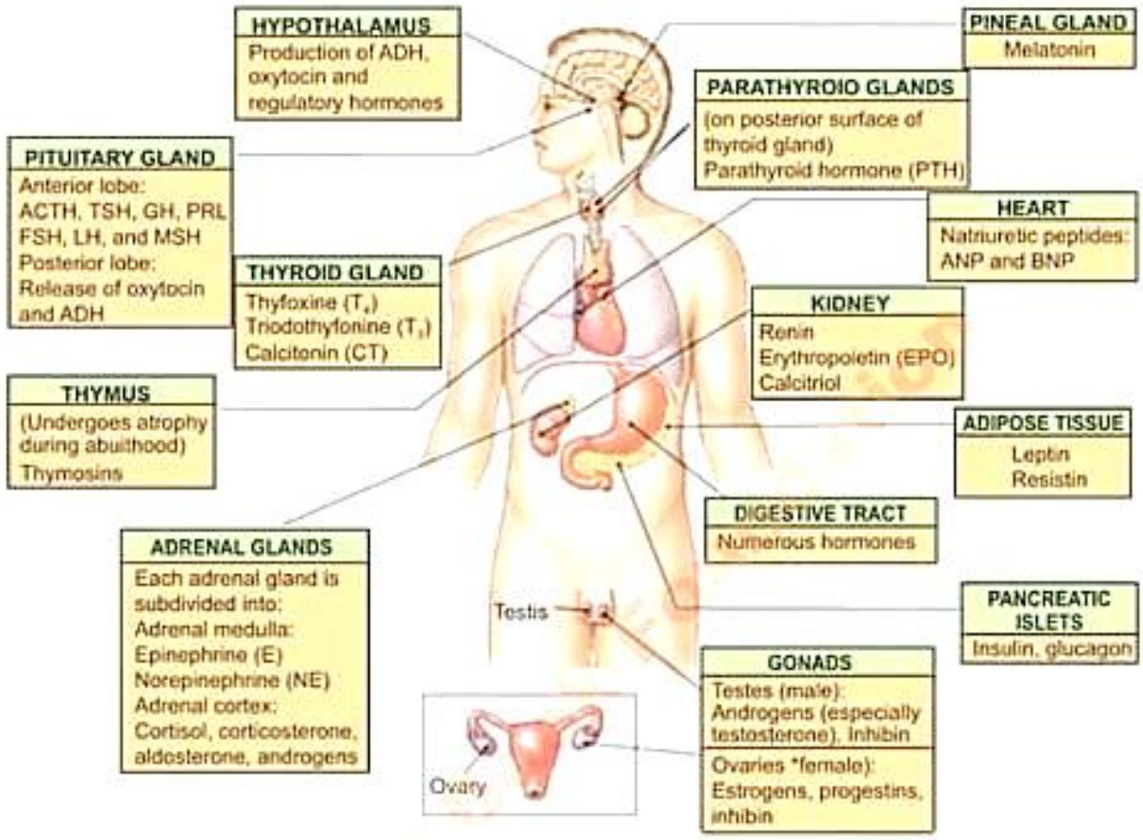


Fig. 18.3 Major endocrine glands and their locations in human

Table 18.1 Hypothalamic hormones and their effect on pituitary gland

Hormone from the hypothalamus	Anterior pituitary response
Growth hormone releasing factors **(GHRF)**	Secretion of growth hormone **(GH)**
Somatostatin	Inhibition of **GH**
Thyrotrophin releasing factor **(TRF)**	Secretion of thyroid stimulating hormone **(TSH)**
Adrenocorticotrophin releasing factor **(CRF)**	Secretion of adrenocorticotrophic hormone **(ACTH)**
Prolactin inhibiting factor **(PIF)**	Inhibits secretion of prolactin
Gonadotrophin releasing hormone **(GnRH)**	Secretion of **FSH** and **LH**

In addition, the neurosecretory cells that arise from the hypothalamus also produce two **primary hormones** i.e., antidiuretic hormone (ADH) and oxytocin which are stored in posterior lobe of pituitary gland and are released from here when needed.

18.2.2 Pituitary Gland

Pituitary gland is located just below the hypothalamus. It is attached to hypothalamus by a stalk called **infundibulum** which is composed of blood vessels and the fibres of neurosecretory cells. Pituitary gland is divided into three lobes, the anterior, posterior and the median.

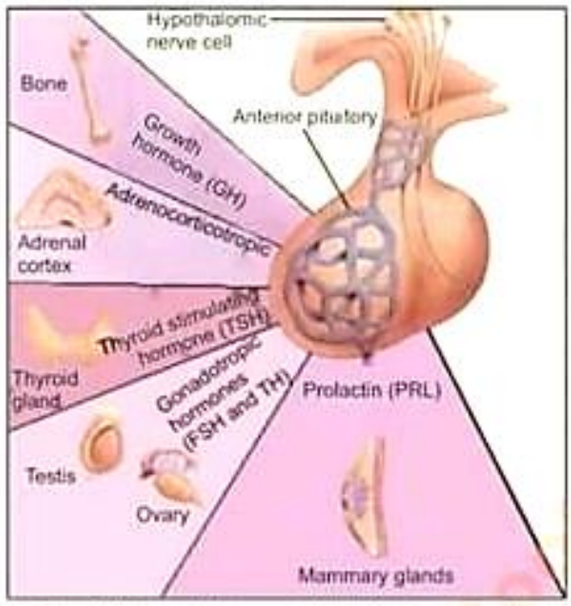


Fig. 18.4 Hypothalamus and anterior pituitary gland

Anterior lobe of pituitary

Classically, the anterior pituitary is considered the **master gland** of the endocrine system because it secretes numerous hormones, many of which regulate the activity of other endocrine glands. It secretes six hormones, all of which regulate the secretory action of other endocrine glands.

Growth hormone (GH) or **Somatotrophic hormone (STH)**. It is released under influence of hypothalamic **growth hormone releasing factor (GHRF)** and are inhibited by **hypothalamic somatostatin**. GH has a direct effect on growth and development. GH stimulates cell growth and cell division. It also stimulates uptake of amino acids into cells and increases rate of protein synthesis.

Deficiency of GH results in **dwarfism** in which development is much slower and individual has short stature, however, the body parts stay in proportion and brain development and IQ are unaffected.

Gigantism is result of over secretion of GH during childhood in which the bones are still capable of growth and person increase in height abnormally. Over secretion of GH in adult life causes **acromegaly** in which bones are no longer capable of increasing in length but grow in thickness. Acromegaly is characterised by enlarging the hands, feet, skull, nose and jawbone.

Thyroid stimulating hormone (TSH): Thyrotrophin releasing factor (TRF) from hypothalamus stimulates the synthesis and release of **thyroid stimulating hormone (TSH)** from the anterior pituitary. TSH regulates the endocrine function of the thyroid gland. It increases the number of cells and secretory activity of the thyroid gland. Over secretion of TSH causes **hyperthyroidism** i.e., excess of thyroxin and its under secretion causes **hypothyroidism** i.e., lack of thyroxin.

Adrenocorticotropic Hormone (ACTH): It is secreted by the release of **corticotrophin releasing factor (CRF)** from hypothalamus which is controlled by steroid level in the blood and by direct nervous stimulation of the hypothalamus as a result of stress e.g., cold, heat,

pain, fright and infections. ACTH acts on adrenal cortex and stimulates the secretion of corticosteroids (cortisone and aldosterone).

Follicle stimulating hormone (FSH), **luteinising hormone** (LH, also called **interstitial cell stimulating hormone**, ICSH in the male) and **prolactin** or **leuteotrophic hormone (LTH),** are all collectively known as **gonadotropic hormones**. These hormones act upon reproductive system and regulate its function.

Median lobe of pituitary

In humans, median lobe of pituitary is not very prominent. It is a thin layer of cells between the anterior and posterior pituitary. It produces **melanocyte stimulating hormone (MSH).** Secretion of MSH is regulated by hypothalamic MSH inhibitory hormone. Melanocyte stimulating hormone increases in humans during pregnancy too. It stimulates the production and release of melanin by melanocytes in skin and hair.

Posterior lobe of Pituitary

Posterior pituitary is not glandular by itself. It does not synthesize any hormone. It is largely made up of axons of neurosecretory cells of hypothalamus.

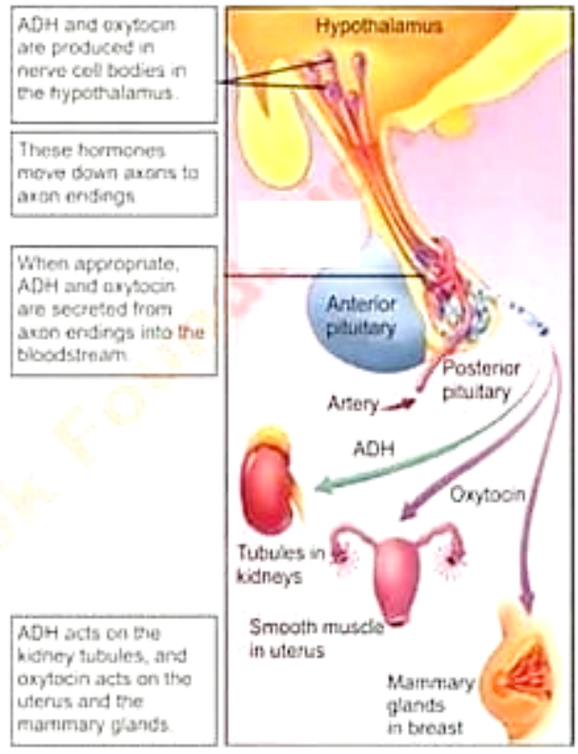


Fig. 18.5 Hypothalamus and posterior pituitary gland

Antidiuretic hormone: Posterior pituitary stores antidiuretic hormone (ADH or vasopressin) and oxytocin. These hormones are released in response to nerve impulses from hypothalamus. **ADH** is produced during the state of dehydration, decreased blood volume and low blood pressure. **Under secretion** of ADH causes **diabetes inspidus** which is characterized by excessive production of diluted urine and frequent thirst. Over secretion may leads to the kidney problems.

Oxytocin: It is released during child birth and in nursing women. During birth it is released in waves, and results in labour contractions. **Over secretion** causes rupturing of uterine wall while **under secretion** of oxytocin inhibits normal labour process. In lactating women, suckling causes the release of oxytocin. During this feeding process it causes the dilation of milk ducts of mother's mammary glands and thus promotes milk ejection.

Science Titbits

Under certain conditions, such as severe blood loss, exceptionally large amounts of ADH are released, causing a rise in blood pressure. The alternative name for this hormone, vasopressin, reflects this particular effect.

18.2.3 Thyroid Gland

Thyroid gland is composed of two lobes which are located on either side of the trachea inferior to the larynx. Thyroid gland produces three active hormones, tri iodothyronine (T3), tetra iodothyronine (T4) or thyroxin, and calcitonin.

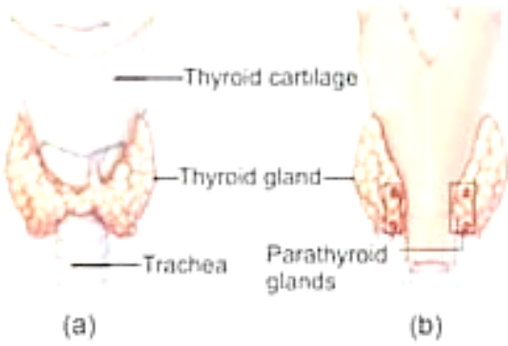


Fig. 18.6 Thyroid and parathyroid gland

T3 and T4: These are iodine containing hormones. **Tri iodothyronine** contains three iodine atoms in structure and **thyroxin** contains four, hence the names T3 and T4. TSH from anterior pituitary stimulates production and release of these hormones. These hormones show a variety of physiological effects: (1) They promote basal metabolic rate of the body. (2) Enhance glucose catabolism and synthesis of cholesterol in the liver. (3) Promote development of nervous system in foetus and infants. (4) They act on muscles for their development and functioning. (5) Promote growth and maturation of skeleton. (6) These hormones also promote normal motility of the gastrointestinal tract.

Hyperthyroidism term is applied to excess of these hormones. **Over secretion** of T3 and T4 causes **Graves' disease**.

Science Titbits

T_4 (also known as thyroxin) is the major hormone, about 90%, secreted by the thyroid; T_3 is only 10%. T_3 is four times more potent than T_4; however, action duration of T_4 is four times more than T_3.

DID YOU KNOW?

Graves' disease is believed to be an autoimmune disease. The serum of patients contains abnormal antibodies that mimic TSH and continuously stimulate thyroxin release. The symptoms include high metabolic rate, rapid and irregular heartbeat, increased breathing rate,increased body temperature, sweating and weight loss despite adequate food intake. Mostly exophthalmia (protrusion of the eyeballs) results from Graves's disease and is a classic symptom of hyperthyroidism.

Science Titbits

Hormones released from kidney

Renin monitors blood pressure and takes corrective action if it drops.

Erythropoietin acts on the bone marrow to increase the production of red blood cells. Stimuli such as bleeding or moving to high altitudes (where oxygen is scarcer) trigger the release of EPO.

Calcitriol acts on the cells of the intestine to promote the absorption of calcium from the diet.

Hypothyroidism is the **under secretion** of thyroxin. In adults, the full-blown hypothyroid syndrome is called **myxedema** which is characterized by low metabolic rate, feeling chilled, puffy eyes, thick and dry skin with hair lost from the scalp and eyebrows, oedema, tongue swelling, constipation; and enlarged thyroid gland i.e., goiter. Myxedema may result due to

deficiency of iodine in diet. **Congenital under secretion** results in a severe **hypothyroidism** in infants called **cretinism** which is characterized by mental retardation with poor physical growth and disproportionate body size. Bone maturation and puberty are severely delayed and infertility is common.

Calcitonin: Excessive Ca^{2+} level of blood stimulate release of calcitonin whereas declining blood Ca^{2+} levels inhibit its secretion. Calcitonin increases the deposition of calcium in bone matrix. Calcitonin inhibits Ca^{2+} absorption by the intestine and decreases its reabsorption by the kidney tubules allowing its excretion in urine.

Calcitonin appears more important in childhood, when the skeleton grows quickly and the bones are changing dramatically in mass, size, and shape. If deficient, Ca^{2+} are not deposited in bones and high blood Ca^{2+} level causes disturbance in the functioning of muscles and nervous system and may lead to kidney stones.

18.2.4 Parathyroid Glands

In human, there are four parathyroid glands. All four glands are located on the thyroid gland. They are small, light coloured masses that stick out from the posterior surface of the thyroid gland. The **parathormone** is the single most important hormone of parathyroid controlling the calcium balance of the blood. Its release is triggered by low blood Ca^{2+} levels and inhibited by high blood calcium levels. Parathormone works antagonistically to the calcitonin.

Over secretion of parathormone is usually a result of a parathyroid gland tumour. Calcium is released from the bones, and bones deform soften and tend to fracture spontaneously. Blood calcium level elevates (hypercalcemia) which depresses nervous system and causes weakness of muscles. Excess calcium salts precipitate in the kidneys leading to stone formation.

Under secretion of parathormone causes **hypocalcemia**. This increases the excitability of neurons. Also it can lead to tetany in which muscles remain in contracted state. If untreated, it can be fatal.

18.2.5 Pancreas

Pancreas is composed of two types of tissues. **Exocrine tissue** produces and secretes digestive juice. **Endocrine tissues** are distributed in the form of patches in the pancreas and these patches are called **Islets of Langerhans**. Islets of Langerhans secrete two hormones **insulin** and **glucagon**.

Science Titbits

Pancreatic **acinar cells** are functional units of the exocrine pancreas. They synthesize, store, and secrete inactive digestive enzymes into the lumen of the acinus.

Insulin is secreted by the **Beta (β) cells** which are larger in number and glucagon is secreted by **alpha (α) cells** which are lesser in number. These cells respond directly to the level of blood glucose. **Insulin** is secreted when the level of blood sugar rises, such as right

after a meal. Its overall effect is to reduce blood glucose level to the normal level by increasing the rate of glucose uptake by most body cells especially skeletal muscles and fat cells. It promotes **glycogenesis** (conversion of glucose to glycogen), increases the use of glucose in cellular respiration, promotes the conversion of excess glucose to fats and inhibits **gluconeogenesis** (glucose synthesis).

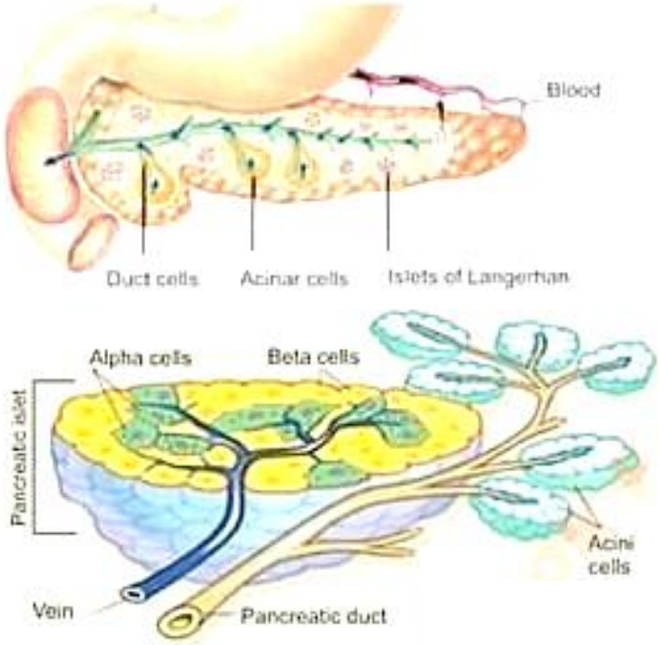


Fig. 18.7: Pancreas and Islets of Langerhans

The under secretion of insulin leads to the metabolic disease known as **diabetes mellitus** which is characterized by high glucose level in the blood and urine. If excess of insulin is produced the utilization of glucose is too great and its level falls in the blood which upsets nerve and muscles functioning.

Glucagon is released by α cells when blood glucose level is low. Sympathetic nervous system also stimulates its secretion. High blood glucose levels, insulin and somatostatin suppress its secretion. Its role is to increase the blood glucose level. It acts antagonistically to the insulin and thus reverses the activities performed by insulin.

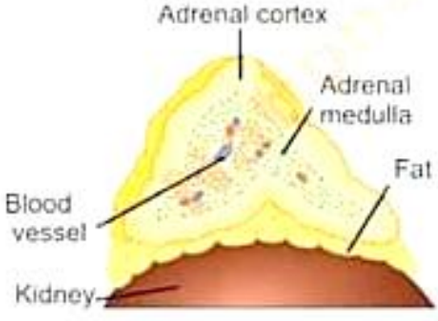


Fig. 18.8 Adrenal gland

18.2.6 Adrenal Glands

Each of the two adrenal glands rests on a kidney. Each adrenal gland is composed of an inner portion called the **medulla** and outer portion the **cortex**.

Epinephrine (adrenaline) and **norepinephrine** are produced by the **adrenal medulla**. Both are released during the state of emergency under the influence of sympathetic nervous system. Both are involved in the body's immediate response to stress. The two hormones exert the same effects in different ways i.e., synergistic effect. Epinephrine is the more potent stimulator of metabolic activities, bronchial dilation and increased blood flow to skeletal muscles and the heart but norepinephrine has the greater influence on peripheral vasoconstriction. The net effect is the rise in blood pressure.

Over secretion of these hormones may cause hypertension and aggressive behaviour during routine life. Under secretion causes failure to combat with emergency situation.

The two major types of hormones produced by the **adrenal cortex** are **glucocorticoids**, e.g., **cortisone**, which help to regulate the blood glucose level and **mineralocorticoids**, e.g., **aldosterone**, which help to regulate the level of minerals in the blood. Both are produced under the influence of ACTH. Under secretion of cortical hormones will lead to **Addison's disease** which is characterized by general metabolic disturbance, in particular, weakness of muscle action and loss of salts. Stress situation, such as cold which may leads to collapse and death. **Over secretion** of cortical hormone cause **Cushing's disease** which is characterized by excessive protein breakdown resulting muscular and bone weakness. Another hormone **androgen** (testosterone) is also produced from adrenal cortex in small amount in both male and female bodies. Its major site of secretion is testis, which are male gonads.

18.2.7 Gonads

Gonads are special type of endocrine glands which beside hormone secretions also produce gametes. Female gonads are ovaries while male gonads are testes.

Ovaries: The ovaries secrete female sex hormones estrogen and progesterone. **Estrogen** is secreted by **Graffian follicle** under the stimulation of FSH but estrogen has negative feedback upon FSH. Estrogen is secreted at the time of puberty and is responsible for secondary sex characteristics in females. It aids in healing and repair of uterine wall after menstruation. Due to its deficiency in the young females, they fail to mature sexually. Deficiency of this hormone in adults leads to sterility. Its over secretion may leads to the development of fibroids (abnormal growth) in uterus and polycystic ovaries.

Progesterone is produced by **corpus luteum** in response to LH during normal menstrual cycle but it is produced and released from placenta during pregnancy. It inhibits further FSH secretion from pituitary, thus preventing any more follicles from ripening. It causes further thickening and vascularisation of the uterus wall for maintaining state of pregnancy. Progesterone suppresses ovulation. Under secretion of progesterone during menstrual cycle, decreases the chance of pregnancy and may cause early menstruation. Under secretion during pregnancy may leads to the miscarriage.

Testes: The male gonads are testes. Testes produce sperm and male sex hormones called **testosterone** which is secreted from interstitial cells among seminiferous tubules under the influence of ICSH. During puberty, testosterone initiates the maturation of the male reproductive organs and the appearance of secondary sex characteristics and sex drive. In addition, testosterone is necessary for normal sperm production and maintains the reproductive organs in their mature functional state in adult males. Under secretion of this hormone causes the development of feminine characteristics and male sterility.

Skills: Interpreting and Communication

- **Explain on what grounds some companies claim that growth is possible in people having short heights.**

If growth hormone is administered to young people before growth of their long bones is complete, it causes long bones to grow and they will grow taller. To accomplish this however, GH would have to be administered over a considerable length of time.

Skills: Interpreting and Communication

- **Explain the role of artificially synthesized steroids in sports and their long term effects on its users.**

Steroids are artificial substances. Steroids are developed in order to do the job of testosterone. It can be classified as either anabolic or androgenic. Anabolic functions include those that promote formation of muscles, vertical growth and regulation of weight gain or loss. Androgenic refers to masculine attributes such as agility, strength, and endurance. By the help of these drugs, sportsmen can become bigger, stronger, more agile, and hence more competitive. Artificial steroid users carry many severe health risks. Major medical problems associated with steroids include a weakened immune system, liver disease, kidney disease, high blood pressure, high cholesterol, increased risk for heart disease, blood clots, strokes, tissue damage and cancer.

Science Titbits

The **pineal gland** is attached to the hypothalamus. Its primary hormone is **melatonin**. It influences daily rhythms called circadian rhythm. The **thymus** reaches its largest size and is most active during childhood. Thymus produces various hormones called **thymosin.** Certain lymphocytes that originate in the bone marrow and then pass through the thymus are transformed into T lymphocytes with the help of this hormone.

18.2.8 Other Endocrine Tissues/Cells

Hormones are also produced by organs or tissues whose function is not primarily an endocrine one. Even nerve cells produce hormones. The hormone **gastrin**, produced by the **stomach wall**, travels in the blood stream but exerts its effect locally, stimulating the production of pepsinogen and hydrochloric acid. **Secretin** and **cholecystokinin** control pancreatic and liver secretions. Both are formed in the cells of duodenal wall. The **placenta** secretes **progesterone,** which maintains pregnancy. **Prostaglandins** are a group of localized hormone. They provide protection during infections. **Endorphins** are produced in the brain. Endorphins bind to pain receptors and so block sensation of pain.

18.3 FEEDBACK MECHANISM

It is a type of interaction in which a controlling mechanism is itself controlled by the product of reactions it is controlling. After receiving the signal, a change occurs to correct the deviation by depressing it with negative feedback or enhancing it with positive feedback.

Positive feedback

In positive feedback an end product speeds up its production. These responses are not homeostatic and are rare in healthy individuals An example of positive feedback is childbirth. The early contractions of labour begin to force the baby's head against the cervix to dilate (open). Stretch-receptive neurons in the cervix respond to this extension by signalling the hypothalamus, which response by triggering the release of the hormone **oxytocin** that stimulates more and stronger uterine contractions. Stronger contractions create further pressure on the cervix, which in turn prompts the release of more hormones. The feedback cycle is finally terminated by the expulsion of the baby and its placenta.

Fig. 18.9 Positive feedback mechanism

Negative feedback

In negative feedback end products results in the reversal of the direction of change and tends to maintain homeostasis. In this system an endocrine gland is sensitive either to the concentration of a substance it regulates or to the concentration of a product from a process it controls. For example if blood glucose becomes too high, beta (β) cells in the islets of Langerhans respond by releasing insulin. Insulin lowers blood glucose by making cell surface membranes more permeable to glucose. It activates transport proteins in the membranes, allowing glucose to pass into the cells. Insulin also activates enzymes inside the cells. Some of these enzymes convert glucose to glycogen. If the levels of blood glucose get too low, alpha (α) cells in the islets of Langerhans secrete glucagon. This hormone fits into the receptor sites on the cell surface membranes, and activates the enzymes inside the cells that convert glycogen to glucose. The glucose then passes out of the cells and into the blood, raising blood glucose levels. In this way, negative feedback mechanism controls blood glucose.

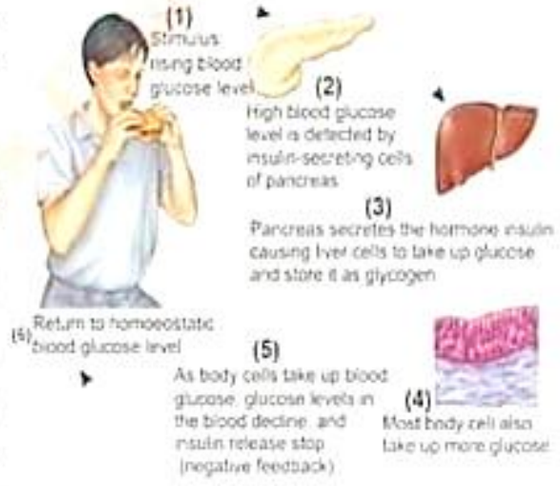


Fig. 18.10 Negative feedback mechanism

Critical Thinking

Is the sensation of thirst associated with a negative or positive-feedback mechanism?

Activity

Use the information in this chapter to construct a chart listing the actions of each hormone discussed. Your chart should include three headings: Endocrine gland, Hormone, Acton of hormone

Exercise

M.C.Qs

1. Select the correct answer

(i) Steroid hormones are secreted by

(A) the adrenal cortex (B) the gonads

(C) the thyroid (D) both A and B

(ii) Examples of posterior pituitary hormones are

(A) FSH and LH (B) prolactin and parathormone

(C) melatonin and prostaglandin (D) ADH and oxytocin

(iii) The primary targets for FSH are cells in the

(A) hypothalamus (B) ovary

(C) thyroid (D) pituitary

(iv) Which of the following controls the activity of all others?

(A) thyroid (B) pituitary

(C) adrenal cortex (D) gonads

(v) Which of the following have antagonistic (opposing) effects?

(A) parathyroid hormone and calcitonin (B) glucagons and thyroxine

(C) growth hormone and epinephrine (D) cortisone and ACTH

(vi) Which of the following hormones has broadest range of targets?

(A) ADH (B) oxytocin

(C) TSH (D) epinephrine

(vii) The pancreas increases its output of insulin in response to

(A) an increase in body temperature (B) changing cycle of dark and light

(C) a decrease in blood glucose (D) and increase in blood glucose

Short Questions

2. Why hormones are called chemical messengers?
3. Where are receptors located in proteinous hormones and steroid hormones?
4. What are neurosecretory cells?
5. Name the hormones of anterior pituitary gland.
6. Why the anterior lobe of pituitary gland is called master gland?
7. Describe the median lobe of pituitary gland.
8. How the secretion of ADH is controlled?
9. Write the differences between:
   (a) exocrine and endocrine glands
   (b) steroid hormones and proteinous hormones
   (c) first messenger and second messenger
   (d) receptor of proteinous hormones and steroid hormones
   (e) hypothyroidism and hyperthyroidism
   (f) calcitonin and parathormone
   (g) beta and alpha cells of Islets of Langerhans
   (h) insulin and glucagon
   (i) diabetes inspidus and diabetes mellitus
   (j) estrogen and progesterone
   (k) positive and negative feedback

Extensive Questions

10. Describe the chemical nature of hormone.
11. Describe the mode of hormone action.
12. Describe the hormones secreted by the anterior lobe of pituitary gland.
13. Describe the hormones secreted by the posterior lobe of pituitary gland.
14. Describe the hormones secreted by tissues and cells.

17

NERVOUS COORDINATION

After completing this lesson, you will be able to

- Recognize receptors as transducers sensitive to various stimuli.
- Trace the path of a message transmitted to the CNS for processing.
- Identify the three neurons (sensory, intermediate, motor) involved in nervous transmission.
- Identify muscles and glands as the effectors.
- Predict from every day experience what various kinds of receptor can be found in human body.
- Describe the detailed structure of a sensory neuron, associative and a motor neuron and relate the specialization in structures with functions.
- Differentiate between myelinated and non-myelinated neurons.
- Explain the function of the three types of neurons with the help of a reflex arc.
- Draw and label the structure of three kinds of neuron.
- Define nerve impulse.
- Describe the generation and transmission of nerve impulse.
- Name the factors responsible for the resting membrane potential of neuron.
- Evaluate from a graph the phenomena of polarization, depolarization and hyperpolarisation of membrane.
- Compare the velocities of nerve impulse in the axon membrane and in the synaptic cleft.
- Describe the structure of synapse.
- Explain synaptic transmission of nerve impulse.
- Classify neurotransmitters as inhibitory and excitatory and list some common examples.
- Basic Organization of Human Nervous System.
- Identify the main components of the nervous system.
- Explain briefly the functions of major divisions of brain.
- Describe the architecture of human brain and compare its sectional view with that of the spinal cord.
- Describe cranial and spinal nerves in man.
- Explain the structure, types and functions of autonomic nervous system.
- Explain the structure and functioning of the receptors for smell, taste and touch / pain.
- Draw a labeled diagram of the human brain.
- Identify different components in the diagram of CNS and PNS.
- Effect of Drugs on Nervous Coordination.
- Define narcotic drugs as agents that interact with the normal nervous activity.
- Compare the use and abuse of drugs with respect to heroine, *Cannabis*, nicotine, alcohol and inhalants like nail polish remover and glue.

- Define and explain the terms; drug addiction and drug tolerance with reference to caffeine and nicotine.
- Associate the effects of drug addiction and tolerance with the functioning of nervous system.
- Describe withdrawal symptoms of alcohol.
- Disorders of Nervous System and Diagnostic Tests
- Classify nervous disorders into vascular, infectious, structural, functional and degenerative disorders.
- Describe the causes, symptoms and treatment one type of each category of disorders outlined above. (e.g., stroke as vascular, meningitis as infectious, brain tumor as structural, headache as functional, and Alzheimer disease as degenerative disorder).
- Explain the principles of the important diagnostic tests for nervous disorders i.e. EEG, CT scan and MRI.
- Conceptualize the activity of brain as an electrical activity, which can be recorded using magnets and tomography.
- Compare the MRI scan of the brain of a sleeping human with that of a fully awake individual.
- Justify the way nervous system helps to coordinate complex and intricate movements of hand to play a piano, or write alphabets.
- Ascertain the effect of nerve gas as an inhibitor of acetylcholinesterase.
- Justify that the development of a modern computer is in fact a product of the understanding of the way nervous coordination occurs in complex organisms like humans.
- Describe how this knowledge has helped humans to treat diseases like epilepsy, paralysis.

Reading

The body of an animal is frequently exposed to variety of stimuli in its daily life. For an appropriate response to a particular stimulus, usually more than one body parts are involved, their activities are coordinated either by nervous system or endocrine system or both. These two types of coordination systems you also have studied in grade X to some extent, but in this chapter we are mainly focusing on the human nervous system.

The system of the body that provides coordination through electric signals among different body parts for the response to a particular stimulus is called **nervous system**. Human nervous system is the most evolved among all the animals. The study of the structure of nervous system is called **neurology**.

Nervous coordination mainly comprises highly specialized cells, called the **neurons**. The function of a neuron is to detect and receive stimuli from different sensory organs (receptors) and then, integrate them to determine the mode of response of the living organism, and then commands for an appropriate response are transmitted to the other organ (effectors). Nervous coordination in higher animals therefore consists of three basic steps i.e., reception of stimulus, processing/analysis of information and response to stimulus.

Receptor act as transducer because it converts one form of energy into another form e.g., rod and cone cell in the retina of eye convert the light energy into nerve impulse (electro chemical energy).

17.1 NEURONS

Neurons are the basic structural and functional unit of the nervous system.

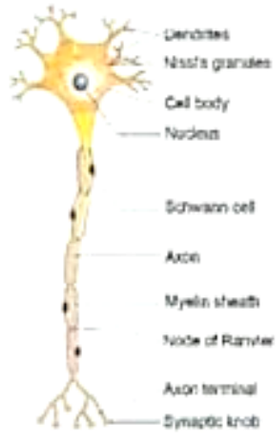


Fig. 17.1 Structure of neuron

17.1.1 Structure of Neuron

Although neurons vary considerably in size and shape, they all have three basic components: a cell body, dendrites and an axon.

Cell body

The cell body is called **neuron cell body**. It contains a mass of granular cytoplasm and cell membrane. The single large nucleus is centrally placed with a prominent nucleolus. Golgi apparatus, mitochondria and other organelles are present. The cytoplasm is characterised by the presence of **Nissl's granules**. These are group of ribosomes and rough ER associated with protein synthesis.

Dendrites

Dendrites are short and thin, often highly branched cytoplasmic extensions that are gradually tapered from their bases to their tips. Axons of other neurons form synapses with the dendrites. The function of the dendrite is to receive stimuli and conduct impulses to the cell body.

Axon

An **axon** is comparatively a long and thick nerve fibre which has a constant diameter and can vary in size from a few mm to more than a metre length. It may be branched or un-branched. Axons terminate by branching to form small extensions with enlarged ends called **presynaptic terminals**. Functionally, axons conduct action potentials from the neuron cell body to the presynaptic terminals, i.e., conduct signal (information) away from the cell body.

Myelin sheath and Schwann cells

Beside neuron, nervous system also consists of **neuroglia** or **glial cells**, which support, protect and nourish the neurons. **Schwann cells** are neuroglial cells in peripheral nervous system. Usually axons are covered by Schwann cells which are strip like cells wrap around axon fibres. Schwann cells are also covered by a fatty substance called **myelin sheath** that acts as an insulator. This is why axons are called **myelinated fibres**. A non-myelinated part of axon between two Schwann cells is called **node of Ranvier.**

Science Titbits

Unipolar neuron has a single process an axon that extends from the cell body and divides into two. In bipolar neurons, the cell body is located between the two processes: an axon and a dendrite e.g., retina of the eye. Multipolar neurons have three or more processes i.e., the several dendrites and one axon. Velocity of impulse in axon fibre depends upon the diameter, length and myelin sheath. The larger and thicker the axon, the faster it transmits information. The myelinated axons transmit information much faster than other neurons.

Types of neurons

However, all neurons vary somewhat in size, shape, and characteristics depending on the function and role of the neuron. Based upon function there are three types of neurons.

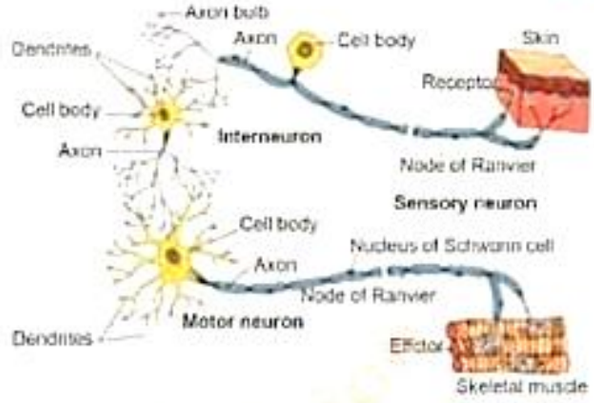


Fig. 17.2: Types of neuron

Sensory neurons conduct impulses towards the central nervous system from the sensory receptors. The cell body is at the end of a short stalk on one side of the main conducting fibre just outside the CNS. The branches at one end are connected to the receptor.

Motor neurons conduct impulses away from the central nervous system. The dendrites make contact with other neurons in the spinal cord. The terminal branches at the far end of the neuron are connected to an effector.

Interneurons occur entirely within the CNS. They convey messages between various parts of the CNS. The axon is comparatively thin and non-myelinated.

17.1.2 Reflex Arc

Reflex action is an immediate, automatic and involuntary response to external and internal environmental changes. The path of the nerve impulse during reflex action is called **reflex arc.**

Example

A typical reflex arc includes five fundamental parts: receptors, sensory neurons, interneuron, motor neuron and effectors. For example if one unexpectedly touches a hot object, the hand is rapidly removed from the source of heat. **Receptors** in the skin of the hand are activated by the heat of the object. The receptors stimulate a **sensory neuron** leading to the spinal cord via a spinal nerve. The cell body of the sensory neuron is outside the cord. The sensory neuron enters a **dorsal nerve root** of the spinal cord. The impulse then crosses a synapse to an **interneuron** which lies completely within the cord. The impulse travels along the interneuron and then passes across a synapse to the dendrites and the cell body of a **motor neuron** which lies within the spinal cord. The motor neuron eventually branches to form synapses with several muscle cells i.e., an effector. The nerve impulses then move along the motor neuron to the muscles, which cause them to contract.

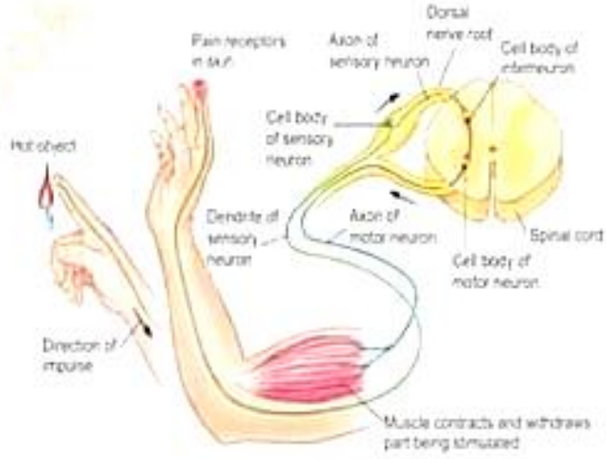


Fig. 17.3: A reflex arc

17.2 NERVE IMPULSE

Nerve impulse is information or signal about a stimulus that is transmitted from receptors to the CNS and from CNS to the effectors. In technical terms a nerve impulse can be defined as a wave of electrochemical changes that travel along the length of neuron, from one end to the other.

17.2.1 Generation and Transmission of Nerve Impulse

Here, word "electrochemical" refers to the electrical potential (a capacity to do electrical work) that exists on neuron membrane. In case of neuron the electrical potential is termed as **membrane potential** which is exhibited in two different forms i.e., Resting Membrane Potential (RMP) and Active Membrane Potential (AMP).

Resting membrane potential

It is characterized by more positive outer surface of neuron membrane than inner surface. This state is also referred as **polarized state** and the neuron is supposed to be at rest. This means that there is an unequal distribution of ions on the two sides of the nerve cell membrane. This potential generally measures about 70 mV (with the inside of the membrane negative with respect to the outside). So, the resting membrane potential is expressed as -70 mV, and the minus means that the inside is negative relative to (or compared to) the outside. It is called a **resting membrane potential** because it occurs when a membrane is not being stimulated or conducting impulses. Resting membrane potential is established by the following factors:

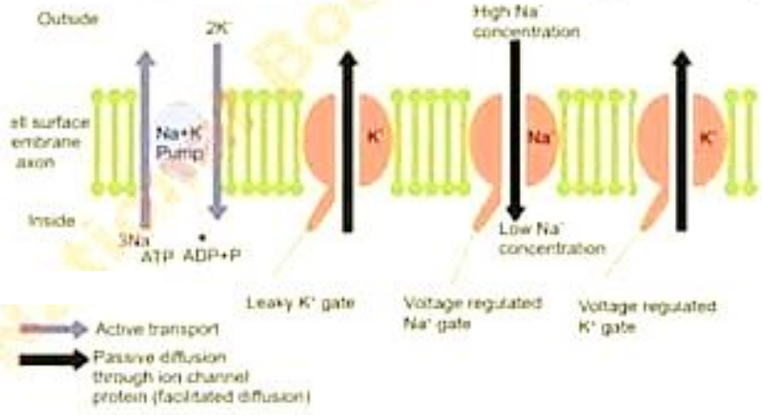


Fig. 17.4: Ionic movement across neuron membrane

Distribution and active movement of Na^+ and K^+ ions

The concentration of potassium (K^+) is 30 times greater in the fluid inside the cell than outside and the concentration of sodium ions (Na^+) is nearly 10 times greater in the fluid outside the cell than inside. These ions are continuously moved against their concentration gradient through sodium-potassium pumps by the expenditure of energy. For every two K^+ that are actively transported inward, three Na^+ are pumped out. So inside becomes more negative than outside of the neuron membrane.

Negative organic ions

There are many types of organic compounds in the neuron cytoplasm that also have negative charges. These ions include some amino acids, many proteins and RNA. Presence of these ions in the neuron cytoplasm makes inside of neuron more negative than outside.

Leakage of K^+ ions

Cell membrane of neuron also has many channel proteins called **gates**. K^+ ions leak continuously through leaky K^+ gates. This also makes more positive outside of neuron than inside.

Overall there are more positive charges on the outside than on the inside. This is known as **resting membrane potential**. This potential will be maintained until the membrane is disturbed or stimulated by a sufficiently strong **stimulus** (threshold), then action potential will be produced.

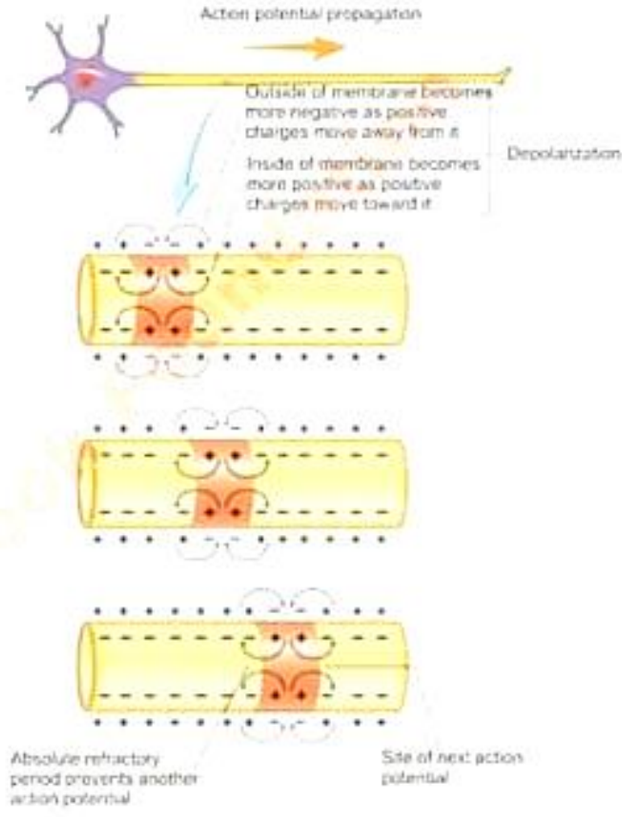


Fig. 17.5: Conduction of nerve impulse

Active membrane potential

Active membrane potential (also called action potential) is characterized by more positive inside of neuron than outside (depolarized state). This happens when positive charges tend to move inside of neuron on receiving a particular stimulus. This electrochemical change appears on a short region of neuron for a brief period of time followed by the recovery of polarized state. In this way a wave of action potential begins to move towards other end of neuron. Action potential is established by the following factors.

Threshold stimulus

If a stimulus is capable to produce action potential in neuron, it is called **threshold stimulus**. If stimulus is not capable to excite or fails to arise any response, it is called **sub threshold stimulus.**

Influx of Na^+ ions

When a neuron fibre is stimulated by threshold stimulus, it causes the opening of voltage regulated Na^+ gates. As a result Na gates permit the influx of Na^+ ions by diffusion. Since there are more Na^+ ions entering than leaving, the electrical potential of the membrane changes from -70 mV towards zero and then reaches to the 50 mV. This reversal of polarity across two sides of membrane is called **depolarization**. This electropositive inside and electronegative outside lasts for about one millisecond till the Na^+ gates are not closed.

Repolarization of neuron fibre

A fraction of second after the sodium gates open, depolarization of the axon membrane causes potassium gates to open. Potassium therefore diffuses out of the cell. Since the potassium is positively charged, this makes the inside of the cell more negative and starts the process of **repolarization.**

Hyper-polarization (More K^+ ions are on the outside than Na^+ ions on the inside)

At the peak of the action potential, the sodium gates start to close again. Sodium permeability therefore declines. The sodium-potassium pump continues to work during this time, so it gradually begins to restore the original resting potential. This repolarization is shown by the falling phase of the action potential spike and results in the membrane potential returning to its original level. In fact, there is a slight overshoot into a more negative potential than the original resting potential. This is called **hyperpolarization**. It is due to the slight delay in closing all the potassium gates compared with the sodium gates. As potassium ions continue to enter the axon their positive charge restores the normal resting potential.

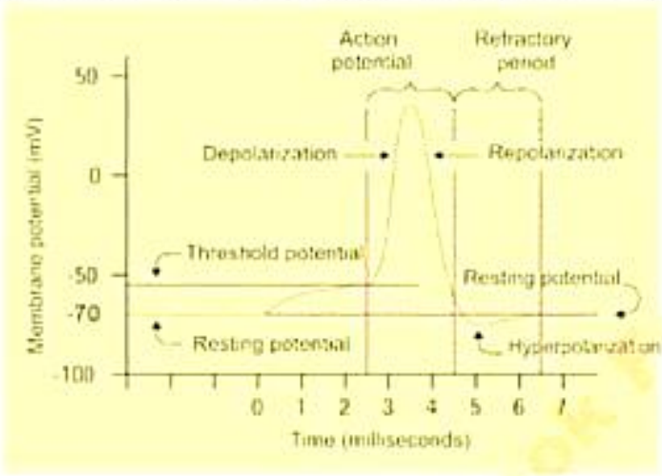


Fig. 17.6: Action potential in a neuron

Refractory period

After an action potential, nerve fibre undergoes a period of recovery in which it regains its original ionic distribution and polarity and prepares itself for the next stimulation. This period of recovery of nerve fibre is called **refractory period**. Although a repolarised neuron fibre has same polarity as that of a polarized neuron fibre but has different ionic distribution. It has more K^+ outside and more Na^+ inside. So the repolarised nerve fibre undergoes a refractory period of few milliseconds during which the original ionic distribution is restored by sodium-potassium pump which actively transports Na^+ ions out and K^+ ions in. This returns the membrane to its resting potential i.e., -70mV. Refractory period lasts for about 4 milliseconds so a neuron can conduct 250 impulses per second.

17.2.2 Velocities of Nerve Impulse

Velocities of nerve impulse in the axon membrane and in the synaptic cleft are variable. In human non myelinated fibres, nerve impulses travel at 1 to 3 metres per second. Myelinated fibres conduct at speeds of up to 120 meters per second. The velocity of nerve impulse is faster in myelinated neuron fibre due to saltatory conduction. Saltatory conduction is the rapid transmission of a nerve impulse along an axon, resulting from the action potential jumping from

one node of Ranvier to another, skipping the myelinated regions of membrane. It is up to 50 times faster than conduction through the fastest unmyelinated axons because they don't have to travel throughout every single space before moving to the next. Another reason that myelinated fibres conduct faster impulse is that myelin sheath acts as an insulating sheath and prevents loss of energy, so myelinated neuron fibres require less energy.

Velocity of nerve impulse also depends upon diameter of neuron fibres. Thick neuron fibres conduct faster impulse than thin fibres because resistance to electrical current flow is inversely proportional to the cross sectional area of the conductor, so with the increase in thickness of neuron fibres there is decrease in resistance of fibre to nerve impulse. The short journey across the synapse takes about a millisecond, longer than a nerve impulse takes to travel the same distance. This time is therefore called **synaptic delay**.

17.3 SYNAPSE

The junction between axon terminal of one neuron and the dendrite of another neuron, where information from one neuron is transmitted or relayed (handed over) to another neuron is called **synapse**.

17.3.1 Structure of Synapse

The neurons are not in direct contact at a synapse. There is a gap, called a **synaptic cleft** between them. A single neuron may form synapses with many incoming fibres of different neurons. A neuron which carries an impulse toward a synapse is called **presynaptic neuron**. A neuron which receives the impulse after it crosses the synapse is a **post synaptic neuron**.

17.3.2 Mechanism of Synaptic Transmission

The movement of impulse across the synapse is called a **synaptic transmission**. It takes place in the formation of a message which is transmitted across the synapse in the form of chemical messenger called **neurotransmitter**. The axons usually have several rounded **synaptic knobs** at their distal

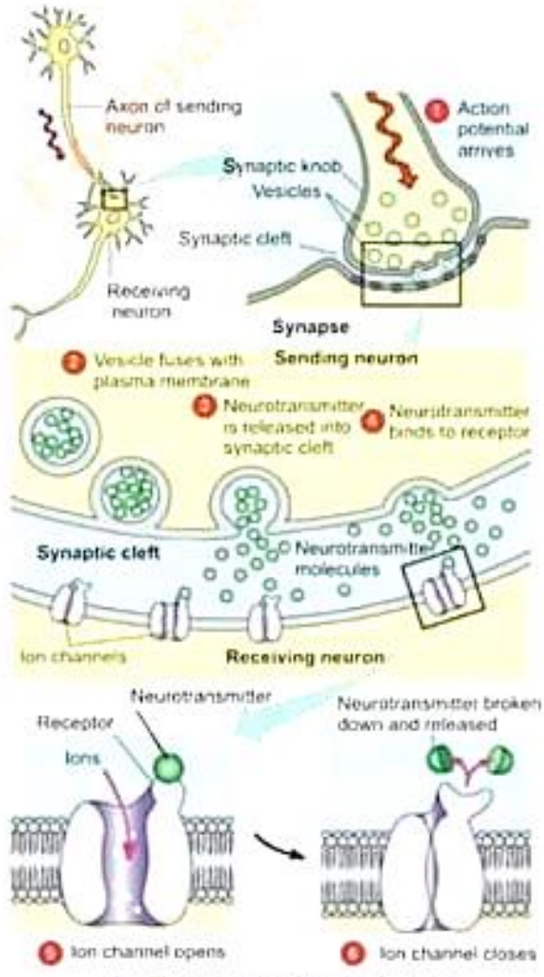


Fig. 17.7: Synaptic transmission

ends, which dendrites lack. These knobs contain numerous membranous sacs, called **synaptic vesicles** and when a nerve impulse reaches a knob, some of the vesicles respond by releasing a neurotransmitter. Fig: 17.8 shows the following numbered sequence: (1) An action potential (red arrow) arrives at the synaptic knob. Calcium channels open in the presynaptic membrane. As the calcium ion concentration inside the bulb is lower than the outside, calcium ions rush in. As the calcium concentration increases, synaptic vesicles move towards the membrane. (2) The neurotransmitter vesicles fuse with the plasma membrane of the transmitting cell. (3) The fused vesicles release their neurotransmitter molecules (green) into the synaptic cleft. (4) The released neurotransmitter molecules diffuse across the cleft and bind to receptor molecules on the postsynaptic cell surface membrane. (5) Binding of neurotransmitters to the post synaptic neuron receptors opens some channels and allows Na^+ ions to diffuse across the post synaptic membrane. As a result post synaptic membrane depolarizes and an action potential is generated. Since this depolarization brings the membrane potential towards threshold level, it is called **excitatory postsynaptic potential (EPSP).** (6) Once the neurotransmitters have acted on the postsynaptic membrane, they are immediately broken down by enzymes, like acetylcholine is hydrolyzed by **acetylcholinesterase** and adrenalin by **monoamine oxidase.**

Science Titbits

In electrical impulses, which are specialized for rapid signal transmission, the cells are separated by the synaptic cleft of only 0.2 nm, so that an action potential arriving at the pre - synaptic side of cleft, can sufficiently depolarize the post synaptic membrane to directly trigger its action potential.

17.4.3 Classification of Neurotransmitters

Neurotransmitters are classified as excitatory and inhibitory.

(a) Excitatory Neurotransmitters

Neurotransmitters that cause increased membrane permeability to sodium ions and, thus, trigger nerve impulses are said to be **excitatory**. **Acetylcholine** is an excitatory neurotransmitter of peripheral nervous system whereas **biogenic amines** (amino acid derivatives) are important neurotransmitters in central nervous system. They include epinephrine, norepinephrine, serotonin and dopamine, all of which also function as hormones. **Epinephrine** and **norepinephrine** increase the heartbeat rate during stress. **Serotonin** and **dopamine** affect sleep, mood, attention and learning.

(b) Inhibitory Neurotransmitters:

Other neurotransmitters cause decreased membrane permeability to sodium ions, thus causing the threshold of stimulus to be raised. This action is called **inhibitory**; because it lessens the chance that nerve impulse will be transferred to an adjoining neuron e.g., amino acids gamma-aminobutyric acid (GABA) and glycine. The **endorphins** are peptides that function as both neurotransmitters and hormones, decreasing our perception of pain.

Science, Technology and Society Connections

Ascertain the effect of nerve gas as an inhibitor of acetylcholinesterase.

Acetylcholine and the enzyme, acetyl cholinesterase, enable muscles to contract and relax. Normally, acetylcholine, a neurotransmitter, when released into the synapse of a muscle elicits the contraction of a muscle and is subsequently broken down by the enzyme, acetylcholinesterase, and relaxation of the muscle can occur. However, sarin, a nerve gas, irreversibly binds to acetylcholinesterase blocking it from breaking down the acetylcholine, thereby causing muscles to remain contracted. There exists a structural similarity between the active sites of the acetylcholine and the sarin molecule that enables the sarin molecule to fit into the acetylcholinesterase molecule. If the muscle is the diaphragm, it would remain contracted and the person would not be able to breathe. Nerve gases are extremely toxic; a small droplet can kill a person. They exist in both liquid and gaseous forms.

17.4 BASIC ORGANIZATION OF HUMAN NERVOUS SYSTEM

The human nervous system consists of central nervous system (CNS) and peripheral nervous system (PNS).The CNS is a coordinating centre and it lies in the midline of the body, whereas, the PNS transmits information from receptors to CNS and transmits orders and commands from CNS to effectors. An outline of divisions of human nervous system is given in figure 17.10.

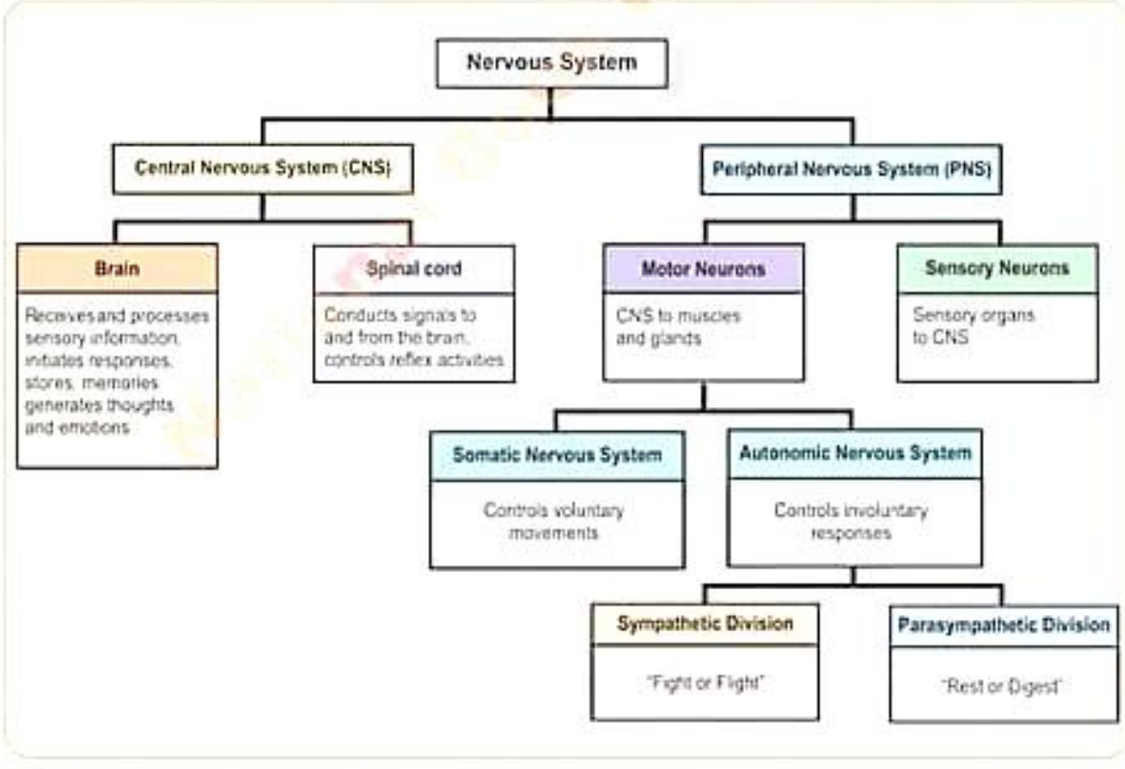


Fig. 17.8: Organization of human nervous system

17.4.1 Architecture of Brain and Spinal Cord and their Functions

Central nervous system

Central nervous system consists of brain and spinal cord, and both are hollow. The brain and spinal cord are covered with three protective membranes called **meninges** (singular: *meninx*). Brain is enclosed within the cranium while spinal cord is enclosed within vertebral column. The three meninges are **dura matter** (next to the cranium), **arachnoid matter** (middle membrane) **pia matter** (next to the nervous tissue). Between the arachnoid and pia matter there is a fluid, the **cerebrospinal fluid** (CSF), which helps to cushion the brain from shock.

Science Titbits

The surface of the cerebrum is called cerebral cortex. Cerebral cortex has many folds or convulsions forming ridges or gyri (singular, gyrus) which are separated by grooves. A shallow groove is called a sulcus (plural, sulci) and a deep groove is called a fissure. The two hemispheres are separated by longitudinal fissure.

The brain

The brain is divided into three part, forebrain, midbrain and hindbrain.

Forebrain consists of cerebrum, thalamus and limbic system.

Cerebrum is the largest part of the human brain. Cerebrum is divided into two **cerebral hemispheres** which are interconnected with each other by a band of axons, called **corpus callosum**. Each hemisphere contains four surface lobes: frontal, parietal, temporal and occipital lobe. Each lobe further contains different functional areas e.g., auditory (hearing) visual area etc. Each functional area consists of three sub-areas i.e., sensory area, association area and motor area. **Sensory area** receives impulses from different body parts. **Association area** interprets or analyzes the incoming information. The **motor area** controls responses of the body. Cerebrum also functions in the analysis and interpretation of memory, reasoning, judgement, thoughts and dreams.

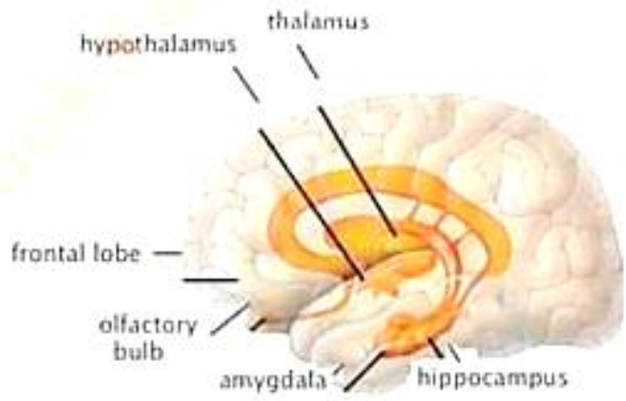


Fig. 17.9: Limbic system

Thalamus is below the cerebrum. It receives all sensory impulses (except sense of smell) and channels them to limbic system and to appropriate regions of the cortex for interpretation. The **limbic system** is a complex set of structures that lies on both sides of the thalamus, just under the cerebrum. It includes the **hypothalamus**, the **amygdala**, the **hippocampus**, and several other nearby areas. On the ventral side of the thalamus is the **hypothalamus**. It maintains homeostasis and contains centres for regulating hunger, sleep, thirst, body temperature, water balance and blood pressure, menstrual cycle and sleep wake

cycle. The hypothalamus also controls the pituitary gland and thereby serves as link between the nervous and endocrine system.

The **amygdalae** are two almond-shaped masses of neurons on either side of the thalamus. They control feeling and emotions of love, hate, anger, fear, rage and sexual arousal.

The **hippocampus** consists of two "horns" that curve back from the amygdala. It appears to be very important in converting things that are "in your mind" at the moment (in short-term memory) into things that you will remember for the long run (long-term memory).

Midbrain is reduced in humans. It acts as a relay station for tracts passing between the cerebrum and the spinal cord or cerebellum. Midbrain contains **reticular formation**, which is a relay centre connecting hindbrain with forebrain.

Hindbrain consists of cerebellum, medulla oblongata and pons. **Cerebellum** controls equilibrium i.e., body position and coordination of the actions of individual muscles to produce complex activities such as walking, running, riding bicycles, doing delicate work with hand. The cerebellum is also involved in learning memory storage for behaviour. **Pons** acts as a bridge between the cerebellum, medulla and cerebrum. It also controls rate and pattern of heartbeat and breathing. **Medulla** controls the automatic functions of the body, such as heartbeat, blood pressure, respiration, swallowing etc.

Brain is hollow structure as it has cavities called **ventricles**. There are four ventricles in the brain.

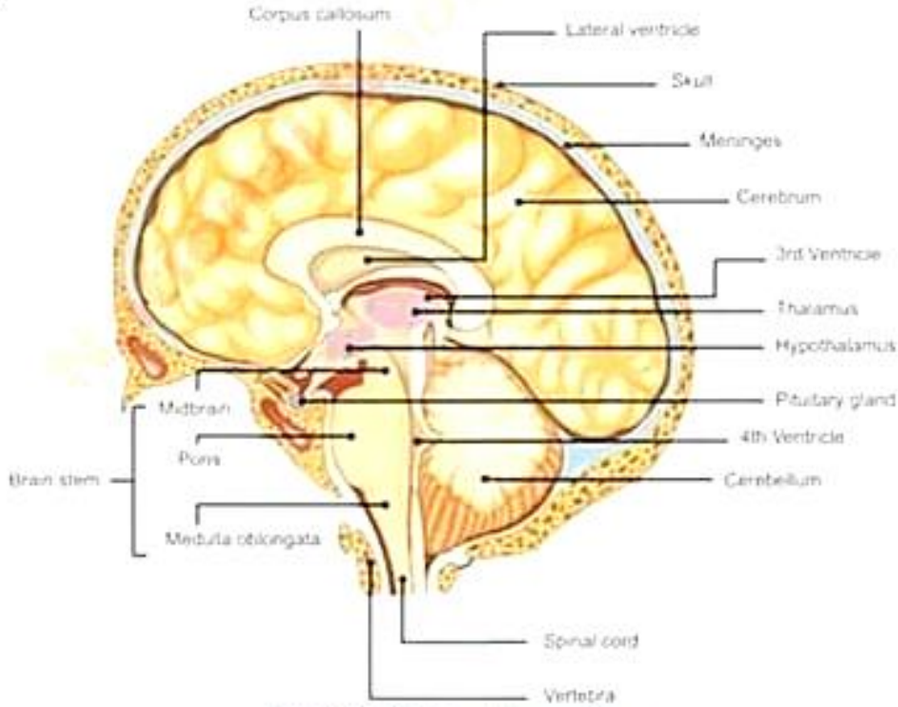


Fig. 17.10: Structure of human brain

Spinal cord

The spinal cord is the most important structure between the body and the brain. The spinal cord extends from the medulla to the level of the lumbar vertebrae. It is a vital link between the brain and the body. A transverse section of the adult spinal cord shows white matter in the periphery, grey matter inside, and a tiny central canal filled with CSF at its centre. **Grey matter** is shaped like the letter "H" or a "butterfly". The grey matter consists of neuron cell bodies and nonmyelinated parts of the fibres. The **white matter** is made up of bundles of myelinated fibres. Several pairs of spinal nerves originate from ventral and dorsal horn of grey matter. Dorsal root of spinal nerves, also contain ganglia present just beside the spinal cord. Arrangement of grey and white matter in brain is opposite to that of spinal cord.

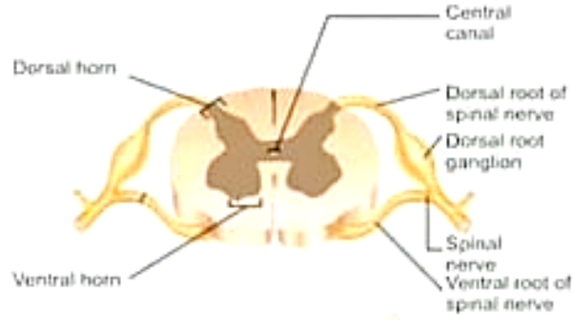


Fig. 17.11: Spinal cord anatomy

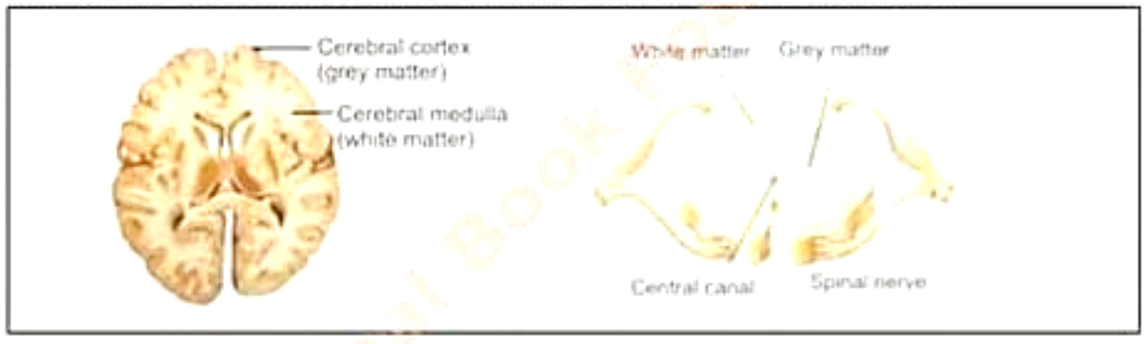


Fig. 17.12: Cross section view of brain and spinal cord

17.4.2 Cranial and Spinal Nerves in Man

The **peripheral nervous system** consists of the nerves that branch out from the central nervous system and connect it to other body parts. The peripheral nervous system includes **cranial nerves** which arise from the brain and the **spinal nerves**, which arise from the spinal cord.

There are twelve pairs of **cranial nerves**. Some of these are sensory nerves, some are motor nerves and others are mixed nerves. Cranial nerves are largely concerned with the head, neck and facial regions of the body. Thirty-one pairs of **spinal nerves** originate from the spinal cord. They are all mixed nerves and they provide two-way communication between the spinal cord and parts of the arms, legs, neck and trunk. Each spinal nerve emerges from the spinal cord by two short branches or roots, which lie within the vertebral column. The **dorsal root** contains the fibres of sensory neuron, which conduct impulses to the spinal cord. The **ventral root** contains the fibres of motor neurons, which conduct impulses away from the cord.

The two roots join just before a spinal nerve leaves the vertebral column. Each spinal nerve serves the particular region of the body in which it is located.

17.4.3 Somatic and Autonomic Nervous System

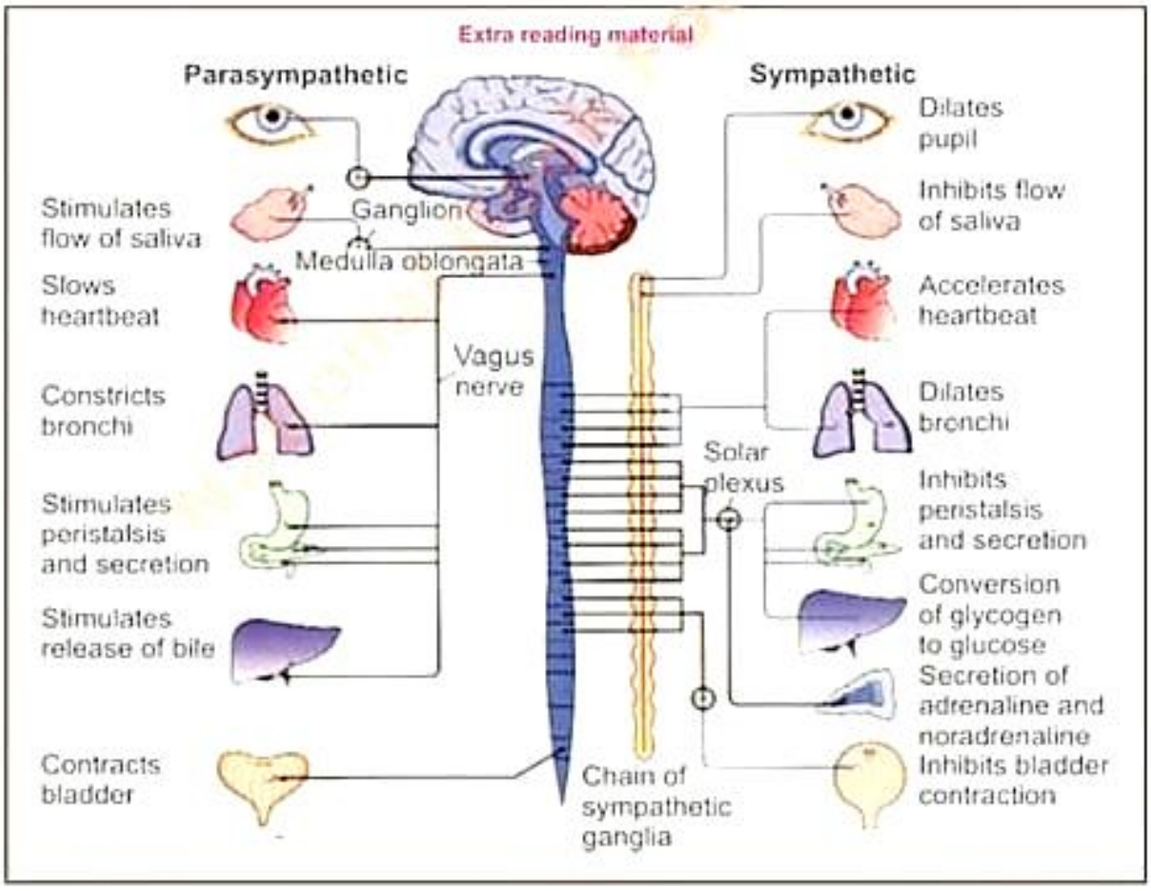
The **peripheral nervous system** can also be subdivided into the somatic and autonomic nervous systems.

Somatic nervous system

Generally, the **somatic nervous system** consists of the cranial and spinal nerve fibres that connect the CNS to the skin and skeletal muscles; it is involved in conscious activities.

Autonomic nervous system

The **autonomic nervous system** includes those fibres that connect the CNS to the visceral organs, such as the heart, stomach, intestines and various glands. It is concerned with unconscious activities. The autonomic system is divided into sympathetic and parasympathetic system. Both of these systems function automatically and usually subconsciously in an involuntary manner.



Sympathetic division

The **sympathetic division** controls various autonomic functions during the state of emergency. It prepares the body for fight or flight response. It consists of only spinal nerves. These nerves arise from first thoracic segment to second lumber segment of the spinal cord.

Parasympathetic division

A few cranial nerves, including the **vagus nerve**, together with nerves that arise from the sacral portion of the spinal cord, form the **parasympathetic division**. It controls various autonomic functions during the state of rest. In short, the parasympathetic system returns the body functions to normal after they have been altered by sympathetic stimulation. In times of danger, the sympathetic system prepares the body for violent activity. The parasympathetic system reverses these changes when the danger is over.

17.5.4 Sensory Receptors and their Working

The body must detect what is occurring inside and outside the body and is performed by sensory receptors. Here we will discuss receptors for smell, tastes, touch and pain.

Olfactory receptors

The smell or olfactory receptors are chemoreceptors, stimulated by chemicals dissolved in liquids. The olfactory organs, which contain the olfactory receptors, are present in the upper part of the nasal cavity. The olfactory receptor cells are neurons. These cells are surrounded by columnar epithelial cells having cilia at the distal ends. Chemicals that stimulate the olfactory receptors enter the nasal cavity as gases. They must dissolve at least partially in the watery fluids that surround the cilia before they can be detected.

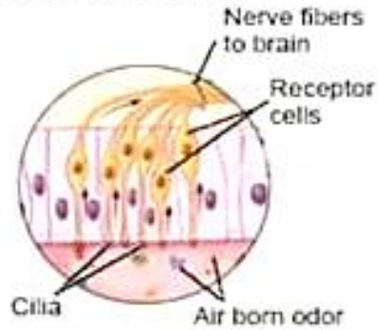


Fig. 17.13: Olfactory receptors in nasal epithelium

Taste receptors

Taste buds occur primarily on the surface of the tongue and are associated with tiny elevations called **papillae**. Each taste bud includes a group of modified epithelial cells, the **taste cells**, which function as receptors. The taste bud has an opening, the **taste pore** on its surface. Tiny projections, called **taste hairs**, protrude from outer ends of taste cells and just protrude through the taste pore. There are four primary taste sensations i.e., sweet, sour, salty and bitter, which are situated at various regions on the tongue. All the four regions overlap at certain places.

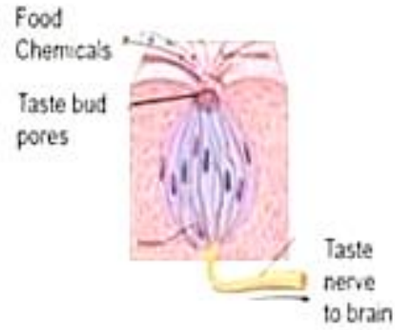


Fig. 17.14: Taste buds on tongue

Sensory receptors in human skin

The dermis of the skin contains receptors for touch, pressure, temperature and pain. **Meissner's corpuscles** and **Merkel disks** are touch receptors. These consist of small, oval masses of flattened connective tissue cells. Two or more sensory nerve fibres branch into each corpuscle. Meissner's corpuscles are especially numerous in the lips, fingertips, palm, and soles. **Paccinian's corpuscles** are also encapsulated nerve endings present in the fatty layer deep into the skin. They are concerned with sensation of pressure. Receptors for touch and pressure are also called **mechanoreceptors**. Skin also has cold and heat receptors to detect the temperature variations.

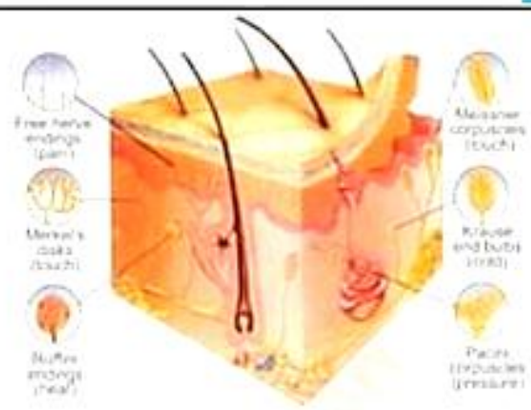


Fig. 17.15: Receptors in human skin

Pain receptors are technically called **nociceptors.** Pain receptors are located at the top of the skin in the epidermis area to detect pain. These receptors are free nerve endings that respond to chemicals released by damaged tissues or excess stimuli of heat or pressure. These receptors are widely distributed throughout the skin and inter tissues, except in the tissue of the brain.

17.5 EFFECTS OF DRUGS ON NERVOUS COORDINATION

A narcotic is a group of substances when administered diminish the perception of pain. Narcotics bind to certain painkilling sites in the brain. With constant use, they build up in the brain and block the production of **endorphins**, the brain's natural painkilling chemicals. Their side effects are inhibition of the endocrine and autonomous nervous system etc. The narcotics are the drugs that act as agents which interact with the normal nervous activity.

17.5.1 Narcotic Drugs

Common narcotic drugs are heroine, *Cannabis*, nicotine, alcohol.

Heroin

Heroin gives a feeling of euphoria along with relief of pain. Side effects can include nausea, vomiting, respiratory and circulatory depression leading to death.

Cannabis

It is the dried flowering tops, leaves and stem of Indian hemp plant *Cannabis sativa*. It includes marijuana and hashish. Usually the users report a mild euphoria, along with alterations in vision and judgement. Intoxication is recognised by the presence of hallucinations, anxiety and depression etc.

Nicotine

It is an alkaloid derived from tobacco. When smoking a cigarette, nicotine is quickly distributed to all body organs. In peripheral nervous system, nicotine stimulates postsynaptic

receptors (like acetylcholine) and leads to increased skeletal muscular activity. It also increases heartbeat rate and blood pressure.

Alcohol

Alcohol act as depressant and slows down the nervous communication. Its short term effects are impairment of vision, judgment and alertness. Long term chronic drinking can damage nervous system, liver, pancreas and development.

Withdrawal symptoms of alcohol

Individuals who eliminate addictive substances from their lives often feel withdrawal symptoms. Alcohol withdrawal refers to a group of symptoms that may occur from suddenly stopping the use of alcohol. The symptoms are feeling of anxiety, irritability, depression, headache, and hallucinations etc.

Inhalants

These are volatile organic chemicals, commonly referred to as "glue sniffing". Inhalant abuse now includes aerosols e.g., hair spray and anaesthetics e.g., ether etc. Inhalants rapidly start euphoria followed by central nervous system depression. Deep breathing of the toxic vapours may result in hallucinations or even death.

17.5.2 Drug Addiction and Drug Tolerance

> **Critical Thinking**
> What measures can be taken to eradicate drug addiction from the society?

Drug addiction: Drug addiction is a dependence on an illegal drug or a medication. You may want to quit, but most people find they can't do it on their own.

Drug tolerance: Drug tolerance is a person's diminished response to a drug, which occurs when the drug is used repeatedly and the body adapts to the continued presence of the drug. For instance, when **nicotine** or caffeine is used for a long time, larger and larger doses must be taken to produce the same effect.

Effects of drug addiction and tolerance on the central nervous system

Drugs are chemicals that interfere with the way neurons normally send, receive, and process information. Some drugs can activate neurons because their chemical structure mimics that of a natural neurotransmitter. Drugs interact with the brain and body to alter moods, emotions, and behaviors by changing brain chemistry. Regions of the brain affected by drug abuse are the brain stem, limbic system, and cerebral cortex.

All depressants work by slowing down the functioning of the central nervous system, while stimulants can produce a number of effects on the body such as increased heart rate, improved concentration, increased respiratory rate etc.

17.6 DISORDERS OF NERVOUS SYSTEM AND DIAGNOSTIC TESTS

The disorders of the nervous system may be classified as vascular, infectious, structural, functional and degenerative. While classifying the site of involvement is also considered. We will discuss here causes, symptoms and treatment of few diseases of the major categories.

17.6.1 Vascular Disorders of the CNS

Any disorder of nervous system which occurs due to abnormality in blood circulation is called vascular disorder of the nervous system e.g., strokes, brain haemorrhage.

Stroke

It occurs due to rupture of small cerebral arteries. **Cause:** The cause and risk factors for stroke include hypertension, cigarette smoking, diabetes mellitus, high alcohol intake, thrombosis, blood disorders, blood embolism and cocaine abuse. **Symptoms:** These include sudden loss of function in one region of brain. Weakness and heaviness occur in arm, leg or face. Paralysis occurs on the side of the body opposite the cerebral infarction (a portion of the tissue that is dying because of blood supply to it has been cut off). Aphasia (inability to express through words) may be present. **Treatment:** Medical treatment is aimed at preventing further attacks and stroke. Anticoagulants and platelet aggregation inhibitor (such as aspirin) is given. Blood pressure management and nursing care is essential.

17.6.2 Infectious Disorders of the CNS

Infections of the central nervous system can be caused by almost any infectious agent, including viruses, bacteria, fungi, protozoa, and Platyhelminthes.

Meningitis

It is an inflammation of the meninges. **Cause:** Bacterial or viral infection of meninges. **Symptoms** usually include stiffness in the neck, headache and fever. In severe cases, meningitis may also cause paralysis, coma or death. **Treatment:** For viral meningitis, there is no specific treatment. Bacterial meningitis is treated with antibiotics and steroids.

17.6.3. Structural Disorders of the CNS

Several disorders disturb the structure of brain are referred as structural disorders, such as tumours.

Tumour

It is an abnormal mass of neuroglial cells produced as a result of uncontrolled cell division. **Cause:** It is caused by mutation which may occur at any age in brain and spinal cord. **Symptoms:** These vary widely, depending on the location of the tumour but may include headaches, severe nerve pain, paralysis, seizures, coma and death. **Treatment:** Surgical removal of tumour.

17.6.4 Functional Disorders of the CNS

Headache

A **headache** is pain anywhere in the region of the head or neck. It can be a symptom of a number of different conditions of the head and neck. The brain tissue itself is not sensitive to pain because it lacks pain receptors. Rather, the pain is caused by disturbance of the pain-sensitive structures around the brain. There are two major categories of headaches i.e., primary headaches (due to the headache condition itself and not due to another cause) e.g., migraine, tension headache; and secondary headaches (due to an underlying structural problem in the head or neck such as bleeding in the brain, tumour, meningitis etc. Several analgesic drugs are available for **treatment** of any kind of headache.

17.6.5 Degenerative Disorders of the CNS

Many diseases cause degeneration in different part of the nervous system without an identifiable external cause. Genetic factors are known to be involved. Example of such diseases is Alzheimer's disease.

Alzheimer's disease

Alzheimer's disease is a slowly progressive disease of the brain that is characterized by impairment of memory and eventually by disturbances in reasoning, planning, language, and perception. Although onset of this disease occurs in aged peoples but it is not particularly associated with aging. **Cause:** There is genetic predisposition, so tends to run in families. **Symptoms:** Most prominent symptom is loss of short-term memory loss. **Treatment:** There is no effective treatment for this disease.

17.6.6 Diagnostic Tests for Nervous Disorders

These days number of diagnostic tests have been developed for nervous disorders. The principle of EEG, CT scan and MRI are discussed here.

Electroencephalography

Neurons within the cerebral cortex continuously generate electrical activity. This activity can be recorded by electrodes attached to precise locations on the scalp, producing **electroencephalogram** and this technique is called **electroencephalography (EEG)**. An EEG pattern is commonly called **brain waves.**

Fig. 17.16: Electroencephalography

Computed tomography scan

Computerized tomography is more commonly known by its abbreviated names, **CT scan**. It is an X-ray procedure that combines many X-ray images with the aid of a computer to generate cross-sectional views and, if needed, three-dimensional images of the internal organs and structures of the body.

A **CT scan** is used to define normal and abnormal structures in the body and/or assist in procedures by helping to accurately guide the placement of instruments or treatments.

Fig. 17.17: CT scan

Magnetic Resonance Imaging

Magnetic Resonance Imaging (MRI) scan is a radiology technique that uses magnetism, radio waves, and a computer to produce images of body structures. The MRI scanner is a tube surrounded by a giant circular magnet. The patient is placed on a moveable bed that is inserted into the magnet. The patient is exposed to strong magnetic field and beam of radio waves. The receiver information is processed by a computer, and an image is produced. The image and resolution produced by MRI is quite detailed and can detect tiny changes of structures within the body.

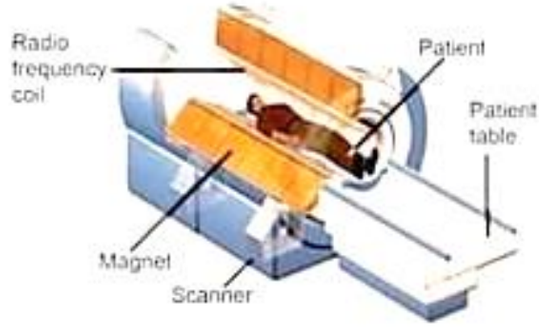


Fig. 17.18: MRI scanner

DID YOU KNOW?

In CT Scan a large donut-shaped X-ray machine or scanner called tomograph takes X-ray images at many different angles around the body. These images are processed by a computer to produce cross-sectional pictures of the body. In each of these pictures the body is seen as an X-ray "slice" of the body, which is recorded on a film. This recorded image is called a tomogram.

Skills: Interpreting and Communication

- **Conceptualize the activity of brain as an electrical activity, which can be recorded using magnet and tomography.**

Scientists have attempted to conceptualize electrical activity of the brain as a reflection of mental processes. Due to recent advances in computer software and hardware it is now possible to sample more electrical information from the brain by using different radiologic imaging technique such as CT scan, MRI Scan and EEG. Therefore, these techniques have become very useful for the diagnosis of brain disorders.

- **Compare EEG of the brain of a sleeping human with that of a fully awake individual.**

During wakefulness, alpha and beta activities are experienced in the human brain. Alpha activities consist of medium frequency waves. Beta activities consist of irregular low amplitude waves which are present when the individual is very alert and attentive. As the individual gets drowsy, brain experiences theta activities. This is the transition stage between wakefulness and sleep. Sleeping stage contains irregular theta activities where sleep spindles (short bursts of waves of 12-14 Hz) and K complexes (sudden sharp wave forms) are present. Then the next stage of sleep contains high-amplitude delta activities 20 to 50 percent of the time.

Exercise

M.C.Qs

1. Select the correct answer

(i) The cell transmits impulses from the
(A) effector organ to the spinal cord (B) receptor cells to the effector organ
(C) receptor cells to the spinal cord (D) spinal cord to the effector organ

(ii) Depolarization of an axon is produced by the movement of:
(A) Na^+ into the axon and K^+ out of the axon. (B) Na^+ into the axon to bond with K^+
(C) K^+ into the axon and Na^+ out of the axon
(D) Na^+ and K^+ within the axon towards the axon terminal

(iii) What will happen if the receptor sites on the post-synaptic membrane are blocked by a drug at the neuromuscular junction?
(A) inhibition of acetylcholine (B) inhibition of cholinesterase
(C) muscle contraction (D) muscle paralysis

(iv) Which of these are the first and last elements in a spinal reflex?
(A) axon and dendrite (B) sense organ and muscle effector
(C) ventral horn and dorsal horn (D) motor neuron and sensory neuron

(v) Impulses travel very rapidly along nerves to the leg of a man. Which fact accounts for the speed at which they travel?
(A) a nerve impulse is an all or none phenomenon
(B) the nerves contain myelinated fibres
(C) there is a high concentration of Na^+ ions inside the axons
(D) there is a potential difference across the axon membranes

(vi) Where are neurotransmitter receptors located?
(A) on the nuclear membrane (B) at nodes of Ranvier
(C) on the postsynaptic membrane (D) in the myelin sheath

Short Questions

2. Why is neuron co-ordination important?
3. Describe the receptors as transducers sensitive to various stimuli.
4. Name the five fundamental parts of human reflex arc.
5. What is the function of neurotransmitter?
6. What characteristics do the brain and spinal cord have in common?
7. What is limbic system?
8. Name the sensory receptors of human skin.
9. How narcotic drugs interact with the normal nervous activity.
10. Find out some of the common withdrawal symptoms of alcohol.
11. Write the differences between:
    (a) thermoreceptors and nociceptors
    (b) axoplasm and axolemma
    (c) neuroglial cells and Schwann cells
    (d) sensory neuron and motor neuron
    (e) reflex action and reflex arc
    (f) resting membrane potential and active membrane potential
    (g) depolarization and repolarization
    (h) repolarization and hyperpolarization
    (i) refractory period and absolute refractory period
    (j) presynaptic neuron and postsynaptic neuron
    (k) axon and dendndrite
    (l) synaptic knob and synaptic vesicles
    (m) pons and medulla
    (n) white matter and grey matter
    (o) myelinated and nonmyelinated nerve fibres
    (p) cranial nerves and spinal nerves
    (q) drug addiction and drug tolerance
    (r) somatic and autonomic nerves system

Extensive Questions

12. What is the basic organization of a nervous system?
13. Describe the structure of a neuron.
14. Describe the three types of neurons and write their functions.
15. Describe the mechanism of synaptic transmission.
16. Write the classification of neurotransmitters.
17. Describe the human brain.
18. Describe the structure of spinal cord with diagram.
19. Describe the somatic and autonomic nervous system.
20. Give an account of narcotic drugs.
21. Describe the cause, symptoms and treatment of:
    (a) Stroke (b) Headache (c) Meningitis
    (d) Tumour (e) Alzheimer disease
22. Explain the principles of the following diagnostic tests of nervous disorders:
    (a) EEG (b) CT scan (c) MRI

16

SUPPORT AND LOCOMOTION

After completing this lesson, you will be able to

- Describe the structure of bone and compare it with that of cartilage.
- Explain the functions of osteoblasts, osteoclasts and osteocytes.
- Identify the main divisions of human skeleton.
- List the bones of appendicular and axial skeleton of man.
- Describe three types of joints i.e. fibrous joints, cartilaginous joints and synovial joints and give example of each.
- Relate the bipedal posture of man with his skeleton and musculature.
- Identify the bones of the pelvic girdles, pectoral girdle, arms and legs by using the model of human skeleton.
- Describe the disorders of human skeleton (disc-slip, spondylosis, sciatica, arthritis) and their causes.
- State different types of fractures (simple, compound and complicated) and describe the repair process of simple fractures.
- Describe the injuries in joints (dislocation and sprain) and their first aid treatment.
- Describe the first-aid treatment for fracture.
- Compare smooth muscles, cardiac muscles and skeletal muscles.
- Explain the ultra-structure of the skeletal muscle.
- Explain the sliding filaments model of muscle contraction.
- Describe the action of antagonistic muscles in the movement of knee joint.
- Explain muscle fatigue, cramps and tetany.
- Differentiate between tetanus and muscle tetany.
- Compare the structure of skeletal, smooth and cardiac muscles with the help of prepared slides.
- Draw a diagram of sarcomere and label its parts.
- Justify how the main functions of the skeleton are to act as a system of rods and levers, which are moved by the muscles.
- Justify why do the muscles pull but do not push.
- Name the techniques for joint transplantation.
- Justify why the use of calcium in teenage and twenties can be a preventive action against osteoporosis.
- Reason out the rigor mortis.
- Relate improper posture to bone/joint problems.

Reading

Some support in living organisms is necessary to uphold and sustain the body against gravity and other external forces. As the living organisms have been increased in size through the process of evolution, the need for support became greater. This was particularly true once living organisms left water and colonized land. The skeleton in animals contributes to this support.

16.1 HUMAN SKELETON

Human skeletal system consists of bones and cartilage. The skeleton acts as a framework that supports soft tissues. It allows free movement through the action of muscles across joints. The study of bones and cartilage is called osteology.

16.1.1 Structure of Bone

An individual bone is composed of a variety of tissues, including bone tissue, cartilage, fibrous connective tissue, blood and nerve tissue. The terminal broad parts are called **epiphysis** and the middle part along the length of bone is called **diaphysis** or **shaft** which also contains a central cavity filled by **yellow bone marrow**. The outer connective tissue around the bone is called **periosteum** and the inner region is called **endosteum**. The

Fig. 16.1 Structure of bone

endosteum further consists of a peripheral part, called **compact** bone and the inner bone mass, called **spongy bone**. Most of the spongy bone is present in epiphysis. The **red bone marrow** is also found in the spaces of spongy bone.

There are three types of cells associated with bone (derived from osteogenic cells) i.e., **osteoblasts** are bone forming cells that synthesize and secrete unmineralized ground substance. Once the osteoblasts are surrounded by matrix, they become the osteocytes. **Osteocytes** maintain healthy bone tissue by secreting enzymes and influencing bone mineral content. They also regulate the calcium release from bone tissue to blood. **Osteoclasts** are bone destroying

Fig. 16.2 Types of bone cells

cells. Osteoclasts perform bone resorption, i.e., they breakdown bone and deposit calcium and phosphate in the blood. The work of osteoclasts is important to the growth and repair of bone.

16.1.2 Structure of Cartilage

Cartilage is not strong as bone. It is present at particular places only. It is more flexible than the bone because the matrix is gel like and contains many collagenous and elastic fibres. The cartilage matrix is covered by a dense layer of collagen fibres, called **perichondrium**. There are many small cavities distributed in the matrix called **lacunae** which contain cartilage cells. The living cells of cartilage are called **chondrocytes**. Unlike other connective tissues, cartilage does not contain blood vessels and the chondrocytes are supplied by diffusion. Because of this, it heals very slowly. Although the human skeleton is initially made up of cartilages and fibrous membranes, most of these early supports are soon replaced by bones. A few cartilages that remain in adults are of three types. **Hyaline cartilage** is found at the ends of long bones and in the nose, at larynx and trachea. **Fibrocartilage** contains wide rows of thick collagenous fibres is found in the disks located between the vertebrae, cartilage of knee. **Elastic cartilage** is found in the ear flaps and epiglottis.

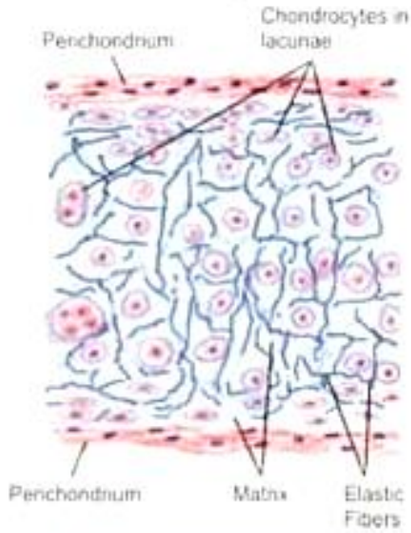


Fig. 16.3 Structure of cartilage

Table 16.1 Comparison between bone and cartilage

Feature	Bone	Cartilage
Collagen	Densely packed	Loosely packed
Cell types	Osteoblast, osteocytes and osteoclasts	Chondrocytes
Blood vessel	Present	Absent
Minerals	Deposit minerals such as calcium, carbonates, phosphates, etc.	No deposition of minerals
External covering	Covered by periosteum	Covered by pericondrium

16.1.3 Divisions of Human Skeleton

Human skeletal system consists of 206 bones which are primarily divided into two division i.e., axial skeleton and appendicular skeleton.

Axial skeleton

Axial skeleton includes those skeletal parts which are present along the central axis of the body, like skull, vertebral column and rib cage.

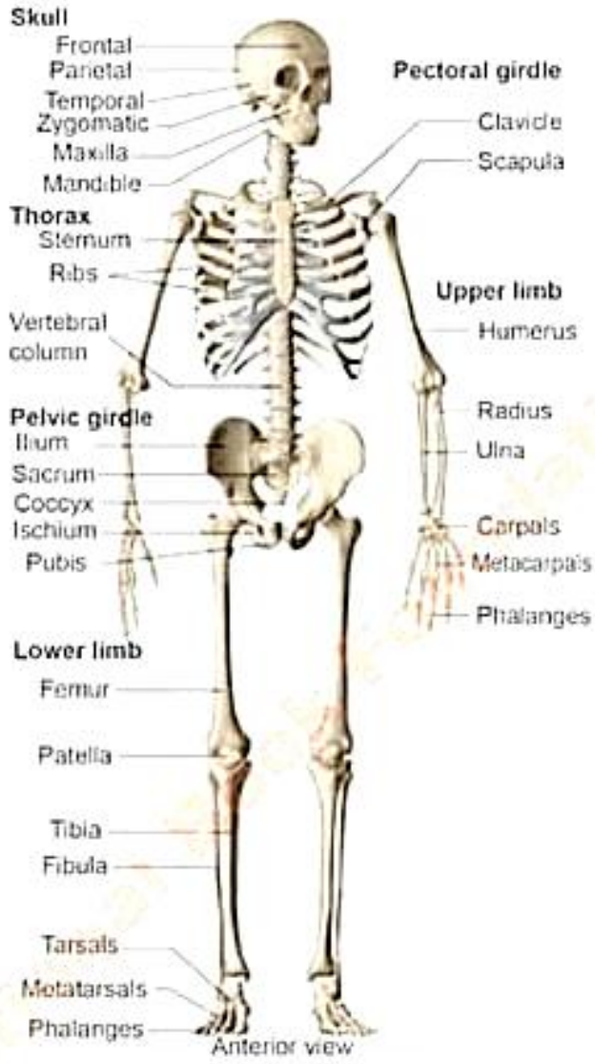


Fig. 16.4 Human skeleton

Head bones

Head contains 29 bones which are divided into four divisions i.e., cranial bones, facial bones, ear ossicles and hyoid bone. Cranial bones form cranium (brain box). Out of 8 cranial bones two are paired i.e., parietal bones (left and right) and temporal bone (left and right) while four are unpaired like frontal bone, occipital bone, ethmoid bone, sphenoid bone. Facial bones are fourteen in number and are attached to the cranium to form face. The six paired bones of face are: lacrimal, zygomatic, nasal bones, inferior nasal concha, maxilla and palatine. The unpaired bones of face are mandible and vomer. Three pairs of middle ear ossicles are malleus, incus and stapes. Hyoid bone is a small single bone which lies at the base of skull below the tongue. It does not articulate with any other bone of head.

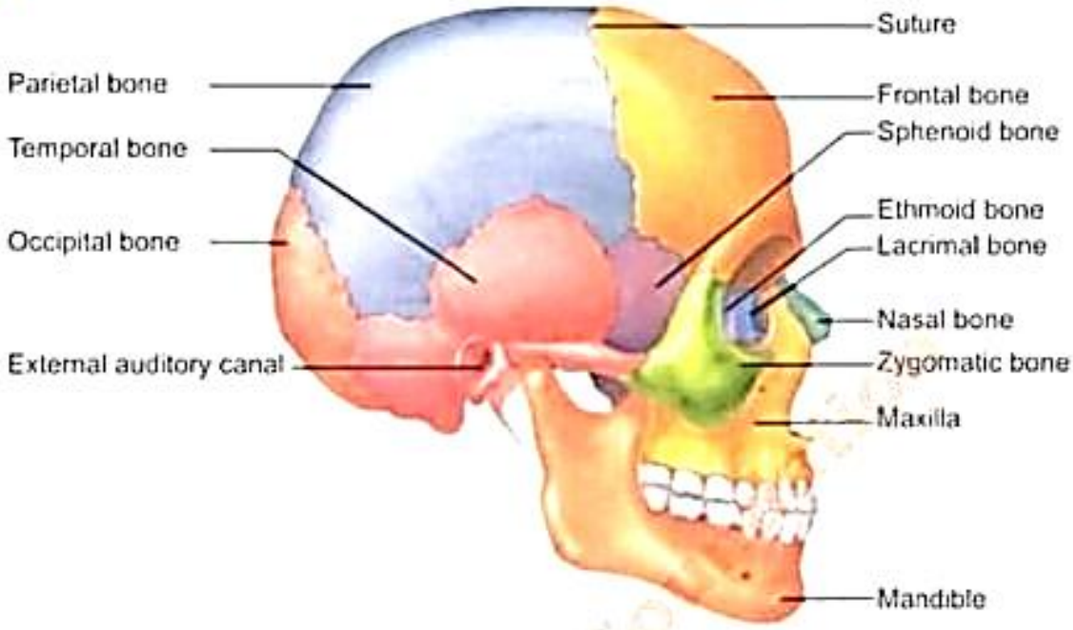
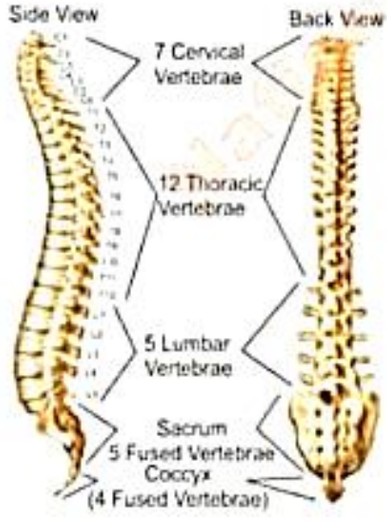


Fig. 16.5 Human Skull (side view)

Vertebral column

The **vertebral column** in human being consists of thirty-three vertebrae. The vertebrae may be divided into following five groups:

(a) Cervical vertebrae - 7, (b) Thoracic vertebrae - 12 (c) Lumbar vertebrae - 5 (d) Sacral vertebrae -5 (e) Coccygeal vertebrae - 4. Cervical vertebrae are the vertebrae of the neck. The **atlas** is the first cervical vertebra. **Axis** is the second cervical vertebra. **Thoracic vertebrae** are rib carrying vertebrae having large spinous processes and are found in chest region. **Lumbar vertebrae** are present in abdominal region. **Sacral vertebrae** are five fused vertebrae forming the **sacrum**. The sacrum articulates with the iliac bones of the hip bone to form the back of the pelvis. **Coccygeal vertebrae** or coccyx are four vertebrae fused in the adults. Sacral and coccygeal vertebrae are together called **pelvic vertebrae.**



Rib cage

The **rib cage** consists of twelve pairs of ribs. The ribs articulate posteriorly with the thoracic vertebrae. Ten ribs are connected anteriorly with sternum either directly or through the costal cartilage. The rib cage provides support for a

semi-vacuum chamber called **chest cavity**. The seven pairs of ribs that attach directly to the sternum are called **true ribs**. The 8th, 9th and 10th are called false ribs, as these three pairs of ribs are attached to the sternum by means of common costal cartilage. 11th and 12th pairs of ribs are known as **floating ribs**, because they do not attach to the sternum.

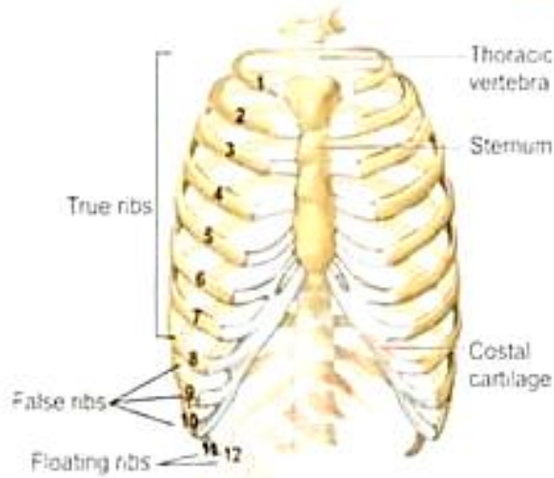


Fig. 16.7 Human rib cage

Appendicular skeleton

Appendicular skeleton includes those skeletal parts which are present in appendages (arms and legs). These are pectoral girdle, pelvic girdle, forelimbs and hind limbs.

Pectoral girdle and Upper limb

Pectoral girdle consists of a pair of **clavicles** and a pair of **scapula**. **Clavicles** are a pair of collar bones. One end of each curved bone articulates with the sternum. The other end articulates with the scapula. **Scapulas** are two shoulder blades.

Upper limb or **Forelimb** consists of humerus, ulna, radius, carpals, metacarpals and phalanges. **Humerus** is a long bone, the end of which has a spherical **head**, which fits into the **glenoid cavity**. **Radius** is a long, outer bone of the forearm (on the thumb side). **Ulna** is a long bone on the inner side of the forearm, and slightly bigger than radius. **Carpals** consist of two rows of eight short bones forming the wrist. The upper row articulates with the radius and forms the wrist joint. **Metacarpals** consist of five bones making up the palm of the hand. Each finger possesses three **phalanges** except thumb which comprises two phalanges, (see figure 16.4).

Pelvic girdle and Lower limb

The **pelvic girdle** is made up of three units the **ileum**, **ischium** and **pubis** which form **coxa**. The two halves of the pelvic girdle are joined at the **pubic symphysis**. A cavity called **acetabulum** is also present.

Lower limb or **Hind limb** consists of femur, patella, tibia, fibula, tarsal, metatarsal and phalanges. **Femur** or the thighbone is a long bone with head, which fits into the acetabulum. **Patella** or the kneecap is embedded in a long tendon which runs over the knee joint. Tibia or shin bone is the large bone in the leg. **Fibula** or outer bone is a thin bone joins the tibia just below the knee joint and just above the ankle. **Tarsal** is made of seven bones which are tightly attached to form the ankle. **Metatarsal** consists of five bones which articulate with the tarsal and phalanges to form the sole of the foot. **Phalanges** are small bones which make up the toes. Each toe of the foot possesses three phalanges except big toe, which comprises of two phalanges.

16.1.4 Joints

A joint or articulation is a place where two bones or bone and cartilage come together. The scientific study of the structure and function of joints is called arthrology. The joints are classified as fibrous joints (immoveable), cartilaginous joints (slightly moveable) and synovial joints (freely moveable).

Fibrous joints

When the adjacent bones are directly connected to each other by fibrous connective tissue consisting mainly of collagen, it is called fibrous joint. In this joint the bones do not have a joint cavity between them. The gap between the bones may be narrow or wide.

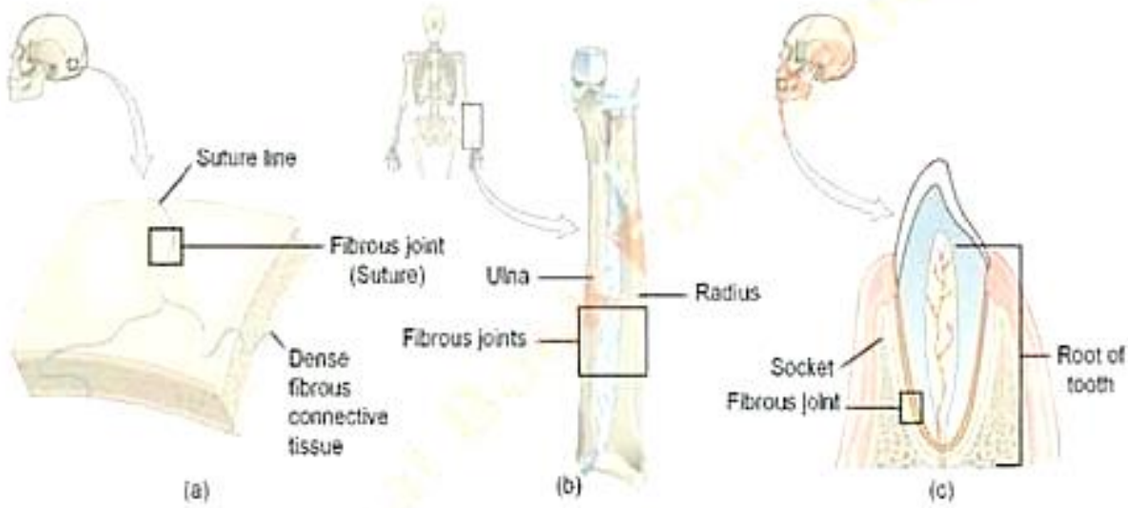


Fig. 16.8 Examples of fibrous joints

Examples: Fibrous joint is found between:

(a) Most bones of the skull called suture.

(b) The shaft regions of the long bones in the forearm and in the leg.

(c) The root of a tooth and the socket in the maxilla or mandible (jawbones).

Cartilaginous joints

At a cartilaginous joint, the adjacent bones are united by cartilage, a tough but flexible type of connective tissue. These types of joints lack a joint cavity and involve bones that are joined together by either hyaline cartilage or fibrocartilage. Cartilaginous joints allow little movement. The examples of cartilaginous joint are:

(a) Costal cartilages that attach ribs to the sternum.

(b) Pubic symphysis and intervertebral disc.

Fig. 16.9: Cartilaginous joint of pubic symphysis

Synovial joints

They are freely moveable joints. The ends of bones are covered with hyaline cartilage and held together by a surrounding, tube like capsule of dense fibrous tissue. The joint capsule is composed of an outer layer of ligaments and an inner lining of synovial membrane, which secretes **synovial fluid**.

Examples: Hinge joint, Pivot joint, Ball-and-socket joint, Gliding joint.

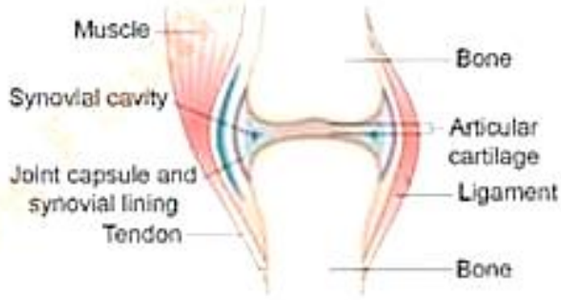


Fig. 16.10 Synovial joint

Science, Technology and Society Connections

Name the techniques for joint replacement.

Many joints of the body can be replaced by artificial joints. Joint replacement is called arthroplasty. Artificial joints are usually composed of metal, such as stainless steel, titanium alloys, in combination of modern plastics, such as high-density polythene, silastic or elastomer. The bone of the articular area is removed on one side. This procedure called partial joint replacement or hemi-replacement technique. When both sides of the articular area are removed it is called total joint replacement technique. The artificial articular areas are glued to the bone with a synthetic adhesive, such as methyl methacrylate.

Science, Technology and Society Connections

- **Relate the bipedal posture of man with his skeleton and musculature.**

Curvatures of vertebral column help to balance the body for bipedal stance. The intervertebral discs lend flexibility to the vertebral column and absorb vertical shock. The structure of the pelvis, in its attachment to the vertebral column, permits upright posture and locomotion on two appendages (bipedal locomotion). Certain muscles are active posture muscles, whose primary function is to work in opposition to gravity. For example, the strong, complex muscles of the vertebral column are adapted to provide support and movement in resistance to the effect of gravity. Thus, the skeleton and muscular systems maintain the bipedal posture of man.

16.2 DISORDERS OF SKELETON

Skeletal deformation may be hereditary e.g., arthritis may be hormonal e.g., osteoporosis or may be due to nutritional deficiency e.g., osteomalacia and rickets. Here we will describe slipped disc spondolysis, sciatica and arthritis.

16.2.1 Common Disorders of Skeleton

Slipped disc

Each intervertebral disc is a cushion like pad which consists of nucleus pulposus and annulus fibrosus. Nucleus pulposus is an inner semifluid which acts as a rubber ball to give a disc its elasticity and compressibility. Annulus fibrosus is the strong outer ring of fibrocartilage, which holds together successive vertebrae. The discs act as shock absorber. Severe or sudden trauma to spines may result in herniation of one or more discs.

The herniated disc or slipped disc usually involves rupture of annulus fibrosus followed by protrusion of the spongy nucleus pulposus. If protrusion presses on spinal cord or on spinal nerves, generate severe pain or even destruction of these nervous structures. 'Slipped disc' is misleading as it is not the whole disc that slides out of the position.

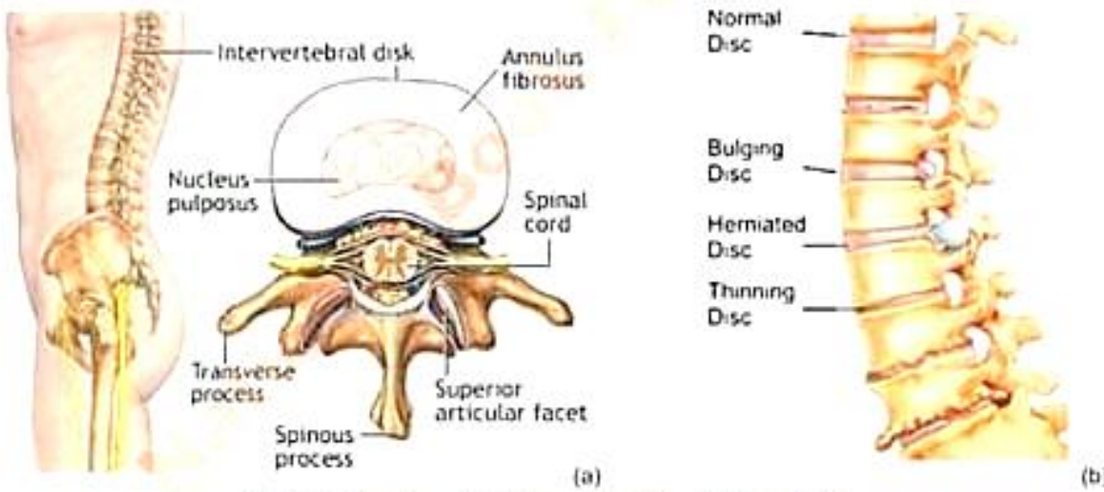


Fig. 16.11 Structure of (a) intervertebral disc (b) slipped disk

Spondylosis

It is the immobility and fusion of vertebral joint. Cervical spondylosis results from chronic cervical degeneration, with herniation of disc and aging.

Sciatica

Sciatica refers to pain, weakness, numbness, or tingling in the leg. It is caused by injury to or pressure on the sciatic nerve. Common causes of sciatica include: Slipped disk, pelvic injury or fracture and tumors.

Arthritis

It is the inflammation of joints. The typical **symptoms** of arthritis include pain after walking which may later occur even at rest, creaking sounds in joint, difficulty in getting up from a chair and pain on walking up and down stairs. There are different types of arthritis. **Osteoarthritis** is a progressive disease in which the articular cartilages gradually soften and disintegrate. It affects knee, hip and intervertebral joints.

Rheumatoid arthritis is the result of an autoimmune disorder in which synovial membrane becomes inflamed due to faulty immune system. **Gouty arthritis** results from a metabolic disorder in which an abnormal amount of uric acid is retained in the blood and sodium urate crystals are deposited in the joints. The most common joint affected is the joint of the big toe.

Science, Technology and Society Connections

Justify the use of calcium in teenage and twenties can be a preventive action against osteoporosis.

Osteoporosis, is a common disease characterised by reduced bone mass and an increased risk of fracture. In normal individuals, bone mass increases during skeletal growth to reach a peak between the ages of 20 and 25 but falls thereafter in both sexes. Osteoporosis, occurs because in the bone resorption exceeds bone deposition. The increased calcium is used to increase bone mass. The greater the bones mass before the onset of osteoporosis, the greater the tolerance for bone loss later in life. For this reason, it is important for adults, especially women in their twenties and thirties, to ingest adequate amounts of calcium.

16.2.2 Bone Fractures

A fracture is the medical term for a broken bone. They occur when the physical force exerted on the bone is stronger than the bone itself. So bones break when they cannot withstand a force or trauma applied to them.

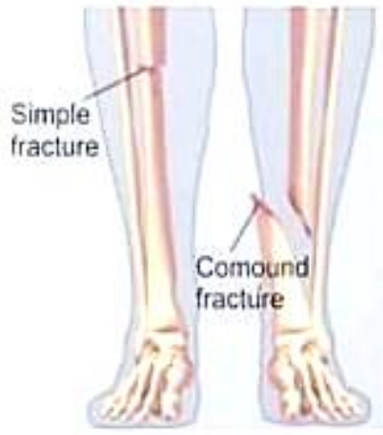


Fig. 16.12 Bone fractures

Common types of fractures

Simple fracture or **closed fractures** are those in which the skin is intact. If the bone ends penetrate the skin and form a wound are called **compound fracture** or **open fracture.** When a fracture damages the adjacent organs it is called **complicated fracture.**

16.2.3 Bone Repair

Bone is a living tissue that undergoes repair following fracture. The repair process of a simple fracture takes place in four major steps.

Haematoma or clot formation

When a bone breaks, blood vessels in the bone, and perhaps in surrounding tissues, are torn and haemorrhage. As a result, a haematoma, a mass of clotted blood, forms at the fracture site. Soon, bone cells deprived of nutrition die, and the tissue at the site becomes swollen, painful, and inflamed.

Fibrocartilaginous callus formation

Within a few days, several events lead to the formation of fibrocartilaginous or soft callus. Capillaries grow into the haematoma and phagocytic cells invade the area and begin cleaning up the debris. A fracture ruptures the periosteum and stimulates the production and release of the numerous osteoblasts. These osteoblasts in conjunction with cartilage forming cells secrete a porous mass of bone and cartilage called **callus** (or cartilaginous callus) surrounding the break. The callus replaces the original blood clot and holds the ends of the bones together. This process takes 3-4 weeks.

Bony callus formation or callus ossification

Within a week, after the formation of soft callus, it is gradually converted into a hard **bony callus** of spongy bone. Bony callus formation continues until a firm union is formed about two months later. Osteoclasts breakdown the cartilage while osteoblasts replace it with bone.

Fig. 16.13: Bone repair

Bone remodelling

It takes place when a compact bone is formed across the fracture line to connect both sides. Usually, more bone is produced at the site of a healing fracture than needed to replace the damaged tissue. However, osteoclasts eventually remove the excess and the final result of the repair is bone shaped very much like the original. The final structure of the remodeled area resembles that of the original unbroken bony region because it responds to the same set of mechanical stressors.

Science, Technology and Society Connections

Relate improper posture to bone/joint problems

Improper posture causes increased stress on joints and their supporting structures resulting in injury, pain and early degeneration of bones and joints.

16.2.4 Injuries to Joints

Torsion or sudden impact to the side of a joint can be devastating. We will discuss here dislocation and sprain.

Dislocation of joints

A dislocated joint is a joint that slips out of place. It occurs when the ends of bones are forced from their normal positions. A severe dislocation can cause tearing of the muscles, ligaments and tendons that support the joint. Symptoms include; swelling, intense pain, and immobility of the affected joint. The most common causes are a blow, fall, or other trauma to the joint. In some cases, dislocations are caused by a disease or a defective ligament. **Rheumatoid arthritis** can also cause joint dislocation. A dislocated joint usually can only be successfully 'reduced' into its normal position by a trained medical professional. Surgery may be needed to repair or tighten stretched ligaments.

Sprain

A sprain is an injury to a ligament. Commonly injured ligaments are in the ankle, knee and wrist. The ligaments can be injured by being stretched too far from their normal position. The sprain should be rested. Sprains can usually be treated conservatively with treatments such as icing and physical therapy. Dressings, bandages, or ace-wraps should be used to immobilize the sprain and provide support.

16.2.5 First aid Treatment for Disorders of Skeleton

Prompt and proper first aid increases the chances of a complete recovery. Usually, the severity of the bone fracture and dislocation of joints depend on its cause and the affected part. If you suspect someone has dislocated a joint or fractured bone, you can help by: (a) immobilizing the fractured bone or dislocated joint but do not attempt to manipulate, pull or re-align the injured joint or bone. Leave this task to a professional (b) If possible; apply ice pack or cold pack over the affected part to reduce swelling. (c) Assist the victim to position of comfort. (d) Provide support to the affected area such as using sling or splints. Listen to what the victim tells you. (e) Dislocations involving the hip, ankle and leg joints and compound fractures require ambulance to transport the victim.

16.3 MUSCLES

The specialized tissues that can undergo contraction and relaxation and provide movements of body parts or whole body are called **muscles**. The study of muscles is called **myology**. They also function to hold body parts in postural positions, movement of body fluids and heat production.

16.3.1 Types of Muscles

There are three types of muscle tissues: smooth, cardiac and skeletal.

Smooth muscles

These are distributed widely throughout the body and are more variable in function than other muscle types. The smooth muscle cells are spindle shaped, with a single nucleus located in the middle of the cell. **Myofilaments** are not organised into sarcomeres. Consequently, smooth muscle does not have a striated appearance. Smooth muscle cells contain

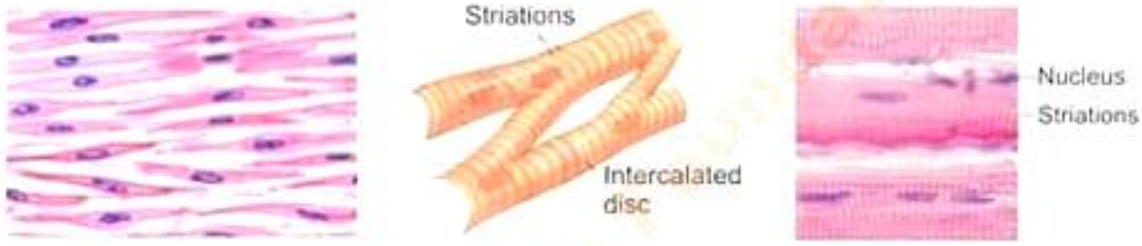
noncontractile **intermediate filaments**. Smooth muscles are involuntary in function. They are found in digestive, reproductive, urinary tract, blood vessels etc.

Cardiac muscles

These are found only in heart. They branch extensively. Cardiac muscles are striated like skeletal muscle, but each cell usually contains one nucleus located near the centre. Adjacent cells join together to form branching fibres by specialised cell-to-cell attachments called **intercalated discs,** which have gap like junctions that allow action potentials to pass from cell to cell.

Skeletal muscles

These muscles are attached to the bone and are responsible for movements of body parts and whole body movements (locomotion).



(a) Smooth muscles (c) Cardiac muscles (b) Skeletal muscles

Fig. 16.14 Types of muscles

Table 16.2 Comparison of three types of muscle tissues

Property	Smooth Muscles	Cardiac Muscles	Skeletal Muscles
Muscle appearance	Unstriped	Irregular striped	Regular striped
Cell shape	Spindle	Branched	Spindle or cylindrical
Number of nuclei	One per cell	One per cell	Many per cell
Speed of contraction	Slow	Intermediate	Slow to rapid
Contraction caused by	Nervous system	Spontaneous	Nervous system
Function	Controls movement of substances through hollow organs	Pumps blood	Move skeleton
Voluntary control	Usually no control	Usually no control	Have control

16.3.2 Structure of Skeletal Muscles

Skeletal muscles or striated muscles show alternate light and dark regions under microscope. Skeletal muscles are composed of muscle fibres or muscle cells. Bundles of muscle fibres are enclosed by collagen fibres and connective tissue. At the ends of the muscle the collagen and connective tissue

Muscles (Extra reading material)
Externally muscle is covered in a connective tissue wrapping called **epimysium**. Each skeletal muscle consists of hundreds to thousands of muscles fibres (muscle cells). Each muscle is divided into discrete bundles of muscle cells called **fascicles**. The fascicle is surrounded by **perimysium**. Each muscle fibre within the fascicle is covered by a layer of connective tissue called the **endomysium**

forms **tendons** which attach the muscle to skeletal elements. Each skeletal muscle fibre is a single cylindrical cell, enclosed by a plasma membrane like structure called **sarcolemma** and has several nuclei. The sarcolemma of muscle fibre cell penetrates deep into the cell to form a hollow elongated tube, the **transverse tubule** (**T-tubule**). The cytoplasm of the muscle fiber is called **sarcoplasm.** It contains **sarcoplasmic reticulum**. Within the muscle fibres are numerous thin **myofibrils** which possess characteristic cross striations.

The myofibrils are 1-2 μm in diameter that run in parallel fashion and extend the entire length of the cell. Each myofibril is composed of two types of myofilaments **thin myofilaments** and **thick myofilaments.**

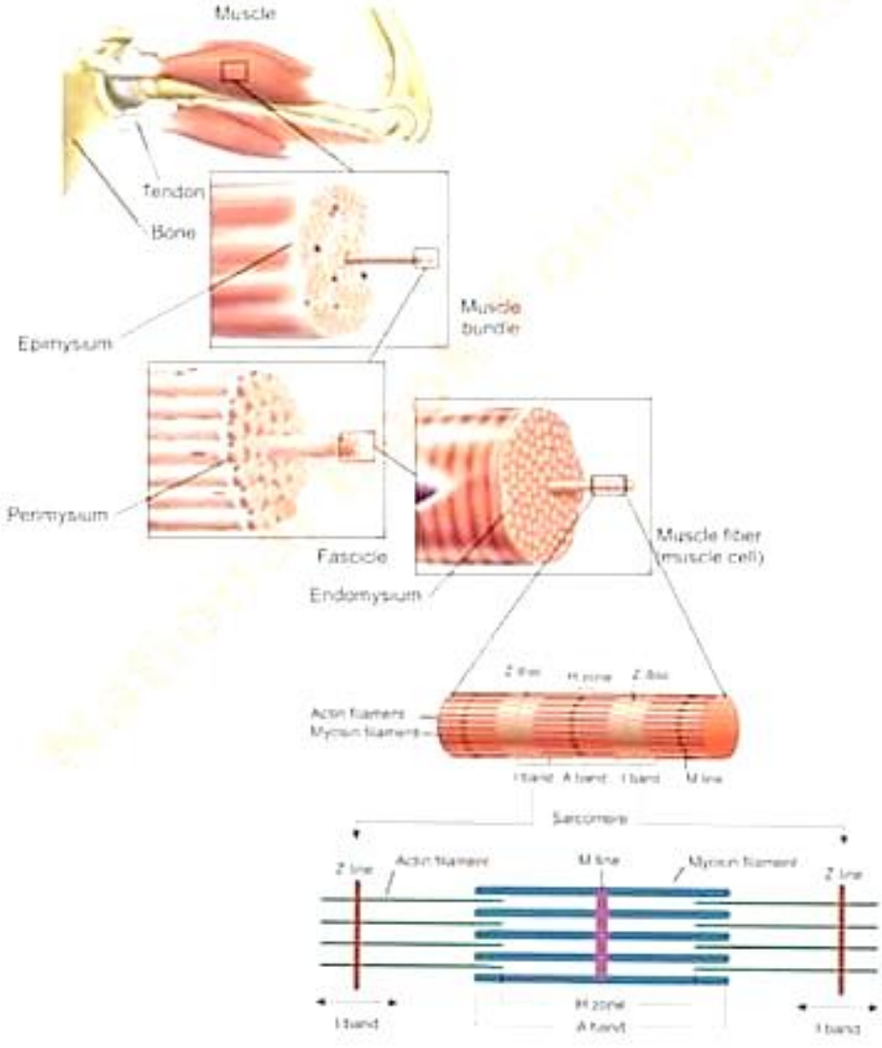


Fig. 16.15 Structure of skeletal muscle

Ultra - structure of skeletal muscles

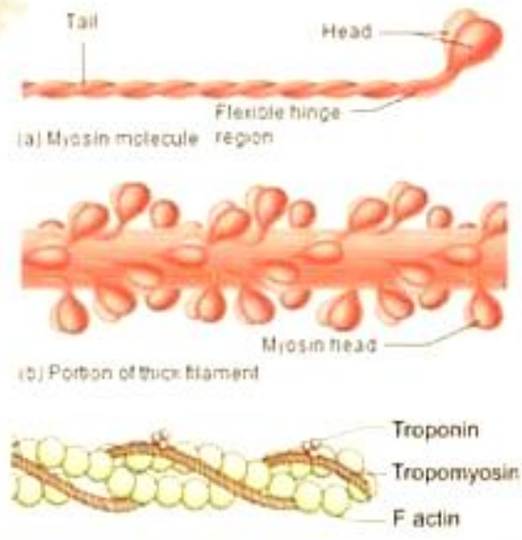
Under a light microscope only the striated nature of the myofibrils can be observed. This is seen as a regular alternation of light and dark bands called the **I bands** and **A bands** respectively, transversed by thin, dark lines. Electron microscope studies clearly indicate that the bands are due to regular arrangement of thin and thick myofilaments. Transversing the middle of each I band is a dark line called the **Z line**. The section of myofibril between two Z lines is called a **sarcomere**, which is a contractile unit. From the Z line thin myofilaments extend in both directions, whilst in the centre of the sarcomere are found thick myofilaments.

In certain regions of the sarcomere, thin and thick myofilaments overlap. Transverse sections in these regions indicate that six thin myofilaments surround each thick myofilament. This arrangement of thin and thick myofilaments results in a number of other bands being recognizable in the sarcomere. The entire length of thick myofilaments constitute the **A band** because they are **anisotropic** that can polarize visible light. Thin myofilaments alone constitute **I band**, which is **isotropic** or nonpolarizing. The centre of the **A band** is lighter than the outer regions in a relaxed sarcomere as there are no overlap between the thin and thick myofilament in this region. It is called the **H zone** (H stands for 'hele' means bright). The H zone itself may be bisected by a dark line, the **M line**. The M line joins adjacent myosin filaments together at a point halfway along their length.

Extra Reading Material

Each myosin molecule consists of six polypeptides which are arranged in such a way that each myosin molecule possesses a tail and two globular heads. Each thick filament contains about 300 myosin molecules bundled together with their tails forming the central part of the thick filament and their heads facing outward and in opposite directions at each end.

The kidney-shaped polypeptide subunits of actin, called globular actin or **G actin**, bear the active sites to which the myosin heads attach during contraction. G actin monomers are polymerized into long actin filaments called **fibrous**, or **F actin**. The backbone of each thin filament appears to be formed by two intertwined actin filaments that look like a twisted double strand of pearls.



Thick myofilaments are 16 nm in diameter and are composed of only **myosin** protein.

The **thin filaments** are 7–8 nm in diameter and are composed of three proteins. Two intertwisted beaded chain of **actin** which form the core of filament. Two strands of **tropomyosin** spiral about the actin core and help stiffen it. In a relaxed muscle fibre, they block myosin binding sites on actin so that the myosin heads cannot bind to the thin filaments.

Troponin is a three-polypeptide complex found at regular intervals on thin myofilaments. One of these polypeptides (TnI) is an inhibitory subunit that binds to actin; another (TnT) binds to tropomyosin and helps position it on actin. The third (TnC) binds calcium ions. Both troponin and tropomyosin help control the myosin-actin interactions involved in contraction.

16.3.3 Muscle Contraction – Sliding Filament Model

The sliding filament theory of contraction states that during contraction the thin myofilaments slide past the thick ones so that they overlap to a greater degree. In a relaxed muscle fibre, the thick and thin myofilaments overlap only at the ends of the A band. But when muscle fibres are stimulated by the nervous system, the myosin heads are attached on to myosin binding sites on actin in the thin myofilaments, and the sliding begins. These links are called **cross bridges** which are formed and broken several times during a contraction, acting like tiny ratchets to generate tension and propel the thin myofilaments toward the centre of the sarcomere.

As this event occurs simultaneously in sarcomeres throughout the cell, the muscle cell shortens. The I bands shorten, the distance between successive Z discs is reduced, the H zone disappears, and the contiguous A bands move closer together but do not change in length.

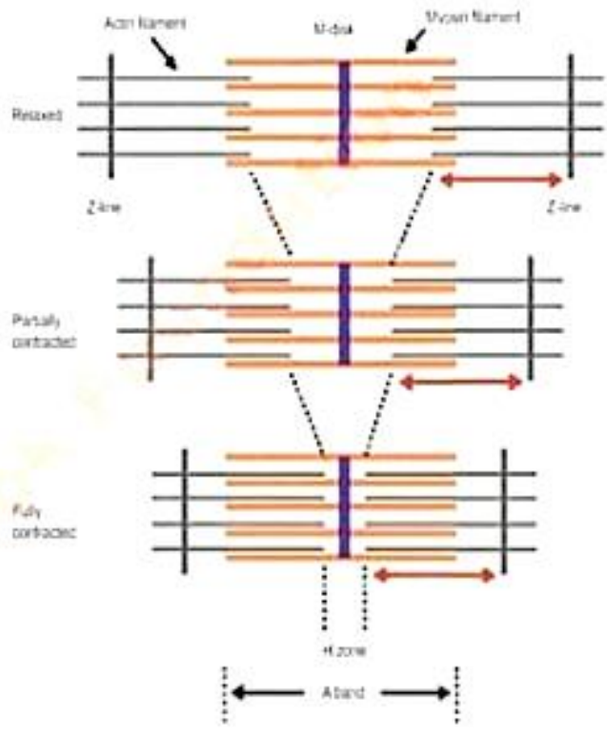


Fig. 16.16 Sliding filament model of muscle contraction

Control of cross bridges

Muscle contraction is initiated by nerve impulse arriving at the neuromuscular junction. The nerve impulse is carried through the sarcolemma to the T tubule then to the sarcoplasmic reticulum (SR). The calcium gates of the SR open releasing calcium into the cytosol. When muscle is at rest the tropomyosin is disposed in such a way that it covers the sites on the actin chain where the heads of myosin become attach. When calcium ions bind with the troponin molecules they cause them to move slightly. This has the effect of displacing the tropomyosin and exposing the binding sites for the myosin head. Once the myosin head has become attached to the actin filament, ATP is hydrolysed and the crossed bridges are broken down. The formation and breakdown of cross bridges occur again and again during the sliding of the filament.

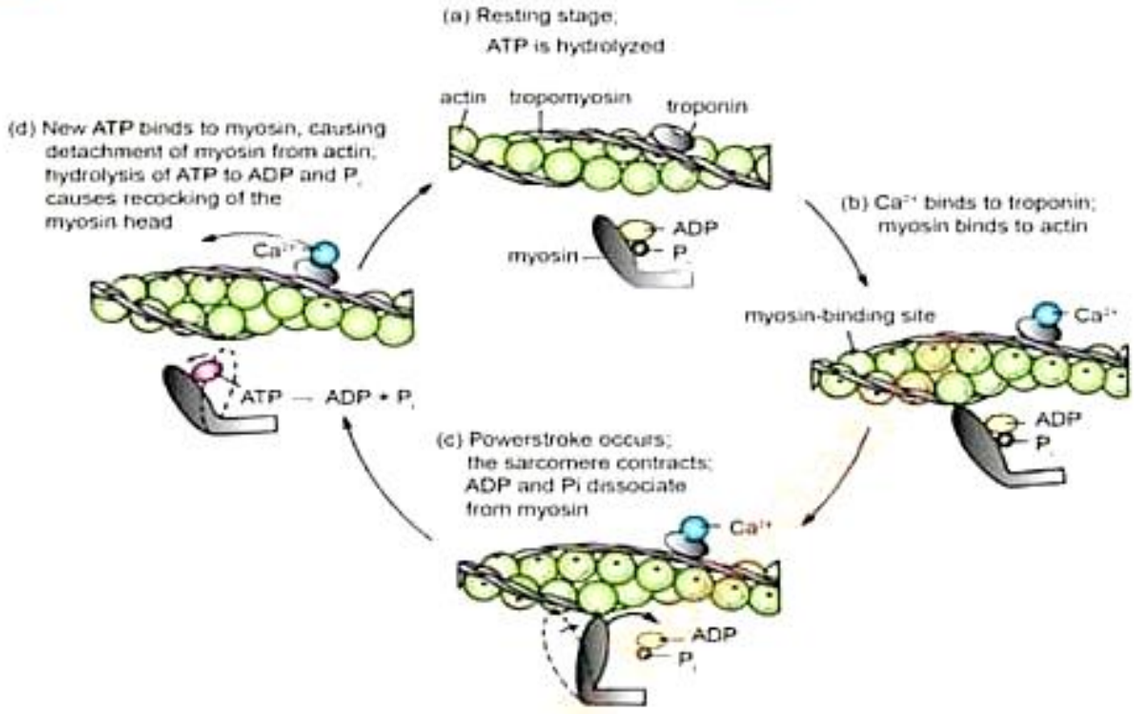


Fig. 16.17 Sliding filament model of muscle contraction

16.3.4 Antagonistic Arrangement of Skeletal Muscles

Bones are attached to the bones through connective tissue called **ligament.** When a muscle contracts one end normally remains stationary and the other end is drawn towards it. The end which remains stationary is called **origin** and that which moves is called **insertion**. Both are the points of attachment to bones. Every muscle has its own origin and insertion. **Belly** is the thick part between origin and insertion which contracts. Normally the bones of insertion is pulled upon when muscle contracts and drawn towards origin, one bone moving on the other at the joints. **Flexor muscle,** when contracts, it bends the bone at joint. **Extensor muscle,** when contracts it straightens the bone at joints. For the movement of the bone in two directions muscles work in pairs. When flexors contract, the extensors relax and vice versa. Such arrangement of muscles is called **antagonistic arrangement.**

Movement in knee joint

Knee or **tibio-femoral joint** is located between the femur and tibia. It is a complex hinge joint that permits limited rolling and gliding movements in addition to flexion and extensions. The flexion is carried out by the flexor muscles. These are **hamstring muscles** present at the back of the upper part of the leg (thigh). The major hamstring muscle is **biceps femoris**. It has two origins, one from pelvic girdle and other from the top of the femur. At its insertion the tendon divides into two portions to attach at the upper part of the tibia and fibula.

The extension is carried out by the extensor muscles which are present in the front of the thigh. The main extensor muscles are **quadriceps femoris**. They originate at the ilium and femur, come together in a tendon surrounding the patella (kneecap), and insert at the tibia. These extend the leg at the knee joint and are important for standing, walking, and almost all activities involving the legs.

Fig. 16.18 Movement at knee joint

16.3.5 Muscle Disorders

There are many problems related to muscle which are generally called muscle disorder. Some common muscle disorders e.g., muscle fatigue, cramp and tetany are discussed here.

Muscle fatigue

When the muscles lose the ability to contract, the physiological state of muscles is called **muscle fatigue**. The other factors which contribute to muscle fatigue are accumulation of lactic acid and ionic imbalance. The cause of extreme fatigue is lactic acid which causes muscle pH to drop and the muscle to ache.

Cramp

It is also known as **tetanic contraction** of entire muscle. It lasts for just few seconds to several hours, causing the muscle to become taut (tightly drawn) and painful. It is most common in thigh and hip muscles. It usually occurs at night or after exercise. It reflects low blood sugar level, electrolyte depletion, dehydration, irritability of spinal cord and neurons.

Tetany

In tetany the body shakes from continuous muscle contraction and convulsion occur due to calcium imbalance. It results in the excitability of neurons and results in loss of sensation. If untreated the symptoms progress to spasm of larynx, paralysis and ultimately death occurs.

Table 16.3 Difference between tetany and tetanus

Tetany	Tetanus
Caused by low calcium level in blood	Caused by infection of *Clostridium tetani*
Results in the excitability of neurons and loss of sensation	Due to severe convulsion the patient dies due to lack of oxygen.

Science, Technology and Society Connections

Justify why do the muscles pull but do not push.

Bones act as the levers, while joints perform as living fulcrums. Muscle, attached to bones by tendons and other connective tissue, exerts force by converting chemical energy into tension and contraction. When a muscle contracts, it shortens, in many cases pulling a bone like a lever across its hinge. Muscles move and by their motions we move. We are capable of performing a wide variety of actions, but despite this, muscle itself moves only by becoming shorter. They shorten and then they rest - in other words, a muscle can pull but it cannot push.

Science, Technology and Society Connections

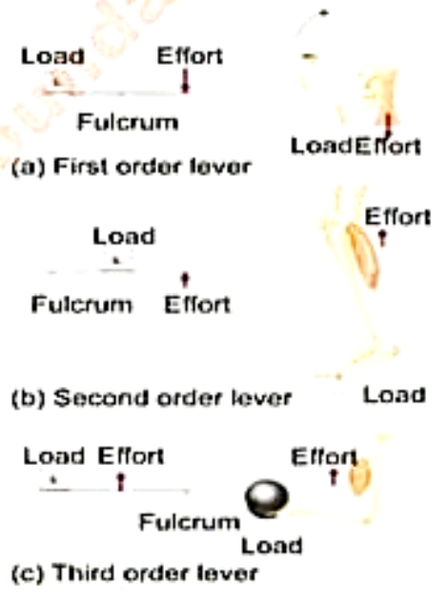
Reason out the rigor mortis.

When death occurs, ATP is no longer made. It is a short-lived chemical and so it runs out fairly quickly. This causes the muscles to lock into position as cross-bridges that formed between actin and myosin filaments before death can no longer be broken. The skeletal muscles undergo a partial contraction that causes the joints to become fixed. This condition, rigor mortis (rigidity of death), happens in all body muscles. It appears about four hours after death and lasts about 34 hours. After this time, muscles proteins are destroyed by enzymes within the cells and so rigor mortis disappears.

Skill: Analyzing and Interpreting

- **Justify how the main functions of the skeleton are to act as a system of rods and levers, which are moved by the muscles.**

A lever is any rigid structure that runs about a fulcrum when force is applied. Levers are generally associated with machines but can also apply to human body. In the body, synovial joints usually serve as the fulcra (sing: fulcrum) (F), the muscles can provide force or effort (E), and the bones act as the rigid lever arms that move the resisting object. There are three kinds of levers, determined by the arrangement of their pairs. In first order lever, the fulcrum is positioned between the effort and the resistance. In the human body the head at the atlanto-occipital joint, straightening of the elbow is an example. In the second order lever, the resistance is positioned between the fulcrum and the effort. Contraction of the calf muscles (E) to elevate the body on the toes, with the ball of the foot acting as the fulcrum is an example in the human body. In the third order lever, the effort lies between the fulcrum and the resistance. The flexion of the elbow is an example.



Activity

1. Identification of the bones of the pelvic girdles, pectoral girdle, arms and legs by using the model of human skeleton
2. Comparison of the structure of skeletal, smooth and cardiac muscles with the help of prepared slides

Exercise

M.C.Qs

1. Select the correct answer

(i) The atlas and axis vertebrae are located in:

(A) lumbar region (B) cervical region

(C) thoracic region (D) pelvic region

(ii) Skeletal muscles contain dark band, which are anisotropoic, are called

(A) A band (B) I band (C) Z band (D) M line

(iii) The acetabulum provides the articular surface for the

(A) humerus (B) femur (C) pelvis (D) fibula

(iv) Scapula is connected with sternum by

(A) ribs (B) carpals (C) clavicle (D) atlas

(v) Which statement correctly describes the smooth muscles?

(A) Unstriated involuntary with spindle shape cells

(B) Unstriated involuntary with multinucleate cells

(C) Unstriated voluntary with uninucleate cells

(D) Striated involuntary with spindle shape cell

(vi) Thin myofilaments consist of

(A) actin, myosin, troponin (B) actin, tropomyosin, troponin

(C) actin, tropomyosin, fibrin (D) actin, myoglobin, troponin

(vii) Which of the following changes occur when skeletal muscle contracts?

(A) The A- bands shorten (B) The I- bands shorten

(C) The Z- lines move further apart (D) The H- zone becomes more visible

(viii) A human internal organs are protected mainly by the

(A) hydrostatic skeleton (B) axial skeleton

(C) exoskeleton (D) appendicular skeleton

(ix) Arm and leg muscles are arranged in antagonistic pairs. How does this affect their functioning

(A) it provides a backup if one of the muscles is injured

(B) one muscle of the pair pushes while the other pulls

(C) it allows the muscles to produce opposing movements

(D) it doubles the strength of contraction

(x) Which of the following bones in the human arm would correspond to the femur in the leg?
(A) radius (B) ulna (C) tibia (D) humerus

(xi) The deep infolding of the muscle fibre membrane is called
(A) sarcoplasmic reticula (B) Z lines
(C) T-tubules (D) sarcomeres

(xii) Bone dissolving cells are called
(A) chondrocytes (B) osteoblasts
(C) osteoclasts (D) osteocytes

(xiii) Which of the following cartilage is found at the end of long bones?
(A) calcified (B) fibrous
(C) elastic (D) hyaline

(xiv) At times ligaments are overstretched or torn. It is called
(A) sprain (B) dislocation (C) fracture (D) tension

(xv) Which ion is essential for muscle contraction?
(A) Na (B) K (C) Ca (D) Cl

Short Questions

2. How do compact bone and spongy bone differ in structure?
3. Name three types of cells associated with bone and write their functions.
4. Compare structure of bone with that of cartilage.
5. Name the bones of axial and appendicular skeleton.
6. Name the bones of cranium.
7. Describe the five groups of vertebrae.
8. What is the structure of the human rib cage.
9. Name the bones that form the (a) pectoral girdle (b) pelvic girdle.
10. Name the bones of upper and lower limbs.
11. What are the main types of joints found in bones?
12. What is fibrous joint?
13. What are the four steps required for bone fracture repair?
14. What skeletal structures are affected from the osteoarthritis?
15. List the major parts of skeletal muscle fibre and write the function of each part.
16. What is Z line and M line and what are their functions?
17. How the arrangement of actin filaments and myosin filaments produce I band, A band and H zone?
18. Describe the antagonistic arrangement of skeletal muscles.
19. Why are ligaments elastic and why does the tendon need to be inelastic?

20. Draw a diagram of sarcomere and label its parts.
21. Write the difference between:
    (a) epiphysium and diaphysium
    (b) periosteum and endosteum
    (c) compact and spongy bone
    (d) axial skeleton and appendicuar skeleton
    (e) true ribs and false ribs
    (f) false ribs and floating ribs
    (g) atlas and axis
    (h) rheumatid arthritis and gouty arthritis
    (i) simple and compound bone fracture
    (j) tropomysin and troponin
    (k) tendons and ligaments
    (l) callus and bony callus
    (m) tetany and tetanus

Extensive Questions

22. Explain the structure of bone with diagram.
23. Explain the structure of cartilage with diagram.
24. Describe the bones of appendicular and axial skeleton of man.
25. Describe the bones of cranium.
26. What are the following common types of disorders of human skeleton:
    (a) Slipped disc (b) Spondylosis (c) Sciatica (d) Arthritis
27. Give a detail account of bone repair.
28. Give an account of the following related to injuries to bones:
    (a) Dislocation of joints (b) Sprain
    (c) First aid treatment for disorders of skeleton
29. Describe three types of muscle tissues in man.
30. Explain the ultra structure of skeletal muscle.
31. Explain the sliding filament model of muscle contraction.
32. Explain the action of antagonistic muscles in the movement of knee joint in man.
33. Give explanation of the following statement:
    (a) Pregnant women should be encouraged to drink milk
    (b) The sutures of the skull are fixed joint.
    (c) The human femur is stronger than humerus.
37. Describe the following muscle disorders:
    (a) Muscle fatigue (b) Cramp (c) Tetany

15

HOMEOSTASIS

After completing this lesson, you will be able to

- Differentiate between osmoconformers and osmoregulators.
- Define osmoregulation.
- Explain the problems faced by osmoregulators.
- Explain the different methods of osmoregulation found in freshwater, marine water and terrestrial habitats.
- List various nitrogenous compounds excreted during the process of excretion.
- Explain the nature of excretory products in relation to habitat.
- Explain different organs of urinary system. Describe the structure of kidney and relate it with its function.
- Explain the detailed structure of nephron.
- Explain the processes of glomerular filtration, selective re-absorption and tubular secretion as the events in kidney functioning.
- Explain that concentration of urine is regulated by counter-current and hormonal mechanisms.
- Justify the functioning of kidneys as both excretion and osmoregulation.
- Compare the function of two major capillary beds in kidneys i.e. glomerular capillaries and peritubular capillaries.
- List urinary tract infections and the bacteria responsible.
- Explain the causes and treatments of kidney stones.
- Outline the causes of kidney failure.
- Explain in detail the mechanism dialysis.
- Define thermoregulation and explain its needs.
- Classify animals on the basis of the source of body's heat i.e. ectotherms and endotherms.
- Classify the animals on the bases of the ability to thermoregulate i.e. poikilotherms and homeotherms.
- Describe the regulatory strategies in man for thermoregulation.

Animals have two environments in their lives, an external environment in which the organism is situated and an internal environment in which the tissues live. The external environment consists of varying conditions of atmosphere, marine or freshwater. The internal environment is formed by the interstitial fluid or tissue fluid that surrounds and bathes all the

tissues and circulating body fluids like lymph or plasma, the liquid part of the blood. Homeostasis is the tendency of an organism or cell to regulate its internal conditions, such as the chemical composition of its body fluids, so as to maintain health and functioning, regardless of outside conditions.

15.1 OSMOREGULATION

The maintenance of constant osmotic conditions (water and solute concentration) in the body is called **osmoregulation.** Animals may be either osmoregulators or osmoconformers with respect to their external environment.

15.1.1 Osmoregulators and Osmoconformers

Osmoregulators

Those animals that can maintain internal osmotic concentrations different from the surrounding medium are called **osmoregulators**. Such animals are hypotonic or hypertonic to their environment. Almost all of the freshwater animals and most of the marine vertebrates are osmoregulators.

Osmoconformers

Those animals that change the osmotic concentrations of the body fluids according to that of surrounding medium are called **osmoconformers**. These are isotonic to their external environment. These include all marine invertebrates, some freshwater invertebrates and some marine vertebrates like *Myxine* (hag fishes) and elasmobranches (sharks and rays).

The unusual higher osmotic concentration than other vertebrates of marine habitat is maintained by high levels of urea and trimethylamine oxide (TMAO) in the blood. These organic substances are called **osmolytes** because they increase the osmotic (solutes) concentration.

Science Titbits

In most vertebrates, the level of urea to this high concentration would damage the proteins because it is chaotropic (denaturing) agent that disrupts non-covalent and ionic bonds between amino acids residues, but the presence of TMAO helps to stabilize these protein molecules against the adverse effects of urea.

15.1.2 Problems Faced by Osmoregulators

Since, freshwater animals live in hypotonic environment, therefore, water constantly enters the body and they also face deficiency of salts, so they have to lose excess water and maintain higher salt concentration than their environment.

On the other hand, most of the **marine teleosts** (bony fishes) are hypotonic to sea water. So these fishes have tendency to lose water to the environment, especially across the gill epithelium. They also have problem of excess of salts in the body due to drinking of sea water.

Terrestrial animals are also hypotonic to the outer environment. Evaporation of water that leads to the dehydration is the major problem faced by these animals.

15.1.3 Osmoregulatory Adaptations in Animals

Freshwater animals

Almost all of the freshwater animals are osmoregulators. These animals are generally hypertonic to their outer environment.

These animals deal with these problems by producing large volume of diluted urine. Their kidney reabsorbs the salts that are required. Salts are also obtained from the food they eat. These animals also actively transport salts from the external dilute medium with the help of special salt cells called **ionocytes**. Ionocytes are found in the amphibian skin and gills of fishes.

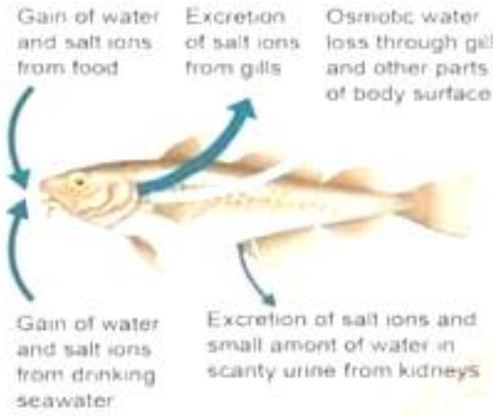


Fig. 15.1 Osmoregulation in marine animals

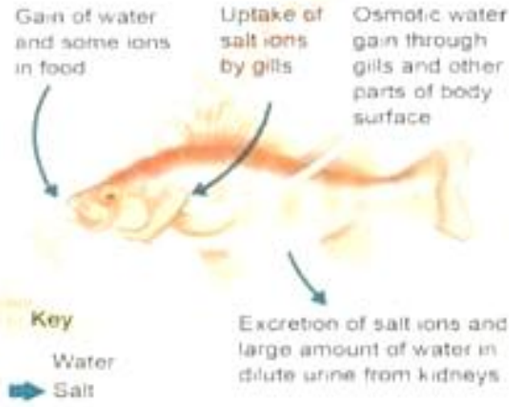


Fig. 15.2 Osmoregulation in freshwater animals

Marine animals

Teleosts (bony fishes) are osmoregulators in marine environment which are hypotonic to their environment. So these fishes have tendency to lose water to the environment, especially across the gill epithelium. In order to replace the water loss, these fishes usually drink large amount of water unlike freshwater fishes.

They also have problem of excess of salts in the body due to drinking of sea water. Among the excess salts, Na^+, Cl^- and some amount of K^+ are removed across the gill epithelium while divalent ions like Mg^{++}, Ca^{++} are excreted by the kidney. Some fishes also have special salt secreting glands in the wall of rectum called **rectal glands** that remove salts into the digestive tract which are then eliminated from the body during egestion.

Terrestrial animals

The successful groups of land animals are arthropods among the invertebrates and reptile, birds and mammals among the vertebrates. The presence of chitinous exoskeleton in arthropods and dead keratinized skin in vertebrates are adaptation to reduce water loss by their bodies.

Desert mammals are very much resistant in this regard. They can tolerate against strong degree of dehydration by special metabolic and behavioural adaptation. This characteristic is called **anhydrobiosis**. Actually, these animals feed upon seeds of desert plants in which large amount of carbohydrate are stored. During the breakdown of these compounds; water is produced as by-product that is utilized by these animals. Best example of such animals is kangaroo rat. They avoid day time heat and emerge at night. 90% of the water that they use is metabolic water derived from cellular oxidation.

15.2 EXCRETION

Metabolism produces a number of toxic by-products, particularly the nitrogen containing compound. The **excretion** is the removal of chemical waste from the body which are produced by the metabolic processes within cells. The nitrogenous excretory products of animals are ammonia, urea and uric acid.

15.2.1 Relationship between Excretory Products and Habitats

The exact nature of excretory product is determined mainly by the availability of water to the organism which is based upon its habitat. The correlation with habitat is: (a) ammonia – aquatic (b) urea – aquatic and terrestrial (c) uric acid – terrestrial.

Ammonia

Ammonia is highly toxic because it tends to raise the pH of body fluids and interferes with membrane transport functions. It is highly soluble in water and diffuses rapidly across cell membrane. It is therefore excreted rapidly. One gram of nitrogen, in the form of ammonia, requires five hundred ml of water to dissolve it to nontoxic level. Such plenty of water can only be afforded by many aquatic organisms, particularly those in freshwater e.g., most fishes, protozoans, sponges, coelenterates. Animals which excrete ammonia as their major nitrogenous waste product are called **ammonotelic**.

Urea

Organisms with less freshwater available, such as some marine organisms and all terrestrial organisms remove their most of the nitrogenous waste in the form of urea. They will often invest some energy to convert the ammonia into urea, which is 100,000 times less toxic than ammonia. One gram of nitrogen, in the form of urea, requires 50 ml of water to dilute it to nontoxic level. Animals which excrete urea as their major nitrogenous waste product are called **ureotelic**.

Critical Thinking

Where do you think the carbon dioxide used in the formation of urea comes from? Where does the remainder of excess carbon dioxide go to be excreted?

Science Titbits

Humans excrete small quantities of uric acid but this is produced from the breakdown of nucleic acid and not from breakdown of proteins. Approximately one gram of uric acid is excreted in urine per day.

Uric acid

Uric acid is a purine even less toxic than urea, and it precipitates from solution, allowing the 4 nitrogen atoms per uric acid molecule to be excreted. One gram of nitrogen, in the form of uric acid, requires just 1 ml of water for its excretion. It has evolved in two groups with major

water loss problems, terrestrial invertebrates and egg-laying vertebrates. These animals are called **uricotelics**.

15.3 EXCRETORY SYSTEM OF MAN

The excretory system (urinary system) consists of kidneys, ureter, urinary bladder and a tubular urethra. The kidneys lie on either side of the vertebral column between the twelfth thoracic and third lumbar vertebrae.

Each **ureter** is a tubular organ about 25 cm long, which begins as the funnel-shaped renal pelvis. It extends downward parallel to the vertebral column to join the urinary bladder. It transports urine from the kidney to the urinary bladder. The **urinary bladder** is a hollow and distensible muscular organ. It is located within the pelvic cavity. It serves as urine reservoir. The **urethra** is a tube that carries urine from urinary bladder to the outside of the body.

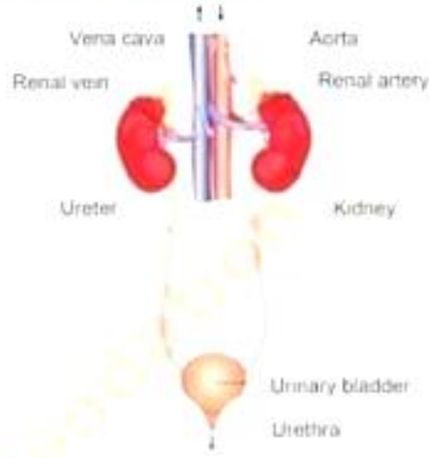


Fig. 15.5 Excretory system of man

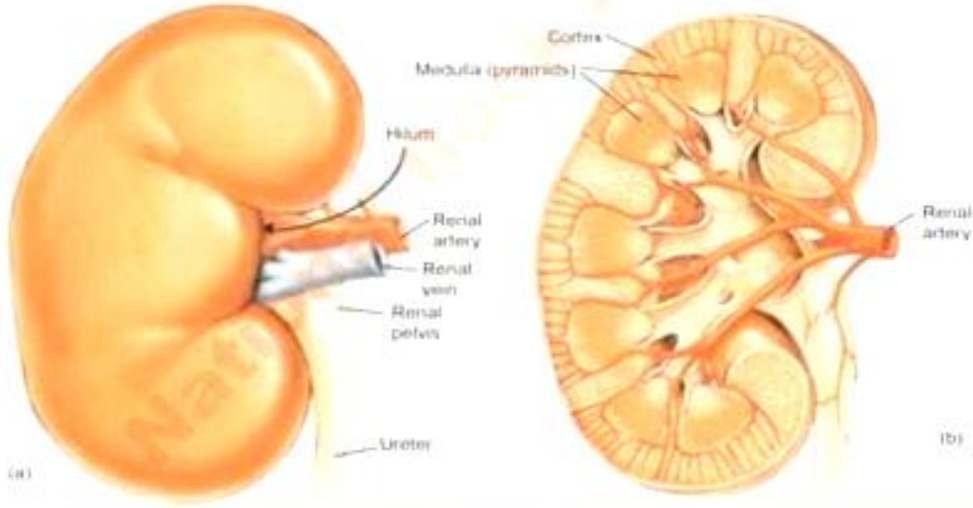


Fig. 15.6 Human kidney: **(a)** external structure **(b)** internal structure (longitudinal section)

15.3.1 Structure and Functions of Kidney

The science concerned with the structure, functions and diseases of the kidneys is called **nephrology**. A kidney is a reddish brown, bean shaped organ with a small surface. A fibrous connective tissue layer, called the **fibrous capsule**, encloses each kidney. The lateral surface of each kidney is convex, but its medial side is deeply concave. The resulting medial depression leads into a hollow chamber called the **renal sinus**. The entrance to this sinus is termed **hilum (hilus)**, where the renal artery and nerves enter and the renal vein and the

ureter exit. The kidney is divided into an outer **renal cortex** and inner **renal medulla** that surrounds the renal sinus. The renal medulla consists of a number of cone-shaped **renal pyramids.** Urine is collected in the renal pelvis and exit the kidney through **ureter**.

Structure of nephron

The **nephron** is the functional unit of kidney. A nephron consists of a **renal corpuscle** and a **renal tubule**. A renal corpuscle is composed of a network of capillaries called **glomerulus** which is surrounded by a thin double-walled structure called **Bowman's capsule.** The Bowman's capsule is an expansion at the closed end of a renal tubule. The **renal tubule** leads away from the Bowman's capsule and becomes highly coiled. This coiled portion of the tubule is called **proximal convoluted tubule.** The proximal convoluted tubule dips toward the renal pelvis into the medulla forming a sharp loop called **loop of Henle**. The loop of Henle consists of a descending limb and an ascending limb. The ascending limb returns to the region of the renal corpuscle, where it becomes highly coiled again, and is called the **distal convoluted tubule** which is connected to the **collecting duct**. The collecting duct receives many nephrons. Many collecting ducts combine together to form larger collecting ducts which empty into renal pelvis.

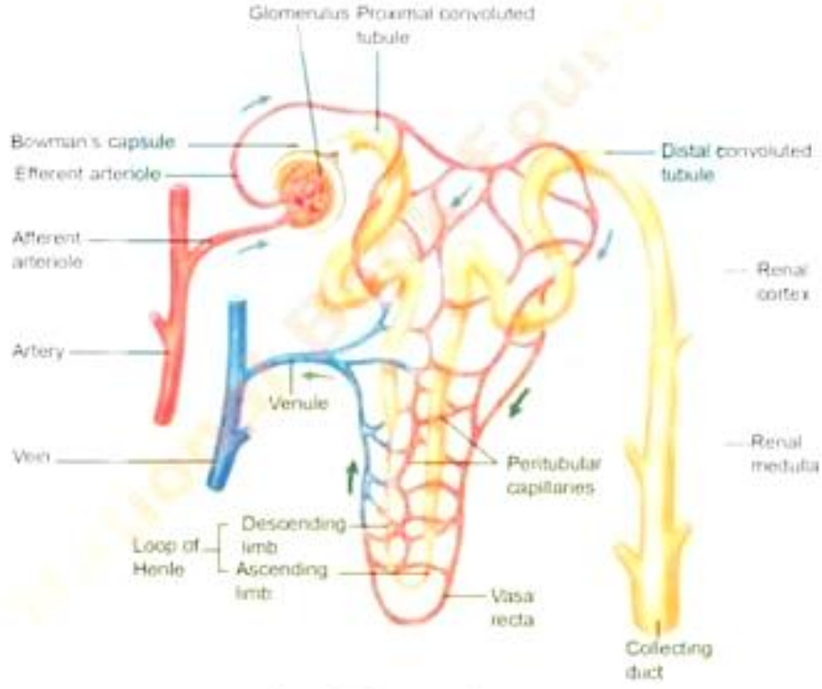


Fig. 15.7 Structure of nephron

Blood circulation to nephron

The renal artery within kidney gives rise branches which project into the cortex and give rise to **afferent arterioles**. The afferent arterioles supply blood to the glomerular capillaries of the renal capsule. **Efferent arterioles** arising from the glomeruli give rise to a plexus of capillaries called the **peritubular capillaries** around the proximal and distal tubules. Specialized part of the peritubular capillaries called **vasa recta** course into the medulla along with the loops of Henle and then back toward

the cortex. The peritubular capillaries drain into renal vein. The renal vein exits the kidney and connects to the inferior vena cava.

Functions of kidney

Kidneys function as excretory as well as osmoregulatory organs. Their excretory functions include the filtration of nitrogenous wastes from the blood and its removal outside the body in the form of urine. Being osmoregulatory organ, these are concern with the formation of diluted urine during the state of flooding and form concentrated urine during the state of dehydration.

Science Titbits

Efferent arterioles have somewhat less diameter than that of the afferent vessel. Therefore, a considerable blood pressure is developed in glomerulus.

Urine formation

The formation of urine involves glomerular filtration, tubular reabsorption and tubular secretion.

Glomerular filtration (pressure filtration) takes place in the **renal capsule** under pressure. The pressure comes from the blood pressure and is known as **hydrostatic pressure**. Glomerular capillaries have exceptionally high blood pressure than any other part of capillary bed in the body. The diameter of efferent arteriole is half as compared to the afferent arteriole so as the blood enters the narrow capillaries, pressure rise. Due to such a high pressure, water and small solute molecules are filtered out of the glomerular capillaries and are collected into the Bowman's capsule. Larger molecules like proteins, as well as red blood cells and platelets are left behind in the blood. The filtered fluid in the capsule is called **glomerular filtrate**. It has a chemical composition similar to that of blood plasma.

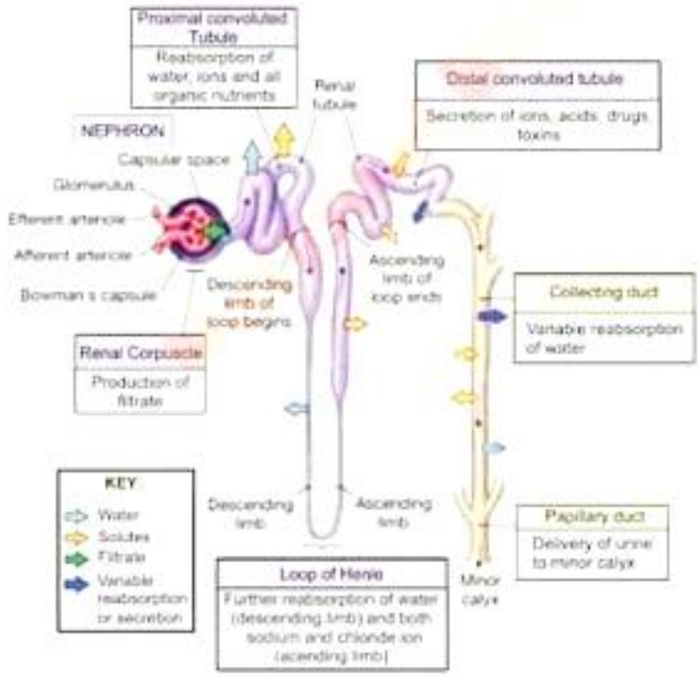


Fig. 15.8 Urine formation

Science Titbits

There are two types of nephrons, cortical nephrons and juxtamedullary nephrons. **Cortical nephrons** are found in the cortex. They have their renal corpuscle in the superficial renal cortex and have relatively short loops of Henle. 70 to 80% nephrons in human kidney are cortical. Under normal conditions of water availability, the cortical nephrons deal with the control of blood volume by forming diluted urine. **Juxtamedullary nephrons** have their renal corpuscle close to the junction of the cortex and medulla. They have long loop of Henle which extends deep into the medulla. These types of nephrons are relatively rare and only comprise 20-30% of the nephrons in the human kidney. When water is in short supply, increased water retention occurs through juxtamedullary nephrons.

Selective reabsorption, is the process by which certain substances that have been filtered out of the blood during ultrafiltration are reabsorbed. These substances include glucose, amino acids, vitamins, inorganic salts and some water. As only certain substances are reabsorbed, it is known as selective reabsorption.

Tubular secretion is the process by which certain substances e.g., ammonium, hydrogen ions are secreted mainly by the tubular epithelial cells of loop of Henle into the lumen of the tubule. However, to some extent this process also occurs in convoluted tubule. The main purpose of this secretion is to maintain the pH of the urine. Normal urine has pH range from 4.8 to 7.5.

The mechanism of urine concentration

Water is reabsorbed along the whole length of the nephron, but the formation of hypertonic urine is dependent on the reabsorption of water from the loop of Henle and collecting duct. This is achieved by **countercurrent multiplier mechanism**. Due to the counter current, filtrate moving in limbs of loop of Henle and the blood moving in the capillaries of **vesa recta**, water is greatly (approx. 99.5%) reabsorbed . As fluid travels up the ascending limb, sodium chloride is transported actively out of the limb into the surrounding area. This movement is controlled by **aldosterone**. This causes increase in the concentration of water in filtrate and decrease in concentration of water in kidney interstitium (space within a tissue or organ). As a result, water passes out of the descending limb by osmosis. This movement of water is also promoted by **anti-diuretic hormone** which is secreted from posterior lobe of pituitary.

Science Titbits

In addition to their excretory and osmoregulatory role, kidneys also help to control the red blood cell formation by secreting the hormone **erythropoietin** and help to regulate blood pressure by secreting the (EPO) enzyme **renin**.

15.4 DISORDERS OF URINARY TRACT

The normal aging process in human affects kidney function in various ways. Urinary tract infections (UTI) are fairly common. **Urology** is the branch of medicine which deals with diseases and abnormalities of urinary tract and their treatment.

15.4.1 Urinary Tract Infection

Although males can get a urinary tract infection, the condition is fifty times more common in women. In general, the higher risk in women is mostly due to the shortness of the female urethra, which is 1.5 inches compared to 8 inches in men. Bacteria from faecal matter at the anal opening can be easily transferred to the opening of the urethra. Almost all parts of the urinary tract are affected by the infection except ureters which are rarely the site of infection. The types of UTIs depending upon the site are: **urethritis** is an infection of urethra, **cystitis** involves the bladder and if the kidneys are infected the infection is called **pyelonephritis**.

Since the infection is caused by bacteria, it is curable by antibiotic therapy. For prevention, one should drink lot of water to flush out bacteria. Personal hygiene is especially important too.

Table 15.1 : Urinary Tract Infection (UTI) Caused by Bacteria

Bacteria	Diseases
1. *E. coli*	1. UTI
2. *N. gonorrhoeae*	2. Urethritis, Gonorrhoea
3. *T. palladium*	3. Syphilis

15.4.2 Kidney Stones

Urinary stones are hard, crystalline mineral materials that stick together to form small "pebbles" within the kidney or urinary tract. They can be as small as grains of sand or as large as golf balls. They may stay in kidneys or travel out of the body through the urinary tract. The condition of having stones in the kidney is termed **nephrolithiasis**.

Science Titbits

There are five major types of urinary stones: **calcium oxalate, calcium phosphate, magnesium ammonium phosphate, uric acid and cystine. Uric** stones are composed of combination of uric acid and calcium oxalate. They are normally 2-3 mm in diameter with either smooth or uneven surface. Branching stone is called **staghorn stone.**

Skills: Initiating and Planning

Hypothesize kidney stone by studying the urine test of relevant patient.

When urine is acidic (low pH) the stone is of calcium oxalate.

When the urine is alkaline (high pH) the stone is of calcium phosphate.

When urine is persistently acidic the stone is of uric acid type.

Kidney stones may be caused by increased calcium level in the blood which is termed as **hypercalcemia.** It, in turn, causes high calcium in the urine, the **hypercalciuria.** Increased oxalate level in the urine is called **hyperoxaluria**. Hypercalciuria and hyperoxaluria cause **calcium oxalate** type of kidney stones which are present in 70% of kidney stone patients. **Hyperuricemia** is the increased amount of uric acid in the blood and it causes **uric acid** type of kidney stones which are found in 10% of kidney stone patients. High concentration of cysteine and phosphates in urine also cause kidney stones. Continuous state of dehydration increases the chances of kidney stone formation.

Extracorporeal shock wave lithotripsy (ESWL) and Percutaneous Nephro Lithotripsy (PCNL) are common methods for kidney stone treatment. In ESWL, an instrument called **lithotripter** is used to generate shock waves from outside the patient's body focused on the stone, breaking it into small pieces. Most of the fragments then pass spontaneously via the urethra.

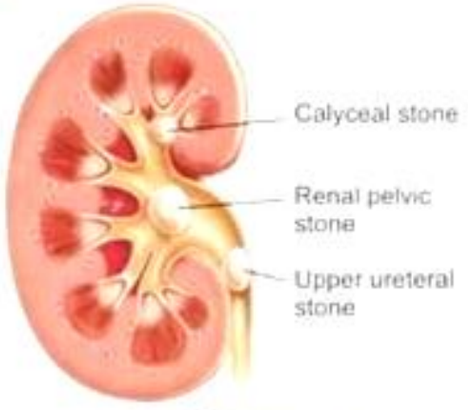


Fig. 15.9 Kidney stone

Fig. 15.10 Extracorporeal shock wave lithotripsy

In case of larger stone PCNL is preferred in which a tube is inserted from the patient's back into the kidney to create a tract. A scope is run through the tract to directly visualize the stone inside the kidney. Ultrasound equipment can then be inserted to breakup the stone. While watching the stone through the scope, the stone fragments can be grasped with special equipment and pulled through the tract out from the kidney. Open surgery is now almost never needed except for large bladder stone.

15.4.3 Kidney Failure

A general term for a decline kidney function particularly the efficiency of the filtering process is called **kidney failure** or **renal failure**. **Chronic renal failure** is the irreversible deterioration in renal function. It is a gradual, slowly progressive and occurs over a period of years.

Chronic renal failure may be caused by: (a) Bacterial infection of the pelvis and surrounding tissue. (b) Nephritis (inflammation of glomeruli). (c) Damage due to high blood pressure. (d) Diabetes mellitus.

Acute renal failure may be caused by: (a) Haemorrhage due to trauma. (b) Vomiting, diarrhoea. (c) Diuresis (excess excretion of urine), sweating. (d) Obstruction of the ureters, bladder or urethra e.g., kidney stone. (e) Severe nephritis.

Science Titbits

Chronic kidney failure can progress to **end-stage renal disease (ESRD)** and **uremia**, which is fatal unless artificial filtering (dialysis) or a kidney transplant.

15.4.4 Dialysis: Mechanism

A procedure to filter toxins from the blood by artificial methods when the kidneys are unable to perform this function is called **renal dialysis.** Dialysis works on the principle of kidneys although it is not as effective, efficient or thorough as the natural processes performed by the kidneys. There are two general types of renal dialysis: haemodialysis and peritoneal dialysis.

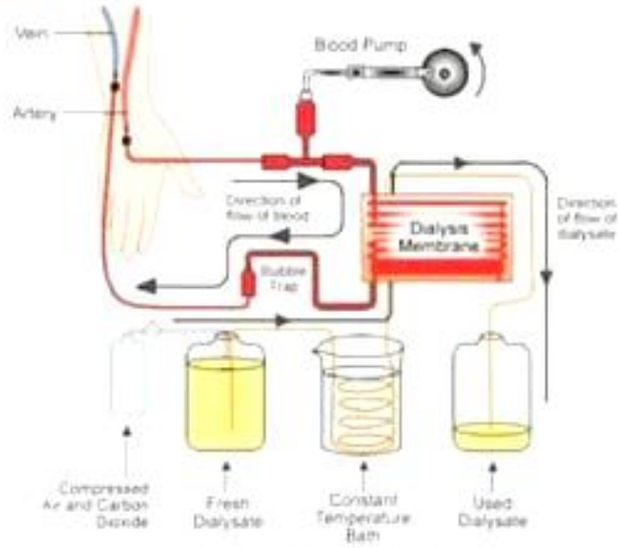


Fig. 15.11 Haemodialysis

Haemodialysis

Haemodialysis removes wastes and water by circulating blood outside the body through an external filter, called a **dialyzer**, which consists of tubes of semipermeable membrane. In this process, a **catheter** is inserted into a blood vessel, usually in the arm. It routes the blood circulation externally through a machine that removes wastes. The cleansed blood then returns to the body through a second catheter. The haemodialysis machine consists of a pump and a container in which a network of synthetic tubes made up of cellophane membrane, called the **dialyzer**, is situated. The blood moves into the tubes of dialyzer from the top through blood pump. After circulating through the dialyzer, blood leaves the machine from the bottom and transfuses (to pour out into another vessel, to transfer to another's vein) back to the body. On the other hand, **dialysate** (dialysis fluid) pour into the machine from bottom, which after circulating around the membranous tube, leaves the machine from the top. The dialysate attracts certain substances-minerals, electrolytes, and waste by-products—to cross the membrane from the blood. The dialysate absorbs these substances.

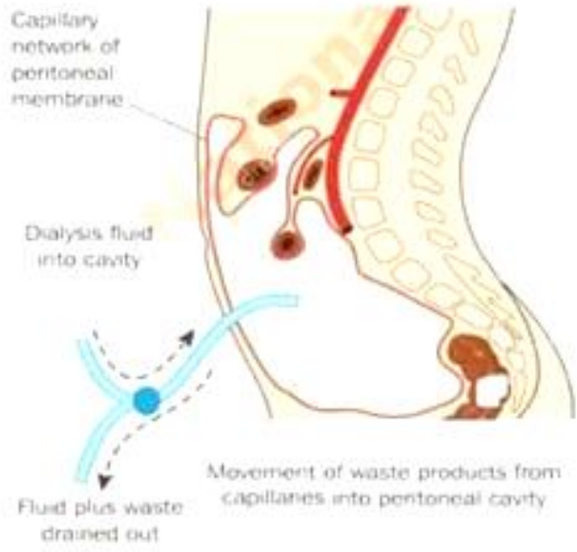


Fig. 15.12 Peritoneal dialysis

Peritoneal dialysis

Peritoneal dialysis involves the use of a natural membrane in the body, the peritoneum, which encloses the abdominal cavity. In this process, two catheters are surgically inserted into the abdominal cavity that serve as the portals (any entrance) through which **dialysate** enters and leaves the cavity. During circulation, when blood passes through the blood capillaries within the peritoneum, the dialysate attracts certain molecules to cross the membrane into the dialysate.

15.4.5 Kidney Transplant: Process and Problems

Kidney transplantation is the surgical procedure of placing a fully functioning kidney into a person with chronic kidney failure.

Principles of kidney transplant

The kidney graft is taken from a deceased donor or from a related or unrelated person. ABO blood group compatibility between donor and recipient is essential. It is usual to select donor kidneys on the basis of human leucocytes antigen (HLA) matching as this improves graft survival. A person can live normally with just one kidney.

Problems associated with kidney transplant

The two problems are rejection and toxic effects of cyclosporine. These problems are usually treated simultaneously by adding extra doses of **steroids**. Patients are required to take medications such as **cyclosporine** etc., to suppress their immune system in order to prevent rejection of the transplanted kidney. If at any point a recipient stops taking the medications, rejection can occur; even ten or fifteen years after the transplant.

DID YOU KNOW?

Dr. Adeeb Rizvi is a pioneer in treating kidney related diseases in Pakistan. He started off very humbly by just a ward in the Civil Hospital Karachi where he treated kidney patients. This 8 bedded ward was established in 1971 and today it has grown into a full-fledged institution called Sindh Institute of Urology and Transplantation (SIUT). Dr. Adeeb Rizvi, is recipient of many awards, locally and internationally. Treatment at SIUT is totally free of cost.

Science Technology and Society Connections

Describe the importance of kidney donation for the benefit of kidney failure patients.

Kidney donation is a relatively safe operation, and many donors will never feel the loss of their second kidney. It's the most expendable of organs. So giving up a kidney causes no disadvantage to your long-term health. In fact, studies have shown, that kidney donors actually live longer than the general population, because donors come from a pool of people in good health.

Just think people have no problem having only one kidney, so we have to ask, why did Allah give us two kidneys? Perhaps it is so you would have an extra one to donate and save a life.

15.5 THERMOREGULATION

Thermoregulation is defined as the maintenance of internal temperature within a range that allows cells to function efficiently. The body works to balance the amount of heat loss to maintain a stable internal temperature. Temperature colder or warmer than the enzymes optimum range, changes the shape of the active site and causes chemical reaction to stop.

15.5.1 Classification of Animals on The Basis of Temperature

Animals can be classified based upon ability to maintain constant body temperature as poikilotherms and homeotherms. **Poikilotherms** are all non-vertebrates, fishes, amphibian and reptiles. These are unable to maintain their body temperature within narrow limits using physiological mechanisms. **Homeotherms** are birds and mammals which are able to maintain a fairly constant body temperature by using physiological mechanisms.

Animals are also classified on the basis of source of body's heat as ectotherms and endotherms. **Ectotherms** animals produce metabolic heat at low level and that is also exchanged quickly with environment. They rely more on heat derived from the environment to raise their body temperature. Examples are most invertebrates, fishes, amphibians and reptiles. **Endotherms** animals produce their own body heat through heat production as by-product during metabolism in muscles, or by the action of hormones that increase metabolic rate. The examples of endotherms are birds and mammals.

15.5.2 Thermoregulatory Strategies in Man

Thermoregulatory centre in human body is located in the hypothalamus which acts as thermostat. It can detect the temperature of the blood that passes through it and, if the

Fig. 15.13 Thermoregulation in human

temperature increases or decreases even slightly, the hypothalamus initiates corrective responses such as sweating or shivering. When we encounter a particularly warm or cold environment, temperature receptors in the skin inform the hypothalamus. They also stimulate the higher, voluntary centres of the brain. This means that we 'feel' changing our clothing or turning the heating up or down. Often, this behavioural response corrects the situation without the need for any physiological response.

Hyperthermia is the body temperature above 37°C. There are two main physiological responses to heat, vasodilation and sweating. **Vasodilation** is the expansion of blood capillaries which lie just beneath the epidermis of the skin.So there is more flow of the blood in blood capillaries of the skin. Sweat glands spread sweat over the skin. Evaporation of sweat from the skin carries heat from the blood thus produces cooling effect.

Physiological responses to cold

Spasmodic contraction of the muscles is called **shivering.** This contraction produces heat which helps to raise the body temperature. **Vasoconstriction** reduces blood flow to the skin. **Piloerection** literally means "erection of skin hair". It traps air in the erected hair which is insulator for the heat. **Increased metabolic rate** is also a physiological response to cold.

Activity

Construct a flowchart that illustrates how wastes are removed by the kidneys.

Exercise

M.C.Qs

1. **Select the correct answer**

(i) Excretion of hypotonic urine in humans is associated best with the

(A) glomerular capsule (B) proximal convoluted tubule

(C) loop of the Henle (D) distal convoluted tubule.

(ii) The walls of the ---------- are made more or less permeable to water, depending on the need to conserve water.

(A) ureter (B) urethra

(C) fibrous capsule D) collecting duct.

(iii) Which of the following will cause a decrease in ADH production?

(A) dehydration (B) an increase in osmotic pressure of blood

(C) drinking water (D) abnormally low blood pressure.

(iv) The function of glomerulus and Bowman's capsule of the nephron is to

(A) reabsorb water into the blood (B) eliminate ammonia from the body

(C) reabsorb salts and amino acids. (D) filter the blood and capture the filtrate

(v) In man, glucose is present in blood plasma but not in urine. This is because glucose molecules are

(A) actively transported from the proximal convoluted tubule to blood capillaries

(B) oxidised to supply energy for ultrafiltration

(C) stored in the kidney (D) too large to enter Bowman's capsule

(vi) Evidence for glomerular filtration in the kidney could be obtained by comparing the sizes of the molecules present in Bowman's capsule with those in the

(A) afferent blood vessel (B) collecting duct

(C) loop of Henle (D) proximal tubule

(vii) The site and principal mechanism for the passage of glucose into the bloodstream in the human kidney is the

(A) collecting duct, by active secretion (B) glomerulus, by selective reabsoption

(C) glomerulus, by ultrafiltration

(D) proximal convoluted tubule, by selective reabsorption

(viii) A drug reduces mitochondrial activity in kidney nephrons. Which chemical will be present in increased amounts in the urine?

(A) ammonia (B) glucose (C) uric acid (D) urea

(ix) The main difference between endotherms and ectotherms is

(A) how they conserve water (B) where from they get most of their body heat

(C) whether they are warm or cold blooded

(D) whether they live on land or in the water

(x) The water content of human blood is regulated by ADH. In which part of the nephron does regulation occur?

(A) ascending limb of loop of Henle (B) descending limb of the loop of Henle

(C) Bowman's capsule (D) proximal convoluted tubule

Short Questions

2. What are osmoregulators and osmoconformers?
3. Name the organs of the urinary system and write their major functions.
4. What is anhydrobiosis?
5. Describe glomerular filtration.

6. Describe the countercurrent multiplier mechanism.
7. Name the parts of a nephron and trace the blood supply to the nephron.
8. Name general processes which are involved with urine formation?
9. Describe urinary tract infection.
10. Name three urinary tract infections and bacteria responsible.
11. What are the causes of kidney failure?
12. By what physical processes do solutes enter or leave the blood during dialysis?
13. Why do blood and dialysate flow in opposite direction?
14. Suggest two problems that might occur if the dialysate was pure water.
15. How animals can be classified on the bases of the ability to thermoregulation?
16. How do blood vessels in the skin help regulate body temperature during hot and cold external condition?
17. Why women are more likely to acquire UTI as compared to men?
18. Write the differences between:
    (a) osmoregulation and osmoconformers
    (b) ammonotelic and ureotelic
    (c) ureotelic and uricotelics
    (d) proximal and distal convoluted tubule
    (e) afferent and efferent arterioles
    (f) hypercalcemia and hyperuricema
    (g) extracorporeal shock wave lithotripsy and percutaneous nephronlithotripsy
    (h) chronic renal failure and acute renal failure
    (i) peritoneal dialysis and haemodialysis
    (j) renal cortex and renal medulla
    (k) vasodilatation and vasoconstriction
    (l) dialyzer and dialysate

Extensive Questions

19. Describe the osmoregulatory adaptations in
    (a) Freshwater animals (b) Marine animals (c) Terrestrial animals
20. Discuss relationship between excretory products and habitats.
21. Describe the structure and function of human kidney.
22. Describe the structure of human nephron.
23. Discuss the 'urine formation 'and mechanism of urine concentration in man.
24. What are kidney stones? Discuss the causes and treatment of kidney stones?
25. What is renal dialysis? Describe the two types of renal dialysis.
26. What is kidney transplantation? Describe principles of kidney transplant. What are the problems associated with kidney transplant?
27. What is thermoregulation? Classify animals on the basis of temperature. What are the thermoregulatory strategies in man?

21

DEVELOPMENT AND AGING

After completing this lesson, you will be able to: This is a 20 days unit

- Describe cleavage and relate it with amount of yolk.
- Explain the events of gastrulation.
- List the tissues and organs formed from the three germ layers.
- State the events of neurulation.
- Describe the formation of neural crest and list the structures that are derived from neural crest cells.
- Define organogenesis.
- Through experimental narration, describe the role of the nucleus and cytoplasm in controlling development.
- Give a brief overview of the work done by Hans Spemann in the discovery of induction.
- Define organizers and differentiate between primary and secondary induction.
- Describe the events of development in human in terms of first, second and third trimesters.
- Describe in brief the development of twins and quadruplets.
- Describe the structural details of placenta and umbilical cord.
- Differentiate the terms gestation and pregnancy.
- Describe the role of fetal and maternal hormones in initiating labor pains and culminating in the birth of baby.
- Define the term premature birth and correlate it with the growth phases in the second and third trimesters.
- Define afterbirth and describe how umbilical cord is detached from the baby.
- Define colostrum and describe the role of prolactin in the production and of oxytocin in the secretion of milk.
- State the hormonal regulation in the end of milk production.
- Compare breast-feeding and bottle-feeding, in terms of advantages and disadvantages.
- Relate the major genetic abnormalities in embryos with spontaneous abortion.
- Disorders during embryonic development.

- Define the term aging.
- Rationalize aging as a part of normal development.
- List the genetic and extrinsic factors responsible for aging.
- State the changes (graying, thinning hair, pigmented patches of skin, slowed movements, fading vision, impaired hearing, reduced ability to adapt to stress and decreased resistance to infections) as primary aging.
- State the changes that are the result of environmental, lifestyle factors such as disease, disuse (lack of exercise), and abuse (smoking, obesity, malnutrition, and exposure to ultra-violet light) as secondary aging.

• Zygote is a single diploid cell.

Reading

• all those changes that takes place from zygote to form new individual. (Development)

Development is a period of extensive growth in size and mass as well as ongoing differentiation of organ system established in embryonic period. Many different systems formed in the developmental period of an organism, such as brain, respiratory system digestive system etc. These are positive changes which lead an organism from its zygotic stage to adult form, and then further growth leads an organism to its full size. However, these positive changes become negative at some stage in the life cycle, which are termed as aging.

21.1 EMBRYONIC DEVELOPMENT

The progressive changes which are undergone before an organism acquires its adult like form constitute the embryonic development. It begins with a series of mitotic divisions in the zygote which leads to a multicellular stage called **embryo**, finally an adult like body is formed. The study of an organism at this stage is called **embryology**. The process of embryonic development comprises following stages: cleavage, gastrulation, organogenesis and growth.

21.1.1 Cleavage (repeated mitotic division)

Following fertilization, the zygote undergoes a mitotic division called **cleavage**.

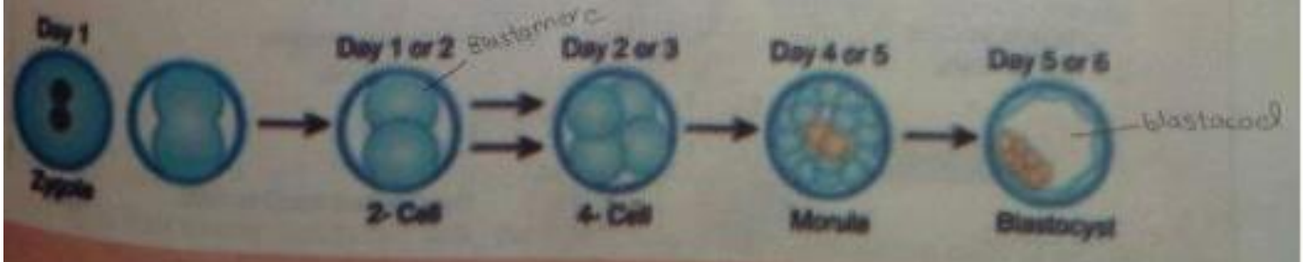


Fig. 21.1 Cleavage in human zygote

MITOSIS (Extra reading material)

Mitosis is a process where a single cell divides into two identical daughter cells (cell division). During mitosis one cell divides once to form two identical cells. The major purpose of mitosis is for growth and to replace wornout cells. Mitosis is divided into five phases:

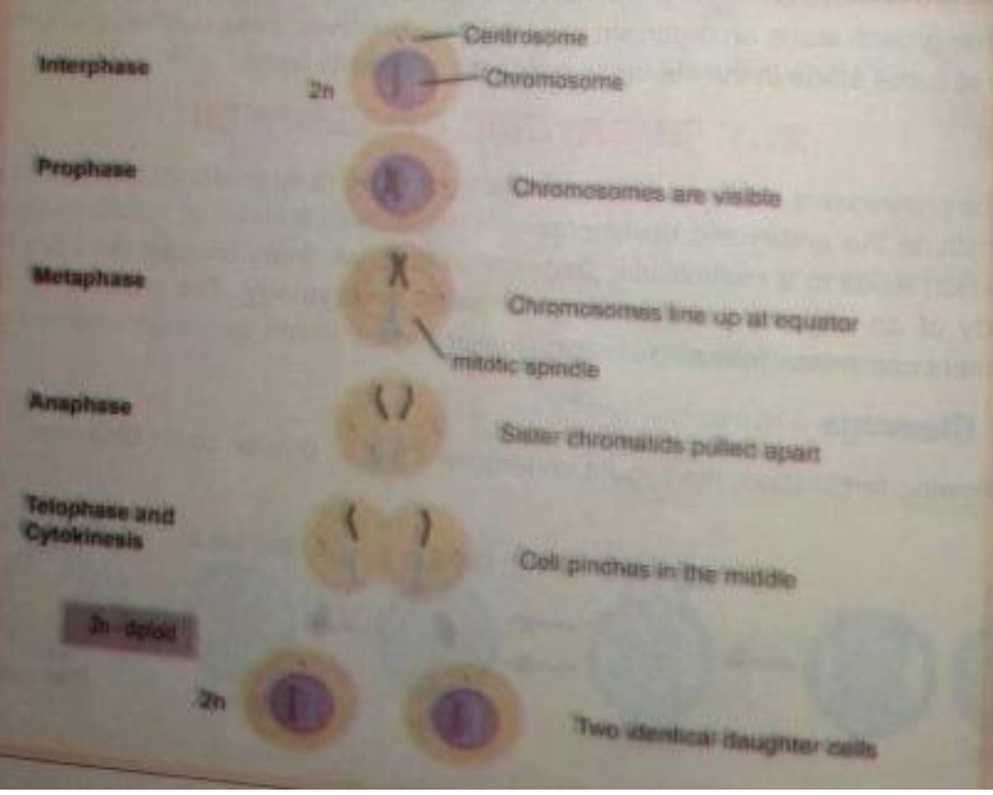
1. **Interphase:** The DNA in the cell is copied in preparation for cell division, these results in two identical full sets of chromosomes. Outside of the nucleus are two centrosomes, each containing a pair of centrioles. During interphase, microtubules extend from these centrosomes.

2. **Prophase:** The chromosomes condense into X-shaped structures that can be easily seen under microscope. Each chromosome is composed of two sister chromatids, containing identical genetic information. At the end of prophase the membrane around the nucleus in the cell dissolves away. The mitotic spindle, consisting of the microtubules and other proteins, extends across the cell between the centrioles as they move to opposite poles of the cell.

3. **Metaphase:** The chromosomes line up neatly end-to-end along the centre (equator) of the cell. The centrioles are now at opposite poles of the cell with the mitotic spindle fibres extending from them. The mitotic spindle fibres attach to each of the sister chromatids.

4. **Anaphase:** The sister chromatids are then pulled apart by the mitotic spindle which pulls one chromatid to one pole and the other chromatid to the opposite pole.

5. **Telophase:** At each pole of the cell a full set of chromosomes gather together. A membrane forms around each set of chromosomes to create two new nuclei. The single cell then pinches in the middle to form two separate daughter cells each containing a full set of chromosomes within a nucleus. This process is known as cytokinesis.



Morula and Blastula

The first division results in the formation of two identical cells called **blastomere**. DNA replication and mitotic division occur repeatedly. The cells get smaller and smaller with each division. Finally a solid ball of small cells, the **morula** is formed. The morula is still about the same size as the zygote. As cleavage or division continues, cells begin to move apart, so that spaces appear among cells in the centre of mass. Cells keep pulling away from the central area, forming a fluid cavity known as **blostocoel.** This hollow-sphere embryo which develops at the end of cleavage is called **blastula**. This embryonic stage in mammals is called **blastocyst**. The blastocyst is a fluid-filled hollow sphere composed of a single layer of large, flattened cells called **trophoblast cells** and a small cluster of 20 to 30 rounded cells, called the **inner cell mass**, located at one side.

Different patterns of cleavage based upon amount of yolk

Many animal eggs contain yolk. The yolk is a mixture of proteins, phospholipids and fats and serves as food for developing embryo. The amount and distribution of yolk vary among different animal groups. Most invertebrates and simple chordates have eggs with relatively small amounts of yolk uniformly distributed through the cytoplasm. Many vertebrates have large amounts of yolk concentrated at one end of the cell known as the **vegetal pole**. The opposite pole is called the **animal pole**. The amount of the yolk in the egg affects the pattern of cleavage. The cleavage is of two types: holoblastic cleavage, meroblastic cleavage.

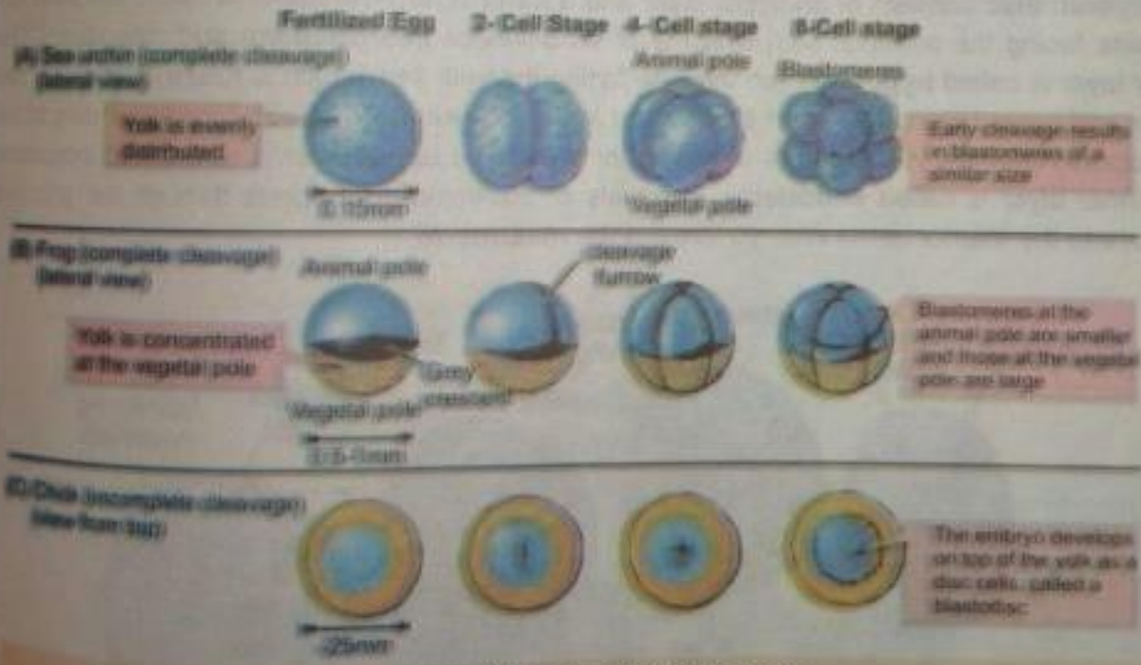


Fig. 21.2 Different patterns of cleavage

Holoblastic cleavage

imp (S.P) Differentiate blw

In eggs with evenly distributed yolk, the entire egg divides, producing cells of roughly the same size. This type of cleavage is termed **holoblastic**. e.g., bony fishes and amphibians.

Meroblastic cleavage

The eggs of reptiles, birds and some fishes have a very large amount of yolk and only a small amount of cytoplasm concentrated at the animal pole. In such eggs, cell divisions take place only in the **blastodisc**, the small disc of cytoplasm at the animal pole. This type of cleavage is termed **meroblastic**.

21.1.2 Gastrulation

Gastrulation is the second major phase of embryonic development which is characterized by differentiation of embryonic **germ layers**. In the process, the embryo is transformed from a hollow ball of cells, the blastula, into a three layered stage called the **gastrula**. The three layers produced in gastrulation are embryonic tissues called **ectoderm**, **endoderm** and **mesoderm**. The mechanism of gastrulation varies somewhat depending on the species.

Gastrulation in human

In human embryo, gastrulation begins about the 15th day after fertilization. During this phase, the **trophoblast** thickens at one point to form a mass of cells called **inner mass cells**. After implantation, the inner cell mass grows and splits, forming two fluid-filled sacs that are separated by a double layer of cells called the **embryonic disc**. One sac is **amniotic sac** which is bounded by the **amnion** and filled by **amniotic fluid**. The other sac is **yolk sac** but it does not contain yolk as human embryo takes nutrition from the mother's body. At this stage, the **embryonic disc** consists of an upper layer and a lower layer. The upper is called **epiblast** (on the side facing the amniotic sac) which later on develops into ectoderm and mesoderm. The lower layer is called **hypoblast** (on the side facing the yolk sac) which is future endoderm. The upper and lower layers split apart slightly and a slit, the **primitive streak** (corresponding to the blastopore) appears in the centre of the upper layer. The upper layer is now called **ectoderm** and inner layer is called **endoderm**. The cells of the ectoderm migrate through the primitive streak into the interior of the embryo forming the **mesoderm**.

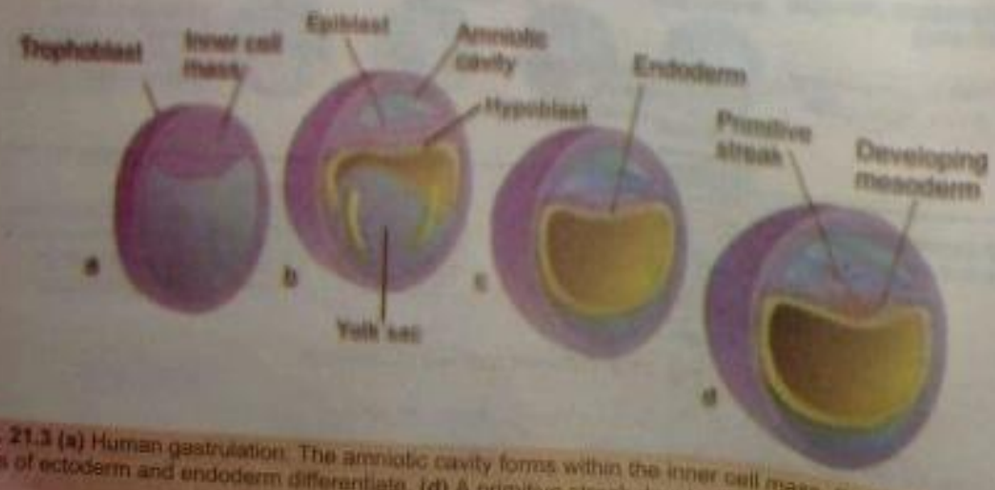


Fig. 21.3 (a) Human gastrulation: The amniotic cavity forms within the inner cell mass, **(b-c)** and in its base, layers of ectoderm and endoderm differentiate. **(d)** A primitive streak develops, through which cells destined to become mesoderm migrate into the interior.

Fate of three embryonic germ layers

The three primary germ layers serve as the **primitive tissues** from which all body organs will derive. Ectoderm fashions structures of the nervous system and the skin epidermis. Endoderm forms the epithelial linings of the digestive, respiratory, and urinogenital systems and associated glands. Mesoderm forms virtually everything else.

21.1.3 Organogenesis

The formation of organs and systems during embryonic development is called **organogenesis**. The first major event in organogenesis is **neurulation**, the differentiation of ectoderm that produces the brain and spinal cord.

Neurulation in human embryo

In human embryo, the process of neurulation begins during the first month of development. The nervous system develops from the middle ectoderm located just above the notochord. The developing notochord induces (stimulates) the overlying ectoderm to thicken, forming the **neural plate** which is seen along the dorsal surface of the embryo. Central cells of the neural plate move downward and form a depression called the **neural groove**. Then neural folds develop on either side of a neural groove. By 22nd day, the continued cell movement brings the superior margins of the neural folds closer together until they meet and fuse forming the **neural tube**. It soon pinches off and becomes covered by surface ectoderm. At this point the embryo is called **neurula**. Later the anterior of the neural tube grows and differentiates into brain; the remainder of the tube develops into the spinal cord.

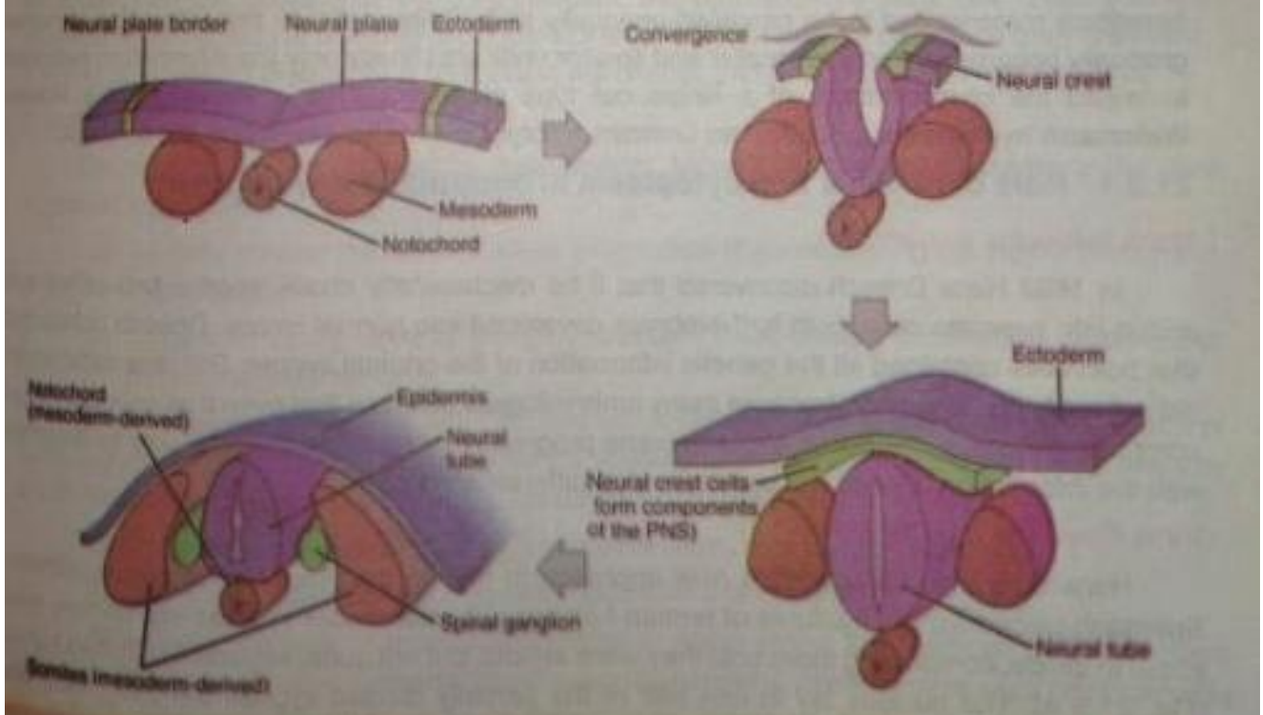


Fig. 21.4 Neurulation and neural crest formation

By the end of the first month of development, the three primary brain vesicles (fore, mid and hindbrain) are obvious. By the end of the second month, all brain flexures are evident, the cerebral hemispheres cover the top of the brain stem and brain waves can be recorded.

Formation of neural crest and its role in development

Various motor nerves grow out of the developing brain and spinal cord, but sensory nerves have a separate origin, the **neural crest** which is developed in the region of the neural plate border. After neural tube closure, the associated neural crest cells migrate widely to the lateral sides of neural tube and give rise to the cranial, spinal, and sympathetic ganglia and associated nerves. Neural crest cells subsequently migrate to various parts of the embryo, forming peripheral nerves, medulla of the adrenal gland, teeth, skull bones and so many other different cell types that some have proposed considering neural crest cells as a **"fourth germ layer"**.

Growth is the most obvious phase of development which is characterized by increase in size and mass of an organism. It begins soon after fertilization and remain continue throughout the development. In human embryo the process of organogenesis is almost accomplished in the embryo of about eight weeks. By the start of third month the human embryo is called **foetus**. The rest of the development period comprises only growth and maturation of organs and systems.

21.2 CONTROL OF DEVELOPMENT

How does a developing embryo generate the multitude of many cell types of a complete multicellular organism from the starting point of a single diploid nucleus of a zygote? To many 19th century embryologists there seemed only one acceptable answer: as cell division ensued, hereditary material had to be parcelled unequally to daughter cells. In this view, the genome gradually became broken into smaller and smaller units until finally only the information required to impart the characteristics of a single cell type remained. This is known as the **Roux-Weismann hypothesis**, after the two German embryologists who developed the concept.

21.2.1 Role of nucleus and cytoplasm in controlling development

Hans Driesch's Experiment

In 1892 Hans Driesch discovered that if he mechanically shook apart a two-celled sea urchin into separate cells, both half-embryos developed into normal larvae. Driesch concluded that both cells contained all the genetic information of the original zygote. Still, this experiment did not settle the argument, because many embryologists believed that even if all cells contained complete genomes, the nuclei might become progressively modified in some way to dispense with the information they do not use in forming differentiated cells.

Hans Spemann's experiment

Hans Spemann introduced a new approach to testing the Roux-Weismann hypothesis. Spemann placed minute ligatures of human hair around salamander zygotes just as they were about to divide, constricting them until they were almost, but not quite, separated into two halves (Fig. 21.5 a). The nucleus lay in one half of the partially divided zygote; the other side was **anucleate**, containing only cytoplasm. The zygote then completed its first cleavage division on

the side containing the nucleus; the anucleate side remained undivided. Eventually, when the nucleated side had divided into about 16 cells, one of the cleavage nuclei would wander across the narrow cytoplasmic bridge to the anucleate side. Immediately, this side began to divide. With both halves of the embryo containing nuclei, Spemann drew the ligature tight, separating the two halves of the embryo. He then watched their development. Usually two complete embryos will be resulted. This showed that a single nucleus selected from the 16-cell embryo contained a complete set of genes; all were equivalent.

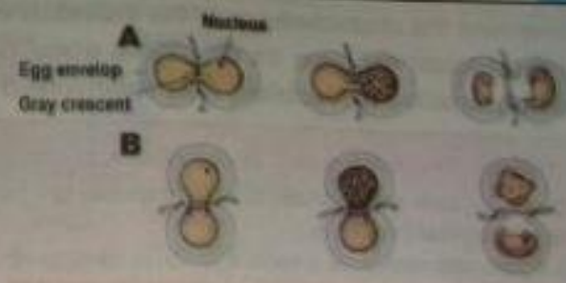


Fig. 21.5 Spemann's delayed nucleation experiments. Two kinds of experiments were performed. A, Hair ligature was used to constrict an uncleaved fertilized newt egg. Both sides contained part of the grey crescent. The nucleated side alone cleaved until a descendant nucleus crossed over the cytoplasmic bridge. Then both sides completed cleavage and formed two complete embryos. B, Hair ligature was placed so that the nucleus and grey crescent were completely separated. The side lacking the grey crescent became an unorganized piece of belly tissue; the other side developed normally.

Sometimes, however, Spemann observed that the nucleated half of the embryo developed only into an abnormal ball of "belly" tissue, although the half that received the delayed nucleus developed normally. Here question arises, why nuclei of 15 cells out of 16 fail to develop and the one which was transferred to non-nucleated side resulted in embryo development? *Explanation*: Spemann discovered, depending on the position of the grey crescent, the **pigment-free area** that appears at the moment of fertilization. If one-half of the constricted embryos lacked a part of the **grey crescent**, it would not develop (Figure 21.5 b), instead will change into undifferentiated mass of cells.

Conclusion

Spemann's delayed nucleation experiments served as compelling evidence for two important conclusions:

a) All cells contain the same nuclear information (thus disproving the Roux- Weismann hypothesis),

b) Cytoplasm in the area of the grey crescent must contain information essential for normal development.

If all nuclei are equivalent, what causes some cells to develop into neurons while others develop into skeletal muscle? In most animals (excluding insects), there are two major ways by which cells become committed to particular developmental fates:

a) Cytoplasmic segregation of determinative molecules during cleavage.

b) Interaction with neighbouring cells (inductive interactions).

All animals use both of these mechanisms to some extent to specify different cell types. However, in some animals cytoplasmic specification is dominant, whereas others rely predominantly on inductive interactions. From all these experiments, it was concluded that both nucleus and cytoplasm play an important role in the development. Nucleus contains all the genes, which

determine the characteristics of the individual while cytoplasm is involved in the selection of genes in a particular cell type as it contains some components called **morphogenetic determinants (determinative molecules)** that are involved in the expression of genes.

Science, Technology and Society Connections

Describe how a blastula is divided into two (by using micromanipulator) to produce twins of the animals for biological research.

A micromanipulator, is a device which is used to physically interact with a sample under a microscope, where a level of precision of movement is necessary that cannot be achieved by the unaided human hand. It may typically consist of an input joystick, a mechanism for reducing the range of movement and an output section with the means of holding a micro tool to hold, inject, cut or otherwise manipulate the object as required. The best example of micromanipulation is the division of blastula in order to produce twin embryos for the purpose of biological research.

21.2.2 Embryonic Induction

The capacity of some cells to evoke a specific developmental response in others is called **embryonic induction**. It is a widespread phenomenon in development which was reported by Hans Spemann and Hilde Mangold in 1924 in a series of classic experiments on amphibian's embryos.

Spemann's experiment

In this experiment, when a piece of dorsal blastopore lip from a salamander gastrula was transplanted into a ventral or lateral position of another salamander gastrula, it invaginated and developed a notochord and somites. It also induced the host ectoderm to form a neural tube. Eventually, a whole system of organs developed where the graft was placed, and then grew into a nearly complete secondary embryo. This creature was composed partly of grafted tissue and partly of induced host tissue. It was soon found that only grafts from the dorsal lip of the blastopore were capable of inducing the formation of a complete or nearly complete secondary embryo. This area corresponds to the presumptive areas of notochord, somites, and prechordal plate. It was also found that only ectoderm of the host would develop a nervous system in the graft and that the reactive ability was greatest at the early gastrula stage and declined as the recipient embryo got older.

Spemann designated the dorsal lip area the **primary organizer** because it was the only tissue capable of inducing the development of a secondary embryo in the host. He also termed this inductive event **primary induction** because he believed it to be the first inductive event in development. Subsequent studies showed that many other cell types originate by inductive interactions, a process called **secondary induction**. For example, cells of the neural plate induce neural crest in the embryo.

Science Titbits

Usually cells that have differentiated, act as inductors for adjacent undifferentiated cells. Timing is important. Once a primary inductor sets in motion a specific developmental pattern in some cells, numerous secondary inductions follow.

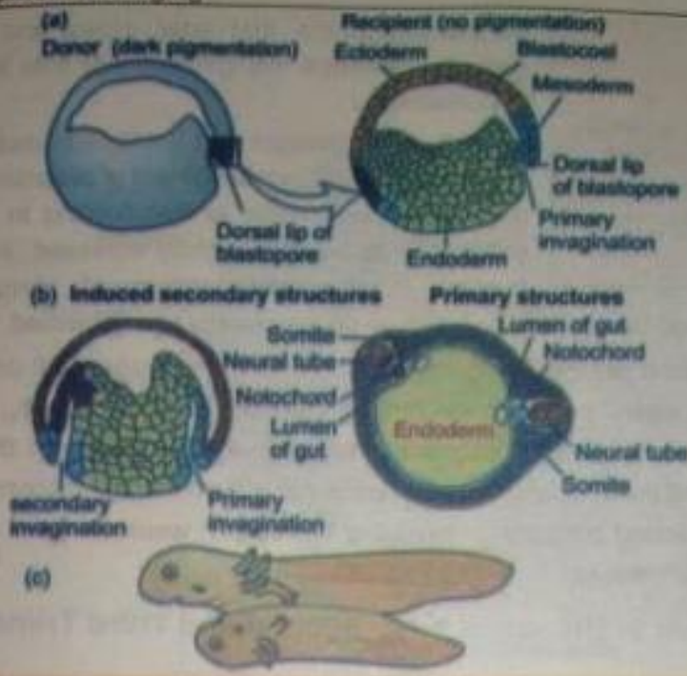


Fig. 21.6 Spemann's experiment showing the demonstration of embryonic induction (a) The blastopore lip from an early gastrula of a pigmented newt embryo was transplanted to the ventral vegetal side of an unpigmented recipient embryo. Because of the differences in pigmentation, the donor and recipient tissues could be distinguished visually. (b) The donor tissue induced the recipient tissue to form a secondary embryonic axis containing a notochord, neural tube, somites, and gut. (c) Eventually, a twinned embryo developed.

21.3 HUMAN EMBRYONIC DEVELOPMENT

Embryonic development begins with fertilization. After fertilization, zygote undergoes changes which leads the formation of foetus and then ultimately to an adult.

Fertilization of an egg by a sperm (also called conception in humans) normally occurs in the proximal part of oviduct. As the zygote passes down oviduct it undergoes **cleavage** which leads to the formation of **blastocyst.** Pregnancy is usually established when blastocyst is implanted into the **endometrium**. If the implantation is successful the embryo begins to secrete **human chorionic gonadotropin** (hCG). This hormone forces the corpus luteum in the ovary to continue to secrete progesterone, thereby maintaining the endometrium and inhibiting FSH production. The **chorion**, one

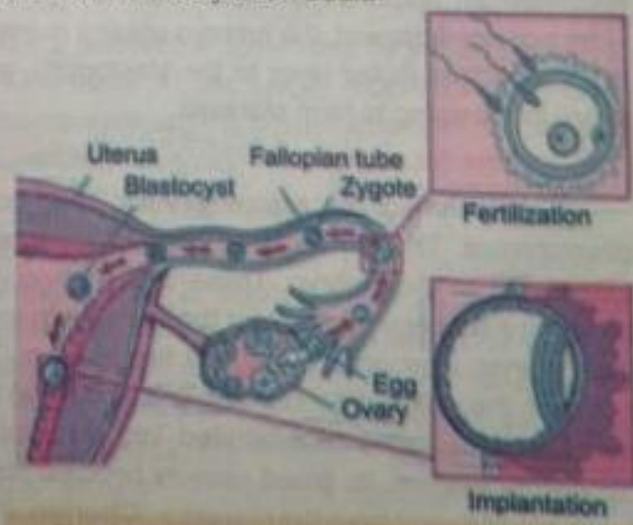


Fig. 21.7 Fertilization and implantation in human

Science Titbits

Pregnancy test, which is readily available in hospitals and clinics, is based on the fact that hCG is present in the blood and urine of a pregnant woman.

of the membrane that later grows and surrounds the embryo, develops villi (projections) that burrows into the endometrium.

It's often thought that fertilization and conception are synonymous with establishment of pregnancy. However, at this stage, the zygote is free-floating in the womb. For pregnancy to be successfully achieved, the embryo must implant in the endometrium of uterus. Sometimes, conception is not followed by implantation, hence no pregnancy is established.

The human **gestation period**, the duration of pregnancy, averages 280 days from the time of the mother's last menstrual period to the birth of the baby or 266 days i.e., about 9 months from the time of conception. Human pregnancy can be divided roughly into **three trimesters**, each approximately three months long. The first trimester is from the last menstrual period (LMP) to the 13th week, the second trimester is from the 14th to 27th week, and the third trimester is from the 28th week to 36th weeks.

21.3.1 Development of Human in First, Second and Third Trimester

First trimester

Since the gestation period starts from last menstrual period (LMP), therefore, fertilization, blastocyst formation and implantation of blastocyst are included in 1st trimester. In addition, 1st trimester also comprises development of placenta, extra embryonic layers and major events of organogenesis.

Fertilization, blastocyst formation and implantation

After fertilization, the zygote undergoes cleavage that leads to the formation of blastocyst. By the end of the 2nd week, implantation of the blastocyst in the endometrium begins. The endometrium responds to implantation by growing over the **blastocyst**. During the first 2-4 weeks of development, the embryo obtains nutrients directly from the endometrium. Meanwhile the trophoblast (outer layer of the blastocyst), grows out and mingles with the endometrium, eventually helping to form placenta.

Extraembryonic membrane

Establishment of extraembryonic membrane is one of the major events in early development. The term extraembryonic membrane is appropriate because these membranes extend out of the embryo. These are: amnion, yolk sac, allantois and chorion. The **amnion** provides a fluid environment for the developing embryo and foetus. Firstly amnion is seen above the embryo; later on it surrounds the embryo. The **yolk sac** appears below the embryo. In humans, the yolk sac contains no yolk and is the first site of red blood cell formation. Part of this membrane becomes incorporated into the umbilical cord. The **allantois** contributes to the circulatory system. Its blood vessels become blood vessels of umbilical cord, which transport foetal blood to and from the placenta. **Chorion**, the outer extra embryonic membrane surrounds the embryo. It becomes the part of the placenta.

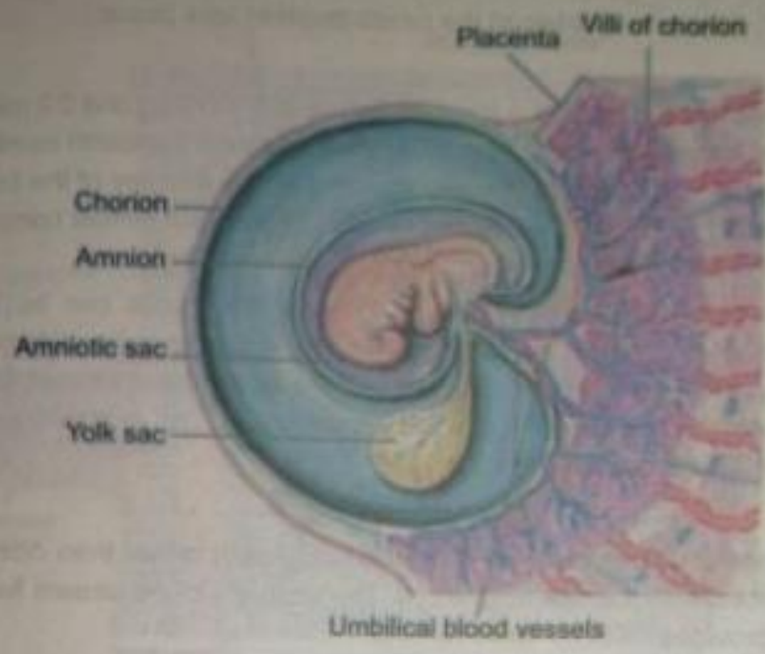


Fig. 21.8 Extraembryonic layers

Placenta development

The placenta is an organ that connects the developing foetus (fetus) to the uterine wall to allow nutrient uptake, waste elimination, and gas exchange via the mother's blood supply. In addition, the placenta is an endocrine organ that secretes hormones, progesterone and estrogen to maintain pregnancy. The placenta begins to develop upon implantation of the blastocyst into the maternal endometrium. The outer layer of the blastocyst becomes the trophoblast, which forms the outer layer of the placenta. The placenta grows throughout pregnancy. However, development of the maternal blood supply to the placenta is completed by the end of the first trimester of pregnancy (approximately 12–13 weeks). At this stage, **hCG** (human chorionic gonadotropin) declines, the corpus luteum degenerates and the placenta completely takes over the production of progesterone, which maintains pregnancy.

Major events of organogenesis

As the development of extraembryonic membranes and the placenta proceeds, the process of gastrulation and development of the embryo is also going on. At the end of the **second month,** the embryo's tail has disappeared and the arms and legs are more developed with fingers and toes apparent. Internally all major organs have been appeared. Embryonic development is now finished. At the end of the embryonic period, all organ system are established and there is a mature and functioning placenta. The embryo is only about 38 mm (1 ½ inch) long. After the first two months of development the embryo is called a **foetus**. During

the **third month** of pregnancy, the foetus grows rapidly. It is about 85 mm (3 ½ inches). At the end of 1st trimester, differences between the sexes begin to take place.

Second trimester

During the second trimester, the foetus grows to about 0.6 kg and 0.3 meter (1 foot) long. The **four month** old foetus looks quite human. Halfway through the fourth month, the foetus can bring the hand together and suck the thumb. At about **8th week** many of the bones are forming by replacing the cartilages. By **15th week**, the sensory organs are almost completely developed and by **16th week** the foetus is actively turning inside the mother.

By the end of the **5th month** the heartbeat of the foetus can be heard through a stethoscope. At the end of the **6th month**, the head is no longer quite as large compared with the rest of the body, the eyelids have separated and the lashes have formed. During the second trimester, the foetus grows to about 30 cm and is very active. The mother may feel movements during the early part of the second trimester.

Third trimester

The third trimester is predominantly a period of growth rather than one of development. The weight of the foetus doubles several times. The mother's blood stream fuels all this growth by the nutrients it provides.

As the end of the development approaches the foetus usually rotates, so the head is pointed toward the cervix. At the end of 9th months, the foetus is about 530 mm (20 ½ inches) long.

Skills: Initiating and Planning

- **Proper nourishment of mother is imperative during the third trimester of pregnancy.**

The third trimester is a period of growth. The mother's blood stream fuels all of this growth by the nutrients it provided. Within the placenta these nutrients pass into the foetal blood supply. If the foetus is malnourished because the mother is malnourished, this growth can be relatively affected. The result is the severely retarded infant, so proper nourishment of mother is necessary.

21.3.2 The Placenta

The placenta is the organ that provides nutrients and oxygen to the embryo and helps dispose of its metabolic wastes, formed of the embryo's chorion and mother's endometrial blood vessels.

Structure of placenta

The structure of the placenta consists of tissue from foetal part and maternal part. The **foetal part** consists of **chorionic villi**. These increase surface area for absorption. The **maternal part** consists of projections from endometrium. The placenta is a relatively large structure, weighing on average about 600 grams when fully formed and measuring 15-20 cm in diameter and 3 cm thick at the centre.

The foetal blood in the capillaries of the chorionic villi comes in close contact with the mother's blood in the tissues between villi. However, they are always separated by a membrane through which substances may diffuse or be actively transported. Maternal and foetal blood does not normally mix in the placenta or any other place.

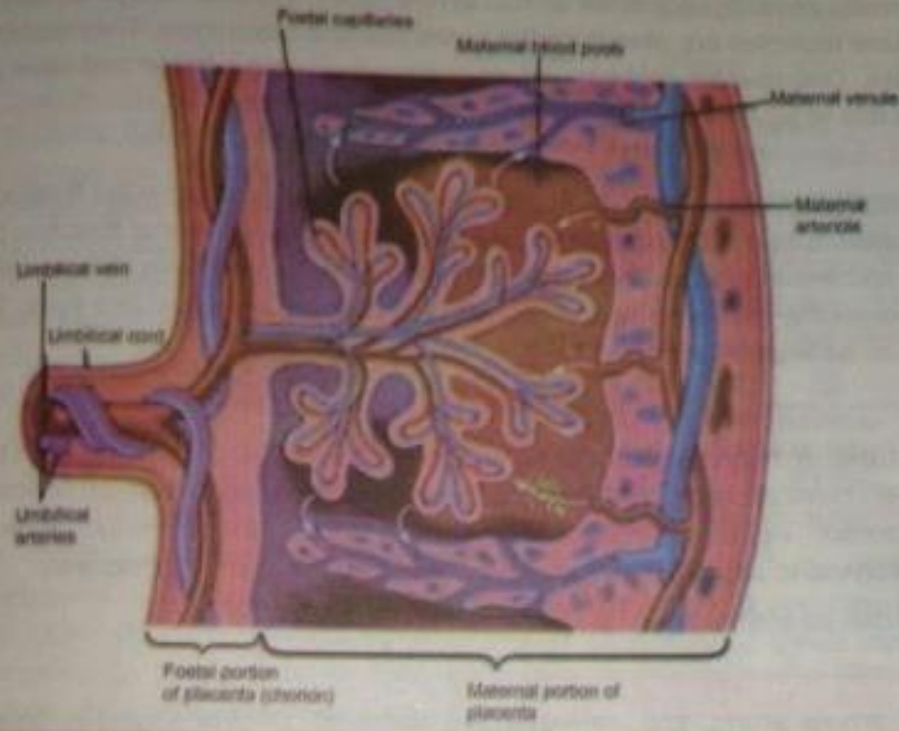


Fig. 21.9 Structure of placenta and umbilical cord

Umbilical cord

As the human embryo grows, the umbilical cord (also called navel string) develops and connects the embryo to the placenta. The umbilical cord is physiologically and genetically part of the foetus and (in humans) normally contains two arteries (the umbilical arteries) and one vein (the umbilical vein). The umbilical vein supplies the foetus with oxygenated, nutrient-rich blood from the placenta. Conversely, the foetal heart pumps deoxygenated, nutrient-depleted blood through the umbilical arteries back to the placenta.

21.3.3 Twins and Quadruplets

A pregnancy of two or more foetuses is called a multiple pregnancy. Multiple foetuses can be the same (identical) or different types (fraternal).

Identical twins

Identical twins or triplets or quadruplets come from a single egg that has been fertilized by one sperm therefore also called **monozygotic**. For unknown reasons, the zygote splits into two or more embryos during the first stage of development. Some identical multiples share the same placenta. However, they usually grow

Fig. 21.10 Identical twins versus fraternal twins

within separate amniotic sacs in the uterus. In rare cases, identical multiples share one amniotic sac. Identical multiples are always of the same sex and blood type. They do not always look exactly alike. One may be right-handed while the other is left-handed and have different hand fingerprint due to exposure of different environment in later life.

Fraternal twins

Fraternal twins or triplets or quadruplets come from multiple eggs fertilized by different sperms therefore also called **dizygotic** or **multiple zygotic**. Fraternal foetuses have separate placentas and amniotic sacs. They can be of different sexes and have different blood types and may look very different from one another, with different coloured hair and eyes. They may also look alike, as siblings often do

21.4 BIRTH AND NURSING

Parturition means birth of the baby. Toward the end of pregnancy, the uterus becomes progressively more excitable, until finally it develops such strong rhythmical contractions that the baby is expelled. There are several factor involved in the onset of this excitation like increased ratio of estrogens to progesterone, foetal hormones and maternal hormones.

21.4.1 Role of Hormones in Controlling Birth

Increased ratio of estrogen to progesterone

Both progesterone and estrogen are secreted in progressively greater quantities throughout most of pregnancy, but from the seventh month onward, estrogen secretion becomes greater than progesterone secretion therefore called increased ratio of estrogens to progesterone. It stimulates the myometrial cells of the uterus to form abundant oxytocin receptors. As a result, the myometrium becomes increasingly irritable, and weak, irregular uterine contractions begin to occur. These contractions are called **Braxton Hicks** contractions or **false labour pains**.

Role of foetal hormones in birth

As birth nears, two more chemical signals cooperate to convert these false labour pains into the real thing. The foetus pituitary gland secretes increasing quantities of **oxytocin**, which might play a role in exciting the uterus. Pituitary gland also secrete ACTH that stimulates the foetal adrenal gland to release **corticosteroids** which affect two regions, first they influence the placenta and cause a decrease in **progesterone** production and second they stimulate the foetal membranes to produce an increased secretion of **prostaglandins**. Both oxytocin and prostaglandins are powerful uterine muscle stimulants, oxytocin causes contraction of the smooth muscles of the myometrium and prostaglandins increase the power of the contractions.

Teacher's Point

Teacher would ask the students to find out twins from their locality and identify the type of twins?

Role of maternal oxytocin in birth

At this point, the increasing emotional and physical stresses activate the mother's hypothalamus, which signals for **oxytocin** release by the posterior pituitary. Together the elevated levels of oxytocin and prostaglandins trigger the rhythmic expulsive contractions of true labour. Once the hypothalamus is involved, a positive feedback mechanism is propelled into action. The greater contractile force causes the release of more oxytocin, which causes greater contractile force, and so on. The release of oxytocin occurs in **"waves"** during labour.

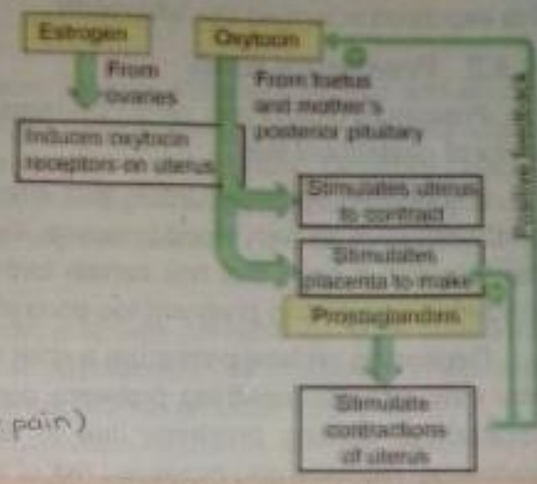


Fig. 21.11 Hormones of birth

Labour process

Braxton Hicks contractions become progressively stronger toward the end of pregnancy; then they change suddenly, within hours, to become exceptionally strong contractions that start stretching the cervix and later force the baby through the birth canal, thereby causing **parturition**. This process is called **labour**, and the strong contractions that result in final parturition are called **labour contractions**. The process of labour has three stages. The first stage is the opening up and thinning of the cervix, ending with complete dilation. The second stage is the expulsion, or delivery, of the baby. Continuous strong contractions force the foetus down and out of the uterus and vagina. The final stage of the labour is delivery of the placenta, which normally follows the baby.

Clamping and cutting of umbilical cord: After the delivery of the baby (second stage of the birth) the umbilical cord is still attached to the baby. The umbilical cord is clamped artificially as early as 1 to 5 minute after the birth of the child. Clamping is followed by cutting of the cord, which is painless due to the lack of any nerves.

Afterbirth: Following the birth of the foetus, usually within 10-15 minutes, the placenta

Breast cancer awareness

Breast cancer affects one in eight women during their lives. Major causes of disease among women are alcohol drinking, obesity, overweight and aversion to breast feeding. Symptoms of breast cancer may include a lump in the breast, a change in size or shape of the breast, and discharge from a nipple. Breast self-exams and mammography can help find breast cancer early, when it is most treatable. One possible treatment is surgery. Other treatments include radiation therapy, chemotherapy, hormone therapy, and targeted therapy. Targeted therapy uses substances that attack cancer cells without harming normal cells. Around 40,000 women dying of it every year in Pakistan

Teacher's Point

The teacher would ask the students to write a brochure to create awareness about breast cancer.

separates from the uterine wall and is expelled by uterine contractions through the birth canal. This expulsion is termed the **'afterbirth'**.

21.4.2 Premature Birth

Premature birth is also called **preterm birth**. It refers to the birth of a baby of less than 37 weeks of gestational age.

It is caused by many factors like: smoking, not getting good prenatal care, having health conditions, such as high blood pressure, diabetes, blood clotting disorders, or infections, being pregnant with a baby that has certain birth defects, being pregnant with a baby from in vitro fertilization and getting pregnant too soon after having a baby.

Depending on how premature a child is when they are born problems experienced by the baby may include: breathing problems due to immature lungs, difficulty in maintaining body temperature, feeding problems due to difficulty in sucking or coordinating breathing and swallowing, jaundice and increased risk of infections.

Premature babies are at higher risk of long-term complications, which may include visual impairment or blindness, hearing impairment. The earlier the baby is born, the more likely that he or she will have these problems.

21.4.3 Lactation / Nursing

Lactation or **nursing** is secretion and yielding of milk by mother after giving birth. The milk is produced by the mammary glands, which are contained within the breasts. The first secretion of the breasts, following birth, is not milk but **colostrum**. This has a yellow colour. It is rich in the protein globulin. It contains little fat and less lactose than the milk. It provides nutrition and contains antibodies, particularly IgA that protects the nursing baby from infections.

Role of prolactin in the production of milk

Rising levels of (placental) estrogen, progesterone, and lactogen toward the end of pregnancy stimulate the hypothalamus to release **prolactin-releasing hormone** (PRH). This in turn stimulates the anterior pituitary gland, which responds by secreting **prolactin**. Prolactin prepares the mammary glands for milk production. True milk production begins after a delay of two to three days of birth. **Serotonin**, a neurotransmitter, is synthesized by the mammary glands. It sends feedback signals to the hypothalamus that slow down prolactin release (and milk production) once the mammary glands are full of milk.

After birth, prolactin release gradually wanes (decrease), and continual milk production depends on mechanical stimulation of the nipples, normally provided by the sucking infant. Mechanoreceptors in the nipple send impulses to the hypothalamus, stimulating secretion of

Teacher's Point

The teacher would ask the students to draw a table to list the events of human development in the first trimester (First, Second and third month), second trimester and third trimester.

PRH. This results in a burst like release of prolactin, which stimulates milk production for the next feeding.

Role of oxytocin in the secretion of milk

The sucking of the baby on the breast stimulates sensory receptors around and in the nipple. Nerve impulses pass from the receptors to the hypothalamus which also stimulate posterior pituitary to release oxytocin via a positive feedback mechanism. Oxytocin causes dilation of milk ducts and thus promotes ejection of milk from the alveoli of the mammary glands. During nursing oxytocin also stimulates the recently emptied uterus to contract, helping it to return to its pre-pregnant size.

As long as milk is removed from breasts, prolactin and oxytocin continue to be released. When nursing is discontinued, the stimulus for prolactin release and milk production ends, and within about one week, the mammary glands lose their capacity to produce milk.

Skills: Analyzing, Interpreting and Communication

- **When oxytocin is involved in the secretion of milk, hypothesize why a new mothers experience cramps in uterus while nursing?**

The stimulus of sucking releases oxytocin. Oxytocin also stimulates contraction of the muscle in the uterus, helping it to recover its normal tone after birth, thus new mother often experience cramps in the uterus while nursing.

Table: 21.1 Comparison of breast-feeding and bottle-feeding

BREAST-FEEDING	BOTTLE-FEEDING
Nutrition	**Nutrition**
Perfect balance of nutrients.	Not efficiently utilized as breast milk.
Contain high level of nutrients.	Nutritional contents depend on proper preparation.
Easily digested and absorbed.	Some babies have difficulty in tolerating certain elements.
ADVANTAGES	**ADVANTAGES**
Baby gets immunity.	Baby does not gets immunity.
DISADVANTAGES	**DISADVANTAGES**
Early breastfeeding may be uncomfortable.	Preparation time varies.
Certain medication can interrupt breastfeeding.	Baby may not tolerate formula well. Always have to carry bottles, formula/mixing items with you.

Science, Technology and Society Connections

Rationalize that nursing is an important bonding time between mother and child as it provides the child with protection by mother's immune system while its own develops.

During the first few weeks of life, a baby's immune system is almost entirely dependent on the mother's breast-milk for immune protection from its environment. The colostrums and mother's milk contains the antibody IgA. When the mother comes in contact with a pathogen, or disease-causing agent, she synthesizes antibodies specific to that agent only. The immune system of a child does not reach its full strength until around the age of five. Demonstrating the unique bond a lactating mother has with her baby, a baby's saliva actually communicates with the mother and literally affects the composition of the milk based on the baby's unique needs. Breast-feeding promotes bonding between mother and baby. Breast-feeding stimulates the release of the hormone oxytocin in the mother's body. "It is now well established that oxytocin promotes the development of maternal behaviour and also bonding between mother and offspring." It is important not to forget the incredible bond that is formed between mother and baby during the months that she is nursing is only the beginning of a deep bond they will share over a lifetime.

21.5 DISORDERS DURING EMBRYONIC DEVELOPMENT

The structural, behavioural, functional and metabolic disorders present at birth are called **embryonic development disorder** or **birth defects**. Following are the four major kinds of birth defects.

1. **Malformation** is a primary structural defect resulting from a localized error of morphogenesis.
2. **Disruption** is specific abnormality that results from disruption of normal developmental processes. It depends on time, not on agent.
3. **Deformation** is an alteration in shape / structure of previously normally formed part.
4. **Syndrome** refers to a group of anomalies occurring together that have a common cause.

21.5.2 Genetic Abnormalities and Spontaneous Abortion

Approximately 50 - 60 % of first trimester and 20% of second trimester miscarriages are due to chromosomal abnormalities within the foetus. The factors leading to this cause include chromosomally abnormal sperm or egg, abnormal cell division of the foetus, and chromosomal abnormalities of mother and / or father.

21.6 POSTNATAL DEVELOPMENT

Further development of a young one after birth is called **postnatal development**.

Stages of Postnatal Development

The five life stages of postnatal development are:

(i) neonatal (ii) infancy (iii) childhood (iv) adolescence or puberty (v) adulthood or maturity

Teacher's Point

The teacher would ask the students to make a list of cancer hospitals in Pakistan.

Skills: Analyzing and Interpreting

* **Using knowledge about the postnatal growth rates of brain and jaw; interpret why a six-month old baby has the same jaw-skull proportion at the time of birth.**

At the time of birth, the 8 bones that make up the cranium are not yet fused together. This means that the skull can flex and deform during birth, making it easier to deliver a baby through the narrow canal. The individual plates of the bone fuse together after about 18 months to form the adult skull.

CANCER (Extra reading material)

Cancer is a class of diseases characterized by out-of-control cell growth. There are more than 100 different types of cancer. Most cancers are named for where they start. For example; lung cancer starts in the lung, and breast cancer starts in the breast. Cancer harms the body when altered cells divide uncontrollably to form lumps or masses of tissue called tumours (except in the case of leukemia where cancer prohibits normal blood function by abnormal cell division in the blood stream). Tumours can be benign or malignant. Benign tumours aren't cancer while malignant ones are. Cells from malignant tumours can invade nearby tissues. They can also break away and spread to other parts of the body. The spread of cancer from one part of the body to another is called metastasis. It is a serious condition that is very difficult to treat. Cancer is often treated with some combination of radiation therapy, surgery, chemotherapy, and targeted therapy.

The **neonatal** period extends from birth to one month. **Infancy** begins at one month and continues to two years of age. **Childhood** begins at two years of age and lasts until adolescence. **Adolescence** begins at around 12 or 13 years of age and ends with the beginning of adulthood. **Adulthood**, or maturity, includes the years between ages 18 to 25 and old age. The process of aging is called **senescence**.

21.6.2 Allometric Growth

Varying rates of growth for different parts of the body during development, resulting in a change of shape or proportion is called **allometric growth**. As a result of simultaneous changes in patterns of growth and development, relative proportions of various structures in human changes. The size of the head in human newborn is large in proportion to the rest of the body. As a human grows and matures, its tarsus (the bones forming the parts of the foot), hands and legs grow more rapidly than head. The brain and skull grow very rapidly after birth, reaching 90% of adult size soon after the age of 6 years. The lymphatic system also develops early. Finally the reproductive systems grow very slowly in childhood. Maximum growth of these organs occurs during puberty.

21.7 AGING

Aging can be defined as the progressive changes over time, leading to loss of physiological function and eventually death. Aging can also be defined as negative physiological changes in our body. The study of aging is called **gerontology**.

21.7.1 Factors Responsible for Aging

There are many theories about what causes aging. Two of these, the genetic and extrinsic factors are considered here.

Genetic factor

Several evidences have been discovered that show aging has a genetic basis. Experiments have shown that human cells will divide less than 100 times outside the body. This is because of short length of **telomeres**, the specific nucleotide sequences at the end of chromosomes. When we are conceived our telomeres are full length but each time a cell divides, the length of telomeres is decreased (like the way lead in a pencil wears down when you write with it). If these sequences are shortened to a critical length, the cell is no longer able to divide. When cells cannot divide, damaged tissues cannot regenerate and we begin to wear out.

Extrinsic factor

Aging is due to years of poor health habits. **Osteoporosis** is the progressive decline in bone density as result fracture is most likely to occur. While there is no denying that osteoporosis occurs as result of aging, certain extrinsic factors are also important. The occurrence of osteoporosis itself is associated with cigarette smoking, heavy alcohol intake and perhaps inadequate calcium intake. A moderate exercise program has been found to slow down the progressive loss of bone mass. If the diet includes fruits and vegetables, good health habits sensible exercise, it will most likely help to eliminate cardiovascular disease, the leading cause of death today.

21.7.2 Signs and Symptoms of Aging

There are different signs and symptoms of aging. Most of these develop gradually and different people possess varying degrees of these signs and symptoms which can be divided into two groups i.e., primary aging and secondary aging.

Primary aging

As aging occurs there are fewer hair follicles, so the hair on the scalp and extremities thins out. Older people experienced a decrease in the number of **melanocytes**, making hair grey and skin pale. In contrast, some of the remaining pigment cells are larger and pigmented blotches appear in skin. The immune system, too no longer performs as it once did and this causes decreased resistance to infection. The other effects of aging are: deafness (in particular lack of ability to hear high notes), fading vision, and reduced ability to adapt to stress. It is important to remember that different individuals can be affected to different extent.

Teacher's Point

The teacher would ask the students to write 'why many changes in your life – both social and physical are dependent on your age'?

Secondary aging

The characteristics that are the result of environmental, life style factors such as diseases, disuse and abuse are considered as secondary aging. The disuse is lack of exercise and abuse includes smoking, obesity, malnutrition and exposure to ultra-violet light.

Science Titbits

Electron shells or orbital, each of which can hold a maximum of two electrons. Thus atoms or molecules with just a single unpaired electron in their orbitals become highly unstable. They are called free radicals (FRS). These FRs can cause oxidation and damage the tissues and nutrients in the body. Thus the formation of FRs caused by oxidation is one of the main causes of aging. Breathing in the environments loaded with polluted particles accelerates the formation of FR in the body. FRs are extremely reactive and alter the structure or any nutrient of tissues or organ that come in their way. Nature has not left us at the mercy of free radicals. This has produced several antioxidants in almost all the types of foods and fruits. Heating food in microwave also create FRs. It's better to use an oven or stove.

(Courtesy: Dr. M. Qudrat-e-Khuda, The Dwan, Islamabad, November 2, 2013)

Science, Technology and Society Connections

List some of the diseases due to aging and what medical science is doing to treat those diseases.

Osteoarthritis: Anti-inflammatory drugs and painkillers are given in the early stages. Later a joint replacement may be necessary.

Osteoporosis: Calcium intake is recommended in the form of medicine or food. Hormone therapy is recommended for most women after menopause if they wish to avoid problem.

Arteriosclerosis: Lifestyle changes, such as eating a healthy diet and exercising, are often the best treatment for atherosclerosis. But sometimes, medication or surgical procedures may be recommended as well.

Activity

1. Identification of the group of vertebrates, through diagrams of different blastula.
2. Identification of the different stages in chick development through observation of prepared slides

Exercise

M.C.Qs.

1. Select the correct answer

(i) How does a zygote differ from an ovum?

(A) a zygote has more chromosomes

(B) a zygote is smaller

(C) a zygote is much larger

(D) a zygote divides by meiosis

(ii) A woman has several miscarriages. Her doctor suspected that a hormonal insufficiency was causing the lining of the uterus to breakdown, as does during menstruation, terminating her pregnancies. Treatment with which of the following might help her pregnancy?

(A) oxytocin

(B) luteinizing hormone

(C) follicle stimulating hormone

(D) progesterone

(iii) In human development, ectoderm cells migrate through the primitive streak to form

(A) endoderm

(B) mesoderm

(C) the chorion

(D) the yolk sac

(iv) The process by which a tissue causes another tissue to differentiate is called

(A) gastrulation

(B) metamorphosis

(C) cleavage

(D) induction

(v) Which of the following has three germ layers?

(A) embryonic disc

(B) blastula

(C) gastrula

(D) trophoblast

(vi) Which of the following consists of both foetal and maternal tissue?

(A) umbilical cord

(B) placenta

(C) amnion

(D) allantois

(vii) Identical twins result from the fertilization of

(A) one ovum by one sperm

(B) one ovum by two sperms

(C) two ova by two sperms

(D) two ova by one sperm

(viii) Which of the following are mismatched?

(A) endoderm; lining of the digestive tube

(B) ectoderm;circulatory system

(C) mesoderm; notochord

(D) mesoderm; reproductive system

Short Questions

2. What is development?
3. What is morula and blastula?
4. What is meroblastic cleavage?
5. List the tissues and organs formed from the three germ layers.
6. What is 'fourth germ layer'?
7. What is Roux-Weismann hypothesis?
8. Describe fertilization and implantation.
9. Describe gestation period and trimesters.
10. What is the basis of the pregnancy test?
11. What are the extra embryonic membranes? Write their functions.
12. Why proper nourishment of mother is imperative during the third trimester of pregnancy?
13. What happens to a sperm and secondary oocyte immediately after fertilization occurs?
14. What are the two very different functions of placenta?
15. How umbilical cord is detached from the baby?

16. How regulation in the end of milk production takes place?
17. What are the four types of birth defects?
18. Describe stages of postnatal development.
19. Rationalize aging as a part of normal development.
20. What are the changes you can recognize as primary aging and secondary aging?
21. List some changes that occur at the cellular level during aging.
22. Define/Describe/Explain briefly:

embryo, embryology, cleavage, blastoderm, morula, blastocoels, blastula, blastocyst, trophoblast, vegetal pole, animal pole, blastodisc, germ layers, gastrula, embryonic disc, primitive disc, epiblast, hypoblast, primitive streak, gastrulation, organogenesis, neuralation, neural tube, neurula, neural crest, foetus, grey crescent, embryonic induction, blastopore, human chronic gonadotropin, gestation period, amnion, yolk sac, allantois, chorion, monozygotic, dizygotic, parturition, Braxton, Hick's contraction, afterbirth, premature birth, lactation, colostrum, prolactin, serotonin, syndrome, allometric growth, gerontology.

23. Write the differences between:
    (a) vegetal pole and animal pole
    (b) blastula and gastrula
    (c) amnion sac and yolk sac
    (d) epiblast and hypoblast
    (e) neurula and rubella
    (f) allantois and chorion
    (g) primary induction and secondary induction
    (h) fertilization and conception
    (i) fertilization and implantation
    (j) monozygotic and dizygotic
    (k) afterbirth and premature birth
    (l) breast feeding and bottle feeding
    (m) neonatal and infancy period
    (n) primary and secondary aging

Extensive Questions

24. What is cleavage? How cleavage can be related with the amount of yolk.
25. What is gastrulation? Explain the events of gastrulation.
26. Define neurulation. State events of neurulation and explain its significance.
27. Describe the formation of:
    (a) Neural crest
    (b) List the structures that are derived from neural crest cells.
28. Describe the experiments of Hans Driesh and Hans Spemann on the role of nucleus cytoplasm in controlling development.
29. What is embryonic induction? Describe the experiments of Hans Spemann and Hilde Mangold on the development of neural tube to explain the embryonic induction.
30. Describe the development in humans in terms of first, second and third trimesters.
31. Describe the structure and role of placenta and umbilical cord.
32. Describe the role of foetal and maternal hormones in initiating labour pains and culminating in the birth of baby.
33. Define the term premature birth and correlate it with the growth phases in the second and third trimester.
34. What is lactation? Describe
    (a) role of prolactin in the production of milk
    (b) roe of oxytocin in the secretion of milk.
35. Describe maternal derived abnormalities.
36. Describe how foetal surgery helps to correct the detected foetal development problems.
37. What is aging? What are the genetic and extrinsic factors of aging?
38. Describe sign and symptoms of aging.
39. Describe the changes that occur during aging at
    (a) system level
    (b) cellular level.
40. State changes that are the result of environmental, life style factors such as disease, disuse and abuse as secondary aging.
41. What changes take place during the aging process?

SECTION 4

Continuity in Life

A sperm entering an ovum

20

REPRODUCTION

After completing this lesson, you will be able to

This is a 07 days unit

- Describe the structures of male reproductive system identifying their functions.
- Explain the principal reproductive hormones of human male and explain their role in the maintenance and functioning of reproductive system.
- Explain the structures of female reproductive system and describe their functions.
- Describe the menstrual cycle emphasizing the role of hormones.
- Describe the causes of female and male infertility.
- Explain that in-vitro fertilization (test tube babies) is one of the methods to solve the problem of infertility.
- Define miscarriage and state its causes.
- Relate miscarriage with abortion.
- Explain AIDS as a worldwide sexually transmitted disease.

Reading

The ability of an organism to produce new offspring of its own type is called reproduction. It is unique characteristic of life as it is not essential for the survival of the individual unlike other characteristics of life; it is however, required for the survival of the species. Without reproduction, the species will cease to exist if all of the members of present generation have died.

20.1 REPRODUCTIVE SYSTEM OF MAN

Human reproduction needs internal fertilization. The reproductive system is unique in two respects. Firstly, the fact that it does not become functional until it is 'turned on' at puberty by the action of sex hormones. In contrast, all other body systems are functional at birth or shortly thereafter. Secondly, the other organ systems of the body exhibit slight differences in male and female while the reproductive system is quite different in male and female.

20.1.1 Male Reproductive System

The main function of male reproductive system is to produce and maintain sperms.

Structure of male reproductive system

The male reproductive system includes: gonads (testes), accessory ducts (epididymis, ductus deferens, ejaculatory duct and urethra), accessory gland (seminal vesicles, prostate gland, bulbourethral glands) and copulatory organ (penis).

Fig. 20.1 Human male reproductive system. (lateral view)

Gonads (Testes)

The testes are male gonads which are situated outside the abdomen within a skin pouch called **scrotum**. Each testis is divided into 250 to 300 lobules. Each lobule contains one to four tightly coiled **seminiferous tubules**.

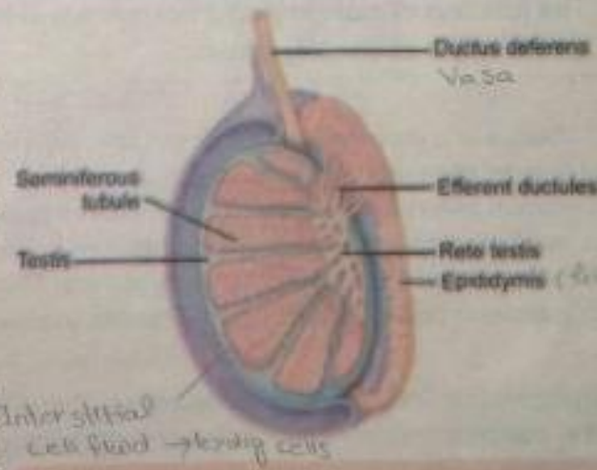


Fig. 20.2 Anatomy of testis

Accessory ducts

Once spermatozoa are produced, they move through the seminiferous tubules and enter a tubular network called the **rete testis** for further maturation. The spermatozoa are transported out of the testis by a series of **efferent ductules**. The **epididymis** is coiled on the outer surface of the testis. The epididymis functions in the transport and storage of the sperms.

Science Titbits

Do you know why testes are relatively located outside the body? The reason is that the process of sperm production, (spermatogenesis) is most efficient around 35°C, two degrees cooler than the body temperature.

Epididymis opens into another duct called **ductus deferens** (sperm duct or vas deferens) which joins with the duct of the seminal vesicle to form the short ejaculatory duct. Each **ejaculatory duct** enters the **prostate gland**, where it empties into the **urethra**.

Urethra is also called **urinogenital duct** as it carries urine as well.

Copulatory organ (Penis)

The human penis consists mainly of tissues that can fill with blood to cause an erection.

Accessory glands

A pair of **seminal vesicles** is located at the junction of sperm duct and ejaculatory duct. The **prostate gland** encircles the urethra just below the bladder. A pair of **bulbourethral gland** (Cowper's gland) is situated at the junction of **ejaculatory duct** and urethra.

Function of male reproductive system

The function of male reproductive system in human reproduction is production of sperms which is also called spermatogenesis

Science Titbits

Semen is a white, sticky mixture of sperm and secretions of accessory glands. The liquid substance in the semen provides nutrients and protection to sperms and acts as a transport medium for sperms. The amount of semen propelled out of the male duct system during ejaculation is about 2–5 ml and there are between 20 to 150 million sperm per ml.

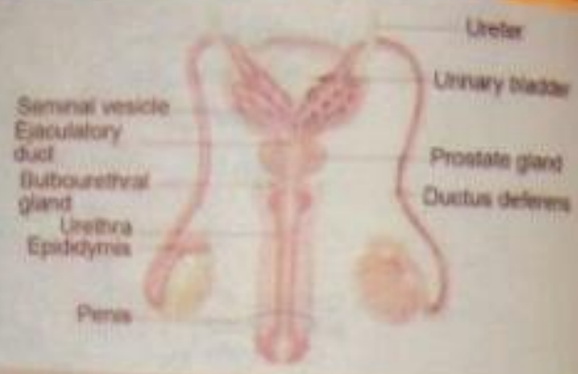


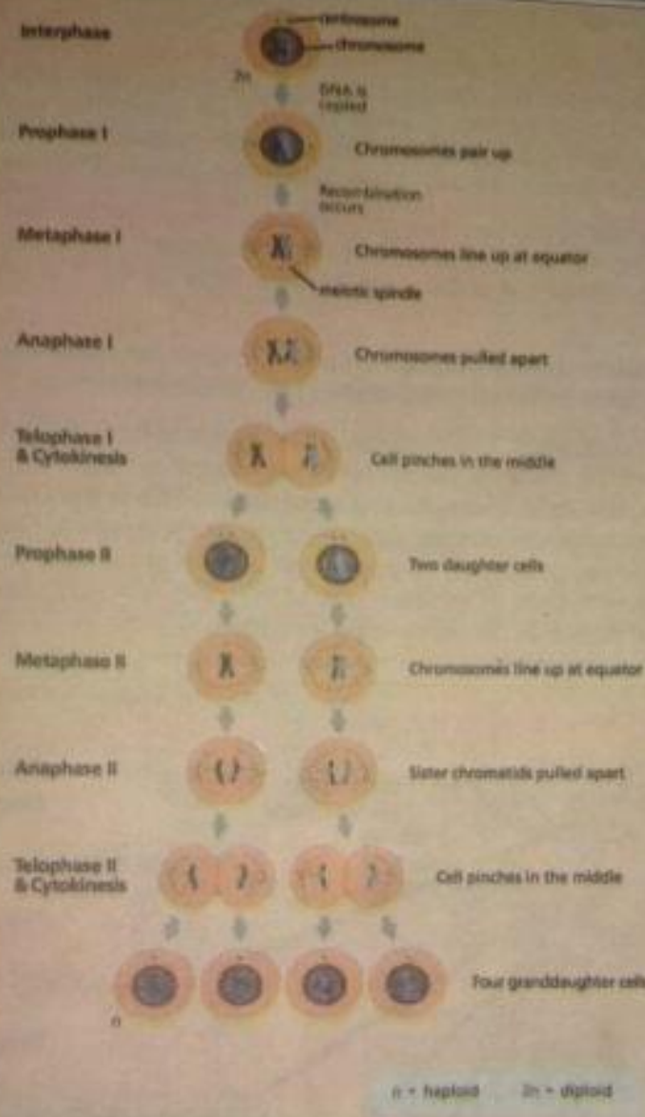
Fig. 20.3 Human male reproductive system. (ventral view)

MEIOSIS (Extra reading material)

Meiosis is a process where a single cell divides twice to produce four cells containing half the original amount of genetic information. These cells are the sex cells – sperm in males, eggs in females. During meiosis one cell divides twice to form four daughter cells. These four daughter cells only have half the number of chromosomes of the parent cell – they are haploid. Meiosis produces sex cells or gameles. Meiosis can be divided into nine stages. These are divided between the first time the cell divides (meiosis I) and the second time it divides (meiosis II).

Meiosis I

1. Interphase: The DNA in the cell is copied resulting in two identical full sets of chromosomes. Outside of the nucleus are two centrosomes, each containing a pair of centrioles. During interphase, microtubules extend from these centrosomes.
2. Prophase I: The copied chromosomes condense into X-shaped structures. Each chromosome is composed of two sister chromatids containing identical genetic information. The chromosomes pair up so that both copies of chromosome 1 are together; both copies of chromosome 2 are together, and so on. The pairs of chromosomes may then exchange bits of DNA in a process called recombination or crossing over. At the end of Prophase I the membrane around the nucleus in the cell dissolves away. The meiotic spindle, consisting of microtubules and other proteins, extends across the cell between the centrioles.



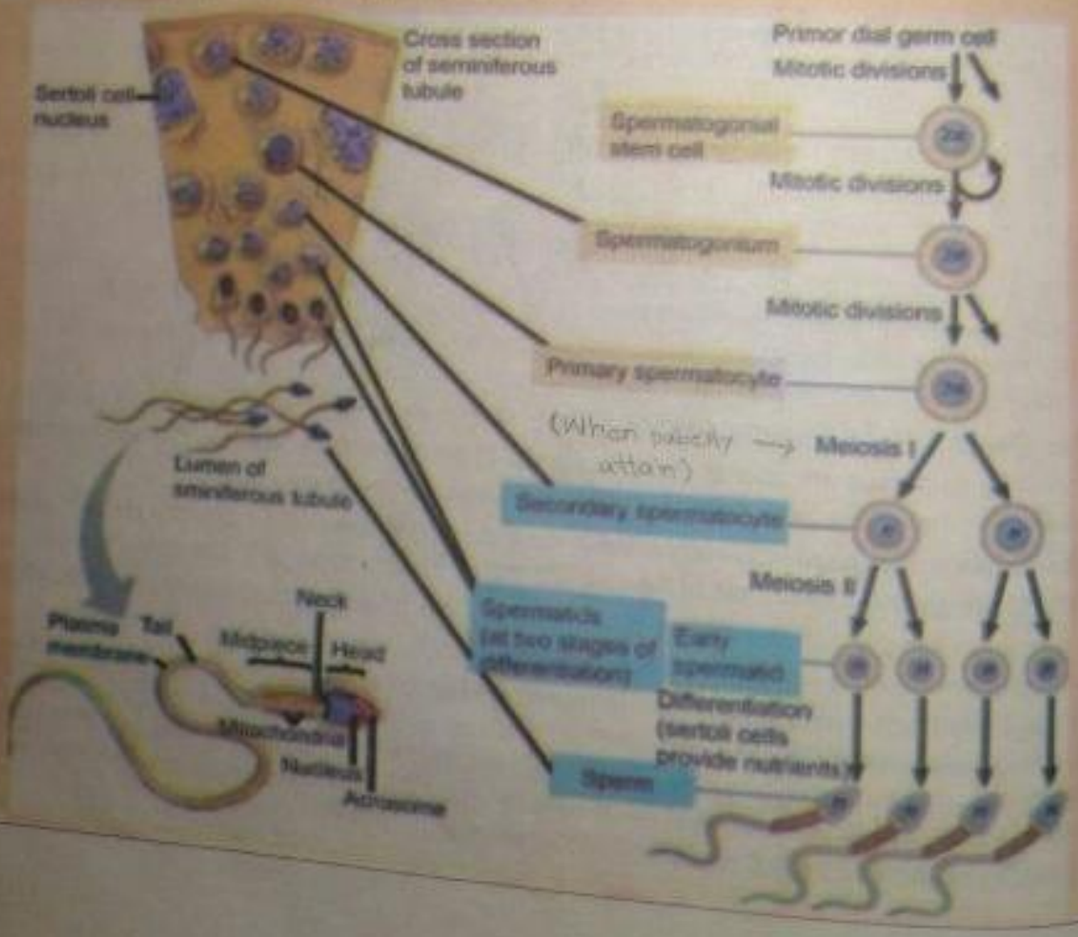
3. Metaphase I: The chromosome pairs line up next to each other along the centre (equator) of the cell. The centrioles are now at opposites poles of the cell with the meiotic spindles extending from them. The meiotic spindle fibres attach to one chromosome of each pair.
4. Anaphase I: The pairs of chromosomes are then pulled apart by the meiotic spindle, which pulls one chromosome to one pole of the cell and the other chromosome to the opposite pole.

   In meiosis I the sister chromatids stay together. This is different to what happens in mitosis and meiosis II.
5. Telophase I and cytokinesis: The chromosomes complete their move to the opposite poles of the cell. At each pole of the cell a full set of chromosomes gather together. A membrane forms around each set of chromosomes to create two new nuclei. The single cell then pinches in the middle to form two separate daughter cells each containing a full set of chromosomes within a nucleus. This process is known as cytokinesis.

Meiosis II

6. Prophase II: Now there are two daughter cells e.g., in human, each with 23 chromosomes (23 pairs of chromatids). In each of the two daughter cells the chromosomes condense again into visible X-shaped structures that can be easily seen under a microscope. The membrane around the nucleus in each daughter cell dissolves away. The centrioles duplicate. The meiotic spindle forms again.
7. Metaphase II: In each of the two daughter cells the chromosomes (pair of sister chromatids) line up end-to-end along the equator of the cell. The centrioles are now at opposites poles in each of the daughter cells. Meiotic spindle fibres at each pole of the cell attach to each of the sister chromatids.
8. Anaphase II: The sister chromatids are then pulled to opposite poles due to the action of the meiotic spindle. The separated chromatids are now individual chromosomes.
9. Telophase II and cytokinesis: The chromosomes complete their move to the opposite poles of the cell. At each pole of the cell a full set of chromosomes gather together. A membrane forms around each set of chromosomes to create two new cell nuclei. This is the last phase of meiosis; however cell division is not complete without another round of cytokinesis. Once cytokinesis is complete there are four granddaughter cells, each with half a set of chromosomes (haploid): in males, these four cells are all sperm cells in females, one of the cells is an egg cell while the other three are polar bodies (small cells that do not develop into eggs).

Spermatogenesis (Extra reading material)

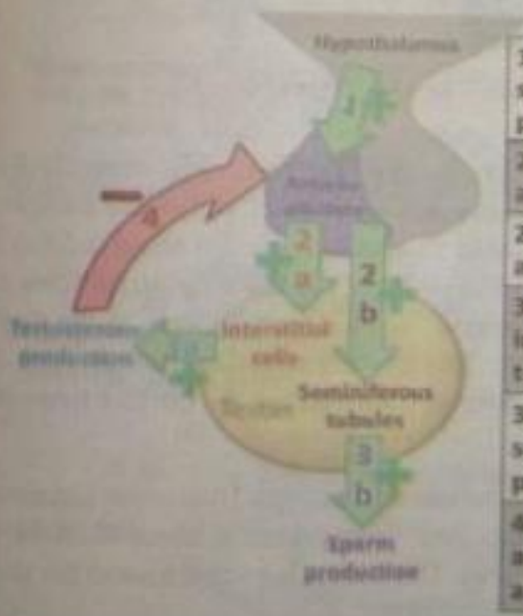


It is the process of sperm formation in males. The process of spermatogenesis takes place in inner epithelium (germinal epithelium) of the seminiferous tubules. During this process, spermatogonia divide by mitosis forming a **primary spermatocyte**.

Each primary spermatocyte undergoes meiosis I, forming two smaller haploid cells called **secondary spermatocytes**. Each secondary spermatocyte after meiosis II produces two daughter cells called **spermatids**. Each spermatid is a round, nonmotile haploid cell which after maturation changes into motile and active **sperms**. During this process a spermatid elongates, sheds its excess cytoplasm, and forms a tail. Every day, a healthy adult male makes about 400 million sperms.

Hormonal control of male reproductive function

Process of spermatogenesis is controlled by hormonal secretions from hypothalamus and pituitary gland. The hypothalamus releases gonadotropin-releasing hormone (GnRH), which controls the release of the anterior pituitary gonadotropins follicle-stimulating hormone (FSH) and luteinizing hormone (LH). FSH stimulates spermatogenesis by stimulating the **sertoli cells** to complete the development of sperms from spermatids. The sertoli cells are elongated cells found in the seminiferous tubules of the testis and they nourish the spermatids. LH stimulates **Leydig cells** to release testosterone. Testosterone causes the growth and development of germinal epithelium to form sperms. **Inhibin** hormone is produced by the **sertoli cells** and serves to control the spermatogenesis at normal rate.



1. Releasing hormones stimulate the anterior pituitary
2a. LH release from anterior pituitary
2b. FSH release from anterior pituitary
3a. LH stimulates the interstitial cells to produce testosterone
3b. FSH stimulates the seminiferous tubules to produce sperm
4. Testosterone inhibits LH and FSH production in the anterior pituitary

Fig. 20.4 Hormonal control of male reproductive system

20.1.2 Female Reproductive System

The reproductive role of the female is far more complex than that of a male. She not only has to produce gametes, but her body must be prepared to nurture a developing embryo for a period of approximately nine months.

Structure of female reproductive system

Female reproductive system consists of gonads (a pair of ovaries), oviducts, uterus and vagina.

Fig. 20.5 Human female reproductive system

Gonads (Ovaries)

Ovaries are female gonads which produce **ova** and release hormones. The paired ovaries flank the uterus on each side and each ovary is held in place within the peritoneal cavity by several ligaments. The **ovaries** are solid, ovoid structures, measure about 3-5 cm long and 2-3 cm wide. Within the ovary are many tiny saclike structures called **ovarian follicles** each of which consists of an immature egg, called an **oocyte**. In adult women, one of the ripening follicles ejects its oocyte from the ovary each month. This event is called **ovulation**. After ovulation, the ruptured follicle is transformed into a glandular structure called the **corpus luteum**.

Oviduct (Fallopian tubes or uterine tube)

The **oviducts** form the initial part of the female duct system. They receive the ovulated **oocyte** and are the site where fertilization generally occurs. Each oviduct is about 10 cm long and transfer developing ovum from ovary towards the uterus. The oocyte is carried toward the uterus.

Uterus

The **uterus** or womb is a hollow, muscular organ, shaped somewhat like an inverted pear. The uterus has three portions: the **fundus**, the **body** and the **cervix**. The oviducts join the uterus just below the fundus and the opening of the cervix leads to the vaginal canal. The wall of the uterus is composed of three layers. The **perimetrium** is the outermost thin covering layer of the uterus. The **myometrium** is the middle thick muscular layer composed of bundles of smooth muscle, which contracts rhythmically during childbirth to expel the baby from the mother's body.

The **endometrium** is the inner spongy lining of the uterine cavity. If fertilization occurs, the young embryo is implanted into the endometrium and resides there for the rest of its development. The main functions of uterus are to receive, retain, and nourish a fertilized ovum. **Cervix** is a narrow entrance to the uterus from the vagina. It is normally blocked by a plug of mucus.

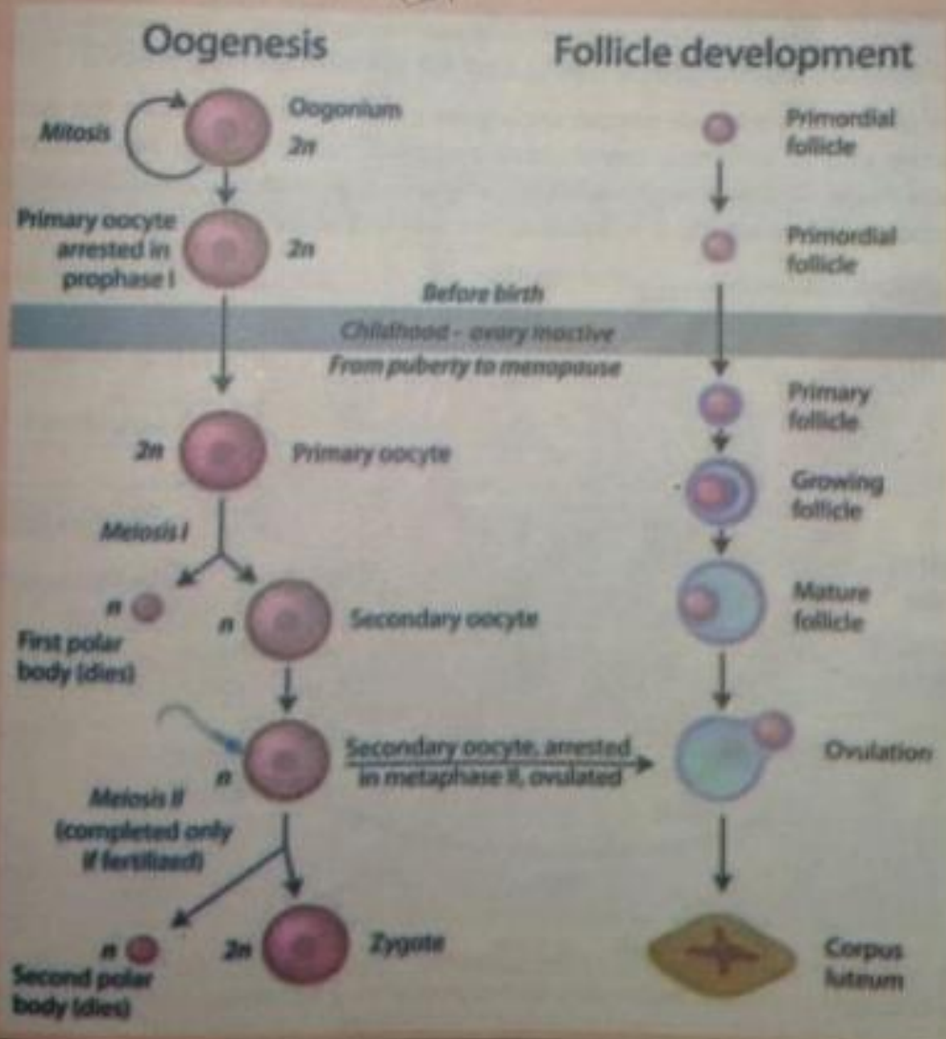
Vagina

The **vagina** is a thin-walled 8–10 cm long tube and extends from the cervix to the body exterior. Vagina is often called the **birth canal** as it provides a passageway for delivery of an infant and for menstrual flow. The urethra is embedded in its anterior wall.

Function of female reproductive system

The organs of the female reproductive system are responsible for production of egg (oogenesis), fertilization, development of embryo and production of female sex hormones.

Oogenesis (Extra reading material)



The process of egg formation in females is called **oogenesis**. This process starts when the individual is at the stage of foetus. In that period the oogonia (diploid stem cells of the ovaries) multiply rapidly by mitosis and then enter a growth phase. Gradually the oogonia are transformed into primary oocytes and become surrounded by a single layer of follicle cells. The primary oocytes begin the first meiotic division, but become arrested late in prophase I and do not complete it. They remain in this state all through childhood; the wait is a long one, at least 10 to 14 years.

At puberty, a small number of primary oocytes are recruited each month, however, only one is selected each time to continue meiosis I, ultimately producing two haploid cells (that are quite dissimilar in size). The larger cell, which contains nearly all the cytoplasm of the primary oocyte, is the secondary oocyte. The smaller cell is called the first polar body. In humans, the secondary oocyte arrests in metaphase II and it is this cell that is ovulated. If an ovulated secondary oocyte is not penetrated by a sperm, it simply deteriorates. But, if sperm penetration does occur, it quickly completes meiosis II (in oviduct), yielding one large ovum and a tiny second polar body. The unequal cytoplasmic divisions that occur during oogenesis ensure that a fertilized egg has ample nutrients for its six- to seven-day journey to the uterus. Without nutrient-containing cytoplasm the polar bodies degenerate and die.

20.1.3 Female Reproductive Cycle and its Hormonal Regulation

The female reproductive system undergoes cyclic events. Therefore the sequence of all reproductive events in female reproductive system is called **female reproductive cycle** or **menstrual cycle.** The female reproductive cycle primarily divided into two phases i.e., **ovarian cycle** (includes those events that occur in ovaries) and **uterine cycle** (includes those events

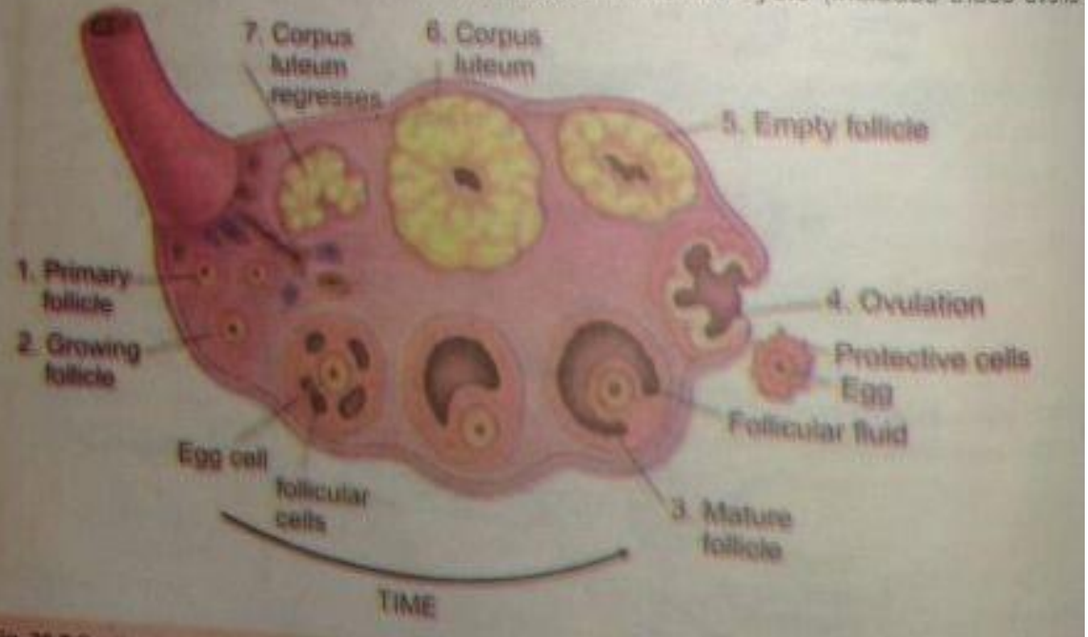


Fig. 20.7 Section through a human ovary showing stages in the development of a mature follicle (or Graafian or vesicular follicle), ovulation and the formation and degeneration of the corpus luteum. Not all the stages would be seen together. The numbers indicate to sequence of the stages.

that occur in uterus). The events of ovarian cycle are very well coordinated with events of uterine cycle by pituitary hormones called **gonadotropins**. Based upon changes and hormonal regulation the cycle can be divided into three phases i.e., menstrual phase, proliferative phase and secretory phase.

Menstrual phase (Days 1 – 5)

In this menstruation phase, the uterus sheds all but the deepest part of its endometrium. The thick, hormone-dependent functional layer of the endometrium detaches from the uterine wall, a process that is accompanied by bleeding for 3–5 days. The detached tissue and blood pass out through the vagina as the menstrual flow. At the beginning of this stage, ovarian hormones are at their lowest normal levels and gonadotropins are beginning to rise. Then FSH levels begin to rise.

Proliferative/pre-ovulatory phase (Days 6–14)

Through the influence of a rise in follicle stimulating hormone (FSH) during the first days of

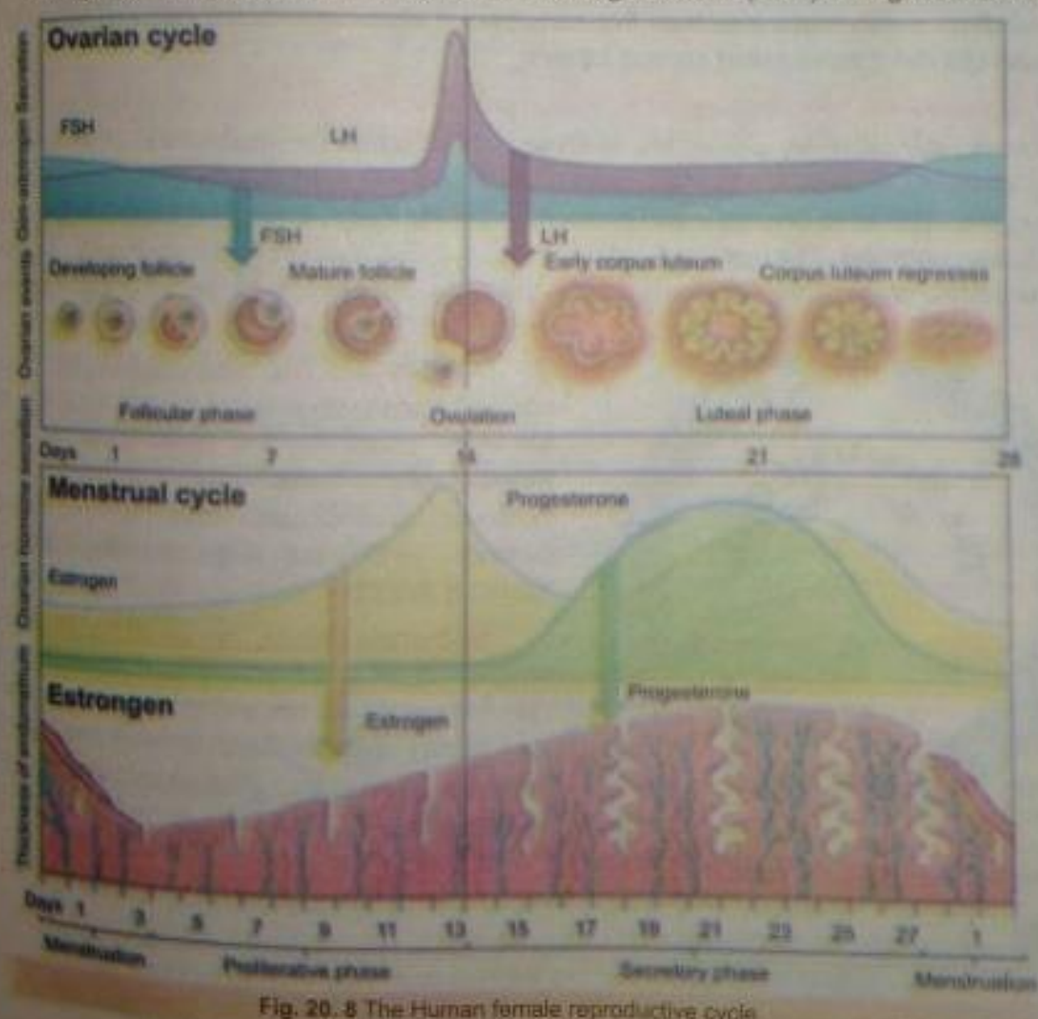


Fig. 20. 8 The Human female reproductive cycle

the cycle, a few ovarian follicles are stimulated. These follicles compete with each other for dominance. As a result all but one of these follicles stop to grow and finally disintegrate (follicle atresia), while one dominant follicle in the ovary continue to mature and becomes **mature follicle** (Graafian or vesicular follisicle), in which **oogenesis** occurs.

FSH also stimulates the graffian follicle to secrete estrogen which in turn governs the vascularization of endometrial lining of uterine wall. Consequently, the endometrium once again becomes velvety, thick and well vascularized. Normally, cervical mucus is thick and sticky, but rising estrogen levels cause it to thin and become crystalline, forming channels that facilitate the passage of sperm into the uterus.

Estrogen has negative feedback upon FSH, therefore, as the concentration of estrogen rises the level of FSH falls. This is a signal for anterior pituitary to release **LH**, at the end of the proliferative stage (day 14) in response to the sudden release of **LH** from the anterior pituitary causes the release of developing egg from the mature follicle into the oviduct, the event is known as **ovulation**, which takes less than five minutes. LH also converts the ruptured follicle to a yellowish glandular mass called **corpus luteum**.

Secretory/post-ovulatory phase (Days 15–28)

During the secretory phase, the endometrium prepares for implantation of an embryo. Rising levels of progesterone from the corpus luteum act on the estrogen-primed endometrium, causing the arteries to elaborate and converting the functional layer to a glandular secretory layer (uterine glands). The uterine glands enlarge, coil and begin secreting nutritious glycogen into the uterine cavity. These nutrients sustain the embryo until it has implanted in the blood-rich endometrial lining.

Fig. 20. 9 The Human female reproductive cycle

If fertilization has not occurred, the corpus luteum begins to degenerate toward the end of the secretory phase as LH blood level declines. Progesterone levels fall, depriving the endometrium of hormonal support and endometrial cells die, setting the stage for menstruation to begin on day 28.

In human beings, menstrual cycle ceases around 50 year of age and it is termed as menopause. Cyclic menstruation is an indicator of normal reproductive life of females and extends between menarche (first menstruation) and menopause.

Teacher's Point

The teacher would ask the students to answer that which hormone is at its peak during ovulation.

20.2 DISORDERS OF REPRODUCTIVE SYSTEM

Infertility cannot be defined precisely because there are varying degrees of infertility. A useful working definition is the failure to achieve pregnancy.

20.2.1 Causes of Male Infertility

The common causes of male infertility are azoospermia, oligospermia, sperm deformities and autoimmune disorder.

a) **Azoospermia:** Azoospermia is the state of having no sperms. It may be caused if sperm ducts are blocked due to infection, injury, etc. The blockage may be congenital (dating from birth).

b) **Oligospermia:** The sperm count below 20 million/ml is called oligospermia.

c) **Sperm deformities:** The changes in shape of sperms are called sperm deformities.

d) **Autoimmune disorder:** In some individuals, the infertility is probably due to an immune response by the male to its own sperms. Antibodies are made which attack the sperm and reduce sperm count.

Science, Technology and Society Connections

Realize the effects of endocrine disrupting contaminants on the reproductive abilities.

Some chemicals, both natural and man-made, can interfere with endocrine glands and their hormones or where the hormones act - the target tissues. These chemicals are called endocrine disrupting contaminants (EDCs) e.g., DDT. The presence of EDCs in our environment raises risk of some human reproductive disorders and some cancers which could be related to disturbance of the endocrine system. EDCs can act in a number of ways in different parts of the body, they may: (a) reduce the production of hormones in endocrine glands (b) affect the release of hormones from endocrine glands, (c) copy or counteract the action of hormones at target tissues, or (d) speed up the metabolism of hormones and so reduce their action.

20.2.2 Causes of Female Infertility

The common causes of female infertility are: failure to ovulate, blocked fallopian tubule, uterus damage, cervical mucus defect and endometriosis etc.

a) **Failure to ovulate:** Sometimes the hypothalamus or pituitary gland fail to produce hormones normally, with the result that either no follicles develop (lack of FSH) or egg release is affected (lack of LH).

b) **Blocked oviduct:** In some females the infertility is due to the diseases causing blockage of fallopian tube. It may be due to infections.

c) **Uterus damage:** Fibroids are benign (non-cancerous) tumours that grow from the walls of the uterus, can cause infertility.

d) **Cervical mucus defect:** During ovulation, mucus in the cervix becomes thinner so that sperm can swim through it more easily. If there is a problem with the mucus, it can make it harder to conceive.

Science Tidbits

Gynaecology is the speciality of medicine concerned with dysfunction and diseases of the female reproductive system. **Obstetrics** is the speciality dealing with pregnancy and childbirth. A physician specialises in both obstetrics and gynaecology.

e) **Endometriosis:** It is a condition where small pieces of the endometrium, start growing in other places, such as the ovaries. This can cause infertility because it becomes difficult for an egg to be released and become implanted into the womb (uterus).

20.2.3 Treatment of Infertility

A number of treatments for infertility are available e.g., surgical, hormone treatments, in vitro fertilization etc.

In Vitro Fertilization (Test-tube baby Technique)

In vitro (glassware) fertilization (IVF), means fertilization outside of the female body. IVF is the most effective types of assisted reproductive technology. It is often used when a woman's fallopian tubes are blocked or when a man produces too few sperm. This is commonly known as the **'test-tube baby'** technique. The technique involves fertilizing one or more eggs outside the body and then transferring the fertilized eggs, known as **'pre-embryos'**, back into the uterus i.e., **embryo transfer**.

In this procedure, the ovary is stimulated with a drug, (having fertility FSH) to produce several eggs. Eggs can be collected from the follicle by sucking out the fluid contents of mature follicles with a fine hollow needle which is inserted through the abdominal wall under general anaesthetic.

Sperms are collected from the male partner and washed in a culture fluid to remove seminal fluid. About 100,000 healthy sperms are added to each egg about six hours after egg collection. This is done in a glass dish or tube. The fertilized eggs are grown for about two days, after which they are usually at 2 to 8 cell stage. After examination under the microscope, the embryo are

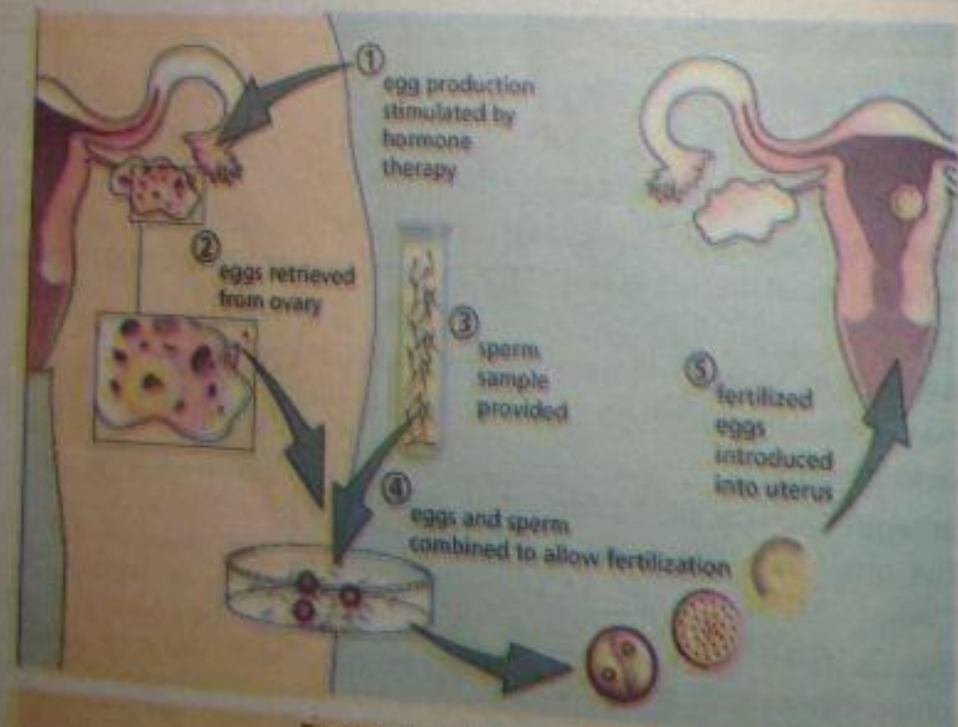


Fig. 20.10 In vitro fertilization

transferred through the cervix into the uterus using a fine plastic tube. She is subsequently treated with progesterone to promote favourable uterine environment for implantation of the embryos.

20.2.4 Miscarriage

It is the act of giving birth spontaneously to a foetus too premature to survive, usually before the 28th week of pregnancy.

Causes of miscarriage

The embryo may implant near the cervix. In this condition, as the placenta grows it may extend partially or completely across the internal cervical opening. As the foetus and placenta continue to grow and uterus stretches, the region of the placenta over the cervical opening may be torn and haemorrhage may occur. Secondly, the normally positioned placenta may tear away from uterine wall accomplished by haemorrhaging. Both of these conditions may result in miscarriage and also be life threatening to the mother. Remember, the live birth which takes place before normal time (EDD or expected date of delivery) is called **pre mature birth**. It is not miscarriage.

Miscarriage versus abortion

The term **abortion** is usually referred to the induced premature termination of a pregnancy. Medically speaking a miscarriage is a **natural abortion** or **spontaneous abortion**.

Science, Technology and Society Connections

Become aware of the ethical implications of abortion.

Abortion is one of the most controversial issues associated with reproduction. It raises ethical issues. The commonly raised ethical issues are:

1. Abortion could be regarded as murder. Christians and Muslims believe that the soul is independent of the body and that it enters the body at the moment of conception.
2. Extra children may impose several financial stresses on an existing family.
3. Many abortions are carried out on foetuses with disabilities such as thallasaemia, Down syndrome etc.

20.3 SEXUALLY TRANSMITTED DISEASES (STDs)

Sexually transmitted diseases are contagious diseases caused by pathogens that are passed from one human to another by sexual contact. The STDs caused by viruses and bacteria. Gonorrhoea and syphilis are caused by bacteria.

AIDS - A Worldwide Sexually Transmitted Disease

AIDS is one of the most serious deadly diseases in human history. AIDS is caused by the human immunodeficiency virus (HIV). HIV destroys helper T-lymphocyte which is the major component of human immune system; therefore the people with HIV begin to get serious infections. The main cause of HIV transmission is that it is a worldwide sexually transmitted disease. As per HIV fact sheet November 2016 released by www.unaids.org/en/resources, there

Teacher's Point

The teacher would ask the students to write a brief research paper on 'sexually transmitted diseases. Students may use library or Internet.

are 36.7 million people living with HIV or AIDS worldwide in 2015. About 35 million people have died from AIDS-related illness since the start of the epidemic till 2015. Every year since 2010, around 1.9 million adults have become newly infected with HIV.

Science, Technology and Society Connections

List the measures that can help to prevent transmission of HIV.

HIV virus is transmitted by exchange of body fluids from infected person to the uninfected. The measures that can help to prevent transmission of HIV are:

1. Refrain from sexual activity and follow Islamic principle of life.
2. Routine testing for HIV can prevent transmission through blood transfusion.
3. Ask the barbers to always use new blades and towels, preferably make shave at home yourself.
4. Use of disposable syringes can avoid transmission of virus by needle.

Activity

Examination of the prepared slides of histology of ovaries and drawing its structure.

Review Questions

1. Select the correct answer

(i) Sertoli cells are found in
- (A) seminiferous tubules
- (B) seminal vesicle
- (C) between interstitial cells
- (D) epididymis

(ii) Fertilization of the ovum normally occurs:
- (A) in distal part of oviduct
- (B) in proximal part of oviduct (anterior)
- (C) along the uterine wall
- (D) successfully in vagina

(iii) Embryo implants in the _____ of the uterus
- (A) perimetrium
- (B) myometrium
- (C) endometrium
- (D) cervix

(iv) Spermatozoa are stored prior to emission and ejaculation in
(A) epididymis
(B) seminal vesicle
(C) urethra
(D) prostate gland

(v) The cervix is a portion of
(A) ovary
(B) vagina
(C) uterus
(D) fallopian tube

(vi) If pregnancy is established, ovulation and menstruation .....throughout gestation period.
(A) remain continued
(B) remain stopped
(C) cannot be affected
(D) none of them

(vii) On which date is a woman most likely to ovulate if the first day of menstrual loss was first March?
(A) 5 March
(B) 14 March
(C) 20 March
(D) 28 March

(viii) If ruptured mature follicle is degenerated without forming corpus luteum, which of the following is expected?
(A) ovulation will not occur
(B) menstruation will not occur
(C) pregnancy is established
(D) none of them

(ix) How does a zygote differ from an ovum?
(A) A zygote has diploid number chromosomes
(B) A zygote is smaller
(C) A zygote consists of more than one cell
(D) A zygote is much larger

Short Questions

2. List the structures of male reproductive system.
3. Explain hormonal control of human male reproductive function.
4. List the structures of female reproductive system.
5. List the structure in order, through which a sperm passes on its way from the seminiferous tubules of the testis to the fallopian tubule of the female.
6. What changes occur in ovulation and menstruation during gestation period.
7. Name the three phase of menstruation cycle and mention the characteristic days.
8. What is the role of the corpus luteum in a menstrual cycle?
9. Name sexually transmitted diseases.
10. What are the actions of GnRH, FSH and LH in the human reproductive functions?
11. Why are so many sperms produced in the male and so few ova produced in the female?
12. Why it is necessary for large numbers of sperms to be produced when only one sperm is required to bring about fertilization?
13. Enlist the reasons of human male infertility?
14. Enlist the reasons of human female infertility?
15. What are the ethical implications of abortion?
16. Explain AIDS as worldwide sexually transmitted disease.
17. Define/Describe/Explain briefly:
    puberty, scrotum, seminiferous tubule, reti testis, efferent ductules, ductus deferens, epididymis, urethra, urinogenital duct, seminal vesicles, bulbourethral gland, spermatogenesis, sertoli cell, leydig cell, myometrium, ova, ovaries, ovarian follicles, oocytes, ovolution, corpus luteum, oviduct, gonadotropins, oogenesis, menarche, menopause, infertility, menstrual cycle, ovarian cycle, uterine cycle, azopermia, oligospermia, sperm deformities, autoimmune disorders, endometriosis, in vitro fertilization, miscarriage, abortion, premature birth.
18. Write the differences between:
    (a) human male and female reproductive system.
    (b) spermatogenesis and oogenesis
    (c) primary and secondary spermatocytes.
    (d) sertoli cell and leydig cell
    (e) spermatids and sperms
    (f) perimetrium and endometrium
    (g) primary oocyte and secondary oocyte
    (h) menarche and menopause
    (i) miscarriage and abortion

Extensive Questions

19. Describe the structure of human male reproductive system. Identify the function of each part.
20. Explain the principal reproductive hormones of human male and explain their role.
21. Explain the structures of human female reproductive system. Identify the function of each part.
22. Describe the events of a menstrual cycle and explain its hormonal regulation.
23. What is infertility? What are the causes of human male and female infertility?
24. Explain "in vitro" fertilization with diagram.

19

BEHAVIOUR

After completing this lesson, you will be able to:

This is a 8 days unit

- Define behavior as the series of activities performed by an organism in response to stimuli.
- Explain relationship between stimuli and behaviour.
- Describe the relationship between heredity and behaviour.
- Explain, through examples, the biological rhythms.
- Define innate (inborn) behaviour.
- Describe examples of innate behavior in terms of taxis shown by unicellular organisms and tropism shown by plants.
- Justify reflexes as a type of innate behavior, by giving examples from man and invertebrates.
- Define instincts and justify these as a type of innate behaviour.
- Justify the fact that each species displays its own characteristic instinctive behavior giving example of migration of salmon.
- Define learning and distinguish between learning and innate behavior.
- Define habituation and illustrate it through the example of squirrels' adjustment in a park.
- Explain imprinting by narrating the work of Lorenz.
- Differentiate habituation and imprinting as reversible and irreversible learned behaviors.
- Describe classical conditioning by narrating the work of Pavlov on salivary reflex in dogs.
- Describe instrumental conditioning (trial-and-error learning) by narrating the work of Skinner on rats' learning.
- Describe latent learning, through the example of a rat in a maze with no reward.
- Interpret Kohler's work on chimpanzee's insight learning to justify that reasoning and planning are involved in the insight learning.
- Differentiate between animal aggregations and animal societies.
- Describe social behavior in terms of hostile and helpful interactions between animals belonging to the same species.
- Describe agonistic behavior and relate it with the maintenance of social order in terms of territories and dominance hierarchies.
- Explain territorial behavior by quoting example of the territories of gorillas.

- Explain dominance hierarchy.
- Define altruism and illustrate it through the organization of a honeybee society.

Reading

Behaviour means the responses of an organism to signals from its environment, including those from other organisms. Animal behaviour is the scientific study of everything animals do. The capacity of behaviour is inherited, but most of the inherited behaviour can be modified by experience. Each organism has a set of adaptive behaviours that fits its life style. In this chapter we will consider the nature of behaviour, innate behaviour, learning and social behaviour.

19.1 THE NATURE OF BEHAVIOUR

The entire pattern of responses made by an organism to the stimuli of its environment is called **behaviour**. In other words, what an organism does after being stimulated is part of its behaviour. The ability of an organism to respond against particular stimuli varies from the relatively simple action of the growth of a plant stem towards a light source, to the complex sexual behaviour patterns of territory defence, courtship and mating seen in birds and mammals. In other words, what a person, animal, plant, or any organism does after being stimulated, is part of its behaviour. In order to cause that response, the stimulus must be sensed, processed, and interpreted.

19.1.1 Relationship between Stimuli and Behaviour

Reception of stimuli

Living organisms have sensors (or senses) that detect forms of energy from the world around them and convert the energy into a signal. The organism then processes or interprets the signal from the sensor, resulting in a response or being ignored as not important.

Interpretation and response to stimuli

Signals may be processed in the brain or outside the brain for example, the skin can detect heat. If the heat is interpreted as dangerously high, the person will jerk away from the source of heat. The signal does not have to reach the brain for the interpretation to cause the response in this situation. On the other hand, the nose of a dog senses the odour from a treat being offered. The signal reaches the brain which interprets the smell as something good to eat. The dog then responds by salivating and perhaps begging for the treat. This response is its behaviour to the stimulus.

Different ways of responses to a stimulus

The response to a stimulus can be **positive**, **negative**, or **ignored** as not important. A positive reaction is that in which one wants more or is attracted to the stimulus. A negative reaction is that in which one wants to avoid the stimulus. *Example:* A person laughs after hearing a funny joke, is a positive response or you make a face after smelling a sour odour, is a negative response. The decision to ignore the stimulus is also a kind of response as a dog pays no attention to sounds from the television or your child ignores your order to clean up his room.

19.1.2 Relationship between Heredity and Behaviour

All behaviours depend on nerve impulses, hormones and other physiological mechanisms such as sensory receptors. Therefore **genes** play a role in the development of behaviours because they direct the development of the nervous system. In addition, autonomic responses depend on specific nerve pathways within central nervous system of an organism. These pathways are neural programs and are genetically determined. Even the capacity to learn is inherited.

Members of a species vary in the expression of certain behaviours because of variations in their genes and these behaviours have survival value in some environments. One example of such behaviour is **curiosity**, some organisms are more curious than others and in some settings curiosity is advantageous for survival. Heredity has important role in intelligence, moodiness, impulsiveness, shyness and all other psychological characteristics.

19.1.3 Biological Rhythms

Biological rhythms are cyclic pattern of physiological changes or changes in activity in living organisms that are in response to periodic environmental changes. The internal mechanism by which such a rhythmic phenomenon occurs and is maintained even in the absence of the apparent environmental stimulus is termed as **biological clock**. The exact nature of the internal mechanism, or "biological clock," that controls such rhythms is not understood.

Biological rhythms may show interval of less than 24 hours or less than a month or less than a year. The **circadian rhythms** are 24 hour-cycle shown by physiological processes in all organisms. These include changes in body temperature, blood pressure, and urine production, sleep/wakefulness cycle, patterns of hormone secretion, digestive secretions and levels of alertness. When the rhythm is synchronized with the day/night cycle it is termed as **diurnal** (behavioural activities of day time) or **nocturnal** (behavioural activities of night

Science Tidbits

Biological rhythms are generated in two ways: Some biological rhythms are directly driven by external environmental stimuli are called exogenous e.g. migration of birds. Rhythms driven by an internal biological clock, rather than by an external process, are called endogenous e.g., core body temperature, sleep-wake cycle etc.

time). For example, animals like honeybees and pigeons, are most active during the day light, hence called **diurnal animals** while animals like owl, bats, pigs are most active during the night, thus, called **nocturnal animals**. However, some animals like the fiddler crab, are busiest during the time of dawn or dusk or both are therefore called **crepuscular animals**.

Monthly rhythms include menstrual cycle in women. **Annual cycles**, or **circannual rhythms**, include bird migrations, reproductive activity, and hibernation of animals.

19.2 INNATE BEHAVIOUR (INBORN OR INSTINCTIVE BEHAVIOUR)

Innate behaviour may be defined as behaviours resulting from genetically determined neural programmes that are part of the nervous system at the time of birth or develop at an appropriate point in maturation. Interestingly, these instinctive or **inborn behaviours** are performed in a reasonably complete form, the first time they are exhibited. A human new born, for example, will turn to suckle when touched on the cheek near the mouth. Innate behaviours are important in the survival of specially those animals that have short life span and poorly developed nervous coordination. All **plant behaviours** are innate in nature. Innate behaviours primarily divided into two types i.e., orientation and non-orientation behaviours.

Science Titbits

The circadian rhythm is controlled by the supra-chiasmatic nuclei (or SCN), a specialized area found in the hypothalamus. It takes information on the lengths of the day and night from the retina, interprets it and passes it on to the pineal gland. In response, the pineal gland secretes the hormone melatonin. Secretion of melatonin peaks at night and ebbs during the day.

19.2.1 Orientation Behaviours

When an animal moves or changes its position or alignment relative to points of the specific directions in response to some stimuli, this behaviour is known as **orientation behaviour**. There are two types of animal orientation behaviours i.e., taxis and tropism.

Science Titbits

Kinesis, is a simple change in activity, or turning rate in response to stimulus. Animals will neither move towards nor away from an environment. An example of this is wood lice, which will slow down in favourable conditions, a humid environment, and speed up in unfavourable conditions, a dry environment. This simple behaviour tends to keep the wood lice in humid areas. Since they slowdown in this environment, they tend to stay there.

Taxis

A **taxis** (plural, taxes) is a directional movement toward or away from a stimulus, such as light, chemicals or heat. e.g., *Euglena* shows positive taxis by moving toward dim light but negative taxis by moving away from intense light.

Tropisms

Tropisms are growth movements related to directional stimuli. **Phototropism** is the responses to the light in which shoots will grow towards a source of light but away from the direction of gravity. **Geotropism** is the responses to gravity. If a plant is placed horizontally, its stem will change its direction and grow upwards

and away from gravity. The roots will change their direction of growth to grow vertically downwards towards the pull of gravity. Plant roots also show **hydrotropism** (responses to water).

Fig. 19.1 Geotropism

19.2.2 Non-Orientation Behaviours

In these behaviours animals do not show particular movement in response to stimuli. These are much more complex than orientation behaviour. These include reflexes, and instincts which consist of emotions, sentiments, habits.

Reflexes

A reflex is an automatic, involuntary response to changes occurring inside or outside the body. A particle of food touching the lining of the windpipe will set off a coughing reflex which cannot be suppressed. In the bright light, the pupil will contract. You cannot stop this reflex and you are not even aware that it is happening. Some reflexes, such as blinking your eye, involve the brain; while others such as withdrawing your hand from a hot object, do not necessarily involve brain. Most insects have simple "startle" reflexes triggered by small disturbances as well as more comprehensive "escape" reflexes triggered by larger disturbances.

Instincts

Instincts are unlearned, inherited fixed action patterns of responses or reactions to certain kinds of stimuli. The term instinct refers to innate or inborn behaviour. Since these are genetically programmed behaviour so these are also referred to as **fixed action pattern**. Instincts are complex behaviour patterns which, like reflexes, are inborn, rather inflexible and valuable at adapting the animal to its environment. The entire body participates in instinctive behaviour and an elaborate series of actions may be involved.

Migration of salmon (An example of instinct)

The upriver salmon migration is one of nature's most exciting dramas. The female builds her nest and deposits her eggs. The male then moves alongside and deposits his sperm, over the eggs. This process is known as **spawning**. The male salmon dies within a few days of spawning. The newly hatched fish live the first part of their lives in freshwater and then migrate to the ocean to spend their adult lives which may be as short as 6 months or as long as 7 years.

Teacher's Point

The teacher would ask the students to find out:

1. How are reflexes important in maintaining body's homeostasis?
2. How do migrating animals navigate?

Migration between fresh and saltwater occurs during every breeding season. When they reach sexual maturity, Pacific salmon may swim hundreds, even thousands, of miles to get back to the stream where they hatched. Only a small percentage of salmon reach their spawning grounds due to many reasons. Since salmon do not feed once they leave the ocean, some will die on the way. Thus migration of salmon is an instinct or inborn behaviour as th young ones doing it to perfection without having seen it done.

Fish migration of salmon

19.3 LEARNING

Learning is a change in behaviour resulting from experience. Unlike innate behaviour, learning involves some choice of responses to a given stimulus. Also, learning is not directly controlled by genes as innate behaviour is. Innate behaviour can become a liability if environment conditions change and an animal's condition cannot change to adapt to new conditions.

Learned behaviour can help an animal become better suited to a particular environment or set of conditions. Unlike innate behaviour, learning is more prominent in those organisms that have comparatively long life span and have well developed nervous coordination. Learning behaviours can be grouped into six categories: (a) habituation (b) imprinting (c) classical conditioning (d) trial and error learning (e) latent learning (f) insight learning.

Habituation

In habituation, an animal learns to ignore a repeated, irrelevant stimulus. We see the buffalos or cows in street city or squirrels in the city parks. These animals have learned by repeated harmless encounters that humans are no more dangerous to them and behave accordingly. This is to their advantage. An example of learning by habituation is the one observed in squirrels: when one of them feels threatened, the others hear its signal and go to the nearest refuge. However, if the signal comes from an individual who has caused many false alarms, its signal will be ignored. Habituation is highly adaptive.

Imprinting

Imprinting is a type of learning in which a very young animal fixes its attention on the first object with which it has visual, auditory, or tactile experience and thereafter follows that object. In experiments, animals and inanimate objects have been used. Imprinting has been intensively studied only in birds, especially chickens, ducks, and geese, but a comparable form of learning apparently occurs in the young of many mammals and some fishes and insects. The specific time during which imprinting develops is called the **critical period**. One result of imprinting is the formation of a strong bond between two animals, often a hatching or other newborn animal and its parent.

We can describe the curious process of imprinting by giving an example. In normal life, geese hatch from eggs in the presence of the mother goose. Soon after hatching, the goslings begin to follow their mother about, as she leads from them to suitable areas to feed or protects

Fig. 19.3 Konrad Lorenz with graylag goose

them. In 1930s, **Konard Lorenz** showed that principal imprinting stimulus in graylag (*Anseranser*) is a nearby object that is moving away from the young. When incubator hatched goslings spent their first few hours with Lorenz rather than with a goose, they imprinted with him and followed him from then on. Furthermore, they showed no recognition of their biological mother or other adults of their own species.

Difference between habituation and imprinting as reversible and irreversible learning behaviour

Habituation is the loss of a response to a stimulus after repeated exposure. It is reversible. For example a snail crawling on a sheet of glass retracts into its shell when glass is tapped. After a pause, it emerges and continues moving. A second tap causes retraction again but it emerges more quickly. Ultimately, tapping has no effect and snail ceases to respond. It is reversible learning behaviour because after some time this habituation will vanish and snail again will show same response. Imprinting is learning that is limited to a specific time period in animal's life and that is irreversible i.e., it remains throughout the life.

Classical conditioning

When an animal learns the same response for two different stimuli which are given to the animal simultaneously is called **classical conditioning**. The response is actually for one stimulus but the same response has become developed for the other stimulus. **Pavlov** paired the meat powder with various stimuli such as the ringing of a bell. After the meat powder and bell (auditory stimulus) were presented together several times, the bell was used alone. Pavlov's dogs, as predicted, responded by salivating to the sound of the bell (without the food). The bell began as a neutral stimulus (i.e., the bell itself did not produce the dogs' salivation). However, by pairing the bell with the stimulus that did produce the salivation response, the bell was able to acquire the ability to trigger the salivation response. In technical terms, the meat powder is considered an unconditioned stimulus (UCS) and the dog's salivation is the unconditioned response (UCR). The bell is a neutral stimulus until the dog learns to associate the bell with food. Then the bell becomes a **conditioned stimulus** (CS) which produces the conditioned response (CR) of salivation after repeated pairings between the bell and food.

Fig. 19.4 Experiment of Pavlov

Instrumental conditioning (Trial and Error Learning or Operant learning)

When an animal learns a response to a particular stimulus after many unsuccessful tries is called **trial and error learning** or operant conditioning. The trial and error learning occurs through experience. In the natural environment, animals are faced with naturally occurring awards and punishment and they learn by experience.

The American psychologist **B. F. Skinner** studied conditioning in rats by placing them in a specially designed box (today called a Skinner box fitted with levers and other experimental devices).

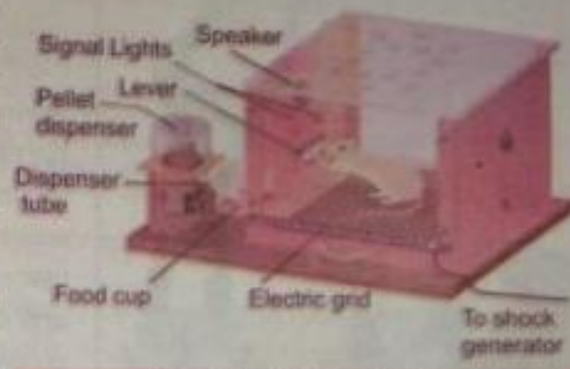


Fig. 19.5 A 'Skinner box'.

Once inside, the rat would explore the box feverishly, running this way and that. Occasionally, it would accidentally press a lever, and a pellet of food would appear. At first, a rat would ignore the lever and continue to move about, but soon it learned to press the lever to obtain food. As the animal learns by chance i.e., trial so this type of learning has been named **'trial and error learning'**.

Latent Learning

When an animal learn a response for a particular stimulus in its routine life without having any punishment or reward, but sometimes, when the animal is particularly exposed to that stimulus, the animal suddenly respond it by quickly recalling the previous experience. This is called **latent learning**.

The American psychologist **K.L. Lashley** used a maze (a network of path). A rat was,put in the maze. The rat explores the maze in order to find the exit. Eventually the rat found the way to exit but it also learned the location of food in the maze. The rat was not hungry at that time so it did not pay attention to the food. Then the same rat was again put in the maze when it felt hungry. The rat because of its previous experience found the food quickly than a rat that has been put without previous exploratory experience.

Fig. 19.6 Maze

Teacher's Point

The teacher would ask the students to give an example how human learn through classical conditioning.

Insight Learning

Insight learning is a much complex type of learning because it requires that an a must respond correctly in first attempt to a particular situation it has never met befo Animals capable of insight learning seem to practice a sort of mental trial and error proce analyzing the possibilities for the solution of problem before setting out to tackle it.

Fig. 19.7 Insight learning

German psychologist **Wolfgang Kohler** wa the first to describe learning by insight, performin extensive experiments on chimpanzees in 192 Kohlar showed that animals must perceiv relationship and manipulate concepts in its mind t solve a problem on the first try. A chimpanzee wa placed by Kohler in a cage containing several boxes and out of reach hung bananas. The chimpanzee solved this problem by stacking the boxes so that it could climb on them to reach the bananas. No past experience provided the chimpanzee with his 'plan of attack'. Somehow the chimpanzee was able to 'think' out of the fact that putting the boxes on top of each other would provide a means for reaching the bananas. He used insight to solve the problem. This type of behaviour is in direct contrast to trial and error learning. Although both forms involve experience, insight or reasoning enables the animal to 'path' other than randomly trying several approaches. In fact this learning is common only among primates (humans, apes, monkeys) and in some other mammals and a few birds.

Skills: Analyzing and Interpreting and Communication

- **Relate different examples of learning behaviours of human with habituation, conditioning, latent learning and insight learning.**

Habituation: In a short time after human dresses in clothing, the stimulus clothing creates disappears from our nervous systems and we become unaware of it. In this way, habituation is used to ignore any continual stimulus.

Conditioning: You almost certainly have many conditioned responses yourself. One that is common in man and particularly easy to demonstrate, is the increased rate of flow of saliva in response to the sound or even the thought of words like lemon, vinegar, tamarind and sweets etc.

Latent learning: If a person knows the route to go to any destination he can reach quicker than a person who does not know the route to reach that place.

Insight learning: You know how reasoning plays a part in modifying human behaviour. For instance you feel hungry and want to go to college cafeteria. You look at the watch and decide not to go yet. Your capacity to reason and the time had shown by your watch make you aware of the fact that (probably) that cafeteria will not be opened for half an hour. Another example of human insight learning is the ability to solve mathematical problems.

Table 19.1: Difference between innate and learned behaviour

Innate behaviour	Leaning behaviour
It comes natural or by default	It should be developed with experience.
It cannot be modified.	It can easily be modified.
It may or may not have the direct involvement of the brain	It has the direct involvement of the brain
These are more common in the animals having short life span.	These are more common in the animals having long life span
It is economical as animals require no time to adapt them.	It is not economical as animals require more time to adapt them

19.4 SOCIAL BEHAVIOUR

Many species of insects and most vertebrates show a variety of (hostile or friendly) group behavioural activities associated with numbers of individuals living together. This is known as **social behaviour**. The cooperation achieved as a result of social behaviour has adaptive significance. It increases the efficiency and effectiveness of the species over that of the other species.

19.4.1 Hostile and Helpful Intraspecific Interaction

In bees hive, hostile interaction is seen among the worker bees. Old worker bee which is unable to perform its duties in hive is killed by other worker bees. On the other hand helpful interaction is found among these bees as different bees have specific duties to perform over all functions of the hive. Worker bees collect nectar and transform it into honey, drones are specific to perform the duty to fertilize the eggs and queen lays eggs.

19.4.2 Animal Society and Aggregation

Organisms living together in organized groups are said to live in societies. A **society**, or social group, is a group of individuals of the same species that interact with each other and influence each other's behaviour in different ways. This behavioural interaction is a key characteristic of society. A hive of bees, a pack of wolves and a school of fishes are examples of societies. Characteristics of a well-organized society include cooperation and division of labour among animals of different sexs, age groups or castes. Such as some members are specialized for finding food, reproduction, rearing and defence

A simple **aggregations** is a group of animals that may be together but do not interact behaviourally. For example huge flocks of birds of many species roosting together in trees are aggregations or a group of zebras, buffaloes are not societies.

19.4.3 Agonistic Behaviour

Agonistic behaviour includes a variety of threats or actual combat that settles disputes between individuals in a population. Agonistic interaction uses a great deal of energy, may cause

injury. Agonistic behaviour is displayed to maintain social order such as territoriality and dominance hierarchy.

(a) Territorial behaviour

Territoriality is the defence of an area by an organism or group of organisms against organisms of the same or different species. There are threat displays between owners of adjacent territories. Despite the apparent conflict and aggression associated with territory formation, actual fighting, which would detrimental to the species, is rare and is replaced by threats, gestures and postures. Once a territory is established through aggressive interactions, relative peace prevails as boundaries are recognized and respected. Territorial behaviour is seen in animals as diverse as worms, arthropods fish, birds and mammals.

Territorial behaviour in mountain gorillas: Gorillas are highly social, relatively non-territorial and live in groups (called troops). The oldest and strongest adult male (called the silverback) is usually the dominant one of the troop and has exclusive breeding rights with the females. Adolescent females transfer to a different troop once they reach about eight years of age. Adolescent males, on the other hand, usually remain in the troop until they can leave and establish a new troop on their own as the silverback. Although gorillas typically aren't aggressive, they do exhibit territorial behaviour by standing upright on their bottom two legs and pounding their chests in order to intimidate whatever threat they have been given. These gestures, however, are more for show and aren't usually violent.

Fig. 19.8 Mountain Gorilla

(b) Dominance hierarchy

In each animal establishes a rank that determines its access to resource. The dominant individuals obtain most access to the resources needed for reproduction, including food, space and mates. In some animals, dominance is a simple function of aggressiveness, which is itself often influenced by sex hormones. An example is pecking order or social hierarchy of chicken. A society of chicken evolves a hierarchy as the result of "pecking" each other. If several hens unfamiliar to one another are put together, they respond by chasing and pecking one another. Eventually, they establish a clear "peck order". The alpha or first ranked hen in the peck order is dominant. She is not pecked by any other hens and can usually drive off all others by threats rather than actual pecking. The alpha hen also has first access to resources such as food, eater and rooting sites. The beta or second ranked hen similarly subdues all others except the alpha and so on down the line to the omega or lowest animal.

Science Titbits

A **pheromone** is a secreted or excreted chemical factor that triggers a social response in members of the same species. Pheromones are chemicals capable of acting outside the body of the secreting individual to impact the behaviour of the receiving individual. There are alarm pheromones, food trail pheromones, sex pheromones, and many others that affect behaviour or physiology.

(c) Altruism

Altruism means the principle of living for the interest of other. Altruism refers to any behaviour that endangers an individual organism or reduces its reproductive success that benefits other members of its species. Altruistic behaviour is common in animal kingdom e.g., female worker bees forgo reproduction and devote their lives to raising the offspring of the hive queen. Here we will illustrate altruism through the organization of a honey bee society.

A honeybee colony is made up of three types of castes: the queen, the workers and the drones. The **queen** lays eggs from which all other bees develop. The **drones** are the male bees that fertilize the eggs of the queen. The drones develop from the unfertilized eggs and are haploid. The **workers** are females that develop from fertilized eggs. Any fertilized egg can develop into either a queen or a worker depending upon how the egg is housed and fed. The workers perform all the tasks of the hive except for mating and laying eggs. They take flights away from the hive to forage food. At the time of mating, many drones follow the queen out of the hive, following her pheromone trail, and mate in flight. After mating, the drones die. Drones that do not mate have no better fate, because the workers pinch them, sting them and then throw them out of the hive as the winter approaches.

Fig. 19.9 Honeybees society

The queen controls the bees with a chemical called **pheromone**. The bees ingest this chemical by licking the queen and then pass the substance around from one another as they pass food. Pheromone renders the worker bees sterile. Workers make half a dozen or more new queen cells in which replacement queens begin to develop. The old queen and a swarm of females and male drones leave to establish a new hive. The first queen to emerge may kill the other candidate queen and assume rule or may create another swarm and leave to establish another hive. Sterile female workers are prevented from producing offspring, yet they spend their lives looking after their brothers and sisters.

Science, Technology and Society Connections

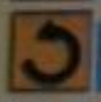
Rationalize why the marine snail, Aplysia, has proved very helpful in the studies of neurobiology and of behaviour pattern.

Aplysia californica is a marine snail. It's simple nervous system, consists of just a few thousand of large neurons. Despite its simple nervous system, however, it is capable of a variety of non-associative and associative learning tasks, including sensitization, habituation, classical and operating conditioning. The nice feature that makes *Aplysia* so attractive for neurobiologists is its large brain cells (neurons). The cell body of one neuron can measure up to 1mm in diameter, which makes it relatively easy to study the physiology of these cells to find out how they accomplish learning.

Activity

Observation of a spider's web and recording the instincts by providing it various stimuli.

Exercise



M.C.Qs

1. Select the correct answer

(i) The responses of an organism to signals from its environment are its
- (A) behaviour
- (B) culture
- (C) releaser
- (D) motor programs

(ii) A form of learning in which a young animal forms a strong attachment to a moving object (usually its parents) within a few hours of birth is
- (A) classical conditioning
- (B) insight learning
- (C) imprinting
- (D) habituation

(iii) In an insect society, such as the honeybee society
- (A) the division of labour is based on biologically determined castes
- (B) all adult members share labour equally
- (C) all adult members have the opportunity to reproduce
- (D) reproduction is altered seasonally among adults

(iv) Working out a mathematics problem is an example of:
- (A) insight learning
- (B) an involuntary act
- (C) an instinct
- (D) reflex

(v) An animal learns to ignore a repeated, irrelevant stimulus. This behaviour is
- (A) classical conditioning
- (B) imprinting
- (C) insight learning
- (D) habituation

(vi) The benefits of territoriality include
- (A) rights to defend a home range
- (B) increased reproductive success
- (C) monogamy
- (D) pair bonding

(vii) When *Dorsophila* were exposed to a particular odor and electric shock at the same time, they started to avoid the odor. This is an example of_____.

(A) classical conditioning

(B) reasoning

(C) imprinting

(D) habituation

Short Questions

2. What are the ways of responses to a stimulus?
3. What is re relationship between heredity and behaviour?
4. What are biological rhythms? How biological rhythms are important to man?
5. What is innate behaviour? Give example.
6. What is learning behavior?
7. Relate examples of learning behaviour of human with: (a) habituation (b) conditioning (c) latent learning (d) insight learning.
8. Write the differences between innate behaviour and learning behaviour.
9. What are the differences between animal society and animal aggression?
10. What is altruism? Give example.
11. Define/Describe/Explain briefly:

    Behaviour, biological clock, circadian rhythms, diurnal anmals, nocturnal animals, monthly cycle, annual cycle, innate behaviour, taxis, tropism, phototropism, geotropism, hydrotropism, reflexes, instinct, fixed action pattern, spawning, dances of bees, habituation, imprinting, classical conditioning, trial and error learning, latent learning, insight learning, social behaviour, society, territoriality, altruism, pheromones.

12. Write the differences between:

    (a) taxis and tropism

    (b) geotropism and hydrotropism

    (c) reflexes and instinct

    (d) habituation and imprinting

    (e) latent learning and instinct learning

Extensive Questions

13. Describe relationship between stimuli and behavior.
14. Describe relationship between heredity and behavior.
15. Explain biological rhythms.
16. Describe orientation behaviour.
17. What are instincts? How can you justify instincts as a type of innate behaviour?
18. Describe migration of Salmon as instinct behaviour.
19. Describe imprinting in young ducks. How is this imprinting adaptive?
20. Describe classical conditioning by narrating the work of Pavlov on salivary reflex in dog.
21. Describe trial and error learning by narrating the work of Skinner on rat's learning.
22. Interpret Kohlr's work on chimpanzee's insight learning.
23. Give an account of territorial behaviour.
24. Discuss dominance hierarchy as agonistic behaviour.
25. Illustrate altruism through the organization of a honey bee society.

27

BIOLOGY AND HUMAN WELFARE

After completing this lesson, you will be able to

This is a 3 days unit

- Explain what is meant by integrated disease management
- Describe vaccination and its importance.
- List some common viral diseases against which vaccination are required e.g. polio, measles, influenza and hepatitis.
- Describe the role of vaccines in preventing polio, measles, hepatitis and tetanus.
- State the schedule of the vaccination against polio, measles, hepatitis and tetanus.
- Describe animal husbandry and the role of life stock in national economy (milk, meat, eggs, wool and other miscellaneous products).
- List the outstanding milk producing breeds of cows and buffaloes.
- Describe different methods adopted for plant improvements (acclimatization, selection, hybridization and back crosses etc).
- Explain home gardening and its importance.
- Identify some seasonal vegetable and fruit plants suitable for home gardening.
- Explain the role of microbes in household food processing, industrial production, sewage treatment and energy generation.

Reading

The theme of this chapter is to make the learners aware of the role of biological sciences for human welfare. They will also be given the fundamental knowledge of the various fields and it would help them in the choice of career. The age which is now passing away is largely the age of physical science, with its inventions and discoveries, which have given us power over the forces of Nature. It is to be hoped that when this War is over the age of physical science will be replaced by an age of biological science.

27.1 VACCINATION AND INTEGRATED DISEASE MANAGEMENT

One of the important contributions of biology for the welfare of mankind is the human disease control. There are several methods of disease control that have been discovered and are being used.

27.1.1 Integrated Disease Management (IDM)

Effective control of a particular disastrous disease or all the common diseases of a population can be achieved by using all relevant, appropriate methods of disease control. Such an approach of disease control is known as integrated disease management.

Procedure

Combating disease by utilizing all methods, as and when required, ensuring the participation of community in this program is very useful way of disease control. This requires an awareness of the community about the severity of the problem, its causes and its remedies. Public awareness can be ensured by using print and electronic media, by arranging seminars in school and colleges, or by person to person communication. In integrated disease management, every available method of disease control is used like preventive measures, drug treatment, vaccination and different kinds of therapies.

Objectives

Actually the real objective is to stop the further spread of disease and to prevent its new onset. This is proved very effective program for elimination and control of the dangerous disease from the human society.

27.1.2 Vaccination and its Importance

Vaccination is the administration of vaccine to stimulate the immune system of an individual to develop artificially induced active immunity against an infectious disease.

A **vaccine** may be intact but inactivated (non-infective) or an attenuated (with reduced infectivity) form of the causative pathogens (bacteria or viruses), or purified components of the pathogen that have been found to be highly immunogenic (e.g. the outer coat proteins of a virus particle). When the body is exposed to the weak or dead organisms (vaccine), the body is triggered to produce antibodies. Since the injected agents are weak or dead, the body does not actually suffer the disease, but an immune response is initiated. Now the body is fully equipped to fight against the actual causative agent like virus or bacteria that attack the body later in life. Vaccination is generally considered to be the most effective method of preventing infectious diseases that were once common in many countries, including polio, measles, mumps and tetanus.

Table 27.1 List of viral diseases against which vaccines are required

1.	Hepatitis A	HepA vaccine protects against hepatitis A.
2.	Hepatitis B	HepB vaccine protects against hepatitis B.
3.	Flu	Flu vaccine protects against influenza.
4.	Measles, Mumps and Rubella	MMR vaccine protects against measles, Mumps and Rubella.
5.	Polio	OPV and IPV vaccine protects against polio.

27.1.3 Role of Vaccines in Preventing Diseases

Vaccine is a non-virulent strain of actual causative agents that can develop immunity in the body against infectious diseases. Vaccines play a very significant role in preventing diseases.

Vaccination against polio

Polio is an infectious disease caused by a virus that lives in the throat and intestinal tract. There are two types of vaccine that protect against polio: inactivated polio vaccine (IPV) and oral polio vaccine (OPV). Widespread use of **polio vaccine** has led to complete elimination of polio cases in almost all the countries. IPV is a shot, given in the leg or arm, depending on age. It may be given at the same time as other vaccines. OPV is given to the children up to the age of 5 years in the form of drops orally.

Science Tidbits

The annual incidence of polio in Pakistan which was estimated to be more than 20,000 cases a year in early 1990's decreased to around 30 cases in 2005 and a few cases in 2016 and 2017. This happened due to regular program of vaccination.

Vaccination against measles

Measles is an infectious viral disease that occurs most often in the late winter and spring. Prior to 1963, almost everyone got measles; however, after the measles vaccine became available, the number of measles cases dropped by 99 %, and the epidemic cycles diminished drastically. Therefore, the best prevention of measles is the measles vaccine i.e., MMR. The MMR vaccine is a live, attenuated, combination vaccine that protects against the measles, mumps and rubella viruses.

Vaccination against hepatitis

Hepatitis means inflammation of the liver and also refers to a group of viral infections that affect the liver. The most common types are Hepatitis A, Hepatitis B, and Hepatitis C. Vaccines for hepatitis A and hepatitis B are available but there is currently no vaccine available for hepatitis C because of the structural characteristics of this virus. **Hepatitis A – vaccine** is made from inactivated whole virus of hepatitis A. **Hepatitis B vaccine** is a recombinant DNA vaccine.

Vaccination against tetanus (lockjaw)

Tetanus (lockjaw) causes painful tightening of the muscles, usually all over the body. It can lead to "locking" of the jaw so the victim cannot open his mouth or swallow. Tetanus leads to death in about 1 in 10 cases. Several vaccines are used to prevent tetanus among peoples of varying age groups. For example **DT** (diphtheria tetanus) vaccine is used for children and adolescents while **Td** is given to the adults. Vaccination is the best way to prevent a tetanus infection casued by *Clostridium tetani*.

Teacher's Point

The teacher would ask the students to write a research paper on "The history of development of immunization".

27.1.4 Schedule of Vaccination of Common Infectious Diseases

The recommended immunization schedule is designed to protect infants and children early in life, when they are most vulnerable and before they are exposed to potentially life-threatening diseases.

Table 27.2 Schedule of vaccination

Disease	Vaccine	Type	Age group
Polio	OPV (Oral Polio Vaccine)	Live vaccine	From birth to 5 years of age
Hepatitis	Hepatitis-B Vaccine	Recombinant DNA	At any age
Measles + Mumps + Rubella	Measles + Mumps + Rubella Vaccine (MMR)	Combination vaccine	Up to one year of age
Diphtheria + Tetanus	Diphtheria toxoid (DT) vaccine Tetanus toxoid (TT) vaccine	Toxoid vaccine	Generally in childhood

Science, Technology and Society Connections

Justify the importance of vaccination campaign observed worldwide to curb the diseases.

There are many lethal diseases which are threatening the health of human throughout the world. Most of them are endemic or epidemic. Fortunately, vaccines are available for most of the diseases which once were thought to be non-curable. But it is necessary that vaccination campaign should be worldwide, so that these diseases could be eradicated from whole world. Otherwise, these diseases will spread from nation to nation and will remain threat to whole world. For example, polio is eradicated completely from most of the world; but in countries like Pakistan still many cases of polio are reported. Government of Pakistan has started an extensive campaign of vaccination against this disease.

Science, Technology and Society Connections

List the objectives of the institutions of the Federal Health Department and UNO working for Integrated Disease Management.

Integrated Disease Management is a method to eradicate any disease by using all relevant, appropriate measures of disease control. Combating the disease by utilizing all the methods, as and when required and ensuring the participation of community in this program is very useful way of disease control. This requires awareness among the community about the severity of the problem, its causes and remedies. Institutions of Federal Health Department and UNO working for Integrated Disease Management fulfil this objective of public awareness and then co-operate different health departments to utilize all available resources and techniques to combat the disease.

27.2 ANIMAL HUSBANDRY

Animal husbandry deals with the care of livestock like cows, buffaloes, sheep, goats, chickens, and horses etc. Taking care of these animals involves feeding and watering them daily and keeping their living space clean. Breeding and birthing the animals, for example, helping deliver a baby calf is called **calving**, is also a big part of the job. Baby animals must be properly monitored and weaned (to accustom a young animal to nourishment other than

mother's milk) from their mothers in a timely manner. Another side of animal husbandry includes preparing the livestock for sale or slaughter and making sure they are ready to go to market. Other necessary tasks include medicating sick animals, monitoring the general health of the animals, herding and branding etc. Animal husbandry is the agricultural practice of breeding and raising livestock.

27.2.1 The Role of Livestock in National Economy

The animal husbandry plays vital role in national economy. Being a country that has a largely rural and agriculture based industry; animal husbandry plays an important role in the economy of Pakistan and is a major source of livelihood for many farmers. It is estimated that there are between 30 to 35 million people in Pakistan's current labour force who are engaged in livestock. Recently role of biotechnology in the field of animal nutrition, reproduction, breeding and management is tremendously improving the animal husbandry.

Livestock products

There are many livestock products which are being used in our daily lives. These products mainly include milk and milk products, meat and meat products, poultry and dung for fuel. In addition the other animal products as eggs, wool, leather goods etc., also contribute to the national economy. Dung excreted by cattle is a vital resource for supplying cooking fuel and soil fertilizers. Sheep are found throughout the grazing lands of central and northern Pakistan. Their wool is exported in large quantities.

Importance of livestock sector for Pakistan

The contribution of livestock to the national economy is estimated in billions. The livestock sector constitutes about 11% of Pakistan's GDP (Gross domestic product) and employs about 17% of the workforce, including most of the poorest people in the country. "The livestock sectors of Pakistan can singlehandedly become a game changer for our economy."

Table 27.3 Some of outstanding breeds of milk producing cows.

S.#	Name of the cow	Milk yield per lactation cycle
1.	Holstein-Friesian	12,700 L
2.	Friesen	7800 L
3.	Ayrshire	7,711L
4.	Jersey	7,260 L

Table 27.4 Some of outstanding breeds of milk producing buffaloes

S.#	Name ofbuffalo	Milk yield per lactation cycle
1.	Nili-Ravi	1500 to 2300 L
2.	Jaffarabadi	1000 to 1200 L
3.	Godavari	1200 to 1500 L
4.	Bhadawari	800 to 1000 L

Teacher's Point

The teacher would ask the students to write a paragraph on "How the livestock sector in Pakistan can

27.3 LATEST TECHNIQUE APPLIED TO ENHANCE CROP AND FRUIT YIELDS

Plant breeding started in the beginning of 20th century after the rediscovery of Mendel's work. Today plant breeding has become a specialized technology based on genetics.

27.3.1 Methods of Plant Breeding for Crop Improvement

There are several methods of plant breeding techniques which can be used for crop and fruit improvements. Some of common methods are: (1) Acclimatization (2) Selection (3) Hybridization (4) Backcross

Acclimatization

The process of introducing new plants from their growing place to new locality with different climate is termed as plant introduction. The adjustment of such plants to their new ...ility is called **acclimatization**.

Plant introduction has been an important basis of agricultural development throughout the ...rld. For example, groundnut was introduced in this subcontinent in the beginning of 19th century from Philippines, papayas from West Indies, potato from South America, date-palm from Brazil. Custard apple, coffee, tea, tobacco etc., have been successfully introduced in this subcontinent.

Selection

Selection is an important step in all breeding experiments and has been practiced by man since the early days of agriculture. The selection involves picking up the better ones out of the entire crop plants. The selected plants are separated from the inferior ones and are favoured by reproducing them under controlled conditions.

Hybridization

Hybridization is the technique of introducing characters of two desirable species into a single offspring (hybrid) by means of artificial pollination. Hybrids are first generation (F_1) crosses between genetically different parents. The term **hybrid** is used to describe the individuals that are heterozygous even for a single gene. Hybrids are known for their vigour, growth, size and yield. As a result of hybridization, hybrid varieties of cereals, oil seeds, pulses, sugar beet, onion, tomato and fruits have been developed.

Heterosis or **hybrid vigour** refers to the exhibition of superiority of the hybrid over both of its parents in one or more traits. It is based on the ability to give higher yield, and disease and pest resistance. It is best suited to plants which can be vegetatively propagated, e.g., sugarcane, mango, apple, guava, rose, dahlias, chrysanthemum, etc. In these plants, the heterohybrids retain their desirable characters indefinitely since there is no chance of segregation as they are multiplied vegetatively.

Backcross

Backcrossing is a crossing of a hybrid with one of its parents or an individual genetically similar to its parent, in order to achieve offspring with a genetic identity which is closer to that

of the parent. It is used in horticulture, animal breeding and in production of gene knockout organisms. Backcrossed hybrids are sometimes described with acronym "BC", for example, an F_1 hybrid crossed with one of its parents (or a genetically similar individual) can be termed a BC_1 hybrid, and a further cross of the BC_1 hybrid to the same parent (or a genetically similar individual) produces a BC_2 hybrid.

27.4 HOME GARDENING

The home garden is an integrated system which comprises different things in its small area: the family house, a living/playing area, a kitchen garden, a mixed garden, a fish pond, stores, an animal house and of course, people. The home garden can be defined as a farming system which combines different physical, social and economic functions on the area of land around the family home.

27.4.1 Importance of Home Gardening

It provides fuel for cooking, wood for building, food, medicinal plants, herbs, spices and flowers. It produces enough nutritious food, including some staple foods, for all the family year round. Sale from home garden produce can make a substantial contribution to a family's income. At any price, the market cannot compete with the flavour of home grown produce, which comes immediately from the plant to the dinner table. Home gardening produces relatively large amounts of food with marginal labour on areas of land too small for field agriculture.

The seasonal vegetable and fruit plants suitable for home gardening

In order to eat fresh fruits and vegetables, it's good to know when and what is available fresh. Here is a seasonality chart that will help you in choice for home gardening. This chart could be slightly different in different parts of the country.

Table 27.5 Seasonal vegetables and fruit plants

Winter **January, February**	Cabbage, Cauliflower, Celery Root, Grapefruit, Mandarin Oranges, Sweet Oranges, Pears, Spinach, Sweet Potatoes
Spring **March, April, May**	Asparagus, Basil, Beans, Berries, Broccoli, Cabbage, Cucumbers, Radish, Mangoes, Okra (lady's fingers), Oranges, Papayas, Peas
Summer **June, July, August**	Corn, Cucumbers, Dates, Figs, Grapes, Mangoes, Okra, Peaches, Chile Peppers, Sweet Peppers, Plums, Tomatoes, Watermelon
Fall **Sept., Oct., Nov.**	Apples, Cabbage, Cauliflower, Cranberries, Cucumbers, Dates, Grapes, Pears, Chile Peppers, Sweet Peppers, Spinach, Sweet Potatoes.

27.5 ROLES OF MICROBES IN HUMAN WELFARE

Microorganisms have a great impact on many areas of biology and general human welfare. Some are beneficial to man while others are harmful. The beneficial functions include production of bread, cheese, antibiotics, vaccines, vitamins, enzymes and many other products. Microorganisms occupy an important position in the ecosystem. They are required

of the various cycles of nature such as carbon, nitrogen, oxygen, and Sulphur that take place in the ecosystem.

27.5.1 Role of Microbes in Household Food Processing

There are many useful applications of microorganisms in the food processing industry. They influence the quality, availability and quantity of food. Microorganisms are used to change one substance to another which is used as food such as milk to yoghurt and cheese, sugar to wine and bread.

Table 27.6 Role of Microbes in Household Food Processing

Source/substrate	Microbe used	Food product
Pasteurized milk	*Streptococcus thermophilus* *Lactobacillus vulgaricus*	Yoghurt
Pasteurized milk	*Streptococcus sp.* *Lactobacillus sp.*	Cheese
Variety of organic compounds	*Penicillium sp.* *Saccharomyces sp.*	Acetic acid i.e., vinegar
Mixture of wheat and soya beans + salt water	*Aspergillus orzae* *Lactobacillus and yeast*	Soya sauce
Cereal grains ground into flour	*Saccharomyces cerevisiae*	Bread

27.5.2 Role of Microbes in Industrial Production

Microorganisms are particularly suitable for industrial processes as: (a) they produce higher ds and have higher specificity than conventional processes. (b) a wide range of chemicals be used and produced (c) some complex chemicals, such as hormones and antibiotics, can manufactured which are difficult to produce by other methods and specific isomers (such as L-amino acids) can be produced.

27.5.3 Role of Bacteria in the Sewage Treatment Process

Sewage treatment or domestic wastewater treatment is the process of removing contaminants from wastewater and household sewage, both runoff (effluents) and domestic. It includes physical, chemical and biological processes to remove physical, chemical and biological contaminants. A sewage treatment plant is nothing more than a giant microbial culture breeding facility where microbes are engaged to work for our benefit. Sewage treatment generally involves three stages, called primary, secondary and tertiary treatment.

Teacher's Point

The teacher would ask the students about the role of microorganism in treating waste water".

Primary treatment

Primary treatment consists of temporarily holding the sewage in a quiescent basin. The settled and floating materials are removed and the remaining liquid may be discharged or subjected to secondary treatment.

Secondary treatment

Secondary treatment removes dissolved and suspended biological matter. It is typically performed by indigenous, water-borne micro-organisms in a managed habitat. It may require a separation process to remove the micro-organisms from the treated water prior to discharge or tertiary treatment.

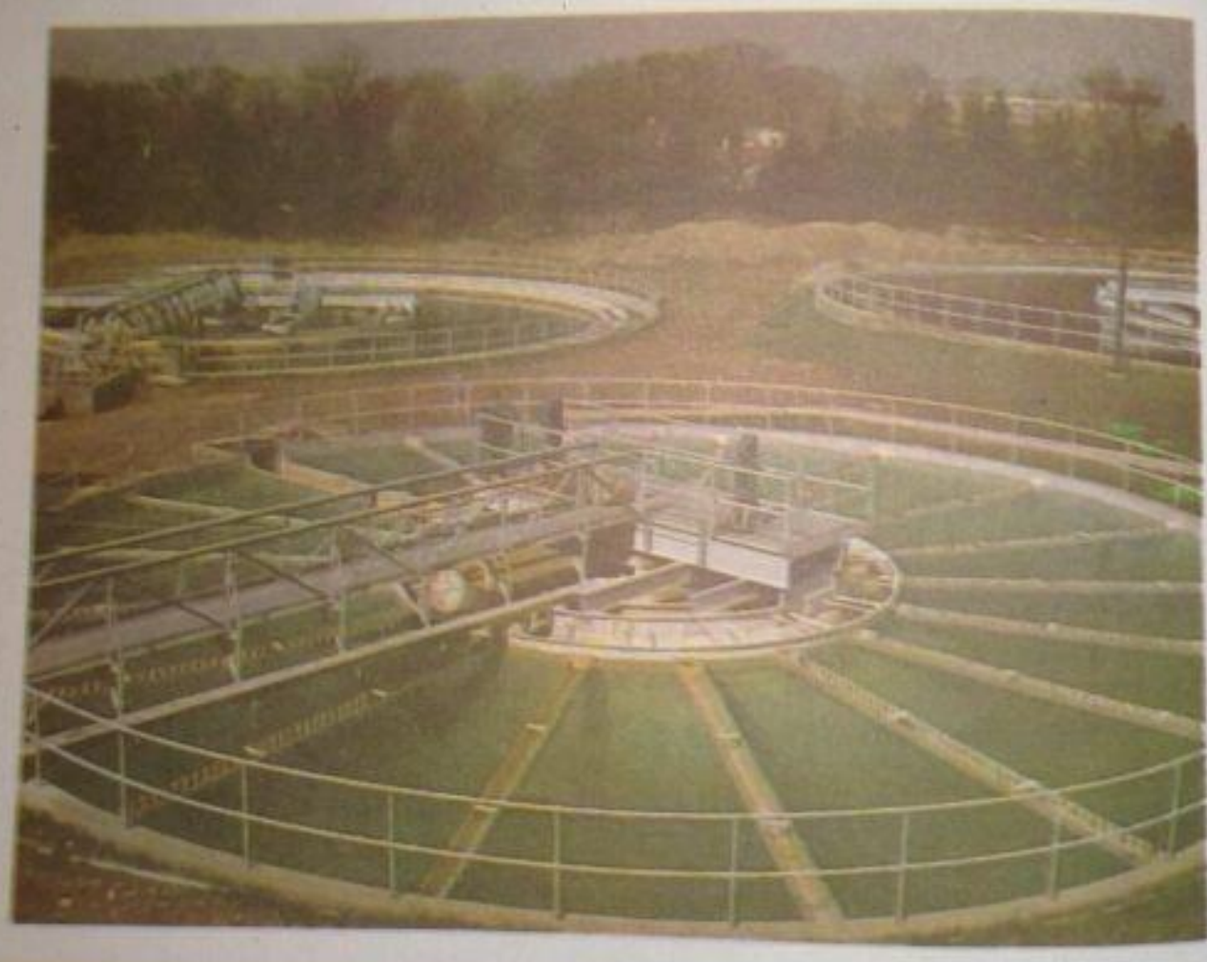


Fig. 27.1 Sewage treatment plant

Tertiary treatment

In tertiary treatment the treated water is sometimes disinfected chemically or physically prior to discharge into a stream, river and wetland etc.

27.5.4 Role of Microbes in Energy Production

There are microbes that clean sewage and generate electricity at the same time. Bacteria have evolved to utilize almost any chemical as a food source. The **sludge** is the precipitate produced by sewage treatment. If it is fed into large, enclosed tanks (anaerobic digesters) and heated initially to about 25°C. Conditions soon become anaerobic and wide range of bacteria in the waste material digest the organic compounds to fatty acids, monosaccharides and amino acids. These are acted on by other bacteria to produce organic acids and alcohols. Finally methanogenic bacteria convert these to methane and water. The methane is drawn off and used as an energy source to provide the heat needed to start the reactions and there is usually substantial surplus for other purposes such as central heating.

Science, Technology and Society Connections

Assess the impact of livestock in boosting up the national economy.

Pakistan being an agricultural based economy has a great potential in livestock. Livestock has a share of 10.6% of Pakistan's total GDP. Livestock sector employs 35 million people and produces almost $500 million products. Vast majority is of small firms of less than 2 hectors that maintain herds of 1 to 3 animals. Pakistan is fortunate to be the home tract of some of the finest natural breeds of livestock as compared with other regional countries.

Activity

Make a brochure to create awareness for vaccination against polio.

Exercise

M.C.Qs

1. **Select the correct answer**

(i) The artificial active immunity is achieved by injecting small amount of antigen called

(A) antibodies (B) toxin

(C) vaccine (D) serum

(ii) Polio virus lives in the ---------- of man

(A) mouth and throat (B) stomach and intestine

(C) mouth and intestine (D) throat and intestine

(iii) Measles is an infectious viral disease that occurs most often in
(A) late winter and autumn
(B) late winter and summer
(C) late winter and spring
(D) winter and rainy days

(iv) The best way to prevent a tetanus is
(A) antibiotic
(B) medication
(C) vaccination
(D) cleaness

(v) Hybridization in plants is the technique of introducing desirable species into a hybrid by means of:
(A) pollination
(B) selection
(C) emasculation
(D) micropropagation

(vi) Which of the following is/are the method of crop improvement?
(A) selection
(B) hybridization
(C) polyploid breeding
(D) all of these

Short Questions

2. Describe vaccination against: (a) measles (b) hepatitis (c) tetanus
3. What is vaccine and vaccination (immunization)?
4. What is the importance of vaccination?
5. Explain the role of livestock.
6. Describe hybridization.
7. Describe backcross.
8. What is the importance of home gardening?
9. Define/Describe/Explain briefly:

   vaccine, vaccination, acclimatization, animal husbandry, calving, mass selection, hybrid, heterosis, backcross, biopolymer, sludge.

10. Write the differences between
    (a) oral polio virus vaccine and hepatitis B vaccine
    (b) acclimitazation and selection
    (c) hybridization and backcross
    (d) yoghurt making and cheese making
    (e) primary and secondary treatment of sewage
    (f) secondary and tertiary treatment of sewage

Extensive Questions

11. Explain 'integrated disease management.
12. Describe vaccination and its importance.
13. Explain the role of vaccine in preventing diseases.
14. Give an account of animal husbandry.
15. Describe the techniques applied to enhance crop and fruit yield.
16. Describe home gardening and its importance.
17. Explain sewage treatment process.
18. Describe the role of microbes in:
    (a) Industrial production
    (b) Sewage treatment
    (c) Energy production

Glossary

A

5′ cap: The 5′ of a pre-mRNA modified by the addition of a cap of a guanine nucleotide.

A band: The broad region that corresponds to the length of the thick filaments of myofibrils.

A site: One of a ribosome's three binding sites for tRNA during translation. The A site holds the tRNA carrying the next amino acid to be added to the polypeptide chain. (A stands for aminoacyl tRNA).

abiotic: Nonliving

ABO blood groups: Genetically determined classes of human blood that are based on the presence or absence of carbohydrates A and B on the surface of red blood cells. The ABO blood group phenotypes, also called blood types, are A, B, AB and O.

abortion: The termination of pregnancy in progress.

acclimatization: Physiological adjustment to a change in an environmental factor.

acetylcholine: One of the most common neurotransmitters, functions by binding to receptors and altering the permeability of the postsynaptic membrane to specific ions, either depolarizing or hyperpolarizing the membrane.

acid precipitation: Rain, snow or fog that is more acidic than pH 5.6

actin: A globular protein which twist helically about each other, forming microfilaments in muscle and other contractile elements in the cells.

action potential: A rapid change in the membrane potential of an excitable caused by stimulus-triggered, selective opening and closing voltage-sensitive gates in sodium and potassium ion channels.

activator: A protein that binds to DNA and stimulates transcription of a specific gene.

adenyl cyclase: An enzyme that converts ATP to cyclic AMP in response to a chemical signal.

adrenal gland: One of two endocrine glands located to the kidneys in mammals. Endocrine cells in the outer portion (cortex) respond to help maintain homeostasis during long term stress. Neurosecretory cells in the central portion (medulla) secrete epinephrine and norepinephrine in response to nervous inputs triggered by short-term stress.

adrenocorticotropic hormone (ACTH): A tropic hormone produced and secreted by the anterior pituitary that stimulates the production and secretion of steroid hormones by the adrenal cortex.

aerobic: containing oxygen, referring to an organism, environment or cellular process that requires oxygen.

agarose: It is a polysaccharide polymer material, generally extracted from seaweed. Agarose is one of the two principal components of agar, and is purified from agar by removing agar's other component, agaropectin.

Agarose is frequently used in molecular biology for the separation of large molecules, especially DNA, by electrophoresis

agonistic behaviour: A type of behaviour involving a contest of some kind that determines which competitor gains access to some resource such as food or mates.

aldosterone: An adrenal hormone that acts on the distal tubules of the kidney to stimulate the reabsorption of sodium (Na^+) and the passive flow of water from the filtrate.

allantois: One of the four extraembryonic membranes: serves as a repository for the embryo's nitrogenous waste.

alleles: Alternative versions of gene that produce distinguishable phenotypic effects.

allopatric speciation: a mode of speciation induced when an ancestral population becomes segregated by a geographic barrier or itself divided into two or more geographically isolated subpopulation.

allopolyploid: A common type of polyploid species resulting from two different species interbreeding and combining their chromosomes.

altruism: Behaviour that reduces an individual's fitness while increasing the fitness of another individual.

alveolus: One of the dead-end, multi-lobed air sacs that constitute the gas exchange surface of the lungs.

Alzheimer's disease: An age related dementia (mental deterioration) characterized by confusion, memory loss and other symptoms.

aminoacyl-tRNA synthetase: An enzyme that joins each amino acid to the correct tRNA.

amniocentesis: A technique of prenatal diagnosis in which amniotic fluid, obtained by aspiration from a needle inserted into the uterus, is analyzed to detect certain genetic and congenital defects in the fetus.

amnion: The innermost of four extra-embryonic membranes, enclosed in a fluid filled sac in which the embryo is suspended.

anaerobic: Lacking oxygen, referring to an organism, environment, or cellular process that lacks oxygen and may be poisoned by it.

androgen: Any steroid hormone, such as testosterone, that stimulates the development and maintenance of the male reproductive system and sex characteristics.

aneuploidy: A chromosomal aberration in which one or more chromosomes are present in extra copies or are deficient in number.

angiotensin II: A hormone that stimulates constriction of precapillary arterioles and increases reabsorption of NaCl and water by the proximal tubules of the kidney, increasing blood pressure and volume.

anterior pituitary: Aso called the adenohypophysis, portion of the pituitary that develops from nonneural tissue, consists of endocrine cells that synthesize and secrete several tropic and nontropic hormone.

anticodon: A specialized base triplet at one end of a tRNA molecule that recognizes a particular complementary codon on an mRNA molecule.

antidiuretic hormone (ADH): A hormone produced in the hypothalamus and released from the posterior pituitary. It promotes water retention by the kidneys as part of an elaborate feedback scheme that helps regulate the osmolarity of the blood.

antiparallel: The opposite arrangement of the sugar-phosphate backbones in a DNA double helix.

archenteron: The endodermal lined cavity, formed during gastrulation process, that develops into the digestive tract of an animal.

artificial selection: The selective breeding of domesticated plants and animals to encourage the occurrence of desirable traits.

aspartate: An amino acid that functions as neurotransmitter.

assisted reproductive technology (ART): Fertilization procedure that generally involve the surgical removal of eggs (secondary oocyte) from a woman's ovaries after hormonal stimulation, fertilizing the egg and returning them to the woman's body.

associative learning: The acquired ability to associate one stimulus with another, also called classical conditioning.

atrial natriuretic factor: A peptide hormone that opposes the renin-angiotensin aldosterone system(RAAS).

autonomic nervous system: A subdivision of the motor nervous system of vertebrates that regulates the internal environment; consists of the sympathetic, parasympathetic and enteric divisions.

autopolyploid: An individual that has more than two chromosome sets, all derived from a single species.

autosome: A chromosome that is not directly involved in determining sex, as opposed to as sex chromosome.

axon: A typically long extension, or process, from a neuron that carries nerve impulses away from the cell body toward target cell.

B

Bartholin's gland: Glands near the vaginal opening in a human female that secret lubricating fluid during sexual arousal.

base pair substitution: A type of point mutation; the replacement of one nucleotide and its partner in the complementary DNA strand by another pair of nucleotides.

behaviour: Everything an animal does and how it does, including muscular activities such as chasing prey, certain nonmuscular processes such as secreting a hormone that attracts a mate and learning.

biogeochemical cycle: Any of the various nutrient circuits, which involve both biotic and abiotic components of ecosystems.

biogeography: The study of the past and present distribution of species.

biological clock: An internal time keeper that controls an organism's biological rhythms. The biological clock marks time with or without environmental cues but often requires signals from environment to remain tuned to an appropriate period.

biomass : The dry weight of organic matter comprising a group of organisms in a particular habitat.

biotechnology: The manipulation of living organisms or their components to produce useful products.

biotic: Pertaining to the living organisms in the environment.

bipolar cell: A neuron that synapses with the axon of a rod or cone in the retina of the eye.

blastocoel: The fluid-filled cavity that forms in the centre of the blastula embryo.

blastocyst: An embryonic stage in mammals; a hollow ball of cells produced one week after fertilization in humans.

blastoderm :An embryonic cap of dividing cells resting on a large undivided yolk.

blastomere: A small cell of an early embryo.

blastopore: The opening of the archenteron in the gastrula that develops into the mouth in protostomes and the anus in deuterostomes.

blastula: The hollow ball of cells marking the end stage of cleavage during early embryonic development.

blotting: A hybridization technique that enables researchers to determine the presence of certain nucleotide sequences in a sample of DNA.

body cavity: A fluid-containing space between the digestive tract and the body wall.

bone: A type of connective tissue, consisting of living cells held in a rigid matrix of collagen fibres embedded in calcium salts.

bottleneck effect: Genetic drift resulting from the reduction of a population, typically by a natural disaster, such that the surviving population is no longer genetically representative of the original population.

Bowman's capsule: A cup-shaped receptacle in the vertebrate kidney that is the initial, expanded segment of the nephron where filtrate enters from the blood.

brainstem: Collection of structures in the adult brain, including the midbrain, the pons, and the medulla oblongata; functions in homeostasis, coordination of movement, and conduction of information to higher brain centres.

breathing: The process involving alternate inhalation and exhalation of air that ventilates the lungs.

breathing control centre: A brain centre that directs the activity of organs involved in breathing.

bronchiole: One of the fine branches of the bronchus that transport air to alveoli.

bronchus: One of a pair of breathing tubes that branch from the trachea into the lungs.

bulbourethral gland: One of a pair of glands near the base of the penis in the human male that secretes fluid that lubricates and neutralizes acids in the urethra during sexual arousal.

C

calcitonin: A hormone secreted by the thyroid gland that lowers blood calcium levels by promoting calcium deposition in bone and calcium excretion from the kidneys.

callus: A mass of dividing, undifferentiated cells at the cut end of a shoot.

carrier: In genetics, an individual who is heterozygous at a given genetic locus, with one normal allele and one potentially harmful recessive allele. The heterozygote is phenotypically normal for the character determined by the gene but can pass on the harmful allele to offspring.

carrying capacity: The maximum population size that can be supported by the available resources, symbolized as K.

cartilage: A type of flexible connective tissue with an abundance of collagenous fibres embedded in chondroitin sulphate.

catastrophism: The hypothesis by Georges Cuvier that each boundary between strata corresponded in time to a catastrophe, such as a flood or drought, that had destroyed many of the species living there at that time.

cDNA library : A limited gene library using complementary DNA. The library includes only the genes that were transcribed in the cells examined.

cellular respiration: The most prevalent and efficient catabolic pathway for the production of ATP, in which oxygen is consumed as a reactant along with the organic fuel.

central canal: The narrow cavity in the centre of the spinal cord that is continuous with the fluid-filled ventricles of the brain.

central nervous system (CNS): In vertebrate animals, the brain and spinal cord.

cerebellum: Part of the vertebrate hindbrain located dorsally; functions in unconscious coordination of movement and balance.

cerebral cortex: The surface of the cerebrum; the largest and most complex part of the mammalian brain, containing sensory and motor nerve cell bodies of the cerebrum; the part of the vertebrate brain most changed through evolution.

cerebral hemisphere: The right or left side of the vertebrate brain.

cerebrospinal fluid: Blood-derived fluid that surrounds, protects against infection, nourishes, and cushions the brain and spinal cord.

cerebrum: The dorsal portion of the vertebrate forebrain, composed of right and left hemispheres; the integrating centre for memory, learning, emotions, and other highly complex functions of the central nervous system.

cervix: The neck of the uterus, which opens into the vagina.

chondrocyte: Cartilage cell that secretes collagen and chondroitin sulphate.

chorion: The outermost of four extraembryonic membranes; contributes to the formation of the mammalian placenta.

chorionic villus sampling (CVS): A technique of prenatal diagnosis in which a small sample of the foetal portion of the placenta is removed and analyzed to detect certain genetic and congenital defects in the foetus.

chromatin: The complex of DNA and proteins that makes up a eukaryotic chromosome. When the cell is not dividing, chromatin exists as a mass of very long, thin fibres that are not visible with a light microscope.

chromosome theory of inheritance: A basic principle in biology stating that genes are located on chromosomes and that the behavior of chromosomes during meiosis accounts for inheritance patterns.

circadian rhythm: A physiological cycle of about 24 hours that is present in all eukaryotic organisms and that persists even in the absence of external cues.

classical conditioning: A type of associative learning; the association of a normally irrelevant stimulus with a fixed behavioral response.

cleavage: The process of cytokinesis in animal cells, characterized by pinching of the plasma membrane. Also, the succession of rapid cell divisions without growth during early embryonic development that converts the zygote into a ball of cells.

cleavage furrow: The first sign of cleavage in an animal cell; a shallow groove in the cell surface near the old metaphase plate.

clone: (1) A lineage of genetically identical individuals or cells. (2) In popular usage, a single individual organism that is genetically identical to another individual. (3) As a verb, to make one or more genetic replicas of an individual or cell.

cloning: Using a somatic cell from a multicellular organism to make one or more genetically identical individuals.

cloning vector: An agent used to transfer DNA in genetic engineering. A plasmid that moves recombinant DNA from a test tube back into a cell is an example of a cloning vector, as is a virus that transfers recombinant DNA by infection.

codominance: The situation in which the phenotypes of both alleles are exhibited in the heterozygote.

codon: A three-nucleotide sequence of DNA or mRNA that specifies a particular amino acid or termination signal; the basic unit of the genetic code.

Coffin - Lowry syndrome: Males with Coffin-Lowry syndrome typically have severe to profound intellectual disability and delayed development. Affected women may be cognitively normal, or they may have intellectual disability ranging from mild to profound. This syndrome is inherited in an X-linked dominant pattern.

community: All the organisms that inhabit a particular area; an assemblage of populations of different species living close enough together for potential interaction.

complementary DNA (cDNA): A DNA molecule made in vitro using mRNA as a template and the enzyme reverse transcriptase. A cDNA molecule therefore corresponds to a gene, but lacks the introns present in the DNA of the genome.

conception: The fertilization of the egg by a sperm cell in humans.

corpus callosum: The thick band of nerve fibres that connect the right and left cerebral hemispheres in placental mammals, enabling the hemispheres to process information together.

cortex: Ground tissue that is between the vascular tissue and dermal tissue in a root or dicot stem.

cortical nephron: Nephrons located almost entirely in the renal coertex. These nephrons have a reduced loop of Henle.

corticosteroid: Any steroid hormone produced and secreted by the adrenal cortex.

cosmid: It is a type of hybrid plasmid that contains a Lambda phage cos sequence. Cosmids' (cos sites + plasmid = cosmids) DNA sequences are originally from the lambda phage. They are often used as a cloning vector in genetic engineering. Cosmids can be used to build genomic libraries. They were first described by Collins and Hohn in 1978.

countercurrent exchange: The opposite flow of adjacent fluids that maximizes transfer rates; for example, blood in the gills flows in the opposite direction in which water passes over the gills, maximizing oxygen uptake and carbon dioxide loss.

countercurrent multiplier system: A countercurrent system in which energy is expended in active transport to facilitate exchange of materials and create concentration gradients. For example, the loop of Henle actively transports NaCl from the filtrate in the upper part of the ascending limb of the loop, making the urine-concentrating function of the kidney more effective.

crossing over: The reciprocal exchange of genetic material between nonsister chromatids during prophase I of meiosis.

cystic fibrosis: A human genetic disorder caused by a recessive allele for a chloride channel protein; characterized by an excessive secretion of mucus and consequent vulnerability to infection; fatal if untreated.

D

Decidua: It is the term for the uterine lining (endometrium) during a pregnancy, which forms the maternal part of the placenta. It is formed under the influence of progesterone and forms highly characteristic cells. Different layers of the deciduas have been described: (a)Compact outer layer (stratum compactum) (b) Intermediate layer (stratum spongiosum) (c)Boundary layer adjacent to the myometrium (stratum basalis).That part of the decidua that interacts with the trophoblast is the decidua basalis (also called decidua placentalis). The remainder of the decidua is termed the decidua parietalis or decidua vera. Also, there is the decidua capsularis, which grows over the embryo, on the luminal side, enclosing it into the endometrium and surrounding the embryo together with decidua basalis

decidua ba·sa·lis: The area of endometrium between the implanted chorionic vesicle and the myometrium, which becomes the maternal part of the placenta. Also called *decidua serotina*.

deletion: (1) A deficiency in a chromosome resulting from the loss of a fragment through breakage. (2) A mutational loss of one or more nucleotide pairs from a gene.

demography: The study of statistics relating to births and deaths in populations.

dendrite: One of usually numerous, short, highly branched processes of a neuron that convey nerve impulses toward the cell body.

depolarization: An electrical state in an excitable cell whereby the inside of the cell is made less negative relative to the outside than at the resting membrane potential. A neuron membrane is depolarized if a stimulus decreases its voltage from the resting potential of -70 mV in the direction of zero voltage.

diabetes mellitus: An endocrine disorder marked by inability to maintain glucose homeostasis. The type I form results from autoimmune destruction of insulin-secreting cells; treatment usually requires insulin injections several times a day. The type II form most commonly results from reduced responsiveness of target cells to insulin; obesity and lack of exercise are risk factors.

diaphragm: A sheet of muscle that forms the bottom wall of the thoracic cavity in mammals; active in ventilating the lungs.

Digitalis lanata: is a species of foxglove. It gets its name due to the texture of the leaves. Digitalis lanata, like some other foxglove species, is highly toxic in all parts of the plant

dihybrid: An organism that is heterozygous with respect to two genes of interest. All the offspring from a cross between parents doubly homozygous for different alleles are dihybrids. For example, parents of genotypes AABB and aabb produce a dihybrid of genotype AaBb.

distal tubule: In the vertebrate kidney, the portion of a nephron that helps refine filtrate and empties it into a collecting duct.

DNA fingerprint: An individual's unique collection of DNA restriction fragments; detected by electrophoresis and nucleic acid probes.

DNA ligase: A linking enzyme essential for DNA replication; catalyzes the covalent bonding of the 3' end of a new DNA fragment to the 5' end of a growing chain.

DNA polymerase: An enzyme that catalyzes the elongation of new DNA at a replication fork by the addition of nucleotides to the existing chain.

dominant allele: An allele that is fully expressed in the phenotype of a heterozygote.

dopamine: A biogenic amine closely related to epinehhrine and norepinephrine.

dorsal lip: The dorsal side of the blastopore.

double helix: The form of native DNA, referring to its two adjacent polynucleotide strands wound into a spiral shape.

Down syndrome: A human genetic disease caused by presence of an extra chromosome 21; characterized by mental retardation and heart and respiratory defects.

Duchenne muscular dystrophy: A human genetic disease caused by a sex-linked recessive allele; characterized by progressive weakening and a loss of muscle tissue.

duplication: An aberration in chromosome structure due to fusion with a fragment from a homologous chromosome, such that a portion of a chromosome is duplicated.

E

E-site: One of ribosome's three binding sites for tRNA during translation. The E site is the place where discharged tRNAs leave the ribosome. (E stands for exit.)

ecological succession: Transition in the species composition of a biological community, often following ecological disturbance of the community; the establishment of a biological community in an area virtually barren of life.

ecology: The study of how organisms interact with their environment.

ecosystem: All the organisms in a given area as well as abiotic factors with which they interact; coimmunity and its physical environment.

ectotherm: An animal such as a reptile (other than birds), fish, or amphibian, that must use environmental energy and behavioural adaptations to regulate its body temperature.

ectothermic: Referring to organisms that do not produce enough metabolic heat to have much effect on body temperature.

ejaculatory duct: The short section of the ejaculatory route in mammals formed by the convergence of the vas deferens and a duct from the seminal vesicle. The ejaculatory duct transports sperm from the vas deferens to the urethra.

electrocardiogram (ECG): A record of the electrical impulses that travel through cardiac muscle during the heart cycle.

electroporation: A technique to introduce recombinant DNA into cells by applying a brief electrical pulse to a solution containing cells. The electricity creates temporary holes in the cells' plasma membranes, through which DNA can enter.

embryo sac: The female gametophyte of angiosperms, formed from the growth and division of the megaspore into a multicellular structure with eight haploid nuclei.

emigration: The movement of individuals out of a population.

endocrine gland: A ductless gland that secretes hormones directly into the interstitial fluid, from which they diffuse into the bloodstream.

endocrine system: The internal system of chemical communication involving hormones, the ductless glands that secrete hormones, and the molecular receptors on or in target cells that respond to hormones; functions in concert with the nervous system to effect internal regulation and maintain homeostasis.

endometrium: The inner lining of the uterus, which is richly supplied with blood vessels.

endorphin: Any of several hormones produced in the brain and anterior pituitary that inhibits pain perception.

endotherm: An animal, such as a bird or mammal, that uses metabolic heat to regulate body temperature.

endothermic: Referring to organisms with bodies that are warmed by heat generated by metabolism. This heat is usually used to maintain a relatively stable body temperature higher than that of the external environment.

epididymis: A coiled tubule located adjacent to the testes where sperm are stored.

epinephrine: A catecholamine hormone secreted from the adrenal medulla that mediates fight-or-flightresponses to short-term stress; also functions as a neurotransmitter.

epistasis: A type of gene interaction in which one gene alters the phenotypic effects of another gene that is independently inherited.

erythropoietin: A hormone produced in the kidney when tissues of the body do not receive enough oxygen. This hormone stimulates the production of erythrocytes.

estrogen: Any steroid hormone, such as estradiol, that stimulates the development and maintenance of the female reproductive system and secondary sex characteristics.

euchromatin: The more open, unravelled from eukaryotic chromatin that is available for transcription.

eumelanin: It is one of three basic types of melanin and the most common type. It is produced in 'black' and 'brown' subtypes or any melanin pigment of darker type.

evolution: All the changes that have transformed life on Earth from its earliest beginnings to the diversity that characterizes it today.

excretion: The disposal of nitrogen-containing waste products of metabolism.

exon: A coding region of a eukaryotic gene. Exons, which are expressed, are separated from each other by introns.

explant culture: In biology, it is a technique used for the isolation of cells from a piece or pieces of tissue.

extraembryonic membrane: Four membranes (yolk sac, amnion, chorion, allantois) that support the developing embryo in mammals birds and reptiles

F

feedback inhibition: A method of metabolic control in which the end product of a metabolic pathway acts as an inhibitor of an enzyme within that pathway.

fermentation: A catabolic process that makes a small amount of ATP from glucose without an electron transport chain and that produces a characteristic end product, such as ethyl alcohol or lactic acid.

fertilization: The union of haploid gametes to produce a diploid zygote.

filtration: The extraction of water and small solutes, including metabolic wastes, from the body fluid into excretory system.

foetus: A developing human from the ninth week of gestation until birth; has all the major structures of an adult.

follicle: A microscopic structure in the ovary that contains the developing ovum and secretes estrogens.

follicle-stimulating hormone (FSH) : A tropic hormone produced and secreted by the anterior pituitary that stimulates the production of eggs by the ovaries and sperm by the testes.

follicular phase: The part of the ovarian cycle during which follicles are growing and oocyte maturing.

forebrain: One of three ancestral and embryonic regions of the vertebrate brain; develops into the thalamus, hypothalamus, and cerebrum.

fossil: A preserved remnant or impression of an organism that lived in the past.

fragile X syndrome: It is an X-linked dominant condition. Fragile X syndrome is caused by a mutation in the fragile X mental retardation 1 gene (*FMR1*) on the X chromosome. Moderate to severe learning difficulties in males with the condition and mild learning difficulties in some females who are carriers. Behavioural problems, including autistic spectrum disorder, are common.

G

gadolinium: a metallic element of the rare earth; symbol **Gd**; atomic no 64; from the French chemist Gadolin; 1760-1852)

gamete: A haploid cell, such as an egg or sperm. Gametes unite during sexual reproduction to produce a diploid zygote.

gametogenesis: The process by which gametes are produced in the mammalian body.

ganglion (plural, ganglia): A cluster (functional group) of nerve cell bodies in a centralized nervous system.

gas exchange: The uptake of molecular oxygen from the environment and the discharge of carbon dioxide to the environment.

gastrula: The three-layered, cup-shaped embryonic stage.

gastrulation: The formation of a gastrula from a blastula.

gate ion channel: A gated channel for a specific ion. When ion channels are opened or closed, the membrane potential of the cell is altered.

gel electrophoresis: The separation of nucleic acids or proteins, on the basis of their size and electrical charge, by measuring their rate of movement through an electrical field in a gel.

gene: A discrete unit of hereditary information consisting of a specific nucleotide sequence in DNA (or RNA, in some viruses).

gene cloning: The production of multiple copies of a gene.

gene flow: Genetic additions to or subtractions from a population resulting from the movement of fertile individuals or gametes.

gene pool: The total aggregate of genes in a population at any one time.

genetic drift: Unpredictable fluctuations in allele frequencies from one generation to the next because of a population's finite size.

genetic engineering: The direct manipulation of genes for practical purposes.

genetic map: An ordered list of genetic loci (genes or other genetic markers) along a chromosome.

genome: The complete complement of an organism's genes; an organism's genetic material.

genomic library: A set of thousands of DNA segments from a genome, each carried by a plasmid, phage, or other cloning vector.

genomics: The study of whole sets of genes and their interactions.

germ layers: Three main layers that form the various tissues and organs of an animal body.

gestation: Pregnancy; the state of carrying developing young within the female reproductive tract.

glial cells: Supporting cells that are essential for the structural integrity of the nervous system and for the normal functioning of neurons.

glomerulus: A ball of capillaries surrounded by Bowman's capsule in the nephron and serving as the site of filtration in the vertebrate kidney.

glucagon: A hormone secreted by pancreatic alpha cells that raises blood glucose levels. It promotes glycogen breakdown and release of glucose by the liver.

glucocorticoid: A steroid hormone secreted by the adrenal cortex that influences glucose metabolism and immune function.

gonadotropin: A hormone that stimulates the activities of the testes and ovaries. Follicle stimulating hormone and luteinizing hormone are gonadotropins.

grey matter: Regions of dendrites and clusters of neuron cell bodies within the CNS.

gross primary production: The total primary production of an ecosystem.

growth hormone (GH): A hormone produced and secreted by the pituitary

H

habituation: A very simple type of learning that involves a loss of responsiveness to stimuli that convey little or no information.

Hardy-Weinberg equilibrium: The condition describing a non-evolving population (one that is in genetic equilibrium).

Hardy-Weinberg theorem: The principle that frequencies of alleles and genotypes in a population remain constant from generation to generation provided that only Mendelian segregation and recombination of alleles are at work.

heat-shock protein: A protein that helps protect other proteins during heat stress. Heat-shock proteins are found in plants, animals, and microorganisms.

heterochromatin: Nontranscribed eukaryotic chromatin that is so highly compacted that it is visible with a light microscope during interphase.

heterozygous: Having two different alleles for a given gene.

hindbrain: One of three ancestral and embryonic regions of the vertebrate brain; develops into the medulla oblongata, pons, and cerebellum.

histone: A small protein with a high proportion of positively charged amino acids that binds to the negatively charged DNA and plays a key role in its chromatin structure.

homeostasis: The steady-state physiological condition of the body.

homologous chromosomes:

Chromosome pairs of the same length, centromere position, and staining pattern that possess genes for the same characters at corresponding loci. One homologous chromosome is inherited from the organism's father, the other from the mother.

homology: Similarity in characteristics resulting from a shared ancestry.

homozygous: Having two identical alleles for a given gene.

hormone: In multicellular organisms, one of many types of circulating chemical signals that are formed in specialized cells, travel in body fluids, and act on specific target cells to change their functioning.

human chorionic gonadotropin (HCG): A hormone secreted by the chorion that maintains the corpus luteum of the ovary during the first three months of pregnancy.

human Genome Project: An international collaborative effort to map and sequence the DNA of the entire human genome.

Hunter's disease: Hunter syndrome, or mucopolysaccharidosis Type II, is a lysosomal storage disease caused by a deficient (or absent) enzyme, iduronate-2-sulfatase. The accumulated substrate in

Hunter's syndrome is heparan sulfate and dermatan sulfate. The syndrome has X-linked recessive inheritance.

hybridization: In genetics, the mating, or crossing, of two true-breeding varieties.

hymen: A dome-shaped rubber cup fitted into the upper portion of the vagina before sexual intercourse. It serves as a physical barrier to block the passage of sperm.

hyperpolarization: An electrical state in which the inside of the cell is more negative relative to the outside than at the resting membrane potential. A neuron membrane is hyperpolarised if a stimulus increases its voltage from the resting potential of -70 mV, reducing the chance that neuron will transmit a nerve impulse.

hypothalamus: The ventral part of the vertebrate forebrain; functions in maintaining homeostasis, especially in coordinating the endocrine and nervous systems; secretes hormones of the posterior pituitary and releasing factors that regulate the anterior pituitary.

hypophosphatemic rickets: It is a form of rickets that is characterized by low serum phosphate levels and resistance to treatment with ultraviolet radiation or vitamin D ingestion.

I

I band: The area near the edge of the sarcomere where there are only thin filaments.

incontinentia pigmenti: It is a genetic disorder that affects the skin, hair, teeth, nails, and central nervous system. It is named due to its microscopic appearance.

immigration: The influx of new individuals from other areas.

immunization: The process of generating a state of immunity by artifical means. In active immunization, a nonpathogenic version of a normally pathogenic microbe is administered, inducing B and T cell responses and immunological memory. In passive immunization, antibodies specific for a particular microbe are administered, conferring immediate but temporary protection. It is also called vaccination.

imprinting: A type of learned behaviour with a significant innate component, acquired during a limited critical period.

induction: the ability of one group embryonic cells to influence to influence the development of another.

in vitro fertilization: Fertilization of ova in laboratory containers followed by artificial implantation of the early embryo in the mother's uterus.

incomplete dominance: The situation in which the phenotype of heterozygote is intermediate between the phenotypes of individuals homozygous for either allele.

Innate behaviour: Behaviour that is developmentally fixed and under strong genetic control. Innate behaviour is exhibited in virtually the same form by all individuals in a population despite internal and external environmental differences during development and throughout their lifetimes.

insertion: A mutation involving the addition of one or more nucleotide pairs to a gene.

insulin: It is a peptide hormone, produced by beta cells in the pancreas, and is central to regulating carbohydrate and fat metabolism in the body. It causes cells in the skeletal muscles, and fat tissue to absorb glucose from the blood.

interneuron: An association neuron; a nerve cell within the central nervous system that forms synapses with sensory and motor neurons and integrates sensory input and motor output.

involution: Cells rolling over the edge of the lip of the blastopore into the interior of the embryo during gastrulation.

islets of Langerhans: Clusters of endocrine cells within the pancreas that produce and secrete the hormones glucagon (alpha cells) and insulin (beta cells).

J

juxtamedullary nephrons: Nephrons with well-developed loops of Henle that extend deeply into the renal medulla.

junk DNA: In genetics, "junk DNA" or noncoding DNA describes components of an organism's DNA sequences that do not encode for protein sequences.

K

karyotype: A display of the chromosome pairs of a cell arranged by size and shape.

Krause end bulbs: Nerve terminals in skin, mucosa of the oral cavity, conjunctiva, and other parts, consisting of a laminated capsule of connective tissue enclosing the terminal, branched, convoluted ending of an afferent nerve fiber; generally believed to be sensitive to touch and pressure. The end-bulbs of Krause were named after German anatomist Wilhelm Krause (1833-1910)

L

lambda phage: It also called Enterobacteria phage λ and coliphage λ, is a type of temperate bacteriophage or bacterial virus that infects the *Escherichia coli* (*E. coli*) species of bacteria. The virus may be housed in the genome of its host via lysogeny.

law of independent assortment: Mendel's second law, stating that each pair of alleles segregates independently during gamete formation; applies when genes for two characters are located on different pairs of homologous chromosomes.

law of segregation: Mendel's first law, stating that each allele in a pair separates into a different gamete during gamete formation.

leading strand: The new continuous complementary DNA strand synthesized along the template strand in the mandatory 5' (3' direction.

learning: A behavioural change resulting from experience.

Leydig cells: These are also known as interstitial cells of Leydig, are found adjacent to the seminiferous tubules in the testicle. They produce testosterone in the presence of luteinizing hormone (LH). Leydig cells are polyhedral in shape, display a large prominent nucleus, an eosinophilic cytoplasm and numerous lipid-filled vesicles.

ligament: It comes from the Latin *ligare* meaning "to bind, tie," which is precisely what a ligament does. Ligaments only connect bones to bones. Ligaments allow for range of motion.

ligand: In biochemistry and pharmacology, a ligand (from the Latin ligandum, binding) is a substance (usually a small molecule), that forms a complex with a biomolecule to serve a biological purpose. In protein-ligand binding, the ligand is usually a signal-triggering molecule, binding to a site on a target protein. In DNA-ligand binding studies, the ligand is usually any small molecule or ion,or even a protein that binds to the DNA double helix.

limbic system: A group of nuclei (clusters of nerve cell bodies) in the lower part of the mammalian forebrain that interact with the cerebral cortex in determining emotions; includes the hippocampus and the amygdala.

linked genes: Genes located close enough together on a chromosome to be usually inherited together.

locus (*plural*, loci):A specific place along the length of a chromosome where a given gene is located.

loop of Henle: In the kidney, the loop of Henle (or Henle's loop, nephron loop or its Latin counterpart ansa nephroni) is the portion of a nephron that leads from the proximal convoluted tubule to the distal convoluted tubule. Named after its discoverer F. G. J. Henle, the loop of Henle's main function is to create a concentration gradient in the medulla of the kidney.

luteal phase: That portion of the ovarian cycle during which endocrine cells of the corpus luteum secrete female hormones.

luteinizing hormone (LH): A tropic hormone produced and secreted by the anterior pituitary that stimulates ovulation in females and androgen production in males.

M

Malpighian tubule: A unique excretory organ of insects that empties into the digestive tract, removes nitrogenous wastes from the hemolymph, and functions in osmoregulation.

medulla oblongata: The lowest part of the vertebrate brain, commonly called the medulla; a swelling of the hindbrain dorsal to the anterior spinal cord that controls autonomic, homeostatic functions, including breathing, heart and blood vessel activity, swallowing, digestion, and vomiting.

menopause: The cessation of ovulation and menstruation.

menstrual cycle: A type of reproductive cycle in higher female primates, in which the nonpregnant endometrium is shed as a bloody discharge through the cervix into the vagina.

menstrual flow phase: That portion of the uterine (menstrual) cycle when menstrual bleeding occurs.

menstruation: The shedding of portions of the endometrium during a uterine (menstrual) cycle.

messenger RNA (mRNA): A type of RNA, synthesized from DNA, that attaches to ribosomes in the cytoplasm and specifies the primary structure of a protein.

midbrain: One of three ancestral and embryonic regions of the vertebrate brain; develops into sensory integrating and relay centers that send sensory information to the cerebrum.

monosomic: Referring to a cell that has only one copy of a particular chromosome, instead of the normal two.

myelin sheath: In a neuron, an insulating coat of cell membrane from Schwann cells that is interrupted by nodes of Ranvier, where saltatory conduction occurs.

myofibril: A myofibril (also known as a muscle fibril) is a basic rod-like unit of a muscle.[1] Muscles are composed of tubular cells called myocytes, also known as muscle fibers, and these cells in turn contain many chains of myofibrils. They are created during embryo development in a process known as myogenesis. Myofibrils are composed of long proteins such as actin, myosin, and titin, and other proteins that hold them together. These proteins are organized into thin filaments and thick filaments, which repeat along the length of the myofibril in sections called sarcomeres. Muscles contract by sliding the thin (actin) and thick (myosin) filaments along each other.

myofilaments: These are the filaments of myofibrils constructed from proteins.There are three different types of myofilaments, thick, thin, and elastic filaments.

myoglobin: An oxygen-storing, pigmented protein in muscle cells.

myosin: A type of protein filament that interacts with actin filaments to cause cell contraction.

N

natural selection: Differential success in the reproduction of different phenotypes resulting from the interaction of organisms with their environment. Evolution occurs when natural selection causes changes in relative frequencies of alleles in the gene pool.

negative feedback: A primary mechanism of homeostasis, whereby a change in a physiological variable that is being monitored triggers a response that counteracts the initial fluctuation.

nephron: The tubular excretory unit of the vertebrate kidney.

nerve: A ropelike bundle of neuron fibers (axons and dendrites) tightly wrapped in connective tissue.

net primary production (NPP): The gross primary production of an ecosystem minus the energy used by the producers for respiration.

neural crest: A band of cells along the border where the neural tube pinches off from the ectoderm. The cells migrate to various parts of the body embryo and form the pigment cell in the skin, bones of the skull, the teeth, the adrenal glands and parts of the peripheral nervous system.

neural tube: A tube of cells running along the dorsal axis of the body, just dorsal to the notochord. It will give rise to the central nervous system.

neuron: A nerve cell; the fundamental unit of the nervous system, having structure and properties that allow it to conduct signals by taking advantage of the electrical charge across its cell membrane.

neurosecretory cells: These are specialized nerve cells that produce and secrete hormones. Well-known examples of neurosecretory cells are oxytocin- and vasopressin-secreting neurons in the hypothalamus and cells in the adrenal medulla. These cells are found in vertebrates and invertebrates.

neurotransmitter: A chemical messenger released from the synaptic terminal of a neuron at a chemical synapse that diffuses across the synaptic cleft and binds to and stimulates the postsynaptic cell.

Nissl body: also known as Nissl or tigroid substance is a large granular body found in neurons. These granules are rough endoplasmic reticulum (RER) with rosettes of free ribosomes, and are the site of protein synthesis. It was named after Franz Nissl, a German psychiatrist who invented the Nissl staining method.

nociceptor: A class of naked dendrites in the epidermis of the skin.

nondisjunction: It is the failure of chromosome pairs or sister chromatids to separate properly during cell division. Nondisjunction ("not coming apart") is an erroneous chromosomal segregation that results in an abnormal number of chromosomes (aneuploidy). There are three forms of nondisjunction involving segregation during the cell cycle: failure of a pair or homologous chromosomes to separate in meiosis I, or the failure of sister chromatids to separate during meiosis II or mitosis. Calvin Bridges and Thomas Hunt Morgan are credited with discovering nondisjunction in *Drosophila* sex chromosomes in the spring of 1910, while working in the Zoological Laboratory of Columbia University.

norepinephrine: also called noradrenaline or 4,5-β-trihydroxy phenethylamine is a catecholamine with multiple roles including those as a hormone and a neurotransmitter.

notochord: A longitudinal, flexible rod that runs along the dorsal axis of an animal's body in the future position of the vertebral column.

nucleic acid probe: In DNA technology, a labeled single-stranded nucleic acid molecule used to tag a specific nucleotide sequence in a nucleic acid sample. Molecules of the probe hydrogen-bond to the complementary sequence wherever it occurs; radioactive or other labeling of the probe allows its location to be detected.

nucleoid: A dense region of DNA in a prokaryotic cell.

nucleosome: The basic, bead-like unit of DNA packaging in eukaryotes, consisting of a segment of DNA wound around a protein core composed of two copies of each of four types of histone.

O

Okazaki fragment: A short segment of DNA synthesized on a template strand during DNA replication. Many Okazaki fragments make up the lagging strand of newly synthesized DNA.

oogenesis: The process in the ovary that results in the production of female gametes.

oogonia: Ovary-specific stem cells.

operant conditioning: A type of associative learning in which an animal learns to associate one of its own behaviors with a reward or punishment and then tends to repeat or avoid that behavior; also called trial-and-error learning.

operon: In genetics, an operon is a functioning unit of genomic DNA containing a cluster of genes under the control of a single promoter. The genes are transcribed together into an mRNA strand and either translated together in the cytoplasm, or undergo trans-splicing to create monocistronic mRNAs that are translated separately, i.e. several strands of mRNA that each encode a single gene product. The result of this is that the genes contained in the operon are either expressed together or not at all. Several genes must be co-transcribed to define an operon.

organogenesis: The development of organ rudiments from the three germ layers.

osteoblast: A bone-forming cell that deposits collagen.

osteon: The repeating organizational unit forming the microscopic structure of hard mammalian bone.

ovarian cycle: The cyclic recurrence of the follicular phase, ovulation, and the luteal phase in the mammalian ovary, regulated by hormones.

ovary: (1) In flowers, the portion of a carpel in which the egg-containing ovules develop. (2) In animals, the structure that produces female gametes and reproductive hormones.

oviduct: A tube passing from the ovary to the vagina in invertebrates or to the uterus in vertebrates.

ovulation: The release of an egg from ovaries. In humans, an ovarian follicle releases an egg during each uterine (menstrual) cycle.

ovum: The female gamete; the haploid, unfertilized egg, which is usually a relatively large, nonmotile cell.

oxytocin: It is a mammalian neurohypophysial hormone. Produced by the hypothalamus and stored

and secreted by the posterior pituitary gland, oxytocin acts primarily as a neuromodulator in the brain. It is released in large amounts after distension of the cervix and uterus during lab or, facilitating birth, maternal bonding, and, after stimulation of the nipples, lactation. Childbirth and milk ejection result from positive feedback mechanisms.

P

P generation: The parent individuals from which offspring are derived in studies of inheritance; P stands for parental.

P site: One of a ribosome's three binding sites for tRNA during translation. The P site holds the tRNA carrying the growing polypeptide chain. (P stands for peptidyl tRNA.)

pain receptor: A kind of interoreceptor that detects pain; also called a nociceptor.

parathyroid gland: Any of four small endocrine glands, embedded in the surface of the thyroid gland, that secrete parathyroid hormone.

parathyroid hormone (PTH): A hormone secreted by the parathyroid glands that raises blood calcium level by promoting calcium release from bone and calcium retention by the kidneys.

parthenogenesis: A type of reproduction in which females produce offspring from unfertilized eggs.

parturition: The expulsion of a baby from the mother; also called birth.

pedigree: A diagram of a family tree showing the occurrence of heritable characters in parents and offspring over multiple generations.

peripheral nervous system (PNS): The sensory and motor neuron that connect to the central nervous system.

peritubular capillaries: The network of tiny blood vessels that surrounds the proximal and distal tubules in the kidney.

perennial: A flowering plant that lives for many years.

primary succession: A type of ecological succession that occurs in a virtually lifeless area, where there were originally no organisms and where soil has not yet formed.

pheomelanin: It is one of three basic types of melanin. I t is a cysteine containing red-brown polymer of benzothiazine units largely responsible for red hair and freckles.

pheromone: In animals and fungi, small, volatile chemical that functions in communication and that in animals acts much like a hormone in influencing physiology and behaviour.

pineal gland: A small gland on the dorsal surface of the vertebrate forebrain that secretes the hormone melatonin.

pituitary gland: An endocrine gland at the base of the hypothalamus; consists of a posterior lobe (neurohypophysis), which stores and releases two hormones produced by the hypothalamus, and an anterior lobe (adenohypophysis), which produces and secretes many hormones that regulate diverse body functions.

placenta: A structure in the pregnant uterus for nourishing a viviparous foetus with the mother's blood supply; formed from the uterine lining and embryonic membranes.

plankton: Mostly microscopic organisms that drift passively or swim weakly near the surface of oceans, ponds, and lakes.

plasmid: A small ring of DNA that carries accessory genes separate from those of a bacterial chromosome, also found in some eukaryotes, such as yeast.

poly-A tail: the modified end of the 3' end of an mRNA molecule consisting of the addition of some 50 to 250 adenine nucleotide.

polygenic inheritance: An additive effect of two or more gene loci on a single phenotypic character.

polymerase chain reaction (PCR): A technique for amplifying DNA in vitro by incubating with special primers, DNA polymerase molecules, and nucleotides.

polyploidy: A chromosomsl alteration in which the organism possesses more than two complete chromosome sets.

pons: Portion of the brain that participates in certain automatic, homeostatic functions, such as regulating the breathing centers in the medulla.

population: A localized group of individuals that belong to the same biological species (that are capable of interbreeding and producing fertile offspring).

positive feedback: A physiological control mechanism in which a change in some variable triggers mechanisms that amplify the change.

posterior pituitary: Also called the neurohypophysis; an extension of the hypothalamus composed of nervous tissue that secretes oxytocin and antidiuretic hormone made in the hypothalamus; a temporary storage site for these hormones.

postsynaptic cell: The target cell at a synapse.

presynaptic cell: The transmitting cell at a synapse.

Protenor bugs: These are plant-feeding bugs that mainly suck the juices out of grasses and sedges.

primary succession: A type of ecological succession that occurs in a virtually lifeless area, where there were originally no organisms and where soil has not yet formed.

primer: A polynucleotide with a free 3' end, bound by complementary base pairing to the template strand, that is elongated during DNA replication.

prolactin (PRL): A hormone produced and secreted by the anterior pituitary with a great diversity of effects in different vertebrate species. In mammals, it stimulates growth of and milk production by the mammary glands.

proliferative phase: That portion of the uterine (menstrual) cycle when the endometrium regenerates and thickens.

promoter: A specific nucleotide sequence in DNA that binds RNA polymerase and indicates where to start transcribing RNA.

prostaglandin (PG): One of a group of modified fatty acids secreted by virtually all tissues and performing a wide variety of functions as local regulators.

proximal tubule: In the vertebrate kidney, the portion of a nephron immediately downstream from Bowman's capsule that conveys and helps refine filtrate.

Punnet square: A diagram used in the study of inheritance to show the results of random fertilization in genetic cross.

R

recessive allele: An allele whose phenotypic effect is not observed in a heterozygote.

refractory periods: The short time immediately after an action potential in which the neuron cannot respond to another stimulus, owing to an increase in potential permeability.

renal artery: The blood vessel bringing blood to the kidney.

renal cortex: The outer portion of the vertebrate kidney.

renal medulla: The inner portion of the vertebrate kidney, beneath the renal cortex.

renal pelvis: Funnel-shaped chamber that receives processed filtrate from the vertebrate kidney's collecting ducts and is drained by the ureter.

renal vein: The blood vessel draining the kidney.

replication fork : A Y-shaped region on a replicating DNA molecule where new strands are growing.

residual volume: The amount of air that remains in the lungs after forcefully exhaling.

respiratory pigment: A protein that transports most of the oxygen in blood.

respiratory surface: The part of an animal where gases are exchanged with the environment.

resting potential: The membrane potential characteristic of a nonconducting, excitable cell, with the inside of the cell more negative than the outside.

restriction enzyme: A degradative enzyme that recognizes and cuts up DNA (including that of certain phages) that is foreign to a bacterium.

restriction fragment: DNA segment resulting from cutting of DNA by a restriction enzyme.

restriction fragment length polymorphisms (RFLPs): Differences in DNA sequence on homologous chromosomes that can result in different patterns of restriction fragment lengths (DNA segments resulting from treatment with restriction enzymes); useful as genetic markers for making linkage maps.

restriction site: A specific sequence on a DNA strand that is recognized as a cut site by a restriction enzyme.

Rh factor: A protein antigen on the surface of red blood cells designated Rh-positive. If an Rh-negative mother is exposed to blood from an Rh-positive fetus, she produces anti-Rh antibodies of the IgG class.

rigor mortis: The stiffness of joints and muscular rigidity of a dead body, caused by depletion of ATP in the tissues. It begins two to four hours after death and lasts up to about four days, after which the muscles and joints relax.

RNA polymerase: An enzyme that links together the growing chain of ribonucleotides during transcription.

RNA processing: Modification of RNA before it leaves the nucleus, a process unique to eukaryotes.

RNA splicing: The removal of noncoding portions (introns) of the RNA molecule after initial synthesis.

S

saltatory conduction: Rapid transmission of a nerve impulse along an axon, resulting from the action potential jumping from one node of Ranvier to another, skipping the myelin-sheathed regions of membrane.

sarcomere: The fundamental repeating unit of striated muscle, delimited by the Z lines.

sarcoplasmic reticulum: A specialized endoplasmic reticulum that regulates the calcium concentration in the cytosol.

Schwann cell: A type of glial cell that forms insulating myelin sheaths around the axons of neurons in the peripheral nervous system.

scrotum: A pouch of skin outside the abdomen that houses a testis; functions in cooling sperm, thereby keeping them viable.

secondary succession: A type of succession that occurs where an existing community has been cleared by some disturbance that leaves the soil intact.

semen: The fluid that is ejaculated by the male during orgasm; contains sperm and secretions from several glands of the male reproductive tract.

second messenger: A small, nonprotein, water soluble molecule or ion such as calcium ion or cyclic AMP, that relays a signal to a cell's interior in response to a signal received by a signal protein.

semiconservative model: Type of DNA replication in which the replicated double helix consists of one old strand, derived from the old molecule, and one newly made strand.

seminal vesicle: A gland in males that secretes a fluid component of semen that lubricates and nourishes sperm.

seminiferous tubule: A highly coiled tube in the testis in which sperm are produced.

sensory neuron: A nerve cell that receives information from the internal and external environments and transmits the signals to the central nervous system.

Serotonin: It is a neurotransmitter found in the digestive tract, the central nervous system, blood platelets and the pineal gland (deep at the center of the brain). It is also known as 5-hydroxytryptamine, which is often abbreviated to 5-HT.

sex chromosome: One of the pair of chromosome responsible for determining the sex of an individual.

sex-linked gene: A gene located on a sex chromosome.

skeletal muscle (striated muscle): Muscle generally responsible for the voluntary movements of the body.

sliding-filament model: The theory explaining how muscle contracts, based on change within a sarcomere, the basic unit of muscle organization, stating that thin (actin) filaments slide across thick (myosin) filaments, shortening the sarcomere. The shortening of all sarcomeres in a myofibril shortens the entire myofibril.

sodium-potassium pump: A special transport protein in the plasma membrane of animal cells that transports sodium out of the cell and potassium into the cell against their concentration gradients.

somatic nervous system: The branch of the motor division of the vertebrate peripheral nervous system composed of motor neurons that carry signals to skeletal muscles in response to external stimuli.

speciation: The origin of new species in evolution.

species: A group whose members possess similar anatomical characteristics and have the ability to interbreed.

sperm: The male gamete.

spermatogenesis: The continuous and prolific production of mature sperm cells in the testis.

stem cell: Any relatively unspecialized cell that can divide during a single division into one identical daughter cell and one more specialized daughter cell, which can undergo further differentiation.

steroid: A type of lipid, characterized by a carbon skeleton consisting of four rings with various functional groups attached.

stratum basalis: Boundary layer adjacent to the myometrium in the deciduous is called *stratum basalis*.

synapse *(sin-apse)*: The locus where one neuron communicates with another neuron in a neural pathway; a narrow gap between a synaptic terminal of an axon and a signal-receiving portion (dendrite or cell body) of another neuron or effector cell. Neurotransmitter molecules released by synaptic terminals diffuse across the synapse, relaying messages to the dendrite or effector.

synaptic cleft: A narrow gap separating the synaptic knob of a transmitting neuron from a receiving neuron or an effctor cell.

synaptic terminal: A bulb at the end of an axon in which neurotransmitter molecules are stored and released.

synaptic vesicle: Membranous sac containing neurotransmitter molecules at the tip of the presynaptic axon.

T

TATA box: A promoter DNA sequence crucial in forming the transcription initiation complex.

taxis: Movement toward or away from a stimulus.

telomere: The protective structure at each end of a eukaryotic chromosome. Specifically, the tandem repetitve DNA at the end of the chromosome's DNA molecule. See also repetitive DNA.

tendon: A type of fibrous connective tissue that attaches muscle to bone.

territoriality: A behaviour in which an animal defends a bounded physical space against encroachment by other individuals, usually of its own species. Territory defence may involve direct aggression or indirect mechanisms such as scent marking or singing. **test cross:** In genetics, a test cross, first introduced by Mendel, involves the breeding of a dominant trait individual with a recessive individual, in order to determine the zygosity of the former by analysing proportions of offspring with the recessive phenotype.

Testicular feminization syndrome: Now more appropriately called the complete androgen insensitivity syndrome, this is a genetic disorder that makes XY foetuses insensitive (unresponsive) to androgens (male hormones). Instead, they are born looking externally like normal girls. Internally, there is a short blind-pouch vagina and no uterus, fallopian tubes or ovaries. There are testes in the abdomen or the inguinal canal. (The inguinal canals are the two passages in the anterior abdominal wall which in men convey the spermatic cords and in women the round ligament of uterus. The inguinal canals are larger and more prominent in men. There is one inguinal canal on each side of the midline.)

testis: (plural testes). The male reproductive organ or gonad in which sperms and reproductive hormones are produced.

testosterone: The most abundant androgen hormone in the male body.

tetanus: The maximal, sustained contraction of a skeletal muscle, caused by a very fast frequency of action potentials elicited by continual stimulation.

thalamus: One of two integrating centres of the vertebrate forebrain. Neurons with cell bodies in the thalamus relay neural input to specific areas in the

cerebral cortex and regulate what information goes to the cerebral cortex.

threshold: The potential an excitable cell membrane must reach for an action potential to be initiated.

thrombus: A clump of platelets and fibrin that blocks the flow of blood through a blood vessel.

thymus: A small organ in the thoracic cavity of vertebrates where maturation of T cells is completed.

thyroid gland: An endocrine gland, located on the ventral surface of the trachea, that secretes two iodine-containing hormones, triiodothyronine (T3) and thyroxine (T4), and calcitonin.

thyroid-stimulating hormone (TSH): A tropic hormone produced and secreted by the anterior pituitary that regulates the release of thyroid hormones.

thyroxine (T4) : One of the two iodine –containing hormones that are secreted by the thyroid gland and help regulate metabolism, development and maturation in vertebrates.

trachea: The windpipe; that portion of the respiratory tube that has C-shaped cartilaginous rings and passes from the larynx to two bronchi.

transcription: The synthesis of RNA on a DNA template.

transgenic: Pertaining to an individual plant or animal whose genome contains a gene introduced from another organism, either from the same or a different species.

translation: The synthesis of a polypeptide using the genetic information encoded in an mRNA molecule. There is a change of language from nucleotides to amino acids.

translocation: (1) An aberration in chromosome structure resulting from attachment of a chromosomal fragment to a nonhomologous chromosome. (2) During protein synthesis, the third stage in the elongation cycle when the RNA carrying the growing polypeptide moves from the A site to the P site on the ribosome. (3) The transport of organic nutrients in the phloem of vascular plants.

tridothyronine (T3): One of the two iodine –containing hormones that are secreted by the thyroid gland and help regulate metabolism, development and maturation in vertebrates.

trisomic: Referring to a cell that has three copies of a particular chromosome, instead of the normal two.

Tris Hcl: Tris, or tris(hydroxymethyl) amino methane, or known during medical use as tromethamine or THAM, is an organic compound with the formula (HOCH2)3CNH2. It is extensively used in biochemistry and molecular biology as a component of buffer solutions such as in TAE and TBE buffers, especially for solutions of nucleic acids. It contains a primary amine and thus undergoes the reactions associated with typical amines, e.g. condensations with aldehydes. In medicine, tromethamine is occasionally used as a drug, given in intensive care for its properties as a buffer for the treatment of severe metabolic acidosis in specific circumstances. Some medications are formulated as the "tromethamine salt" including hemabate (carboprost as trometamol salt), and "ketorolac trometamol"

triplet code: A set of three-nucleotide-long words that specify the amino acids for polypeptide chains.

tropic hormone: A hormone that has another endocrine gland as a target.

trophoblast: The outer epithelium of the blastocyst, which forms the foetal part of the placenta.

U

uniformitarianism: Charles Lyell's idea that geologic processes have not changed throughout Earth's history.

urea: A soluble nitrogenous waste produced in the liver by a metabolic cycle that combines ammonia with carbon dioxide.

ureter: A duct leading from the kidney to the urinary bladder.

urethra: A tube that releases urine from the body near the vagina in females and through the penis in males; also serves in males as the exit tube for the reproductive system.

uric acid: An insoluble precipitate nitrogenous waste excreted by land snails, insects and many reptiles and birds.

urinary bladder: The pouch where urine is stored prior to elimination.

uterine cycle: The changes that occur in the uterus during the reproductive cycle of the human female; also called the menstrual cycle.

uterus: A female organ where eggs are fertilized and/or development of the young occurs.

V

vaccine: A harmless variant or derivative of a pathogen that stimulates a host's immune system to mount defenses against the pathogen.

vagina: Part of the female reproductive system between the uterus and the outside opening; the birth canal in mammals; also accommodates the male's penis and receives sperm during copulation.

vasa recta: In the blood supply of the kidney, the vasa recta renis (or straight arteries of kidney, or straight arterioles of kidney) are a series of straight capillaries in the medulla (Latin: vasa, "vessels"; recta, "straight"). They lie parallel to the loop of Henle.

vasoconstriction: A decrease in the diameter of superficial blood vessels triggered by nerve signals that contract the muscles of the vessel walls.

vasodilation: An increase in the diameter of superficial blood vessels triggered by nerve signals that relax the muscles of the vessel walls.

ventilation: Any method of increasing contact between the respiratory medium and the respiratory surface.

ventricle: (1) A heart chamber that pumps blood out of a heart. (2) A space in the vertebrate brain, filled with cerebrospinal fluid.

vestigial organ: A structure of marginal, if any, importance to an organism. Vestigial organs are historical remains of structures that had important functions in ancestors.

villi of chorion frondosum: In the early weeks of development, villi cover the entire surface of the chorion. As pregnancy advances, villi on the embryonic pole continue to grow and expand, giving rise to the *chorion frondosum* (bushy chorion). The chorion frondosum forms up the placenta together with the decidual plate.

vital capacity: The maximum volume of air that a respiratory system can inhale and exhale.

vitamin D resistant rickets: It is defined by its resistance to the vitamin D treatment generally used in deficiency rickets.

vocal cord: One of two small bands of muscle within the larynx. These muscles vibrate to produce the voice. The vocal cords form a "V" inside the larynx, a 2-inch-long, tube-shaped organ in the neck. When we talk, the vocal cords tighten up and move closer together. Air from the lungs is forced between them and makes them vibrate, producing the sound of our voice. The tongue, lips, and teeth form this sound into words.

vulva: Collective term for the female external genitalia.

W

white matter: Tracts of axons within the CNS.

wild type: An individual with the normal (most common) phenotype.

X

Xerosere: It is a plant succession which is limited by water availability. It includes the different stages in a xerarch succession. Xerarch succession of ecological communities originated in extremely dry situation such as sand deserts, sand dunes, salt deserts, rock deserts etc.

Y

yeast artificial chromosome (YAC): A vector that combines the essentials of a eukaryotic chromosome—an origin for DNA replication, a centromere, and two telomeres—with foreign DNA.

Z

Z lines: The border of a sarcomere.

zero population growth (ZPG): A period of stability in population size, when the per capita birth rate and death rate are equal.

zygote: The diploid product of the union of haploid gametes in conception; a fertilized egg.

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He published four research papers in Science Journals of Pakistan on Butterflies stan. He is co-author and managing author of more than twenty-five textbooks on Science and Biology as well as Biology Practical Notebooks. He has travelled to pore, Thailand, Indonesia, India, Bangladesh, UAE, Saudi Arabia, Egypt, Italy, UK, Qatar, USA and Nigeria. He has also served as a National Consultant, Education, JICA sponsored project for the promotion of Student Centred and Based (SCIB) learning, National Institute of Science and Technical Education, ry of Education, Islamabad.

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قومی ترانہ

پاک سَر زمین شاد باد! کشورِ حسین شاد باد!
تو نشان عزمِ عالی شان ارضِ پاکستان
مرکزِ یقین شاد باد!

پاک سر زمین کا نظام قوّتِ اُخوّتِ عوام
قوم ، مُلک ، سلطنت پائندہ تابندہ باد!
شاد باد منزلِ مُراد!

پرچمِ ستارہ و ہلال رہبرِ ترقی و کمال
ترجُمانِ ماضی، شانِ حال جانِ استقبال
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SECTION 6

Application of Biology

Biotechnology

26

BIOTECHNOLOGY

After completing this lesson, you will be able to

- Define gene cloning and state the steps in gene cloning.
- Describe the techniques of gene cloning through recombinant DNA technology.
- Explain the role of restriction endonucleases and DNA ligases in gene cloning.
- Describe the selection and isolation of the gene of interest.
- Explain the properties and the role of vectors in recombinant DNA technology.
- State the steps for the integration of DNA insert into the vector.
- Briefly state the technique applied for the selection of the vectors that take up the DNA insert.
- Describe the steps involved in gene amplification through polymerase chain reaction.
- Give the concept of genomic library, gene bank and probes.
- Describe the principles of Gel Electrophoresis as being used in gene sequencing.
- Explain the Sanger-Coulson method of DNA sequencing.
- Introduce the automated DNA sequencing as based on the Sanger-Coulson method.
- Describe the purposes and mechanism of DNA analysis.
- Describe the terms of genome analysis, genome map and genetic markers.
- State the history of the human genome project
- Describe the goals of the human genome project.
- Predict some of the possible benefits that can be derived after the completion of the human genome project.
- Define following terms related to plant tissue culture; explants, callus, plantlets.
- Explain tissue culture and differentiate between the organ culture and cell culture.
- Briefly describe the techniques used for animal tissue culture.
- State the objectives of the production of transgenic bacteria, transgenic plants and transgenic animals.
- Describe how biotechnologists are able to combat health problems by producing vaccines.
- State the role played by biotechnology in disease diagnosis (DNA/RNA probes, monoclonal antibodies).
- Explain the current methods employed for gene therapy (ex-vivo and in-vivo methods).
- Explain the role of successful gene therapy for cystic fibrosis.
- Explain the scope and importance of biotechnology in promoting human welfare.
- List the hazards of using genetically modified organisms.

Reading

Biotechnology deals with the application of biology and biological concepts to science and engineering for the welfare of mankind. It is the crossroad of the biological sciences with other major disciplines of science, from organic chemistry to mechanical engineering. The history of biotechnology is as old as history of man. When the first human beings realized that they could plant their own crops and breed their own animals, they learned to use biotechnology. But the term biotechnology was introduced by the end of 20th century. Now a days the field of biotechnology becomes a vast field but this chapter touches a brief part of biotechnology, called **genetic engeering**, which deals with manipulation or alteration in genetic material of an organism.

26.1 CLONING OF GENES

Gene cloning is the act of making copies, or clones, of a single gene. Once a gene is identified and cloned, it can be used in many areas of biomedical and industrial research. There are two possible ways of cloning of gene: recombinant DNA technology and polymerase chain reaction (PCR).

26.1.1 Recombinant DNA Technology

Recombinant DNA technology is a series of procedures that are used to join DNA segments from different sources. This is an *in vivo* (in living cells) method which is used when gene cloning is required at industrial scale. The main advantage of this method is the production of product of gene beside copies of gene. It involves the selection and isolation of desired gene (gene of interest), inserting it in a suitable vector and the transformation of a suitable host by the recombinant DNA.

Components / Tools of recombinant DNA technology

The cloning of gene through recombinant DNA technology requires gene of interest, molecular scissors, molecular carrier or vector, molecular glue and expression system.

i) Gene of Interest

The gene of interest is the gene which is to be cloned. It can be obtained by one of the three possible ways: (a) artificial gene synthesis is the process of synthesizing a gene *in vitro* (in glassware) without template DNA samples with the help of DNA synthesizer machine. (b) Gene of interest can also be obtained by synthesizing it from its mRNA. Synthesis of gene from mRNA is carried out by **reverse transcriptase** enzymes which are naturally found in retroviruses. The DNA formed by this process is called **complementary DNA** (cDNA). (c) In most of the cases the gene of interest is directly cleaved from a chromosomal DNA by using particular DNA scissors called restriction endonucleases.

ii) Molecular scissors (Restriction endonuclease)

Restriction endonucleases are enzymes that cleave the phosphodiester bonds of both strands of duplex DNA at specific sequences. In 1970, the first restriction enzyme was isolated. Many different restriction endonucleases have been isolated so far.

Naturally restriction enzymes are found in bacteria, where they appear to serve as host-defence role because they chop up and inactivate **("restrict")** the DNA of infecting viruses.

Each restriction enzyme cleaves DNA at specific sequence of DNA called **recognition sites** or **restriction sites**. These sites have **palindromic sequences**. A palindromic sequence is a four to eight base pairs in DNA in which nucleotides are arranged symmetrically in reverse order.

Restriction enzymes either make **staggered cut** or **blunt cut**. A staggered cut is one in which the resulting duplex fragments show single stranded projected ends called **sticky ends**. While in blunt cut the resulting duplex fragments do not show such sticky ends. In the fig 26.1, the recognition site is boxed in yellow and the cut sites indicated by red triangles.

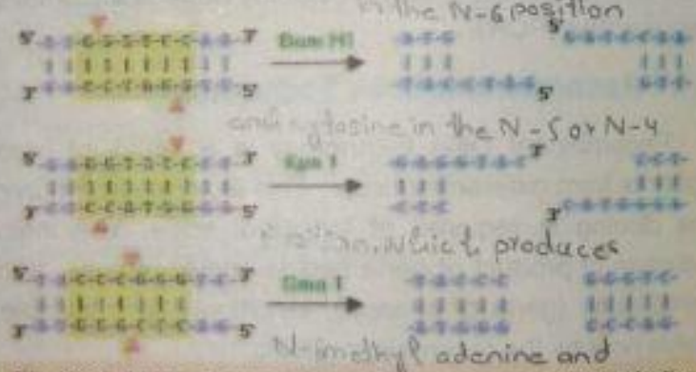


Fig. 26.1 Mode of action of restriction enzymes. (a) Production of 5′ sticky ends, (b) Production of 3′ sticky ends, (c) Production of blunt ends. Bam H-I, Kpn-I and Sma-I are different restriction enzymes

Critical Thinking

If restriction enzymes are capable of destroy the DNA of bacteriophage, why they do not chew up their host genomic DNA? The restriction enzymes that form sticky ends are more useful in genetic engineering. Why?

Science Tidbits

By convention, restriction enzymes are named after their host of origin. For example, EcoRI was isolated from *Escherichia coli*, Hind II and Hind III from *Haemophilus influenzae*, and XhoI from *Xanthomonas holcicola*.

iii) Molecular carriers or vectors

Vectors are another major component required to make a recombinant DNA (rDNA) molecule for gene cloning. Vectors act as a vehicle for carrying foreign DNA into a host cell for multiplication. Usually small circular DNA molecules of bacterial origin are used as cloning vectors. A DNA molecule should possess the following essential characteristics to act as a cloning vector: (a) Origin of replication site, (b) antibiotics resistant genes, (c) restriction sites of different enzymes. Example of vectors are: Plasmid, lambda phage DNA, Cosmid (combination of plasmid and phage DNA), Yeast artificial chromosomes (YACs) etc.

iv) Molecular glue (DNA Ligase)

This enzyme is responsible for the formation of the phosphodiester linkage between two adjacent nucleotides and thus joins two double-stranded DNA fragments; therefore it is called

(ii) Because they ensure that human DNA fragment is inserted into the plasmid in the right direction. The ligation process is fusing of DNA

molecular glue. In rDNA experiments, **DNA ligase** is used to join two different DNA fragments (plasmid/vector and the foreign DNA) that are annealed by the sticky ends.

fragments. requires less DNA when the DNA has sticky ends.

vi Expression system

A suitable organism that can act as host for the recombinant vector to express (multiplication) is called **expression system**. Therefore, the selection of suitable expression system always depends upon the type of vector which is being used while making recombinant DNA. The most important character of an ideal expression system is its short generation time and simplicity of its genetic system. So bacterial cells can act as an ideal expression system.

Mechanism of recombinant DNA technology

Cloning of the desired gene through recombinant DNA technology involves the formation of recombinant DNA (gene of interest + vector DNA), transformation of a suitable expression system by the recombinant DNA, and the identification of transformed clones.

(I) Formation of recombinant DNA

The first step in the construction of a recombinant DNA, is the isolation and purification of vectors and gene of interest. First, digest the vector DNA (e.g., plasmid) with same restriction enzyme by which gene of interest is cleaved so that compatible sticky ends can be produced. Next both, vector and gene of interest are incubated together in the presence of DNA ligase

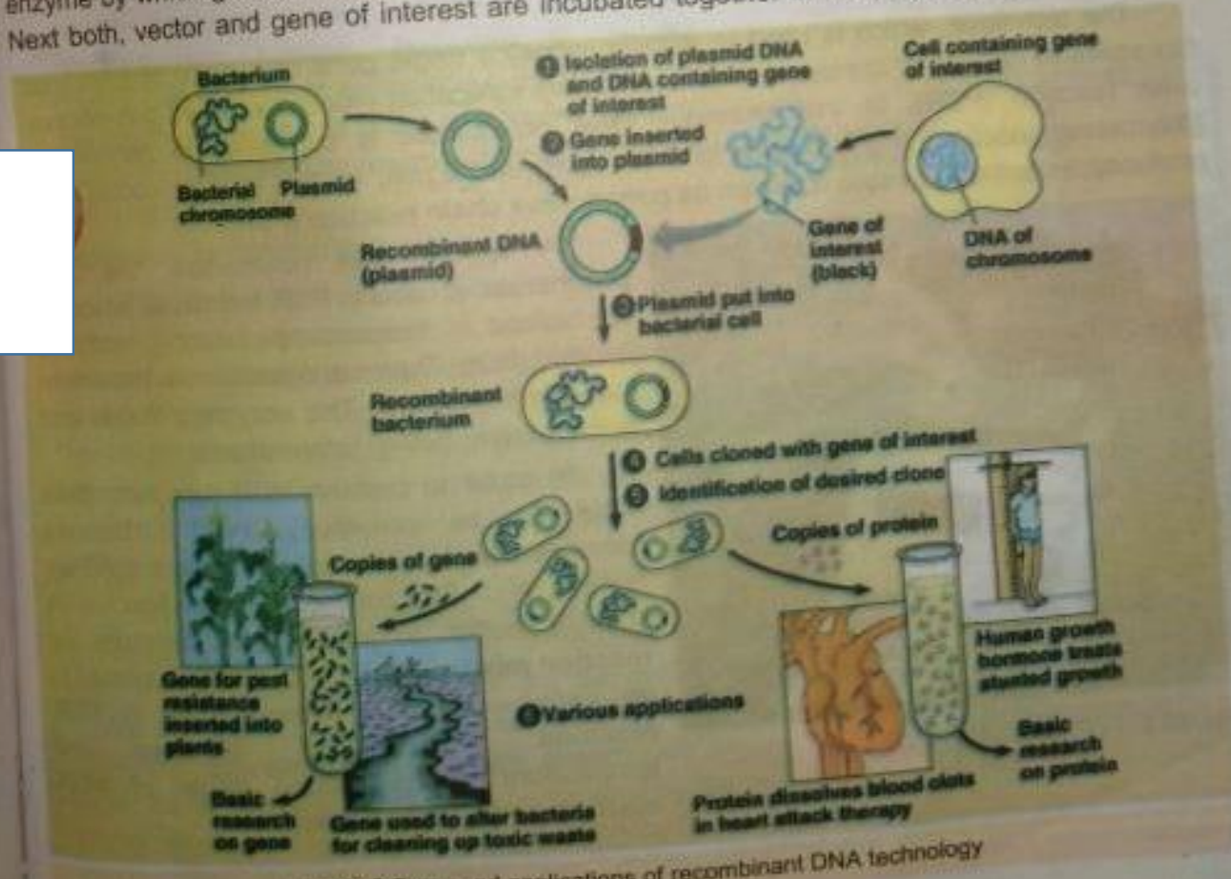


Fig. 26.2 Steps and applications of recombinant DNA technology

which connects them by forming phosphodiester linkage. This results in the formation of recombinant DNA molecule of vector and the gene of interest.

(ii) Transformation of expression system

Here transformation refers to the insertion of recombinant DNA into the expression system which can be performed by putting the expression system (bacterial cells that already contain no plasmids) and recombinant plasmids into the same medium. Bacterial cells take up recombinant plasmid, especially, if they are treated with calcium chloride which make them more permeable. Thereafter, as the cell reproduces, a bacterial clone forms and each new cell contains at least one plasmid. Therefore, each of the bacterial cell contains the gene of interest, which will express itself and make its product. ampicillin(antibiotic)

(iii) Identification of transformed clone

The transformed clone can be identified by adding a particular antibiotic (for which resistant gene is found in plasmid) into the medium. As the transformed clone has got resistance against the antibiotic, so it remains alive and continues to grow, whereas all the untransformed clones are killed by the antibiotic. From this transformed bacterial clone, the cloned gene can be isolated for further analysis or its protein product can be separated and used for various purposes.

26.1.2 Polymerase Chain Reaction (PCR)

The technique, which is used to amplify (clone) a single gene or a piece of DNA into thousands to millions of copies by means of *in vitro* replication process is called polymerase chain reaction (PCR). In this technique DNA polymerase is compelled to polymerize (polymerase reaction) a given piece of DNA again and again, so that multiple copies are produced, thus, the technique is known as **polymerase chain reaction (PCR)**.

Fig. 26.3 PCR machine (Thermocycler)

A special DNA polymerase, the *Taq* polymerase is used in PCR technique, which is specialized temperature-tolerant enzyme isolated from *Thermus aquaticus*, a bacterium found in hot springs. This enzyme is stable and active at near-boiling temperatures.

In order to perform PCR, template DNA (DNA to be amplified), free nucleotides (deoxyribo-nucleoside triphosphates or dNTPs), primers and *Taq* polymerase are dissolve in suitable buffer to make **PCR mixture** or **reaction mixture**. The PCR mixture is placed in an instrument called **thermocycler** or **PCR machine**. Thermocycler regulates the temperature during various steps of PCR reaction according to the need.

Mechanism of PCR Reaction

PCR cycle consists of three steps: **denaturation**, **primer annealing**, and **extension** or **polymerization** each requires a specific temperature. The time duration, temperature and sequence of the steps have to be programmed in the thermocycler.

Denaturation

In the denaturation step, the template is heated to 94°C for one minute. At this high temperature the DNA undergoes complete denaturation and the double-stranded DNA (dsDNA) becomes single-stranded DNA (ssDNA). Each single ssDNA can act as the template for the in vitro DNA synthesis.

Primer annealing

The next step is the primer annealing. In this step the two primers, the forward primers and the backward primers, anneal or hybridize to the single-stranded template DNA at its complementary regions. Annealing is usually carried out at a lower temperature depending on the length and sequence of the primers. In standard cases it is 54°C and approximate time required for this step is 2 minutes.

Extension or Polymerization

The final step in each cycle is the primer extension or polymerization in which the *Taq* polymerase synthesizes new DNA strands to the 3' ends of primers using dNTPs. The optimum temperature for carrying out the primer extension reaction or polymerization of dNTPs is standardized at 72°C. This step takes just one minute to be completed.

At the end of first cycle one target DNA molecule is converted in to two molecules. The second cycle immediately starts with the denaturation by heating at 94°C, so that all the newly synthesized DNA are also denatured to single strands, which again act as templates. It will again be followed by the primer annealing and extension and thus the cycle of denaturation, primer annealing, and extension

Science Tidbits

PCR Technique was invented by Kary Mullis in 1983; later on he was awarded the Nobel Prize in Chemistry in1993

Critical Thinking

1. Why heat is used in PCR technique to denature the target DNA instead of using DNA helicase and DNA gyrase enzymes?
2. Why human DNA polymerase cannot be used in PCR technique?
3. Why already synthesized primers are used in PCR technique instead of using primase

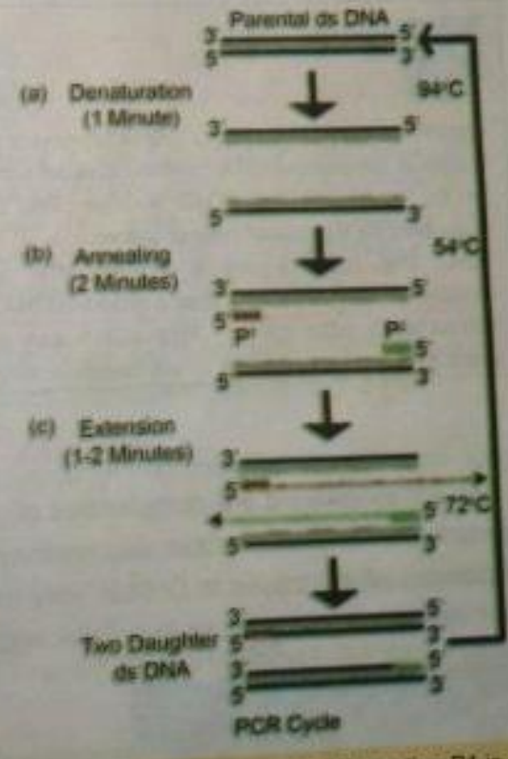


Fig. 26.4 Mechanism of PCR reaction P1 is forward primer and P2 is backward primer

continues resulting in the amplification of the selected DNA sequence at an exponential rate i.e., the number of existing DNA molecules become doubled after each cycle.

The application of polymerase chain reaction.

PCR has application in almost all areas of molecular biology, genetics, and in clinical areas.

1) PCR is an efficient diagnostic technique used for the detection of specific genotypes of infectious agents.
2) Reactions for DNA sequencing are also simplified by introducing the PCR method.
3) DNA fingerprinting is also made simple by PCR as described above.
4) The genetic mutations responsible for certain genetic diseases and cancers can be detected using PCR tools. Early detection of genetic disease is even possible in embryonic conditions or even in sex cells—sperm and egg

26.1.3 Genomic Library

Genome: a complete set of gene of an individual.

A genomic library is a collection of bacterial or bacteriophage clones, each containing at least one copy of every DNA sequence in a genome of an organism. In single library, the entire genome of an organism is represented as a set of DNA fragments inserted into a vector molecule. Collection of genomic libraries of different organisms is called gene bank.

A specific complementary **probe** is used to search a particular gene of interest from genomic library if it is required for further analysis, A DNA probe is a small, fluorescently or radioactively labelled single stranded DNA molecule.

Complementary DNA Library (cDNA Library) is the collection of clones of DNA, which are the complementary copies of messenger RNA (mRNA) isolated from the particular cells. Messenger RNA is the starting material for the construction of cDNA libraries. This DNA library represents only those genes which are being expressed by a group of cells or tissues.

Since the mRNAs are produced after splicing, they are devoid of introns. Therefore, the complimentary copy of these mRNAs (cDNA) represents only the exons or the coding regions of the actual eukaryotic genes. This cDNA can be directly inserted into an expression vector and the protein can be expressed in the bacterial systems.

26.2 DNA SEQUENCING

To understand the complexities of gene structure, its expression, its regulation, protein interactions, and molecular mechanisms of genetic diseases, the detailed and exact sequences of the bases in DNA is very essential. One of the important tools used in various DNA sequencing techniques is gel electrophoresis.

Teacher's Point

The teacher would tell the students that PCR is used to make multiple copies of genes then ask "How many copies of the DNA will be after five cycles if the PCR starts with single template DNA duplex".

26.2.1 Gel Electrophoresis

Gel electrophoresis is a technique used in molecular biology to separate different sized fragments of charge bearing polymers (proteins, RNA or DNA) under the influence of electric field in a semisolid gel medium of agarose or polyacrylamide. The molecules being sorted are dispensed into a well in the gel material. The gel is placed in an electrophoresis chamber, which is then connected to a power source. When the electric current is applied, the different sized molecules begin to move to the opposite pole through the gel.

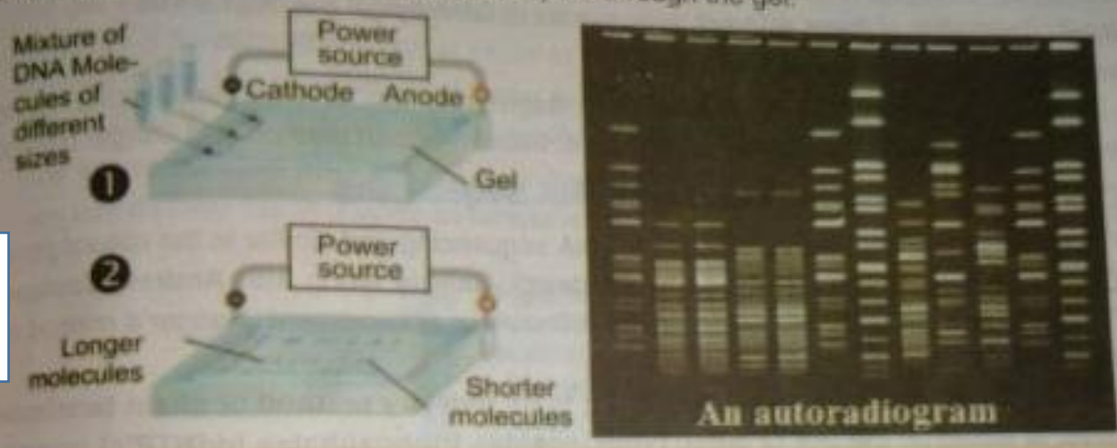


Fig.26.5 Gel electrophoresis

Principle of movement in the gel

The movement of the fragments is primarily dependent upon size because the distance, a DNA fragment travels, is inversely proportional to its length so the smaller fragments move faster through the gel matrix than larger fragments. However, the movement of the fragments also depends upon charges, number of strands (single or double) and shape of the molecules (linear or circular) and the concentration of the gel (pore size).Therefore, after sometimes the different sized molecules have been separated into distinct bands on the gel.

Science Tidbits

The types of gels most commonly used for DNA electrophoresis are agarose (for relatively large DNA molecules i.e., more than 50 nucleotide) and polyacrylamide (for high resolution of short DNA fragments i.e., less than 50 nucleotide).

Visualization of fragments

To visualize DNA or RNA, the gel is placed on an ultraviolet transilluminator. Now you can observe that some bands are thick and some are thin, thick bands represent the high concentration of same sized fragments while thin bands show low concentration.

If a particular sized fragment is to be used for further analysis, the piece of gel containing that band can be cut and its DNA can be purified again. DNA bands can also be transferred from gel to the nitrocellulose membrane for autoradiography (X ray imaging). Be aware that

Teacher's Point

The teacher would ask the students that how do DNA probes help to identify individuals"?

DNA will diffuse within the gel over time, and examination or photography should take place shortly after cessation of electrophoresis.

26.2.2 Major Steps in DNA Sequencing Techniques

The mechanism of any DNA sequencing method is based upon three steps:

Step-1: To generate piece of DNA of different sizes all starting from the same point and ending at different points.

Step-2: Separation of these different sized pieces of DNA by gel electrophoresis.

Step-3: Reading of sequence from the gel.

For generation of different sized DNA fragments, two different sequencing methods were developed during the late 1970s. They are: Maxam-Gilbert method and Sanger method.

26.2.3 Sanger – Coulson Method of DNA Sequencing

This method is widely used method of DNA sequencing and similar to the natural process of DNA replication. It was developed by **Frederick Sanger** along with **Andrew Coulson** in 1977. They were awarded Nobel Prize in 1980 on this achievement. Sanger's method now became the standard because of its practicality.

Sanger's method, which is also referred to as **dideoxy method** or **chain termination method**, is based on the use of **dideoxynucleosides triphosphates (ddNTP's)** commonly known as **dideoxynucleotides** in addition to the normal nucleotides (dNTP's) found in DNA. Dideoxynucleotides are essentially the same as common nucleotides except, they contain a hydrogen group on the 3′ carbon instead of a hydroxyl group (OH). These modified nucleotides, when integrated into a sequence, prevent the addition of further nucleotides. This occurs because an OH group is required at 3′ end of growing chain in order to make phosphodiester bond with next incoming nucleotide. In this way they are used to terminate replication processes.

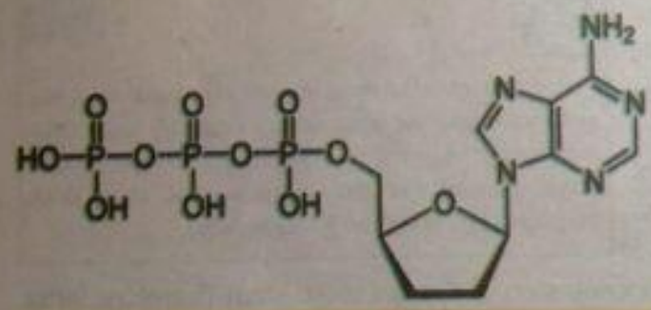


Fig.26.6 (a) Dideoxyribonucleotide

Procedure

(1) Before the DNA can be sequenced, it has to be denatured into single strands using heat because only one strand that acts as template is required in this procedure. Now the template strand is tagged with a known sequence at 3′ end, so that a complimentary primer can bind on the known sequence.

(2) Tagged target DNA, primers, normal free nucleotides and Taq polymersase dissolved in an appropriate buffer are equally added in four separate tubes each containing a different dideoxynucleotide. Now all these test tubes are placed in PCR machine so that sequencing reaction can start. As the DNA is synthesized, nucleotides are added

on to the growing chain by the DNA polymerase. However, on occasion a dideoxynucleotide is incorporated into the chain in place of a normal nucleotide which results in a chain-terminating event. For example in the tube contusing ddATP, only those fragments will be produced that will terminate on "A". Same mechanism takesplace in other tubes.

(3) Once these reactions are completed, the DNA is once again denatured in preparation for gel electrophoresis. The contents of each of the four tubes are run in separate lanes on a polyacrylamide gel in order to separate the different sized bands from one another. After the contents have been run across the gel, the gel is then exposed to either UV light or X-Ray, depending on the method used for labelling the DNA.

(4) The sequence read from the gel is complementary to the actual template DNA. Now you can deduce the sequence of template DNA.

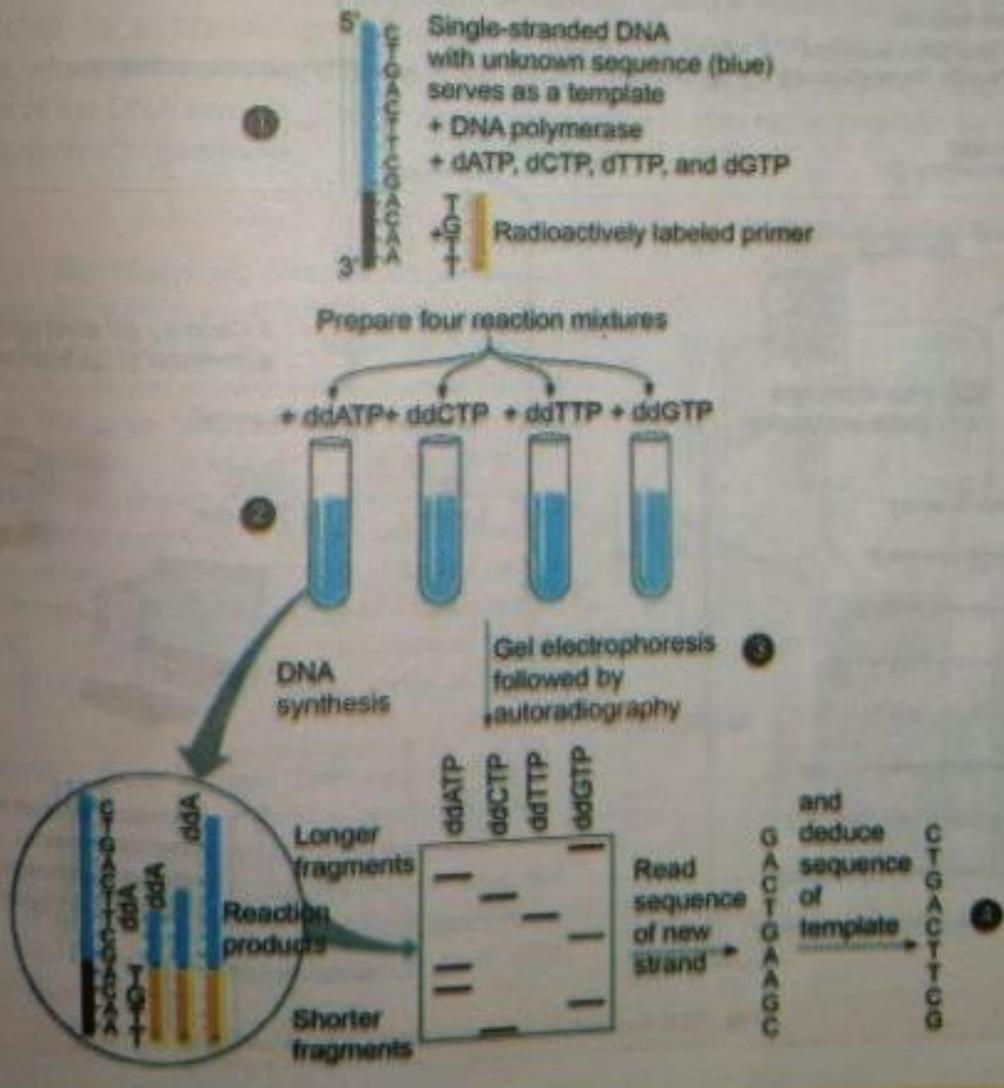


Fig. 26.7 Sanger's method

Science Trivia

Although Maxam and Gilbert published their chemical sequencing method two years after the ground-breaking paper of Sanger and Coulson on plus-minus sequencing. Maxam–Gilbert sequencing rapidly became more popular, since purified DNA could be used directly while the initial Sanger method required that each read start be cloned for production of single-stranded DNA. However, with the improvement of the chain-termination method, Maxam-Gilbert sequencing has fallen out of favour due to its technical complexity prohibiting its use in standard molecular biology kits, extensive use of hazardous chemicals, and difficulties with scale-up.

26.2.4 Automated DNA Sequencing

Automatic sequencing machines have greatly improved the quality as well as the speed of the sequencing process. The basic principle of sequencing is quite same in manual and automated DNA sequencing except few differences.

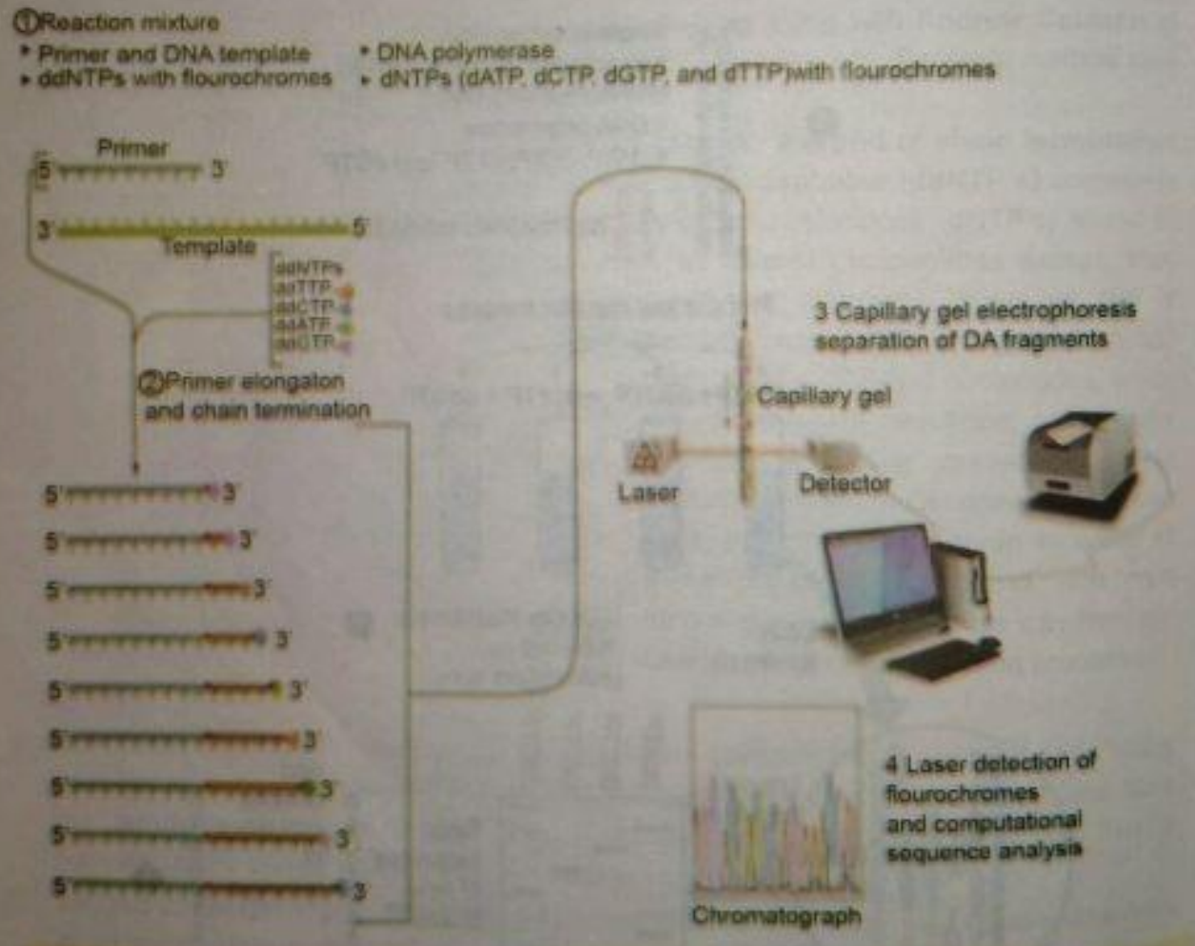


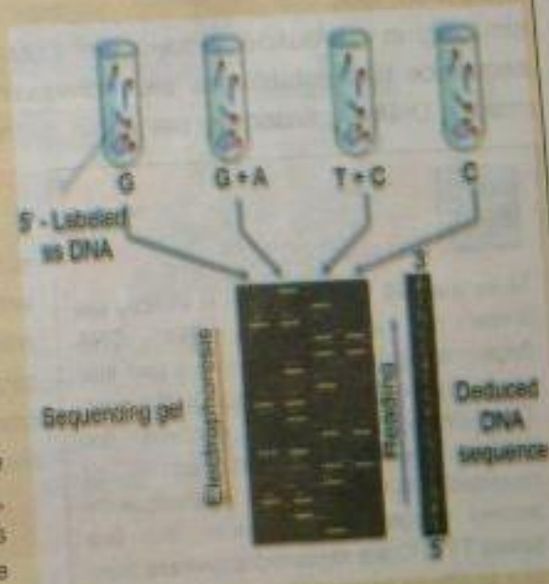
Fig. 26.8 Procedure of automated DNA sequencing

There is no need for radiolabeling and autoradiography. The use of fluorescently labelled ddNTPs (dideoxynucleotide triphosphates) has made the reading very easy, convenient, and automatic with the help of UV laser detectors. Thus, it has greatly improved the speed of sequencing. Each of the four types of ddNTPs can be labelled with a specific dye, so that a specific colour can be attributed to the presence of a particular nucleotide or base. Since fluorescent dyes of four different colours are used so there is no need to perform reaction in separate tubes, all 4 reactions can be run in the same tube which greatly increase the speed and ease of sequencing. There is no need to run the reaction mixtures in separate lanes of electrophoresis gel during the separation of different sized fragments after sequencing reaction. The reaction mixtures can be electrophoresed on a single lane instead of four by using **capillary array electrophoresis**. After electrophoresis, we don't even have to 'read' the sequence from the gel. The computer does that for us. After electrophoresis the coloured bands can be monitored using UV-laser beams. The laser beams excite the fluorescent dyes and result in the emission of specific spectral waves (coloured light), which are recorded by a specific photoelectric device. The data thus generated is fed to a computer, where the emission data from the gel is converted into a corresponding nucleotide sequence of the DNA sample. The nucleotide sequence is also represented in specific peaks indicating each nitrogen base.

Maxam-Gilbert Method of DNA Sequencing (Extra Reading Material)

In 1976-1977, Allan Maxam and Walter Gilbert developed a DNA sequencing method which is also called chemical cleavage method because it is based on chemical modification of DNA and subsequent cleavage at specific bases. The DNA to be sequenced must be amplified by using PCR technique. The multiple copies are denatured first so that the two strands can be separated from each other, one strand of each copy is purified and labelled with isotopic phosphate at 5' end. These labelled, single stranded DNA are divided into four samples and are tagged as G, A+G, T+C, and C. Each tube contain certain chemicals which cleave phosphodiester bonds at specific point.

In the sample "G" the unknown DNA fragment will be cleaved from all those point where G is present. Similarly, in sample "A+G" the fragment will be cleaved from A as well as from G and so on in other samples. After the completion of reactions, the products are run through gel electrophoresis and finally the sequence is read from autoradiogram of gel pattern.



26.3 DNA ANALYSIS

DNA profiling (also called **DNA testing**, **DNA typing**, or **genetic fingerprinting**) is a technique employed by forensic scientists to assist in the identification of individuals by their respective nucleotide sequence of DNA. This method was emerged in the 1980s. The first DNA fingerprint was made in 1985.

26.3.1 Purposes/Applications of DNA Analysis

Today DNA analysis has wide range of application in different fields of life. It can be used to: (a) Identify potential suspects who's DNA may match evidence left at crime scenes. (b) Identify crime and catastrophe victims. (c) Establish paternity and other family relationships. (d) Detect bacteria and other organisms that may pollute air, water, soil and food. (e) Match organ donors with recipients in transplant programs.

26.3.2 Mechanism/Procedure of DNA analysis

There are several techniques that can be used for DNA analysis. **Restriction Fragment Length Polymorphism (RFLP)** was one of the first methods used in DNA analysis. RFLP (pronounced as "rif-lips") refers to the different sized fragments of DNA produced by a particular restriction enzyme. Every person has a unique set of RFLPs because the restriction site of a particular enzyme is always different in number and distribution in all human on earth except the monozygotic (identical) twins. Therefore, RFLPs of any two persons, when compared, one can easily analyse their individuality. However, the entire human have 99% similarity in nucleotide sequence of their genomes, this is the only 1% difference in genome sequence that establishes the individuality of every person. Following are the key steps to make a DNA fingerprint by using this method.

Science Titbits

More than 99 % of the DNA is exactly the same in all humans. But DNA fingerprinting focuses only on the part that tends to differ from one person to the next. Throughout the human genome are **tandem repeats** - short regions of repeated DNA- that differ subsequently among people. For example, the five bases TTTTC are repeated anywhere from four to fifteen times in tandem in different people, and the bases (CGC) are repeated five to fifty times in tandem. By examining many tandem-repeat sites, researchers found out that each person carries a unique combination of repeat numbers.

Collection of DNA samples

For DNA analysis, very small fraction of DNA is sufficient because it can be amplified several times with the help of PCR. Therefore, it can be collected even from a small trace of blood or from the cells of single hair root. DNA samples can also be collected from mummified organisms or from fossils when evolutionary relationship has to be studied.

Placement and separation of RFLP

Placement of RFLP is the digestion of DNA samples by a particular restriction enzyme which produces a set of different sized DNA fragments (RFLPs). The mixture of RFLPs is loaded in polyacrylamide gel and run for electrophoresis; fragments of various lengths begin to move at different rate from negative to positive pole within the gel. When

the movement is stopped, the gel is proceeded for further treatments in order to observe banding pattern.

Denaturation of RFLP fragments

In this step, first, the electrophoresed gel is placed into an alkaline solution (typically containing sodium hydroxide) to denature the double-stranded DNA. The denaturation may improve binding of the negatively charged DNA to a positively charged membrane and separating it into ssDNA for later hybridization to the probe.

Blotting

In this method, a sheet of nitrocellulose (or alternatively nylon) membrane is placed on top of the gel. Ion exchange interactions bind the ssDNA to the membrane due to the negative charge of the DNA and positive charge of the membrane. The membrane is then baked in a vacuum or regular oven at 80 °C for 2 hours to permanently attach the transferred DNA to the membrane.

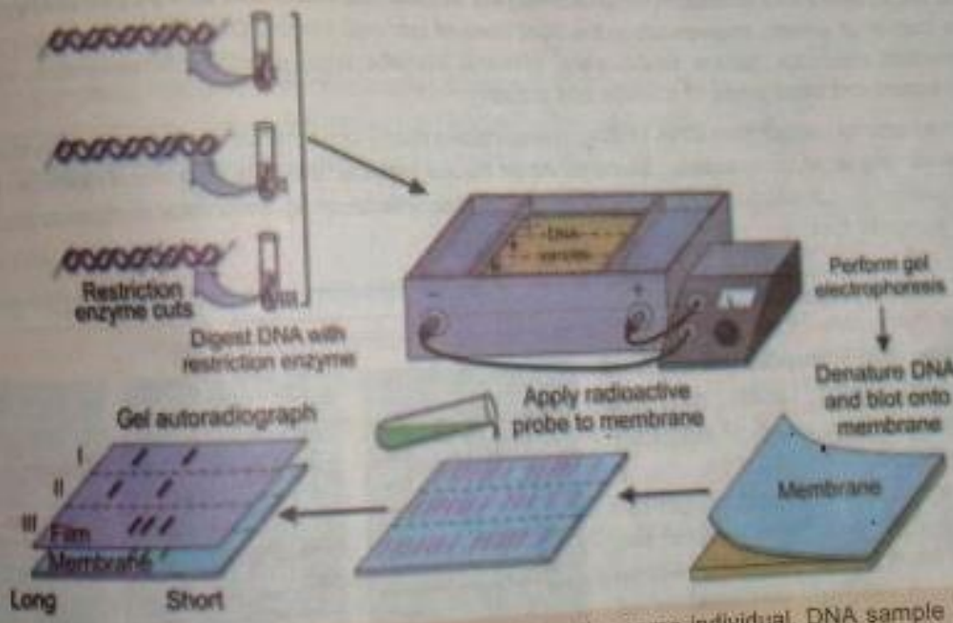


Fig. 26.9 RFLP analysis. DNA samples I and II are from the same individual. DNA sample III is from a different individual. Notice, therefore, that the restriction enzymes cuts are different for sample III. Gel electrophoresis separates the DNA fragments according to their length because shorter fragments migrate farther in an electrical field than do longer fragments. The fragments are denatured and transferred to membrane where a radioactive probe can be applied. The resulting pattern (the DNA fingerprint) can then be detected by autoradiography.

Teacher's Point

The teacher would ask the students to "Explain why RFLPs can serve as genetic markers even though they produce no visible phenotypic differences. *Hint*: RFLPs are inherited in Mendelian fashion and variation in RFLPs among individuals can be detected by southern blotting.

Labelling of RFLP fragments

The membrane is then exposed to radioactive probes which hybridize with denatured DNA (ssDNA) fragments in all bands. Since, the DNA probes are radioactively labelled so they can be detected by autoradiography.

Autoradiography

After hybridization, excess probes are washed from the membrane and the pattern of hybridization is visualized on X-ray film by exposing the membrane to an X-ray source. This technique is known as autoradiography. The banding pattern which was originally obtained in the gel due to the separation of RFLPs, is now developed on an X-ray film.

Science, Technology and Society Connections

State the importance and limitation of DNA analysis in forensic medicine and palaeontology

From the frontier of genetic engineering to the front lines of criminal justice, DNA testing has become a crucial tool in medical research, patient health care, criminal identification, paternity determination, evolutionary history of fossils and other areas of science and industry.

Despite the benefits gained from DNA testing, it also raises many contentious issues in the private sector and the academic and legal communities. Some of these issues include: the lack of uniform regulation for quality control, violation of constitutional rights to privacy and discrimination by insurance companies based on an individual's genetic history.

Skills: Analyzing, Interpreting and Communication

- **Analyze and interpret the DNA of a child with that of two individuals in a case of disputed parenthood.**

Parental testing is the use of genetic fingerprinting to determine whether two individuals have a biological parent-child relationship. A paternity test establishes genetic proof whether a man is the biological father of an individual, and a maternity test establishes whether a woman is the biological mother of an individual. The current techniques for paternal testing are using PCR and RFLP.

Here is a child's paternity and maternity can be clearly seen written in its DNA profile using three different restriction enzymes. Half of the child's (c) makers come from its mother (m), and half from its father (f). An unrelated individual is shown in the last lane (u)

26.4 GENOME MAPS

The **genome** is a collection of all the genes found in one complete set of chromosome. So a diploid organism has two copies of genome while egg or sperm has one.

26.4.1 Genome Analysis

Just like the road maps and street maps of a city, which guide us to reach a specific location, the **genome analyses** are used by the scientists searching for a specific gene somewhere within the vast genome.

Due to rapid development of genome studies, a new branch of biotechnology has emerged called **genomics** which deals with exploration and analysis of complete DNA sequence of an organism's genome.

Genome maps

These analyses help to develop two broad categories of maps: **genetic maps** and **physical maps**, which are being used for genome analyses. A genetic map shows the sequence of gene loci along the length of chromosomes while physical map represents the sequence of nucleotides in the DNA.

Science Tidbits

VNTRs, or variable number of tandem repeat polymorphisms, occur in non-coding regions of DNA. This type of marker is defined by the presence of a nucleotide sequence that is repeated several times. In each case, the number of times a sequence is repeated may vary.

Genetic markers

Just like interstate maps have cities and towns that serve as landmarks, genetic maps have landmarks known as **genetic markers**, or **"markers"** for short. Examples of markers are RFLPs, variable number of tandem repeats, short tandem repeats and single nucleotide polymorphism.

26.4.2 The Human Genome Project (HGP)

The Human Genome Project (HGP) is an international scientific research project which is based on the exploration and analysis of human genome.It was originally founded by the U.S. Department of Energy and the National Institutes of Health in 1990.

Historical background

Due to widespread international cooperation and advances in the field of genomics (especially in sequence analysis), as well as major advances in computing technology, a **'rough draft'** of the genome was finished in 2000. On-going sequencing led to the announcement of the essentially complete genome in April 2003, 2 years earlier than planned, but the sequence of the last chromosome was published in the journal Nature in May 2006. Finally it came to know that human genome comprises **3.2 billion** nucleotides and approximately **20,000-25,000 genes**.

Science Tidbits

In 1090, US government had established National Human Genome research Institute (NHGRI), who had completed HGP in 2003. James D. Watson was appointed as first director of this institute but at the time of completion of the project, the institute was being led by Dr. Francis Collin.

James Watson

Teacher's Point

The teacher would ask the students to evaluate the potential impact on the Human Genome project on both scientific thought and society.

Major goals and benefits of HGP

Major goals and objective of this project were to (a) construct genetic and base sequence map of human genome (b) store this information in databases, (c) improve tools for data analysis, (d) transfer related technologies to the private sector, and (e) address the ethical, legal, and social issues (ELSI) that may arise from the project. Human genome project can benefit in improved diagnosis of disease, earlier detection of genetic predispositions to disease, rational drug design, gene therapy, control systems for drugs etc.

Science, Technology and Society Connections

Justify why the human genome project is regarded as the most ambitious project ever undertaken by man.
Although the Human Genome Project was initially started by the U.S. government in 1990 but later on Welcome Trust (U.K.) became a major partner; additional contributions came from Japan, France, Germany, China, and others. Since the current applications of genome research address national needs in molecular medicine, waste control and environmental clean-up, biotechnology, energy sources and risk assessment. Therefore HGP is regarded as the most ambitious project ever undertaken by man.

Science, Technology and Society Connections

Describe the major findings that have arisen from the human genome project.
Some of the findings of this project are: (1) Repetitive sequences are stretches of DNA sequences that are repeated many times (2) Chromosome 1 has most genes (2968) and chromosome Y has least number of genes. (3) The function of over 50% of discovered genes are unknown.(4) The count of no. of nucleotides.(5) Number of genes (6) In light of the findings of the Human Genome Project that we actually have less genes in our genome, or on our chromosomes, than originally thought.

26.5 TISSUE CULTURE

Growth of single cell or group of cells in a glassware on artificial medium under aseptic conditions is called **tissue culture**. Many somatic plant cells, including some fully differentiated types (e.g., leaf mesophyll), contain intact nuclear, plastid and mitochondrial genomes and have the capacity to regenerate into whole plants. This phenomenon is **totipotency**.

26.5.1 Methods of Plant Tissue Culture

Tissue culture is often a generic term that refers to both organ culture and cell culture. The initial plant part which is used to develop tissue culture is called **explant**. It may be complete organ (seed, leaf, and twig) or single cell (protoplast) or a piece of tissue. **Plantlets** are young plants which are developed during tissue culturing. On the basis of explant tissue culture is variously called **cell culture** or **organ culture**.

There are several methods of tissue cultures have been developed which are primarily based upon type of explant used e.g., meristem, anther, ovary, embryo culture etc.

26.5.2 Animal Cell Culture

Unlike plant and microbial cells, the animal cells can grow only to a limited generations even in the best nutritive media. This growth also depends on the sources of tissue isolated. For example, neurons cannot divide and grow while fibroblast can divide and grow in culture to

some generations. After completing several generations they die. These animal cells cultures are used in recombinant DNA technology, genetic manipulations, cancer research and in a variety of industrial processes such as production of vaccines, monoclonal antibodies, pharmaceutical drugs etc.

There are two major techniques of animal cell cultures i.e., anchorage-dependent and anchorage-independent. Adherent cells are **anchorage-dependent** and propagate as a monolayer attached to the cell culture vessel. Most cells derived from tissues are anchorage-dependent. Since these cells grow for limited generations so they are also called **finite cell line**. Suspension cells can survive and proliferate without being attached to a substratum, therefore, called **anchorage-independent**. Hematopoietic cells (derived from blood, spleen, or bone marrow) as well as some transformed cell lines and cells derived from malignant tumours can be grown in suspension. Since these cells grow for unlimited generations so they are also called **continuous cell lines**.

26.6 TRANSGENIC BACTERIA, PLANTS AND ANIMALS

The free living organisms in the environment that have had a foreign gene inserted into are called **"genetically modified (GM)," "genetically engineered (GE),"** or **"transgenic organisms."** Bacteria were the first transgenic organisms, first transgenic bacterium was produced in 1978. Many transgenic organisms such as animals, plants, and bacteria have been produced.

26.6.1 Transgenic Bacteria

Unlike other organisms bacteria can be easily transformed due to their simple genetics. The first example of this occurred in 1978 when version of the human insulin gene was inserted into the bacterium *Escherichia coli* to produce synthetic "human" insulin. The transgenic bacteria are not only being used to produce different human proteins, they are also being used in improvement of plant growth, removal of environmental pollutants and extraction of metals from low grade ores.

26.6.2 Transgenic Plants

The first field trials of genetically engineered plants occurred in France and USA in 1986, when tobacco plants were engineered to be resistant to herbicides. In most cases the aim of developing transgenic plant is to

Both strains of *Pseudomonas syringae* occur naturally, but recombinant DNA technology has allowed for the synthetic removal or alteration of specific genes, enabling the creation of the ice-minus strain. Modifying *P. syringae* may have unexpected consequences for climate. A study has shown that its ice nucleating proteins may play an important part in causing ice crystals to form in clouds. If humans increase the frequency of bacteria lacking these proteins then it could effect cloud cover.

The first genes available for genetic engineering of crop plants for pest resistance were known as Bt genes from a bacterium ***Bacillus thuringiensis***. These are specific to particular group of insect pests and are not harmful to other useful insects like butter flies and silk worms. Transgenic crops with Bt genes (e.g. cotton, rice, maize, potato, tomato, brinjal, cauliflower, cabbage, etc.) have been developed. This has proved to be an effective way of controlling the insect pests and

roduce a new trait to the plant which does not occur naturally in this species. Examples clude resistance to certain pests, diseases or environmental conditions or the production of a rtain nutrient or pharmaceutical agent.

6.3 Transgenic Animals

A transgenic animal is one that carries a foreign gene that has been deliberately inserted to its genome. Genetic engineering has also been used to improve the traits of farm animals. addition, these animals are also used to produce drugs. Such transgenic animals which also oduce drugs are called **transpharmer animals**.

cience, Technology and Society Connections

Predict the application of genetic engineering in crop improvement.

There has been a consistent increase in the global area planted to transgenic crops from 1996 to 2005. About 90 Mha was planted in 2005 to transgenic crops with high market value such as herbicide tolerant soybean, maize, cotton, and canola; insect resistant maize, cotton, potato, and rice; and virus resistant squash and papaya. With genetic engineering, more than one trait can be incorporated into a plant. Transgenic crops with combined traits are also available commercially. These include herbicide tolerant and insect resistant maize and cotton.

26.7 BIOTECHNOLOGY AND HEALTHCARE

Biotechnology has made a huge difference in human health care and has now enabled cientists to develop products which can give quicker and more accurate tests, therapies that ave a lot less side effects and vaccines which are safer than ever before.

6.7.1 Role of Biotechnology in Treatment and Diagnosis of Diseases

Biotechnology is used in three different ways in the development of vaccine: (a) eparation of a pure antigen using a specific monoclonal antibody. (b) Synthesis of an antigen ith the help of a cloned gene. (c) Synthesis of peptides to be used as vaccines.

Many human diseases can be diagnosed by using products of biotechnology like onoclonal antibodies and DNA/RNA probes.

6.7.2 Gene Therapy

Gene therapy is a technique for correcting defective genes responsible for disease evelopment. Researchers may use one of several approaches for correcting faulty genes, like normal gene may be inserted into a nonspecific location within the genome to replace a non-nctional gene. This approach is most common.

echanism of gene therapy

In gene therapy treatment, normal gene is either delivered directly into the body *(in vivo)* into the cells outside the body then these transgenic cells are again implanted into the body

Teacher's Point

The teacher would ask the students to "Explain how a transgenic plant differs from a hybrid plant?

(ex vivo). In both cases, a "normal" gene is inserted into the genome to replace an "abnormal," disease-causing gene. A carrier molecule called a vector must be used to deliver the therapeutic gene to the patient's target cells. Currently, the most common vector is a virus that has been genetically altered to carry normal human DNA.

Some of the different types of viruses used as gene therapy vectors are Retroviruses, Adenoviruses, Herpes simplex viruses.

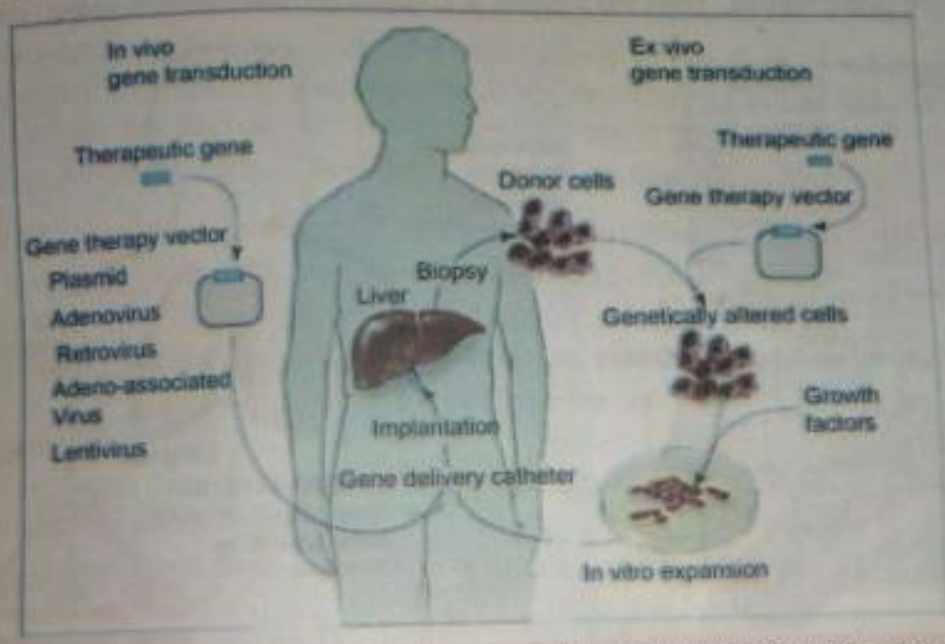


Fig. 26.10 Mechanism of gene therapy: An in vivo approach to gene therapy delivers the therapeutic nucleic acid directly to the patient. The gene is packaged in one of several vectors and delivered with a device to a target organ. In the illustration (left), the gene is incorporated into a plasmid and delivered to the liver via a catheter in the portal vein. An ex vivo approach involves harvesting cells from the tissue of interest, transducing them with a gene in vitro, and re-administering the genetically altered cells to the patient. Gene transduction in vitro may be mediated by the same vectors as those used in ' in vivo ' gene transduction.

26.7.3 Role of Gene Therapy for Cystic Fibrosis

Cystic fibrosis is an inherited disease which affects the mucus and sweat glands. People with severe symptoms can have serious lung and digestive problems.

Cystic fibrosis (CF) involves a defect in the **cystic fibrosis trans membrane conductance regulator (CFTR)** gene that encodes a protein by which the movement of salt and water is controlled in and out of body cells. In people with cystic fibrosis, the gene does not work effectively. As a result, cells that line the passageways of the lungs, pancreas and other organs produce abnormally thick, sticky mucus. This mucus obstructs the airways and glands which causes the characteristic signs and symptoms of cystic fibrosis. On the other hand, in normal persons, mucus is watery. It keeps the linings of certain organs moist and prevents them from drying out or getting infected.

Gene Therapy of Cystic fibrosis

In 1989, experts discovered the gene that causes cystic fibrosis and identified it as the cystic fibrosis trans membrane conductance regulator or CFTR. The discovery of this defective gene posed new possibilities of a cure.

An in vivo method of treatment is being tried. Liposomes-microscopic vesicles that spontaneously form when lipoproteins are put into a solution have been coated with the gene needed to cure cystic fibrosis. Then the solution is sprayed into the patient's nostril.

Fig. 26.11 Effects of cystic fibrosis

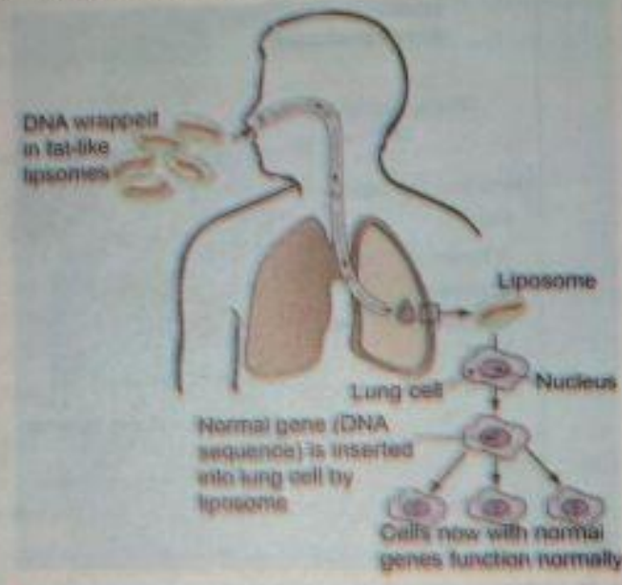


Fig. 26.12 An attempt of in vivo gene therapy for cystic

26.8 SCOPE AND IMPORTANCE OF BIOTECHNOLOGY

Biotechnology is "the science of applied biological process". It is one of the fastest growing field in the area of research and development. It is also called a **technology of the future** or **technology of tomorrow** because of its unprecedented impacts on the human and the universe as a whole.

26.8.1 Scope and Importance of Biotechnology in Promoting Human Welfare

Biotechnology is going to be very important in our daily life with the passage of time. The ultimate objective of biotechnology is to provide welfare for human being.

In future, there is the possibility of developing of **biological computers or biochips.**

Teacher's Point

The teacher would ask the students that "What is the advantage of using stem cells for gene therapy? *Hint:* stem cells continue to reproduce themselves.

essentially miniaturized laboratories can perform hundreds or thousands of simultaneous biochemical reactions. Biochips enable researchers to quickly screen large numbers of biological analysis for a variety of purposes, from disease diagnosis to detection of bioterrorism agents.

In recent years, use of microbial inoculants as a source of **biofertilizers** (nutrient inputs of biological origin for plant growth) has become a hope for most of countries, as far as economic and environmental viewpoints are concerned.

A new and exciting sub-branch of biotechnology is the field of **nanotechnology**. Nanotechnologists are imparting their expertise in the development of such nano particle that can be used for efficient drug delivery at the target cells and in the diagnosis of diseases.

28.2 Concerns about the Genetically Modified Organisms (GMOs)

The main areas of consideration for safety aspects in biotechnology are the following:

(a) How to dispose-off spent microbial biomass and purify the effluents from biotechnological processes?

(b) The toxicity of the allergy associated with microbial production.

(c) How to deal with the increase in the number of antibiotic resistant pathogenic microorganisms?

(d) How to evaluate the pathogenicity of the genetically engineered microorganisms to infect humans, plants and animals?

(e) How to prevent contamination, infection or mutation of the processed strains?

Science Tidbits

Most of the countries of the world are signatories to the Biological Weapons Conventions of 1972. As a signatory, it is a voluntary pledge by a nation "never to produce microbial or other biological agents or toxins, whatever may be their method of production, for use in wars. However, many people have expressed their concerns about the possible use of genetic manipulations for military purposes in the near future.

Science, Technology and Society Connections

- **Justify the need of genetic counselling.**

Genetic counselling is a service that provides information and advice about genetic conditions. Counselling is conducted by healthcare professionals who have been specially trained in the science of human genetics (a genetic counsellor or a clinical geneticist). The counsellor will discuss the risks, benefits and limitations of genetic testing with you. They will also explain how the information found as a result of genetic testing could have implications for both you and your family.

- **Investigate careers that require an understanding of biotechnology and genetic engineering.**

Many careers require an understanding of biotechnology and genetic engineering. Such as: Healthcare professionals, Teachers of biological sciences, biomedical engineers, crime lab analyst, crime scene investigator, environmental impact analyst, forensic scientist, genetic engineer, molecular biologist etc.

- **Describe briefly the accomplishments of the renowned genetic engineers working in private and public sector institutions in her or his province.**

It is advised to the administration that they should arrange study tour of student to public and privet sector institutions of genetic engineering so that students can meet with renowned genetic engineers and can directly ask about their accomplishments.

- **Suggest measure she/he would take to solve related problems by using knowledge gained in this chapter.**

Students should evaluate themselves to know what they have learnt in this chapter and should apply this knowledge to solve related problems under the guidance of their class teacher.

- **Describe the role of Genetic Screening.**

Genetic screening includes all those diagnostics tests which are used to determine whether a person or a new born baby is at risk of genetic diseases or not. Generally there are two types of genetic screening, screening of children and adults, and screening of unborn children. Genetic screening of children and adults has two purposes: first it can confirm whether the person has a mutated gene of certain disease or characteristics. The second purpose is to test adults to see if their children will be at risk of certain disease. Knowing that one or both parents carries a dominant allele for a genetic disease might affect the decision parents make about having children , sometimes this kind of genetic screening is used for approval of marriage licenses in some countries, like Denmark.

Activity

1. Make a model of DNA probe and restriction enzymes.

Exercise

M.C.Qs

Select the correct answer

(i) Which of the following tools of recombinant DNA technology is incorrectly paired with its use?

(A) restriction enzyme --- production of RFLPs

(B) DNA ligase --- enzyme that cuts DNA, creating the sticky ends of restriction fragments.

(C) reverse transcriptase --- production of cDNA from mRNA

(D) electrophoresis --- separation of DNA fragments

(ii) Plants are more readily manipulated by genetic engineering than are animals because

(A) plant genes do not contain interon

(B) plant cells have larger nuclei

(C) a somatic plant cell can often give rise to a complete plant.

(D) genes can be inserted into plant cells by microinjection.

(iii) A paleontologist has recovered a bit of tissue from the 400 year old preserved skin of an extinct bird. the researcher would like to compare DNA from the sample with DNA from living birds. Which of the following would be most useful for increasing the amount of the bird DNA available for testing?

(A) a RFLP analysis

(B) polymerase chain reaction (PCR)

(C) electroporation

(D) Southern hybridization

(iv) Which of the following sequences in double-stranded DNA is mostly likely to be recognized as a cutting site for a restriction enzyme?

(A) AAGG / TTCC (B) AGTC / TCAG (C) GGCC / CCGG (D) ACCA / TGGT

(v) A plasmid

(A) is used as DNA vector (B) is a type of bacteriophage

(C) is a type of cDNA (D) is a retrovirus

(vi) DNA molecules with complementary sticky ends associate by

(A) covalent bond (B) hydrogen bond

(C) ionic bond (D) disulphide bond

(vii) Which technique rapidly replicates specific DNA fragments without cloning?

(A) gel electrophoresis (B) cDNA libraries

(C) genetic probe (D) polymerase chain reaction

(viii) The PCR technique uses
(A) heat resistant DNA polymerase (B) reverse transcriptase
(C) DNA ligase (D) restriction enzymes

(ix) RFLP is a (an):
(A) introns (B) exons
(C) anticodon (D) genetic marker

(x) The dideoxynucleotides (ddATP, ddTTP, ddGTP and ddCTP) are important in DNA sequencing because they:
(A) cause premature termination of growing DNA strand
(B) are used as prime
(C) cause the DNA fragments that contain them to migrate more slowly through the gel
(D) are not affected by high temperatures

(xi) If eukaryotic DNA contains five cleavage sites for a particular restriction enzyme, how many fragments will be produced upon complete digestion of the DNA with that enzyme:
(A) 2 (B) 4 (C) 6 (D) None

(xii) Gel electrophoresis separates nucleic acid on the basis of difference in
(A) length (molecular weight) (B) charge
(C) nucleotide sequence (D) relative proportion of adenine and guanine

(xiii) Which of the following is not the property of probe:
(A) Fluorescently labeled (B) radioactively labeled
(C) Double stranded DNA (D) Complementary to the gene of interest

(xiv) A genomic DNA library
(A) represents all the DNA in a specific chromosome
(B) is made using reverse transcriptase
(C) is stored in a collection of recombinant bacteria
(D) is a DNA copy of mature mRNAs

(xv) Which of the following is genetic marker which is useful in DNA fingerprinting
(A) Probe (B) Primer (C) RFLP (D) Exon

Short Questions

2. What is the role of restriction endonucleases in gene cloning?
3. What are molecular carriers?
4. What is the role of restriction DNA ligases in gene technology?
5. What are the three steps on which mechanism of any DNA analysis sequencing method is based?
6. What are the applications of DNA analysis?
7. What is genomics?
   What are the concerns about genetically modified organisms?

9. Define following terms:
(i) Biotechnology (ii) genetic engineering (iii) gene cloning
(iv) palindromic sequence (v) recombinant DNA technology (vi) DNA ligase,
(vii) polymerase chain reaction (viii) primers (ix) Taq polymerase
(x) genomic library (xi) DNA probe (xii) gel electrophoresis
(xiii) DNA sequencing (xiv) capillary array electrophoresis (xv) DNA profiling
(xvi) genome (xvii) genome maps (xviii) genetic markers
(xix) tissue culture (xx) explants (xxi) cell culture
(xxii) transgenic organisms (xxiii) gene therapy (xiv) biochips.

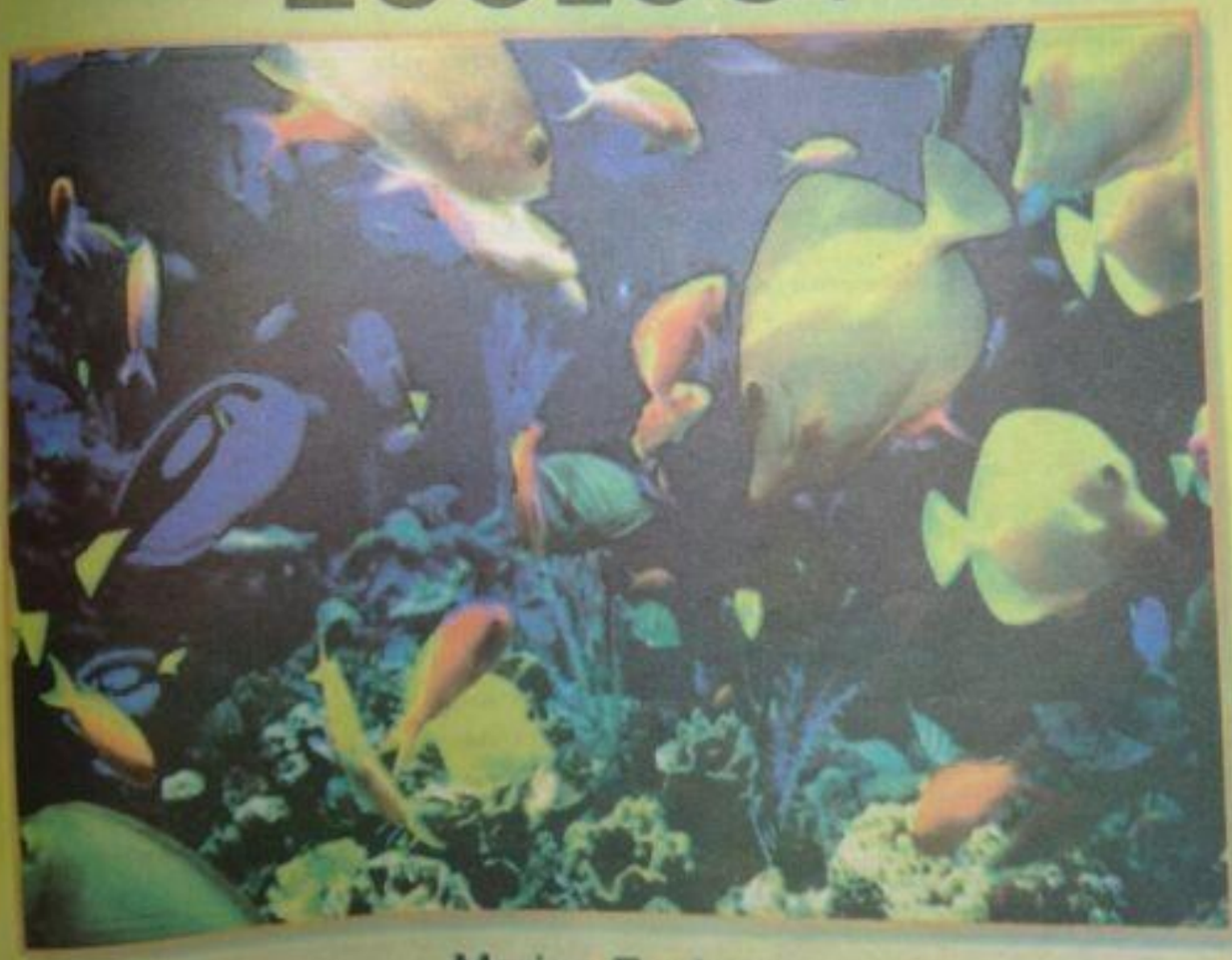
10. Write the difference between:
(a) biotechnology and genetic engineering (b) staggered and blunt cut restriction enzymes
(c) genome and chromosome (d) blotting and clotting
(e) genetic map and physical maps of genome
(f) trangenic organisms and hybrid organisms
(g) biotechnology and nanatechnology

Extensive Questions

11. Describe the components of recombinant DNA technology under the following heads:
(a) Gene of interest (b) Molecular scissors
(c) Molecular carriers (d) Molecular glue
(e) Expression system
12. Describe the mechanism or procedure of recombinant DNA technology.
13. What are polymerase chain reactions discuss its components and mechanism?
14. (a) Describe "Gel electrophoresis" as being used in gene sequencing.
(b) What is the principle of movement of in the gel?
(c) How the fragments are visualized?
15. Explain Sanger-Coulson method of DNA sequencing.
16. Explain automated DNA sequencing as based on the Sanger-Coulson method.
17. (a)Describe mechanism or procedure of DNA analysis.
(b) What are the purpose/applications of DNA analysis?
18. What is gene therapy? Explain the mechanism of gene therapy.
19. What is cystic fibrosis? What is the role of gene therapy for cystic fibrosis?
20. Discuss the scope and importance of biotechnology in promoting human welfare.
21. What are the ethical, legal and social implications of using biotechnology?

SECTION 5

ECOLOGY



Marine Ecology

25

MAN AND HIS ENVIRONMENT

After completing this lesson, you will be able to:

- Define biogeochemical cycles and locate the primary reservoirs of the chemicals in these cycles.
- Describe water cycle in detail.
- Define the terms aquifers and water table.
- Describe nitrogen cycle in detail.
- Define the terms of nitrogen-fixation, nitrification, de-nitrification and ammonification.
- Describe productivity in terms of gross primary productivity and net primary productivity.
- Explain the flow of energy in successive trophic levels.
- Interpret the pyramids of number, biomass and energy.
- Define ecological succession as the process through which ecosystems change from simple to complex.
- Describe primary and secondary succession.
- Differentiate between xerarch and hydrarch succession.
- Explain the xerarch succession on a bare rock starting from the small pockets of lichens to the vegetations of flowering plants.
- Describe characteristics of a population, such as growth, density, distribution, carrying capacity, minimum/viable size.
- Explain, using demographic principles, problems related to the rapid growth of human populations and the effects of that growth on future generations (e.g., relate the carrying capacity of the Earth to the growth of populations and their consumption of resources).
- Analyze the role of the department of population welfare, government of Pakistan in controlling the growing population of Pakistan.
- Relate the need of the nuclear power to the scarcity of fossil fuels.
- State the problems of using nuclear power (surety of safe operation and safe disposal of the wastes).
- Describe the causes of the increasing concentration of carbon dioxide in the world's atmosphere.
- Correlate the increasing CO_2 concentration with the global warming and describe its log term effects.
- Explain the causes and effects of acid rain.
- Describe the composition of the ozone layer and its role in protecting the life on earth.
- State the sources of chlorofluorocarbons and their role in the depletion of ozone.
- Explain the effects of ultraviolet radiation as a serious human health concern.
- Narrate the incidence when one of the four reactors of the Chernobyl nuclear power plant blew up in 1986.

25 Man and His Environment

- Distinguish between renewable and non-renewable environmental resources.
- Describe how man is responsible for the depletion of environmental resources.
- Describe the conventional and non-conventional energy resources.
- Analyze the efforts of various government departments and NGOs to educate people for the protection of environmental resources.

Reading

This chapter aims at enhancing the level of understanding about the basic concepts of ecology. This will enable the students to be well informed of the activities of the very large and growing human population that is threatening the stability of the ecosystem. This chapter introduces how energy and materials flow through natural systems. We begin by discussing how elements essential for life are cycled between organisms and the physical environment. At the end of this chapter, we will discuss human impacts on environment and environmental resources and their depletion.

25.1 BIOGEOCHEMICAL CYCLE

Every organism must require nutrients for its survival. These nutrients are obtained from the environment. The movement of these nutrients in the ecosystem is cyclic one. This flow of nutrients from environment to the organisms and back to the environment is called the **biogeochemical cycle**.

(Nutrient cycle)

25.1.1 Primary Reservoirs of Nutrients

Let us look first at a general model of nutrient cycling that includes the main reservoir of elements and the processes that transfer elements between reservoirs. Each reservoir is defined by two characteristics: whether it contains organic or inorganic materials and whether or not the materials are directly available for use by organisms.

Fig. 25.1 A general model of nutrient cycle. Arrows indicate the process that move nutrients between reservoirs

The nutrients in living organisms themselves and in detritus. (reservoir a) are available to other organisms when consumers feed and when detritivores consume non-living organic matter. Some material move from the living organic reservoir to the fossilized organic reservoir (reservoir b) long ago, when dead organisms were buried by sedimentation over millions of years, becoming coal, oil or peat (fossil fuels). Nutrients in these deposits cannot be assembled directly.

Inorganic materials (elements and compounds) that are dissolved in water present in soil or air (reservoir c) are available for use. Organisms assimilate materials from this reservoir directly and return chemicals to it through the relatively rapid processes of cellular respiration, excretion, and decomposition. Although organisms cannot directly tap into the inorganic elements tied up in the rocks (reservoir d), these elements may slowly become available through weathering and erosion. Similarly, unavailable organic materials move into the available reservoir of inorganic nutrients when fossil fuels are burned, releasing exhaust into the atmosphere.

5.1.2 Water Cycle

The major reservoir of water is the ocean, which contains over 97% of the available water. The water cycle is driven by solar energy which evaporates water and by gravity which draws the water back to Earth in the form of precipitation i.e., rain, snow, dew, sleet (a rain mingled with snow or hail).

Fig. 25.2 Water cycle

Water in environment

Water falling on land takes more varied paths. Some is evaporated from the soil, lakes and streams. A portion runs off the land back to the oceans.

When rain water falls, some of the water sinks or percolates into the ground and saturates the earth to a certain level. The top of the saturation zones is called **water table**. Whenever the Earth contains basin or channels, water will appear to the level of the water table. The water within the **basins** is called **lakes** and **ponds** and water within the **channels** is called **streams** or **rivers.** Sometimes ground water is also located in underground rivers called **aquifers**.

Water in living bodies

Because the bodies of living things are roughly 70% water, some of the water in the water cycle enters the living communities of ecosystems. It is absorbed by the roots of plants and much of this is evaporated back to the atmosphere from their leaves. A small amount is combined with carbon dioxide during photosynthesis to produce high-energy molecules. Eventually these are brokendown during cellular respiration, releasing back to the environment. Heterotrophs get water from their food or by drinking.

25.1.3 Nitrogen Cycle

Nitrogen is required by all living organisms for the synthesis of organic molecules such as amino acids, nucleic acids and proteins. The nitrogen cycle is the movement of nitrogen between the earth and the atmosphere. It consists of a series of processes that convert nitrogen gas to organic substances and these back to nitrogen in nature. It is a continuous cycle maintained by the decomposers and other bacteria. The nitrogen cycle can be brokendown into four types of reactions i.e., decomposition. (ammonification and nitrification), nitrogen fixation, assimilation and de-nitrification.

Decomposition

Decomposition of organic nitrogen compounds is the **first source** of soil nitrates. It occurs in two steps: (a) ammonification (b) nitrification.

Ammonification: The nitrogenous wastes of animals and nitrogenous compounds of dead organisms are decomposed by saprophytic soil bacteria and fungi to form simple substances like water, carbon dioxide, amino acid and energy. The amino acids are converted into ammonia or ammonium ions. Production of ammonia or ammonium compounds in the decomposition of organic matter by microorganisms is called **ammonification.** Ammonification occurs in the soil, in an aerobic environment.

Nitrification: Some ammonia escapes into the soil but much of it and ammonium ions are converted into nitrates by nitrifying bacteria. It is accomplished by two groups of nitrifying bacteria. The first group of bacteria e.g., *Nitrosomonas* converts ammonia to nitrites and the second group of bacteria e.g., *Nitrobacter* converts nitrites to nitrates. This process is called

Teacher's Point

The teacher would ask the students to answer that 'how evaporation and transpiration are related'?

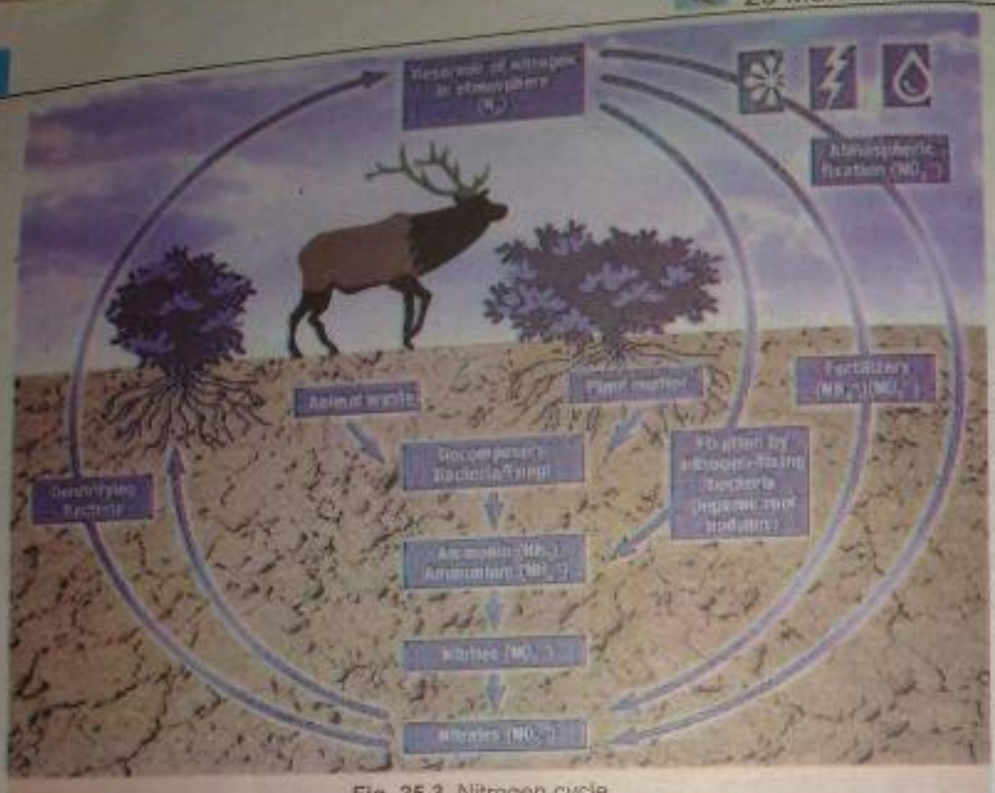


Fig. 25.3 Nitrogen cycle

nitrification. Nitrification takes place only in well aerated soils because the bacteria responsible for it are aerobic.

Nitrogen fixation

Nitrogen gas is composed of two atoms of nitrogen linked by a very strong triple bond. This makes it chemically unreative and large amounts of energy are required to break the bond. Nitrogen gas can be fixed in three ways.

Atmospheric fixation: The nitrogen fixation that occurs spontaneously by lightning is called atmospheric fixation; a small amount (5-8 %) only is fixed in this way. Lightning allows nitrogen and oxygen to combine to produce various oxides of nitrogen. These are carried by the rain into the soil where they can be used by plants.

Industrial fixation: The synthesis of nitrogen containing fertilizers is called industrial fixation.

Biological fixation: Nitrogen-fixing bacteria fix 60 % of nitrogen gas in the atmosphere. The reduction of nitrogen gas to ammonia is energy intensive. It requires 16 molecules of ATP and

Teacher's Point

The teacher would ask the students that using a flowchart; trace the flow of energy in a simple marine food chain. Then, show where nitrogen cycled through the chain when the top level carnivore dies and is decomposed.

a complex set of enzymes to break the bonds so that the nitrogen can combine with hydrogen. Its reduction can be written as:

$$N_2 + 3H_2 \longrightarrow 2NH_3$$

Only a relatively few bacteria (the nitrogen-fixing bacteria) are able to carry out this reaction. Fixed nitrogen is made available to plants by the death and lysis of free-living nitrogen-fixing bacteria e.g., *Azotobacter* (aerobic) and *Clostridium* (anaerobic) or from the symbiotic association of some nitrogen-fixing bacteria with plants e.g. *Rhizobium*.

Assimilation

It is the process of utilization of nitrogenous compounds in living bodies. Many microorganisms are able to utilize free nitrogen directly from atmosphere but plants obtain nitrogen in the form of inorganic nitrogenous compounds like ammonia and nitrates from the soil, whereas animals take their nitrogen from the eating of plants or other animals.

Denitrification

Nitrogen can be lost as a result of the activities of certain soil bacteria; in the absence of oxygen these bacteria breakdown nitrates releasing nitrogen back into the atmosphere and using the oxygen for their own respiration. This process is known as **denitrification** and such bacteria are called denitrifying bacteria e.g., *Pseudomonas* reduce nitrates in the soil to gaseous state.

$$\underset{\text{(Nitrate)}}{NO_3} \longrightarrow \underset{\text{(Nitrite)}}{NO_2} \longrightarrow \underset{\text{(Nitrogen oxide)}}{NO + N_2O} \longrightarrow \underset{\text{(Nitrogen gas)}}{N_2}$$

25.2 THE FLOW OF ENERGY

Ecosystems are transformers of energy. By grouping the species in a community into trophic levels of feeding relationship, we can follow transformation of energy in the ecosystem. Species are classified to trophic levels on the basis of their main source of nutrition and energy. This trophic classification is one of function and not of species.

25.2.1 Concept of Trophic Levels

In an ecosystem the organisms are arranged in different feeding groups, each is known as **trophic level**. At the first trophic level (T_1), primary producers (plants, algae and some bacteria) use solar energy to produce organic plant material through photosynthesis. Herbivores or primary consumers make up the second trophic level (T_2). Predators, the secondary consumers that eat herbivores comprise the third trophic level (T_3); if larger predator i.e., tertiary consumers are present, they represent still higher trophic levels. Organisms that feed at several trophic levels (omnivores) are classified at the highest of the trophic levels (T_4) at which they feed. Decomposers, which include bacteria, fungi, molds, and detritivores such as worms, and insects, breakdown wastes and dead organisms and return nutrients to the soil, occupy the fifth trophic level (T_5).

25.2.2 Concept of Productivity

The ultimate source of energy for our ecosystem is Sun (solar energy). Only 1% of solar energy is incorporated into the ecosystem. The total amount of solar energy which is fixed by the producers during photosynthesis is called **gross primary productivity (GPP)**. On the other hand the amount of energy that remains available for plant growth after subtracting the fraction that plants use for respiration is termed as **Net primary productivity (NPP)** or **biomass**.

Productivity in land ecosystems

Productivity in land ecosystems generally rises with temperature up to about 30°C, after which it declines and is positively correlated with moisture. On land primary productivity thus is highest in warm, wet zones in the tropics where tropical forest biomes are located. In contrast, desert shrub ecosystems have the lowest productivity because their climates are extremely hot and dry.

Productivity in aquatic ecosystems

In the oceans, light and nutrients are important controlling factors for productivity. In oceans, light penetrates only into the uppermost level of the oceans, so photosynthesis occurs in surface and near-surface waters. Marine primary productivity is high near coastlines. Among aquatic ecosystems, algal beds and coral reefs have the highest net primary production while the lowest rates occur in the open due to a lack of nutrients in the illuminated surface layers.

25.2.3 Energy Flow between the Trophic Level

Ecosystems are transformers of energy. On average about 10 % of net energy production at one trophic level is passed on to the next level.

Factors affecting energy flow

Processes that reduce the energy transferred between trophic levels include respiration, growth, reproduction, defaecation and non-predatory death (organisms that die but are not eaten by consumers). The nutritional quality of

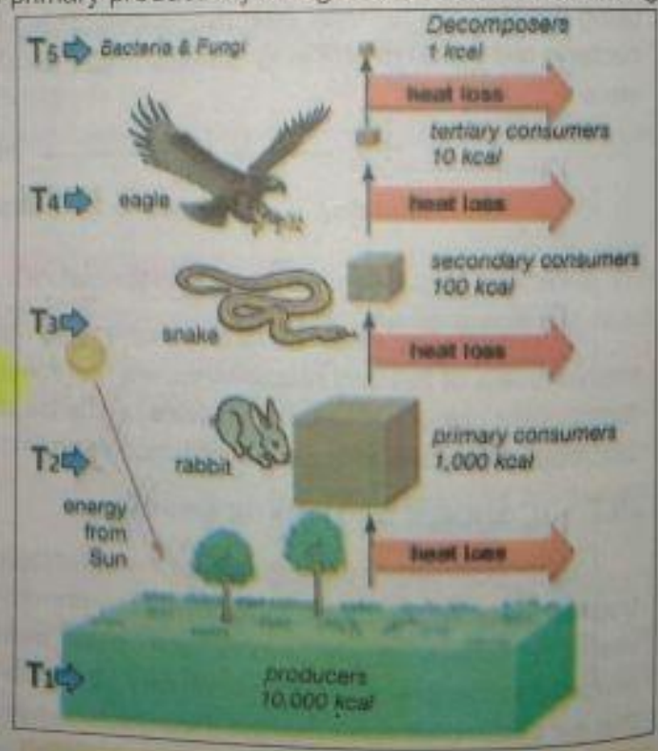


Fig. 25.4 Energy flow in ecosystem

Teacher's Point

The teacher would ask the students that what proportion of energy is transferred from one trophic level to the next in an ecosystem.

material that is consumed also influences how efficiently energy is transferred because consumers can convert high-quality food sources into new living tissue more efficiently than low-quality food sources.

Significance of decomposers in energy flow

The low rate of energy transfer between trophic levels makes decomposers generally more important than producers in terms of energy flow. Decomposers process large amounts of organic material and return nutrients to the ecosystem in inorganic form, which is then taken up again by primary producers. Energy is not recycled during decomposition, but rather is released, mostly as heat.

Ecological pyramids

Ecologists compare trophic levels by determining the number of organisms, the biomass or the relative energy found at each level. If these data are graphed, the graph has a pyramid shape, which is called **ecological pyramids**. Ecological pyramids are therefore, graphical representation of ecological data.

Pyramid of energy: A pyramid of energy indicates the energy contents in the biomass of each trophic level. These pyramids show that most energy dissipates into the environment when going from one trophic level to another. Less energy reaches each successive trophic level from the level beneath it because some of the energy at the lower level is used by those organisms to perform work while some of it is lost. Energy pyramids explain why there are few trophic levels. Food webs are short because of the dramatic reduction in energy contents that occurs at each successive trophic level.

Fig. 25.5 Ecological pyramids

Teacher's Point

The teacher would ask the students to 'Draw an energy pyramid for a five-step food chain. If 100 percent of the energy is available at the first trophic level, what percentage of the total energy is available at the highest trophic level?'

Pyramids of biomass: A pyramid of biomass illustrates the total biomass at each successive trophic level. Biomass is a quantitative estimate of the total mass, or amount of living material; it indicates the amount of fixed energy at a particular time. Units of measure vary; biomass may be represented as total volume, dry weight or live weight. Typically, the pyramids illustrate a progressive reduction of biomass in succeeding trophic levels.

Pyramids of numbers: A pyramid of numbers shows the number of organisms at each trophic level in a given ecosystem with greater numbers illustrated by a wider pyramid. In most pyramids of numbers, each successive trophic level is occupied by fewer organisms. Thus, the number of herbivores is greater than the number of carnivores.

25.3 ECOLOGICAL SUCCESSION

A community may begin when new habitat is created, as when a volcanic island rises out of the area or when rock and soil is deposited by a retreating glacier. New communities also may form in regions that have been disturbed, as by fire or hurricane. Species that arrive early in new or disturbed habitat tend to be replaced later by other species which in turn may be replaced by other species. Earlier species are replaced by later species because the replaced species are better able to grow and reproduce under the environmental conditions of the area. The process by which species are replaced over time is called **ecological succession**. Individual successions are known as **seres** and the development phases are called **seral stages**.

25.3.1 Kinds of Succession

There may be two kinds of ecological successions: primary succession and secondary succession.

Primary succession

Primary succession is the change in species composition over time in a habitat that was not previously inhabited by organisms. Bare rock surfaces, such as recently formed volcanic lava and rock scraped clean by glaciers, are examples of sites where primary succession might occur. (Pioneers)

Secondary succession

Secondary succession is the change in species composition over time in a habitat already substantially modified by a pre-existing community. Soil is already present at the sites. The common examples of sites where secondary succession occurs are: (a) abandoned farm fields undergo secondary succession as they revert to forest (b) Succession in forest area where vegetation has been devastated by fire, flood, cyclone etc.

Ecological succession which begins in pond, lakes, and marshes or elsewhere in water are termed **hydrarch** and different stages are called **hydroseres**. Succession initiated on bare rocks, sand dunes, rocky slopes etc. where there is deficiency of water, are termed **xerarch** and different stages of development are collectively called **xerosere**.

Fig.25.6 Secondary succession

Stages of succession

The first requirement in the process of succession in any bare area is the migration of plants and animals from surrounding areas and their aggregation. These migrants are called **pioneers**. The pioneers become successful in taking hold of the soil. They increase in number. By their death and decay, the pioneers increase organic matter, moisture and nitrogen content of the soil. The enriched soil now becomes suitable for the growth of next group of invaders. These are called **seral communities** and constitute one **seral stage**. By their activities, these serals modify the environment and changed environment becomes unsuitable for their growth and a new group of plants and animals invade the environment. The end product of succession after several seral communities is the **climax community** or relatively stable community.

25.3.2 Xerarch Succession

Various stages and process of **xerarch succession** which results in the development of climax community may be described briefly as follows:

Crustose-lichen stage

It is the pioneer stage. On bare rocks only crustose-lichen (crust-like lichens) can grow. These are slow growers and can withstand extreme desiccation. When there is rain they absorb water like sponges and decompose rock by secreting acids. Important members of this stage are *Licanora* and *Rhinodina*.

Foliose-lichen stage

On the little soil, which is accumulated on the rock there appear species of foliose-lichens (leaf-like lichens) and by their activities there collects a thin layer of soil. Important members of this stage are *Permelia* and *Dermatocarpom*.

Moss stage

When sufficient amounts of soil have been accumulated in the minute crevices and depressions in the rock, xerophytic mosses begin to appear. The mosses increase the amount of soil. By their death and decay, a mat may be formed on the rock surface. This can hold greater amount of water and along with the soil makes habitat suitable for herbs.

Herb stage

Herbaceous weeds, mostly annuals invade the rock. Their roots penetrate deep down, secrete acids and enhance the process of weathering. Leaf litter and death of herbs add humus to the soil. Shading of soil results in decrease in evaporation and there is a slight increase in temperature. As a result the xeric conditions begin to change and biennial and perennial herbs and xeric grasses begin to inhabit.

Fig.25.7 Xerarch succession on bare rock (primary succession)

Shrub stage

On the soil appear xeric shrubs. These shrubs (low woody plants smaller than a tree with little or no trunk, bush) may start from seeds or invade from adjacent areas by rhizomes. These make the condition unsuitable for herbs and overshadow them. The herbs are unable to compete and hence are replaced by shrubs. Early invasion of shrub is slow but once a few bushes have become established, birds invade the area and help disperse the seeds. This results in dense growth, shading the soil and making conditions unfavourable for the growth of herbs which then begin to migrate.

Tree stage

Change in environment favours colonization of tree species. The tree saplings begin to grow among the **shrub** and establish themselves. The trees form canopy and shade the area. Shade-loving plants continue to grow as secondary vegetation. Leaf litter and decaying roots weather the soil further and add humus to it making the habitat more favourable for growth of trees.

Climax stage

The first species of trees are relatively xeric. As the weathering process continue and the soil deepens, the xeric trees in turn give place to mesophytic species of trees. Ultimately, a forest may develop.

Skills: Analysing and interpreting

- **Justify the fact that humans are often responsible for secondary succession**

Humans often interfere with succession. They replace complex ecosystems with simple ecosystems. These ecosystems are often designed to meet human needs. Simple ecosystems have less biodiversity than complex ecosystems. They are often not sustainable. Human disturbance i.e., agriculture, forestry, development, can

25.4 POPOULATION DYNAMICS

Population dynamics is concerned with the studies of long-term and short-term changes in population size and the factors that regulate population size such as:

Inflow: births, immigration.

Overflow: lower recruitment, higher mortality, poor condition, increased emigration, habitat degradation.

Outflow: Culling(to pick and destr oy individuals e.g., seal, deer), predation, natural deaths, accidents, emigration. In other words, the way that the numbers and structure of an animal population vary over time and the factors which cause variations, are described by population dynamics.

25.4.1 Characteristics of a Population

A **population** is a group of individuals of a single species living in the same general area. Members of a population rely on the same resources, are influenced by similar environmental factors and have a high likelihood of interacting with and breeding with one another. The characteristics of a population are: growth, density, distribution, carrying capacity and viable size.

Growth

Increase in the number of individual of a population is called **population growth**. Consider what happens if a few individuals enter an unoccupied area. Assuming there is enough food and that predation and disease are not too severe, reproduction will occur and the number of individuals will increase. At first, there may be a **lag phase** as the individuals settle into their new environment. As reproduction gets underway, the population shows **exponential growth**. The population **size doubles** at regular intervals. During exponential phase the population is said to grow **geometrically**.

Density

Density is the number of individuals per unit area or volume e.g., the number of mulberry trees per square kilometre in Islamabad or the number of *E.coli* bacteria per millilitre in a test tube.

Teacher's Point

The teacher would ask the students to write (creative writing) using the information from the section 25.5 to write a story about an ecosystem that is disturbed ad undergoes succession. Hint: Include a flow chart with your story to show the main stages of change.

The dispersion of individuals in a population is called **distribution**. There are three distribution patterns found in different populations like **clumped** (individuals aggregate in patches), **uniform** (individuals are evenly spaced) or **random distribution** (unpredictable spacing among individuals)

There is a limit to the number of individuals that can occupy a habitat. The **carrying capacity** is defined as the maximum population size that a particular environment can support. Carrying capacity is not fixed but varies over space and time with the abundance or limiting resources.

The Department of Population Welfare is providing Family Planning and Reproductive Health Services through various programmes. (1) Family Welfare Centres, (2) Mobile Service Units, (3) Reproductive Health Services (RHS) Centres are being run by various NGOs with assistance of the Population Welfare Department.

The Ministry of population welfare, Government of Pakistan is responsible for the control of population growth in Pakistan. Population Welfare Program is an on-going program launched by the ministry of population, Pakistan since 1960. The first ever Population Policy in Pakistan was announced on 11th July, 2002.The Population Policy is wide in scope. It states a commitment to reduce the incidence of unwanted fertility, promote small family norm, make an investment in youthful population and focus on male involvement. The overall vision of Population Policy is to achieve population stabilization replacement level i.e., a total fertility rate of 2:1 children per woman by 2020, increase awareness of adverse consequences of rapid population growth at the national, provincial, district and community level and promote family planning as an entitlement based on informed and voluntary choice and attain reduction in infertility.

- **Investigate the effects of human population growth on the environment and the quality of life**

Population growth effect environment and quality of life in following ways: (a) Inadequate fresh water. (b) Depletion of natural resources. (c) Increased levels of air pollution, water pollution, and soil contamination. (d) Deforestation and loss of ecosystems. (e) Changes in atmospheric composition and consequent global warming. (f) Irreversible loss of arable land and increase in deforestation, mass species extinction. (g) High infant and child mortality. (h) Increased chance of the emergence of new epidemics and pandemics, starvation and malnutrition (i) Poverty coupled with inflation in some regions and a resulting low level of capital formation. (j) Low life expectancy occurs in countries with fastest growing populations.

Teacher's Point

The teacher would ask the students to:

- Make a hypothetical j-shape exponential population growth to show that under ideal conditions with unlimited resources, a population will grow exponentially.
- Write the factors that might cause the carrying capacity of a population change.

Science, Technology and Society Connections

Outline the advances in medical care and technology that have contributed to an increase in life expectancy, and relate these developments to demographic issues.

Advances in medical care and technology have contributed to an increase in life expectancy during maternity cases. A good example of how advances in technology have changed health outcomes over time is in the treatment of pre-term babies. Changes in technology, including special ventilators, artificial pulmonary surfactant to help infant lungs develop, neonatal intensive care and steroids for mother and/or baby, helped decrease mortality, with an overall increase in life expectancy of low-birth weight baby. On one hand, it is beneficial aspect of advances in medical care and technology but on the other hand, it is supposed to be one of cause of rapid growth of population.

25.5 HUMAN IMPACTS ON ENVIRONMENT

Humans are a part of the natural environment. Population growth leads to the loss of natural habitat. Deforestation causes loss of species of fauna and flora, oxygen production and carbon dioxide elimination. Ozone layer depletion, water pollution, global warming, desertification, increased erosion of land, is directly caused by human activities such as use of nuclear fuel, industrialization, urbanization, transportation etc.

25.5.1 Nuclear Power

Nuclear power is the use of sustained nuclear fission to generate heat and electricity. As reported in 2005, nuclear power provided 6.3% of the world's energy and 15% of the world's electricity. The scarcity of fossil fuels which is not available in all the countries, is the reason for the development nuclear power stations.

Fig. 25.8 A boiling water reactor

Advantages of nuclear power

(a) Nuclear power costs about the same as coal, so it's not expensive to make. (b) Does not produce smoke or carbon dioxide, so it does not contribute to the greenhouse effect. (c) Produces huge amounts of energy from small amounts of fuel. (d) Produces small amounts of waste. (e) Nuclear power is reliable.

Disadvantages of using nuclear power

The two main problems using nuclear powers are surety of safe operation and safe disposal of the wastes.

Surety of safe operation: To achieve optimum safety, in nuclear plants prevention, monitoring and action i.e., to mitigate consequences of failures are followed. These are: (a) High-quality

Teacher's Point

The teacher would ask the students

- Why do you think age-structure diagrams can help predict future population trends?
- To write a paragraph on the trends of human population in Pakistan from 1950 to 2018

design and construction. (b) Comprehensive monitoring and regular testing to detect equipment or operator failures. (c) Prevention of significant radioactive releases.

Safe disposal of the wastes: Radioactive wastes are wastes that contain radioactive material. Nuclear waste is a cause for concern because it is not bio-degradable, meaning it does not decompose naturally under the effect of the atmosphere. Secondly, it causes a number of health hazards for anyone who comes into contact with the radiation from this waste. Therefore, some safe measure should be used for disposal of nuclear waste which may include **deep ocean disposal, deep geological burial, nuclear waste recycling,** reprocessing and solidification processes.

Chernobyl nuclear power incidence

The Chernobyl disaster was a nuclear accident. It occurred on 26 April 1986 at the Chernobyl Nuclear Power Plant in Ukraine. An explosion and fire released large quantities of radioactive contamination into the atmosphere which spread over much of Western USSR and Europe. Approximately over 500,000 workers were affected apart from the 57 direct deaths in the accident itself. United Nations Scientific Committee on the Effects of Atomic Radiation predicted in 2005 that up to 4,000 additional cancer deaths related to the accident would appear "among the 600 000 persons receiving more significant exposures. Do you know what happened in Japan in 2011?

25.5.2 Carbon Dioxide and Global Warming

Due to various human activities such as burning of fossil fuel in motor vehicles and industrial process, amount of CO_2 is increase in atmosphere. CO_2 absorb high energy radiations and thus result in increase in atmospheric temperature. This rise in temperature is known as **global warming.**

Causes of increasing concentration of CO_2 in atmosphere

Human activities are mainly responsible for increasing the amount of CO_2 in the atmosphere. Burning of fossil fuels, such as coal, oil and natural gas for industry, driving our transport, heating our homes and generating electricity are the major sources of human emission. The burning of wild lands, forests are increasing CO_2 in the atmosphere. Deforestation does increase the amount of CO_2. There has been a marked increase in the CO_2 percentage in the atmosphere since industrial

Fig.25.9 Greenhouse effect (global warming)

Teacher's Point

The teacher would ask the students to name the greenhouse gases that cause greenhouse effect, which helps maintain Earth's temperature range.

revolution. Though it is not the main cause, humans and other animals also increase CO_2 in the atmosphere as they breathe out CO_2. In addition, eruption of volcanoes off and on, is another cause to increase of CO_2 in the world atmosphere.

CO_2 concentration and the global warming

At the moment, the amount of CO_2 in the air is increasing. It makes up about 0.04% of the air now, compared with 0.03% in the mid-twentieth century. What does this matter? It is very likely that the raised atmospheric CO_2 levels are causing the average temperatures on Earth to increase, a process called global warming or greenhouse effect. CO_2 is one of a number of greenhouse gases.

Long term effects of global warming

The more carbon dioxide there is in the atmosphere, the more reflected infra-red radiation is trapped and the warmer the Earth becomes. An increase of only 1.3°C would make the world warmer than at any time in the past 100,000 years. A worst-case scenario suggests that the warming would be great near the poles. The resultant melting polar ice might raise sea level by an estimated 100 m, gradually flooding areas 150 km (or more) inland from the current coastline. A warming trend would also alter the geographical distribution of precipitation, making major agricultural areas drier.

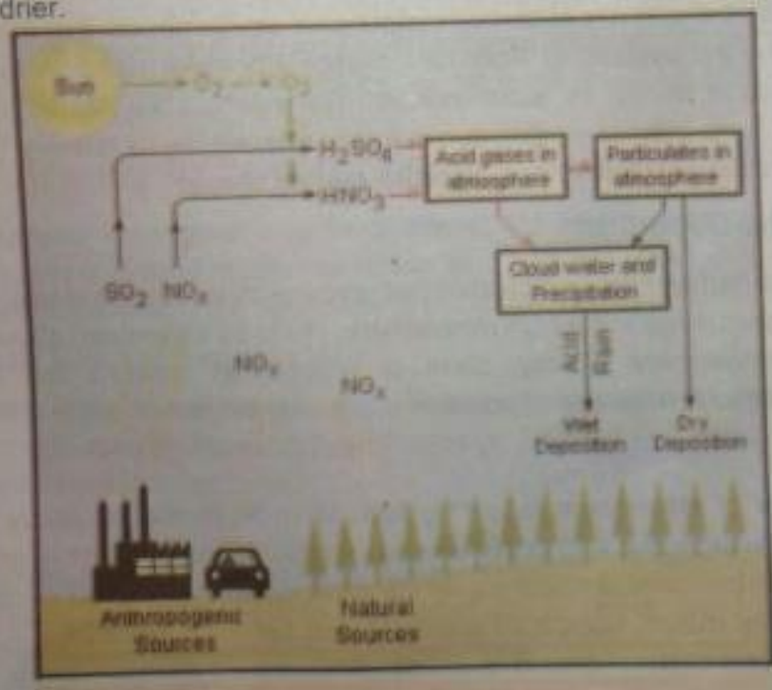


Fig. 25.10 Acid rain

Teacher's Point

The teacher would ask the students to design an experiment to show that how does acid rain affect the germination of seeds?

25.5.3 Acid Rain

The water in the atmosphere has become polluted with sulphur dioxide and oxides of nitrogen. The gases react with water vapour and oxygen in the air. Sulphuric acid (H_2SO_4) and nitric acid (HNO_3) are formed. The water vapour with its contents becomes part of a cloud. The water vapour condenses and falls to Earth as **acid rain** which may be hundreds of miles away from the source of pollution. The acid precipitation also falls as snow or as dry 'micro' particles, mixing with water when it reaches surface on the ground.

Causes of acid rain

Although sulphur and nitrous oxides are produced naturally during volcanic eruption and forest fires, humans produce more than half of these chemicals from burning of coal by electricity generating plants, industrial boilers and large smelters that obtain metals from ores. In addition, nitrogen oxides are emitted by automobiles. The causes of acid rain to human activities are called anthropogenic sources.

Effects of acid rain

Some of the effects of acid rain are: (a) It increases soil acidity, (b) Acid rain damages life in farms and forest. (c) It kills aquatic organisms and prevents their successful reproduction. (d) It also increases the quantity of certain metals such as Aluminium that may prove toxic to many organism in the ecosystem.(e) Acid rain causes extensive damage to buildings and stone structures which is known as 'stone cancer'. For example Taj Mahal (build by Mughal King Shahjahan) at Agra, India is erodjng due to fumes released by oil refinery. (f) Acid rain adversely affects the nervous, respiratory and digestive system of man.

25.5.4 Ozone Depletion

Ozone is a highly reactive molecule that contains three oxygen atoms. It is constantly being formed and broken down in the high atmosphere, (10 to 50 kilometres) above Earth, in the region called the **stratosphere**. Today, there is widespread concern that the ozone layer is deteriorating due to the release of pollution containing the chemicals chlorine and bromine.

Composition of ozone layer

It is a layer of atmosphere extending from 10 to 50 kilometres above Earth. In pure form, ozone is a bluish, explosive and highly poisonous gas. Ozone is a form of oxygen that is human-made pollutant in the lower atmosphere. In the stratosphere, the normal concentration of ozone is about 0.1 part per million, compared with 0.02 part per million in the lower atmosphere. This ozone –enriched layer is called the **ozone layer**.

Formation of ozone layer

A small fraction of the radiant energy produced by the Sun is called ultraviolet or UV radiation. Ozone is formed in the atmosphere when ultraviolet radiation from

Fig. 25.11 Ozone layer depletion

the Sun splits one oxygen molecule into two oxygen atoms. The atomic oxygen then combines with another oxygen molecule to form ozone (O_3).

Causes of ozone layer depletion

Most of the UV radiations are filtered out by ozone in the stratosphere. In the stratosphere, there is a group of commercially important compounds called **chlorofluorocarbons** (CFCs). These have been used as propellants in aerosol cans, coolants (e.g., Freon) in air conditioners and refrigerators, foam (e.g., Styrofoam) for insulation and packing and cleaners in the electronic industry. Ultraviolet radiation breaks CFCs and similar compounds into chlorine, fluorine and carbon. Under certain stratospheric conditions, chlorine and fluorine are capable of reacting with ozone, converting it into molecular oxygen.

Role of Ozone layer in protecting the life on earth

In 1985, it was discovered that the springtime levels of stratospheric ozone over Antarctica had declined by over 40% since 1977. A hole had been punctured in Earth's protective shield. Ozone molecules in the stratosphere absorb incoming solar ultraviolet radiation and this protects life on Earth.

Effects of ultra violet radiation on human health

With depletion of the ozone layer, UV radiations reach Earth's surface. Excessive exposure to UV radiation is linked to a number of human health problems, including sunburn, premature aging of skin, skin cancer and cataracts. Photosynthesis by phytoplankton is also affected and reduced by ultraviolet radiation.

25.6 ENVIRONMENTAL RESOURCES AND THEIR DEPLETION

Wherever humans have gone they have altered the environment to their need. Living organisms are important natural resources. Earth is a self-contained unit. The future of the mankind depends on how wisely the resources of the Earth are used.

25.6.1 Kinds of Natural Resources

Environment is complex body of resources. These resources are the element of nature that serves as primary resources for the existence of humans. These may be described as renewable resources and non-renewable resource.

Renewable resources

Renewable resources are produced by natural systems that replace themselves quickly enough to keep pace with consumptions. Examples are air, water, land and wild life. Living organisms use them. They are constantly replaced by natural cycles e.g., water cycle, carbon cycle, oxygen cycle, nitrogen cycle etc,.

Teacher's Point

The teacher would ask the students to evaluate the impact of environmental research on the problem of ozone depletion. How did research identify the cause of the problem? To what action did this research lead?

Non-renewable resources

Non-renewable resources formed at a rate much slower than their environment consumption. These are exhaustible and cannot be replaced if destroyed. Examples are various metals, non-metallic minerals, coal, oil and natural gas etc.

25.8.2 Conventional and Non-conventional Energy Sources

The energy sources are categorized into two types i.e. conventional and non-conventional.

Conventional energy sources

The energy source that has been used from ancient times is called **conventional sources of energy**. The examples of conventional energy sources are fossil fuels (coal, natural gas, oil), firewood and sources of energy i.e., electricity, are coal, oil, wood, peat and uranium.

The advantages of conventional sources of energy e.g., fossil fuels are that these are inexpensive and require established technologies that can produce energy around the clock. The disadvantages of conventional sources of energy are that they have a limited supply because eventually the nuclear elements and fossil fuels will be used up. In addition, burning fossil fuels release significant amounts of greenhouse gases and contribute to acid rain.

Fossil fuels include coal, oil and gas. They provide 95% of the energy requirement. They are nonrenewable. They are called fossil fuels because they are remains of plants and animal that lived millions of years.

Nuclear energy is the energy obtained by fission of radioactive atom. This energy is used to produce electricity in nuclear reactors. The primary nuclear fuel is U^{235}. The advantage of nuclear energy is that it emits large amount of energy. The disadvantages are that it generates radioactive waste and expensive.

Non-conventional energy sources

Non-conventional energy sources or unusual sources of energy are the new sources of energy which are still not in common use. Their contribution to the national power is nominal. These are: Solar power, Hydro-electric power (dams in rivers), Wind power, Tidal power, Ocean wave power, Geothermal power (heat from deep under the ground), Ocean thermal power (the difference in heat between shallow and deep water), Biomass (burning of vegetation to stop it producing methane), Bio-fuel (producing ethanol (petroleum) from plants, Bio-gas. It is also known as **renewable energy sources**.

The advantages of non-conventional source of energy are that these are abundant in nature, pollution free and eco-friendly. These sources can be renewed with minimum effort and energy. The disadvantages of nonconventional energy sources are that these are often limited to producing energy only under certain circumstances such as sunny days for solar panels and windy days for windmills.

Depletion of resources

Resource depletion is a term referring to the exhaustion of raw materials within a region. Resources are commonly divided between renewable resources and non-renewable resources.

Use of either of these forms of resources beyond their rate of replacement is considered to be resource depletion.

Causes of resource depletion: Man is the main cause of resource depletion. Its activities are continuously consuming natural environmental resources with the pace beyond the pace of their renewal. The factors through which man is depleting natural resources include over-consumption/excessive or unnecessary use of resources, non-equitable distribution of resources, overpopulation, slash and burn agricultural practices, technological and industrial development, erosion, irrigation, mining for oil and minerals and pollution or contamination of resources.

Science Tidbits

The conventional energy sources are the non-renewable resources in Pakistan that includes: thermal energy and nuclear energy. Thermal sources in Pakistan are coal, oil, natural gas. The non-conventional sources of energy in Pakistan are wind energy, hydroelectric power solar energy and biogas.

2.6.3 Protection of Environmental Resources

Natural resources are vital to our existence. Our health and well-being are closely linked to the quality of our air, water, soils and biological resources. Our landscapes, seascapes and wildlife are inseparable from our culture and inspire art and literature. Our economy and key industrial sectors are directly and indirectly reliant on functioning ecosystems. Many people believe that natural resources have their own intrinsic value, that is, they are important for their own sake regardless of their functional value.

Role of government

The governments control and develop forests, dams, major irrigation system, power stations, railways, ports, roads, mines and industries of the country. The ministry of environment is entrusted with planning, protection and coordination of environment and forestry programmes. The ministry is involved in conservation and survey of flora, fauna, forests and wildlife, prevention and control of pollution, aforestation, regeneration of degraded areas and protection of overall environments. The assessment of environmental impact prior to implementing any project which can damage environment is taken up by ministry of environment. To educate the people every year, Earth day on 22nd of April and tree plantation week are observed.

Role of NGOs

NGOs can play a very important role in environmental protection and management and creating mass awareness toward environment. They have made people aware of the environmental problems, which are caused due to neglect and uncontrolled exploitation of natural resources. Some of the NGO's are working for environmental awareness while some are working in research field.

Teacher's Point

The teacher would ask the students to write the sources of energy in Pakistan.

Science, Technology and Society Connections

Justify why science education has become necessary for everyone to understand the basis of man's continued existence and the steps man has to take to save and improve life.

People must be educated to: Pay attention to how you use water. Leave your car at home. Walk or ride your bike to work, school and anywhere you can. Turn off lights when you're not in the room and unplug appliances when you're not using them. Use CFC free products. Use renewable energy sources. Switch to compact florescent or LED light bulbs. **Reuse** means instead of throwing things away, try to find ways to use them again. For example we can use glass bottles after sterilization. **Recycle** means regeneration of waste materials into other useful items. For example any of the things we use every day, like paper bags, soda cans, and milk cartons, are made out of materials that can be recycled.

Science, Technology and Society Connections

Investigate the carriers related to the study of environmental resources.

Environmental attorney, Environmental engineer, Environmental health officer, Environmental impact analyst, Environmental toxicologist, Environmentalist, Natural resource manager, Population biologist, Wildlife conservation officer, Watershed project manager, Natural resource manager etc.

Activity

Draw a concept map that shows how population grows. Include the following terms: exponential growth, logistic growth, birth-rate, death rate, immigration, emigration. Add any other terms that you think are useful to complete the map.

Exercise

M.C.Qs

lect the correct answer

Which is one group of organisms that is able to fix atmospheric nitrogen into forms usable by living organisms?

(A) plants (B) fungi
(C) insects (D) bacteria

) Producers of an ecosystem are:

(A) autotrophs (B) absorptive heterotrophs
(C) ingestive heterotrophs (D) none of them

ii) A population's carrying capacity

(A) can be accurately calculated.
(B) generally remains constant over time.
(C) may change as environmental conditions change.
(D) can never be exceeded.

(iv) Which of the following is also called greenhouse effect?

(A) ozone layer depletion (B) global warming
(C) acid rain (D) all of them

(v) Ozone layer is found in:

(A) troposphere (B) stratosphere
(C) both a and b (D) none of them

(vi) The main cause of the recent increase in the amount of CO_2 in the Earth's atmosphere

(A) increase worldwide primary production
(B) increased worldwide standing crop biomass
(C) the rapidly growing human population
(D) the burning of larger amounts of fossil fuels

(vii) Which of these ecosystems has the lowest net primary production per square metre?

(A) a salt marsh (B) an open ocean
(C) a costal reef (D) a grassland.

(viii) Which of the following is the graphical representation of ecological data of an ecosystem?

(A) pyramids (B) succession
(C) niche (D) habitat

(ix) Which of these levels of ecological study involves both abiotic and biotic components?
(A) organisms (B) populations
(C) ecosystem (D) conservation ecology

(x) Why are ecosystems dependent on a continual supply of solar energy?
(A) carnivores have a greater biomass than producers.
(B) decomposers process the greatest amount of energy in an ecosystem.
(C) energy transformation results in a loss of usable energy to the environment.
(D) energy cycles within and between ecosystems.

(xi) Nutrient cycle always involve
(A) rocks as reservoir
(B) movement of nutrients through the biotic community
(C) the atmosphere as an exchange pool
(D) loss of the nutrients from the biosphere.

(xii) How do nitrogen-fixing bacteria contribute to the nitrogen cycle?
(A) return nitrogen(N_2) to the atmosphere
(B) change ammonium to nitrate
(C) change N_2 to ammonium
(D) absorb nitrate from the soil

Short Questions

2. Describe water in environment.
3. Describe water in living bodies.
4. Describe the role of microorganisms in nitrogen cycle
5. How nitrogen gets from the air to a plant?
6. What natural areas or situations might favour denitrification?
7. What is the concept of trophic levels?
8. What are factors affecting energy flow?
9. What is the significance of decomposers in energy flow?
10. Describe productivity in land ecosystem.
11. Describe productivity in aquatic ecosystem.
12. Why is an ecosystem's primary production lower than gross primary production?
13. What are ecological pyramids?
14. Why is only a small portion of the solar energy that strikes Earth's atmosphere stored by primary producers?

15. Why does the production pyramid have the same general shape as the biomass pyramid in most ecosystems?
16. What happens to the sun's energy as it travels through the food chain.
17. During succession, how might the early species facilitate the arrival of other species?
18. Why food chains are usually short?
19. How can clear cutting a forest damage the water quality of nearby lakes?
20. Enlist the characteristics of a population?
21. Investigate the effects of human population growth on the environment.
22. What is the need of nuclear power and what are the problems of using nuclear power?
23. Narrate the incidence when one of the four reactors of the Chernobyl nuclear power plant blew up in 1986. Can you name any such happening in Japan in 2011?
24. What is the relationship between carbon dioxide concentration and global warming?
25. What are the causes of acid rain?
26. What are the long term effects of global warming?
27. Why should people be concerned that ozone layer in the stratosphere is being depleted?
28. What are the effects of ultra violet radiation on human health?
29. Describe how man is responsible for the depletion of environmental resources?
30. Define/Describe/Explain briefly:
ecology, biogeochemical cycle, water table, aquifers nitrogen cycle, ammonification, nitrogen fixation, nitrification, de-nitrification, atmospheric fixation, industrial fixation, biological fixation, assimilation, trophic level, gross primary production, net primary production, biomass, ecological pyramids, pyramid of energy, pyramid of number, ecological succession, seres, seral stage, hydrach, hydrosere, xerach, xerosere, pioneers, seral communities, seral stage, climax community, population, carrying capacity, demography, acid rain, ozone layer, stratosphere, fossil fuels, nuclear energy.
31. Write the difference between:
(a) basins and channels
(b) ammonification and denitrification
(c) industrial fixation and biological fixation
(d) nitrification and denitrification
(e) primary and secondary succession
(f) hydrosre and xerosere
(g) xerarch and hydrarch succession
(h) non-renewable and renewable resources
(i) conventional and non-conventional energy sources
(j) fossil fuels and nuclear fuel

Extensive Questions

32. Describe and locate the primary reservoirs of the biogeochemical cycle.
33. Describe in detail how water is cycled within ecosystems.
34. Give a detail account of nitrogen cycle
35. Give an account of the roles of bacteria in the nitrogen cycle.
36. Describe productivity in terms of gross primary productivity and net primary productivity.
37. What are trophic levels? Explain the flow of energy in successive trophic level.
38. Describe and interpret pyramids of number, biomass and energy.
39. Differentiate between xerarch and hydrarch succession and explain the xerarch succession.
40. Describe growth, density, distribution, carrying capacity, minimum/viable size as the characteristics of a population.
41. Analyze the role of the department of population welfare, government of Pakistan in controlling the growing population of Pakistan.
42. What is nuclear power? What are the advantages and disadvantages of nuclear power?
43. What are the causes of increase of concentration of carbon dioxide in atmosphere?
44. What is global warming? Describe the causes and effects of global warming.
45. How acid rain is produced? What are the causes and effects of acid rain?
46. (a)What is the composition of ozone layer? What is its role in protecting the life on Earth? (b) What are the sources of chlorofluorocarbons and their role in the depletion of ozone?
47. Describe conventional and non-conventional energy resources.
48. Discuss the efforts of various government departments and NGOs to educate people for the protection of environmental resources.

24

EVOLUTION

After completing this lesson, you will be able to

- Describe creationism and the theory of evolution as two contradictory ideas.
- Explain how biogeography provides an evidence for evolution.
- Describe the evidences of evolution that come from paleontology, comparative anatomy and molecular biology.
- Differentiate between convergent and divergent evolution on the basis of inheritance of the homologous and analogous structures.
- Describe the theories that have been put forwarded about the mechanism of evolution of eukaryotes from prokaryotes.
- Justify Lamarck as an early proponent of evolution.
- Describe the theory of inheritance of acquired characters, as proposed by Lamarck.
- Outline the steps of the evolution of the giraffe, as illustrated in Lamarckism.
- State the drawbacks in Lamarckism.
- Briefly describe the observations Darwin made during his voyage on HMS Beagle.
- Explain the theory of natural selection as proposed by Darwin.
- Describe the ideas of Charles Lyell, James Hutton and Thomas Malthus that contributed in the early development of Darwinism.
- Describe the role of Alfred Wallace in motivating Darwin to publish the theory of natural selection.
- Justify, on the grounds that both Wallace's and Darwin's papers were published in the *Journal of the proceedings of the Linnaean Society*, why the theory was attributed to Darwin.
- Describe the assumptions of the Hardy-Weinberg theorem and relate these to the factors that change the allelic frequencies of the population.
- Explain the concept of genetic drift (neutral selection).
- Define the concept of speciation and explain the mechanisms of speciation (allopatric, parapatric and sympatric speciation).

Reading

Man has always been curious to know how, when and where life originated; and how the diverse forms of animals and plants came into existence. Many scientific and non-scientific theories have been proposed so far regarding the origin of life on earth in which two schools of thoughts have gained more recognition like special creation and biochemical evolution, which one is true, is not the scope of this chapter. Here, you are going to learn basic concept of these two ideas.

4.1 THE EVOLUTION OF THE CONCEPT OF EVOLUTION

The two major and contradictory ideas accounting for the origin of life on Earth are:

(a) special creation, (b) theory of evolution.

4.1.1 Concept of Special Creation

The supporters of special creation are called **creationists**. They have believed that, during limited period, God created the universe and man as supernatural event at a particular time in the past. This theory explains that every species was individually created by God in the form in which it exists today and is not capable of undergoing any change. They reject any other possible views and rely absolutely on inspiration, meditation and divine revelation.

4.1.2 Concept of Evolution

The supporters of evolution are called **evolutionists**. They have believed that the universe and man did not always exist in their present form; neither are they the product of a sudden creative act, but rather the result of innumerable changes from the lower to the higher, each step in advance being an evolution from a pre-existing condition. Modern biologists believe that the Earth is over five billion years old. Then about 3.5 billion years ago, life began. According to evolutionist, life on Earth had emerged as a unicellular prokaryote then with the passage of time variation have been accumulated, new species came into existence. The current biodiversity including man is the descendent of the earliest unicellular prokaryote that might had originated spontaneously.

Extra reading material

Conclusion: Faith accepts things for which there is no evidence in scientific sense. This means that logically there can be no intellectual conflict between scientific and theological accounts of creation, since they are mutually exclusion realm of thought. Scientific truth to the scientists is tentative, but theological truth to the believer is absolute. Since then process of special creation occurred only once and therefore cannot be observed. Science concern itself only with observation phenomena and such will never be able to prove or disprove special creation.

4.1.3 Origin of life according to concept of evolution

Evolutionists believe that first living being on earth belong to a group of prokaryotes, the Archaeobacteria, now known as Archaea. Evolutionists hypothesized that approximately more than 3.5 billion years ago the first living being originated on earth on earth in hot water springs (called hydrothermal vents) through spontaneous reaction among different inorganic and organic molecules. This hypothesis is called **vent hypothesis**.

The evolutionists also concluded that the early atmosphere of earth was reduced (without oxygen), hot and ozone less, therefore, frequent exposure of ultra violet radiation was there. The early prokaryotes were absorptive heterotrophs and then chemosynthetic autotrophs were came into existence.

The primitive earth environment had very little nutrients that would have limited early life. If life were to continue, another source of nutrients was needed so the photosynthetic habit was

probably involved in organisms. The first photosynthetic organisms probably used hydrogen sulphide as a source of hydrogen for reducing carbon dioxide to sugar. Later, water served this same purpose and oxygen liberated by photosynthetic reaction began to accumulate in the atmosphere. Earth and its atmosphere slowly began to change. Ozone in the upper atmosphere began to filter ultraviolet radiation from the sun, the reducing atmosphere slowly became oxidizing atmosphere, and at least some organisms began to utilize oxygen. About 420 million years ago, enough protective ozone had built up to make life on land possible. Ironically, the change from a reducing atmosphere to an oxidizing atmosphere also meant that life could no longer arise abiotically.

24.2 EVOLUTION FROM PROKARYOTES TO EUKARYOTES

As per fossil record the eukaryotes appeared 1.9 to 2.1 billion years ago. They arose from prokaryotes.

24.2.1 Theories about Evolution of Eukaryotes

There are two theories that have been put forward about the mechanism of evolution of eukaryotes from prokaryotes: (a) Membrane invagination theory, (b) Endosymbiosis theory.

Membrane invagination theory

A number of alternative hypotheses for the origin of eukaryotic cells have been suggested. One of these proposes that the prokaryotic cell membrane folded inward invaginated to enclose copies of genetic material. This several double membranous entities within a single cell were formed. These entities could then have evolved into the eukaryotic nucleus, mitochondrion and chloroplast.

Endosymbiosis theory

The endosymbiosis theory (*symbiosis* means living together and *endo* means 'within') was proposed by **Lynn Margulis**. The first step in the evolution of eukaryotic cell is thought to have occurred when a large anaerobic amoeboid prokaryote ingested a small aerobic bacterium and stabilized its prey as an endosymbiont rather than digesting it. This aerobic bacterium developed into a mitochondrion, the site of aerobic respiration in eukaryotic cells. Possession of such mitochondrion like endosymbiont conferred the advantage of aerobic respiration on its host. Flagella may have derived through the ingestion of prokaryotes similar to **spirochetes**.

Chloroplasts are thought to be derived from symbiotic photosynthetic bacteria. In addition, both mitochondria and chloroplasts are similar in size to bacteria, have their own DNA, have ribosomes similar to those of bacteria, and produce a limited portion of their own enzymes and proteins. To explain these observations it is suggested that the endosymbionts must have transferred, over time, some of their genes to the host nucleus and thus relinquished their independence for the sake of symbiotic relationship.

Regardless of the exact mechanism involved, the emergence of the eukaryotic cell led to a dramatic increase in the complexity and diversity of living organisms on Earth. At first, organisms were capable of existing only as independent single cells. Later, however, some

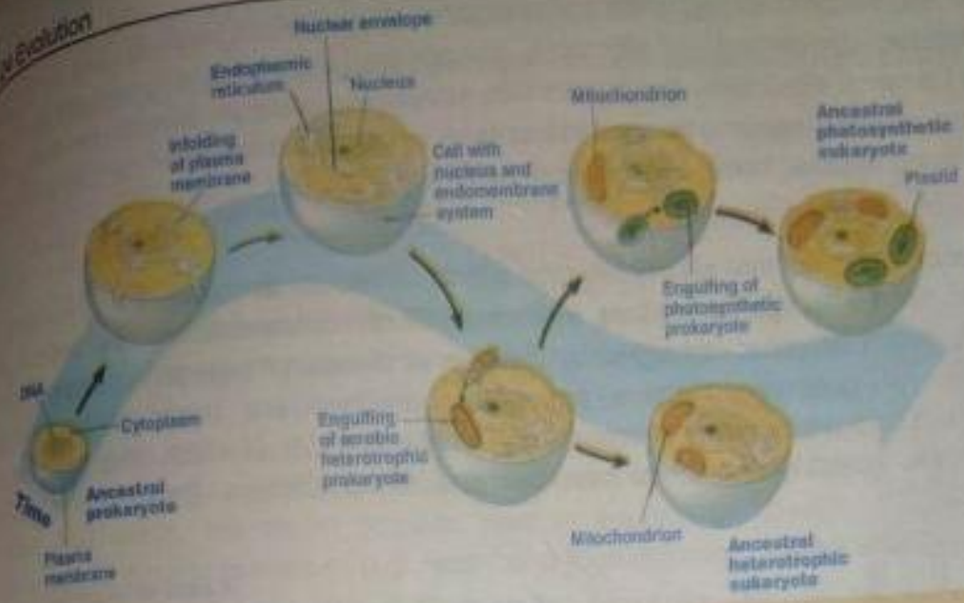


Fig. 24.1 Evolution of eukaryotic cell from prokaryotic cell

volved into multicellular organisms in which various cells became specialized for many different unctions. These multicellular forms became adapted to life in a great variety of environment.

24.3 LAMARCKISM

Jean-Baptiste Chevalier de La Marck, (1744-1829), is often known simply as **Lamarck**. He was a French naturalist, soldier and biologist. Lamarck was an early proponent of the idea that evolution occurred and proceeded in accordance with the natural laws. Lamarck is regarded as a premier authority on invertebrate zoology. He is remembered, at least as a taxonomist of considerable stature. In 1809, he published a book *Philosophie Zoologique* (Zoological Philosophy).

Fig: 24.2 Lamarck

24.3.1 Main Points of Lamarckism

The ideas about evolution presented by Lamarck are known as **Lamarckism**. He pictured evolution as a "ladder of life" from simplest to the most complex animals. Man was the top rung of this ladder. Lamarck did little in the way of explaining the origin of this ladder as a whole. However, he did offer an explanation for the origin of adaptations to the environment. Lamarck's contribution to science is important, because he was the first to propose that organisms undergo change over time as a result of some natural phenomena.

Lamarck's explanation of evolution revolved around two basic assumptions. The first one is the use and disuse of organs. Lamarck's second assumption is the inheritance of acquired characteristics.

Use and disuse of organs

Lamarck believed that some organs which are more frequently used by an organism are

developed and become strong while some organs which are not properly used by an organism are deteriorated, diminished and ultimately disappeared in successive generations. Among the examples Lamarck cited were the blacksmith developing a bigger bicep in the arm that works the hammer; giraffe stretching its neck to increase length to eat leaves of the tree and the snakes which are living in small holes and crevices have lost their legs.

Inheritance of acquired characteristics

Lamarck believed that characteristics which individual acquired during its lifetime were passed on to the offspring of that individual. Such characteristics are called **acquired characters** which are often emerged by the use or disuse of organs. According to Lamarck through several generations these acquired characters are continuously inherited and accumulated. Gradually a group of organisms would be produced which would be better able to "cope" with the environment due to inherited acquired characters. Evolution, in other words, would occur.

Evolution of giraffe neck

An example often used to illustrate Lamarck's hypothesis involves the evolution of the giraffe's long neck from short necked ancestors. In Lamarckian terms, this process would have occurred as follows. Each giraffe, during its lifetime, would try to reach the leaves at the top of trees. Each animal would constantly stretch its neck in order to attain this goal. As these individuals reproduced, the results of neck stretching (an acquired characteristic) would be passed on to future generation. Each offspring would be born with a slightly longer neck than those of its parents. Thus long necked giraffes gradually evolved.

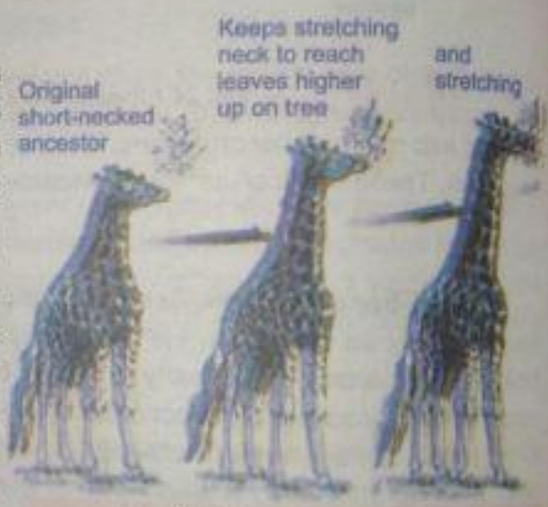


Fig. 24.3 Evolution of giraffe neck

24.3.2 Drawbacks of Lamarckism

The anatomical, biochemical and behavioural characteristics that an individual organism displays as it develops through life is known as its phenotype. However, the phenotype that an individual actually develops is somewhat conditional and is based on two key factors:

(1) The fixed genetic potential of the organism (or its genotype; this refers to the specific qualities of its genetic material, or DNA.)

(2) The environmental conditions which an organism experiences as it grows.

The first point of Lamarckism i.e., use and disuse of organs may be acceptable but the characteristics which are acquired through this process during the life time have no genetic bases and therefore cannot be inherited to the next generation. Actually the scientists of that time were unaware of the mechanism of inheritance which are proposed by Mendel in 1865.

Skills: Initiating and Planning

- **Hypothesize whether Lamarck was criticized in his day for advocating the ideas of evolution or for the mechanism he proposed.**

In 1802, society in general was un-accepting the ideas of evolutionary change, and evidence for evolution had not been developed thoroughly enough to convince most scientists. Thus, Lamarck was criticized in his day more for advocating ideas of evolutionary change than for the mechanism he proposed for that change.

24.4 DARWINISM

Study of God with respect to the universe.

Charles Darwin was born in England in a wealthy family. His father was a prominent physician. He joined Cambridge University to study theology, even so attended many lectures in biology and geology. He was only 22 in 1831 when he accepted the position of naturalist abroad on the HMS *Beagle*, a British Naval ship about to sail around the world. His major mission was to expand the navy's knowledge of natural resources e.g., water and food in foreign lands.

Fig.24.4 Charles Darwin

24.4.1 Darwin's Observations During his Voyage

The **Beagle** left Plymouth, England and cruised slowly along the east and west coasts of South America. He collected and catalogued thousands of plant and animal specimens and kept notes of his observations. The **Beagle** spent almost two months at the Galapagos (means tortoise) Islands. The islands are 965 kilometres west of Ecuador. Here Darwin made observations that were most important in the development of his ideas about evolution. finches (Birds) Types

Observations about South American mainland

He noticed that flora and fauna of different region of the continent had a definite South American stamp, very distinct from the life form of Europe. Further the South American fossils that Darwin found, though clearly different from modern species, were distinctly South American in their resemblance to the living plants and animals of the continent.

Observations about Galapagos islands

He compared the animals and plants of the Galapagos with those of the South American mainland. He was particularly impressed by their similarities and wondered why the organisms of the Galapagos should resemble those from South America more than those from other islands in different parts of the world. Moreover, although there were similarities between Galapagos and South American species, there were also distinct differences. The common birds were group of **finches**. Closely related species had beaks of very different sizes and shapes, adapted for feeding on completely different kinds of food. Darwin collected 14 types of finches on the Galapagos which are although quite similar but seemed to be different species. Some were unique to individual

Teacher's Point

The teacher would ask the students to answer that how did Lamarck's hypothesis of evolution contribute to scientific thought?

islands, while other species were distributed on two or more islands that were close together.

Darwin pondered these observations and tried to develop a satisfactory explanation for the distribution of species among the islands. Darwin perceived the origin of new species and adaptations as closely related processes. A new species would arise from an ancestral form by the gradual accumulation of adaptations to different environment, separated from original habitat by geographical barriers. Over many generations, the two populations could become dissimilar enough to be designated as separate species.

Fig.24.5 Map shows the journey of HMS Beagle around the world

Fig. 24.6 Darwin's theory of finches at Galapagos Islands

24.4.2 Development of the Theory of Evolution

Darwin began formulating his theory of natural selection in the late 1830s but he went on working quietly on it for many years. He wanted to amass a wealth of evidence before publicly presenting his idea. In 1842, Darwin wrote for himself, a brief 35-page sketch of his theory. Two years later he enlarged this into an essay of 230 pages, which he showed to his friends, but did not published it. For the next fifteen years, Darwin continued to collect facts to support his ideas.

The bases for the development of Darwin's theory of evolution were not only his observations about unique distribution of organisms in different regions of the world but he was also inspired by the work of many other scientists of that time. Therefore, the ideas of these scientists also contributed in the early development of Darwinism.

Contribution of Charles Lyell and James Hutton

In the early 1830's, **Charles Lyell** published a book *Principle of geology*. Darwin took this book on the voyage. This book presented arguments to support a theory of geological change proposed by **James Hutton** called **theory of uniformitarianism**. Lyell pointed out that the mountains, valleys, deserts, rivers, lakes and coastlines could have come through the action of existing forces and natural conditions. A river slowly carves a valley. Mountains are worn down to hills and finally plains. The slow pace of these geological processes, which still occurs today, indicated that Earth had to be much older than generally believed – a possibility that fired Darwin's imagination. If the earth of today is so old and so changed, what was it like thousands of years ago? Did it have the kinds of life we now have? What other life may have existed?

Contribution of Thomas R. Malthus

Darwin returned to England in1836. Soon afterwards, he read a work written by the English political economist **Thomas R. Malthus** (1766-1834), An Essay on the Principle of Population. Malthus noted that human populations have the capacity to increase **exponentially** (1⇒2⇒4⇒8⇒16) and food supply has the capacity to increase **arithmetically** (1⇒2⇒3⇒4⇒5). Such a relation could result only in a struggle for food and hence for existence itself.

Contribution of Alfred Russell Wallace

In 1858 Darwin received a letter from a fellow naturalist, **Alfred R. Wallace** (1823-1913), who was travelling at that time in Malays. Wallace enclosed an essay that he had written, and he asked Darwin to read it and then forward it to Lyell. In the essay, Darwin found, almost in his own terms, the theory of the origin of species by means of natural selection. Darwin almost yielded to Wallace the honour of being the first man to announce the theory. However, his friends (Charles Lyell and Joseph Dalton Hooker) arranged to present the two papers under joint authorship using a single title; *On the Tendency of Species to Form Varieties; and on the Perpetuation of Selection*. The papers were presented to the Linnaean Society in London on July1, 1858.

Why the theory was attributed to Darwin?

Alfred Russell Wallace was the man who motivated Darwin to publish his book *The Origin of Species by Means of Natural Selection*. It appeared in November of 1859. Only a passing reference to man's place in evolution was mentioned in *The Origin of Species*. Twelve years later, Darwin's *Descent of Man* was published. This was about the evolution of man. Darwin was much more willing to explore the implications of natural selection, particularly in relation to humans, than Wallace was. In addition, Wallace was a champion of rather radical social causes and later openly embraced spiritualism - all elements that resulted in the down-play of his role in the discovery of natural selection.

24.4.3 Darwin's Theory of Natural Selection

In his book *'The origin of species'* Darwin developed two main points i.e. (i) Descent with modification, (ii) Natural selection and adaptation.

(i) Descent with modification

Darwin believed and perceived unity in life, with all organisms related through descent from some common ancestors and that adaptation to various environments results diversity. In the Darwinian view, the history of life is like a tree, with multiple branching and re-branching from a common trunk all the way from the tips of the living twigs, symbolic of current diversity of organisms. At each fork of the evolutionary tree is an ancestor to all line of evolution branching from that fork.

(ii) IntraSpecific Struggle
(iii) Interspecific Struggle

(ii) Natural selection and adaptations

Natural selection refers to the differential reproductive capacities among the individuals of a population which indicates that some individuals of a population are capable to reproduce while others are not. Darwin's mechanism of evolution by natural selection consists of four observations about natural world.

(iii) Environmental Struggle

Over production: Each species has the capacity to produce more offspring than will survive to maturity. Through reproduction, natural populations may exponentially increase in number over time.

Variations: The individuals in a population exhibit variation in their traits. Some of these traits improve the chances of an individual's survival and reproductive success, whereas other traits do not.

Science Titbits

"Variation is a feature of natural populations and every population produces more progeny than its environment can manage. The consequences of this over production is that those individuals with the best genetic fitness for the environment will produce offspring that can more successfully compete in that environment. Thus the subsequent generation will have a higher representation of these offspring and the population will have evolved."An extract from Darwin's book "on the origin of species through natural selection"

Struggle for existence: Over production leads to the competition among the individuals of a population for the limited resources food, water light, growing space. Because there are more individual than the environment can support, not all will survive to reproductive age. Other limits on population growth include predators and disease causing organisms. The struggle may be interspecific, intraspecific and environmental.

Survival of the fittest: Those individuals that possess the most favourable combination of characteristics are most likely to survive and reproduce, passing their heritable traits on to the next generation. For example if there is sudden flood only those organisms that can swim or respire in water, have a better chance to survive and other will die or if there is an earthquake the flying animals have a better chance of survival. This is called **natural selection**. It is also referred as the survival of the fittest. The fittest individuals are those that reproduce most successfully in the environment.

The processes of natural selection thus cause an increase of favourable alleles and a decrease of unfavourable alleles within the population. Over succeeding generations, individual members become better adapted to local conditions, thus, leading to the evolution of new species.

24.5 NEO-DARWINISM

When Lamarck and Darwin put forward their ideas, practically nothing was known about heredity. The emergence of population genetics has provided a clear understanding of inheritance and variation among the individuals of a population and firm support for Darwinian theory. This reappraisal of the theory of natural selection in terms of modern population genetics is sometimes called **Neo-Darwinism**.

24.5.1 Evidences of Evolution

In this section, you will learn about evidences of evolution from biogeography, palaeontology, comparative anatomy and molecular biology.

Evidence from Biogeography

The study of geographical distribution of plants and animals on Earth is called **biogeography**. Biogeography gives evidence of prehistoric climates, habitats and animal distribution pattern. Biogeographic studies show that life-forms in different parts of the world have distinctive evolutionary history.

Specific pattern of distribution: Darwin noticed that South America lacked rabbits, even though the environment was quite suitable to them. He concluded that there are no rabbits in South America because rabbits originated somewhere else and they had no means to reach South America.

Factors inhibiting distribution of organisms: Bio-geographical studies show that species have restricted distribution from the centre of origin due to some kind of barrier like physical such as an ocean, desert or mountain, environmental such as an unfavourable climate; or ecological such as the presence of organisms that compete with it for food as shelter.

Evidence from Palaeontology

Palaeontology is the science of discovery, identification and interpretation of fossils. The succession of fossil forms is strong evidence in favour of evolution. It provides a visual record in a complete series showing the evolution of a species.

Oldest known fossils: For instance evidence from other modern biological sciences places prokaryotes as the ancestors of all life and predicts that prokaryote should precede all eukaryotic life. In the fossil record, indeed, the oldest known fossils are prokaryotes.

Chronological sequence of vertebrate fossil: There is chronological sequence of the different classes of vertebrates in the fossil record. Fossil fishes the earliest vertebrates, with amphibians next, followed by reptiles then mammals and birds. This sequence is consistent with the complexity of their organ system.

Teacher's Point

The teacher would ask the students that "how the process of survival of the fittest related to population's environment?

Sometimes, the fossil record allows us to trace the history of one particular organism, such as modern day horse *Equus*. The earliest horses had four toes. Over the time the number of toes reduced to three, in the modern horses to one, a large central toe that ends in a hoof. The evidences of fossil record support the common descent hypothesis.

Science, Technology and Society Connections

Describe and analyze examples of technology that have extended or modified the scientific understanding of evolution (e.g., the contribution of radiometric dating to the paleontological analysis of fossils).

Geologists and palaeontologists, use several techniques to determine the actual ages of rocks and the fossils they contain. The most common method is called **radiometric dating** or often called **radioactive dating**, is based on the fact that living organisms contain certain radioactive isotopes in certain ratios. For instance, living organisms have the same constant ratio of ^{14}C, a radioactive isotope to ^{12}C, a stable isotope, as does Earth's atmosphere. However, when an organism dies, its ratio of ^{14}C to ^{12}C starts to drop, because ^{14}C decays to other chemical elements, and the organisms no longer obtains any ^{14}C from the atmosphere. Each radioactive isotope has a fixed rate of decay known as half-life. The **half-life** is the amount of time it takes for one half of the initial amount of the parent radioactive isotope, to decay to the daughter isotope. For example, ^{14}C has a half-life of 5600 years, meaning that half of the ^{14}C in a specimen decays in about 5600 years, half remaining ^{14}C decays in the next 5600 years and so on, until all the ^{14}C is gone. Knowing both the half- life of a radioactive isotope and the ratio of radioactive to stable isotope in a fossil enables us to tell how old the fossil is. For instance, if a fossil has a ^{14}C to ^{12}C ratio half that of atmosphere, it is about 5600 years old; a fossil with one-fourth the atmosphere's ratio is about 11,200 years old.

Evidence from Comparative Anatomy

Comparative study of the anatomy of groups of animals or plants reveals that certain parts of the organisms have similarities in structures while others have similarities in functions.

Homologous organs represent divergent evolution: Body parts that are similar in structure but different in function because they were inherited from a common ancestor are called homologous structures and their similarity is called homology. This pattern of evolution in which different species have been evolved from common ancestors at different habitats is known as divergent evolution.

For example, the basic structure of all the flowers is same. Similarly the limb-bone pattern of all tetrapods from amphibian to mammals has the same structural plan. It is called pentadactyl limb. Vertebrate forelimbs are used for flights (birds and bats), orientation during swimming (whales and dolphins) running (horses), climbing (arboreal lizard), or swinging from tree branches, yet all vertebrate forelimbs contain the same sets of bones organized in similar ways despite their dissimilar functions.

Teacher's Point

The teacher would ask the students to answer that darwin found fossils of many organisms that were different from any living species. How would this finding have affected his understanding of life's diversity?

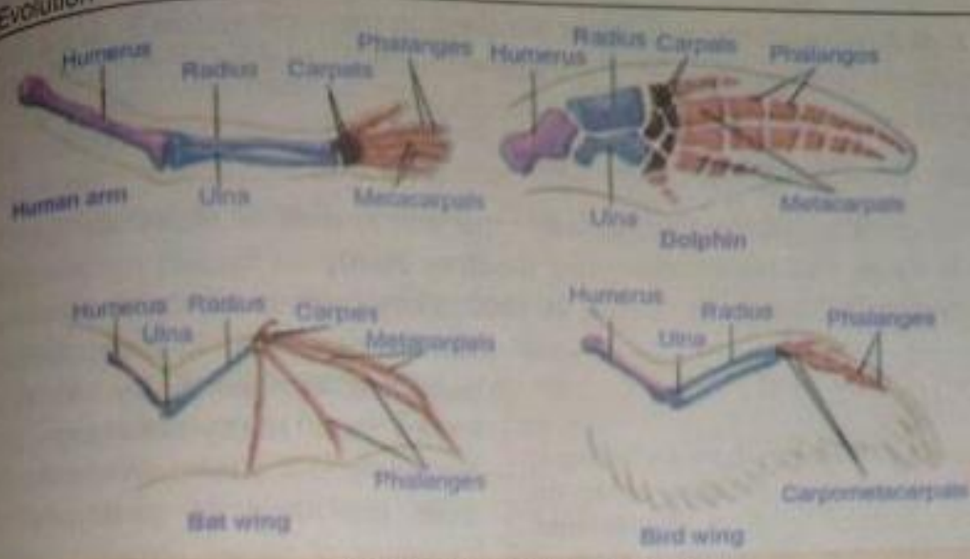


Fig: 24.7 Homologous structure

Science, Technology and Society Connections

List the vestigial structures found in man and categorize them in homologous or analogous structures.

Human vestigial organs are those organs that have lost all or most of their original functions through evolution. There are about 90 such structures present in our body. (1) **Vermiform appendix** is a vestige of the cecum. (2) **Coccyx** or **tailbone** is the remnant of a lost tail.(3) The **wisdom teeth** are vestigial third molars that human ancestors used to help in grinding down plant tissue. (4) Humans have **ear muscles** that are minimally developed and non-functional, but some people are able to move their ears in various directions.

Skills: Analyzing and Interpreting

- **What factors have contributed to the dilemma that pharmaceutical companies face in trying to develop new antibiotics because so many micro-organisms are resistant to existing antibiotics?**

Antibiotic resistance is a type of drug resistance where a microorganism is able to survive exposure to an antibiotic. While a spontaneous or induced genetic mutation in bacteria may confer resistance to antimicrobial drugs, genes that confer resistance can be transferred between bacteria. Thus a gene for antibiotic resistance which had evolved via natural selection may be shared. Evolutionary stress such as exposure to antibiotics then selects for the antibiotic resistant trait.

Analogous organs represent convergent evolution: On the other hand the organs which are similar in function but differ in structure are called analogous organs, e.g., wings of the bird and butterfly. Analogous structure are of evolutionary interest because they demonstrate that population with separate ancestries may adapt in similar ways to similar environmental demands. This pattern of evolution in which different species have been evolved from different ancestors at a common habitat is known as convergent evolution.

Evidence from Molecular Biology

Almost all living organisms use the same basic biochemical molecules, including DNA, ATP and identical enzymes. Further, organisms utilize the same DNA triplet code and the same twenty amino acids in their proteins. Organisms even share the same type of interons.

Cytochrome 'c' is a molecule that is used in the electron transport system in all the organisms. There is obviously no functional reason why these elements need to be similar, but their similarity can be explained by descent from a common ancestors.

24.5.2 Hardy-Weinberg Theorem

The mathematical relationship between the frequencies of alleles and genotypes in population was developed independently by **Godfrey Hardy** an English mathematician, and **Wilhelm Weinberg**, a German physician, in 1908. They pointed out that the frequencies of various genotypes in a population can be described mathematically which is now known as **Hardy-Weinberg principle**. According to the Hardy-Weinberg principle, "both the ratios of genotypes and the frequency of alleles remain constant from one generation to the next in a sexually reproducing population, provided other conditions are stable."

Science Titbits

The branch of biology that deals with the mechanism of inheritance and the origin of variations among the individuals of a population is known as **population genetics**. A population is defined as a group of individuals of the same species that live together at the same area at the same time. For example a group of 500 pea plants in a field. The total number of individuals of a population in per unit area is called population density. The genetic constitution of a population, i.e., the sum total of all the different genes in the population, is known as the **gene pool**. The term gene pool can also be applied to the total number of all the genes or alleles for a particular trait like flower colour etc. For example the gene pool of flower colour in the given population consists of 1000 genes. The total number of a particular gene or allele for a particular phenotype in the population is called allele/gene frequency. For example: gene/allele frequency of a dominant allele (A) or recessive allele (a) in the population. The genotype frequency refers to the total number of individuals of a particular genotype in the population. For example: the total number of dominant homozygotes (AA) or recessive homozygotes (aa) or heterozygotes (Aa). If all the individuals of a population are homozygous for a particular gene/allele, the gene/allele is called fixed gene/allele.

Conditions/assumptions for stability

As long as any population remains free of outside interference, it will remain in genetic equilibrium i.e., ratios and proportions of gene frequencies and genotype frequencies will remain same. Real populations like *Drosophila*, however, are rarely free from outside influences and they never totally meet the following five basic conditions for stability:

(a) Reproduction must be totally random. (b) There must be no gene flow. (c) Populations must be large. (d) There must be no mutations. (e) There must be no selection.

In an ideal population left undisturbed by any of the five conditions listed above, there would be complete genetic equilibrium. The population would always contain the same type of genes represented in the same ratios within the same phenotypes.

Over long periods of time, this population would never change. Actually, in the real world, this does not happen. A definition of evolution, therefore, could be; *"Deviations from genetic equilibrium leads to evolution"*

Factors that change allele frequencies

The evolution of a species to occur, the gene frequencies of that population must undergo change but under certain conditions these frequencies may remain constant over time. However, a number of factors can lead to change in allele frequencies. They are as follows.

a) **Migration/gene flow:** It is the movement of individuals from one population to another. Whether it is movement of foreign individuals into the population (**emigration**) or it is outward movement of individuals form the population (**immigration**), in both cases allele frequencies will change accordingly.

b) **Mutation:** It is a major source of variations. New alleles are arisen due to mutation but it occurs so rarely that it alone does not change allele frequency much.

c) **Non-random mating:** It is mating among specific group of individuals in a large population. Although new alleles cannot be developed by non-random mating but it can cause an increase in homozygous genotypes.

d) **Selection:** Some individuals leave behind more progeny than others, and the rate at which they do so is affected by their inherited characteristics. This is called **selection**. It may be artificial selection or natural selection. In artificial selection breeder select for the desirable traits while in natural selection, the environment plays this role. In both cases frequencies of alleles can be affected.

Science Tidbits

The Hardy-Weinberg equation is not only important in population genetics, public health scientists also use it to estimate the parentage of people carrying for certain diseases. Estimating the frequency of a harmful allele is useful for any public health programs dealing with genetic disease.

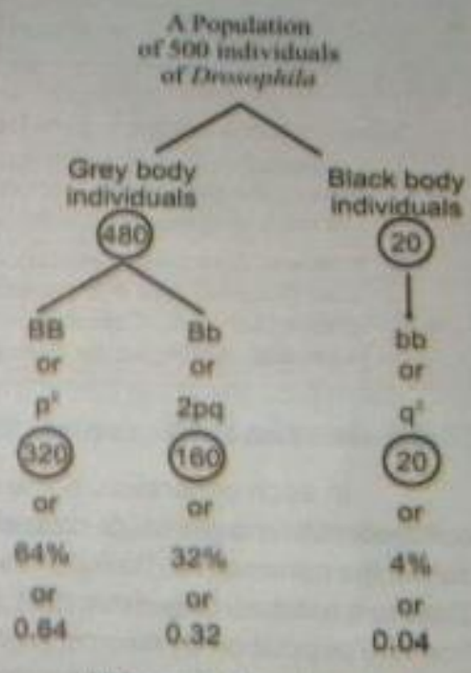


Fig. 24.8 Example of Hardy-Weinberg equilibrium in population

y-Weinberg equation

A general formula called the Hardy-Weinberg equation can be used for calculating the encies of alleles and genotypes in a population which is in genetic equilibrium or Hardy-berg equilibrium. If all alleles of a given locus are the same in the population, the frequency t allele is one. Such allele is called **fixed allele**. In fruit flies the allele for grey body ***B*** is ant over the allele for black body colour ***b***. Because only two alleles ***B*** and ***b*** exist for the

Teacher's Point

Teacher would ask the students to explain the impact of a growing population on society and environment

locus, the sum of their frequencies must be equal to one. If we let ***p*** represent the frequency of the dominant ***B*** allele and ***q*** the frequency of the recessive ***b*** allele in the population, then we can summarize their relationship with a simple binomial equation, $p + q = 1$. (A binomial equation is an algebraic expression that consists of two quantities connected by plus or minus sign) When we know the value of either p or q, we can calculate the value of the other.

$P = 1 - q$ and $q = 1 - p$, Because $p + q = 1$, then $(p + q)^2 = 1$. This binomial equation can be expanded to describe the relationship of the allele frequencies to the genotypes in the population. When it is expanded, we obtain the frequency of the offspring genotypes:

p^2	+	$2pq$	+	q^2	=	1
(Frequency of BB)		(Frequency of Bb)		(frequency of bb)		(all individuals in a population)

Skills: Analyzing and Interpreting

- **Solve problems related to gene frequencies using the Hardy-Weinberg equation**
  1. **Problem:** The allele for grey body colour **B** is dominant to black body colour **b**. There are 30% recessive alleles in the gene pool of population of 1000 individuals. Calculate the number of grey body individuals and black body individuals in the population.
  2. **Problem:** Suppose that in a population of 1000 individuals 50% of people are homozygous for free ear lobes (Free ear lobe **F** is dominant to the attached ear lobes **f**) whereas 16% are homozygous for attached ear lobes. Calculate the frequencies of the **F** and **f** alleles and also workout the number of heterozygous individual for free ear lobe.

24.6.2 Genetic Drift (Natural Selection)

In each generation, some individuals may, just by chance, leave behind a few more descendants (and genes, of course!) than other individuals. The genes of the next generation will be the genes of the "lucky" individuals, not necessarily the healthier or "better" individuals. That, in a nutshell, is **genetic drift**. It happens to all populations. One allele may be eliminated from the population by chance, regardless of whether that allele is beneficial, harmful or of no particular advantage or disadvantage. Although genetic drift occurs in both large and small populations, a large population is expected to suffer less. When a population is small there is a greater chance that some rare genotypes may be lost in the next generation, if few individuals fail to reproduce. Genetic drift can decrease genetic variation within a population although it tends to increase the genetic differences among different population.

Fig. 24.9 Genetic drift

The two important causes of genetic drift are: (a) bottleneck effect (b) founder effect.

Bottleneck effect

The reduction of population size with some specific allele and genotype due to natural disaster is called **bottleneck effect**. For example, events such as earthquakes, floods, or fires may kill large

bers of individuals unselectively, ducing a small surviving population l is unlikely to have the same etic makeup as the original ulation but have different portions and ratios of allele and type frequencies.

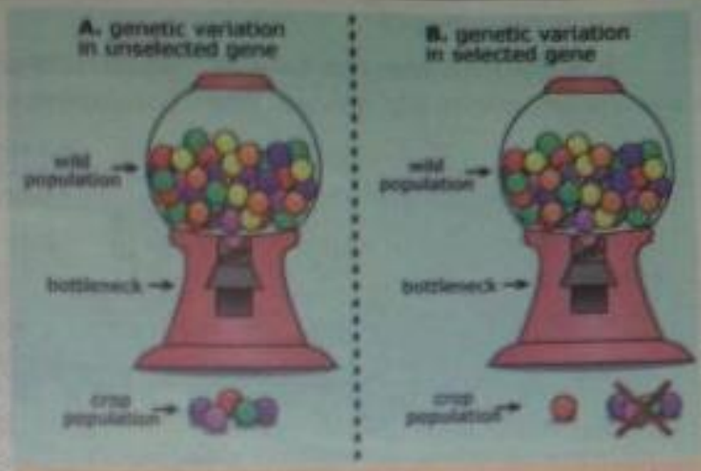


Fig. 24.10 A bottleneck shows the reduction in population size with specific genotypes and alleles

nder effect

The founder effect is a icular example of the influence of dom sampling. It is defined as: e establishment of a new ulation by a few original founders an extreme case, by a single ized female) which carry only a all fraction of the total genetic variation of the parental population."

If a small sample of individuals is taken from a larger population, there is a chance that an ele will be lost. In the case of two alleles, *A* and *a*, if the founding population are all *aa* mozygous then the *A* allele will be lost and the new population will be genetically nomorphic. In fact, the founding effect is quite ineffective at reducing genetic variation - even he founding population is very small, even less than 10, it will usually possess both alleles.

6.3 Speciation

The evolutionary process by which new biological species arise is called **speciation**. There e three modes of speciation: allopatric speciation, sympatric speciation and parapatric eciation. Each idea is based on the degree to which populations undergoing this process are ographically isolated from one another.

mpatric speciation

It occurs when populations of a species that share the same habitat become roductively isolated from each other. It ll commonly occurs through yploidy. A tetraploid individual cannot te with a diploid individual, creating roductive isolation. Sympatric eciation is rare. It occurs more often ong plants than animals, because a ploid plant can fertilize itself and ate offspring. For a tetraploid animal to roduce, it must find another animal of same species but of opposite sex that also randomly undergone polyploidy.

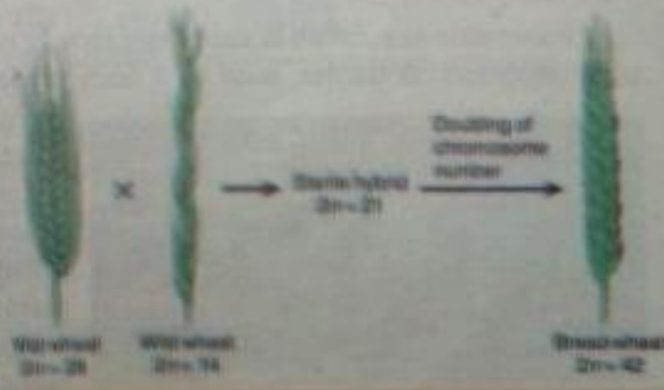


Fig. 24.11 Sympatric speciation in wheat

Allopatric speciation

It is the most common form of speciation. It occurs when populations of a species become geographically isolated. When populations become separated, gene flow between

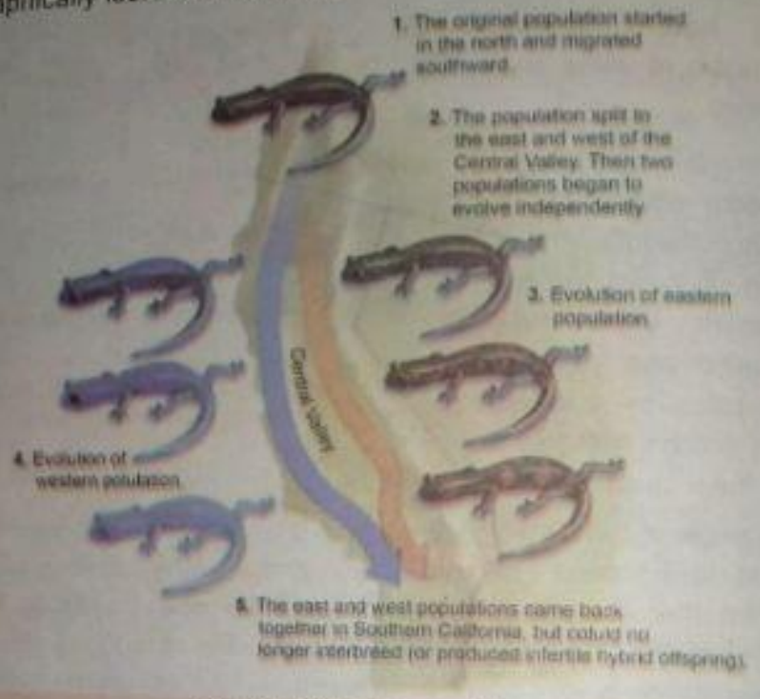


Fig. 24.12 Allopatric speciation

them ceases. Over time, the populations may become genetically different in response to the natural selection imposed by their different environments. If the populations are relatively small, they may experience a founder effect: the populations may have contained different allelic frequencies when they were separated. Selection and genetic drift will act differently on these two different genetic backgrounds, creating genetic differences between the two new species.

Parapatric speciation

Parapatric speciation is extremely rare. It occurs when populations are separated not by a geographical barrier, such as a body of water, but by an extreme change in habitat. While populations in these areas may interbreed, they often develop distinct characteristics and lifestyles. Reproductive isolation in these cases is not geographic but rather temporal or behavioural. For example, plants that live on boundaries between very distinct climates may flower at different times in response to their different environments, making them unable to interbreed.

Fig. 24.13 Parapatric speciation

Interesting Facts

mbulocetus natans was present in early cetacean ould walk as well as swim. It is a transitional fossil that hales have evolved from land-living mammals. *us natans* lived from the early to middle Eocene (50 to years ago). When the animal was alive, Pakistan was a ion of India, which was then a large island in the Indian *mbulocetus natans* was recovered from the Upper ormation of Pakistan (near Islamabad) in 1993 by G.M. Thewissen and Sayed Taseer Hussain.

as described by Thewissen, Hussain, and Mohammad Arif in 1994. Scientists consider *Ambulocetus* to y whale because it shares underwater adaptations with them: it had an adaptation in the nose that to swallow underwater, and its bones had a structure like those of whales, enabling it to hear well r. In addition, its teeth are similar to those of early cetaceans. It was the size of a sea lion.

Activity

t different homologous and analogous structures through observations in plants.

Exercise

M.C.Qs

ct the correct answer

Using the Hardy-Weinberg Principle, which expression represents the frequency of he homozygous recessive genotype?

A) P^2 (B) 2Pq (C) q^2 (D) q

The process of ----- and------ generate variation, and ------- produces adaptation to he environment.

A) sexual recombination ----- natural selection ----mutation
B) genetic drift----mutation ---- sexual recombination
C) mutation ------- sexual recombination ----natural election
D) mutation------ natural selection-----genetic drift

atural selection is sometimes described as "survival of the fittest." Which of the llowing most accurately measures an organism's fitness?

A) its mutation rate
B) how many fertile offspring it produces
C) its ability to withstand environmental extremes
D) how much food it is able to make or obtain.

(iv) Which of the following is a true statement about Charles Darwin?
(A) He was the first to discover that living things can change, or evolve.
(B) He based his theory on the inheritance of acquired characteristics.
(C) He worked out the principles of population genetics.
(D) He proposed natural selection as the mechanism of evolution.

(v) In science, the term theory generally applies to an idea that
(A) is a speculation lacking supportive experiments
(B) attempts to explain many related phenomena
(C) is synonymous with what biologists mean by hypothesis
(D) is considered a law of nature

(vi) The smallest biological unit that can evolve over time is
(A) a specie
(B) an individual organism
(C) an ecosystem
(D) a population

(vii) Which of the following ideas is common to both Darwin's and Lamarck's theories of evolution?
(A) Adaptation results from different reproductive success.
(B) Evolution drives organisms to greater and greater complexity.
(C) Evolutionary adaptation results from interactions between organisms and their environment.
(D) The fossil record supports the view that species are fixed.

(viii) Which of the following pairs of structures is least likely to represent homology?
(A) the wings of a bat and the forelimbs of a human
(B) the haemoglobin of a baboon and that of a gorilla
(C) the brain of a cat and that of a dog
(D) the wings of a bird and those of an insect

(ix) All organisms share the same genetic code. This commonality is evidence that
(A) evolution is occurring now
(B) convergent evolution has occurred
(C) all organisms are descended from a common ancestor
(D) evolution occurs gradually

(x) Which of the following is an example of vestigial structure in human?
(A) human tailbone
(B) nipple on male mammals
(C) sixth fingers found in some humans
(D) human knee cap

Short Questions

What is the concept of special creation?
What is the concept of evolution?
Explain the chronological sequence of vertebrate fossil.
What is half life?
What is convergent evolution?
What is divergent evolution?
How analogous organs represent convergent evolution?
Make a list of the vestigial structures found in man and categorize them in homologous or analogous structures.
What factors have contributed to the dilemma that pharmaceutical companies face in trying to develop new antibiotics because so many microorganisms are resistant to existing antibiotics?
Describe the evidence from molecular biology in support of theory of evolution?
Describe membrane invagination theory.
Justify Lamarck as an early proponent of evolution.
Describe the drawbacks of Lamarckism.
Describe Darwin's observations about South American mainland.
What is the contribution of Charles Lyell and James Hutton in the development of the theory of evolution?
What is the contribution of Thomas R. Malthus in the development of the theory of evolution?
What is the contribution of Alfred Russel Wallace in the development of the theory of evolution?
Why the theory of evolution was attributed to Darwin?
What is meant by 'descent with modification?"
What is neo –Darwinism?
What is Hardy Weinberg principle?
Describe genetic drift.
What are the factors that change allele frequencies?
Define/Describe/Explain briefly.

evolution, radioactive dating, homology, analogy, Lamarckism, theory of uniformitarianism, Neo-Darwinism, Hardy Weinberg principle, bottleneck effect, founder effect, speciation, sympatric speciation, allopatric speciation, parapatric speciation.

26. Write the difference between
   (a) creationists and evolutionist
   (b) homologous and analogous organs
   (c) Lamarckism and Darwinism
   (d) New-Darwinism and Hardy and Weinberg principle
   (e) genetic drift and bottleneck effect
   (f) sympatric and allopatric speciation
   (g) allopatric and parapatric speciatin

Extensive Questions

27. How can you describe that the theory of evolution are two contradictory ideas?
28. What is biogeography? How biogeography provides an evidence of evolution?
29. What is paleontology? How paleontology provides an evidence of evolution?
30. Explain the contribution of radiometric dating to the paleontological analysis of fossils.
31. How evidences from comparative anatomy support theory of evolution?
32. Describe the two theories regarding the evolution of Eukaryotes.
33. Give a detail account of Lamarckism. What is its drawback?
34. Describe the development of theory of evolution.
35. Describe Darwin's observations about Galapagos Island.
36. Describe Darwin's theory of natural selection.
37. Explain Hardy and Weinberg theorem.
38. Describe genetic drift.
39. What is speciation? Describe the three types of speciation.

23

CHROMOSOME AND DNA

After completing this lesson, you will be able to

This is a 12 days unit

- Critically analyze the history of chromosomal theory with reference to Correns' work.
- Critically analyze the experiments of T. H. Morgan in support of the above-mentioned theory.
- Annotate the detailed structure of a chromosome.
- Describe the concept of gene and gene locus.
- Explain the concept of alleles as the alternative forms of a gene.
- Narrate the experimental work of Griffith and Hershey-Chase, which proved that DNA is the hereditary material.
- Describe the three models proposed about the mechanism of DNA replication.
- Narrate the work of Meselson and Stahl to justify the semi-conservative replication as the correct method of replication.
- Describe the events of the process of DNA replication.
- Explain DNA stability and variability as two characters of the replicating DNA molecule.
- Describe the central dogma of gene expression.
- Define gene and genetic code.
- Describe the characteristics of genetic code (universal, triplet, non-overlapping, degenerate, punctuated).
- Differentiate between the terms genetic code and codon.
- Explain the mechanism of transcription.
- Explain why the length of transcribed m-RNA molecule (in Eukaryotes) shortens as it enters the cytoplasm for translation.
- Describe the mechanism of protein synthesis.
- State the difference between protein synthesis in prokaryotes and eukaryotes.
- Suggest possible ways in which the synthesized protein can be used within or outside a cell that synthesized it.
- State the importance of the regulation of gene expression.
- Describe the negative control of gene expression by repressor proteins.
- Describe the positive control of gene expression by activator proteins.
- Relate gene expression with introns and exons.
- Define mutation and identify various sources of mutation.
- Differentiate between natural and induced mutations and mutagens.
- Justify that most mutations are harmful.

- Rationalize that mutations might be a contributing factor towards evolution.
- Describe the symptoms, causes and possible available treatments of some of the chromosomal mutations. (Down's, Klinefelter's and Turner's syndrome)
- Describe the symptoms, causes and possible available treatments of some of the gene mutations. (Sickle cell anemia, Phenylketonuria)

Reading

Gregor Mendel's "hereditary factors" were purely an abstract concept when he proposed their existence in 1865. At that time, no cellular structures were known that could house for these imaginary units. With the advancement of molecular genetics, now it is clearly known that genes, the Mendel's "factors" are specific segments of DNA and are located in chromosomes. It is now possible to determine their locations within chromosomes, to understand their mode of expression and to observe the nature of mutation in them. This chapter deals with all these molecular aspects of genes and chromosomes.

23.1 CHROMOSOMAL THEORY OF INHERITANCE

Mendel and Darwin's work laid the foundation for formulating a testable, research-based theory of heredity. In 1900, however, Mendel's work was "re-discovered" by three European scientists, Hugo de Vries, Carl Correns and Erich von Tschermak.

23.1.1 Origin of Chromosomal Theory of Inheritance

The chromosomal theory of inheritance is the idea that "genes, the units of inheritance, are found in the chromosomes, so chromosomes act as carriers of heredity".

1865 → Mendelian genetics
1882 → Discovery of Chromosome

Contributions of Walther Fleming and Waldeyer

Emergence of chromosomal theory of inheritance is linked with the discovery of chromosomes which were first observed by German embryologist Walther Fleming in 1882, when he was examining the rapidly dividing cells of salamander larvae. The term "**chromosome**" was proposed by Waldeyer, which literally means coloured bodies. Since their discovery, chromosomes have been found in the cells of all eukaryotes. However, in prokaryotic cell, its single DNA molecule is also referred as chromosome.

1900 → genes present on chromosome (but no evidence)
1902 → parallel behaviour

Stranded

Contributions of Carl Correns

For the first time, the relationship of heredity units with chromosomes was put forward in 1900 by a German geneticist **Carl Correns**, in one of the paper announcing the rediscovery of Mendel's work, but he had no supportive evidences for this idea.

Contributions of Walter Sutton and Theodor Boveri

The actual credit of this theory goes to both **Walter Sutton** (an American who at that time was a graduate student) and **Theodor Boveri** (a German biologist). In 1902, these scientists recognized independently that the behaviour of Mendel's factors (genes) is parallel to the behaviour of chromosomes at meiosis.

In addition to the evidences (given in table 23.1) based on parallel behaviour between genes and chromosome during meiosis, we can also analyze the mechanism of sexual reproduction, which involves the initial union of two cells, egg and sperm. If Mendel's model is correct, then these two gametes must make equal hereditary contributions. Sperm, however, contains little cytoplasm and during fertilization it only contribute nucleus to the zygote. Therefore, the hereditary units must reside within the nucleus of the gametes, whereas chromosomes are also found in the nucleus. Beside above mentioned parallel behaviour between genes and chromosome during meiosis, this observation also indicates that genes would be present in chromosomes.

Table 23.1 Parallel behaviour of genes and chromosomes during meiosis.

Behaviour of chromosomes (During meiosis)	Behaviour of genes (mendelism)
1. Diploid cells (before meiosis) have two copies of each chromosome (homologous pairs) while gametes (after meiosis) have only one. For example, in pea plant diploid cells have 7 pairs of homologous chromosomes while gametes have single 7 chromosomes.	1. According to the Mendel, diploid cells have two copies of each gene (pair of alleles) while gametes have only one. For example, in pea plant, diploid cells have a pairs of alleles for each gene like *Rr*, *Yy*, and *Tt*, while gametes have single *R* or *r*, *Y* or *y* and *T* or *t*
2. Homologous pairs of chromosomes segregate during meiosis.	2. According to the Mendel, pair of gene for each trait also segregates from each other during meiosis.
3. During meiosis, each pair of homologous chromosomes orient on the metaphase plate independently of any other pair so that in anaphase each pair assort independently of the other.	3. According to the Mendel, alleles of one gene pair also assort independently to the alleles of other gene pair during meiosis. e.g. *RrYy* genotype as a result of independent assortment can form four type of gametes i.e., *RY*, *Ry*, *rY*, and *ry*.

Many investigators of that time pointed out a serious objection on Sutton's theory. According to that, let we accept that Mendelian traits are determined by the factors (genes) located on chromosome, and if the genes are segregated due to segregation of chromosome and independent assortment of genes is reflected by the independent assortment of chromosome in meiosis, why is it that number of genes that assort independently of one another in a given kind of organism is often much greater than the number of chromosome pairs that the organism possesses.

Contributions of T. H. Morgan

After some years the objection was cleared after the discovery of linkage by the historical experimentation of **T. H. Morgan** in 1910 on *Drosophila*. Morgan's work with respect to the gene

inheritance of eye colour in *Drosophila* (you have already studied in the previous chapter) explains that genes for eye colour in *Drosophila* are located in X chromosome.

Thus, the chromosomal theory of inheritance was not the work of a single scientist, but rather the collaborative result of multiple researchers working over multiple decades. Scientists were finally able to confirm what they had long suspected, that chromosomes were indeed the physical carriers of hereditary information.

T. H. Morgon

23.1.2 Morphologyof Chromosomes

Chromosomes are thick thread like structures that appear in nucleus during cell division. In an interphase cell, chromosome become uncondensed and look like very fine network called **chromatin network**.

Structure of chromosome

A typical chromosome consists of two strands called **chromatids**; each is made up of a long DNA molecule which is highly coiled along with histone proteins. The chromatids contain genes for different traits. Generally both chromatids are attached with each other at a point known as **centromere** or **primary constriction**, so each chromosome shows two **arms** (region from centromere to an end). Some chromosomes may have another point of union along the length of chromatids, called **secondary constriction** or **nucleolar organizer**. It gives rise to nucleoli during interphase. At least, one pair of homologous chromosomes possesses nucleolar organizer region. Beside secondary constriction, the end becomes a knob like structure called **satellite**. This region has a useless sequence of DNA called **junk DNA**. The terminal ends of chromosomes are called **telomeres** which prevent the two chromosomes to attach with each other from their ends.

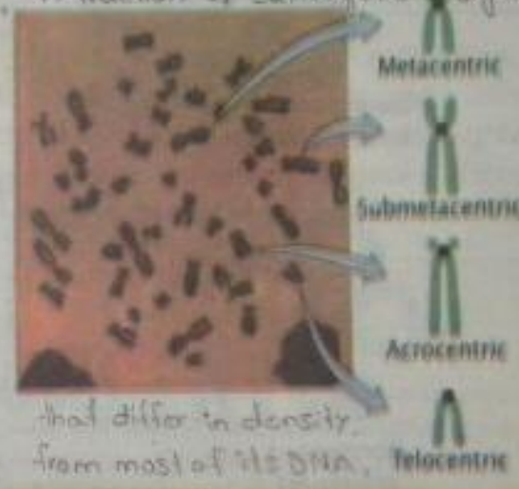


Fig. 23.1 Types of chromosomes

On the basis of position of centromere along the length, a chromosome may be called **metacentric:** centromere located in the centre, **submetacentric:** centromere located slightly away from the centre, **acrocentric:** centromere located near the end, and **telocentric:** centromere located at an end.

Composition and organization of chromosome

Generally a chromosome is made up of 40% DNA and 60% protein. In the cell, which is ready to divide, the chromosome has two identical DNA molecules i.e., each chromatid has one DNA molecule. An average sized human chromosome has approximately 5 cm long DNA which consist of about 140 million nucleotides. So can you imagine how such a huge molecule fit into

a tiny chromosome? DNA is a negatively charged molecule because of phosphate groups therefore it has strong affinity to **histone proteins**, which are positively charged unlike many other proteins due to the abundance of some basic amino acid such as arginine and lysine.

During S phase of cell cycle, DNA and histones are completely disorganized from each other, but after DNA is replicated, both DNA and histones begin to organize again and the process of condensation remains continue till the cell undergoes division and the chromosome are appeared. The organization of chromosomes occurs in **four levels**:

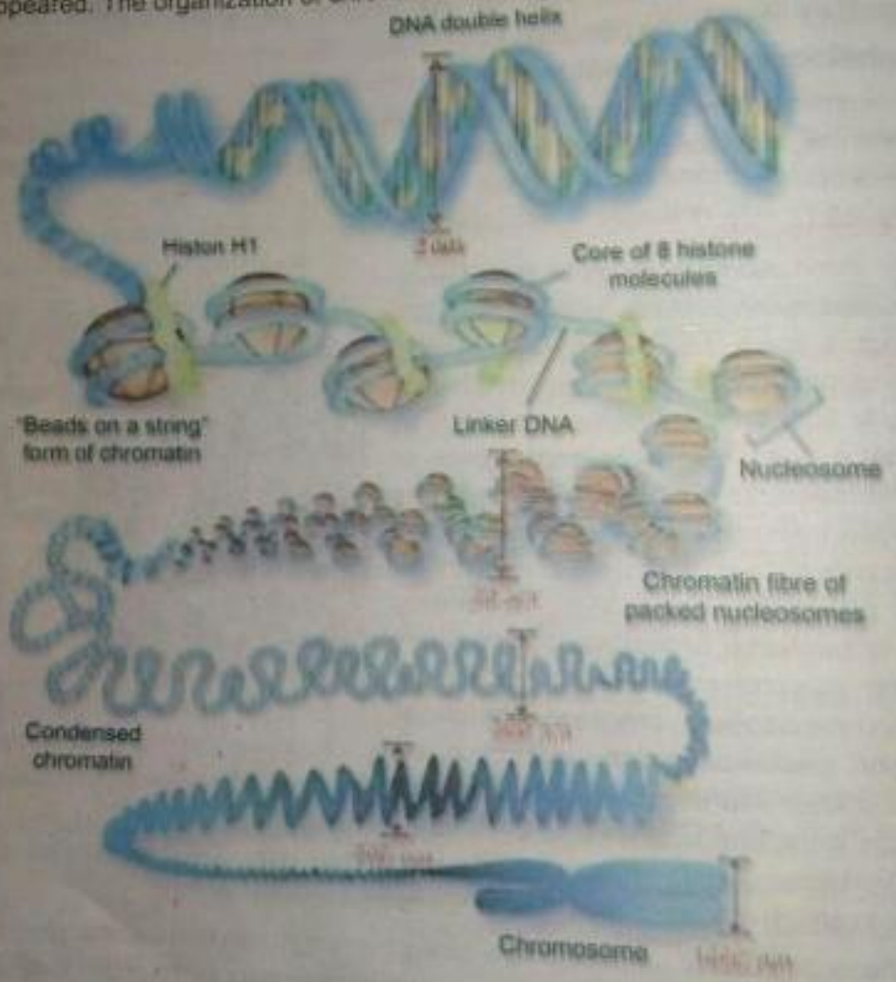


Fig. 23.2 Levels of organization of chromosomes

Teacher's Point

The teacher would ask the students to make a table to show how the structure of chromosomes in eukaryotes is different from the structure of chromosomes in prokaryotes?

Just after the completion of DNA replication, each DNA molecule of enormous length begins to coil around a histones core. Approximately every 200 nucleotides of the duplex DNA wrap twice around the core of eight histones, thus forming a complex known as **nucleosome**. One histone molecule is associated with a small segment of DNA (linker DNA) between every two nucleosomes. In this way the whole DNA (2nm thick) is turned to a chain of beads like appearance called **nucleosome string** (10 nm thick).

Many species have two sets of chromosomes in their somatic cells, hence called **diploid**, while some may have more than two even numbers of sets of chromosomes; they are called **polyploids (tetraploid, hexaploid)**. The term **"haploid"** is referred to the number of chromosome exactly half than the somatic number of chromosome. Gametes and spores are usually haploid cells. A haploid cell may be **monoploid** (one set), **diploid** (two sets), **triploid** (three sets), and etc.

Chromosomes in human and wheat

Name of Species	Somatic Number	Haploid	Monoploid (n)
Haman	46 (2n)	23 (n)	23
Wheat	42 (6n)	21 (3n)	7

Immediately, the nucleosome string begins to coil again about its axis to form yet another thick fibre of 30 nm, called chromatin fibre. During G_1 and G_2 phases, chromosomes are found in this level of organization. The chromatin fibre shows two regions i.e., heterochromatin and euchromatin. **Heterochromatin** is highly condensed and unexpressed region while **euchromatin** is non-condensed and the genes of this region are also expressed. When cell undergoes division, euchromatin is also condensed so that a uniform chromatin fibre is established.

When cell division begins, the higher order coiling of chromatin fibre gives rise supercoil which has diameter of 300 nm. Immediately supercoil establish into the chromatid of 700nm as result of further coiling around itself.

The number of chromosome varies from species to species and usually it is a characteristic feature of many species. *Penicillium*, a fungus, has only one pair of chromosome, while some **ferns** have more than 500 pairs.

23.1.3 Concept of Gene

The heredity units what we call genes today, were proposed by Mendel as **"elementens"**. Later on **Wilhelm Johansson** introduced the term **"gene"** for the basic unit of heredity in 1909.

As far as its physical and chemical nature is concerned, nowadays it is believed that **a gene is** composed of nucleotide sequence of "a short segment of DNA which encodes the sequence of amino acid of a particular polypeptide."

These segments of DNA are found in chromosomes. The particular place of the

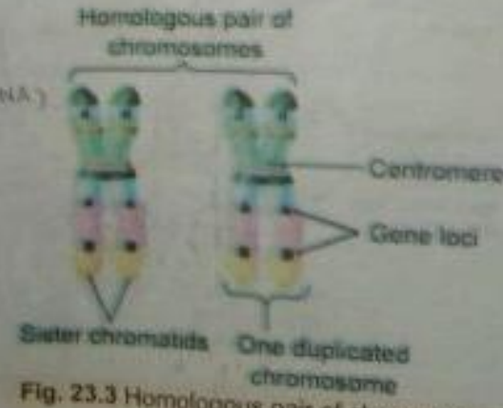


Fig. 23.3 Homologous pair of chromosomes

chromosome where a gene is located is called **locus** (plural: *loci*). The two chromosomes making up a matched pair are called **homologous chromosomes** because both carry genes controlling the same inherited characteristics. For example, if a gene for eye colour is located at a particular locus on one chromosome, then the homologous chromosome will also have a gene for eye colour (though perhaps for a different colour) at that locus. The alternative form of the gene on the same locus is called **allele.**

23.2 DNA AS THE HEREDITARY MATERIAL

When the chromosomal theory of inheritance had been confirmed in 1910, geneticists started thinking over the issue of the form, heredity units are found in the chromosome. It was known that chromosomes contain both DNA and protein. On which of these was the heredity information written? Over a period of 30 years, starting in the late 1920s, a series of investigators addressed this issue.

23.2.1 Griffith's Experiment

In 1928, British microbiologist Fredrick Griffith made a series of unexpected observations while experimenting with *Streptococcus pneumonae* which are found in two types. One of its types has a polysaccharide capsule, its colony appears as smooth or shiny, and hence it is called **S-type.** The other type forms a rough colony due to the absence of polysaccharide capsule; this is called **R-type.**

Fig. 23.4 Griffith's Experiment

When Griffith injected healthy mice with a strain of S-type, all the mice died but when he injected similar mice with a strain of R-type, the mice showed no ill effect. On the basis of these observations Griffith made a hypothesis that virulent effect of S-type might be associated with polysaccharide capsule as the R-type that lack a capsule, appeared non virulent.

As a control experiment, Griffith injected heat killed S-type into another group of mice to see if the polysaccharides capsule itself had a virulent effect. The mice remained perfectly healthy.

As a final control, he blended living R-type with heat killed S-type, both of these strains were already confirmed as non-virulent or benign. When he injected this mixture into the healthy mice unexpectedly, the injected mice developed disease symptoms and many

of them died. The blood of the dead mice was found to contain large number of living S-type virulent bacteria which had surface protein characteristics of the living (previously R) strain.

On the basis of these unexpected observations, Griffith concluded that somehow the information specifying the polysaccharide capsule and virulence had passed from the heat killed S-type bacteria to the living R-type once in the control mixture, transforming them into living S-type virulent bacteria that killed the mice. This transfer of genetic material from one organism to another, by which genetic make-up of recipient is altered, is called **transformation.**

Experiments of Avery, MacLeod and McCarty (Extra reading material)

The agent responsible for transforming R-type to S-type went undiscovered until 1944. In a classic series of experiments, Oswald Avery along with Colin Macleod and Maclyn McCarty characterized what they referred to as the "transforming principle". They first took extract filtrate of heat killed S-type bacteria which contained carbohydrates, lipids, protein, RNA and DNA. They divided this extract into four samples. First sample of this extract was mixed with live R-type cells and injected into the mice; they observed that transformation had occurred. It means that filtrate of heat killed S-type bacteria must contain the active factor, responsible for transformation. The second sample of the extract was treated with proteases (to remove all proteins), mixed with live R-type cells and injected into the mice; now they observed that transformation still had occurred. They concluded that protein is not responsible for transformation. The third sample of the extract was treated with RNAase (to remove all RNA contents), mixed with live R-type cells and injected into another group of the mice; since transformation still had occurred so RNA could not be the transforming agent. When fourth sample of the extract, treated with DNAase (to remove all DNA contents), mixed with live R-type cells and injected into another group of the mice; they observed that no transformation had occurred this time. Therefore, it has been confirmed that the active factor must be the DNA, which caused transformation of live R-type into live S-type in Griffith's experiment.

Avery's Experiment

Teacher's Point

The teacher would ask the students that using experiments of Griffith, Avery or Hershey and Chase as an example, develop a flow chart that shows how the scientists used scientific process.

23.2.2 Hershey and Chase Experiment

Soon after the Avery's results, another very convincing experiment on bacteriophages was performed by Alfred Hershey and Martha Chase in 1952.

Bacteriophages are the viruses that attack upon bacteria, their body consists of DNA and protein. During infection, they multiply in the host and their many copies are emerged within 20-25 minutes. It was not known till 1952 that either DNA or protein which possesses hereditary information of bacteriophages. Even, scientists were not sure that during infection, the whole viral particle enters the host body or only its DNA or protein get entry. In 1952, Hershey and Chase set out an experiment for this purpose.

They labelled the DNA of bacteriophages with a radioactive isotope of phosphorus, ^{32}P, and also labelled their protein coats with radioactive isotope of sulphur, ^{35}S. The labelled viruses are permitted to infect bacteria. Soon after the infection, bacterial cells were separated from media contents with the help of centrifugation technique. Then media contents and bacterial cells were analysed for the activity of ^{32}P and ^{35}S. In this analysis, ^{32}P was found in the bacterial cells while ^{35}S was found in the medium. These observations clearly showed that during infection, ^{32}P labelled DNA of bacteriophage was injected into the bacterial cell while its ^{35}S labelled protein coat remained outside. Subsequently, many viral particles released outside the host. Based on these observations, Hershey and Chase claimed that the virus DNA, not the virus protein, was responsible for directing the production of new viruses.

Fig. 23.5 Hershey and Chase Experiment

Teacher's Point

The teacher would ask the studerits to answer 'why did Hershey and Chase grow viruses in cultures that contained both radioactive phosphorous and radioactive sulphur? What might have happed if they had used only one radioactive substance?

Describe the paradoxical nature of DNA, as a tool of geneticists and forensics

Any type of organism can be identified by examination of DNA sequences unique to that species. To identify individuals, forensic scientists have scaned 13 DNA regions, or loci, that vary from person to person and use the data to create a DNA profile of that individual (sometimes called a DNA fingerprint). There is an extremely small chance that another person has the same DNA profile for a particular set of 13 regions. Some Examples of DNA uses for Forensic Identification: (1) Identify potential suspects whose DNA may match evidence left at crime scenes (2) Exonerate persons wrongly accused of crimes (3) Identify crime and catastrophe (Sudden disaster or misfortune) victims (4) Establish paternity and other family relationships etc.

Interphase: It is actually a period of diverse activities. The activities at interphase make the next mitosis possible. Interphase generally lasts at least 12 to 24 hours in mammalian tissue. During this period, the cell is constantly synthesizing RNA, producing protein and growing in size. Interphase can be divided into 4 steps:

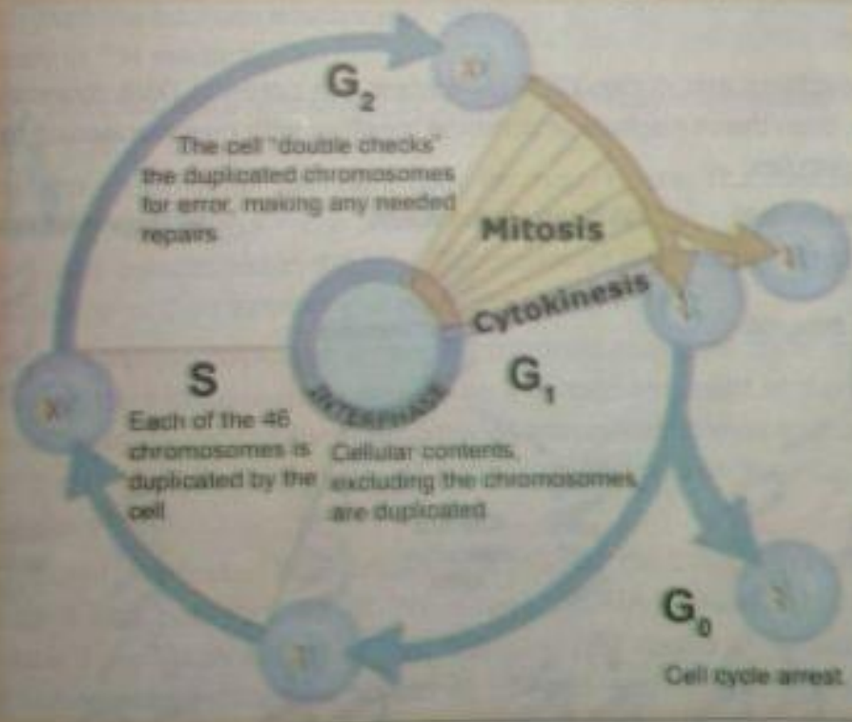
Gap 0 (G0): There are times when a cell will leave the cycle and quit dividing. This may be a temporary resting period or may be permanent (e.g. neuron).

Gap 1 (G1): Cells increase in size in Gap 1, produce RNA and synthesize protein.

S Phase: To produce two similar daughter cells, DNA replication occurs during this S (synthesis) phase.

Gap 2 (G2): The cell will continue to grow and produce new proteins.

Mitosis or M Phase: Cell growth and protein production stop at this stage in the cell cycle. The cell divides into two similar daughter cells. Mitosis is much shorter than interphase, lasting perhaps only one to two hours.



Teacher's Point

The teacher would ask the students to Interpret the experiment in which radio isotopes labeled DNA can be traced in the progeny of an organism.

23.3 DNA REPLICATION

The process of self-synthesis of DNA molecule is called DNA replication. This process occurs only once in S-phase during the life cycle of a cell. The molecule of DNA which is replicated is called parent DNA, while the molecules, produced in this process are called daughter DNA. A parent DNA molecule after replication gives rise two daughter DNA molecule.

23.3.1 Models of DNA Replication

How duplex DNA can replicate? Scientists tried to find the answer of this after the discovery of DNA structure. Over all three different models to explain the replication process were presented i.e., semi-conservative, conservative and dispersive model.

Semi-conservative model

According to this model, the two parental DNA strands separate and each of these strands then serves as a template for the synthesis of a new DNA strand. The result is two DNA double helices, both of which consist of one parental and one new strand.

Conservative model

This model states that after replication has occurred one daughter molecule contains both parental DNA strands, and the other daughter molecule contains DNA strands of all newly-synthesized material.

Dispersive model

This model explains that during DNA replication, the parental DNA molecule is dispersed into its nucleotide, then these nucleotide combine together with new nucleotide to form two new daughter DNA molecules.

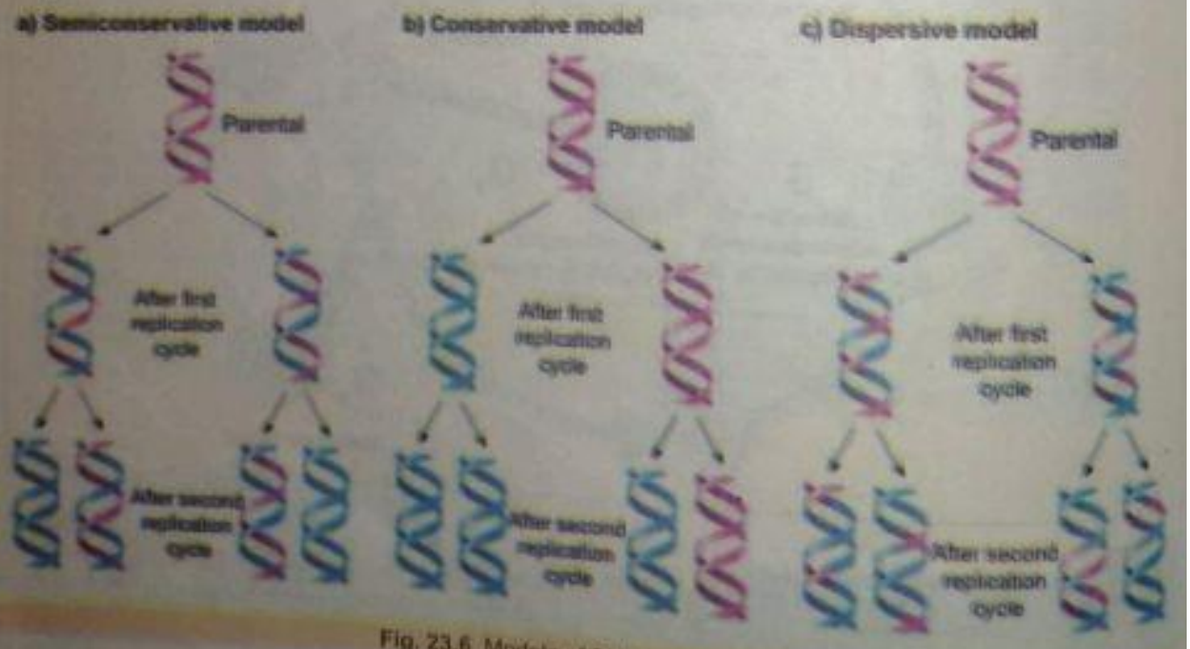


Fig. 23.6 Models of DNA replication

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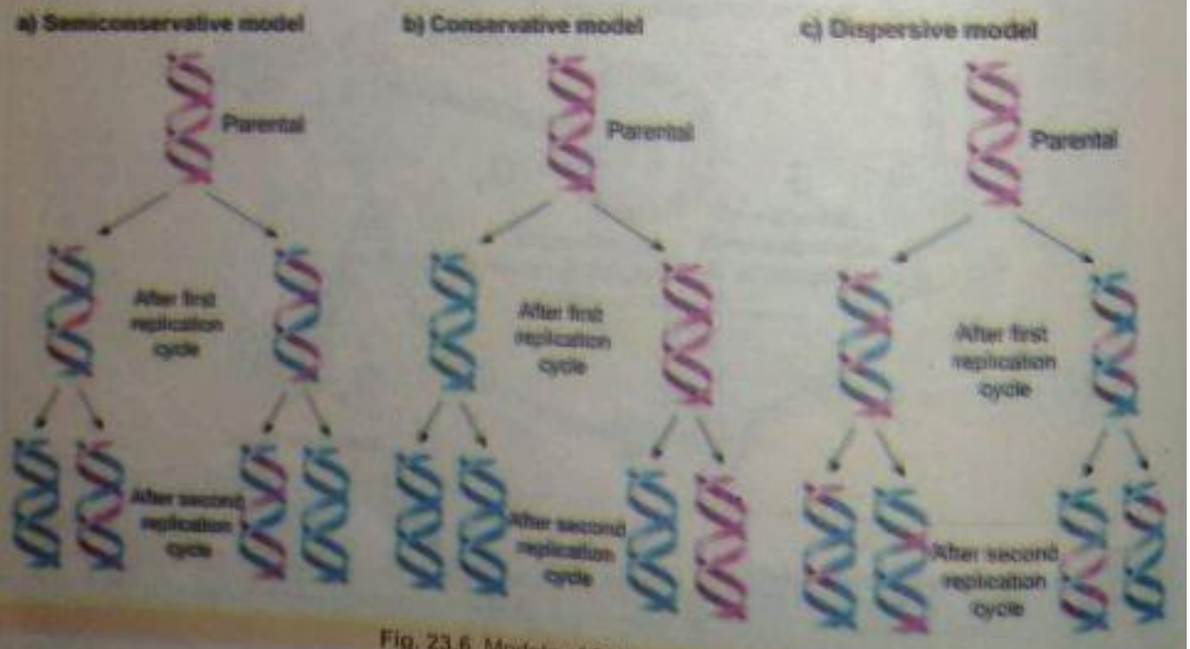

Fig. 23.6 Models of DNA replication

23.3.2 Meselson and Stahl Experiment

The three models of DNA replication were evaluated by Mathew Meselson and Franklin Stahl of the California Institute of technology in 1958. Later on, they were awarded Nobel Prize. In this experiment, it was concluded finally that the replication of DNA occurs according to semi-conservative model.

They grew bacteria in a medium containing heavy isotope of nitrogen, ^{15}N, which became incorporated into the bases of bacterial DNA. After several generations, the bacteria were shifted to three separate plates, which were already poured with medium containing ^{14}N.

Three DNA samples were taken from bacteria shifted from ^{15}N medium to the ^{14}N medium. First sample was obtained from first plate just after the transfer of culture; called sample at 0 minute, the second sample was taken from second plate after 20 minutes, called sample at 20 minutes, and third sample was taken from third plate after another 20 minute, called sample at 40 minute. In addition to these, a control sample was also taken from the bacteria which were grown separately in ^{14}N medium.

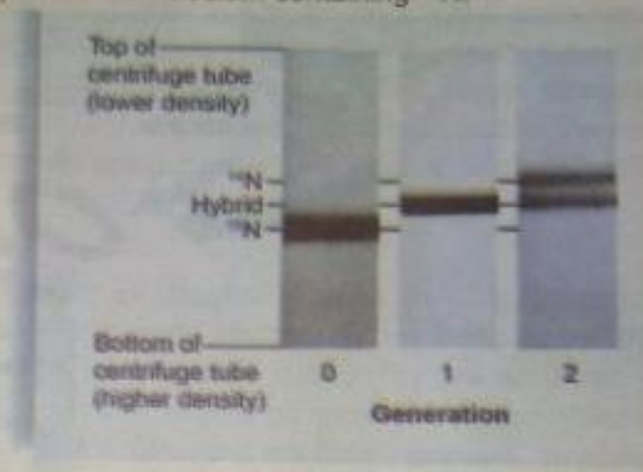

Fig. 23.7 Results of Meselson-Stahl Experiment

The DNA samples were dissolved in cesium chloride (CsCl) solution and then spun at a very high speed in an ultra-centrifuge for many hours. The cesium and chloride ions tend to be pushed by centrifugal force towards the bottom of the tube. Ultimately a gradient of Cs^+ and Cl^- ions was established in the tube. Molecules of DNA were settled down and formed sediments to the level of their appropriate densities in test tubes.

DNA of control sample appeared lightest as it formed sediment at the top of test tube, while DNA of sample at 0 minute appeared heaviest as it formed sediment at the bottom of test tube. The DNA of sample at 20 minute formed sediment intermediate level to that of control sample and sample at 0 minute whereas sample at 40 minute had two sediments, one at the top and other at intermediate level.

Meselson-Stahl interpreted their results as it follows: the DNA of control sample appeared lightest because it had both strands of ^{14}N, whereas DNA of sample at 0 minute appeared heaviest because it had both strand of ^{15}N, but after first round of replication each daughter duplex was a hybrid possessing one strand of ^{14}N and one of ^{15}N, so it formed sediment at intermediate level. When this hybrid duplex replicated in second round of replication, it contributed ^{15}N strand to form another hybrid duplex and ^{14}N strand to form a light duplex containing both ^{14}N strands that is why this sample formed two sediments. On the basis of above mentioned results, they claimed that the DNA replication is semi conservative.

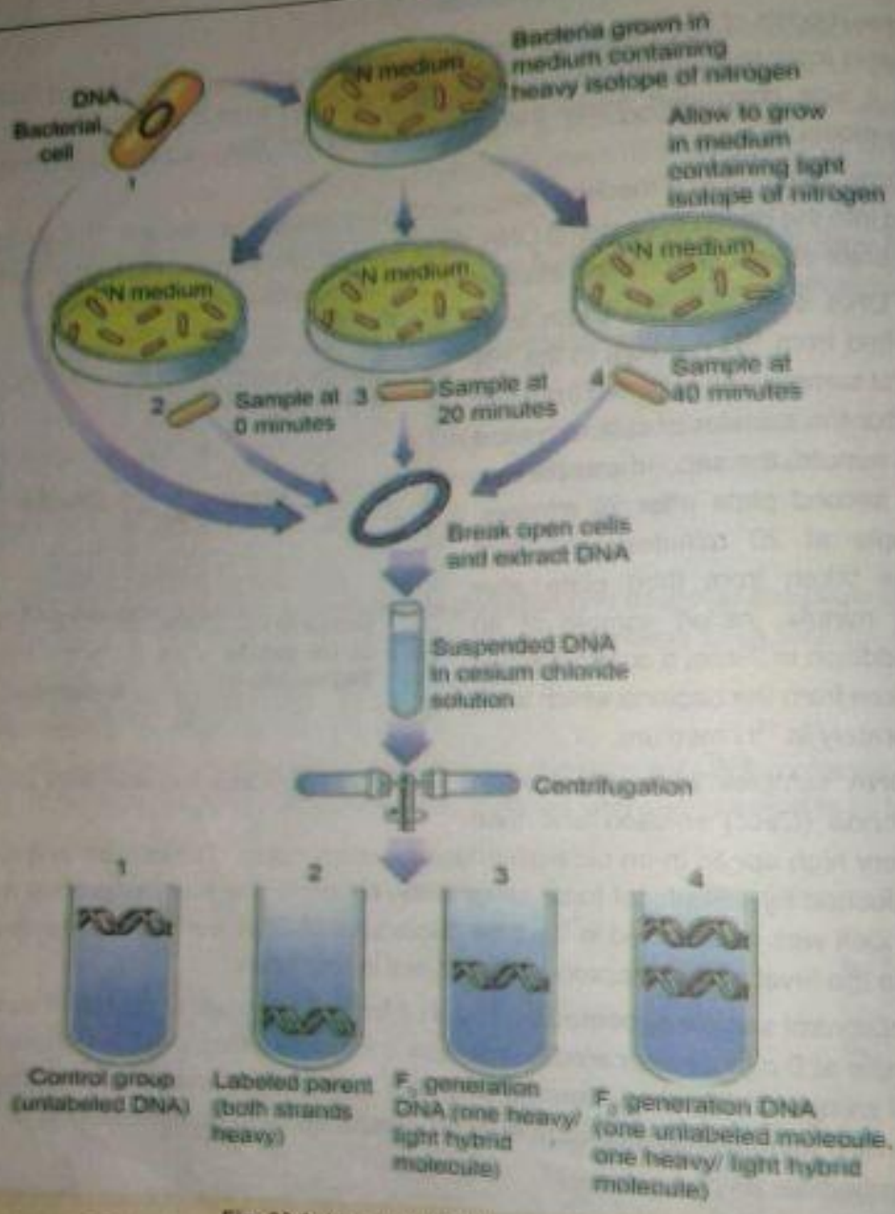

Fig. 23.8 Meselson-Stahl experiment

23.3.3 Mechanism of DNA Replication

Although DNA replication is a continuous process but here we are going to discuss it in different phases for the sake of convenience.

Initiation phase

The initiation phase is characterized by the formation of **replication bubble** and **replication fork**, which are formed at a particular site, called **origin of replication** site. It is a

specific sequence of nucleotides along the length of DNA from where process of replication begins. In eukaryotic DNA, there may be more than one origin of replication sites but in prokaryotic DNA there is only one origin of replication.

Replication bubble is formed when **DNA helicase** and **DNA gyrase (topoisomerase)** enzymes work at origin of replication. DNA helicase opens the turns of DNA duplex by causing breakdown of the base pairs of DNA so that two strands of DNA can be separated. DNA gyrase works slightly ahead the DNA helicase and facilitate in unwinding of the DNA duplex by reducing the tension created during unwinding process. Due to the action of these two enzymes, the two strands of DNA duplex gradually separate from each other and give a bubble like appearance at origin of replication, called **replication bubble**. After the breakdown of base pairs, the single strands of DNA are prevented to pair up again by specific proteins called **single stranded binding (SSB) proteins**. Both single strands of DNA will act as template strand in the next phase and direct the synthesis of daughter strands along themselves. Each side of replication bubble is now termed as **replication fork**.

Extension / Polymerization phase

Extension or polymerization is the formation of daughter strands (leading or lagging strands) along the template strands. The daughter strands are actually synthesized by **DNA polymerase** enzyme. This enzyme cannot work unless some nucleotides are arranged on template. For this purpose, **primase** enzyme is involved to arrange some RNA nucleotides on template strands. Such short fragments of few RNA nucleotides are called **primers**, which act as start site for the activity of DNA polymerase. The primase and DNA helicase enzymes are found in the form of a complex, called **primosome**.

After the establishment of primers, synthesis of daughter strands begins by the DNA polymerase enzyme. There are three different forms of this enzyme:

a) **DNA polymerase-I:** It catalyses the replacement of RNA primers by DNA nucleotides in termination phase of replication so it provides a support to the DNA polymerase-III in the main replication process.

b) **DNA polymerase-II:** It is involved in the repairing process of DNA damages during the life time of a cell.

c) **DNA polymerase-III:** It is the main enzyme that synthesizes both daughter strands along the template during replication process.

Mechanism of DNA polymerase-III activity

DNA polymerase-III cannot initiates replication process. It can add a nucleotide onto only a preexisting 3'-OH group. Therefore, it needs a primer to perform its polymerase activity. It always adds nucleotide at 3' end of primer so the direction of replication becomes 5' to 3' end.

Teacher's Point

The teacher would ask the students to make a Venn diagram that compares the process of DNA replication in prokaryotes and eukaryotes.

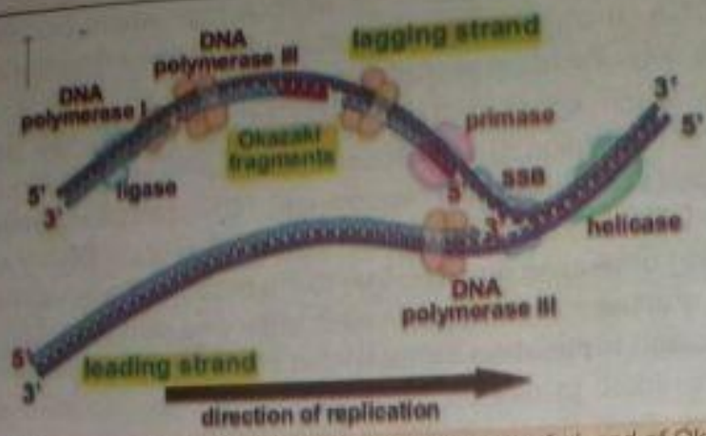

Fig. 23.9 Process of DNA replication: Showing replication fork and of Okazaki fragments

One unit of DNA polymerase-III works on one template and continuously synthesize a daughter strand towards replication fork. This continuously growing daughter strand is called **leading strand**. The second unit work on other template. It synthesizes another daughter strand away from replication fork. As the two units are interlinked so the second unit is allowed to polymerize daughter strand up to a specific length, then it has to jump back (100 to 200 nucleotides in eukaryotes and 1000 to 2000 nucleotides in prokaryotes) to a new primer to perform polymerization again. Therefore, this daughter strand grows discontinuously away from the replication fork by forming short fragments interrupted by primers, called **Okazaki fragments** (named after Japanese scientist who discovered them). This discontinuously growing strand is called **lagging strand**.

One of DNA polymerase-III subunit also possesses ability to remove wrong nucleotide if it is added mistakenly. This ability is called **proofreading**.

Termination phase

Termination phase is characterized by the replacement of primers by DNA nucleotides and joining of Okazaki fragments in lagging strand to form a continuous strand.

The replacement of primers by DNA nucleotides is carried out by **DNA polymerase-I** that has dual function i.e. beside **polymerase** it also acts as **exonuclease**. It is attached to the 3' end of Okazaki fragment where it adds DNA nucleotide so that it can extend while on the other hand it cleaves nucleotide from 5' end of primer. In this way primers are removed and each **Okazaki fragment** is extended up to the next Okazaki fragment but they do not join together.

The joining of Okazaki fragments is carried out by **DNA ligase** enzyme that finally constructs phosphodiester bonds between Okazaki fragments so a continuous strand is formed.

DNA is the genetic material which can replicate. The genetic material is in the form of specific sequences of nucleotides along the DNA strands. The DNA inherited by an organism

Teacher's Point

The teacher would ask the students to interpret how DNA conserves one strand during replication.

Science, Technology and Society Connections

Describe how various scientists in the field of biotechnology or genetic engineering have used DNA replication

The Polymerase Chain Reaction (PCR) is the most revolutionary technique used in biotechnology which is based upon in vitro DNA replication. This is an extremely sensitive means of amplifying small quantities of DNA. This is very important in the detection of low level bacterial infections or rapid changes in transcription at the single cell level, as well as the detection of a specific individual's DNA in forensic science. It can also be used in DNA sequencing, screening for genetic disorders, site specific mutation of DNA, or cloning or sub cloning of DNAs.

forms specific traits by directing synthesis of a protein that acts as catalyst and catalyses a specific chemical reaction of the cell. Thus a gene expresses itself in a protein or enzyme that controls the development of a specific character or function. We can say that proteins are link between the genotype and phenotype. Gene expression or protein synthesis includes transcription and translation.

23.4 GENE EXPRESSION

The idea that DNA makes protein via an intermediate RNA is known as the **central dogma of molecular genetics**. Information can only flow from DNA to protein and not from proteins to DNA. In other words, changes in DNA may change the resulting protein, but changes in proteins cannot feedback and change the DNA.

23.4.1 Transcription

Transcription is the synthesis of RNA from DNA. It is the first step of gene expression. It occurs in G_0, G_1 and G_2 phases of cell cycle. Unlike DNA replication, it requires only one enzyme to be completed i.e., RNA polymerase. However, it is a continuous process; for convenience we can divide it into three phases: initiation, elongation and termination.

Initiation phase

Transcription begins with the binding of RNA polymerase at **promoter region**, a regulatory region of the gene that comprises binding sites for the attachment of RNA polymerase. In prokaryotes, these binding sites are TATAAT also called -10 sequence and TTGACA also called -35 sequence, whereas in eukaryotes, TATA (TATA box) also called -25 sequence and CAAT (CAAT box) also called -70 sequence. Names of these sequences (-10, -35 or -25, -70) refer to their position in promoter region that these sequences are located approximately how many nucleotides before the transcription start point of the gene.

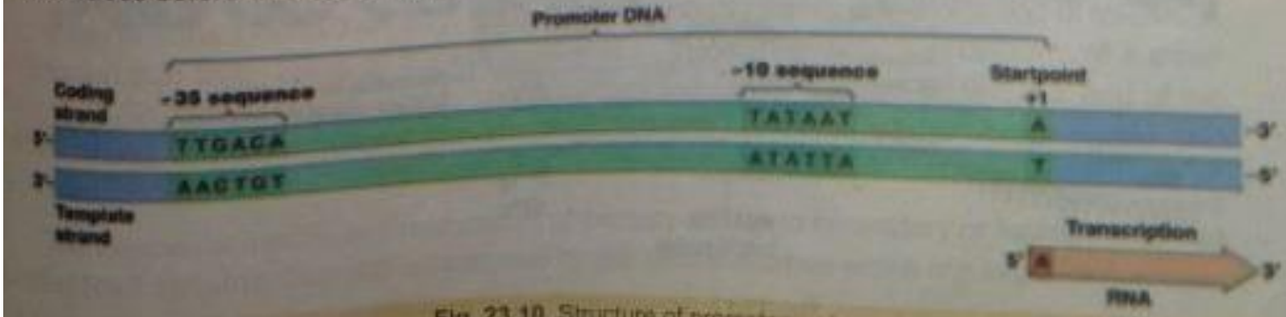

Fig. 23.10 Structure of promoter region

RNA polymerase consists of four subunits: alpha (∝), beta (β), beta and sigma (σ), only the first three subunits are required for polymerase activity and are considered the **core enzyme** while the sigma factor is required for RNA polymerase to bind on the promoter. The core enzyme and sigma factor together constitute a **holoenzyme**. Once RNA polymerase is attached on promoter, the sigma factor is removed, and the core enzyme catalyses the remaining process. It is similar to the DNA polymerase in that it also adds nucleotides to the 3′ end of the growing polypeptide chain but unlike DNA polymerase it does not require primer to perform polymerase activity. In prokaryotes, only one type of RNA polymerase is found while in eukaryotes, there three types of RNA polymerases, namely RNA polymerase-I, which synthesize rRNA; RNA polymerase-II, which synthesize mRNA; RNA polymerase-III which synthesize tRNA.

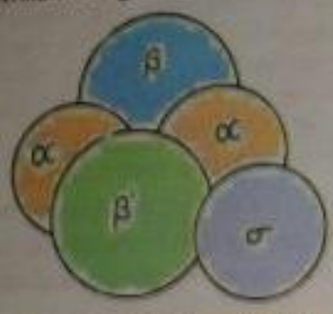

Fig. 23.11 Structure of RNA polymerase

As the RNA polymerase binds to the promoter, DNA duplex become unwind, base pairs are broken down, and a bubble like structure, the **transcription bubble** appears.

Elongation phase

As the RNA polymerase binds to promoter, it begins to arrange and polymerise the ribonucleosides triphosphates (rNTP) or ribonucleotides complementary to the template strand of the DNA. It does not require primer to initiate polymerization. One of the two strand of the gene acts as **template** for transcription. This template strand is also called **antisense** because mRNA is complementary to this strand. The other strand of the gene is called **coding** or **sense strand**. In elongation phase, RNA polymerase keeps on moving from 5′ to 3′ end of RNA towards the terminator region of the gene, beside it transcription bubble also moves along the DNA, leaving the growing RNA strand protruding from the bubble. This event continues till the RNA polymerase reaches the terminator region of the gene.

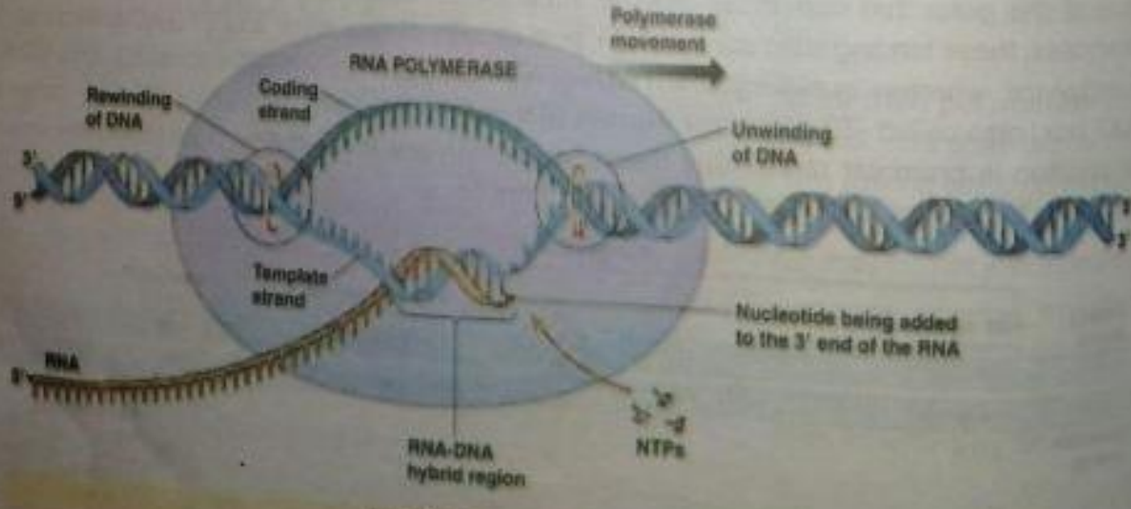

Fig. 23.12 Elongation phase of transcription

Termination phase

The sequence of terminator region of the gene stops the synthesis of mRNA. The terminator region consists of a series of GC base pairs followed by a series of AT base pairs. The part of mRNA which is transcribed in this region, projects to form a loop like structure called **GC hairpin** followed by a small tail of poly U nucleotides. The GC hairpin causes the RNA polymerase to stop the synthesis of RNA.

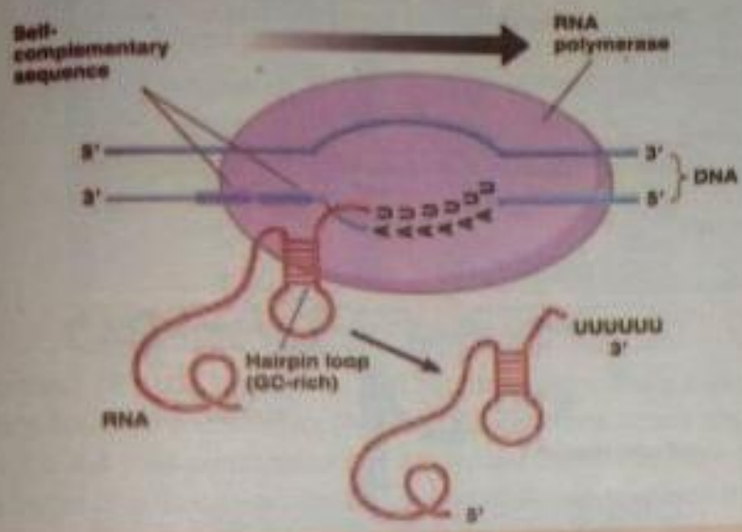

Fig. 23.13 Termination of transcription

23.4.2 Post-Transcriptional Modification of mRNA

In prokaryotes, there is no delay between transcription and translation as the mRNA emerges from DNA it undergoes translation due to the absence of a definite nucleus. On the other hand in eukaryotic cells the pre-mRNA (newly synthesized mRNA) has to be modified into mature or functional mRNA because transcription occurs in nucleus while translation in cytoplasm so mRNA has to move from nucleus to cytoplasm and during this journey, enzymes like phosphatases and nucleases may degrade it. Furthermore, eukaryotic mRNA also contains some non-coding region called **introns** which are removed or spliced during this process. Post transcriptional modification therefore involves two events, addition of a cap and tail to protect it from degradation and RNA splicing to remove non- coding sequences.

A **cap** is in the form of **7-methyl GTP**, which is linked from its 5′ to the 5′ end of mRNA. A modification also takes place at the opposite end of the RNA transcript in the form of a small chain of 30-500 adenine nucleotides, called **poly-A tail**, which is attached to the 3′ end of the mRNA. These two modifications prevent the mRNA to be degraded by phosphatases and nucleases.

The removal of introns and maturation of primary mRNA to secondary or functional mRNA is called **RNA splicing**. Splicing is catalysed by the **spliceosomes** which is a large RNA-protein

complex. Later on the spliced **exon fragments** are joined together with the help of RNA ligase enzyme.

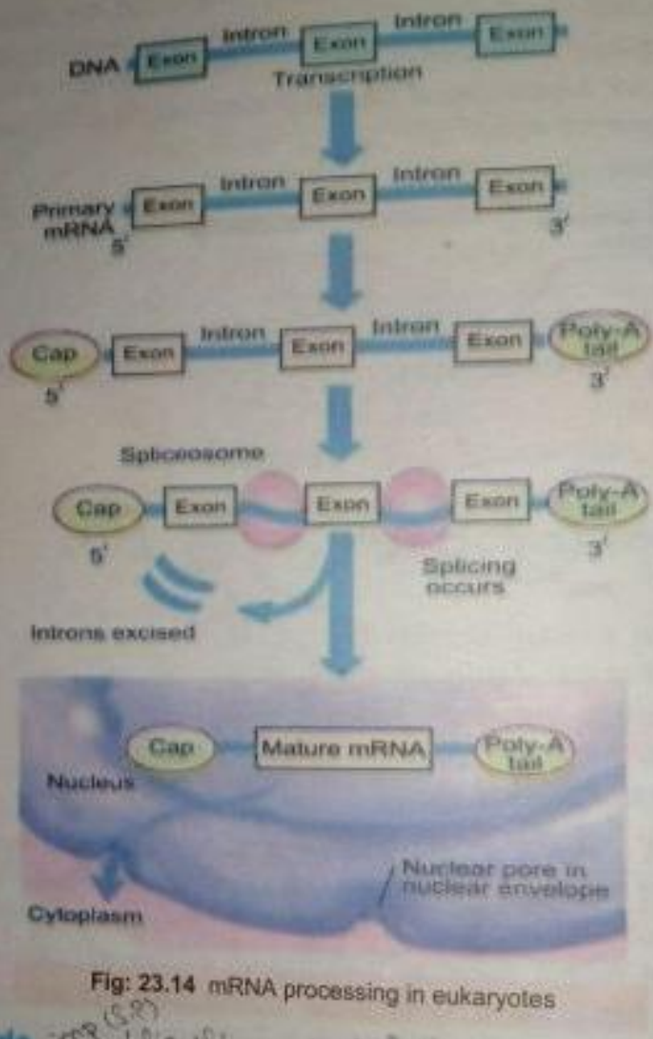

Fig: 23.14 mRNA processing in eukaryotes

23.4.3 Genetic Code

It has been discussed earlier in this chapter that a **gene** is a specific segment of DNA that encodes the synthesis of a particular polypeptide (protein). Therefore, the order of amino acids in a polypeptide is according to the sequence of nucleotides in any part of a DNA. The relationship between amino acids sequence and nucleotide sequence is called **genetic code**. In other words the information in DNA for the synthesis of protein is called as **genetic code** which is transcribed / copied into mRNA.

The mRNA transcript consists of a random sequence of only four kind of nitrogenous bases (A, G, C and U). Now the question is that "how can a code with four letter alphabet specify a protein which at any given point contains one of twenty different amino acids"? Clearly a single base cannot specify a single amino acid, if one base specifies a single amino acid then only four different amino acids could be encoded for, and proteins containing only four different amino

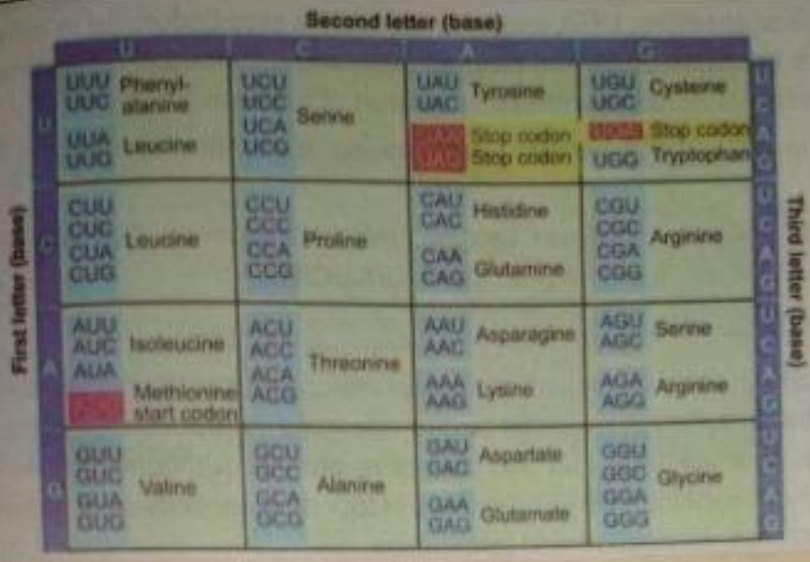

Fig.23.15 Types of codons

acids would be formed. Nor is it feasible for just two bases to specify a single amino acid since only 16 amino acids could be encoded for (4 X 4 = 16). But three bases are sufficient. With three bases a total of 4 X 4 X 4 = 64 combinations are possible; each is specific for a particular amino acid. Therefore a triplet of bases along the length of mRNA that specifies a particular amino acid is called as **codon**.

There are total 64 codons. Three of these codons act as **stop codons** (UGA, UAG, and UAA), one of these must be present at the end of mRNA that indicates that the message is over. Since, these three codons do not encode any amino acid, hence called **non sense codon**, while all the other sequences that encode specific amino acids are called **sense codons**. One of these sense codons (AUG = methionine) also acts as **start codon** which must be present at the beginning of all the sequences that code for amino acid chains.

Genetic code has following characteristics:

1) Some amino acids are only encoded by a single codon; while others are encoded by up to four codons and amino acids leucine and serine are encoded by six codons. This property is known as **code degeneracy**.
2) The genetic code is **universal**. It is the same in almost all the organisms. For example AGA specifies arginine in bacteria, in humans and all other organisms whose genetic code has been studied. Because of the universality of codon the genes can be transferred from one organism to another and be successfully transcribed and translated in their new host. The study of genetic code of mitochondrial DNA however, showed that genetic code is not that

Teacher's Point

The teacher would ask the students that 'using the genetic code, identify the amino acids that have the following messenger RNA strand strand codes: UGGCAGUGC

universal. For example: UGA codon is normally a stop codon but in mitochondria it reads as tryptophan. Likewise, AUA was read as methionine instead of isoleucine and AGA and AGG for termination of protein synthesis is instead of arginine.

3) There is no punctuation between each codon. It means that there is no gap between two codons.

4) The genetic code is non-over lapping. mRNA sequence AUGAGCGCA is not read as AUG/UGA/GAG etc. it will be read as AUG/AGC/GCA.

23.4.4 Translation

Translation is second step of gene expression. In translation, messenger RNA (mRNA) produced by transcription is decoded by the ribosome to produce a specific amino acid chain or polypeptide, that will later fold into an active protein. Although translation is a continuous process but for convenience we will discuss it in four phases: activation of amino acids, formation of initiation complex, elongation and termination.

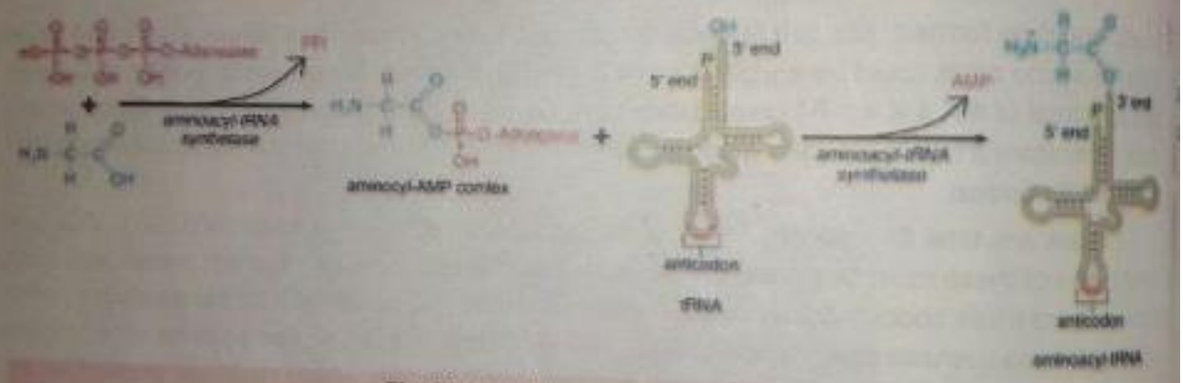

Fig. 23.16 Activation of amino acids

Activation of amino acids

Activation of amino acids refers to the binding of free amino acids dispersed in cytoplasm to the 3′ end of particular tRNA molecules, in this way a complex is formed called **aminocyl-tRNA** complex. This binding is catalyzed by aminocyl-tRNA synthase (activation enzyme). Various amino acids that are to take part in polypeptide formation have been continuously activated throughout the process of translation.

Formation of initiation complex

Process of translation actually begins with the formation of **initiation complex.** It is formed by the combination of ribosomal subunits, mRNA and first aminocyl-tRNA complex. First a tRNA molecule carrying a chemically modified methionine (called N-formyl methionine) binds to the smaller ribosomal subunit. This binding is controlled by an enzyme called **initiation factor.** At the same time 5′ end of mRNA molecule also binds to the smaller sub unit of ribosome with the help of another initiation factor. Initiation complex is completed when larger subunit of ribosome is also placed upon smaller subunit.

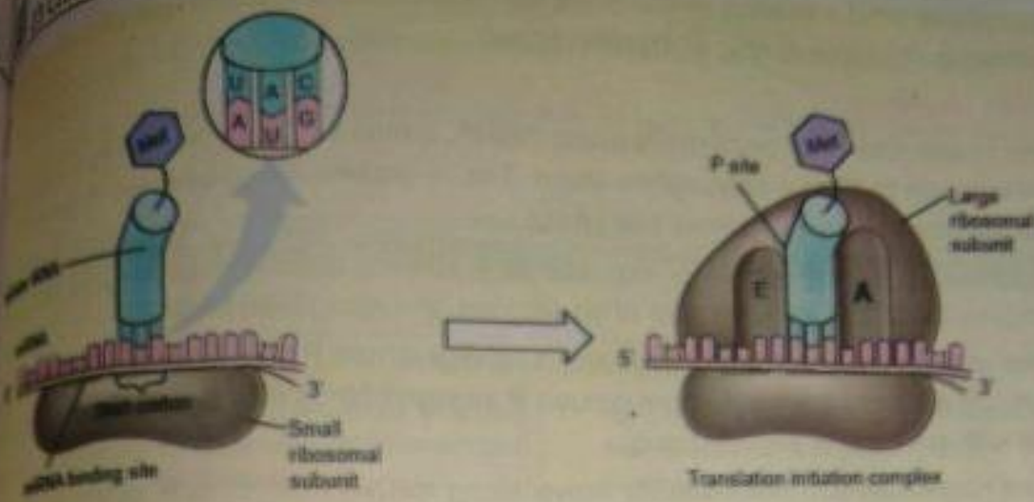

Fig. 23.17 Formation of initiation complex

The region of smaller ribosomal subunit where first aminocyl-tRNA complex is attached is called **P site** (peptidyl site), here peptide bonds will be formed between successive amino acids during elongation phase. Nearby two other sites are also established. **A site** (aminocyl site)

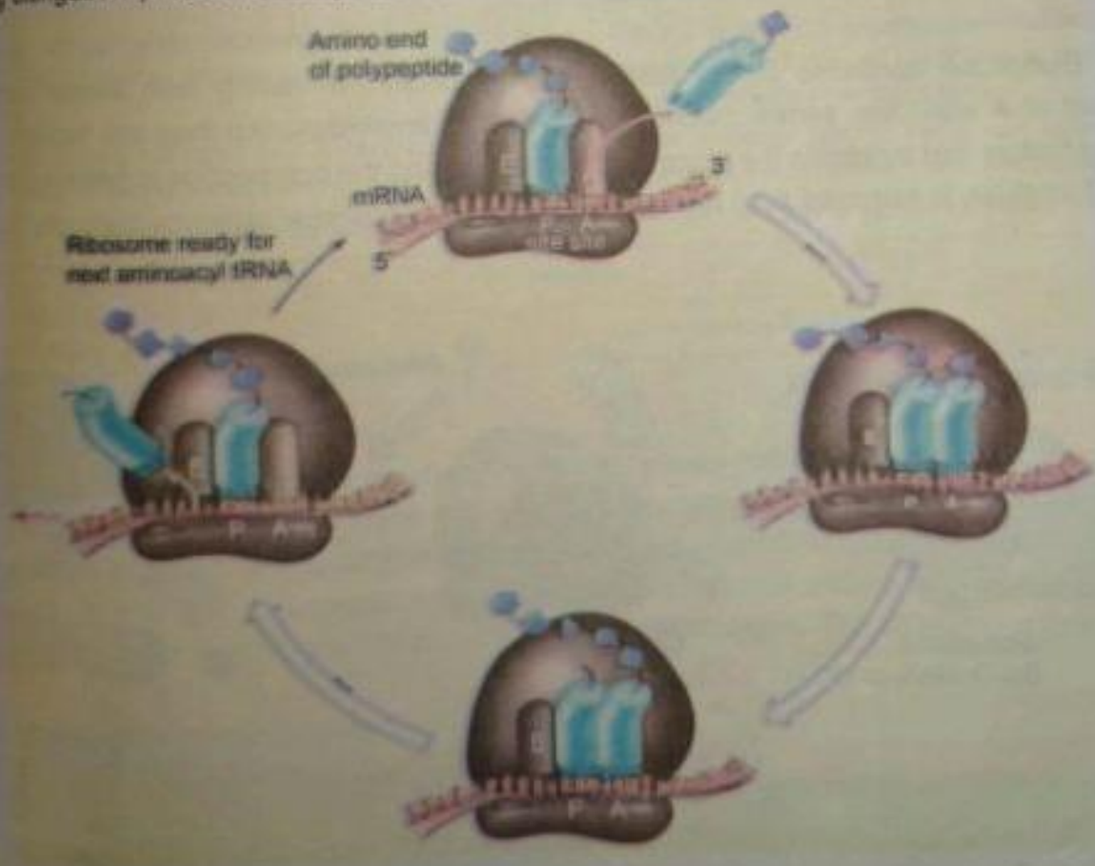

Fig. 23.18 Chain elongation in translation process

where successive tRNAs bearing amino acids will be attached and **E site** (exit site) where empty tRNAs will leave ribosome during elongation phase.

Polypeptide elongation

In this phase ribosomal units move along mRNA, amino acids are brought by tRNAs, which are joined together to form a polypeptide chain. This is accomplished by three steps which are repeated again and again throughout this phase.

a) Whichever codon of mRNA is exposed at A site, its anticodon bearing aminocyl-tRNA complex binds to it with the help of an enzyme, the elongation factor.

b) Then an enzyme **peptidyl transferase** is emerged from P site. It removes the amino acid (may be a chain) from tRNA present on P site and binds it to the newly coming amino acid with the help of peptide bond.

c) Then ribosomal sub units slightly move along mRNA from 5′ to 3′ direction so that a new codon is exposed at A site. This movement is called **translocation**. As a result, the empty tRNA is reached at E site to leave the ribosome, while the other tRNA bearing a chain of amino acid is shifted from A site to P site, and another codon is exposed to A site.

These three steps are repeated again and again until the stop codon is reached at A site.

Termination

Elongation continues in this fashion until a chain-terminating **non sense codon** is exposed at **A site**. Non sense codons do not bind to any tRNA, but they are recognized by release factors that terminate the process of translation and the polypeptide is released from the tRNA. The tRNA is released from the ribosome and the ribosomal subunits separate from the mRNA.

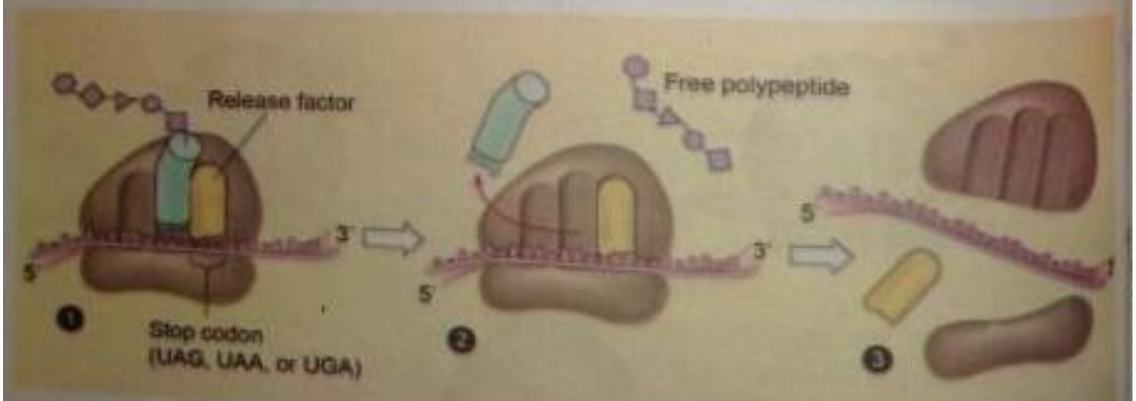

Fig. 23.19 Termination of translation

How many types of tRNA molecules are necessary for living cell, if the genetic code is a triplet code?

If the genetic code is triplet code then approximately 64 types of tRNA molecules are necessary for living cell.

Table 23.2 Difference between protein synthesis (translation) in prokaryotes and eukaryotes

PROKARYOTES	EUKARYOTES
Structure of ribosomes	
Ribosomes are smaller in size consist of a 50S large subunit and a 30S small subunit, which bind together to form 70S ribosome.	1. Ribosomes are larger in size consist of a 60S large subunit and a 40S small subunit, which bind together to form 80S ribosome.
Site of synthesis	
Ribosome occur in cytosol.	2. Ribosome occur in cytosol and in rough endoplasmic reticulum.
Initiation phase	
The initiating amino acid is modified i.e. N-formyl methionine.	3. The initiating amino acid is methionine. It is not modified.
A specific purine-rich sequence on the 5' side is required to distinguish initiator AUG from internal ones.	4. There is no need of a specific purine-rich sequence instead the AUG nearest the 5' end of m RNA is usually selected as the start site.
mRNA is poly cistronic i.e., it can serve as a template for the synthesis of several proteins.	5. mRNA is mono cistronic i.e., it can serve as a template for the synthesis of only one protein.
Only one type of initiation factor is used.	6. More than one type of initiation factors are used.
Termination	
Two release factors are used.	7. Single release factors is used.

23.5 REGULATING GENE EXPRESSION

The overall process by which genetic information flows from genes to proteins that is from genotype to phenotype is called **gene expression**. Regulation of gene expression is the control of the **amount** and **timing** of appearance of a gene. All the cells of a living body must continually turn genes ON and OFF in response to signals from their external and internal environment. The mature human body is composed of about 200 different cell types. The differences between cell

Teacher's Point

The teacher would ask the students to:

(i) Make a Venn diagram that compares the process of protein synthesis in prokaryotes and eukaryotes.

(ii) Make a list of all proteins that have been studied or referred to till now.

called as **exon** and which as introns. Regulatory proteins which are specific to a cell type control intron-exon choices by binding to regulatory sequences within the primary transcript.

23.6 MUTATIONS

A gene mutation is a permanent change in the DNA sequence that makes up a new allele in the population. Mutations range in size from change in a single DNA nucleotide to a large segment of a chromosome or whole chromosome or sometimes changes in the number of chromosome. The agents that cause mutations are called **mutagens** while the organism or cell in which mutation occurs is called **mutant**.

23.6.1 Sources and Types of Mutation

There are many sources of mutation which are basis deferent type of mutation.

Sources of mutagens

The **sources of mutation** may be physical, chemical and biological agents.

Physical mutagens: These include radiation of any kind i.e., UV, gamma rays, X-rays, radioactive rays, high temperature alterations.

Chemical mutagens: These include colchicine, mustard gas, nitrous gas, acridine orange, reactive radioactive isotopes, and free oxygen particles etc.

Biological mutagens: These may include certain viruses, transposons and errors that occur during meiosis or DNA replication.

Types of mutation

Creation or origin of mutation is called **mutagenesis**. Based upon mutagenesis, there are two types of mutations. (a) The mutations which occur naturally and automatically due to internal or external factors are called **spontaneous mutations**. (b) The mutations which are produced by external factors for the establishment of new varieties of organism are called **induced mutations**.

(a) Mutations are of two types on the basis of where the mutations occur and to what extent. A mutation that causes change of single or few nucleotides in the DNA is called **point mutation**.

(b) The mutations that cause change in the structure or number of chromosome are called **chromosomal mutations** or **aberrations**.

23.6.2 Importance of Mutation

A mutation may be harmful or useful.

Harmful aspects of mutation

A mutation that decreases the fitness of the organism in the environment is called **harmful mutation**. A mutation is sometimes a form of adaptation. Sometimes mutations could possibly be harmful to cause serious effect. There are many such examples like (a) born without a part of the brain (b) cancer is a form of a harmful mutation (c) the developmental abnormalities, such as microcephaly, cleft palate, cases of abnormal number of chromosome (Down syndrome,

Klinefelter's syndrome, Turner's syndrome etc.) and heredity disorders like sickle cell anaemia, phenylketonuria etc.

Useful aspects of mutation

Mutations are also considered as contributing factors towards evolution. Changes in the genes controlling development can have major effects on the morphology of the adult organism. Because these effects are so significant, scientists suspect that changes in developmental genes have helped bring about large-scale evolutionary transformations. Developmental changes may help to explain, for example, how some hoofed mammals evolved into ocean-dwellers, how water plants invaded the land, and how small, armoured invertebrates evolved wings. Mutations are considered as the driving force of evolution, where less favourable (or deleterious) mutations are removed from the gene pool by natural selection, while more favourable (beneficial or advantageous) ones tend to accumulate.

23.6.3 Chromosomal Mutation

Changes in structure or number of chromosomes are called **chromosomal mutations** or **chromosomal aberrations**. Here we will discuss symptoms, causes and possible treatments of some of the chromosomal mutations e.g., Down syndrome, Klinefelter's syndrome and Turners syndrome.

Down syndrome

Down syndrome is characterized by 2n+1 as these persons have an extra copy of 21st chromosome (trisomy 21). **Symptoms** include abnormal shaped smaller sized head, rounded inner corner of the eyes instead of pointed, excess skin at the nape (back of the neck) of the neck; flattened nose; single crease in the palm of the hand; small ears; small mouth; wide, short hands with short fingers; white spots on the coloured part of the eye, excessive space between large toe and second toe etc. The affected people may be male or female. It is **caused** by autosomal non-disjunction, in which 21st chromosomal pair fails to segregate properly during maternal meiosis and result into the formation of an egg having 24 chromosomes. The fertilization of such egg by a normal human sperm containing 23 chromosomes produces a child having 47 chromosomes (2n+1) with three copies of 21st chromosomes therefore it is also called as **trisomy 21**

Fig. 23.23 Down syndrome karyotype

Teacher's Point

The teacher would ask the students to write a paragraph comparing and contrasting gene mutations. *Hint:* To organize your ideas, use a compare contrast table. The column heads might be: Definition, Types and Effect.

as it is chromosomal abnormal. There are no **treatments** for Down syndrome as it is chromosomal abnormality. But it can be managed to some extent by taking few measures like: (a) Regular checkups and screening, (b) Medications, (c) Surgery (d) Counseling and support.

Klinefelter syndrome

It is characterised by 2n+1 (44+XXY). **Symptoms** include several feminine characters (although they are males) like sparse body hair, enlarged breasts, and wide hips. In almost all men the testicles remain small. Their voices may not be as deep. Abnormal body proportions (long legs, short trunk, and shoulder equal to hip size). It is **caused** by non-disjunction of sex chromosome pair during maternal meiosis.

They have an extra X chromosome (44+XXY). These patients show trisomy in sex chromosomes. For **treatment** they can be given testosterone for sexual development, like males, around the age of puberty, it can help a boy have more normal body development. The treatment usually continues throughout a man's life but does not help infertility.

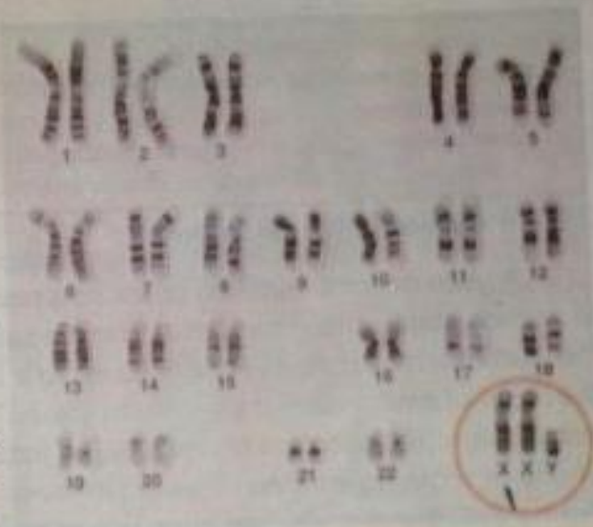

Fig. 23.24 Klinefelter syndrome karyotype

Turner syndrome

It is characterized by 2n-1 (44+XO).The affected individuals are females. **Symptoms** include shorter than average height, infertility, webbed neck, a low hairline at the back of the neck, abnormal bone development (especially the bones of the hands and elbows); a larger than usual number of moles on the skin; **edema** or extra fluid in the hands and feet. It is **caused** by non-disjunction of sex chromosome pair during maternal meiosis.

The normal females have two X chromosomes, but in Turner syndrome one of the X chromosomes is absent i.e., (44+XO = 45). There is no specific **treatment** as it is chromosomal abnormality. However, growth hormone treatment can improve growth and influence a girl's final adult height, especially if treatment is started early enough in childhood.

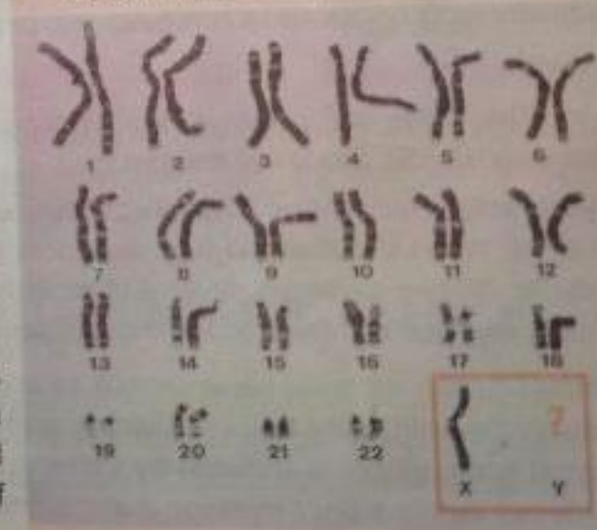

Fig. 23.25 Turner syndrome karyotype

Estrogen replacement treatment helps the girls to develop the physical changes of puberty, including breast development and menstrual periods. This treatment is often started when a girl reaches about age 12 or 13.

Science, Technology and Society Connections

Explain how harmful mutations have been eradicated by nature

A harmful mutation is a mutation that decreases the fitness of the organism. The process of eradication of harmful mutation is done by natural selection. The process of DNA repair is an important way in which the body protects itself from disease. Every harmful mutation is eliminated from the population by premature death or reduced reproductive success, a 'genetic death'.

Skills: Interpreting and Planning

Make a list of some commonly occurring minor mutations in humans.

- Some commonly occurring minor mutations in humans are sickle cell anemia, phenyl ketonuria, thallassaemia, colour blindness, haemophilia etc.
  Pakistan is inherited because of between cousins marriages.

23.6.4 Gene/Point Mutations

A gene or point mutation arises as a result of a chemical change in an individual gene. An alternation in the sequence of nucleotides in the part of a molecule that corresponds to a particular gene changes the order of amino acids making up a protein. These may include base substitution (replacement of one base by another), insertion (addition of one or more bases), and deletion (removal of one or more bases). Frame shift mutations occur when one or more nucleotides are either inserted or deleted from DNA. This results in the completely new sequence of codons and a non-functional protein.

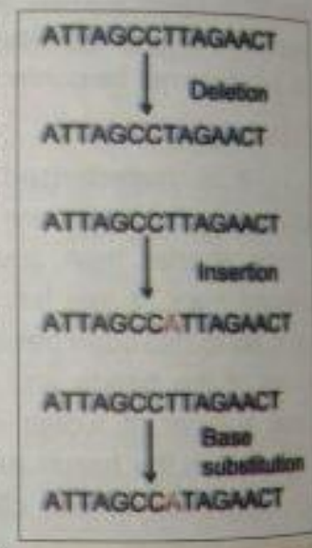

Fig. 23.26 Types of Point Mutation

Sickle cell anaemia

The sickle cell anaemia is also known as Haemoglobin SS disease (Hb SS); Sickle cell disease.

Symptoms: These include painful episodes, which can last from hours to days. When the anaemia becomes more severe, symptoms may include: fatigue, paleness, fever and rapid heart rate, shortness of breath, jaundice and damage to various organs.

Cause: Sickle cell anaemia is caused by a defect in haemoglobin. Actually the sixth amino acid i.e., glutamic acid in the beta chain of the normal haemoglobin is replaced by valine in the haemoglobin of a sickle cell due to a point mutation (base substitution) in which single **thymine** is replaced with adenine at the position that codes for

Critical Thinking

How sickle cell disease and phenyl ketonuria are inherited as the autosomal recessive trait?

Teacher's Point

The teacher would ask the students to justify why mutations prevail in a population and are inherited. *Hint:* marriages between cousins.

AC } Sickle cell anaemia
SS }

glutamic acid. The abnormal sickle cell haemoglobin is called Hb^S which have lost the oxygen carrying capacity. Due to this haemoglobin the shape of the red blood cells is changed. The red blood cells become shaped like crescents or sickles. These cells deliver less oxygen to the body's tissues. Sickle cell anaemia is due to homozygous recessive gene. One can get the disease if inherited from both parents.

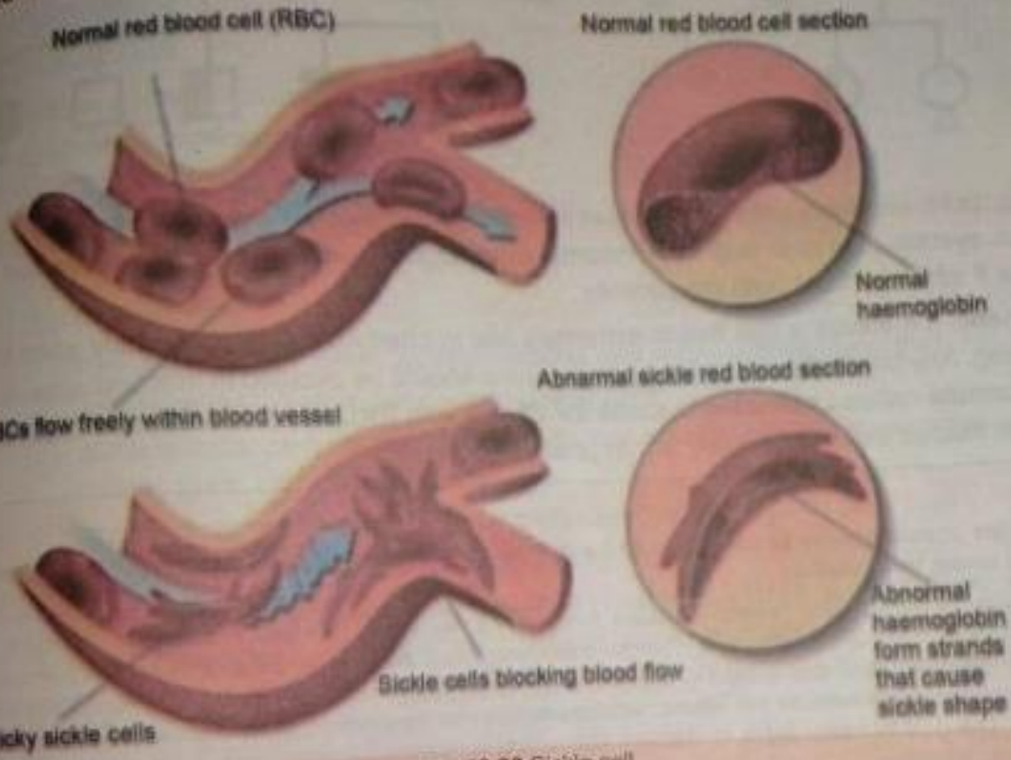

Fig. 23.26 Sickle cell

Treatment: Treatment for a sickle cell anaemia includes blood transfusions, medicines to relieve pain and plenty of fluids. Bone marrow or stem cell transplants can cure sickle cell anaemia. Folic acid supplements should be taken.

Phenylketonuria (PKU)

A rare condition in which a baby is born without the ability to properly breakdown an amino acid called **phenylalanine** is called **phenyl ketonuria**.

Symptoms: These include lighter skin, hair, and eyes than brothers or sisters without the disease, delayed mental and social skills, head size significantly below normal, hyperactivity, jerking movements of the arms or legs, mental retardation, seizures, skin rashes and tremors etc.

Cause: In a healthy baby, **phenylalanine** is converted into **tyrosine** by the enzyme **phenylalanine hyrdoxylase**. Due to a **point mutation**, this enzyme becomes defective and converts phenylalanine into toxic phenylketones that accumulate and damage the central

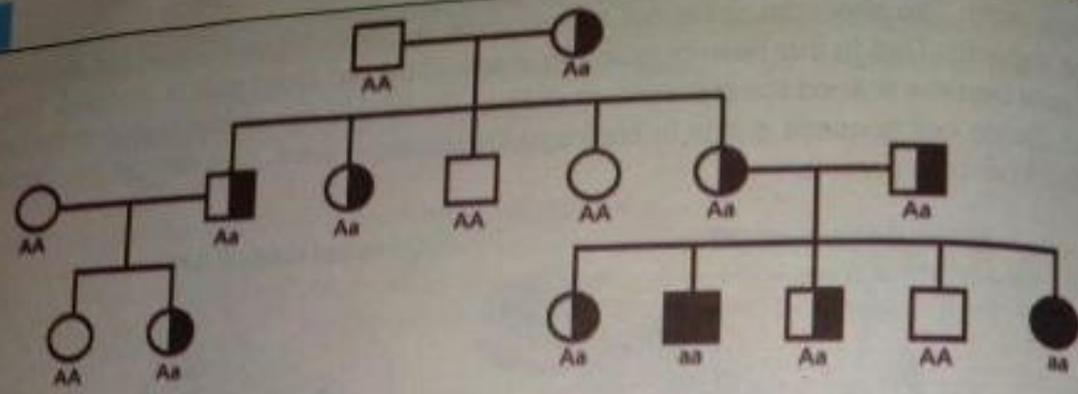

Fig. 23.27: Inheritance pattern of Sickle cell anaemia and phenylketonuria as autosomal recessive trait

nervous system. It is also due to autosomal homologous recessive gene and one can get the disease if inherited from both the parents.

Treatment: It involves a diet that is extremely low in phenylalanine, particularly when the child is growing. Any products containing phenylalanine should be avoided in milk and eggs. A special infant formula called **Lofenalac** is made for infants with PKU. It can be used throughout life as a protein source that is extremely low in phenylalanine.

Science, Technology and Society Connections

Suggests possible ways to save lives or treat genetic diseases (like diabetes) through the knowledge gained under this heading

Lives can be saved from genetic diseases by avoiding marriages between the carriers therefore genetic counseling, screening before birth and proper treatment during pregnancy are suggested. Furthermore, bone marrow transplantation and gene therapy are some other options for the treatment of sickle cell anaemia, thalassemia, phenylketonuria etc. Blood transfusions can also be made in case of thalassemia and sickle cell anemia. The goal of treatment is to manage and control symptoms because there is no permanent treatment for genetic diseases.

Activity

Make a model of DNA replication.

Exercise

M.C.Qs.

Select the correct answer

1. (i) Chromosomes are made of:
   (A) DNA + Pectin
   (B) RNA + DNA
   (C) DNA + Histone
   (D) DNA only

   (ii) In his work with pneumonia-causing bacteria and mice, Griffith found that
   (A) heat-killed pathogenic cell caused pneumonia
   (B) some substance from pathogenic cells was transferred to non pathogenic cells, making them pathogenic.
   (C) the polysaccharide coat of bacteria caused pneumonia.
   (D) bacteriophage injected DNA into bacteria.

   (iii) A biochemist isolates and purifies molecules needed for DNA replication. When DNA is added, replication occurs, but each DNA molecule consists of a normal strand paired with numerous segments of DNA a few hundred nucleotides long. What has probably been left out of the mixture?
   (A) DNA polymerase
   (B) DNA ligase
   (C) Okazaki fragments
   (D) primase

   (iv) What is the basis for the difference in how the leading and lagging strands of DNA molecules are synthesized?
   (A) the origins of replication occur only at the 5′ end.
   (B) DNA ligase works only in the 3′ - - 5′ direction.
   (C) polymerase can only work on one strand at a time.
   (D) DNA polymerase can join new nucleotides only to the 3′ end of a growing strand.

   (v) Synthesis of a new DNA strand usually begins with
   (A) an RNA primer
   (B) DNA ligase
   (C) a DNA primer

(vi) A sub metacentric chromosome has centromere
(A) in the centre of chromosome
(B) near an end of chromosome
(C) slightly away from the centre of a chromosome
(D) at the end of a chromosome

(vii) The elongation of the leading strand during DNA synthesis
(A) progresses away from the replication fork
(B) occurs in the 3′/-5′/ direction
(C) produces Okazaki fragments
(D) depends on the action of DNA polymerase

(viii) Whose pioneering work on the gene used the bacteriophage to show that DNA was the part of this virus which went into the bacterial cell for replication?
(A) Watson and Crick
(B) Meselson and Stahl
(C) Hershey and Chase
(D) Beadle and Tatum

(ix) If you want to label amino acids but not DNA, which of the following radioactive isotopes would you use?
(A) ^{18}F (B) ^{35}S (C) ^{14}C (D) ^{32}P

(x) Semi conservative replication refers to the fact that
(A) genetic information contained in the transforming molecule
(B) only the DNA of the bacteriophage enters the bacterial cell
(C) certain base pair with specific bases
(D) one DNA strand remains while a new one is made

(xi) In eukaryotic cells, transcription cannot begin until
(A) the two DNA strands have completely separated and exposed the promoter
(B) several transcription factors have bound to the promoter
(C) the DNA introns are removed from the template
(D) DNA nucleases have isolated the transcription unit

(xii) The anticodon of a particular tRNA molecule is
(A) complementary to the corresponding triplet in mRNA
(B) complementary to the corresponding triplet in rRNA
(C) the part of the tRNA bonds to specific amino acid
(D) catalytic, making the tRNA

Short Questions

1. Who rediscovered Mendel's work?
2. Define chromosomal theory of inheritance.
3. What is the contribution of Walther Flemming and Waldyer in the " origin of chromosomal theory of inheritance" .
4. What is the contribution of Carl Correns in the " origin of chromosomal theory of inheritance"
5. What is the contribution of T.H.Morgan in the " origin of chromosomal theory of inheritance"
6. On the basis of position of centromere describe the four types of chromosomes.
7. What is chromatin fibre? What is the difference between the two regions of chromatin fibre?
8. Write the functions of polymerase I,II and III.
9. Describe central dogma of gene expression.
10. Describe the four characteristics of genetic code.
11. Explain why the length of transcribed m-RNA (in Eukaryotes) shortens as it enters the cytoplasm for translation.
12. Interpret how many types of t-RNA molecules are necessary for a living cell, if the genetic code is triplet code.
13. Make a list of some commonly occurring minor mutations in human.
14. Suggest possible ways in which the synthesized protein can be used within or outside a cell that synthesized it.
15. Define/Describe/Explain:

chromosome, chromatin network, chromatids, centromere, nuclear organizer, junk DNA, telomere, metacentric chromosome, submetacentric chromosome, acrocentric chromosome, telocentric chromosome, histone proteins, nucleosome, homologous chromosomes, gene, allele, semi conservative nature of DNA replication, gene expression, Okazaki fragments, leading strand, lagging strand, proof reading, polymerase, DNA ligase, primers, primosome, replication fork, transcription, translation, template, sense strand, holoenzyme, introns, genetic code, translocation, mutation, mutant, mutagens, spontaneous mutation, chromosomal mutation, sickle cell anemia, phenylketonuria.

17. Write the differences between:
    (a) metacentric and submetacentric chromosome
    (b) acrocentric and telocentric chromosome

(c) nucleosonme and primosome
(d) heterochromatin and euchromatin
(e) gene and allele
(f) conservative and dispersive model of DNA replication
(g) DNA helicase and DNA gyrase
(h) DNA polymerase I and II
(i) DNA polymerase II and III
(j) leading strand and lagging strand of replication fork
(k) translation and transcription in gene expression
(l) intron and exon
(m) start codon and stop codon
(n) non sense codon and sense codon
(o) point mutation and chromosomal mutation
(p) harmful and useful aspects of mutation
(q) Down syndrome and Klinefelter syndrome
(r) Klinefelter syndrome and Turners syndrome

Extensive Questions

18. What is the contribution of Walter Sutton and Theodor Boveri in the "origin of chromosome theory of inheritance"?
19. Compare the parallel behaviour of genes and chromosomes during meiosis.
20. Describe the structure of chromosome.
21. Describe in detail the composition and organization of chromosome.
22. Describe the experiment of Griffith to prove that DNA is the hereditary material.
23. Describe the experiment of Hershey – Chase to prove that DNA is the hereditary material.
24. Describe the three model proposed about the mechanism of DNA replication.
25. Describe the work of Meselson and Stahl to justify the semi-conservative replication as the correct method of replication.
26. Describe the events of the process of DNA replication and explain DNA stability and variability as two characters of the replicating DNA molecule.
27. Describe the mechanism of polymerase III activity.
28. Explain the mechanism of transcription.

Describe the mechanism of protein synthesis.

Write the difference between protein synthesis in prokaryotes and eukaryotes.

What is the importance of the regulation of gene expression?

Describe the negative control of gene expression by repressor proteins and the positive control of gene expression by activator proteins.

Explain post-transcription modification of mRNA.

(a) Define mutation and identify various sources of mutations

(b) Differentiate between natural and induced mutations.

(a) Explain the harmful and useful aspects of mutations.

(b) Rationalize that mutations might be a contributing factor towards evolution.

Describe the symptoms, causes and possible treatment of the following chromosomal mutations:

(a) Down syndrome

(b) Klinefelter's syndrome

(c) Turner's syndrome

Describe the symptoms, causes and possible treatment of the following genetic mutations:

(a) Sickle cell anaemia

(b) Phenylketonuria

22

INHERITANCE

After completing this lesson, you will be able to:

- Associate inheritance with the laws of Mendel.
- Explain the law of independent assortment, using a suitable example.
- Express limitations in the law and its usefulness.
- State the scope of independent assortment in variation.
- Evaluate that inheritance of genes and their mixing during fertilization is based on mathematical probabilities.
- Describe the exceptions to the Mendel's laws of inheritance.
- Explain incomplete dominance and exemplify it through the inheritance of flower color in 4 0' clock plant.
- Differentiate between incomplete dominance and co-dominance.
- Describe multiple alleles and state the alleles responsible for the trait of ABO blood groups.
- Explain the case where two alleles have equal dominance and through the genetics of human blood group of AB.
- Name the various human blood group systems.
- Associate multiple alleles with the ABO blood group system.
- Investigate the reasons for O-ve individual as the Universal donor and AB +ve as the Universal recipient.
- Describe the occurrence of some other blood group systems.
- Associate the positive and negative blood groups with the presence and absence of Rh factor.
- Justify why Rh incompatibility could be a danger to the developing foetus and mother.
- Explain *Erythroblastosis foetalis* in the light of antigen-antibody reaction.
- Suggest measures to counter the problem of *Erythroblastosis foetalis* before it occurs.
- Explain the terms; polygenic and epistasis.
- Describe polygenic inheritance, using suitable examples from plants (grain color in wheat) and animals (skin color in man).
- List at least five polygenic traits discovered in humans.
- Relate polygenic inheritance with epistasis.
- Give one example of epistasis from mammals (coat color inheritance in *Labrador retrievers*) and one from plants (pigment phenotype in sweet pea) and justify modified Mendelian ratios.
- Describe the terms gene linkage and crossing over.

- Explain how gene linkage counters independent assortment and crossing-over modifies the progeny.
- Exemplify the concept of gene linkage by quoting the example of wing length and width of abdomen in *Drosophila melanogaster*.
- Suggest why linkage could be observed / evaluated only if the number of progeny is quite large.
- Explain the XX-XY mechanism of sex determination in *Drosophila* and mammals.
- Describe the XX-XO and ZZ-ZW sex determination systems and evaluate by studying the karyotype.
- Identify the difference between homogametic and heterogametic conditions in the karyotype of male and female humans.
- Identify male and female individuals from the karyotype of Drosophila and man.
- Solve the genetics problems related to XX-XY, XX-XO and ZZ-ZW sex determination.
- Differentiate between autosomes and sex chromosomes from the karyotype.
- Describe the concept of sex-linkage.
- Explain the inheritance of sex-linked traits (eye color) in *Drosophila*.
- Describe the sex-linked inheritance of male characters due to Y -chromosome and the effect of Hollandric genes.
- Describe sex-influenced and sex-limited traits with common examples from human genetics.
- Describe the X- linked disorders with reference to the patterns of inheritance.
- Name some of the sex-linked disorders of man and drosophila.
- Critically analyze the inheritance of Haemophilia, colour blindness and muscular dystrophy.

Reading

The similar characteristics that pass from parents to their offspring are collectively called **heredity**. The resemblance, however is not complete, offspring differ from each other and their parents in many respects. These differences are known as **variations**. Both similarity and differences are the parts of inheritance which play a significant role in the formation of new species. The science, which deals with mechanism of the heredity and variation, is called **genetics**. Since genes control the heredity and variations, so the term genetics is also referred to the **study of genes**.

22.1 MENDELIAN INHERITANCE

The science of genetics originated in the year 1900 with the rediscovery of an article originally published in 1866 by an Augustinian monk named Gregor Johann Mendel. Mendel was the first who successfully explained the mechanism of inheritance during his research work on pea plant.

22.1.1 Association of Inheritance with Laws of Mendel

In the 1860's, Mendel discovered mechanism of inheritance based on his experimental work with pea plants. The **first step** of his experimental work was to develop true or purebred varieties or lines. A plant that produces all the offspring of its own phenotype upon self-fertilization is called **true** or **pure breeding plant**. For example, if a round seed shaped plant is self-fertilized, and all of its offspring are also round seed shaped, the parent as well as offspring both will be true or pure breeding.

Gregor Mendel

A true or pure breeding plant can be developed by repeated self-fertilization through successive generations until the true breed is achieved. Mendel studied seven pairs of contrasting traits also called **Mendelian parameters**, for each contrasting phenotype, he developed a true breeding plant.

In second step, he performed **hybridization**, a cross fertilization between two individuals having contrasting phenotypes. For example, a cross fertilization of round seed shaped plant with wrinkled seed shaped plant (**monohybrid cross**) or a cross fertilization of round shape and yellow coloured seed plant with wrinkled shape and green coloured seed plant (**dihybrid cross**) are kinds of hybridization. The study of inheritance of single trait and then the combination of two or three traits one by one, was the secret of success of Mendel. Based upon observations of these experiments, Mendel proposed two generalizations, which are commonly known as **Mendel's laws of inheritance**.

INHERITANCE OF SINGLE TRAIT (MONOHYBRID CROSS)

Mendel studied inheritance of single trait in monohybrid cross in which two plants are crossed that differs on single trait.

Procedure and observations

For the study of inheritance of single trait (such as seed shape), he crossed a pure breeding round seed shaped plant with a pure breeding wrinkled seed shaped plants. In such hybridization experiments, he observed that "F_1" (**first filial generation**), was comprised entirely of individuals exhibiting only one of the parental phenotype (round seed shape). However, when this generation was interbred (self-fertilized), its offspring, the "F_2" (**second filial generation**), showed a **3:1** ratio i.e., three individuals had the same trait as F_1 inherited (round seed shape) and one individual had the phenotype that failed to inherit in F_1 generation (wrinkled seed shape).

Mendel got similar results and the same 3:1 ratio in offspring of monohybrid crosses for all the seven contrasting pair of traits.

Mendel proceeded a step ahead. He self-fertilized F_2 plants to get the F_3 generation. He observed that 1/3 of F_2 round produced only round (appeared true or pure breeding like P_1 round), while 2/3 of F_2 round produced both round and wrinkled in 3:1 (appeared non-pure breeding like F_1 round); but F_2 wrinkled produced only wrinkled (pure breeding).

Interpretations of the results

Based upon these observations, Mendel concluded that each contrasting form (phenotype) of a trait, e.g., roundness or wrinkledness of seed was determined by **particulate hereditary factors**, which he called **"elementens"** (now called genes). These factors carrying hereditary information were transmitted from parents to offspring through gametes. Each pea plant had a pair of these factors (now called **alleles**, the alternative form of gene on the same locus), one derived from the male parent and other from the female parent. The pair of these factors (now called **genotype**) controlled the expression of the trait (now called **phenotype**). He designated the term **"dominant"** to the factor that was expressed in F_1 generation while the factor that was failed to express, termed as **"recessive"**. These factors were represented by alphabetical symbols e.g., the dominant factor was represented by capital case letter (**"R"** for round seed shape) and the recessive factor by small case letter (**"r"** for wrinkled seed shape)

The true breeding round seed plant of P_1 generation carried "RR" alleles while the true breeding wrinkled seed plant of P_1 generation carried "rr" alleles. When both the alleles of a gene pair are same, the organism is said to be **homozygous** for that gene pair. An individual with homozygous genotype is called **homozygote**.

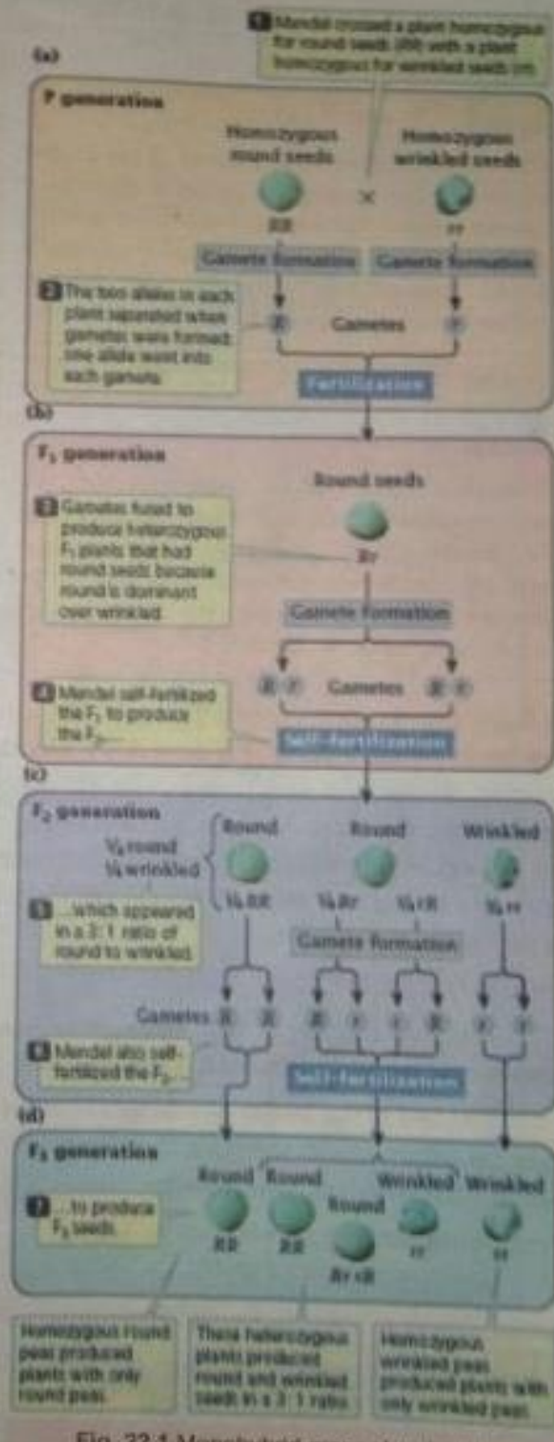

Fig. 22.1 Monohybrid cross showing the inheritance of single trait

Mendel inferred that the factor of a pair (alleles) separated from each other during gamete formation so that each gamete got only one factor for each trait. Therefore, half of the gametes got one allele and the other half carried the other allele. Fertilization was random. When male gamete carrying factor "R" fertilized female gamete with factor "r", the complete set of two factors "Rr" for the trait was restored in zygote. Zygote developed into F_1 offspring that was **heterozygous** "Rr" because the two alleles of its gene pair were different from each other. An individual with heterozygous genotype is called **heterozygote**. F_1 offspring "Rr" was a **monohybrid** for seed shape; it was round in phenotype but heterozygous in genotype. Its alleles also segregated during gamete formation.

Science Titbits

R.C. Punnet devised what is known as the Punnett square for summarizing the fusion of gametes in genetic crosses. Punnett was Professor of Genetics at Cambridge University. He wrote a large number of papers between 1900 and 1958, most of which helped to confirm and extend Mendel's work.

Science Titbits

Observation, assumption, experimentation and creativity, all of them are evident of Mendel's approach. The experiments performed by Mendel were elegant and his conclusions constitute foundation of the modern science of Genetics. Mendel is therefore appropriately called, the father of Genetics.

Punnett square indicates that ¼ of F_2 progeny would have been "RR" (homozygous round), ¼ + ¼ = ½ "Rr" (heterozygous round), and ¼ "rr" (homozygous wrinkled). Mendel actually observed 3:1 phenotypic ratio in F_2. His phenotypic data of F_2 can also be explained on the basis of 1:2:1 genotypic ratio of F_2. Mendel compared the result of all the seven separately studied characters, and found them strikingly similar; therefore, he became able to formulate **law of segregation**. According to law of segregation, "the two coexisting alleles for each trait in an individual segregate (separate) from each other at meiosis, so that each gamete receives only one of the two alleles". Alleles unite again at random fertilization of gametes when zygote is formed.

INHERITANCE OF TWO TRAITS (DIHYBRID CROSS)

The inheritance of two traits simultaneously can be studied in dihybrid cross (a cross between two individuals, which are different on two traits).

The two of the seven characters Mendel studied were seed colour and shape. Seeds shape may be either round (dominant) or wrinkled (recessive) and colour of the seed may be either yellow (dominant) or green (recessive).

Procedure and observations

When he crossed a homozygous round yellow (RRYY) plant with homozygous wrinkled green (rryy) plant, in F_1 generation all the offspring were produced with both dominant

Teacher's Point

The teacher would ask the students to answer that "why true breeding pea plants were important for Mendel's experiment"?

phenotypes i.e., round yellow. In order to analyse the genotype of F_1 plants, he self-fertilized them and produced F_2 generation. He was expecting that dominant and recessive combinations would be produced in 3:1 in F_2 generation as he had obtained in monohybrid cross, but he observed that offspring were produced in four phenotypic combinations i.e., round yellow, round green, wrinkled yellow and wrinkled green in the ratio of 9:3:3:1. The outcome of recombinant phenotype such as round green and wrinkled yellow, were surprising.

Interpretations of the results

Based upon these observations, Mendel concluded that the F_1 offspring (round yellow) were dihybrid i.e., heterozygous (RrYy) for both traits. The key incidence in the experiment was happened when F_1 plants self-pollinated and produced F_2 offspring. An F_1 plant can produce four classes of gametes (RY, rY, Ry, and ry) in equal quantities. If the sperms of the four classes are crossed with eggs of the four types, there will be 16 (4X4) equally probable ways in which the alleles can combine in F_2 generation as shown in the Punnet square in figure 22.2. These combinations make up four phenotypic categories with a ratio of 9:3:3:1.

Mendel tested his seven pea characters in various dihybrid combinations and always observed as 9:3:3:1 phenotype ratio in F_2 generation. The results of Mendel's dihybrid experiments are the basis of for what we now call **law of independent assortment,** which states that "each pair of alleles assort independently of other pairs of alleles during gamete formation". In other words, the alleles of each pair of contrasting trait have equal probability to assort with the alleles of other pair.

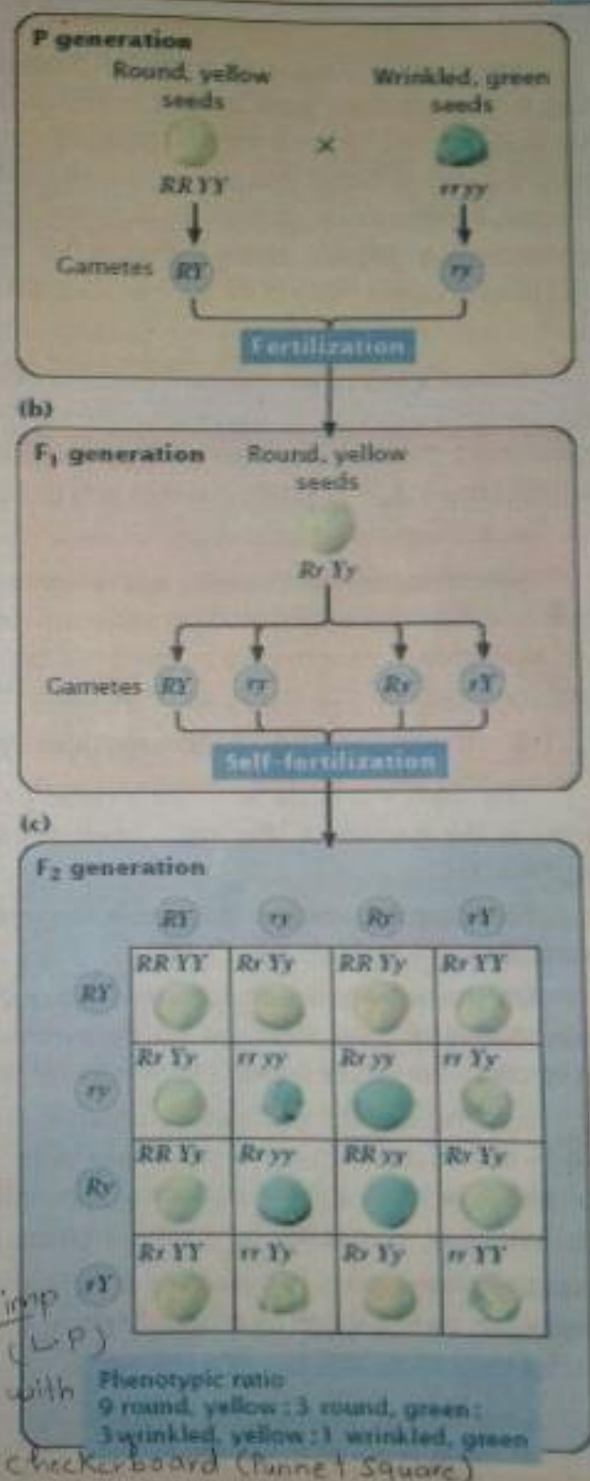

Fig. 22.2 Dihybrid cross showing the inheritance of single trait

Limitations of law of independent assortment

Although Mendel's work forms the basis of heredity, it does not cover all situations. The fact is that Mendel's work applies to diploid organisms, and not all organisms are diploid. Moreover, genes on the same chromosome could not be expected to assort independently. An offspring that inherited one trait would also inherit the other, unless crossing over occurred. Further, if genes are located on the X chromosome, the pattern of distribution in the succeeding generations is different. Males (because they have only one X chromosome - their other is Y, and does not carry many of the genes) are more likely to show recessive X linked characteristics.

Usefulness of law of independent assortment

Mendel's law of independent assortment explains that if two parents have unique set of traits which are desired to express in one individual, so it is possible only because of independent assortment. Furthermore, if two parents have such traits which they do not want to be expressed in their offspring. This is also possible only because of independent assortment.

Scope of independent assortment in variation

Beside mutation and crossing over (which are sources of variation), independent assortment of traits is also a major source of variations in successive generations. It is only due to the crossing over and independent assortment of the traits that the characteristics may appear in new combination in next generation which is often seems necessary for adaptations in varying environment.

22.1.3 Inheritance and Mathematical Probabilities

The chance to occur an event is called **probability**. The probability (P) that an event will occur is the number of favourable cases (a) divided by the total number of possible cases (n): $P = a/n$

For example, when we toss a coin, the probability (P) of onset of head or tail (a) out of two possibilities (n) is $P=a/n$ i.e., $P=1/2$

Mendel had a firm background of mathematics. He understood that the segregation of allele pairs during gamete formation on the reforming of pairs at fertilization obey the rules of probability. Let us see how the rules of probability apply to inheritance.

In genetics, the inheritance of a specific phenotype in a cross also has certain probability like in monohybrid cross the probability of an offspring to inherit dominant phenotype in F_1 generation is 100% and the probability of inheritance of recessive phenotype in F_1 generation is 0%. Whereas, probability of dominant phenotype in F_2 generation is $\frac{3}{4}$ and probability of recessive phenotype in F_2 generation is $\frac{1}{4}$. The combined probability of two or more independent events can be calculated by **product rule**.

Product Rule

According to this rule the probability of round yellow phenotype in F_2 generation of a dihybrid cross is equal to the product of individual probabilities of round (3/4) and yellow (3/4) phenotypes i.e., $P = 3/4 \times 3/4 = 9/16$

Skills: Analyzing, Interpreting and Communication

- **Hypothesize that in a dihybrid inheritance pattern of colour and texture of pea seed, the two traits are not interdependent.**

It has already been observed that characters like colour of pea seed and texture of pea seed are inherited independently of each other. The explanation lies in the behaviour of the chromosomes at meiosis. Independent assortment requires that genes concerned are carried on different chromosomes: for example the alleles of the gene for seed colour are located on one pair of chromosomes and the alleles of the gene for texture of the seed on another pair of chromosome.

Skills: Analyzing, Interpreting and Communication

- **Solve at least 4 genetic problems, to illustrate the law of independent assortment.**

22.1 PKU and albinism are two autosomal recessive disorders, unlinked in human beings. If couple, each of them heterozygous for both traits, produce a child, what is the chance of their having a child with (a) PKU? (b) albinism? (c) Both traits?

22.2 For any gene with a dominant allele C and recessive allele c, what proportions of the offspring from a CC X Cc cross expected to homozygous dominant, homozygous recessive and heterozygous?

22.3 Two tall, yellow seeded pea plants were crossed, and some dwarf, green seeded plants resulted. (a) What were the genotypes of the parent plants? (b) What possible genotypes might there be among the tall, yellow-seeded offspring?

22.4 A TtYy pea plant self-pollinates and one seed is picked at random for planting. (a) What is the chance that the seed will produce a tall, green seeded pant? (b) If it turn out to be tall and yellow-seeded what is the chance that its genotype is TTYY.

22.2 EXCEPTIONS TO MENDELIAN INHERITANCE

Since Mendel's time, our knowledge of the mechanisms of genetic inheritance has grown immensely. For instance, it is now understood that in how many different ways, alleles interact with their contrasting partner alleles at the same locus. These relationships between the contrasting alleles at the same locus in heterozygous state are called **dominance relations**. Although Mendel had observed only one form of dominance relation (**complete dominance**) but later on many geneticists became able to explain several exception to the Mendelian inheritance that could not be explained on the basis of complete dominance. These exceptions are said to have non-Mendelian inheritance patterns. For example incomplete dominance, co-dominance and multiple alleles.

22.2.1 Incomplete Dominance

Studies of the inheritance of many traits have shown that member of a pair of alleles may not be completely dominant to other. For example red and white are common flowers in Japanese four O'clock (*Mirabilis jalapa*). Each colour phenotype produces same phenotypes

Teacher's Point

The teacher would ask the students to write a brief research paper on 'Mendel's era about genetics were the beginning of a new era of biology'

when these plants are self-pollinated. Without knowing which is dominant, we might predict that all would have red flowers or all would have white flowers. In 1899 this cross was first made by German botanist **Carl Correns**, who found that all F_1 offspring have pink flowers. When two of these pink flowered plants are crossed, red flowered, pink-flowered and white-flowered offspring appear in a ratio of 1:2:1. The pink-flowered plants are clearly the heterozygous individuals and neither the red allele nor the white allele is completely dominant. When the heterozygous has a phenotype that is intermediate between those of its two parents, the dominance relation between the parental genes is said to be **incomplete dominance.**

Fig. 22.3 Incomplete dominance in 4-o'clock plants

Genetic Problem 22.5: *When pink flower four O'clock plant is crossed with a red flower plant what is the probability of: (a) Red flower plant (b) pink flower plant (c) ratio of pink flower to red flower plant?*

22.2.2 Co-dominance

Another variation on dominance relationships between alleles is called **co-dominance**, in which both contrasting alleles at the same locus express independently without influencing each other, so the phenotype of both the alleles become apparent. For example, the human MN blood group is determined by two co-dominant alleles (L^M and L^N) for two specific molecules located on the surface of red blood cells, the M and N molecules. A single gene locus at which two allelic variations are possible, determines the phenotypes of this blood group. Individuals homozygous for L^M allele (L^ML^M) have red blood cells with only M molecules; individuals homozygous for L^N allele (L^NL^N) have red blood cells with only N molecules; but both M and N molecules are present on the red blood cells of individuals heterozygous for *M* and *N* alleles (L^ML^N).

Science Tibits

When the dominance is not complete, the capital case and small case letters will not be used to represent the genes, instead, only capital case letter differentiated by numeric figures will be used to represent the phenotypes.

Table 22.1 MN blood groups system in example of co-dominance

Blood Group phenotype	Antigen	Genotype
M blood	M antigen	L^ML^M
N blood	N antigen	L^NL^N
MN blood	Both antigen	L^ML^N

Science, Technology and Society Connections

Evaluate incomplete and co-dominance as variations of Mendel's research.

Many patterns of inheritance which cannot be explained on the basis of Mendel's laws alone were discovered in plant and animals. Such patterns of inheritance are described as non-Mendelian inheritance. Incomplete dominance is a type of interaction where both the alleles of a given trait express as a blend (mixture) as against a normal Mendelian pattern where one allele is dominant over the other. As a result of this blending, an intermediate character is expressed. Co-dominance represents a situation where two allelic genes when present together in an individual, express their traits independently instead of showing a typical dominant recessive relationship. As a result the heterozygous progeny of the F_2 generation show a phenotype that is different from both the homozygous parent.

Table 22.2 Difference between incomplete dominance and co-dominance

Incomplete dominance	Co-dominance
1. In heterozygous state, both genes blend their phenotypic effects.	1. In heterozygous state, both genes independently express their phenotypic effects.
2. The heterozygotes show an intermediate phenotype between the two parental phenotypes.	2. The heterozygotes show both parental phenotypes at a time.
3. Example: Flower colour of 4 O'clock plant.	4. Example: Human MN blood and AB blood groups.

Multiple Alleles

So far we have discussed inheritance patterns involving only two alleles per gene. But many genes have more than two alleles all of them are called **multiple alleles**. Multiple alleles are produced by gene mutation. All the multiple alleles of a trait occupy the same locus. Some traits may be controlled by as many as 100 or more multiple alleles but each individual carries only two of them as each locus is twice represented in a diploid individual. For example, if a trait is controlled by 3 multiple alleles i.e. A_1A_2 and A_3but every individual carries any two of them like A_1A_1, A_1A_2, A_1A_3, A_2A_2, A_2A_3 or A_3A_3. The ABO blood groups in humans are one example of multiple alleles.

22.3 BLOOD GROUP SYSTEMS

There are a number of different blood group systems found in human. The International Society of Blood Transfusion has recognized up to 30 major blood group systems. These systems are characterized by the presence or absence of specific molecules, called **antigens** that are situated on the surface of the red blood cells. Most antigens are glycoprotein molecules. Two main blood group systems are ABO system and Rh (Rhesus) system. These two systems are more significant because incompatibility between donor and recipient's blood with respect to these two systems may become dangerous to life.

There are more than two hundred minor blood groups (belong to the rest of blood group system other than ABO and Rh systems) that usually do not complicate the blood transfusions. These are known as **rare blood types**. One example of such blood group systems other than "ABO" and "Rh" is already given in co-dominance in the form of MN blood group system.

22.3.2 ABO Blood Group System

A well-known example of multiple alleles is the ABO blood group system in human. Karl Landsteiner discovered it in 1901. ABO blood groups are also found in many other primates such as apes chimpanzees, bonobos, and gorillas.

Antigens of ABO system

There are several surface markers on the RBC membrane, which are generally called **antigens**. ABO blood group system is based upon two such antigens i.e., A antigen and B antigen which are glycoprotein in nature. If A antigen is present on the surface of RBC, the blood group is called **type A** and if B antigen is present, the blood group is called **type B**. If both antigens are present, the blood group is **type AB** and if none of these are present, the blood group is **type O**.

Science Tidbits

ABO Blood type antigens are not only found on the surface of red cells. They are also normally secreted by some people in their body fluids, including saliva, tears, and urine. Such persons are called secretors. Whether someone is able to secrete them is genetically controlled by a dominant secretor gene "Se" presents on chromosome 19.

Genetic basis of ABO system

ABO blood group is controlled by autosomal single polymorphic gene *I* (which stands for isohaemagglutinogen). The four blood types result from various combinations of three different alleles, symbolized as I^A (for the ability to make substance A) I^B (for B) and *i* (for neither A nor B). Their dominance relations are very interesting, alleles I^A and I^B are completely dominant over the allele *i*, while I^A and I^B are co-dominant to each other because each expresses in equally in heterozygous (I^AI^B) state to produce AB phenotype. Therefore I^AI^A or I^Ai genotypes will produce phenotype A. Similarly I^BI^B or I^Bi produces phenotype B. The homozygous *ii* will produce phenotype O.

The blood groups alleles start their expression at early embryonic stage and keep on expressing themselves till death. Therefore, the blood group phenotype of a person never changes throughout life.

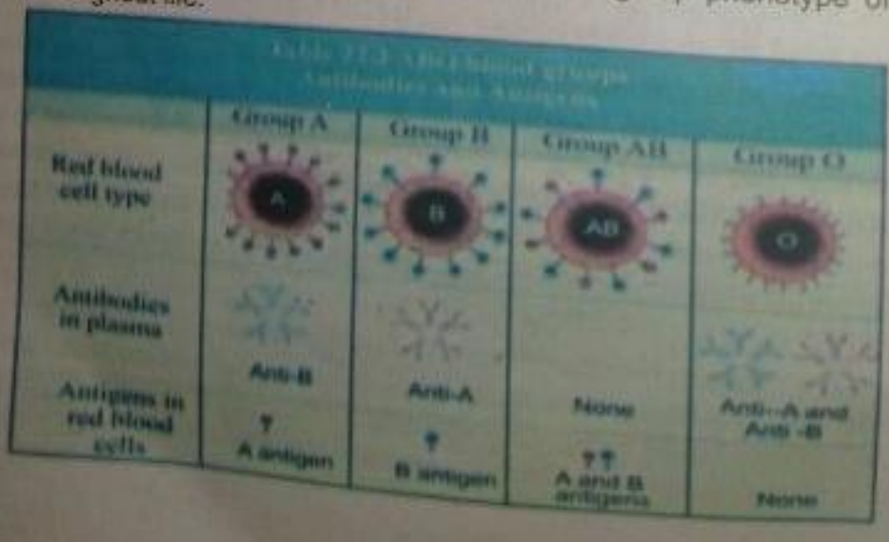

Table 22.4 Multiple alleles and ABO blood groups

Blood group (phenotype)	Antigen	Genotypes	Antibodies	Transfusions
A	A	I^AI^A or I^Ai	B	A and O
B	B	I^BI^B or I^Bi	A	B and O
AB	Both	I^AI^B	None	Any
O	None	ii	Both	Only O

Antibodies of ABO system

It has been observed that if wrong transfusion is carried out, the recipient's blood start **agglutination** (antigen-antibody reaction) and clumping occurs. This is due to the presence of antibodies against wrong antigen. These antibodies are produced in the absence of their corresponding antigens. For example, those with **type A** blood have **anti-B** antibody in their plasma. Similarly, **type B** people have **anti A** antibody and those with type **AB blood** have no anti A or anti B antibodies in their plasma. **Type O** individuals have **no antigens** but have both anti A and anti B antibodies in the plasma. These antibodies of ABO system do not require any stimulus to produce their production begins from embryonic life and remain continue throughout life.

Transfusion principle

When transfusions are carried out between two incompatible (different) blood groups, antigens of donor react with the antibodies (also called **agglutinins**) of the recipient, then the red blood cells clumps with one another. The clumping of red blood cells is known as **agglutination**. Therefore, the transfusions are carried out on the basis of donor's antigens and recipient's antibodies. Due to these limitations the persons with type A blood group can receive blood from type A or from type O because they have anti B antibody so they cannot be given any blood carrying B antigen. Similarly, the persons with type B blood group can receive blood from type B or from type O but those with type AB blood group can receive any blood since they do not have antibody to react with donor's blood, hence they are called **universal recipient.** The persons with type O blood group can receive blood only from type O because they have both antibodies that can react with any antigenic blood (type A, B or AB) but they can donate to any one as they do not have any antigen to interfere with recipients' blood. Therefore they are called **universal donor.**

Fig. 22.4 Blood Transfusion Model

Genetic Problem 22.6: *A person has type A blood group while his wife has type B, they have four children each with different blood group of ABO system. Explain that how is it possible?*

Genetic Problem 22.7: *A woman with blood type B has a child with blood type O. What are the genotypes of the mother and child? Which genotypes could the father not have?*

Science, Technology and Society Connections

Derive an idea to get alternatives of blood transfusion

Some approaches are available that can decrease the need for a blood transfusion. The options currently available are:

1. Volume expanders are used to prevent or treat the shock associated with loss of body fluids. The most common volume expanders used include salt water (normal saline) and saline with some chemicals added (Ringer's solution).
2. Hematopoietic growth factors encourage the bone marrow to make more red blood cells. These growth factors can be made in the laboratory and given to people with low blood cell counts.
3. Erythropoetin is a naturally occurring hormone produced by the kidneys. It stimulates the body to produce more red blood cells and is used to treat anaemia. It is widely used as a transfusion alternative.
4. Aprotinin is a drug that is given prior to heart surgery to reduce the risk of bleeding and the need for transfusion.

Science, Technology and Society Connections

Justify why a recessive blood group allele 'i' is more frequent in the population.

The cross between blood group O *(ii)* and O *(ii)* will have blood group O *(ii)* in all the offspring. The cross between heterozygous I^Ai and I^Ai will produce 25% offspring having blood group O (*ii*), and likewise heterozygous I^Bi and I^Bi will produce 25% offspring having blood group O (*ii*). Cross between heterozygous I^Ai and I^Bi will produce 25% offspring having blood group O *(ii)*. That's why blood group allele 'i' is more frequent in the population. For example Australia 40%, Canada 39%, Iceland 47%, Ireland 47% UK 37%, USA37%.

Science, Technology and Society Connections

Justify blood donation as a service to suffering humanity.

Blood donation is a social responsibility. The donor is donating for it as it will be used in saving lives of human beings. As The Quran says in Surah 5 verse 32 *"if anyone saves a life, it shall be as though he had saved the lives of all mankind."* Millions of people owe their lives to people whom they will never know or meet in their lifetime. They are none other than those people, who have donated their blood freely and without any reward - voluntary blood donors. Voluntary unpaid donors are the foundation of a safe blood supply which saves millions of human beings from the jaws of untimely death. Blood donation is a noble, selfless service. It gives the donor a feeling of joy and contentment. Also this is an expression of love for Mankind, as blood knows no caste, colour, creed, religion or race, country, continent or sex. Do you know that 'one unit of blood can save three lives rather than one?

22.3.3 Rh Blood Group System

The Rh blood group system is clinically the most important blood group system after ABO. The name of this system (Rh) is derived from Rhesus monkey, because its antigen was first discovered in it by Landsteiner in 1930s.

Antigens of Rh blood group system

The Rh blood group system is characterized by the presence or absence of **"D" antigen** also called **Rh factor**. The persons having this antigen are called Rh positive and those in which it is absent are called Rh negative. The D antigen incompatibility between donor and recipient can cause problem not only during blood transfusion but it is also a relevant cause of the **haemolytic disease** of the newborn or **erythroblastosis foetalis**.

Genetic basis of Rh blood group system

Rh blood group system is mainly controlled by *"D"* gene which determines the formation of *D* antigen or Rh factor, while its alternative allele *"d"* inhibits the formation of Rh factor. *D* is completely dominant over *d*, therefore, persons having genotypes *"DD"* or *"Dd"* have *D* antigen (Rh factor) on their RBC and are Rh positive. Persons with genotype *"dd"* do not have Rh factor and are Rh negative.

Table 22.4 Rh-Blood groups system

Blood group (phenotype)	Rh-Antigen/factor	Genotypes	Anti Rh-Antibody	Transfusions
Rh^{+ve}	Present	*DD* or *Dd*	Not produced	Rh^{+ve}, Rh^{-ve}
Rh^{-ve}	Absent	*dd*	Produced (if stimulated)	Rh^{-ve}

The blood group O^{-ve} (O negative) is actual universal donor because it has no antigen of ABO system and of Rh system which can interfere with recipient's blood, whereas, AB^{+ve} (AB positive) is actual universal recipient because it has neither anti-A and anti-B nor anti-Rh antibodies, therefore, it cannot resist any donor's blood.

Anti Rh-antibody and Transfusion principle

Rh blood group system also has a mechanism of antibody production i.e., anti-Rh antibody, which is produced in Rh negative blood. Unlike ABO antibody production mechanism, the production of anti Rh antibody is different in the sense that it always requires a stimulus in the form of exposure to Rh factor for its production.

An Rh negative person does not produce anti-Rh antibodies unless he is exposed to Rh antigen. Rh positive donor is totally incompatible for Rh negative recipient. If Rh negative person receives an Rh antigen through wrong Rh positive blood transfusion, he will begin to produce anti-Rh antibodies against Rh antigens. Once the mechanism of anti-Rh antibody production is stimulated, then it remains continue for whole life. Rh negative blood, clear of any anti-Rh antibody from a donor who has never been exposed to Rh antigen can be transfused to Rh positive recipient.

22.3.4 Erythroblastosis Foetalis

Erythroblastosis foetalis develops in a foetus, when anti-Rh antibodies produced by the mother pass through the placenta.

Teacher's Point

The teacher would ask the students to build a thematic chart for the blood groups of their class fellows and identify the antigens present in the blood.

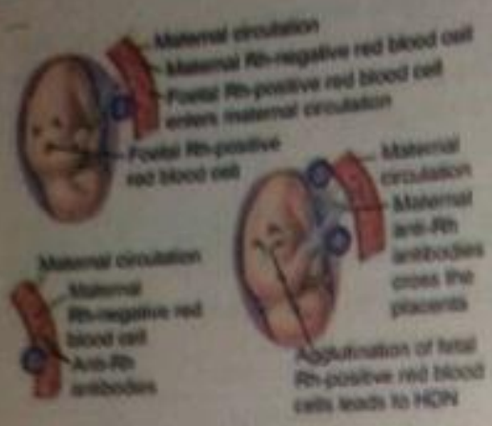

Fig. 22.5 Maternal-Foetal Rh-incompatibility

Science Titbits

An **erythroblast** is a type of red blood cell which still retains a cell nucleus. It is the immediate precursor of a normal erythrocyte.

Science Titbits

Incompatibilities between ABO blood types are less common than those of the Rh factor and tend to be less severe. Sometimes a mild ABO incompatibility protects the baby against the most severe Rh incompatibility. If O^{-ve} mother conceives A^{+ve} or B^{+ve} baby, any foetal A or B type RBC entering the mother's blood are quickly destroyed by her anti-A or anti-B antibodies before she can form anti-Rh antibodies.

Rh-negative mother is given an injection of Rh antiserum (serum containing anti-Rh antibodies) during early pregnancy (1st trimester) and immediately after birth within 72 hours of delivery. This causes any of the baby's red blood cells that may have crossed into the mother's blood to be destroyed before sensitizing the mother's immune system to produce maternal anti-Rh antibodies. The injected antiserum disappears before the next pregnancy. This has to be done with each pregnancy whether it ends in a delivery or an abortion.

Problems and complications in foetus

When mother's anti-Rh antibodies seep through the placenta into the blood circulation of foetus, they start haemolysis (breakdown of blood). As this destruction continues, the foetus becomes anaemic. The anaemic foetus starts to release many immature erythroblasts into his blood stream; therefore, the disease is called **erythroblastosis foetalis**. This anaemia may leads to abortion or still birth. Even if the pregnancy continues, the liver and spleen of the foetus swell as they rapidly produce RBCs. The breakdown product of RBC called **bilirubin** also accumulates in the foetus. Bilirubin damages brain cells and turns skin and whites of the eye yellow of the foetus. This condition is called **jaundice**. So the baby if born alive, suffer from severe haemolytic anaemia and jaundice. Such baby's blood should be immediately replaced by Rh-negative blood free of anti-Rh antibodies.

Causes and risk factors

Erythroblastosis foetalis most commonly happens when a woman with Rh-negative blood gets married to a man with Rh-positive blood and conceives a baby with Rh-positive blood (maternal-foetal Rh incompatibility). If the man's genotype is *DD*, all of their offspring will have *Dd* genotype and will be Rh positive. If the man's genotype is *Dd*, half of their offspring with *Dd* genotype will be Rh positive. There is always chance of erythroblastosis foetalis whenever an Rh-positive foetus is conceived by Rh-negative mother.

Genetic Problem 22.8: *An Rh negative woman is married to an Rh positive man whose father was also Rh negative. What are the possible genotypes of each person in the family, what are the chances that their child will be affected with erythroblastosis foetalis?*

22.4 POLYGENIC INHERITANCE AND EPISTASIS

Some traits have large number of alternative phenotypes that have small and less striking difference so they have continuous variations such as height, weight, intelligence and skin colour in human; and grain colour in wheat. Such traits cannot be encoded by a single gene with two alleles. Even a few multiple alleles of a single gene cannot make a large number of phenotype. Such traits are encoded by alleles of two or more different gene pairs found at different loci, all influencing the same trait in an **additive way** i.e., the intensity of phenotype depends upon the number of particular effect causing alleles. These quantitative traits are therefore called **polygenic traits.** All the genes that control a quantitative trait are called **polygenes**, which have a small positive or negative effect on the character. Polygenes supplement each other and sum of positive and negative effect of all individual polygenes produce quantitative phenotype of a continuous varying traits.

Fig. 22.6 Range of colours in wheat grain colour

22.4.1 Wheat Grain Colour

Wheat grain colour is a good example of polygenic (multiple gene) inheritance. Wheat grains show a continuous variation in colour from white to dark red. Approximately seven different phenotypes (given in the table 22.5) are found in wheat population all over the world.

The genetics of wheat grain colour was studied. When a homozygous dark red grain plant was crossed with a homozygous white grain plant, all F_1 grains had light red colour intermediate between two parental shades. It seemed as if it was a case of incomplete dominance. But when F_1 grains were grown to mature plants and crossed with each other, F_2 grains had exactly seven shades of colour in the following ratio:

Table 22.5 Range of phenotypes in wheat grain colours

Dominant alleles	6	5	4	3	2	1	0
Range of phenotypes	Dark red	Moderately dark red	Red	Light red	Pink	Light Pink	White
Ratio	1	6	15	20	15	6	1

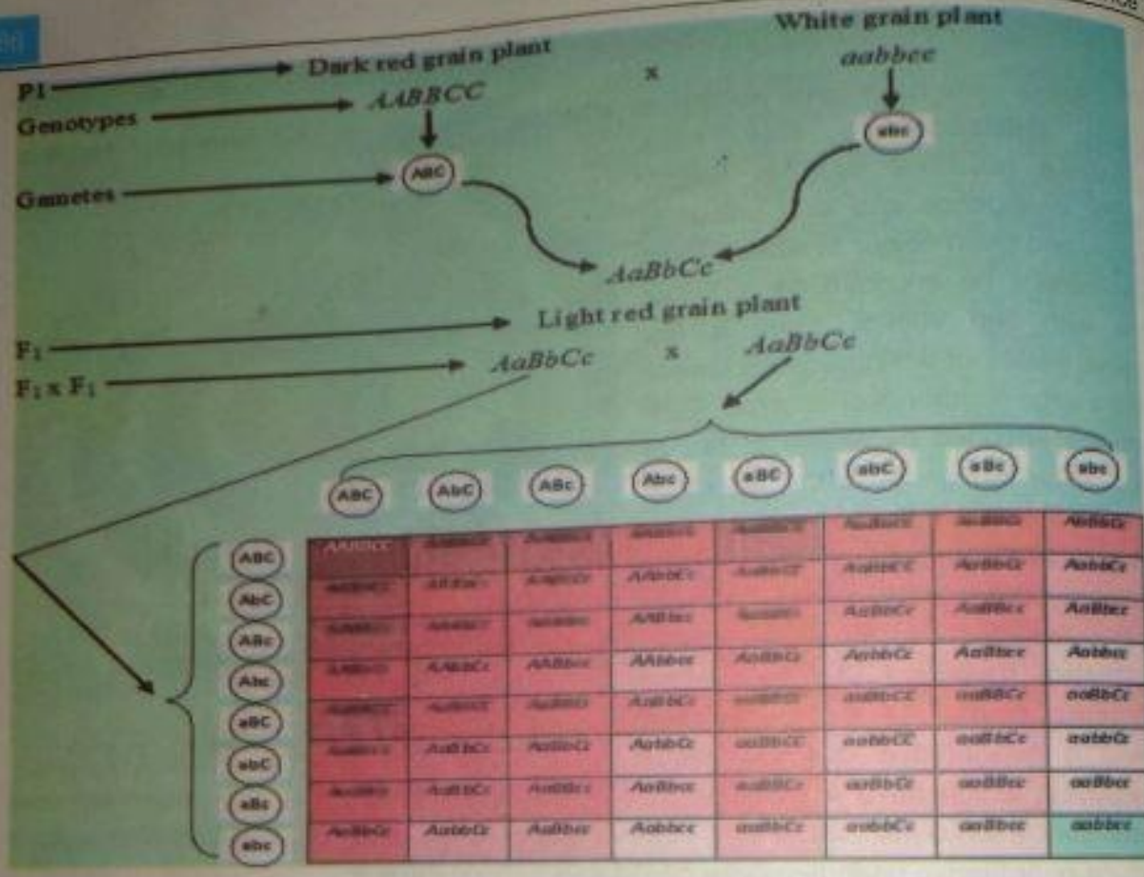

Fig. 22.7 Inheritance of wheat grain colour

Finally it was identified that actually three different gene pairs, Aa, Bb, Cc at three different loci contribute to the wheat grain colour. Each individual would contain six alleles for the trait. Alleles A, B and C codes for an equal amount (dose) of red pigment, which is a positive effect. But none of a, b and c encode red pigment, which is a negative effect. If all the six allele code for red pigment (AABBCC), the grain is dark red; when none of the six allele encodes red pigment (aabbcc), the grain is white. When a grain has one allele for red pigment (Aabbcc or aaBbcc or aabbCc) its colour is light pink; if it has two alleles for the pigment (AaBbcc or aaBbCc or AabbCc) it is pink, if it has three pigment alleles (AaBbCc or AABbcc or AabbCC), it will be light red. Similarly four alleles colour dose (AABBcc or aaBBCC or AAbbCC) will make red and five alleles colour dose (AABBCc or AABbCC or AaBBCC) will produce moderately dark red grain. Thus colour phenotypes of grains depend upon the number of pigment producing alleles (A, B, and C). Environmental factors like light, water and nutrient also influence the amount of grain colour.

22.4.2 Inheritance of Human Skin Colour

Human skin colour is a good example of polygenic (multiple gene) inheritance. Skin colour is largely determined by the amount of melanin the skin produces. Dark-skinned individuals

produce more melanin than light-skinned individuals. At least three genes regulate the amount of melanin produced.

1. **Gene *A*** is involved in the permanent survival, proliferation and migration of melanocytes.
2. **Gene *B*** encodes the enzyme **tyrosinase**, which is involved in the production of melanin from tyrosine.
3. **Gene *C*** is primarily responsible for determining whether pheomelanin or eumelanin (see glossary) is produced in humans.

Each gene has two forms, the dark-skin alleles are represented by capital case letters (*A*, *B* and *C*) and light-skin allelesare represented by small case letters (*a*, *b* and *c*). No allele is completely dominant to the other and heterozygotes exhibit an intermediate phenotype (incomplete dominance). *A*, *B*, and *C* act as dark-skin alleles in the genotype, because they add pigment by increasing melanin production. On the other hand *a*, *b*, and *c* act as light-skin alleles in the genotype because they inhibit melanin production. There are seven different shades of skin colour ranging from very light (*aabbcc*) to very dark (*AABBCC*); most individuals have the intermediate skin colour (*AaBbCc*). This *AaBbCc*, genotype would be characteristic of a **mulatto** (an offspring of a black and a white parent). In the cross between two mulatto genotypes (*AaBbCc X AaBbCc*), each parent produces eight different types of gametes and these gametes combine with each other in 64 different ways resulting in a total of seven skin colours. The skin colours can be represented by the number of capital letters, ranging from zero (no dark skin alleles) to six (all dark skin allele).

22.4.3 Epistasis

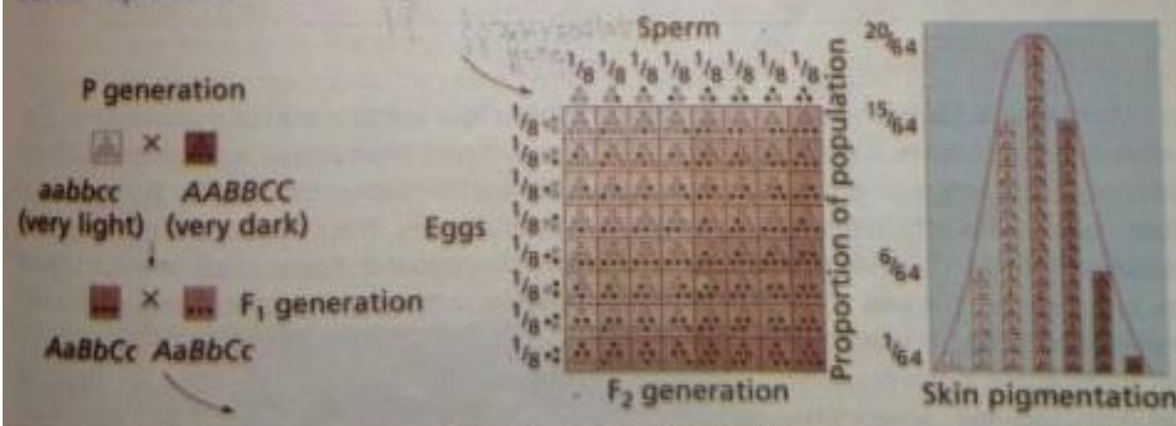

Fig. 22.8 Inheritance of human skin colour

An example of nonallelic interactions is epistasis, which can be defined as the phenomenon in which the effect caused by the genes at one locus interferes with or hides the effect caused by another gene at another locus. In such interactions, the gene which suppresses or masks the effect of action of a gene at another locus is known as **epistatic gene** or **inhibiting gene** and the gene which is suppressed is known as **hypostatic gene.**

Epistasis must not be confused with dominance. Dominance is the relationship between alleles of the same gene occupying the same locus, but epistasis is the interaction between different genes occupying different loci. Following table further illustrate the difference between epistasis and dominance.

Table 22.6 Difference between dominance and epistasis

Dominance	Epistasis
1. It involves the single pair of alleles.	1. It involves the two pair of alleles.
2. A gene suppresses the expression of its own allele.	2. A gene suppresses the expression of allele of another gene.
3. Only the recessive allele is suppressed.	3. It suppresses the effect of both dominant and recessive alleles of another gene.
4. The effect is only due to dominant allele.	4. Both dominant and recessive allele can become epistatic.

Science Titbits

The expression of ABO genotypes (locus is on chromosome 9) also depends upon another gene H (locus is on chromosome 19) that encodes a particular H substance. The H substance is a precursor to the A and B antigens. For instance, the I^B allele must be present to produce the B enzyme that modifies the H substance to become the B antigen. It is the same for the I^A allele. However, if only recessive alleles for the H substance are inherited (hh), the H substance will not be produced. Subsequently, the A and B antigens also will not be produced. The result is an O phenotype by default since a lack of A and B antigens is the O type. This seemingly impossible phenotype result has been referred to as a **Bombay phenotype** because it was first described in that Indian city.

Relationship of epistasis with polygenic inheritance

In epistasis the expression of a gene is controlled by the expression of another gene. For example, in mice there are different genes for fur colour, but there is also a gene that controls whether or not any pigment is produced at all. This is very much related to the phenomenon where one trait is affected by the expressions of many genes, the polygenic inheritance. For example, people vary greatly in traits such as height, skin colour and nose length because there are many genes that can affect the growth of a person. The only distinction between the two phenomena is that in epistasis the traits show discontinuous variations among alternative phenotype whereas, in polygenic inheritance the traits show a long range of alternative phenotypes with continuous variations.

Fig. 22.9 Coat colour in the *Labrador retriever*

Coat colour in the Labrador retriever

Coat colour in *Labrador retriever* (a highly popular type of dog) is an excellent example of epistasis in animals. They have coats of three basic colours: yellow, black, and chocolate. The mode of inheritance of coat colour is autosomal (not related to the sex of the dog), with the information (genes) for black and brown at a different location in the chromosomes (locus) from the information (genes) for yellow.

The allele for black coat colour (B) is dominant to the chocolate colour (b). Therefore, a puppy will only be chocolate if each parent contributes the chocolate alleles (bb). If one (Bb) or both (BB) parents contribute the black (dominant) gene, the puppy will be black (BB or Bb).

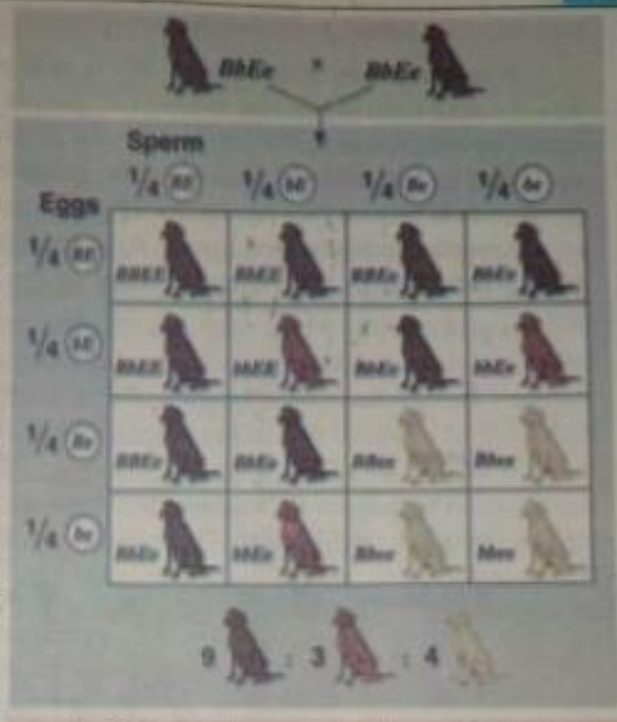

Fig. 22.10 Inheritance of coat colour of *Labrador retriever*

The gene that determines yellow coat colour is at a different location, (locus) in the DNA from the black versus chocolate gene. In order to be yellow, a *Labrador* must have two recessive copies of the yellow gene (ee). In this case, the yellow colour genes become epistatic and completely inactivate the black or brown genes, and the puppy is yellow. This means both parents contributed a yellow gene (e). However, if only one (Ee) or no (EE) yellow genes are contributed, the puppy will be either black or chocolate, determined as explained above by what is on the black/chocolate gene. Two yellow Labradors (ee) can only have yellow puppies (ee), since they both have two copies of the yellow gene and that is all they can contribute. If a black *Labrador*, homozygous for both gene pairs (BBYY) is crossbreed with a yellow (bbee) partner, all the offspring will be black (BbEe). If such black Labradors, heterozygous for both gene pairs (BbEe) are interbreed (as shown in figure 22.10), all three coat colours (black, chocolate and yellow) are expected in offspring in 9:3:4 ratio.

Genetic Problem 22.9: *When two chocolate coloured Labradors were crossed, a yellow puppy was born, what is the probability of yellow coat coloured puppy if the parents are again crossed?*

Continuous Variation
1. Height, Skin colour

Discontinuous Variation
1. Blood group

Teacher's Point

The teacher would ask the students to write a paragraph on 'the importance of the dog *Labrador retriever*'

Inheritance of flower colour (pigment phenotype) of sweet pea (*Lathyrus odoratus*)

A best example of epistasis in plants is flower colour in sweet pea, *Lathyrus* odoratus. Its flower colour exist in two phenotypes i.e., **purple** (dominant) and **white** (recessive). The purple colour develops due to the production of a purple pigment, the **anthocyanin**. Its production is controlled by two different gene loci. The presence of at least single dominant allele of both the gene pairs is required for production of anthocyanin. The dominant allele of one gene "A" acts on a colourless precursor (substrate A) to produce an intermediate colourless product which on getting activated by the dominant allele of the second gene "B" results in the formation of

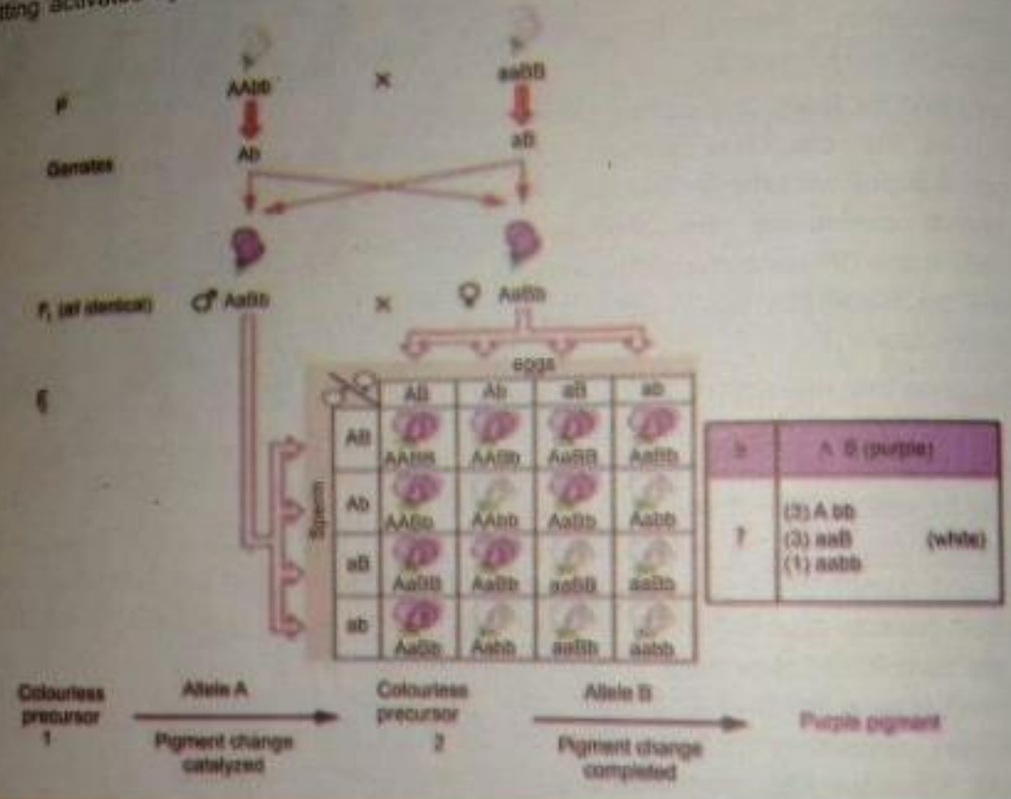

Fig. 22.11 Inheritance of flower colour of Sweet pea (*Lathyrus odoratus*)

anthocyanin pigment leading to production of purple coloured flowers. Thus, dominant alleles "A" and "B" complement each other to produce purple colour. This type of interaction is also called **complementary gene interaction** because it involves the cooperative interaction of both the genes. On the other hand, if any one locus has homozygous recessive genotype AAbb or aaBB, that will interfere with dominant alleles and hides their expression, the result will be white flower. The recessive alleles in this case are epistatic in nature and this type of epistasis is called **duplicate recessive epistasis**.

If a white flower plant (AAbb) is crossed with another white flower plant (aaBB), all of the F_1 offspring will be purple flower plants. When these F_1 purple flower plants are self-fertilized then the F_2 ratio 9:3:3:1 would be altered to 9:7 and represents duplicate recessive epistasis.

22.5 GENE LINKAGE AND CROSSING OVER

Mendel didn't know anything about the physical nature of genes or that genes are part of chromosomes, because nature of chromosomes were not even discovered until long after his experiment were concluded.

22.5.1 Gene Linkage

The number of genes in a cell is far greater than the number of chromosomes. In fact each chromosome has hundreds and thousands of genes. Genes located on the same chromosome that tend to be inherited together in genetic crosses are said to be **linked genes**. The phenomenon of staying together of more than one gene on the same chromosome is called **gene linkage**. If genes are linked on autosomes, their linkage is called **autosomal linkage**. Similarly, if they are linked on sex chromosome, their linkage is called **sex linkage**. All the linked genes found on the same homologous pair of chromosome form a group, known as **linkage group**, so the number of linkage groups in an organism are equal to the number of homologous pair of chromosomes in that organism. The linked genes tend to be inherited together (*en bloc* inheritance) in the offspring, so usually they do not show recombination and do not assort independently in the offspring. So the ideal Mendelian ratio of independent assortment is deviated.

Detection of gene linkage

(Gene linkage can easily be detected by performing a **test cross** between two gene pairs (dihybrid test cross). In such type of test cross, a heterozygous individual for two traits (F_1) is back crossed with its recessive parent (P_1). If all four phenotypic combinations (parental and recombinants) are produced in equal 1:1:1:1 ratio, then there would be no linkage between the genes. If this ratio is deviated i.e., more parental types and less recombinant types, this indicates the **incomplete** or **partial linkage**; but if only parental types are produced, **complete** or **tight linkage** is believed. In a typical dihybrid cross, the complete or tight linkage inhibits the outcome of recombinant types and disturbs 9:3:3:1 ratio of independent assortment, as a result, only parental combinations are produced in 3:1. To see how linkage between genes affects the inheritance of two different characters, let's take an example from T. H. Morgan's experiments on *Drosophila*.)

Morgan's Experiment

T. H. Morgan studied about 85 pair of contrasting trait in fruit fly *Drosophila melanogaster*. Two of them were wing length and width of the abdomen. Allele for **long wings (Vg)** is dominant over **short** or **vestigial wing (vg)**. Similarly, allele for **broad abdomen (A)** is dominant over **narrow abdomen (a).** Morgan crossed a fly with long wings and a broad abdomen with one having vestigial wings and a narrow abdomen. The F_1 offspring all had long wings and broad abdomens. Then two of these flies were mated. In the F_2 generation about ¾ of the offspring had long wings and a broad abdomen and nearly all the remaining flies about ¼ of the total had vestigial wings and a narrow abdomen.

Gene linkage encounters independent assortment

Morgan's results were very different from the results he expected based on the law of independent assortment i.e., 9:3:3:1. *What had happened?* From his data Morgan concluded that the genes for abdomen width and wing length were located on the same chromosome so they did no assort independently during meiosis. Instead, they inherited together. Therefore no recombinant types were produced and the standard ratio of independent assortment 9:3:3:1 is modified to 3:1

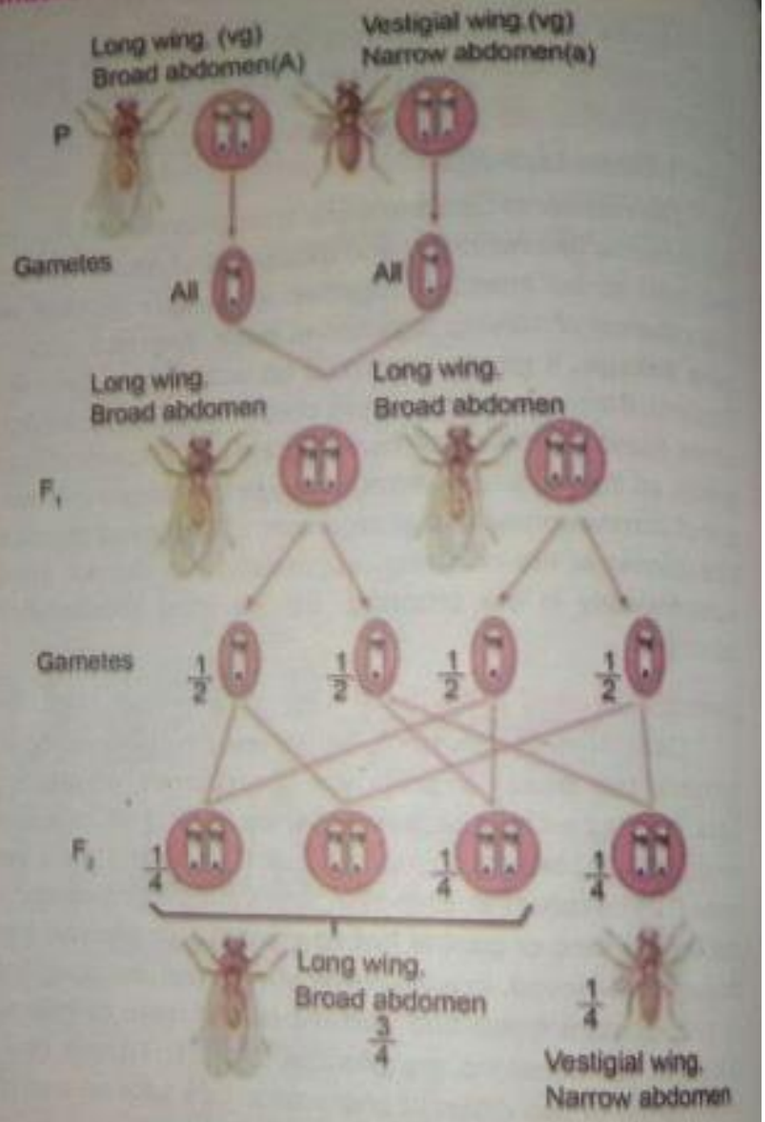

Fig.22.12 Linkage in fruit fly

Effect of number of progeny on detection of linkage

Gene linkage could be observed or evaluated only if the number of progeny is quite large because probability is used to determine the kinds of gametes produced and the chances of their combining. The larger the number of individuals, the greater is the likelihood that the laws of probability will hold. A small sample may not produce the results indicated by the laws of probability. Linkage can be recognized when an excess of parental type offspring and a few of recombinant type.

Teacher's Point

The teacher would ask the students to 'explain why alleles for long wing and broad abdomen in *Drosophila* inherited together' include the idea of gene linkage. To organize your idea, draw cause effect diagram that should show what happens to the two alleles during meiosis.

22.5.2 Crossing Over

Subsequent experiments demonstrated that the process which is responsible for the recombination of linked genes is crossing over. In crossing over, an exchange of maternal and paternal chromatid parts occurs while homologous chromosomes are paired during prophase of meiosis I. The recombinant chromatids resulting from crossing over may bring alleles together in new combinations, so when they are distributed in different gametes, a wide variety of gametes are produced. Therefore, crossing over leads to recombination of genes and thus responsible for genetic variability in sexually reproducing organisms. Percentage of crossing over or recombinant frequency has helped in locating the genes on chromosomes, determining their sequence and preparation of chromosomal maps.

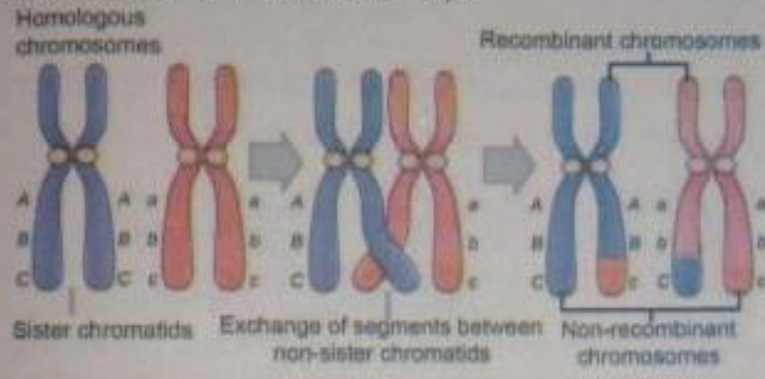

Fig. 22.13 Crossing over

22.6 SEX DETERMINATION

Although the anatomical and physical differences between women and men are numerous, the chromosomal basis for determining sex is rather simple. A clear picture of the genetic basis of sex determination emerged after the discovery of sex chromosome.

22.6.1 Genetic Identification of Sex Phenotypes

Most of the animals and plants have a genetic difference in male and female individuals which is reflected in their particular array (arrangement) of chromosomes i.e., **karyotype**. Let us see this difference and identify the male and female individuals in *Drosophila* and human.

Chromosomes in *Drosophila*

There are four pairs i.e., 8 chromosomes in *Drosophila*. Three pairs of chromosomes are identical in male and female, which are called **autosomes**. The fourth pair of chromosome is different in male and the female and determines genders are called **sex chromosomes**. In females both the sex chromosomes are identical, so these are called **XX chromosomes**. In males one is rod shaped and is like the sex chromosome of the female so it is also called **X-chromosome**, while the other chromosome hooked shape and it is called **Y-chromosome**.

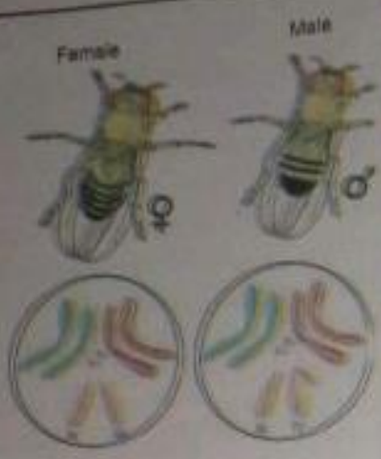

Fig. 22.15 Chromosomes in *Drosophila*

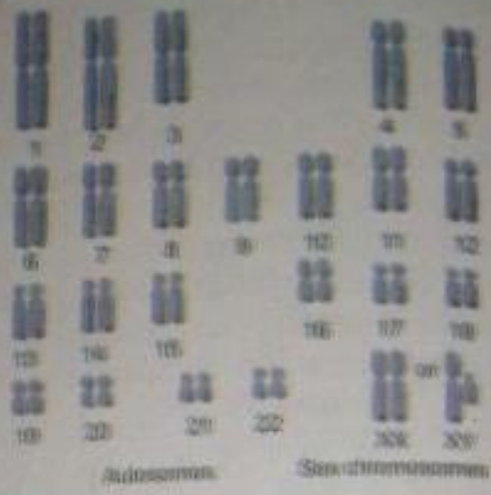

Fig. 22.16 Chromosomes in human

Chromosomes in Man

In humans there are 23 pairs of chromosomes, 22 pairs of which are autosomes, while one pair is sex chromosome. In female it is XX and XY in males. Human females have two copies of the X chromosome. All eggs cells produced by a woman contain one X chromosome. In males half of the sperms contain an X chromosome and half contain Y chromosome. The sex chromosome carried by the sperms is therefore determines the gender of the child. If a sperm carrying X-chromosome fertilizes the egg, the child will be a girl, but if a sperm carrying a Y chromosome fertilizes the egg, the child will be a boy.

Science, Technology and Society Connections

Technique employed for embryonic screening

Embryo screening checks an embryo to see if it's carrying any alleles for a genetic disease. A number of techniques are now available to test for chromosomal and gene mutations in human foetuses.

Amniocentesis: Before the start of the procedure, a local anesthetic can be given to the mother. A needle is usually inserted through the mother's abdominal wall, then through the wall of the uterus, and finally into the amniotic sac. With the aid of ultrasound-guidance, a physician punctures the sac in an area away from the foetus and extracts approximately 20ml of amniotic fluid. After the amniotic fluid is extracted, the foetal cells are separated from the sample. The cells are grown in a culture medium, then fixed and stained. Under a microscope the chromosomes are examined for abnormalities e.g.,Down syndrome (trisomy 21).

Aminocentesis

22.6.2 Patterns of Sex Determination

There is a wide variety of sex determining mechanisms but three patterns are more common.

XY – XX Type

This pattern of sex determination is found in *Drosophila*, man and many other organisms. Male is XY and female is XX. Male being heterogametic produces two types of sex-determining sperms. Half of the sperms carry X-chromosome and the other half carry Y – chromosome. Chances for both types of sperms are equal.

Female being homogametic produces only one type of eggs, each with an X chromosome. Sex of the offspring is determined by the type of sperm. If an X – carrying sperm fertilizes the egg, the zygote will be XX, and a female offspring is produced. If a Y – carrying sperm fertilizes the egg, the zygote will be XY, and a male offspring will be produced. The sex-ratio between male and female offspring is 1:1. Sex ratio indicates chances of the sex of the offspring. Chances for a son or daughter in human birth are equal.

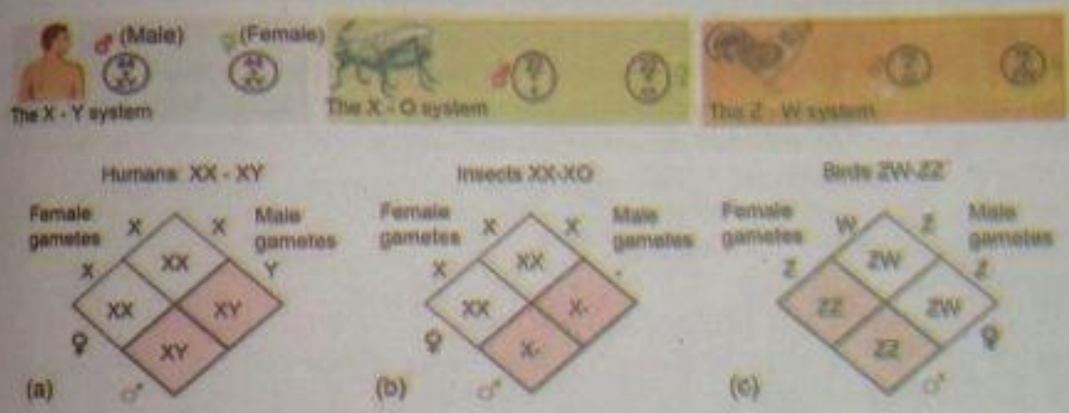

Fig. 22.17 Sex determination in animals

XO – XX Type

This pattern of sex determination is found in grasshopper and Protenor bug. Male is XO because it has only one X chromosome. The other sex chromosome is missing entirely. Male is heterogametic because it forms two types of sperms; half of the sperms have X without any sex chromosome. (A gamete without any sex chromosome is called **nullo gamete.**)

Female is XX, because it has two X chromosomes. It is homogametic, as it forms only one type of eggs. Every egg carries an X chromosome. Sex of the offspring depends on the kind of sperm that fertilizes the egg. If an X-carrying sperm fertilizes the egg, an XX female offspring is produced. If the nullo sperm fertilizes the egg, an XO male offspring is produced. Sex ratio between male and female offspring is 1:1.

XX – XY type or WZ – ZZ Type

This type of sex – determination pattern is common in birds, butterflies and moths. It is the reverse of XY – XX system. Here the female is heterogametic XY but the male is homogametic XX. Female produces two kinds of eggs X and Y in equal proportions. All sperms are alike, each carrying an X – chromosome. It is the kind of egg that determines the sex of offspring. When an X – carrying egg is fertilized by the sperm, a male offspring is produced, but when a Y – carrying egg is fertilized by the sperm a female offspring is produced. Sex ratio is 1:1.

Comparison of sex determination in *Drosophila* and Humans

Although both *Drosophila* and humans follow the same XY – XY sex determining pattern, yet there is a difference between the two. Presence of 'SRY' gene on Y chromosome is essential for triggering the development of maleness in humans. Absence of Y chromosome simply leads to the female development path. XO Turner's syndrome in humans produced through non-disjunction is a sterile female. But in *Drosophila* XO is a sterile male. Similarly XXY individual produced through non disjunction gametes in humans is a sterile male called Klinefelter's syndrome, but the same XXY set of chromosomes in *Drosophila* produces a fertile female.

There is a close genic balance between genes of different chromosomes. *Drosophila* has an X chromosome-autosome balance system. Its Y chromosome appears to have little influence on sex. Here actually the X chromosome is female determining and the autosomes are male determining. Sex of an individual depends more on the number of X chromosomes relative to the number of sets of autosomes. An X: A ratio of 1.00 or higher produces female whereas an X: A ratio of 0.5 or lower produces males.

Species	XX	XY	XO	XXY
Drosophila	♀	♂	♂	♀
Humans	♀	♂	♀	♂

Fig. 22.18 Comparison of sex determination in Drosophila and human

Genetic Problem 22.10: *Why are the chances for a husband and wife having either a boy or girl 50:50?*

Genetic Problem 22.11: *A woman produced four daughters in successive pregnancy. What are the prospects of her getting a son in the fifth pregnancy if it occurs?*

Genetic Problem 22.12: *If both parents are A^{+ve} and have an O^{-ve} child. Calculate the probability of A^{+ve} daughter and A^{+ve} son in next pregnancy?*

Genetic Traits (SRY gene) (Stand for + function)

Investigators have found that the Y chromosome has an SRY gene (sex-determining region Y gene). When this region in gene is lacking from the Y chromosome the individual is a female even though the chromosomal inheritance is XY. This gene is Y linked because it is found only on the Y chromosome

22.7 SEX LINKAGE

Traits controlled by alleles on the sex chromosomes are said to be **sex-linked** and the phenomenon is called **sex linkage**. An allele that is only on the X chromosome is **X-linked** and the allele that is only on the Y chromosome is **Y- linked**. Some traits are controlled by such genes which have alleles found on both X and Y, such traits are called **X-Y linked** or **pseudo autosomal** because unlike other sex linked traits they have just like autosomal mode of inheritance. Very few alleles have been found on the Y chromosome.

22.7.1 Sex Linkage in *Drosophila*

Consider, for example, the inheritance of eye colour in *Drosophila*, where females are XX and males are XY. Organisms having normal characteristics are called **wild types**. Organisms having not the normal characteristics are called **mutant**. The wild type *Drosophila* has bright red eyes. T.H. Morgan raised cultures of fruit fly *Drosophila* to study different traits.

First, Morgan crossed the white-eyed male to a red eyed female, about 1237 flies were produced in F_1 generation and all were red eyed. Now, would the white-eye trait reappear in the F_2 progeny as Mendel had predicted? In the F_2, there were 3470 red-eyed flies and 782 white-eyed flies. In this experiment he observed two deviations from what Mendel had explained. There was a slightly deviated phenotypic ratio in F_2 generation from standard 3:1 ratio of monohybrid cross and secondly that all the white-eyed flies in F_2 generation were male.

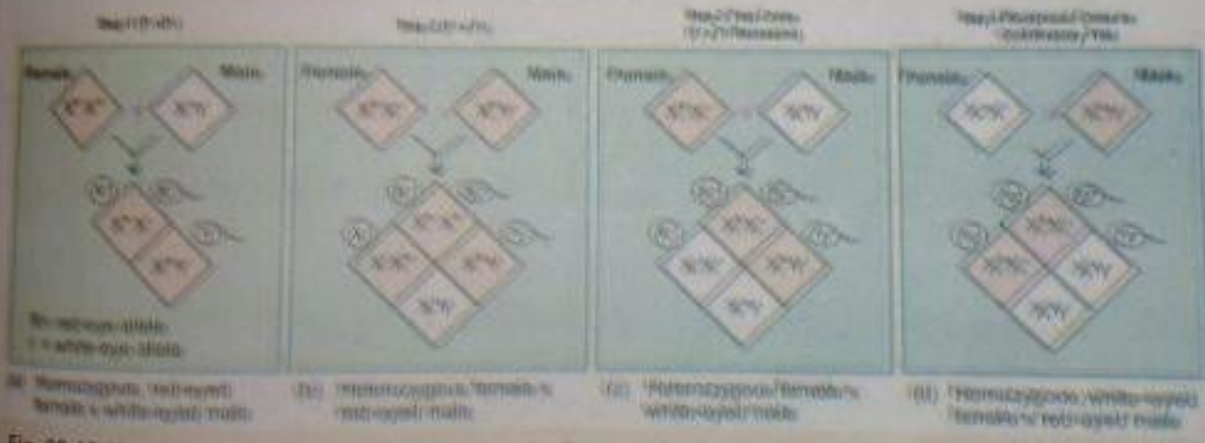

Fig. 22.19 Morgan's Experiments on eye colour in *Drosophila*. Letter R represents the dominant allele for red eye and letter r represents the recessive white eye. Because the alleles are carrier on the X chromosome, these are shown as superscript to the letter X. Thus the red eyed male fruit flies have genotype X^RY, white eyed males are X^rY. The Y chromosome does not have a gene locus for eye colours Therefore, the phenotype of the male results entirely from this single X linked genes. In the female X^RX^R and X^RX^r have red eyes and X^rX^r flies have white eyes.

Teacher's Point

The teacher would ask the students to:

- Trace the karyotype of a human being to observe and count the number and shape of chromosomes.
- Differentiate between autosomes and sex chromosomes from the karyotype (fig: 22.16).

Once he thought that perhaps the white-eyed trait somehow killed female flies preferentially? Morgan preferred a straightforward test: if any of the F_2 females carried the white-eye trait but did not show it, then it should be revealed by a test cross to the recessive parent. It was crossing red-eyed F_2 females back to the original white-eyed male; he obtained 129 red-eyed females, 132 red-eyed males, and 88 white-eyed females, 86 white-eyed males. Again, this was a rather poor fit to the expected 1:1:1:1 ratio due to a deficiency in recessives. The important thing, however, was that there were fully 88 white-eyed female flies. This observation finally discarded his doubt that the white-eyed trait can kill female flies.

Appearance of white eyed female provided an opportunity for a further confirmatory test. Morgan mated a white-eyed female with a red-eyed male. All female offspring had red eyes, and all male offspring had white eyes.

Morgan's conclusion

A simple explanation of his results, Morgan concluded that the white-eye trait gene resided on the X chromosome. Morgan had only to assume that the Y chromosome did not have this gene (it was later shown to carry almost no functional genes). Knowing from his previous crosses that white-eye is a recessive trait, the results he obtained could be seen to be a natural consequence of Mendelian segregation.

22.7.2 Sex – Linkage in Humans

Humans have many X – linked traits of which some like haemophilia and colour blindness are recessive while others like hypophosphatemic or vitamin D resistant rickets are dominant. X – linked dominant is a trait which is determined by an X linked dominant gene, while X – linked recessive is a trait that is determined by as X – linked recessive gene. Their patterns of inheritance are very different from each other.

Experimental mating is not practically possible in humans. Mode of inheritance of human traits can be traced through pedigrees.

X-Linked Recessive Inheritance

Females possessing one X-linked recessive mutation are considered carriers and will generally not manifest clinical symptoms of the disorder. All males possessing an X-linked recessive mutation will be affected (males have a single X-chromosome and therefore have only one copy of X-linked genes). All offspring of a carrier female have a 50% chance of inheriting the mutation. All female children of an affected father will be carriers (daughters possess their fathers' X-chromosome). No male children of an affected father will be affected (sons do not inherit their fathers' X-chromosome). *Examples*: Haemophilia A and B, Colour Blindness and Testicular feminization

X-Linked dominant inheritance

A male or female child of an affected mother has a 50% chance of inheriting the mutation and thus being affected with the disorder. All female children of an affected father will be affected (daughters possess their fathers' X-chromosome). No male children of an affected father will be affected (sons do not inherit their fathers' X-chromosome). *Examples:* Hypophosphatemic rickets.

Linked inheritance

Y linked traits are called **holandric traits**. The genes located on the Y chromosome, whose alleles are absent on the X chromosome are Y-linked genes or holandric genes (also hemizygous). The Y-linked genes are located only on the non-homologous region of Y chromosome. Their phenotypic expression is observed only in the males. They are inherited from father to son, as son only receives Y-chromosome from the father. The examples of Y-linked characters in man are: hypertrichosis (growth of hair on the rim of pinna), porcupine man (straight hair on the body) and webbing of toes. Recently two more genes have been discovered, testis determining factor (TDF) and minor histocompatibility gene (H-Y).

Fig. 22.20 Hairy Pinna

(formation of testis)

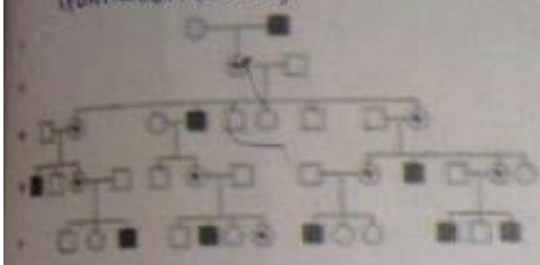

Fig. 22.21 Pedigree of recessive X linked trait

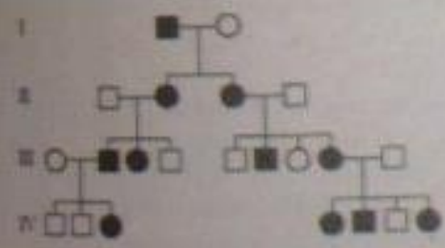

Fig. 22.22 Pedigree of dominant X linked trait

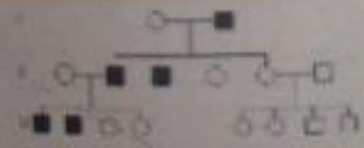

Fig. 22.21 Pedigree of Y-linked trait

Science Titbits

The study of inherited Mendelian traits in humans must rely on observations made while working with individual families. Classical cross fertilization breeding experiments as performed by Mendel are not allowed in humans. Human geneticists are not allowed to selectively breed for the traits they wish to study. One of most powerful tools in human genetic studies is pedigree analysis. When human geneticists first began to publish family studies, they used a variety of symbols and conventions. Now there are agreed upon standards for the construction of pedigrees.

22.7.3 Sex Linked Disorders in Human

Human have several disorders which are caused by the mutation in sex chromosomes as *Drosophila* has white eye colour. The inheritance patterns of some sex liked disorders in human are discussed here.

Genetics of haemophilia

Haemophilia is a rare X – linked recessive trait. Haemophiliac's blood fails to clot properly after an injury, because it has either a reduction or malfunction or complete absence of blood

clotting factors. It is a serious hereditary disease because a haemophiliac may bleed to death even from minor cuts. Haemophilia is of three types: A, B and C. Haemophilia A and B are X– linked recessive, but haemophilia C is an autosomal recessive trait. Being X – linked recessives, haemophilia A and B affect men more than women, but haemophilia C affects both the sexes equally because it is autosomal. Chances for a man to be affects by haemophilia A and B are double than a woman. A woman can suffer from haemophilia A or B only when she is homozygous for the recessive allele, but a man with just one recessive allele will display the trait. Haemophilia A and B zigzag from maternal grandfather through a carrier daughter to a grandson. It never passes direct from father to son. Gene for normal is H and gene for haemophilia A is h.

Genetic Problem 22.13: *Can two normal parents have haemophiliac child? Work out the probability.*

Table 22.6 Haemophilia

S.No	Types of Haemophilia	Cause	Percentage	Inheritance
1.	Haemophlia A	Abnormality of blood clotting factor VIII	80%	Recessive X linked
2.	Haemophilia B	Disturbance in blood clotting factor IX	20%	Recessive X linked
3.	Haemophlia C	Reduction in blood clotting factor XI	Less than 1%	Recessive autosomal

Science, Technology and Society Connections

Justify effectiveness of some the treatments of haemophilia

Cases of **mild haemophilia A**, can be treated using a type of medication called desmopressin. Desmopressin is a synthetic hormone. Desmopressin works by stimulating the production of clotting agent VIII and is usually given by injection. **Haemophilia B** which is caused by a lack of clotting agent IX is treated with a medication called nonacogulfan which is an engineered version of clotting agent IX. **Haemophilia A** is treated by using a synthetic version of clotting agent VIII , called octocogalfa, which is another type of genetically engineered purified protein.

How does blood clot?

Within seconds of cutting a blood vessel, the damaged tissue causes platelets to become 'sticky' and clump together around the cut. These 'activated' platelets and the damaged tissue release chemicals. These chemicals then react with other chemicals and proteins in the plasma, called clotting factors. Next to a cut a complex series of reactions involving these clotting factors then happens quickly. Each reaction triggers the next reaction. This is called a cascade. The final step of this cascade of chemical reactions is to convert factor I (also called fibrinogen - a soluble protein) into thin strands of a solid protein called fibrin. The strands of fibrin form a meshwork and trap blood cells which form into a solid clot. There are 13 known clotting factors which are called by their Roman numbers - factor I to factor XIII.

Teacher's Point

The teacher would ask the students to explain why are sex-linked disorders more common in males than females?

Genetics of colour-blindness

Normal trichromatic colour vision is based on three different kinds of cone in the retina, each sensitive to only one of the three primary colours, red, green or blue. Each type of cone cell has specific light absorbing proteins called **opsins**.

The genes for red and green opsins are on X chromosome, while the gene for blue opsin is present on autosome 7. Mutations in opsin genes cause colour-blindness like dichromacy and monochromacy.

Dichromacy: A dichromate can perceive two primary colours but is unable to perceive the one whose opsin is missing due to mutation. **Protanopia** is red blindness; **deuteronopia** is green blindness, while tritanopia is blue blindness.

Monochromacy: A monochromate can perceive only one colour. Monochromacy is true colour-blindness. Blue cone monochromacy is an X – linked recessive trait in which both red and green cone cells are absent. That is why it is also called red – green colour-blindness. It is a common hereditary disease.

Like any sex – linked recessive trait, it also zigzags from maternal grandfather through a carrier daughter to a grandson. It never passes direct from father to son. This type of colour-blindness is more common in men than women, because chances for a male to be affected by it are double than a female.

Fig. 22.22 Red/green colorblindness

Genetic Problem 22.14: *When a carrier women get married with normal man, what will be the probably colour blind son in their children?*

Genetics of muscular dystrophy

Muscular dystrophy, as the name implies, is characterized by a wasting away of the muscles. The most common form is **Duchenne muscular dystrophy**. It is a sex-linked recessive disorder. The symptoms appear in early childhood, when the child begins to have difficulty standing up and rises to a standing position in a characteristic way as shown in (figure 22.23). He is inevitably wheelchair bound by the age of 12. Eventually, he becomes severely wasted, and normal breathing becomes difficult. Death usually occurs by age of 20, therefore, affected males are rarely fathers. The recessive allele remains in the population by passage from carrier mother to carrier daughter. The gene whose mutation causes this disorder has been mapped. It codes for a protein called **dystrophin**, which is present in the normal muscle but

Fig. 22.23 Muscular dystrophy

missing in Duchenne patient. The lack of dystrophin causes calcium to leak into the cell, which promotes the action of an enzyme that dissolves muscle fibres.

22.7.4 Sex Related Traits 2(R)

Sex related traits are those which are associated with maleness or femaleness. These traits are not necessarily being sex linked. These traits may be controlled by sex linked or autosomal genes. They are of two different types i.e., sex limited traits and sex influenced traits.

Sex limited traits IMP (P.2) Differentiate b/w Sex limited traits and

A sex limited trait is a type of sex related trait which is confined to only one sex due to anatomical differences. Such traits affect a structure or function of the body present in only males or in only female. Example: Genes for milk yield in dairy cattle affect only cows. Similarly beard growth in human is limited to men. A woman does not grow a beard herself but she can pass the genes specifying heavy beard growth to her son.

Sex influenced trait IMP (P.2) Sex influenced trait.

Sex influenced traits are also the type of sex related traits. These occur in both males and females but they are more common in one sex. Such traits are controlled by an allele that is expressed as dominant in one but recessive in the other. This difference in expression is due to hormonal difference between the sexes. The amounts of body hair, muscle mass and male pattern balding are sex influenced traits. For example, many more men than women are bald. It is inherited as an autosomal dominant trait in males but as an autosomal recessive trait in females. A heterozygous male is bald but a heterozygous female is not. A woman can be bald only when she is homozygous recessive.

Table 22.7 Pattern baldness in humans

Genotype	Phenotype in male	Phenotype in female
B_1B_1	Bald	Bald
B_1B_2	Bald	Normal hairs
B_2B_2	Normal hairs	Normal hairs

Science, Technology and Society Connections

Suggest ways to save lives through the knowledge gained in this chapter.

Genetic research helps identify diseases and health problems that are more likely to be influenced by genetic factors as well as to assess the risk of a particular disease in an individual. These researches are known as genetic tests. When a genetic test confirms a high risk of certain condition, an expert in the field determines preventive measures to reduce the risk of that particular disease. Genetic testing is very reliable, however, it cannot tell you for sure whether you will develop a particular disease or not.

Teacher's Point

The teacher would ask the students to write on 'how to overcome human hereditary diseases'.

Science, Technology and Society Connections

Describe how the field of genetics has progressed to a more applied science.
With the discovery of DNA, the field of genetics has progressed as more applied science than just a theoretical subject. Many DNA based techniques are now widely used in various fields of life. Such as DNA sequencing technique, DNA fingerprinting, PCR etc. Genetic engineering or recombinant DNA technology is an applied field of genetics.

Science, Technology and Society Connections

Justify the effectiveness of some of the treatments of haemophilia.
Treatment for haemophilia today is very effective. The missing clotting factor is injected into the bloodstream using a needle. Bleeding stops when enough clotting factors reach the spot that is bleeding. With an adequate quantity of treatment products and proper care, people with haemophilia can live perfectly healthy lives. Without treatment, most children with severe haemophilia will die young. An estimated 400,000 people worldwide are living with haemophilia and only 25% receive adequate treatment. The World Federation of Haemophilia is striving to close this gap.

Exercise

M.C.Qs

1. Select the correct answer

(i) Mendel's law of independent assortment can be demonstrated by
(A) test cross (B) back cross
(C) monohybrid cross (D) dihybrid cross

(ii) Investigators do a dihybrid cross between two heterozygous and get about a 3:1 ratio among the offspring. The reason must be due to
(A) polygenes (B) pleiotropic genes
(C) linked genes (D) epistatic gene

(iii) Whether an allele is dominant or recessive depends on
(A) how common the allele is relative to other alleles
(B) whether it is inherited from the mother or the father
(C) which chromosome it is on
(D) whether it shows expression in heterozygous state or not

(iv) All the offspring of a white hen and a black rooster are grey. The simplest explanation for this pattern of inheritance is
(A) linkage (B) sex linkage
(C) independent assortment (D) incomplete dominance

(v) A man who has type B blood and a woman who has type A blood could have children of which of the following phenotypes?
(A) A or B only (B) AB only
(C) AB or O only (D) A, B, AB or O

(vi) A heterozygous red-eyed female *Drosophila* mated with a white-eyed male would produce
(A) red-eyed females and white males in the F1
(B) white eyed-females and red eyed males in the F1
(C) all white-eyed females and half red and half white eyed males in the F1
(D) half red and half white-eyed female as well as males in the F1

(vii) When mother's anti-Rh negative antibodies seep through placenta into blood circulation of foetus they start _____ of RBC of the foetus
(A) plasmolysis (B) crenation
(C) haemolysis (D) deplasmolysis

(viii) All chromosomes other than sex-chromosomes are called
(A) autosome (B) mesosome
(C) polysome (D) lysosome

(ix) If a man of M blood group marries a woman of N blood group all their children will have
(A) M blood group (B) N blood group
(C) O blood group (D) MN blood group

(x) Rh blood group system is encoded by three genes C, D and E which occupy------ tightly linked loci
(A) four (B) three
(C) five (D) two

(xi) Albinism is a recessive gene. A woman with albino father marries an albino man. The proportion of her progeny is:
(A) 2 normal: 1 albino (B) all normal
(C) all albino (D) 1normal:1 albino

(xii) Phenomena of an allele of one gene suppressing the activity of allele of another gene is called
(A) dominance (B) epistasis
(C) suppression (D) inactivation

(xiii) If red eyed (dominant) fly is mated with white eyed (recessive) fly, the ratio of red to white eyed in F_2 generation would be
(A) 3:1 (B) 2:2
(C) 2:1 (D) 1:3

(xiv) Blood group in human beings are controlled by
(A) 4 alleles in which A is dominant
(B) 3 alleles in which A and B are co-dominant and i is recessive
(C) 3 alleles in which none is dominant
(D) 3 alleles in which A is recessive

(xv) Genes located on same locus but having different expressions are
(A) multiple alleles (B) oncogenes
(C) polygenes (D) codominants

Short Questions

2. In monohybrid cross why did only about one fourth of Mendel's F_2 plants exhibit the recessive trait.
3. List the basic four principles of genetics that Mendel described in his experiment.
4. Hypothesize that in a dihybrid inheritance pattern of colour and texture of pea seed, the two traits are not interdependent.
5. What are the exceptions to the Mendel's laws of inheritance?
6. What is probability? How probability does relate to genetics?
7. Write the differences between incomplete dominance and co-dominance.
8. Describe the multiple alleles and state the alleles responsible for the traits of ABO blood groups.
9. Why can multiple alleles provide many different phenotypes for a trait?
10. Name various human blood group systems.
11. Describe the antigens of ABO blood group system.
12. Describe the antibodies of ABO blood group system.
13. Why Rh-incompatibility could be a danger to the developing foetus and mothers?
14. Investigate the reasons for O^- (negative) individuals as the Universal donor and AB^+ (positive) as the Universal recipient.
15. Why a recessive group allele "*i*" is more frequent in population.
16. List at least five polygenic traits discovered in humans.
17. Write the differences between dominance and epistasis.
18. What is gene linkage?
19. Explain crossing over.
20. Describe XX-XY type pattern of sex determination.
21. Why is human male referred as heterogametic?
22. What is sex linkage?
23. Describe Y-linked inheritance in man.
24. Name some of the sex-linked disorders of man and *Drosophila*.
25. How gene linkage encounters independent assortment?
26. Under what circumstances is it possible for father and son to suffer from haemophilia?
27. Explain dichromacy.
28. Explain monochromacy.
29. What is a sex limited trait?
30. What is sex influenced trait?
31. Describe how the field of genetics has progressed to a more applied science.

32. How do multiple alleles and polygenic traits differ?
33. Solve the following:
    (a) *Problem*: Show how the "test cross" works, using round-seeded pea of unknown generation.
    (b) *Problem*: if a four-o'clock plant having red flowers is crossed with a white- flowered four o'clock, the F_1 plants are all pink flowered. From this evidence alone, what ratios would you expect in these crosses: red crossed with red? red X pink? pink X white? white X white?
    (c) *Problem*: Suppose a man with blood type A marries a woman who has type AB blood. What blood types would you expect to find among their children? In this problem we have no way of knowing whether the man is homozygous or heterozygous for I^A allele. What would tell you which of the genotype he has?
    (d) *Problem*: Two white sheep produce a black offspring. What is the parent's genotype for the colour? What is the probability that their next offspring will be black?
    (e) *Problem*: Albinism (lack of pigment) in man is caused by a recessive gene. If normal parents have an albino child, what is the probability that their next child will be normal for colour?
34. Define/ Describe/Explain briefly:
    heredity, variations, genetics, true breeding plants, Mendelian parameters, hybridization, Mendel's law of segregation, Mendel's law of independent assortment , probability, products rule, incomplete dominance , co-dominance , alleles, multiple alleles, rare blood type, antigen, antibodies, agglutinin, agglutination , universal recipient , universal donor, erythroblastosis foetalis, bilirubin, polygene, mulatto, duplicate recessive epistasis, gene linkage, crossing over , recombinant frequency, karyotype, amniocentesis , nullo gamete, wild type, mutant, holandric traits, opsin, dichromacy, protonopia, deuteronopia, monochromacy
35. Write the differences between:
    (a) genes and alleles
    (b) alleles and multiple alleles
    (c) monohybrid and dihybrid cross
    (d) first and second filial generation
    (e) genotype and phenotype
    (f) homozygous and heterozygous,
    (g) homozygous and hemizygous
    (h) incomplete dominance and co-dominance
    (i) dominance and epistasis
    (j) sex determination in *Drosophila* and human
    (k) X-linked and Y-linked inheritance
    (l) sex limited traits and sex influenced traits
    (m) pattern of baldness in human male and female.

Extensive Questions

36. How inheritance can be associated with the laws of Mendel?
37. Explain Mendel's law of inheritance of monohybrid cross and interpret the results.
38. Describe law of independent assortment under the following heads:
    (a) Limitations law of independent assortment
    (b) Usefulness law of independent assortment
    (c) Scope of law of independent assortment
39. What is probability? How can you evaluate that the inheritance of genes and their mixing during fertilization is based on mathematical probabilities?
40. Explain incomplete dominance with reference to the inheritance of flower in 4 O'clock plant.
41. Give a detailed account of ABO blood group system.
42. (a) Associate the positive and negative blood groups with the presence and absence of Rh factor.
    (b) Explain *Erythroblastosis foetalis* in the light of antigen-antibody reaction.
43. Suggests prevention and treatment of *Erythroblastosis foetalis*.
44. Explain polygenic inheritance with reference to wheat grain colour.
45. Explain polygenic inheritance with reference to inheritance of human skin colour.
46. What is epistasis? Explain relationship of epistasis with polygenic inheritance.
47. Explain epistasis with reference to the inheritance of coat colour in *Labrador retriever*.
48. Explain epistasis with reference to the inheritance of flower colour (pigment phenotype) of sweet pea, *Lathyrus odoratus*.
49. What is gene linkage? How gene linkage can be detected? Explain Morgan's experiment. Suggest why linkage could be observed or evaluated if the number of progeny is quite large.
50. Explain Morgan's experiment to demonstrate the role of crossing over in gene linkage.
51. Explain genetic identification of sex phenotypes in *Drosophila* and man.
52. Explain the technique employed for embryonic screening.
53. Describe: (a) XY –XX (b) X – XO (c) WZ-ZZ sex determination system.
54. Explain sex linkage in *Drosophila*.
55. Explain sex linkage in man.
56. Explain genetics of haemophiulia.
57. Explain genetics of colour blindness.
58. Explain genetics of muscular dystrophy.
59. Describe sex related traits.