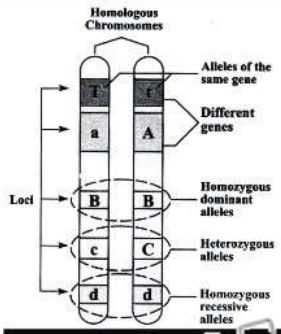


Chapter 22

Variation and Genetics

22.1 GENES, ALLELES AND GENE POOL

Gene

Gene is the basic unit of biological information.

These are actually parts of DNA comprising its base sequences.

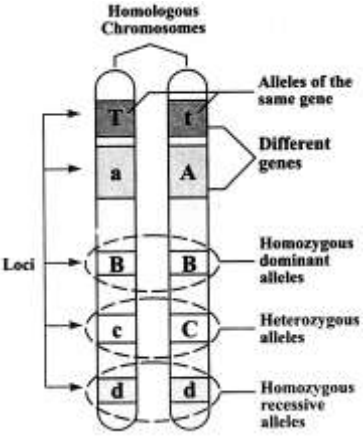


Fig. 22.1 Allelic pairs on a homologous pair of chromosomes

Characteristics of Gene

(i) The position of a gene on the chromosome is called its locus.

(ii) Hereditary characteristics pass from parents to offspring through genes in their gametes.

(iii) Genes are responsible for producing startling inherited resemblances as well as distinctive variations among generations.

(iv) The genes are passed to the next generations as intact parental combination between generations.

(v) Sometimes, variation are produced due to shuffling, mutation or juggling in gene.

(vi) Genes form pairs on pairs of homologous chromosomes.

(vii) One member of a gene pair is located on one homologue, and the other member on other homologue.

Allele

Partners of a gene pair are called alleles.

Characteristics of Allele

(i) Each allele of a gene pair occupies the same gene locus on its respective homologue.

(ii) Both alleles on one locus, may be identical or different from each other.

Phenotype and Genotype

Definitions

(i) Phenotype is the form of appearance of a trait.

(ii) Genotype is the genetic complement for a particular trait in an individual.

Jumping genes do not settle peacefully on their loci, they keep on hopping on different loci on the same chromosome or other chromosomes.

Example

A flower may be red or white in colour. Flower colour is trait and red and white are its two phenotypes. Red is represented by 'R' and white by 'r'.

Population

Any group of interbreeding organisms of the same species that exist together in both time and space is called a population.

Gene Pool

All the genes/alleles found in a breeding population (group of sexually interbreeding organism of same species that exist together in both time and space) at a given time are collectively called the gene pool. It is the total genetic information encoded in the total genes in a breeding population existing at a given time.

Beanbag Genetics

If we imagine population not as a group of individuals, but as a group of individually segregating and randomly assorting alleles, we can understand the concept of "beanbag genetics"

The alleles are like beans in a beanbag. The entire beanbag full of beans is the gene pool of the population.

In the beanbag approach we can imagine the entire gene pool comprising all the alleles for all the different traits at once, or we can just focus on some subset, such as all the alleles for a single trait.

For convenience, we can focus on the gene pool for a single particular trait. A sample population of 100 diploid plants, some of which bear red flowers, others bearing white flowers has a sum total of 200 of all the different alleles (R or r) for flower colour trait as its gene pool.

QUESTIONS RELATED TO ABOVE ARTICLE

Define gene pool. Explain the concept of gene pool in a same population.

(Exercise Question xv)

22.2 MENDEL'S LAWS OF INHERITANCE

Introduction

Gregor Johann Mendel (1822-1884) laid the foundation of classical genetics by formulating two laws of heredity (law of segregation and law of independent assortment). He was a priest. He performed series of breeding experiments on garden pea (*Pisum sativum*) in his monastery garden for eleven years (1854-1865).

Reasons for Selection of Pea Plant

Mendel selected *Pisum sativum* for his experiment due to following reasons:

i) It is easy to cultivate and grows well in garden.

ii) Its flowers are hermaphrodite and self-fertilizes but can also be cross-fertilized.

iii) Time gap between generations was very short so many generations can be grown in short period of time.

iv) It has sharply distinct traits. Each trait has two clear cut alternative forms or varieties. E.g. seed shape had round or wrinkled phenotype.

Contrasting Pair of Traits

Trait	Dominant	Recessive
Plant Height	Tall (6–7 feet)	Short (9–18 inches)
Flower colour	Purple	White
Flower Position	At leaf junctions (axial)	At tips of branches (terminal)
Pod Colour	Green	Yellow
Pod Shape	Inflated	Constricted
Seed Colour	Yellow	Green
Seed Shape	Round	Wrinkled

Fig. 22.2 Seven traits of garden pea studied by Mendel

22.2.1 Mendel's Law Of Segregation

STATEMENT

According to Mendel's 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."

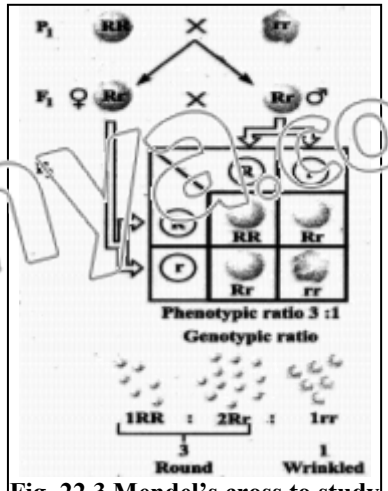


Fig. 22.3 Mendel's cross to study single trait inheritance in pea.

EXPLANATION

Mendel first established true-breeding lines or varieties for each trait.

A ***true-breeding*** *variety* upon self-fertilization always produced offspring identical to the parents. For example a true-breeding round seed plant produces only round seeds. Similarly, a true breeding wrinkled seed plant produces only wrinkled seeds.

Mendel studied one trait at time to develop law of segregation.

Such a cross in which only one trait is studied at a time is called *monohybrid cross*.

Phenotypic Expression

Different steps performed by Mendel to develop law of segregation are described in sequence as following

P_1 Cross

He cross-fertilized a true breeding round seed male plant with a true breeding wrinkled seed female plant.

He called it **first parental** generation (P1). Their offspring were called F_1 or **first filial** generation. All F_1 offspring were round like one of the parent.

Round dominated wrinkled.

Its dominance was complete because no offspring intermediate between parents was found. He called the trait that appeared in F_1 as dominant while the trait, which was masked as recessive.

F_1 Cross

Mendel allowed self-fertilization among F_1 monohybrids to raise F_2 progeny. As a result of monohybrid cross, 3/4 of F_2 were round and 1/4 wrinkled. Mendel got similar result and same 3:1 ratio in offspring of monohybrid cross for all seven contrasting pairs of traits.

F_2 Cross

He self-fertilized F_2 plants to raise F_3. He noted that 1/3 of F_2 round produced only round, while 2/3 of F_2 round produced both round and wrinkled in 3:1 ratio but F_2 wrinkled produced only wrinkled. He concluded that 1/3 of F_2 rounds were true-breeding like P_1 round and 2/3 of F_2 round were monohybrid like F_1 round.

Mendel's Interpretations

Mendel proposed

- Each contrasting form of a trait e.g. roundness or wrinkledness of seed was determined by particulate hereditary factors, which he called ***'elementen'***.
- These factors carrying hereditary information were transmitted from parents to offspring through gametes.
- Each pea plant had a pair of these factors, one derived from male parent and other from female parent. Both of these factors together controlled expression of a trait.

Genetic Expression

Mendel designated dominant factor with a capital letter and recessive factor with a small letter e.g. 'R' for roundness factor and 'r' for wrinkledness factor.

Johannsen renamed them as ***'gene'***.

The true-breeding round seed plant of P_1 generation carried 'RR' alleles while true-breeding wrinkled seed plant of P_1 carried 'rr' alleles.

When both the alleles of a gene pair in an organism are same, the organism is ***homozygous*** for that gene pair. An individual with a homozygous genotype is a ***homozygote***.

Mendel inferred that each factor separated from pair so that a gamete received only one member. When male gamete carrying factor (R) fertilized female gamete with factor (r), the complete set of the two factors (Rr) for the trait was restored in zygote. The zygote developed into F_1 offspring that was heterozygous 'Rr'.

When two alleles of a gene pair are different then it is called ***heterozygous*** and such an individual with a heterozygous genotype is called ***heterozygote***.

F_1 offspring (Rr) a monohybrid for seed shape was round in phenotype but heterozygous in genotype. Its alleles also segregated during gamete formation.

Punnett square indicates that 1/4 of F_2 progeny would have been 'RR' (homozygous round), 2/4 'Rr' (heterozygous round) and 1/4 'rr' (wrinkled).

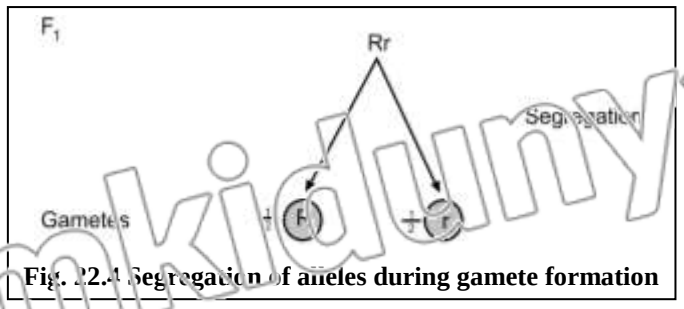


Fig. 22.4 Segregation of alleles during gamete formation

Conclusion

All these results support Mendel's law of segregation i.e.

- Alleles are present in form of pairs in an individual.
- Alleles separate during gamete formation from one another.
- Each gamete receives only one member of the pair.
- Actual number is restored after fertilization when gametes fuse to form zygote.

Further

- Phenotype ratio of Mendel's law of segregation is 3:1 ratio.
- Genotype ratio of Mendel's law of segregation is 1:2:1

QUESTIONS RELATED TO ABOVE ARTICLE

Define and explain Mendel's law of Segregation?

Explain with example Mendel's law of segregation. (SWL 2021, MTN 2022)

Define Mendel's law of segregation. Explain it with an example. (Exercise question ii)

Test Cross

Definition

Test cross is a mating in which an individual showing a dominant phenotype is crossed with an individual showing a recessive phenotype.

Importance

It is used to find genotype (homozygosity or heterozygosity) of phenotypically dominant parent.

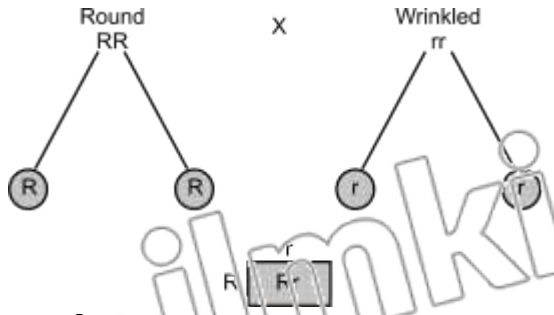
Explanation

This cross was used by Mendel to find genotype of individual.

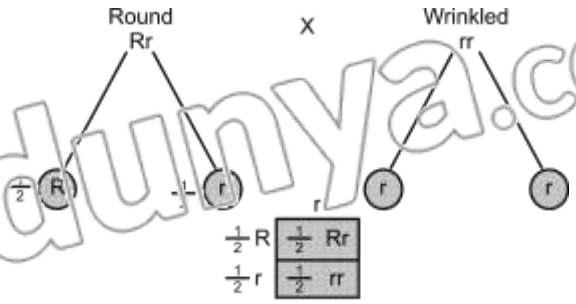
A phenotypically round seed could be homozygous (RR) or heterozygous (Rr).

- If phenotypically round seed plant is crossed with homozygous recessive and all progeny is of round seed then tested phenotypically dominant individual is homozygous.
- 1/2 round seed and 1/2 wrinkled seed progeny then tested phenotypically dominant individual is heterozygous.

Case I	Case II
If the seed is homozygous round (RR), it will grow into a pea plant that forms all gametes with only 'R' allele. Wrinkled seed plant is always homozygous recessive. It will form all gametes with 'r' allele. Fertilization will result in 100% round seed progeny.	If the seed is heterozygous round (Rr), it will grow into a plant that forms half the gametes with 'R' and half with 'r' allele. Wrinkled seed plant will form only 'r' type of gametes. Fertilization will result into 50% round and 50% wrinkled seed progeny. Even a single wrinkled seed in the progeny is a convincing proof for heterozygous nature of the round parent.



Result
All round seed progeny.
The tested phenotypically dominant individual is homozygous.



Result
½ round seed and
½ wrinkled seed progeny.
The tested phenotypically dominant individual is heterozygous.

Fig. 22.5 Test cross of a round seed

QUESTIONS RELATED TO ABOVE ARTICLE

Define and explain test cross.

Define test cross. Explain it with example. **(GRW 2017, LHR 2019)**

22.2.2 Mendel's Law Of Independent Assortment

Statement

According to Mendel's law of independent assortment

"When two contrasting pairs of traits are followed in the same cross, their alleles assort independently into gametes."

Proof of Law of Independent Assortment by Dihybrid Cross

During these experiments, Mendel decided to study the inheritance of two traits simultaneously.

- He crossed true-breeding round and yellow seed plants with true-breeding wrinkled and green seed plants. All the F_1 **dihybrids** were round yellow seeded due to dominance.
- Then he made a dihybrid cross by allowing self-fertilization among F_1 dihybrids. In F_2 alongwith **two parental combinations** (round yellow and wrinkled green), two new phenotypic combinations (round green and wrinkled yellow) were also found. A clear cut 9:3:3:1 phenotypic ratio was found in F_2.

Conclusion

From appearance of these new recombinant phenotypes of F_2, Mendel inferred that

- Some sort of shuffling of alleles had occurred during gamete formation.
- He called this mechanism as independent assortment of alleles into gametes.
- He concluded that the alleles for seed shape and colour were not bound to remain in parental combinations forever i.e. 'R' with 'Y' and 'r' with 'y' rather these were free to assort independently.
- The distribution of alleles of one trait into gametes has no influence on the distribution of alleles of the other trait. Thus the chance for a plant to be round or wrinkled is independent of its chance of being yellow or green.

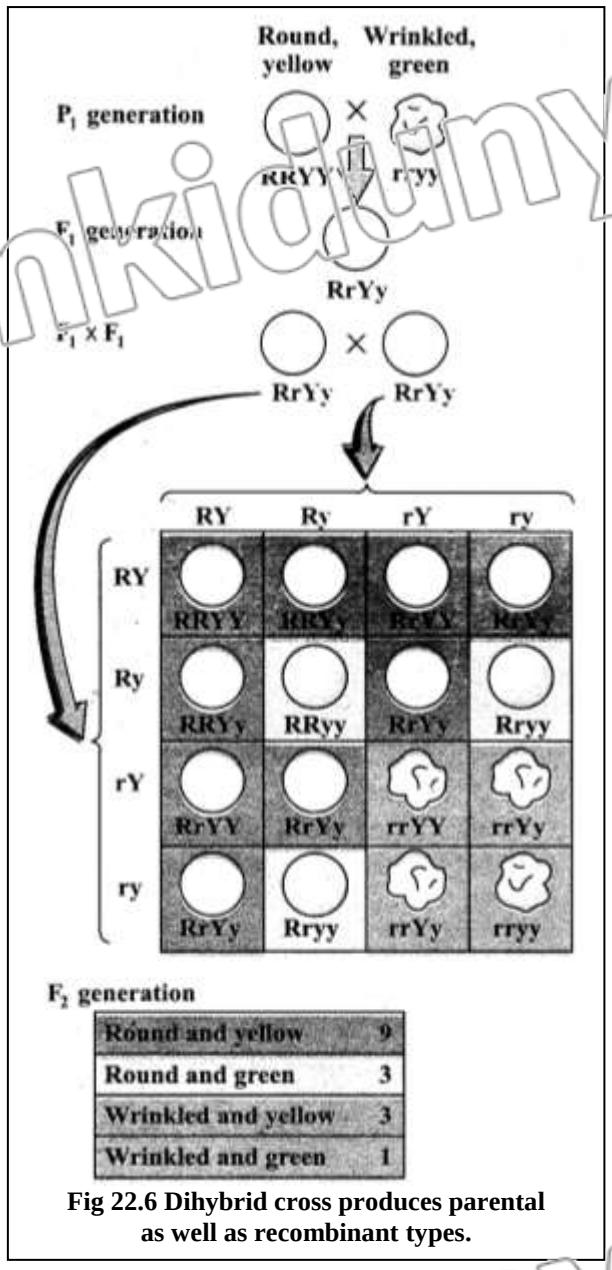


Fig 22.6 Dihybrid cross produces parental as well as recombinant types.

PROBABILITY AND PRODUCT RULE

Probability is chance of an event to occur.

Two independent events occur during law of independent assortment

(i) In F_2 offspring of a monohybrid cross the independent chance for a seed to be round is 3/4 or it to be wrinkled is 1/4.

(ii) Inheritance of seed colour is another separate event. The independent chance in F_2 of a monohybrid cross for a seed to be yellow is 3/4 or it to be green is 1/4.

When two independent events are occurring simultaneously like in dihybrid cross, the ratio of each joint phenotypic combination can be obtained by multiplying the probabilities of individual phenotypes. It is called product rule. ***The product rule state that:***

"The joint probability that both of the independent events will occur simultaneously is equal to the product of individual probabilities of each event."

Event No. 1	Event No. 2	Both events at a time
Seed shape	**Seed colour**	**Seed shape and colour**
Independent probability to be	Independent probability to be	Joint probability of being
Round = ¾	Yellow = ¾	Round yellow = ¾ x ¾ = 9/16
Round = ¾	Green = ¼	Round green = ¾ x ¼ = 3/16
Wrinkled = ¼	Yellow = ¾	Wrinkled yellow = ¼ x ¾ = 3/16
Wrinkled = ¼	Green = ¼	Wrinkled green = ¼ x ¼ = 1/16

Limitation of Law of Independent Assortment

Independent assortment of genes depends upon independent assortment of their chromosome. All the genes present on a homologous pair of chromosomes are linked to each other in the form of a linkage group. These cannot assort independently.

Only those contrasting pairs of traits can assort independently whose alleles are riding on non-homologous chromosomes.

Pea has seven homologous pairs of chromosomes. The allelic pair for each of the seven characters studied by Mendel were luckily on different homologous pair of chromosomes. If he had studied an eighth character, its alleles would have been linked to alleles of another trait on the same homologous pair and could have never assorted independently.

Rediscovery of Mendel's Work

Mendel presented his findings to Brunn Society for the study of Natural Sciences in 1865. His work lay neglected for 34 years. In 1900, 16 years after his death three botanist; Correns, De Varies and Tschermach independently rediscovered and acknowledged his work.

Activity

Normal individuals have melanin pigment in their skin, hair and eyes. Albinos totally lack pigment in their bodies. Albinism is a recessive trait in humans. Two normal parents have an albino child. What is the probability that their next child will also be an albino?

Solution

Normal allele is represented by "N" and recessive alleles for albino is represented by "n".

Normal person = NN, Nn

Albino person = nn.

As the both of the parent are normal, so their genotype may be NN or Nn. In case of NN, they cannot produce a recessive character. So their genotype is Nn.

According to the law of segregation, following cross takes place

Father X **Mother**

Nn Nn

Gametes

	N	n
N	NN	Nn
n	Nn	nn

Progeny

It gives 3:1 ratio according to law of segregation.

Conclusion

The result show that ¼ of the progeny of the above parent will be albino while ¾ will be normal.

QUESTIONS RELATED TO ABOVE ARTICLE

Describe the law of independent assortment with an example. (LHR 2017)

Describe the Mendel's law of independent assortment with an example. (Exercise Question iii)

Define probability. Derive 9:3:3:1 F_2 ratio of independent assortment through product rule. (Exercise Question iv)

22.3 DOMINANCE RELATIONS

DEFINITION

Dominance is a physiological effect of an allele over its partner allele on the same gene locus.

TYPES

There are four types of dominance relations among alleles. Each relation shows a different style of functional effect on each other.

1. Complete dominance
2. Incomplete dominance
3. Codominance
4. Over dominance

22.3.1 Complete Dominance

Definition

When one allele (R) is completely dominant over the other (r), presence of the recessive allele is functionally hidden so that heterozygote (Rr) has the same round phenotype as (RR) homozygote then it is called complete dominance.

Example

The contrasting pairs of alleles for all the seven characters chosen by Mendel are examples of complete dominance.

22.3.2 Incomplete Dominance

Definition

When the phenotype of the heterozygote is intermediate between phenotypes of the two homozygotes, it is called incomplete or partial dominance.

It was first described by Carl Correns in 1899 by working on a flowering plant named 4 O' clock.

Features

- There is no true dominant allele, the usual capital and small letter distinction for dominant and recessive is not used.
- Both the alleles are represented by same letter but are numbered differently.
- Phenotype ratio is the same as the genotype ratio so there is no need of test cross.

Example

Cross-I

When a true breeding red flowered 4 O' clock plant was crossed with a true breeding white flowered plant, in F_1 hybrids, all had pink flowers having a shade intermediate between those of the parents due to an intermediate amount of pigment in petals.

Cross--II

When Correns self-fertilized F_1 pink, the F_2 showed all three phenotypes of flowers in the ratio of 1 red 2 pink 1 white.

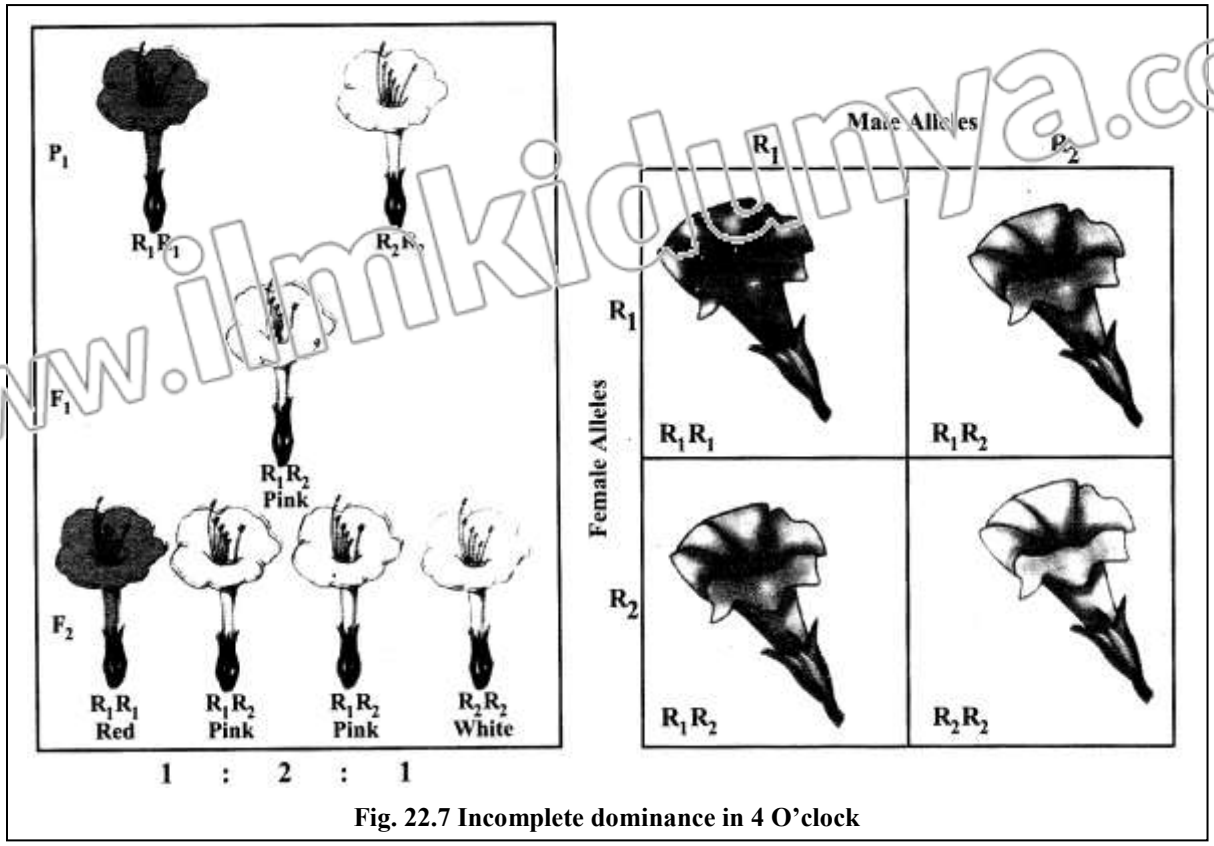


Fig. 22.7 Incomplete dominance in 4 O'clock

QUESTIONS RELATED TO ABOVE ARTICLE

Write a note on incomplete dominance. **(GRW 2018, MTN 2019)**

What is incomplete dominance? Explain with examples. **(BWP 2021)**

What is incomplete dominance? Explain with examples. **(Exercise Questions i)**

22.3.3 Codominance

Definition

Codominance occur when both the alleles express independently in heterozygote (A_1A_2) and form their respective products X and Y the codominant heterozygote would have both substances at the same time such alleles are called codominant alleles.

Different alleles of a gene that are both expressed in a heterozygous condition are called codominant.

Example

MN blood group is the most suitable example of codominance as discussed below.

MN BLOOD GROUP SYSTEM

Discovery

MN blood types in man were discovered by Landsteiner and Levine on the basis of specific antigens present on RBC.

Blood Types

There are three general phenotypes i.e. M, N and MN.

- M phenotype has antigen M that is produced by gene L^M.
- N phenotype has antigen N that is produced by allele L^N.
- MN phenotype has both M and N antigen, simultaneously produced by their alleles L^M and L^N.

PHENOTYPE	GENOTYPE	ANTIGEN ON RBC
M	L^ML^M	M
N	L^NL^N	N
MN	L^ML^N	M and N

If a man of M blood group marries a woman of N blood group, all their children will have MN blood group.

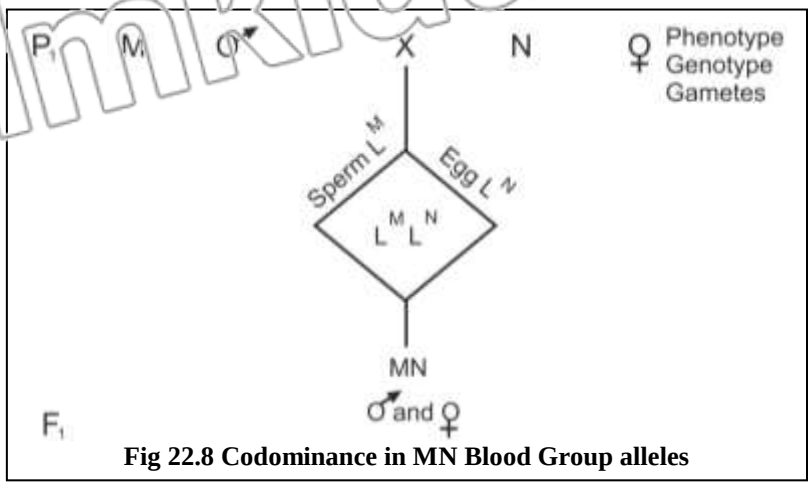


Fig 22.8 Codominance in MN Blood Group alleles

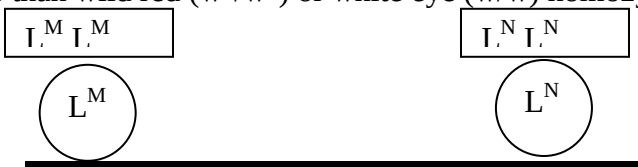
22.3.4 Over Dominance

Definition

Such type of dominance in which phenotype of heterozygote exceeds over phenotypic expression of both the homozygotes is called over dominance.

Example

In fruit fly *Drosophila*, the heterozygote (w+/w) has more quantity of fluorescent pigments in eyes than wild red (w^+/w^+) or white eye (w/w) homozygotes.



QUESTIONS RELATED TO ABOVE ARTICLE

What is incomplete dominance? Explain it with an example.

Explain codominance with the help of MN blood group system in man. (GRW 2021)

What is codominance? Explain the phenomenon of codominance with an example. (Exercise Question v)

22.4. MULTIPLE ALLELES

Definition

All such altered alternate forms of a gene, whose number is more than two, are called multiple alleles.

Features

i) Gene mutations may produce many different alleles of a gene.

ii) Some gene may have as many as 300 alleles.

iii) Any two of these multiple alleles can be present in the genome of a diploid organism.

iv) A haploid organism or a gamete can have just one of them in its genome.

Example

The ABO blood group system and Rh blood group system in man are the most suitable examples of the multiple alleles.

22.4.1 ABO – The First Discovered Multiple Allelic Blood Group System in Man

Discovery

ABO blood group system was first discovered by Karl Landsteiner in 1901.

Phenotypes of ABO system

ABO system has four different phenotypes, which are distinct from each other on the basis of specific antigen on the surface of RBC.

(i) **Blood group A** It has antigen A.

(ii) **Blood group B** It has antigen B.

(iii) **Blood group AB** It has both antigens A and B.

(iv) **Blood group O** It has neither antigen A nor B.

Genetics of ABO system

Bernstein explained the genetic basis of ABO system in 1925.

ABO blood group system is encoded by a single polymorphic (with many forms) gene I on chromosome 9. It has three multiple alleles I^A, I^B and i.

Antigens in the blood

(i) Allele I^A specifies production of antigen A.

(ii) Allele I^B specifies production of antigen B.

(iii) Allele i does not specify any antigen.

Dominance Relations

Following are dominance relations

i) Alleles 1^A and I^B are codominant to each other. So they are expressed equally in I^AI^B heterozygote to produce AB phenotype.

ii) Allele i is recessive to both I^A and I^B.

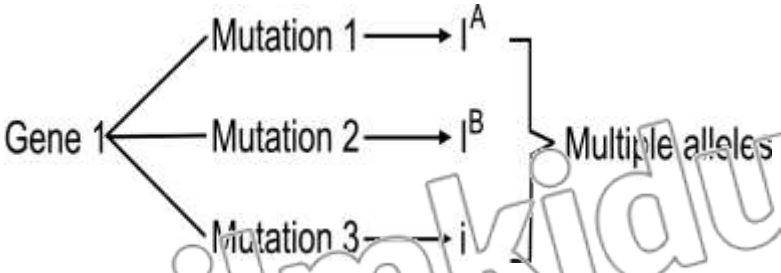
iii) Therefore, I^AI^A or I^A i genotypes will produce phenotype A.

iv) Similarly, I^BI^B or I^B i produces phenotype B.

v) The homozygous or I^B I^B produces phenotype B.

vi) The homozygous ii will produce phenotype O.

Blood group 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.



A and B antigens can also be present in saliva and other body fluids of some persons called secretors. Secretors have dominant secretor gene 'Se' on chromosome 19.

BLOOD GROUP	GENES	COMBINATION	ANTIGEN	ANTIBODIES
A	I^AI^A	Homozygous	A	Anti-B
	I^Ai	Heterozygous	A	Anti-B
B	I^BI^B	Homozygous	B	Anti-A
	I^Bi	Heterozygous	B	Anti-A
AB	I^AI^B	Heterozygous	A & B	No
O	ii	Homozygous	No	Anti-A & Anti-B

Antibodies in ABO system

Anti-A and Anti-B antibodies appear in plasma during the first few months after birth. They are naturally occurring in the absence of corresponding antigen.

The blood serum containing antibodies is called ***antiserum***.

1. The blood serum of A phenotype contains anti-B antibodies.

2. The blood serum of B phenotype contains anti-A antibodies.

3. The blood serum of AB phenotype has neither anti-A nor anti-B antibodies.

4. The blood serum of O phenotype contains both anti-A and anti-B antibodies.

Importance

1) **Blood Transfusion**

- **Blood group A** can be transfused only into A and AB recipients because they do not have anti-A antibodies.
- **Blood group B** can be transferred only into B and AB recipients as they do not have anti-B antibodies.
- **Blood group AB** can be transfused only into AB recipients because they have neither anti-A nor anti-B antibodies.
- **Blood group O** can be transfused into all other phenotypes.

Donor's antibodies are quickly absorbed by other tissues or greatly diluted in recipient's blood stream.

- O blood group individuals are called **universal donors**.
- AB blood group individuals are called **universal recipients**.

The blood samples of donor and the recipient are cross-matched for compatibility before giving transfusion. If incompatible blood is transfused, then it may cause;

i) Agglutination

It leads to serious results because clumped cells cannot pass through fine capillaries.

ii) Hemolysis

In it either the antibodies of the recipient destroy the RBC of donor or the antibodies of the donor hemolyze the RBC of the recipient.

2) Determining Paternity

Genetic analysis of blood groups helps in solving cases of disputed parentage but does not provide sufficient evidence. It can only be used to prove that an individual is not the parent of a particular child e.g.

i) A child with phenotype AB cannot be child of a parent of phenotype O.

ii) A man of B phenotype cannot be father of a blood type A child whose mother is of phenotype O. His father could either be A or AB phenotype.

Blood type data is not sufficient evidence for disputed paternity. Most modern DNA finger printing test is more seriously considered for a legal decision.

Activity

Two new born babies get mixed up in the nursery of a hospital. Baby I is type B and baby II is of type O.

Determine their parentage from the phenotypes of these two couples. Mr. Haris is type A and Mrs. Haris is type AB. Mr. and Mrs. Bilal are both of type A.

Solution

Baby	Blood group	Possible genotype
Baby I	B	I^BI^B,I^Bi
Baby II	O	ii

The data of parents shows that;

Parent	Blood group	Possible genotype
Mr. Haris	A	I^AI^A, I^Ai
Mrs. Haris	AB	I^AI^B
Mr. Bilal	A	I^AI^A, I^Ai
Mrs. Bilal	A	I^AI^A, I^Ai

1. **Cross between Mr. Haris and Mrs. Harris**

Mr. Haris I^AI^A/ I^Ai X Mrs. Haris I^AI^B

Gametes I^A, i I^A, I^B

Progeny I^AI^A,I^Ai, I^Bi, I^AI^B

Children can be with blood groups A, B and AB but not with O.

2. **Cross between Mr. and Mrs. Bilal**

Mr. Bilal I^AI^A/ I^Ai X Mrs. Bilal I^AI^A/ I^Ai

Gametes I^A, i I^A, i

Progeny I^AI^A, I^Ai, ii

The cross shows children can be with blood group A or O but cannot be B or AB.

Result

These crosses show that Baby I (with blood group B) belongs to Mr. and Mrs. Haris while Baby II (with blood group O) belongs to Mr. and Mrs. Bilal.

22.4.2 Rh Blood Group System

Introduction

ABO blood type is further differentiated by a + or – sign. This positive or negative sign refers to the presence or absence of another blood group system antigen called **Rh factor.** Rh Blood group system is defined on the basis of Rh factor present on the surface of RBC. This system is named Rh after Rhesus monkey, because its antigen was first discovered in it by Landsteiner in 1930s.

Genetics of Rh System

It is encoded by three genes C, D and E. They occupy two tightly linked loci. Alleles of gene D occupy one locus called locus D while genes C and E alternatively occupy the other locus. The D locus is of prime importance.

Gene D has two alleles i.e. D and d. D is completely dominant over d.

Phenotypes of Rh System

- Persons having genotype DD or Dd have Rh factor on their RBC and are Rh^+.
- Persons with genotype dd do not have Rh factor and are Rh^-.

Antibodies of Rh System

Antibodies are not present either in Rh^+ or Rh^- persons. Anti–Rh antibody production requires a stimulus by the human Rh antigen itself.

An Rh–person does not produce anti–Rh antibodies unless he is exposed to Rh antigen.

Blood Transfusion

Rh^- donor is totally incompatible for Rh recipient. If an Rh person receives Rh antigen through wrong Rh^+ blood transfusion, he will begin to produce anti-Rh antibodies against Rh antigens. Rh blood, clear of any anti Rh antibody from a donor who has never been exposed to Rh antigen, can be transfused to Rh^+ recipient.

22.4.2 a. Erythroblastosis Foetalis (Maternal-Foetal Rh Incompatibility)

Introduction

It results when an Rh^- woman, married to an Rh^+ man, conceives a child who is Rh^+.

Chances

- If the man's genotype is DD, all of their offsprings (Dd) will be Rh^+.
- If the man's genotype is Dd, half of their offspring with Dd genotype will be Rh+.

Mechanism

Different steps involved are

i) RBC of Rh^+ foetus cross the placental barrier and enter into Rh^- mother's blood stream, the mother's immune system reacts to the foetal Rh antigen stimulus by producing a large number of anti-Rh antibodies.

ii) Mother's anti-Rh antibodies seep through placenta into blood circulation of foetus, they start hemolysis (brake down/busting) of RBC of foetus.

iii) Due to destruction of RBC, foetus becomes anaemic.

iv) Anaemic foetus starts to release many immature erythroblasts into his blood stream. That is why this disease is called erythroblastosis foetalis.

v) Anaemia may lead to abortion or still birth.

vi) If pregnancy continues, the liver and spleen of the foetus swell as they rapidly produce RBC.

vii) The breakdown product of RBC called bilirubin also accumulates in foetus. Bilirubin damages his brain cells and turns his skin and whites of eye yellow. This condition is called **jaundice**. If such baby is born alive, it may suffer from severe hemolytic anaemia and jaundice. Such baby's blood should be immediately replaced by Rh^- blood free of anti-Rh antibodies.

High Risk of Incompatibility after First Pregnancy

The first Rh incompatible pregnancy may not face much problems if very few of foetal antigens cross placenta into maternal circulation and the amount of maternal antibody production is not very high. But when placenta detaches at birth, a large number of foetal cells enter mother's blood stream and stimulate production of large amount of anti–Rh antibodies by the mother. These anti – Rh antibodies persist in mother's blood for a long time and are persistent risk for the next Rh^+ foetus.

Control of Incompatibility

- Rh sensitization of Rh⁻ mother is avoided by a simple therapy. She is given an injection of Rh antiserum during early pregnancy and immediately after birth. The Rh–antibodies in the Rh antiserum will destroy Rh^+ RBC of the foetus before they stimulate production of maternal anti–Rh antibodies. The injected antiserum disappears before the next pregnancy.
- Sometimes a mild ABO incompatibility protects the baby against a more severe Rh incompatibility. If O^- mother conceives A^+ or B^+ 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.

Activity

An Rh^- woman is married to an Rh^+ man whose father was also Rh^-.
What is the probable risk of erythroblastosis foetalis in their babies?

Solution

Genotype of the mother is dd
The father of the man was Rh^-, Thus the genotype of the father is Dd.

Cross

Father	X	Mother
Dd	↓	dd

Gametes

D, d d

Progeny

Dd (Rh^+), dd (Rh^-)

Result

The result of the cross shows that the 50% of the baby will be Rh^+ and 50% will be Rh^-. Therefore, there is 50% chance of erythroblastosis foetalis in their babies.

QUESTIONS RELATED TO ABOVE ARTICLE

Explain the ABO blood group system. (LHR 2018, BWP 2019)
Explain the genetic basis of human blood groups. (SGD 2019)
Discuss Rh blood group system in man. (MTN 2021)
What are multiple alleles? Explain with an example. (MTN 2021, FSD 2021)
Write note on mother-fetal RH incompatibly (FSD 2021)
Write a note on Rh blood group system. Give the principle of its transfusion. (DGK 2022)
Define multiple alleles. Describe multiple allelic blood group system of man. (Exercise question vi)
What is Rh factor? Describe the genetic basis of Rh blood group system of man. (Exercise question vii)
What is erythroblastosis foetalis? Discuss the adverse effect of Rh-incompatibility. Also suggest a therapy to avoid Rh sensitization of an Rh negative mother married to an Rh positive man. (Exercise question viii)

22.5 EPISTASIS

Definition

When an effect caused by a gene or gene pair at one locus interferes with or hides the effect caused by another gene or gene pair at another locus, such a phenomenon of gene interaction is called **epistasis**.

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.

Example

Bombay phenotype is an example of epistasis.

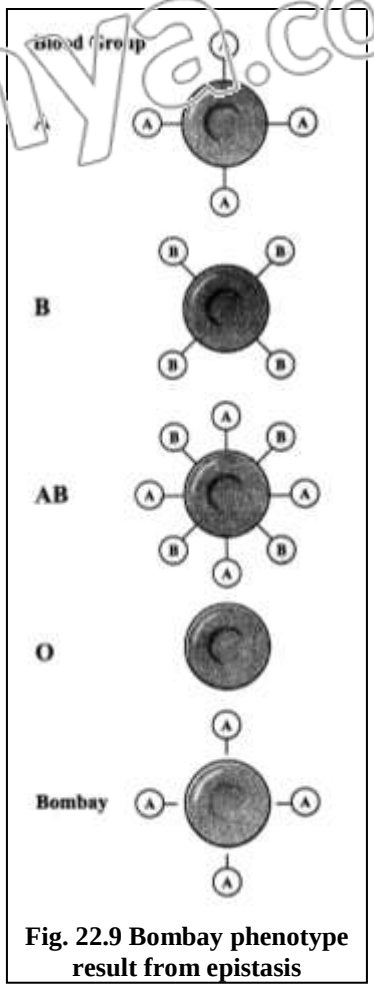


Fig. 22.9 Bombay phenotype result from epistasis

Bombay Phenotype

Definition

Such a blood phenotype which is different from genotype is called Bombay phenotype.

Mechanism

The expression of ABO blood type antigens by I^A or I^B gene depends upon the presence of another gene H.

(i) ABO locus is on chromosome 9.

(ii) H locus is on chromosome 19.

- H gene (dominant) changes a precursor substance into substance H.
- It produces an enzyme that inserts a sugar onto a precursor glycoprotein on the surface of RBC.
- Antigen A or antigen B specified by I^A or I^B gene could attached to this sugar of substance H

The recessive allele h cannot insert sugar molecule to glycoprotein. Therefore, hh individuals lack the site of attachment for antigen A or antigen B. Their RBC lack A and B antigens although they do not lack I^A and I^B genes. They are phenotypically like O but are not genotypically O.

Activity

A student of biology learns about ABO blood types. He knows that he is type O, and his father is type A and mother is type AB. He wonders how his blood type could have arisen. Suggest how type A and AB parents could produce a child of blood type O.

Solution

First possibility if the A is homozygous

I^AI^A ↓ I^AI^B

Gametes I^A, I^A I^A, I^B

Progeny I^AI^A, I^AI^B

50% children with blood group A and 50% with blood group AB.

Second possibility if the A is heterozygous

I^Ai ↓ I^AI^B

Gametes I^A, i I^A, I^B

Progeny I^AI^A, I^AI^B, I^Ai, I^Bi

Result

The above cross shows that it is not possible that two parents with blood group A and AB produce O blood group. They always produce blood group A or blood group B.

QUESTIONS RELATED TO ABOVE ARTICLE

What is Epistasis? Explain it with example of Bombay phenotype. (GRW 2019, GRw 2022, RWP 2022)

What is epistasis? Explain your answer with an example (BWP 2022)

Define epistasis. Explain epistasis gene interaction with examples.

(Exercise Question ix)

22.6 PLEIOTROPY

Definition

When a single gene affects two or more traits, the phenomenon is called pleiotropy. Such a gene with multiple phenotypic effects is called pleiotropic.

Examples

1. White eye gene in *Drosophila* also affects the shape of sperm storing organs (spermathecae).

2. Genes that affect growth rate in humans also influence both weight and height.

3. In cats, the dominant allele W not only makes fur pure white but also causes deafness. In w homozygous normal pigmented cats, melanocytes produce pigment of fur and also contribute to hair cells in inner ear that sense sound.

When a cat gets W allele, its melanocytes fail to develop properly. Melanocyte failure causes change in both phenotypes i.e. white fur and deafness.

QUESTIONS RELATED TO ABOVE ARTICLE

Explain epistasis and pleiotropy

Explain pleiotropy with the help of examples. (RWP 2021)

What is a pleiotropic gene? Discuss pleiotropy with examples. (Exercise Question x)

22.7 CONTINUOUSLY VARYING TRAITS

Phenotypic Expression

Phenotypic expression of traits has two aspects i.e.

1. Qualitative

2. Quantitative

1. Qualitative Traits

Definition

Such traits that vary qualitatively are called qualitative traits.

Features

- Qualitative differences (variations) are large and more obvious.
- Some traits like pea seed shape show **discontinuous qualitative variations** with two, sharply distinct phenotypes.
- Their frequency diagram forms asymmetric distribution curve, with much greater frequency of phenotypes at one end than at the other end.
- Mostly traits are controlled by single gene/gene pair or some multiple alleles.

Examples

- Mendelian traits of pea plant show two **phenotypes Round or wrinkled**
- Flower colour in 4 O' clock plant has three phenotypes Red, pink, and white.
- ABO blood group system has four phenotypes A, B, AB and O.

Tongue Rolling as Example

Some people can roll their tongue into a distinct U shape when they extend it out of their mouth. They are called rollers. This ability is due to a single dominant gene.

2. Quantitative Traits (Continuously Varying Trait)

Such traits that vary quantitatively are called quantitative traits or continuously varying trait.

A continuously varying trait is encoded by alleles of two or more different gene pairs found at different loci, all influencing the same trait in an additive way. These quantitative traits are called **polygenic** traits and their genes are **polygenes.**

Features

- Quantitative differences are small and less striking.
- These traits are controlled by more than one pair of genes and show many phenotypes.
- Each polygene has a small positive or negative effect on the character. Polygenes supplement each other and sum of positive and negative effects of all individual polygenes produce quantitative phenotypes of a **continuously varying trait.**
- Frequency histogram of quantitative traits shows bell shaped curve in which maximum phenotypes are present in the mid. The horizontal or X axis indicates the range of different phenotypes of a trait while vertical or Y axis indicates the number of individuals or their percentage.

Examples

i) Wheat Grain Colour

Wheat grains vary in colour from white to dark.

Experiment of Nilsson-Ehle

Nilsson-Ehle studied the genetics of wheat grain colour.

When he crossed a true breeding dark red grain plant with a true breeding white grain plant, all F1 grains had light red colour, intermediate between two parental shades. It seemed to be a case of incomplete dominance.

But when F1 grains were grown to mature plants and crossed with each other, F2 grains had exactly seven shades of colour in the ratio of 1 dark red : 6 moderately dark red : 15 red : 20 light red : 15 pink : 6 light pink : 1 white.

Genetics

Three different gene pairs (Aa, Bb, Cc) at different loci contribute to wheat grain colour.

- Alleles A, B and C code for an equal amount of red pigment, which is a **positive** effect.
- But none of a, b and c encode red pigments, which is a no (zero) dose **negative** effect.

Phenotypes In Relation To Genotypes

- If all the six alleles code for red pigment (AABBCC), the grain is **dark red**.
- When none of the six alleles encode 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**.
- Four alleles colour dose (AABBcc or aaBBCC or AAbbCC) will make **red**.
- Five alleles colour dose (AABBCc or AABbCC or AaBBCC) will produce moderately **dark red** grain.

Fig 22.10 Colour variation in wheat grains is a polygenic trait.

Thus the colour phenotype of the grain is the sum of the individual effects of all the six alleles.

Environmental factors like light, water and nutrients also influence the amount of grain colour. Environmental variations make the distribution of phenotypes more smooth and continuous.

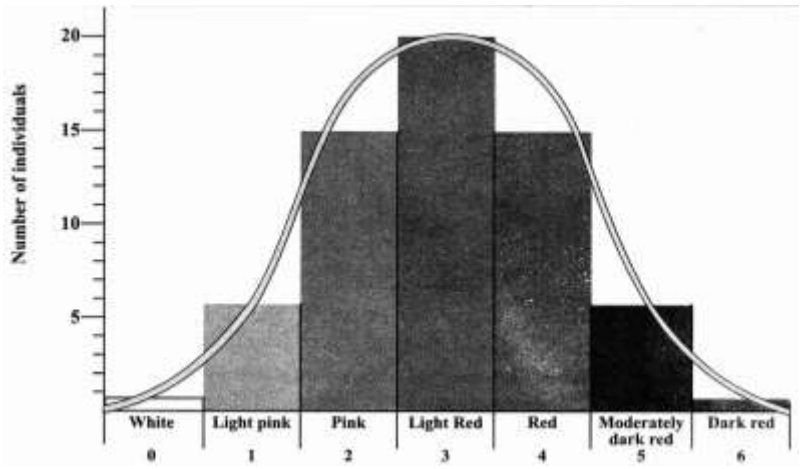


Fig. 22.11 Number of pigments – contributing alleles in F2

ii) Human Skin Colour

It is also a quantitative trait which is controlled by three to six gene pairs. The greater the number of pigment specifying genes, the darker the skin. A child can have darker or lighter skin than his parents.

iii) Human Height

It is a more complex polygenic trait. The perfectly continuous variation in range of human heights produces a smooth bell-shaped curve. A few people are very tall or very short, but most individuals fall in the average or mean value.

This trait is controlled by many pairs of genes at different loci. Even multiple alleles may be possible at each locus.

More the number of alleles for shortness, the shorter the height will be. Similarly, greater the number of alleles for tallness, the taller the height will be.

Affect of Environment

Environment also has a strong influence on height, intelligence and skin colour in humans. Constant exposure to sun darkens skin. Poor nutrition prevents achieving genetically determined height. Healthy and encouraging social environment promotes intelligence.

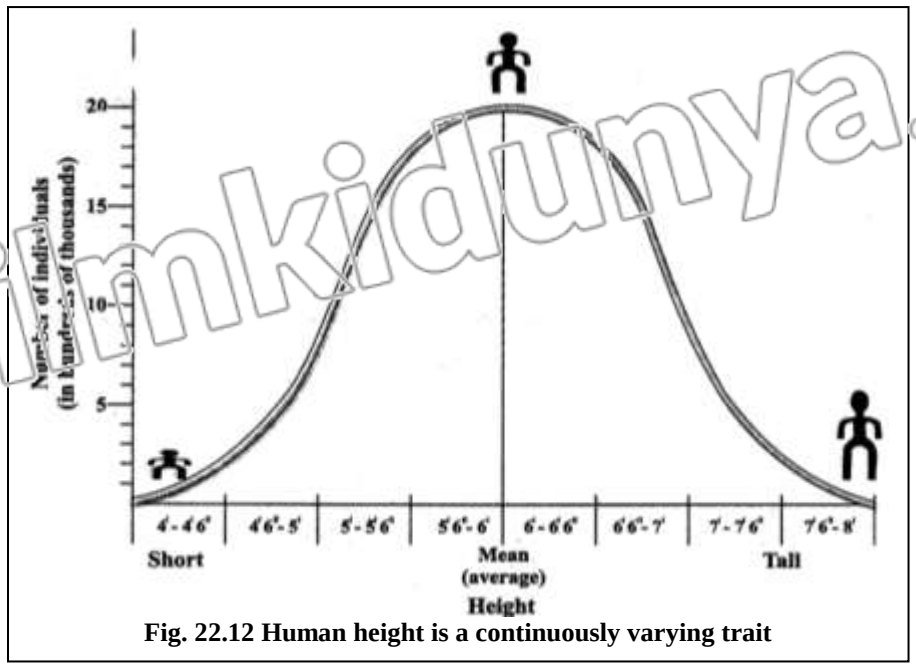


Fig. 22.12 Human height is a continuously varying trait

Activity

Study continuous variations in height and discontinuous variation in tongue rolling ability of man and record your observations as histograms.

Solution

Frequency Histograms

A frequency histogram is a simple graph. It shows variations.

- The horizontal or X axis indicates the range of different phenotypes of a trait within a population.
- The vertical or Y axis indicates the number of individuals or their percentage in the population.

Frequency Histograms of Tongue Rolling

Some people extend their tongue out of their mouth and roll it into U shape. They are called rollers. This character is due to a single dominant gene. It is a discontinuous variation.

So it is inherited in simple Mendelian fashion. Its frequency diagram from asymmetric distribution curve. This curve show much greater frequency of phenotypes at one end and then at the other end.

Fig 23.13 Roller

Frequency Histograms of Human Height

Human height is a continuously varying trait. A frequency diagram of heights of humans in a large population shows that so many phenotypes are found in it. The categories of these phenotypes blend into one another. It forms a smooth bell-shaped normal distribution curve.

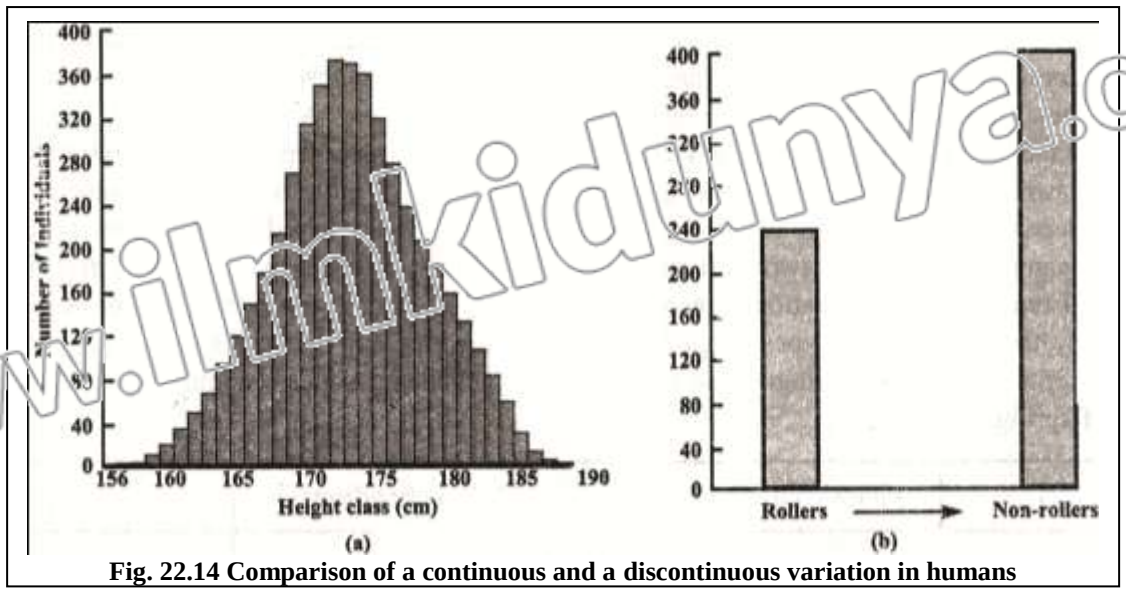


Fig. 22.14 Comparison of a continuous and a discontinuous variation in humans

Self-Work

Measure the heights of a large number of students in your college in cms (centimeter). Also note the ability of each student as roller on non-roller. Record your observation in a table like this.

Sr. No	Name	Height in cm	Roller / non-roller

Representing each measurement class as a bar with its height proportional to the number of individuals in each class, plot the graph (Fig. 22.14)

QUESTIONS RELATED TO ABOVE ARTICLE

What is a pleiotropic gene? Discuss pleiotropy with examples.

What are polygenes? Explain polygenic inheritance. (Exercise Question xi)

22.8 GENE LINKAGE

Definition

Phenomenon of staying together of all the genes of a chromosome is called gene linkage.

Features

i) It is a physical relationship between genes.

ii) A chromosome carries its linked genes en bloc in the form of linkage group.

iii) The number of linkage groups corresponds to the number of homologous pairs of chromosomes. Man has 23 linkage groups.

iv) Linked genes whose loci are close to each other do not obey Mendel's law of independent assortment, because these cannot assort independently during meiosis.

v) Gene linkage also minimizes the chances of genetic recombination and variations among offsprings.

Examples

- Genes for colour blindness, haemophilia, gout etc form one linkage group on human X chromosome.
- Genes for sickle cell anaemia, leukemia and albinism make another linkage group on human chromosome 11.

QUESTIONS RELATED TO ABOVE ARTICLE

Describe the phenomenon of gene linkage.

22.9 CROSSING OVER

Definition

Crossing Over is an exchange of segments between non-sister chromatids of homologous chromosome during meiosis.

Importance

Crossing over reduces the chances of gene linkage.

Explanation

Consider one pair of homologous chromosome. The homologous chromosomes pair up lengthwise, point to point and locus to locus. One homologue carries genes 'A' & 'B', the other homologue has 'a' & 'b'. Crossing over occurs at 4 strand stage (tetrad) between non-sister chromatids with formation of chiasma. Exchange of chromosome segments logically means exchange of DNA, i.e. genes or alleles. Alleles recombine and after separation four types of gametes are formed.

- Two with parental combinations of linked genes i.e. AB and ab.
- Two with recombination of genes i.e. Ab and aB.

If crossing over does not occur, only two parental types of gametes are formed. Parental types of gametes produce parental types of offsprings, while recombination gametes produce recombinant types of offsprings.

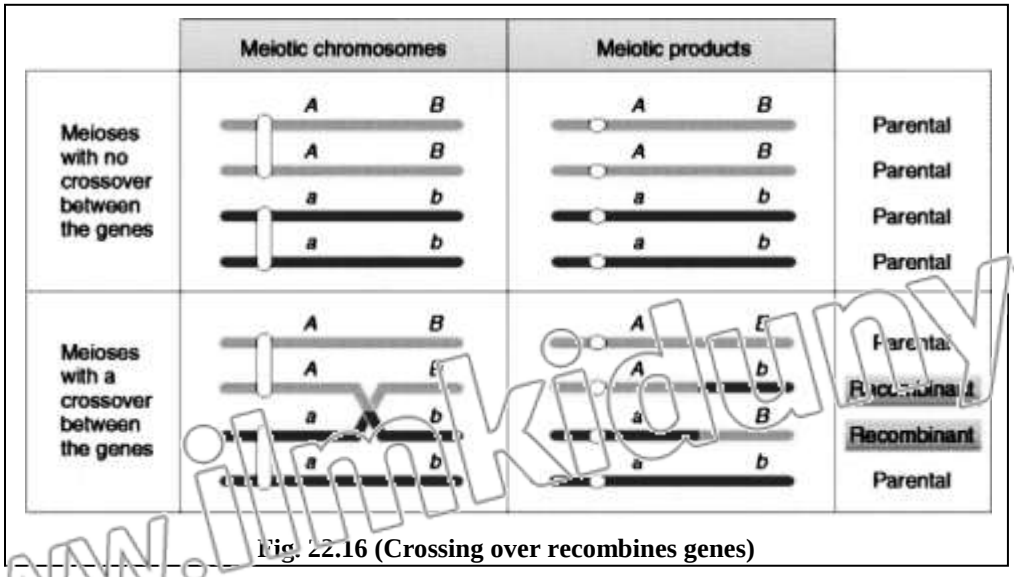


Fig. 22.16 (Crossing over recombines genes)

22.9.1 Cross Over or Recombination Frequency

Definition

It is the proportion of recombinant types between two gene pairs as compared to the sum of all combinations.

Formula

$$\text{Recombination Frequency} = \frac{\text{Recombinant Types}}{\text{Sum of all combinations}} \times 100$$

Calculation of Recombination Frequency

The recombination frequencies between two linked genes can be calculated by backcrossing the heterozygote to a homozygous double recessive.

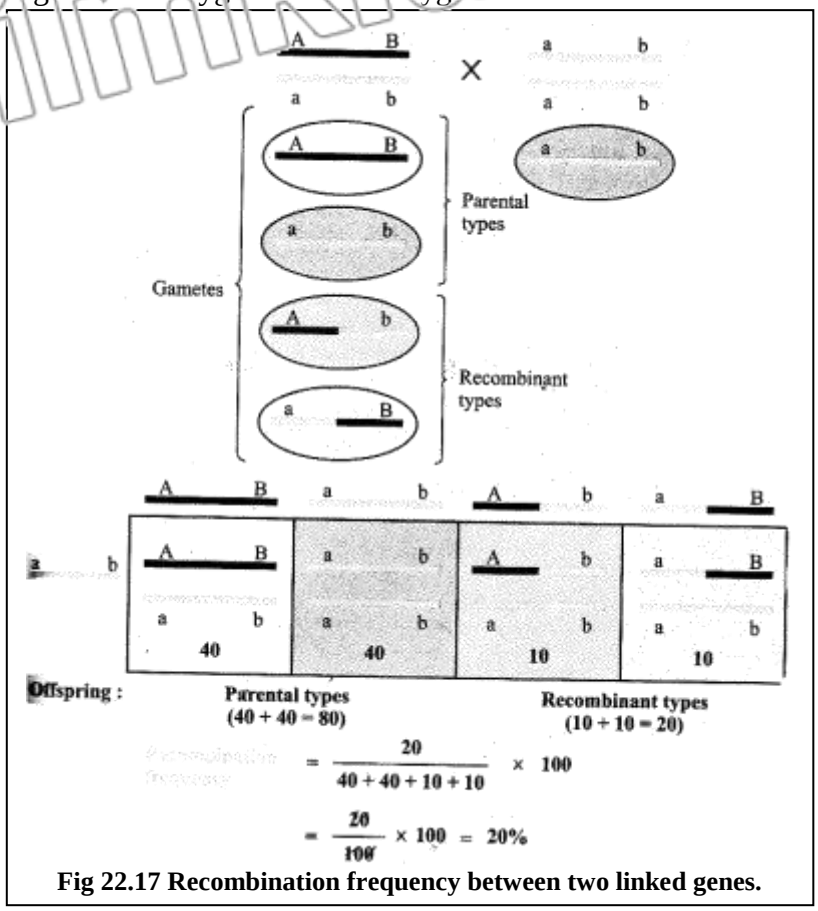
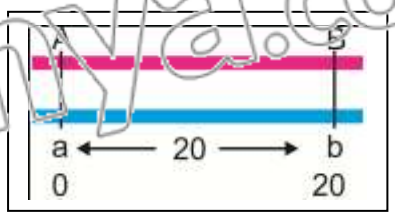


Fig 22.17 Recombination frequency between two linked genes.

Mapping of Gene

The recombination frequency is directly proportional to the distance between the linked gene loci. Genes can be mapped on a chromosome on the basis of their recombination frequencies. If 1% of recombination frequency is equal to 1 unit map distance, the two linked genes A and B with a 20% recombination frequency must be 20 units apart.



Importance of Crossing Over

Crossing over produces genetic variations among offspring. Genetic variations lead to tremendous variations in their traits. Variations provide raw material for evolution by letting them adapt successfully to the changing environment.

QUESTIONS RELATED TO ABOVE ARTICLE

Explain the process of crossing over with the help of diagram. (LHR 2022)

What is crossing over? Define recombination frequency and explain its significance. (Exercise Question xii)

22.10 SEX DETERMINATION

SEX CHROMOSOME & AUTOSOME

Sex Chromosomes

Such chromosomes which have genes for determination of sex are called sex chromosomes.

Autosomes

All chromosomes other than sex chromosomes are called autosomes.

Autosomes do not carry any sex determining gene.

Discovery

Sex chromosomes were first discovered by T. H. Morgan in *Drosophila.*

Examples

1) In Drosophila

In fruit fly, *Drosophila melanogaster*, there are four homologous pairs of chromosomes. The chromosomes of the three homologous pairs are similar in both of the sexes, but fourth pair is different.

- The female has two similar rod-shaped X-chromosomes in the fourth pair
- The male has one rod shaped X-chromosome but the other a morphologically different J-shaped Y-chromosome in the fourth heteromorphic pair.

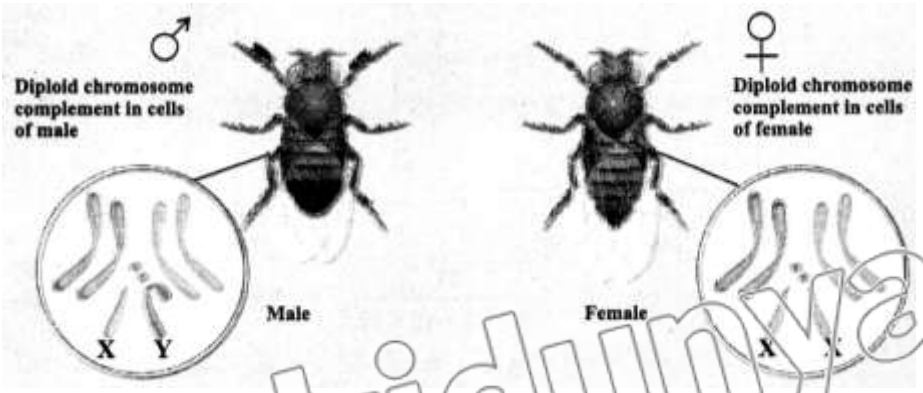


Fig. 22.18 Chromosomes of ♂ ♀ *Drosophila melanogaster*

2) In Human

Humans have 46 chromosomes in the form of 23 pairs. 22 pairs are of autosomes and one pair is of sex chromosome.

Autosome pairs are common in both the sexes but the 23rd sex chromosome pair is different in male and female.

- Woman has two similar X-chromosomes in her 23rd pair. Thus, she is XX.

- A man has an X-chromosomes along with a much shorter Y-chromosome in his 23rd pair. The 23rd pair in man is heteromorphic. Thus, he is XY.

  SRY is the male determining gene. It is located at the tip of short arm of Y-chromosome. Its name SRY stands for 'sex determining region of Y.

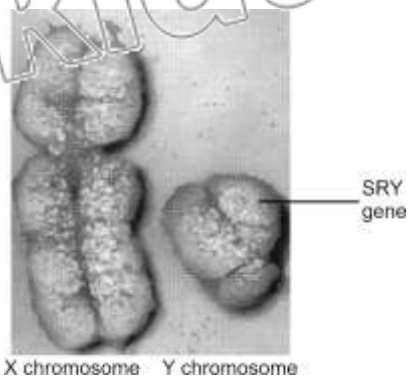


Fig. 22.19 Sex Chromosomes of a man

3) In Grasshopper

In some grasshoppers, males and females have different number of chromosomes.

- The female has 24 chromosomes in the form of 11 pairs of autosomes and a pair of X chromosomes. Thus, female is XX.
- Male grasshopper has 23 chromosomes. He has 11 pairs of autosomes and only one X chromosome. The other member for sex chromosome pair is entirely missing in male. Thus, male is XO and female is XX.

4) In *Ascaris incurva*

Ascaris incurva (a round worm) has compound sex chromosomes.

- Female has 42 chromosomes in the form of 8 pairs of compound X along with 13 pairs of autosomes (16 + 26).
- Its male has 35 chromosomes comprising 8X plus one Y along with 13 pairs of autosomes (8 + 1 + 26).

QUESTIONS RELATED TO ABOVE ARTICLE

Explain the phenomenon of sex determination in humans. (LHR 2019)

What are sex chromosomes? Discuss the chromosomal patterns of sex determination in organisms. (Exercise Question xiii)

22.10.1 Patterns of Sex Determination

22.10.1 a Sex Determination In Animals

There is a wide variety of sex determining mechanisms, but three patterns are more common.

1) XO-XX Type

Examples

This pattern of sex determination is found in grasshopper and *Protenor bug.*

Features of Male

- Male is XO because it has only one X chromosomes. The other sex chromosome is missing entirely.
- Male is heterogametic because it forms two types of sperms; half the sperms have X chromosome while the other half are without any sex chromosome. A gamete without any sex chromosome is called nullo gamete.

Features of Female

- 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 Determining Gamete

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 is 1:1.

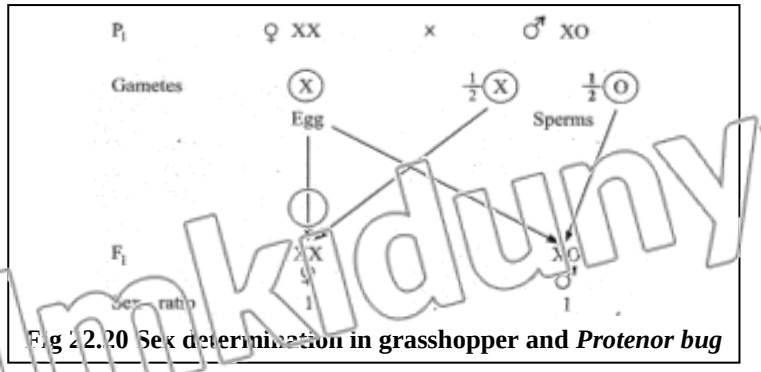


Fig 22.20 Sex determination in grasshopper and *Protenor bug*

2) XY-XX Type

Examples

This pattern of sex determination is found in *Drosophila*, man and many other organisms.

Features of Male

- Male is XY.
- Male being heterogametic produces two types of sex determining sperms. Half the sperms carry X-chromosome and the other half carry Y-chromosome.

Features of Female

- Female is XX.
- Female being homogametic produces only one type of eggs, each having an X-chromosome.

Sex Determining Gamete

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.
- Sex ratio between male and female offspring is 1:1.

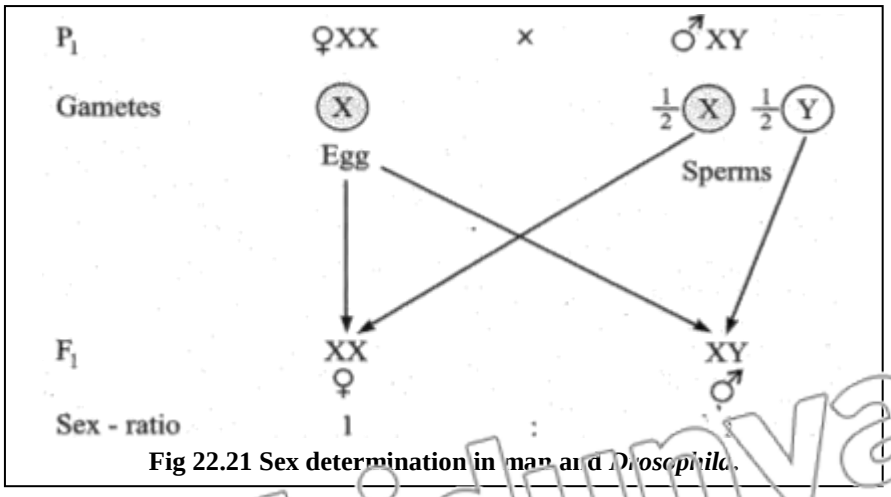


Fig 22.21 Sex determination in man and *Drosophila*.

3) ZZ-ZW Type

Examples

This type of sex-determination pattern is common in birds, butterflies and moths.

It was discovered by J. Seiler in 1914 in moth.

It is the reverse of XY-XX system.

Features of Male

Male is homogametic (ZZ). All sperms are alike carrying a Z-chromosome.

Features of Female

Female is heterogametic (ZW). Female produces two kinds of eggs Z and W in equal proportions.

Sex Determining Gamete

It is the kind of egg that determines the sex of offsprings.

- When a Z carrying egg is fertilized by the sperm, a male offspring is produced.
- When a W-carrying egg is fertilized by the sperm, a female offspring is produced.
- Sex Ratio is 1:1.

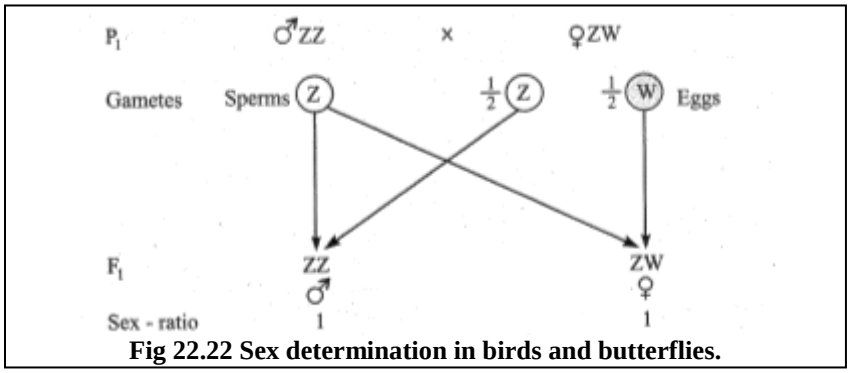


Fig 22.22 Sex determination in birds and butterflies.

COMPARISON OF CHROMOSOMAL DETERMINATION OF SEX BETWEEN DROSOPHILA AND HUMANS

FEATURE	MAN	*DROSOPHILA*
Female determining chromosome	X	X
Male determining chromosome	Y	Autosome
Sex determining system	Sex chromosome (SRY on Y)	X-chromosome autosome balance
Turner's syndrome (XO)	Sterile female	Sterile male
Klinefelter's syndrome (XXY)	Sterile male	Fertile female

Species	XX	XY	X0	XXY
Drosophila	♀	♂	♂	♀
Humans	♀	♂	♀	♂

Fig. 22.23 Comparison of sex determination in man and *Drosophila*

In drosophila, an X: A ratio of 1.00 or higher produces female whereas an X: A ratio of 0.5 or lower produces males.

QUESTIONS RELATED TO ABOVE ARTICLE

Compare chromosomal determination of sex in *Drosophila* and human.

Explain different patterns of sex determination in animals.

Explain XO-XX and ZZ-ZW types of sex determination.

Discuss pattern of sex – determination is animals?

(SGD 2022)

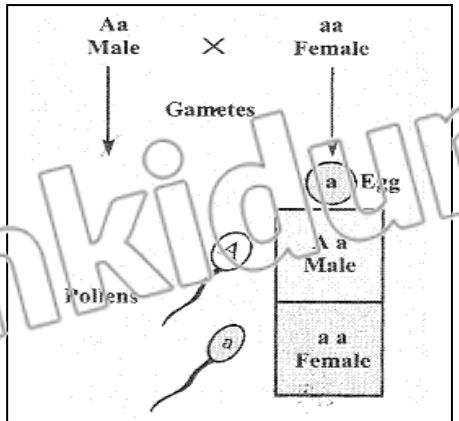


Fig. 22.24 Pollens determine Sex

22.10. 1 b Sex Determination In Plants

Sex Determination in Plants

Most of the plants are monoecious but some are dioecious (having plants of separate sexes) e.g. Ginkgo.

In it, male plant produces flowers with only stamens and female plants produce flowers with only carpels.

Mechanism of Sex Determination

- Some dioecious plants have a difference of sex chromosomes between the sexes. These have an X-Y system. These plants typically exhibit an X chromosome-autosome balance system for sex determination.
- Many other sex determining mechanism are also seen in dioecious plants.

Work of Correns

Correns (1907) discovered that pollens of certain plants were sex-determining. All eggs are of one type. Pollens of the two types are produced in equal number. One kind of pollen after fertilizing the egg produces male plant whereas the other kind of pollen after fertilization produces female plant.

22.10.1 c Sex Determination In Yeast

Yeast and other eukaryotic microorganisms do not have sex chromosome. These depend on genic system for determination of sex. In this system the sexes are specified by simple allelic differences at a small number of gene loci e.g. a and α are the two mating types (sexes) of yeast, controlled by MAT a and MAT α alleles respectively.

QUESTIONS RELATED TO ABOVE ARTICLE

What is crossing over? Define recombination frequency and explain its significance.
Explain sex determination in plants.
Describe the process of sex determination in plants and yeast.

(RWP 2019, SGD 2021)

22.11 SEX LINKAGE

22.11.1 Sex Linkage in Drosophila

Thomas Hunt Morgan (1910) provided experimental evidence in support of chromosomal theory of heredity through discovery of sex linkage in fruitfly Drosophila.

Reasons for Selection of Drosophila

Drosophila is a very useful organism for genetic studies for many reasons.

i) Easy collection & culturing

The tiny fly is found hovering around rotten fruits. It can be easily collected and cultured on mashed bananas and other fruits. It does not need large spacious cages and lives happily in ordinary glass bottle of jams and marmalades. It eats yeast that grows on mashed banana.

ii) Sexual dimorphism

Male and female *Drosophila* show sexual dimorphism i.e. these are morphologically distinct from each other.

- Male is smaller in size with black rounded abdomen male has sex combs on front legs.
- Female is larger with pointed abdomen.

iii) Short generation time

It has generation time of just two weeks. It lays a large number of eggs, which hatch out into fertile offspring. Many generations can be raised in a relatively short time.

iv) Excellent for genetic studies

It is perfectly suited for genetic studies.

- It shows a large number of distinct contrasting traits. Morgan and his colleagues studied pattern of inheritance of more than about 85 traits of drosophila.
- Its larvae are excellent material for dissection for chromosome study.
- It has only eight chromosomes. Salivary gland cells have giant chromosomes in their nuclei. These have banding pattern corresponding to genes.

v) Part of human genome project

The entire genome of Drosophila has been successfully sequenced as part of human genome project.

Morgan's Experiments & Crosses

Morgan raised cultures of Drosophila flies to study different traits, such as colour of the eye. Normal fruit flies, the **wild type,** have bright red eyes. One of his coworkers Calvin Bridges, observed an unusual white eye mutant male fly.

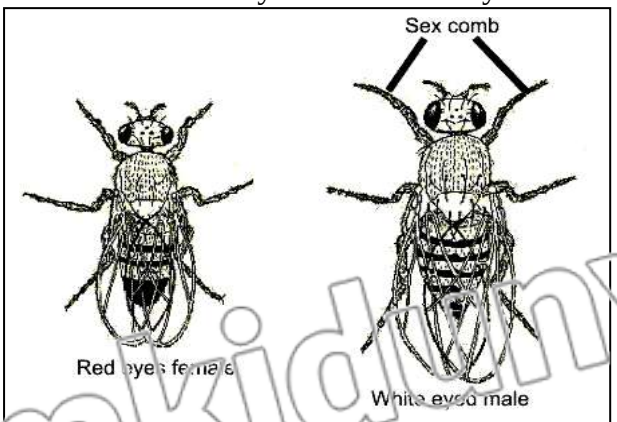


Fig. 22.25 Wild type red eyed female and mutant white eyed male Drosophila.

Step 1 Normal Cross

Morgan mated white eyed male with a wild type red eyed female. All 1237 offspring of this cross had red eyes.

Morgan concluded that red eye is dominant trait.

Step 2 Normal Cross

Morgan allowed males and females of F_1 generation to mate and produce F_2 generation. He counted 2459 red-eyed females, 1011 red-eyed males and 782 white eye males among F_2.

Observations

Different observations got by this cross were;

- Offspring produced due to this cross were red-eyed females, red-eyed males and white-eyed males.
- The proportion of 3470 red eyed to 782 white eyed flies did not perfectly fit into Mendelian 3:1 ratio.
- The number of recessive phenotype individuals was too small.
- All the white-eye flies were only males. There was no white eye female in F_2.

Conclusion

The inheritance of eye color somehow seemed to be related to the sex of offspring. Morgan proposed that

i) The gene for eye color is located on X-chromosome.

ii) The alleles for eye color are present only on X-chromosome. There is no corresponding allele for this trait on Y-chromosome.

iii) Single recessive allele on X-chromosome can express itself in males because Y-chromosome is empty for that gene. Males are hemizygous as they carry just one allele on their only X- chromosome. Females have two X-chromosomes, each carrying an allele of the trait. Females can be homozygous or heterozygous.

Symbol 'w' represents recessive allele for white eye and 'w^+' designates its wild type allele for red eye.

Now we can relate these crosses with genotype.

The genotype of the parents of P_1 cross were $X^{W+}X^{W+}$ for red eye female and $X^{W}Y$ for white eye male.

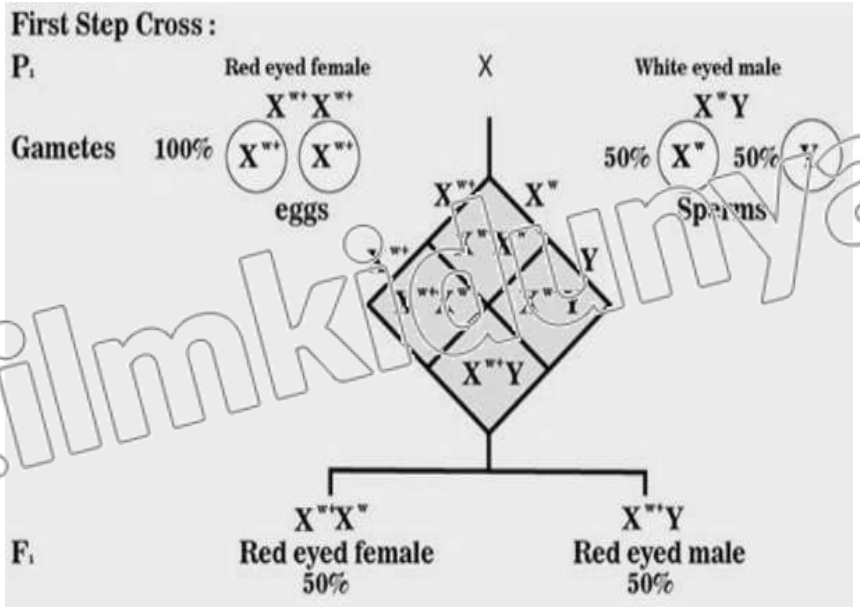


Fig. 22.26 (a)

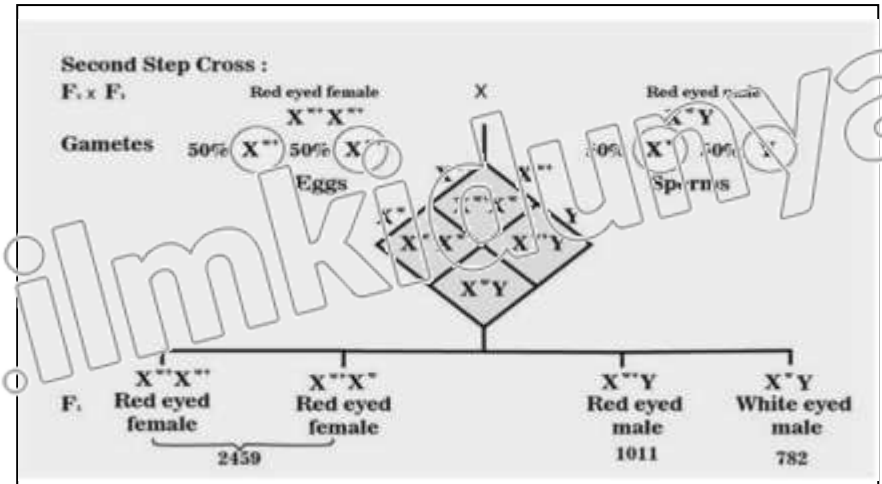


Fig. 22.26 (b)

Step 3 Test Cross

Morgan wanted to test his hypothesis.

He crossed the P_1 white eyed male (X^wY) with one of its own daughters (the red-eyed heterozygous female from F_1 generation).

Out of total offspring of this cross, half female offspring had red eyes and half had white. Similarly, half the males had red eyes and half had white.

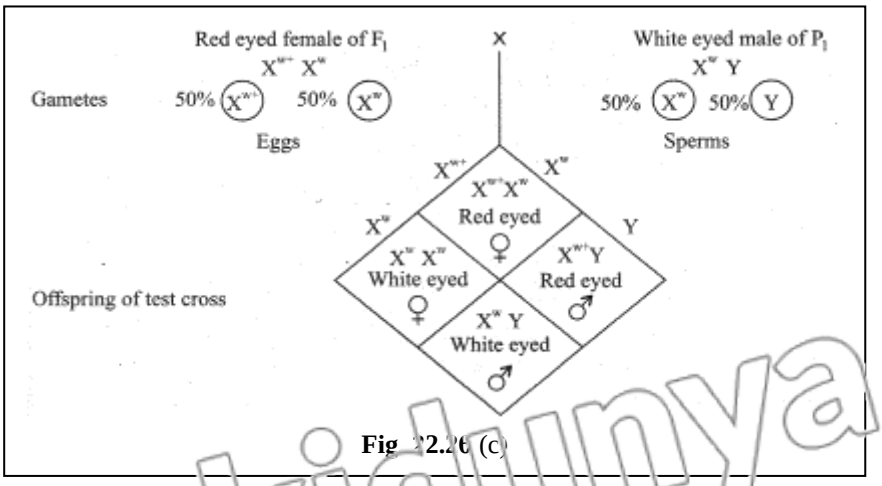


Fig. 22.26 (c)

Step 4 Reciprocal Cross/ Confirmatory Test

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.

When F_1 red-eyed females and white-eyed males were mated to produce F_2, then half of the F_2 females had red eyes and half white eyes. Similarly, half of the F_2 males had red eyes and half white. This F_1 x F_1 cross was exactly like step 3 test cross.

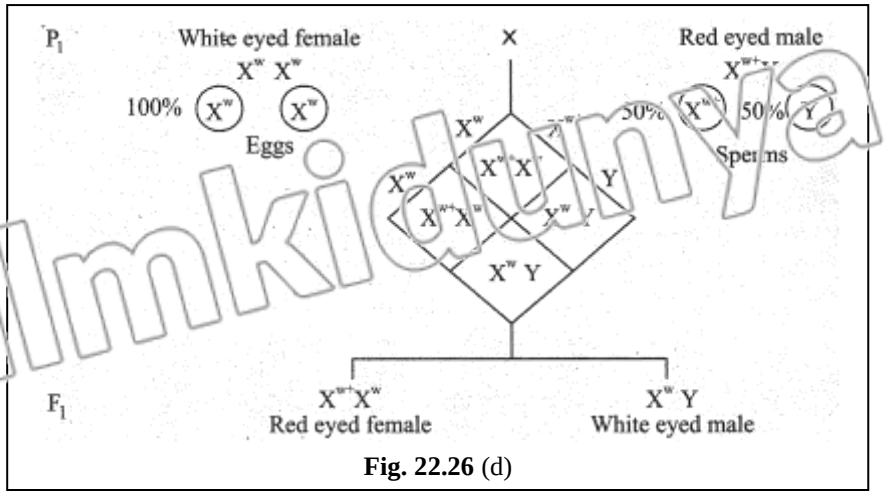


Fig. 22.26 (d)

Different Traits of Organisms

1) X-Linked Trait

Definition

A trait whose gene is present on X-chromosome is called X-linked trait.

Features

- X-linked traits are commonly referred as sex-linked traits.
- A gene present only on X-chromosome, having no counterpart on Y-chromosome, is called X-linked gene.
- An X-linked trait passes in crisscross fashion from maternal grandfather (P_1) through his daughter (F_1) to the grandson (F_2).
- It never passes direct from father to son because a son inherits only Y-chromosome from father.

2) Y-Linked Trait

Definition

Y-chromosome is not only completely inert. It does not carry a few genes which have no counterpart on X-chromosome. Such genes are called Y-linked genes and their traits are called Y-linked trait.

Features

- Such genes which are present only on Y-chromosome are called Y-linked genes.
- Example is SRY gene on Y-chromosome of man that determines maleness.
- Y-linked traits are found only in males.
- These traits directly pass through Y-chromosome from father to son only. As females do not normally inherit Y-chromosome, such traits cannot pass to them.

3) X- and Y-Linked Trait

Definition

Such a trait whose genes are located both on X and Y chromosomes is called X- and Y-linked trait.

Features

- Such genes which are located both on X and Y chromosomes are called X- and Y-linked genes or pseudoautosomal genes.
- Their pattern of inheritance is like autosomal genes.
- Example is bobbed genes as in drosophila.

QUESTIONS RELATED TO ABOVE ARTICLE

Explain sex linkage in drosophila with crosses.
Discuss sex-linkage in humans with one example. (LHR 2018)
Define sex linkage. Discuss X-linked dominant inheritance in humans. (DGK 2019)
What is X-linked recessive inheritance? Explain it with an example. (LHR 2021)
What is sex-linkage? Explain T.H. Morgan's study of sex-linkage in Drosophila. (Exercise Question xvi)
Compare pattern of inheritance of an X-linked dominant trait with an X-linked recessive trait in humans. (Exercise Question xvii)

22.1.2 Sex Linkage in Humans

Different sex-linked traits in humans are described here briefly. Mode of inheritance of human traits can be traced through pedigree.

Many X–linked traits in man are also found X–linked in other mammals like mouse, rabbit, dog, sheep, horse, donkey, cattle, kangaroo and chimpanzee. Was the mammalian X–chromosome conserved throughout mammalian evolution?

1) X-LINKED TRAITS

The pattern of X-linked traits is further divided into

A) X-linked recessive trait
B) X-linked dominant trait

A) X-Linked Recessive Trait

Definition

Such a trait that is determined by an X-linked recessive gene is called X-linked recessive trait.

Examples

(i) Haemophilia
(ii) Colour blindness
(iii) Testicular feminization syndrome

(i) HAEMOPHILIA

Definition

It is a hereditary disease in which patient's blood fails to clot properly after an injury.
It is a rare X-linked recessive trait.

Cause

Effect:

It results due to either reduction or malfunction or complete absence of blood clotting factors.
It is a serious hereditary disease because a haemophilic may bleed to death even from minor cuts.

Types

It is of three types i.e. A, B and C

1. Haemophilia A and B are non-allelic recessive sex-linked, but hemophilia C is an autosomal recessive trait.
2. 80% haemophilics suffer from haemophilia A due to abnormality of factor VIII, about 20% suffer from hemophilia B due to disturbance in factor IX but less than 1% suffer from hemophilia C due to reduction in factor XI.
3. Haemophilia A and B affect men more than women (being X-linked) but hemophilia C affects both the sexes equally (being autosomal).

Genetics

Haemophilia A and B zigzag from maternal grandfather through a carrier daughter to grandson. It never passes direct from father to son.

Gene for normal is 'H' and for hemophilia A is 'h'.

A woman can suffer from hemophilia A or B only when she is homozygous for the recessive allele but a man with just one recessive allele will show the trait.

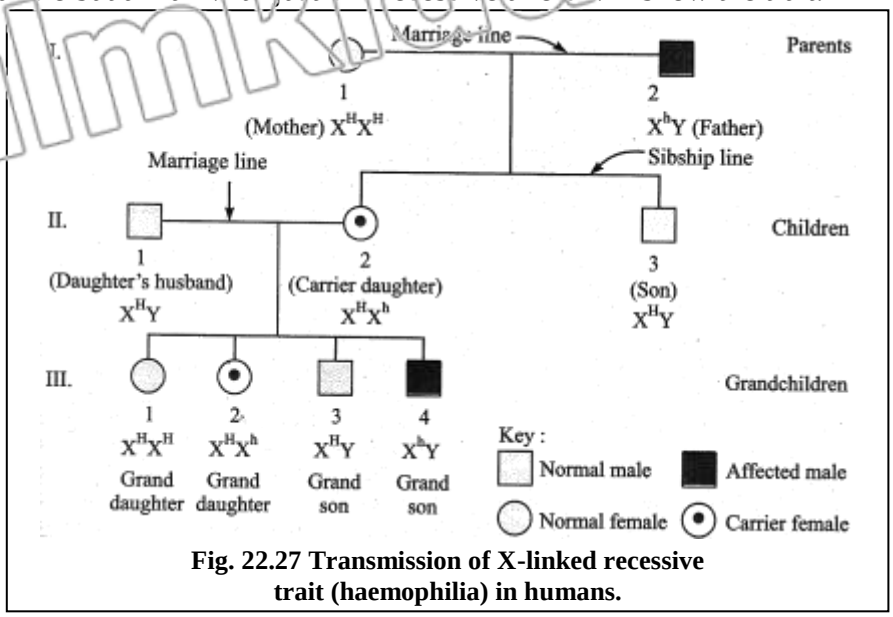


Fig. 22.27 Transmission of X-linked recessive trait (haemophilia) in humans.

Activity

Cases of Haemophilia A are reported in Queen Victoria's family. Pedigree of Queen Victoria's family **(figure)** indicates the Queen Victoria was a carrier mother, because she gave birth to an affected son Prince Leopold. Prince Leopold passed on this recessive X – linked trait in typical zigzag fashion through his carrier daughter (III – 1) to his grandson Rupert (IV – 1). Assign genotype to each individual. Can you explain how Alexis (IV – 3) became haemophiliac?

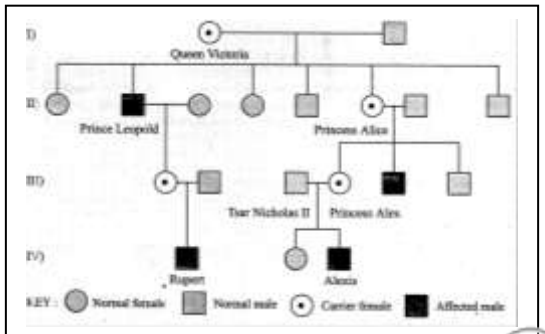


Fig. 22.28 Pedigree of Queen Victoria's family showing cases of Haemophilia A

Genotypes of the Individuals

(i) Queen Victoria, carrier (I – 1) X^HX^h

(ii) Her son, Prince Leopold (II – 2) X^hY

(iii) Daughter of Prince Leopold, carrier (III – 1) X^HX^h

(iv) His grandson Rupert (IV – 1) affected X^hY

Prince Alexis also inherited the X^h chromosome from Queen Victoria through her carrier grandmother Princess Alice and his carrier mother Princess Alex.

(i) Genotype of her grandmother (II – 6) X^HX^h

(ii) Genotype of her mother (III – 4) X^HX^h

(iii) Genotype of Prince Alexis (IV – 3) X^hY

(ii) COLOUR BLINDNESS

Definition

It is a hereditary disease in which person cannot differentiate between different colours.

Genetics

Normal trichromatic colour vision is based on three different kinds of cone cells in retina. Each cell is sensitive to only one of the three primary colours i.e. 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.

Types

Mutations in opsin genes cause three types of colour-blindness.

a) Dichromacy

A dichromate can perceive two primary colours but is unable to perceive the one whose opsins are missing due to mutation.

(i) Protanopia is red blindness.

(ii) Deuteranopia is green blindness.

(iii) Tritanopia is blue blindness.

b) Anomaly

Some people can detect red and green but with altered perception of the relative shades of these colours. They have abnormal but still partially functional opsins.

- Protanomalous and deuteranomalous for red and green weakness respectively.

c) Monochromacy

A monochromat can perceive only one colour. Monochromacy is true colour-blindness.

Red-Green 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. It also zigzags from maternal grandfather through a carrier daughter to grand son. It is more common in men than women because chances for a male to be affected by it are much more than a female.

(iii) TESTICULAR FEMINIZATION SYNDROME

It is a rare X-linked recessive trait. Although the persons affected by this trait have a male set of XY chromosome, yet this gene on their X chromosome develops them physically into females. They have breast, female genitalia, a blind vagina but no uterus. Degenerated testes are also present in abdomen. Such individuals are happily married as female but are sterile.

It is an androgen insensitivity syndrome. Male sex hormone testosterone has no effect on them.

Activity

A sex-linked recessive allele "c" produces red – blindness. Its normal dominant allele is "C". A normal woman whose father was red-blind marries a red-blind man. What proportion of their children can have normal colour vision?

Solution

Genotypes

Mother: As the father of the woman was colour blinded, so she must be carrier with genotype X^CX^c.

Father: As the father is affected his genotype will be X^cY

Cross Conclusion

$X^CX^c \times X^cY$

Gametes: X^C X^c X^c Y

Progeny

X^CX^c	X^cX^c	X^CY	X^cY
Normal	Affected	Normal	Affected

Conclusion

The cross shows that 50% of the children will have normal color vision, while 50% will be color blind.

B) X-Linked Dominant Inheritance

Definition

Such a trait that is determined by X-linked dominant gene is called X-linked dominant trait.

Genetics

Its pattern of X-linked dominant inheritance is different from X-linked recessive. It is more common in females than males.

All daughters of an affected father, but none of his sons are affected. Any heterozygous affected mother will pass the trait equally to half of her sons and half of her daughters.

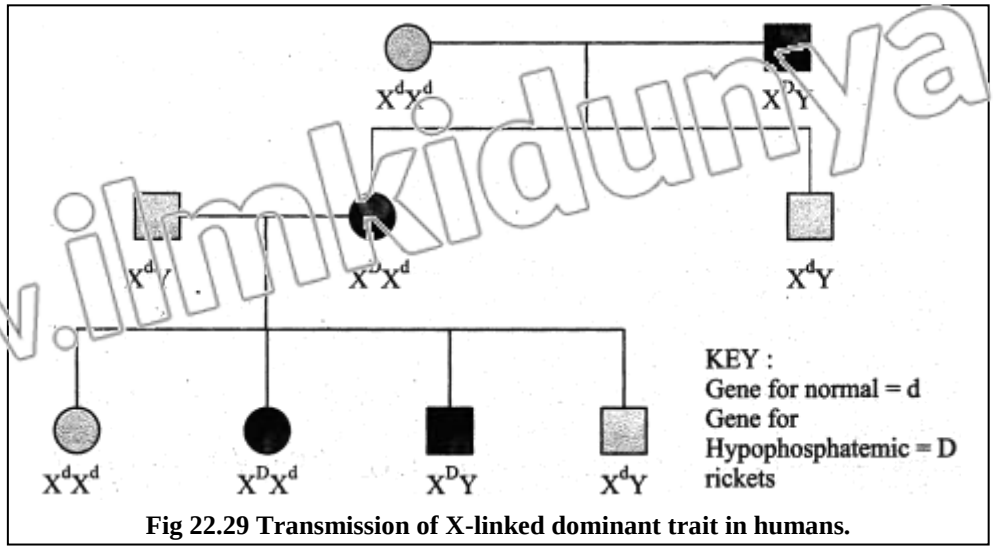


Fig 22.29 Transmission of X-linked dominant trait in humans.

Example

Hypophosphatemic rickets is an example of X–linked dominant trait. It does not result from vitamin D deficiency (as in dietary rickets) but its cause is genetic communication failure at molecular level.

2) Y-LINKED TRAIT

Genetics

It passes through Y chromosome from father to son only. Such traits cannot pass to daughters because they do not inherit Y chromosome. All sons of an affected father are affected by Y-linked trait.

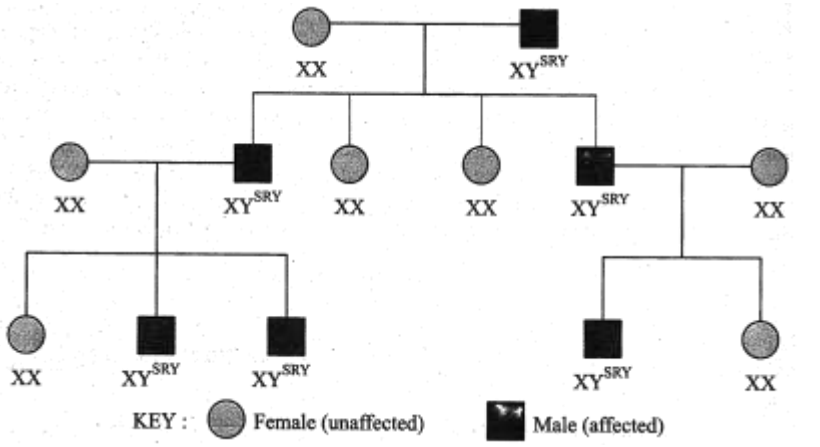


Fig 22.30 Y-linked inheritance in man.

Example

Maleness is a Y-linked trait. 'SRY' gene on Y chromosome determines maleness in man. It is male sex switch which triggers developmental process towards maleness after 6 weeks of pregnancy.

3) SEX LIMITED TRAIT

Definition

A sex-limited trait is limited to only one sex due to anatomical differences.

Genetics

Such trait affects a structure or function of the body present in only males or only females. These traits may be controlled by sex-linked or autosomal genes.

Example

- Genes for milk yield in dairy cattle affects only cows.
- Beard growth in humans is limited to men. A woman does not grow a beard herself, but she can pass the genes specifying heavy beard growth to her sons.

4) SEX INFLUENCED TRAIT

Definition

Sex influenced trait occurs in both males and females, but it is more common in one sex.

Genetics

It is controlled by an allele that is expressed as dominant in one sex but recessive in the other. This difference in expression is due to hormonal difference between the sexes.

Example

Pattern baldness is an example of sex influenced trait in humans. 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.

Activity:

A man is 45 years old and bald. His wife also has pattern baldness. What is the risk that their son will lose his hair?

Solution

Genotypes

Man

Baldness is dominant in man. So, a man can be homozygous and heterozygous bald BB or Bb.

Wife

Baldness is recessive in women. So, a bald wife must be homozygous recessive bb.

1. In case of homozygous dominant man.

	Man	×	Women
	BB	⇓	bb
Gametes	B	⇓	b
Offsprings		Bb	

Conclusion

In this case, genotypically all the offsprings will be heterozygous. Thus, all the boys will be affected by baldness, while all the girls will be normal.

2. In case of heterozygous male

	Man	×	Women
	Bb	⇓	bb
Gametes	B b	⇓	b
Offsprings	Bb		bb

Conclusion

In this case, 50% offspring will be heterozygous dominant and 50% will be recessive. Bb boys will be bald, but Bb girls will be normal. While bb boys will be normal, but bb girls will be bald. Thus, both boys and girls will be 50% bald and 50% normal.

QUESTIONS RELATED TO ABOVE ARTICLE

Discuss sex linkage in humans with one example.

Describe the genetics of color-blindness in humans. (SWL 2019, DGK 2019)

Discuss the genetics of colour-blindness or haemophilia. (Exercise Question xix)

22.12 DIABETES MELLITUS AND ITS GENETIC BASIS

Introduction

Diabetes mellitus is a hereditary disease.

It is actually a heterogeneous group of disorders which are characterized by elevated blood sugar level. Diabetics are unable to metabolize blood sugar in their body. They pass glucose in their urine.

Complications

Diabetes is the leading cause of kidney failure, adult blindness, lower limb amputation and heart diseases.

Types

There are two major types of diabetes

A) Type I (IDDM)

B) Type II (NIDDM)

A) Type I

Introduction

- It is also called insulin dependent diabetes mellitus (IDDM).
- It is also called Juvenile diabetes because it usually occurs in early age before 40.
- It arises due to deficiency of pancreatic hormone insulin that normally routes blood glucose to cells for use.

Mechanism

It is an autoimmune disorder. The immune system backfires and manufactures auto antibodies against body's own cells. Sometimes, specific viral infections activate autoimmune response. T-cells of immune system attack pancreas and destroy insulin producing β-cells.

Treatment

Diabetics of type I must receive exogenous (from outside source) insulin to survive

Genetics

The insulin gene is located on short arm of chromosome 11. Polymorphism and genetic variations within this locus are responsible for diabetes type I susceptibility. It is recessive single gene trait it is a multifactorial inheritance is associated with several alleles.

B) Type II

Introduction

- It is also called non-insulin dependent diabetes mellitus (NIDDM).
- It accounts for 90% of all diabetic patients.
- It occurs among people over the age of 40 and is more common among obese.

Mechanism

These persons produce some endogenous insulin themselves, but their body cells gradually fail to respond to insulin and cannot take up glucose from blood. They develop a sort of insulin resistance.

Obesity increases insulin resistance

BLOOD PRESSURE

Blood pressure is also an example of multifactorial trait. There is a correlation between systolic and diastolic blood pressure of parents and their children. This correlation is partly due to genes common in them. Blood pressure is also influenced by environmental factors such as diet, stress and tension.

Treatment

As exercise reduces obesity, it indirectly increases insulin sensitivity and improves glucose tolerance.

MODY

About 2-5% of type II diabetics get the disease early in life before 25 years of age. It is called maturity onset diabetes of the young (MODY).

Genetics

MODY can be inherited as an autosomal dominant trait.

Cause

- About 50% of cases of MODY are caused by mutations in glucokinase gene. Glucokinase enzyme usually converts glucose to glucose–6–phosphate in pancreas.
- MODY can also be caused by mutations in any of the four other genes which encode transcription factors involved in pancreatic development and insulin regulation. But these four MODY genes do not play any significant role in adult onset type II.

QUESTIONS RELATED TO ABOVE ARTICLE

Explain diabetes mellitus and its genetic basis.
(Exercise Question xviii, GRW 2021, FSD 2019)

Explain in detail diabetes mellitus and its types. (LHR 2017, 2021)

What is diabetes mellitus? Discuss diabetes type I disease. (SWL 2022, FSD 2022)

KEY POINTS

Gene

The smallest part of DNA with specific base sequence which control specific character is called gene.

Alleles

Partners of a gene pair are called alleles.

Phenotype

The form of appearance of a trait is called phenotype.

Genotype

The genetic complement of an individual is called genotype.

Clotting factors in haemophilia

13 factors are involved in the clotting of blood. Different factor are missing in different types of haemophilia. These factors are labeled as. Factor II, IV and VIII etc.

Back cross

The cross of an individual to any of the parent is called back cross.

Punnett Square

It is a square diagram that used to predict the genotype of a particular cross or breeding experiment, to determine the probability of an offspring having a particular genotype.

Polymorphism

A discontinous genetic variation resulting in the occurrence of several different forms or types of individuals among the members of single species. A polymorphism is a DNA sequence variation that is common in population.

Multifactorial

Involving a number of factors especially genetic or environmental factors.

Agglutination

Is the clumping of particles. The process in which an antigen is mixed with its corresponding antibody.

EXERCISE

Q 1

Fill in the blanks.

i) _____ is the basic unit of biological information.

ii) A sudden change in the structure of a gene is called _____.

iii) _____ is the chance of an event to occur.

iv) A cross among monohybrids is a _____ cross.

v) An individual with a homozygous genotype is called _____.

vi) Different alleles of a gene that are both expressed in a heterozygote are called _____.

vii) When a heterozygote exceeds the phenotypic expression of both the homozygotes, the phenomenon is called _____.

viii) When a single gene affects two or more traits, the phenomenon is called _____.

ix) A gene with multiple phenotypic effects is called _____.

x) The phenomenon of staying together of all the genes of a chromosome is called _____.

xi) _____ minimizes the chances of genetic recombination.

xii) _____ is an exchange of segments between non-sister chromatids of homologous chromosomes during meiosis

xiii) All chromosomes other than sex chromosomes are called _____.

xiv) _____ is the maleness determining gene in man.

xv) Type _____ of diabetes mellitus is non-insulin dependent.

xvi) Polygenic inheritance with environmental influence is called _____ inheritance.

Ans

i) Geneii) Mutation
iii) Probability
iv) Monohybrid
v) Homozygote
vi) Codominant
vii) Overdominance
viii) Pleiotropy
ix) Pleiotropic
x) Gene linkage
xi) Crossing over
xii) Crossing over
xiii) Autosomes
xiv) SRY
xv) II
xvi) Multifactorial

Q 2 Write whether the statement is true or false and write the correct statement if false.

i) In grasshopper, the male has XY and the female has XX types of sex chromosomes. **(False)**
In drosophila, the male has XY and the female has XX types of sex chromosomes.

ii) Pea is normally a self fertilizing plant. **(True)**

iii) Dihybrids are offspring of the parents who differ in one contrasting pair of trait. **(False)**
Dihybrids are offspring of the parents who differ in two contrasting pair of trait.

iv) X-linked traits pass direct from father to son. **(False)**
Y-linked traits pass direct from father to son

v) A person suffering from blue cone monochromacy cannot see blue colour. **(False)**
A person suffering from blue cone monochromacy can see blue colour.

vi) In birds and moths eggs determine sex. **(True)**

vii) A homozygote forms all gametes of the same type. **(True)**

viii) The allele for a sex limited trait is dominant in one sex but recessive in the other. **(False)**
The allele for a sex influenced trait is dominant in one sex but recessive in the other.

ix) Pattern baldness is a sex influenced trait. **(True)**

x) Carriers of haemophilia show no symptoms of the disease. **(True)**

Q 3 Encircle the correct answer from the multiple choices.

i) When a single gene has multiple phenotypic effects, the phenomenon is called:
(a) Codominance
(b) Epistasis
(c) Pleiotropy
(d) Sex-linkage

ii) What happens when both alleles of a gene pair independently express in a heterozygote?
(a) Dominance
(b) Incomplete dominance
(c) Over dominance
(d) Codominance

iii) A heterozygote offspring quantitatively exceeds the phenotypic expression of both the homozygote parents due to:
(a) Dominance
(b) Incomplete dominance
(c) Over dominance
(d) Codominance

iv) How many gene pairs contribute to the wheat grain colour?
(a) One (b) Two
(c) Three (d) Four

v) Who for the first time found white eye mutant in *Drosophila*?
(a) Morgan (b) Bridges
(c) Correns (d) De Varies

vi) Which of the following trait is transmitted directly from an affected father to only his sons?
(a) Autosomal (b) X-linked
(c) Y-linked (d) X and Y linked

vii) Which phenomenon reduces the chances of genetic recombination and variations among off springs?
(a) Linkage
(b) Crossing over
(c) Independent assortment
(d) Dominance

viii) Which of the following traits is not sex-linked recessive?
(a) Haemophilia
(b) Colour blindness
(c) Hypophosphatemic ricket
(d) tfm syndrome

ix) Which of these traits zigzags from maternal grandfather through a carrier daughter to a grandson?
(a) Autosomal
(b) X-linked
(c) Y-linked
(d) X and Y linked

x) When a haemophilic carrier woman marries a normal man, who among her offspring may be affected?
(a) All her children
(b) All her daughters
(c) Half of her daughters
(d) Half of her sons

xi) What is the risk of a colour-blind child in a family when mother is colour-blind but father is normal?
(a) 100% (b) 75%
(c) 50% (d) 25%

xii) What is the risk of a colour-blind child in a family when father is colour-blind but mother is normal?
(a) zero% (b) 25%
(c) 50% (d) 100%

Answer key

i	c	vi	c	xi	c
ii	d	vii	a	xii	a
iii	c	viii	c		
iv	c	ix	b		
v	a	x	d		

Q 4 Short Questions.

i) Differentiate between:

Ans:

Phenotype	Genotype
Physical appearance of a trait is called phenotype.	Genetic make-up of a trait is called genotype.

Homozygous	Heterozygous
When both alleles of a gene pair are similar, then it is called homozygous.	When both alleles of a gene pair are different, then it is called heterozygous.
E.g. XX in human females.	E.g. XY in human males.

Autosome	Sex chromosome
Chromosomes that do not specify sex are called autosomes.	Chromosomes that determine sex are called sex chromosomes.
e.g. 22 pairs of autosomes in human.	e.g. 2 pairs of sex chromosomes.

Allele	Multiple Allele
Alternate forms of a gene pair are called alleles.	Alleles that are more than two in number are called multiple alleles.
E.g. RRYY round or yellow for seed color trait.	e.g. ABO system I^A, I^B, i.

Incomplete dominance	Codominance
Such dominance in which phenotype of heterozygote is intermediate between two homozygotes.	Such dominance in which phenotype of heterozygote shows effects of both alleles is called codominance.
E.g. Pink color flower in 4'0 clock.	e.g. MN blood group system

Continuous variation	Discontinuous variation
Such variations which do not show clear differences and are related closely are called continuous variations.	Such variations which show clear differences are called discontinuous variations.
e.g. height, weight, skin color etc.	E.g. tongue rollers.

Gene	Allele
Gene is basic unit of biological information. Part of DNA comprising of specific sequence of nucleotides specific for each trait.	Partners of a gene pair are called alleles.
E.g. Gene for round seed shape.	e.g. RRYY

Monohybrid	Dihybrid
Offspring of the parents who differ in one contrasting pair of trait is called monohybrid.	Offspring of the parents who differ in two contrasting pair of traits is called dihybrid.
e.g. Round and yellow trait in pea plant.	e.g. Round and yellow, wrinkled and green in pea plant.

Dominance	Epistasis
Physiological effect of an allele over its partner allele on same gene locus is called dominance.	When an affect caused by a gene or gene pair at one locus interferes with or hides the effect caused by another gene or gene pair at another locus, it is called epistasis.
e.g. Complete dominance (round and wrinkled seed cross). Incomplete dominance (4'0 clock flower plant).	e.g. Bombay phenotype.

psp	Y-linked Trait
Such a trait whose genes are present only on X chromosome is called X-linked trait.	Such a trait whose genes are present only on Y chromosome is called Y-linked trait.
e.g. Gene for hemophilia.	e.g. Gene for beard growth.

Sex limited trait	Sex influenced trait
A trait that is limited to only one sex due to anatomical differences is sex limited trait.	A trait that is dominant in one sex and recessive in other is sex influenced trait.
e.g. Milk production in cows.	e.g. Male pattern baldness.

Dominant trait	Recessive trait
A trait that appears in F1 is called dominant trait.	A trait that is masked by dominant in F1 and reappears in F2 is called recessive trait.

Wild type	Mutant
Normal form of a species is called wild type.	Such type which has been produced due to any mutation is called mutant type.
e.g. Red eye female *Drosophila*.	e.g. White eye male *Drosophila*.

ii) What is a gene pool?

Ans: All the genes alleles of a trait in a breeding population collectively constitute gene pool.

iii) Was pea a lucky choice for Mendel? What would have happened if he had studied an eight character?

Ans: Pea was used as experimental by other investigators also before Mendel. If Mendel would have studied an eight character, then he would had failed to develop law of independent assortment.

iv) What is test cross? Why did Mendel devise this cross?

Ans: The cross which is used to determine the genotype of an individual is called test cross. Mendel devised test cross to test the genotype of an individual showing a dominant phenotype.

v) What would happen if alleles of a pair do not segregate at meiosis? How would it affect the purity of gametes?

Ans: If segregation does not occur, then chromosome number will be affected in next generation. Due to absence of segregation, variations will not be present.

vi) If the alleles do not assort independently, which type of combination is missing in the progeny?

Ans: Recombinant types will be missing and only parental combinations will be present.

vii) Why has each gamete equal chance of getting one or the other allele of a pair?

Ans: Because of independent assortment.

viii) Does the dominant allele modify the determinative nature of its recessive partner? What sort of relationship do they have?

Ans: Yes, they have physiological relation of dominance.

ix) Which type of traits can assort independently?

Ans: Such traits whose alleles are riding on non-homologous chromosomes. For example seed color and shape in pea plant.

x) Why does the blood group phenotype of a person remain constant throughout life?

Ans: Due to their predetermined genetic relation. ABO blood group system has four different phenotypes which are distinct from each other on the basis of specific antigens on the surface of RBC's. These antigens are encoded by single polymorphic gene I on chromosome 9. Once these antigens start expressing during early life of an individual, they will express throughout a life without any change.

xi) What is universal blood donor?

Ans: Phenotype O can be used as donor for small transfusions to A, B and AB recipients because donor's antibodies are quickly absorbed by other tissues or greatly diluted in the recipient's blood stream. O blood group individuals are called universal donors.

xii) How can you protect the baby against Rh – incompatibility?

Ans: Sometimes, a mild ABO incompatibility protects the baby against a more severe Rh incompatibility. If O^- mother conceives A^+ or B^+ baby any foetal A or B type RBC entering the mother's blood are quickly destroyed by her anti – A or B antibodies, before she can form anti–Rh antibodies.

xiii) Which types of genes do not obey law of independent assortment?

Ans: Such genes which are present on same chromosome and are linked i.e. linked genes.

xiv) How can linked genes be separated from each other?

Ans: Linked genes can be separated through crossing over.

xv) What is multifactorial inheritance?

Ans: When polygenic inheritance is influenced by environment then it is called multifactorial inheritance.

xvi) What is MODY?

Ans: About 2% - 5% of type II diabetics get the disease early in life, before 25 years of age. It is called maturity onset diabetes of the young (MODY). MODY can be inherited as an autosomal dominant trait. About 50% of cases of MODY are caused by mutations in glucokinase gene.

xvii) Can a child have more intelligence (IQ score) than his parents?

Ans: Intelligence is controlled by polygene. Similarly environment can promote intelligence. So a child can have more intelligence (IQ score) than his parents.

Q 5 Extensive Questions.

i) What is incomplete dominance? Explain with examples.

Ans (see article 22.3.2)

ii) Define Mendel's law of segregation. Explain it with an example.

Ans (see article 22.2.1)

iii) Define Mendel's law of independent assortment. Explain it with an example.

Ans (see article 22.2.2)

iv) Define probability. Derive 9:3:3:1 F2 ratio of independent assortment through product rule.

Ans (see article 22.2.2)

v) What is codominance? Explain the phenomenon of codominance with an example.

Ans (see article 22.3.3)

vi) Define multiple alleles. Describe multiple allelic blood group system of man.

Ans (see article 22.4.)

vii) What is Rh factor? Describe the genetic basis of Rh blood group system of man.

Ans (see article 22.4.2)

viii) What is erythroblastosis foetalis? Discuss the adverse effect of Rh-incompatibility. Also suggest a therapy to avoid Rh sensitization of an Rh negative mother married to an Rh positive man.

Ans (see article 22.4.2.a)

ix) Define epistasis. Explain epistatic gene interaction with examples.

Ans (see article 22.5)

x) What is a pleiotropic gene? Discuss pleiotropy with examples.

Ans (see article 22.6)

xi) What are polygenes? Explain polygenic inheritance.

Ans (see article 22.7)

xii) What is crossing over? Define recombination frequency and explain its significance.

Ans (see article 22.9)

xiii) What are sex chromosomes? Discuss the chromosomal patterns of sex determination in organisms.

Ans (see article 22.10)

xiv) Compare chromosomal determination of sex between Drosophila and human.

Ans (see article 22.10.)

xv) Define gene pool. Explain the concept of gene pool in a same population.

Ans (see article 22.1)

xvi) What is sex linkage? Explain T.H. Morgan's study of sex-linkage in drosophila.

Ans (see article 22.11)

xvii) Compare pattern of inheritance of an X-linked dominant trait with an X-linked recessive trait in humans.

Ans (see article 22.11)

xviii) Explain diabetes mellitus and its genetic basis.

Ans (see article 22.12)

xix) Discuss the genetics of colour-blindness or haemophilia.

Ans (see article 22.11.2)