CHAPTER
22

INHERITANCE

Key Concepts

22.1 Mendelian inheritance
22.2 Exceptions to mendelain inheritance
22.3 Blood group system
22.4 Poly genetic inheritance and epitasis
22.5 Gene Linkage and crossing over
22.6 Sex-determination
22.7 Sex Linkage

EXERCISE

SECTION I: Multiple Choice Questions

Select the correct answer from the following choices.

1. Mendels law of independent assortment can be demonstrated by:
   (a) test cross (b) back cross
   (c) monohybrid cross (d) dihybrid cross

2. Investigators do a dihybrid cross between two heterozygous and get about a 3:1 ratio among the offspring. The reason must be due to:
   (a) polygenes (b) pleiotropic genes
   (c) linked genes (d) epistatic gene

3. Whether an allele is dominant or recessive depends on:
   (a) how common the allele is relative to other alleles
   (b) whether it is inherited from the mother or the father
   (c) which chromosome it is on
   (d) whether it shows expression in heterozygous state or not

4. All the offspring of a white hen and a black rooster are grey. The

(a) linkage
(b) sex linkage
(c) independent assortment
(d) incomplete dominance

5. A man who has types B blood and a woman who has type A blood could have children of which of the following phenotypes?
(a) A or B only
(b) AB only
(c) AB or O only
(d) A, B, AB or O

6. A heterozygous red-eyed female Drosophila mated with a white-eyed male would produce:
(a) red-eyed females and white males in the F_1
(b) white-eyed-females and red eyed males in the F_1
(c) all white-eyed females and half red and half white eyed males in the F_1
(d) half red and half white-eyed female as well as male in the F_1

7. When mothers anti-RH negative antibodies seep through placenta into blood circulation of foetus they start _____ of RBC of the foetus.
(a) plasmolysis
(b) crenation
(c) haemolysis
(d) deplasmolysis

8. All chromosomes other than sex-chromosomes are called:
(a) autosome
(b) mesosome
(c) polysome
(d) lysosome

9. If a man of M blood group marries a woman of N blood group all their children will have:
(a) M blood group
(b) N blood group
(c) O blood group
(d) MN blood group

10. Rh blood group system is encoded by three genes C, D and E which occupy _____ tightly linked loci:
(a) four (b) three (c) five (d) two

11. Albinsim is a recessive gene. A woman with albino father marries an albino man. The proportion of her progeny is:
(a) 2 normal: 1 albino
(b) all normal
(c) all albino
(d) 1 normal: 1 albino

12. Phenomena of an allele of one gene suppressing the activity of allele of another gene is called:
(a) dominance
(b) epistasis
(c) suppression
(d) inactivation

13. If red eyed (dominant) fly is mated with white eyed (ressessive) fly, the ratio of red to white eyed in F_2 generation would be:
(a) 3:1 (b) 2:2 (c) 2:1 (d) 1:3

(a) 4 alleles in which A is dominant
(b) 3 alleles in which A and B are co-dominant and i is recessive
(c) 3 alleles in which none is dominant
(d) 3 alleles in which A is recessive

15. Genes located on same locus but having different expressions are:

(a) multiple alleles (b) oncogenes
(c) polygenes (d) condominants

Answer

1	(d)	2	(c)	3	(d)	4	(d)	5	(d)
6	(d)	7	(c)	8	(a)	9	(d)	10	(d)
11	(d)	12	(b)	13	(a)	14	(b)	15	(a)

SECTION II: Short Questions

Give short answers of the following questions.

Q1. What is a scope of independent assortment in variation?

Answer

Beside mutation and crossing over (which are sources of variation), independent assortment of trail is also a major source of variations in successive generations. It is only due to crossing over and independent assortment of trails that the characteristics may appear in new combination in next generation which is often seems necessary for adoptions in varing environment.

Q2. Differentiate between: Incomplete dominance and codominance, gene and allele polygenic and epistasis.

Answer

Codominance

1) Definition

Different alleles of a gene that are both expressed in a heterozygous condition are called codominant and the phenomenon is called codominance.

Explanation

The phenotype of heterozygote is distinct in quality from those of the two homozygotes. It is not an intermediate quantitative expression like incomplete dominance. Each allele of the gene pair is associated with a different substance, e.g.,

$$\text{Allele } A_1 \xrightarrow{\text{produce}} \text{Subs tance X}$$

$$\text{Allele } A_2 \xrightarrow{\text{produce}} \text{Subs tance X}$$

Codominace occurs when both the alleles express independently in heterozygote (A_1A_2) and form their respective products X and Y. The codominant heterozygote

2) Incomplete dominance

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

Explanation

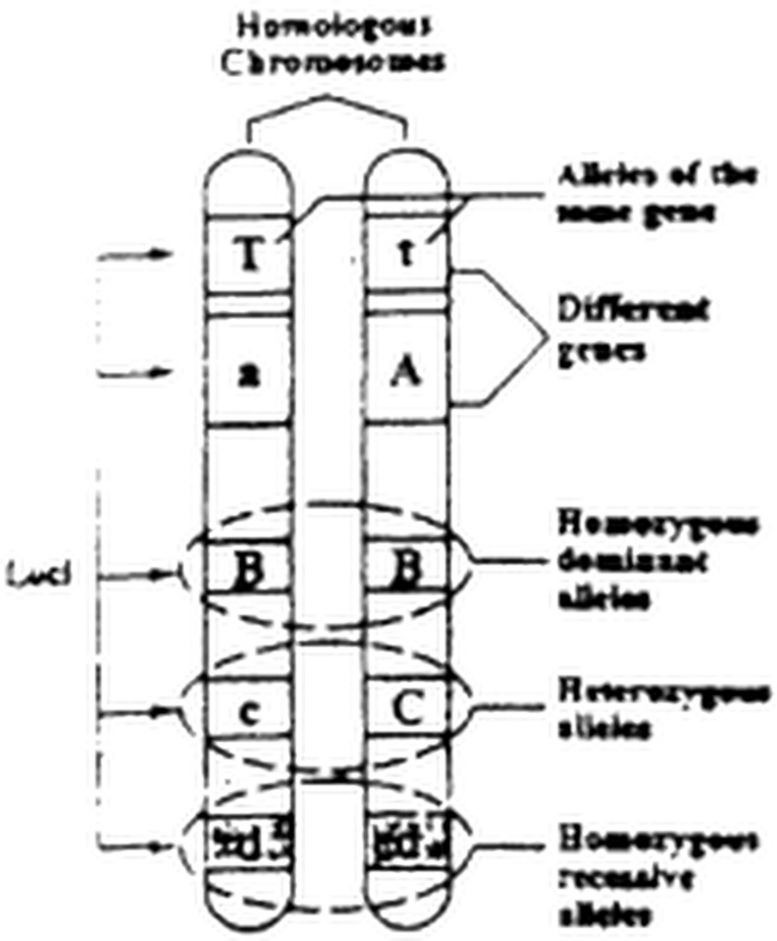
In 1899 Carl Correns was working on a flowering plant named 4 O'clock. When he crossed a true breeding red flowered plant with a true breeding white flowered 4 O'clock, all the F_1 hybrids had pink flowers. This novel phenotype had a shade intermediate between those of the parents due to an intermediate amount of pigment in petals.

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. Red was homozygous for red alleles, and white was homozygous for white alleles.

But when allele for red and allele for white are present together in the same plant, neither of them masked the effect of other: rather these alleles showed incomplete dominance in the form of pink colour.

3) Genes

Gene is the basic unit of biological information. Hereditary characteristics pass from parents to offspring through genes in their gametes. In fact DNA stores all sorts of biological information coded in the sequence of its bases in a linear order, and genes are actually parts of DNA comprising its base sequences.



Function of genes:

Genes are responsible for inherited resemblances as well as distinctive variations among generations. When these pass in the form of intact parental combination between generations, inherited similarities are conserved; but when these shuffle, mutate or juggle (fit in) with each other, variations emerge.

5) Epistasis

When and effect caused by a gene or gene pair at one locus interfers with or hides the effect caused by another gene or gene pair at another locus such as phenomenon of gene interaction is called epistasis.

In such interactions, the gene which suppresses or masks the effect of action of a gene at another locus is known as epistatic gene or inhibiting gene and the gene which is suppressed is known as hypostatic gene.

6) Polygenic inheritance

The traits which cannot be encoded by a single gene with two alleles. Even a few

influencing the same trail in an additive away. These quantitative traits are called polygenic traits and inheritance called polygenic inheritance. All traits which control quantitative traits called polygenes, which have positive or negative effect on characters.

Q3. Give an example of incomplete dominance codominance, gene, alleles, multiple alleles, polygenic and epistasis.

Answer

I) Incomplete Dominance

When neither of the two alleles expresses independently in heterozygous state, rather a blend of expression of both alleles is appeared is called incomplete dominance. In 1899 Carl Correns was working on a flowering plant named 4 O'clock. When he crossed a true breeding red flowered plant with a true breeding white flowered 4 O'clock, all the F, hybrids had pink flowers. This novel phenotype had a shade intermediate between those of the parents due to an intermediate amount of pigment in petals. When Correns self-fertilized F_1 pink, the F_2 showed all three phenotypes of flower in the ratio of 1 red: 2 pink: 1 white. Red was homozygous for red alleles, and white was homozygous for white alleles. But when allele for red and allele for white were present together in the same plant, neither of them masked the effect of other, rather these alleles showed incomplete dominance in the form of pink colour. As there is no truly dominant allele, the usual capital and small letter distinction for dominant and recessive trait is not necessary. Both the alleles are represented by the same letter "R" but are numbered differently to distinguish white from red.

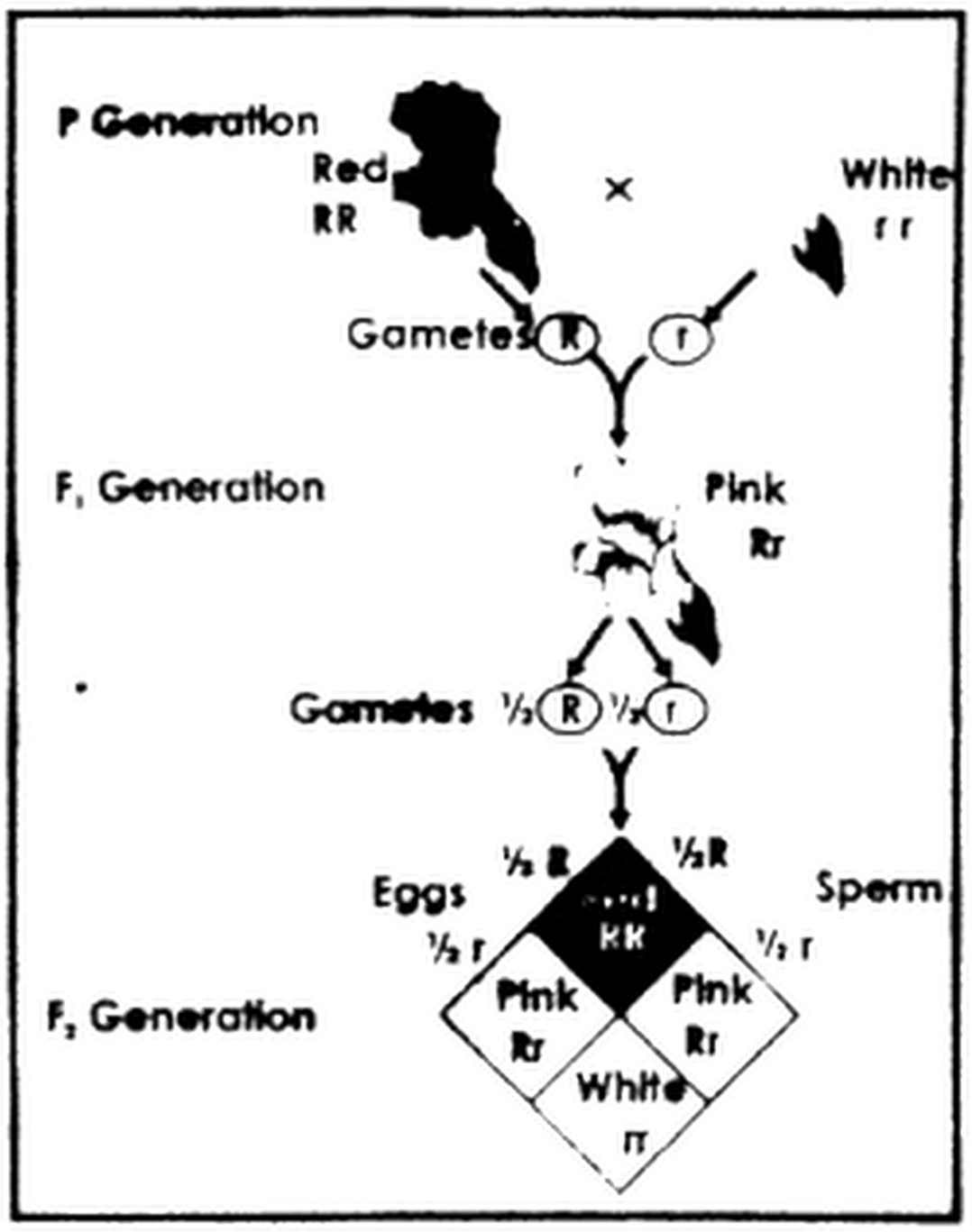


Fig. Incomplete dominance

Allele for red is designated as R, and the allele for white as R_2. Punnett square indicates that the phenotypic ratio is the same as the genotypic ratio.

ii) Co-Dominance

Co-dominance is the equal expression of both alleles that result in a mixed phenotype. This kind of co-expression is most easily seen in some biochemical phenotypes like blood types heterozygose.

homozygotes. This phenotype is not intermediate. Each allele of gene pair is associated with different substance.

Allele A_1, $\xrightarrow{\text{Produce}}$ Substance X

Allele A_2, $\xrightarrow{\text{Produce}}$ Substance Y

Co-dominance occurs when both the alleles express independently in heterozygote ($A_1.A_2$) and form their respective products X and Y. The co-dominant heterozygote would have both substances at same time.

MN Blood Type

Landsteiner and Levine discovered MN blood types in man on basis of specific antigens present on RBC. The function of these antigens is to produce the specific antibodies. Basically there are two types of antigens. M antigen which is produced by gene L^M for M phenotype and N antigen produced by L^N for N phenotype. While MN antigens are produced simultaneously by allele L^M and L for MN phenotype.

Phenotype	Genotype	Antigens on RBC
M	$L^M L^M$	M
N	$L^N L^N$	N
MN	$L^M L^N$	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.

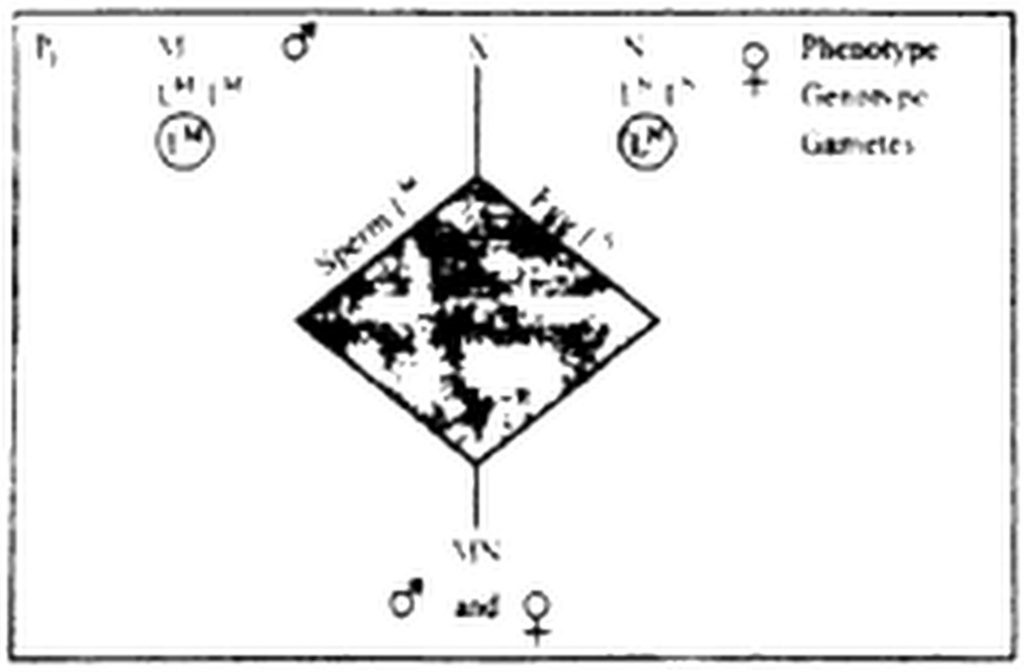


Fig. Codominance in MN Blood group alleles

ii) Multiple alleles

All the altered alternate formes of gene, whose number is more than two, are called multiple alleles.

ABO, Blood Group System In Man

It is the first discovered as multiple allelic system.

A person having antigen A has blood group A; a person having antigen B has blood group B;a person having both the antigens A and B has blood group AB; but a person having neither antigen A nor B would have blood group O.

Bernstein explained the genetic basis of ABO system in 1925. This blood group system

alleles I^A, I^B, and I.

Antigen	Blood group	Genotype
A	A	I^AI^A or I^Ai
B	B	I^BI^B or I^Bi
AB	AB	I^AI^B or I^Bi

Allele I^A specifies production of antigen A and allele I^B specifies production of antigen B but allele i does not specify any antigen. Allele I^A and I^B are codominant to each other, because each expresses equally in I^A I^B heterozygote to produce AB phenotype. But allele (i) is recessive to both I^A and I^B.

iv) Gene

The smallest part of DNA is called genes which is a basic unit of biological information.

Explanation

The biological information in DNA is stored and coded in the sequence of its bases. The position on a chromosome where a gene is located is often referred to as a locus.

- As genes are passed from one generation to another so it can pass the same characters from parents to offsprings and can thus produce inherited resemblances but they are also responsible for producing may distinctive variations as well in next generation. Variations are produced due to the shuffling or mutation of the genes.

v) Alleles

A single gene may have alternative form which is called allele, or partners of gene pair are called allele.

Explanation

Each allele of a gene pair occupies the same gene locus on its respective homologue. Both alleles on one locus may be identical or different from each other. The specific alleles or genes contained in a cell are called *genotype*. The physical trait that occurs as a result of specific genotype is referred as *phenotype*.

Example

The concept of phenotype and genotype can be understood by taking an example of flower colour. Flower colour is a trait and red and white are its two phenotypes. Each form of expression is determined by different allele of colour gene. Allele R is the determiner for redness while 'r' is determiner for whiteness.

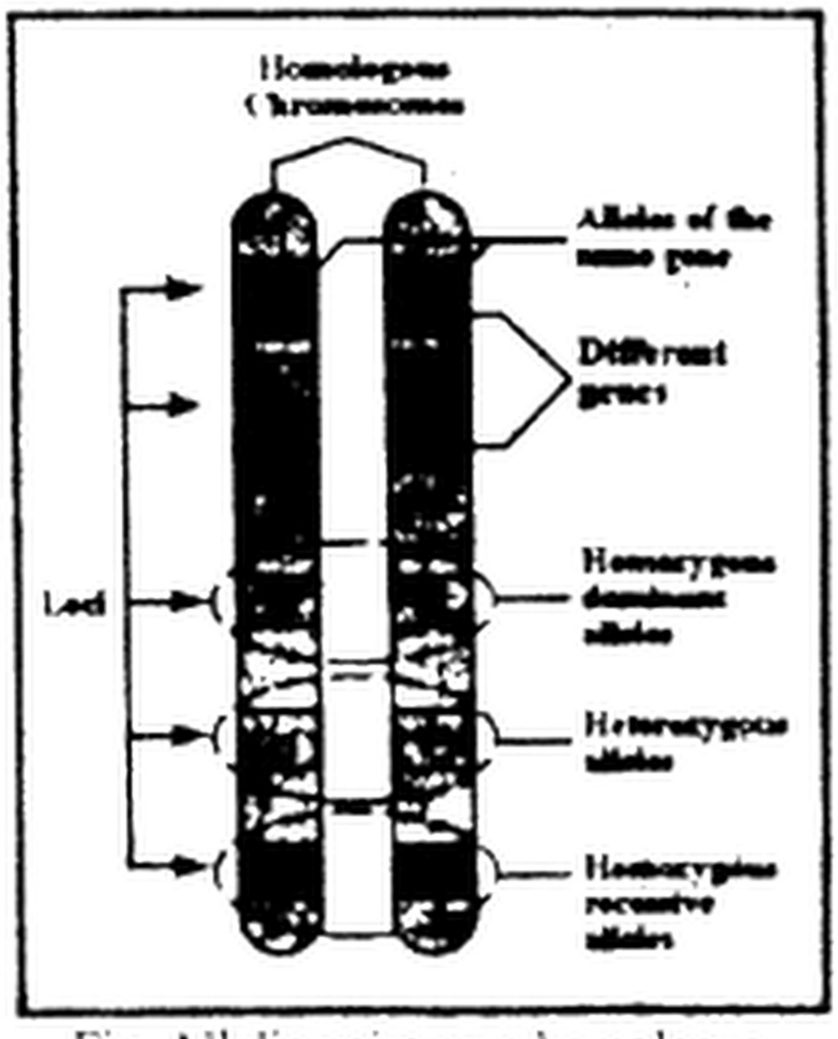


Fig. Allelic pairs on a homologus pair of chromosomes

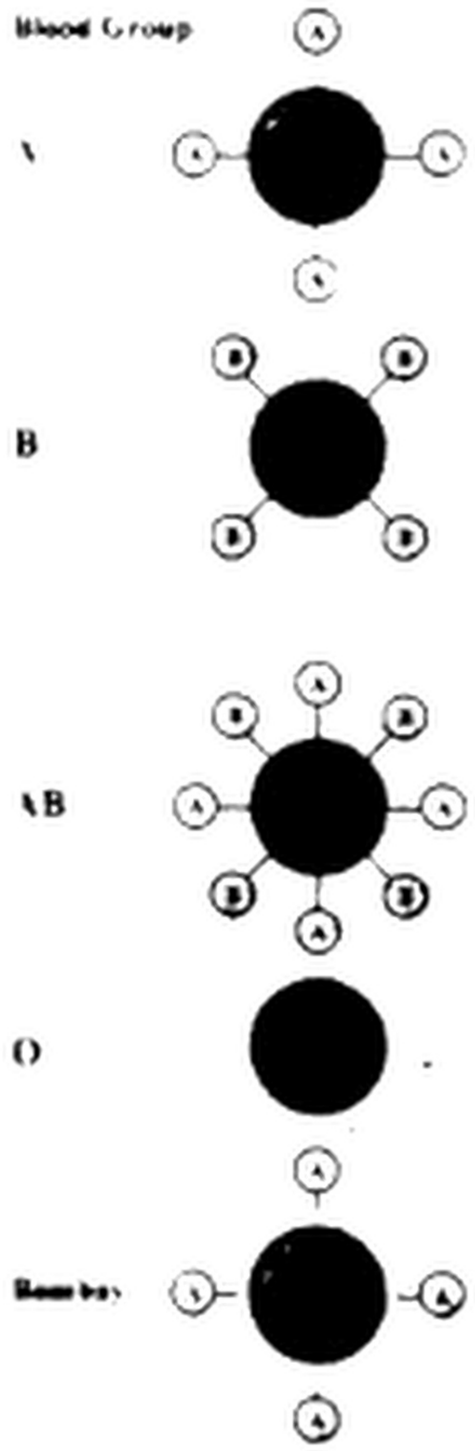
iv) Epistasis

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 the gene interaction is called epistasis.

Epitasis must not be confused with dominance. Dominance is the relationship between alleles of the same gene occupying the same locus but epitasis is the interaction between different gene occupying loci. Following are the two examples.

Bombay phenotype

The expression of ABO blood type antigen by I^A or I^B gene depends upon the presence of another gene H. ABO locus is on chromosome 9, while H locus is on chromosome 19. H gene changes a precursor substance into substance H. It produces an enzyme that inserts a sugar on to a precursor glyocoprotein on the surface of RBC only the antigen or antigen B specified by I^A or I^B gene could attach 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 thus B and B antigens cannot adhere to their RBC and fall away. 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. Their phenotype is called Bombay phenotype.



vii) Polygenic Inheritance

Even a few multiple alleles of a single gene cannot make a large number of phenotype. In fact, 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 therefore called polygenic traits.

All the genes that control a quantitative trait are called polygenes which have a small positive or negative effect on the character. Polygenes supplement each other and sum of positive and negative effect of all individual polygenes produce quantitative phenotype of a continuous varying traits.

Example

Wheat grain colour

Wheat grain colour is a good example of polygenic (multiple gene) inheritance. Wheat grains show a continuous variation in colour from white to dark red.

Approximately seven different phenotypes are found in wheat population all over the world. Some grains are white, some are deep red but most grains have shades in between from light pink to moderately dark red. 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 F_1 grains had light red colour intermediate between two

It seemed as if it was a case of incomplete dominance. But when F_1 grains were grown to mature plants and crossed with each other, F_2 grains had exactly seven shades of colour in the following ratio:

Dark red	Moderately Dark red	Red	Light red	Pink	Light Pink	White
1	6	15	20	15	6	1

Three different gene pairs, Aa, Bb, Cc at three different loci contribute to the wheat grain colour. Each individual would contain six alleles for the trait. Alleles A, B and C codes for an equal amount (dose) of red pigment, which is a positive effect. But none of a, b and c encode red pigment, which is a negative effect.

If all the six allele code for red pigment (AABBCC), the grain is dark red; when none of the six allele encodes red pigment (aabbcc), the grain is white. When a grain has one allele for red pigment (Aabbcc or aaBbcc or aabbCc) its colour is light pink; if it has two alleles, for the pigment (AaBbcc or aaBbCc orAabbCc) it is pink, if it has three pigment alleles (AaBbCc or AABbcc or AabbCC), it will be light red. Similarly four alleles colour dose (AA BBcc or aaBBCC or AAbbCC) will make red and five alleles colour dose (AABBCc or AABbCC or AaBBCC) will produce moderately dark red grain.

Thus colour phenotypes of grains depend upon the number of pigment producing alleles (A, B, and C). Environmental factors like light, water and nutrients also influence the amount of grain colour.

Q4. Name any four human blood group systems.

Answer

Blood groups are determined by the type of antigens. There are four different phenotypes of blood groups.

1) A 2) B 3) AB 4) O

Q5. Associate multiple alleles with ABO blood group system.

Answer

All the altered alternate from of gene, whose number is more than two, are called multiple alleles.

Multiple alleles are produced by gene mutation. Some genes may have as many as 300 alleles, but individuals have only two of those alleles. Why? Because individuals have only two biological parents. We inherit half of our genes (alleles) from mother, and the other half from father, so we end up with two alleles for every trait in our phenotype.

An excellent example of multiple allele inheritance is human ABO blood group system. It is the most well-known blood group system which was discovered in 1901 at the University of Vienna by Karl Landsteiner in the process of trying to learn why blood transfusions sometimes cause death and at other times save a patient. In 1930, he received the Nobel Prize for his discovery of blood types.

chimpanzees, baboons, and gorillas. There are four principal types: A, B, AB, and O, there are two antigens and two antibodies that are mostly responsible for the ABO types. The specific combination of these four components determines an individual's type in most cases.

Table: ABO blood groups

ABO Blood type	Antigen (A or B)	Antibodies (Anti-A or Anti-B)	Donors
A	A	Anti-B	A,O
B	B	Anti-A	B, O
AB	Both A&B	None	A,B,AB,O
O	None	Both Anti-A or Anti-B	Only O

For example, people with type A blood will have the A antigen on the surface of their red blood cells (as shown in the table above). As a result, anti-A antibodies will not be produced by them because they would cause the destruction of their own blood.

Q6. Why O^+ individuals are regarded as universal donor and AB^+ individuals are called universal recipients.

Answer

The persons with blood group O can receive blood only from type O because they have both antibodies that can react with any antigenic blood (type A, B or AB). But can donate blood to anyone as they do not have any antigen to interfere with recipient blood. Therefore are called universal donar.

The persons with blood group AB can receive any blood since they do not have antibody to react with donars blood hence they are called universal recipients.

Q7. What is Rh factor?

Answer

ABO blood group system is further differentiated by a +ve or –ve sign. This positive or negative sign, refers to the presence or absence of antigen called Rh–factor.

Q8. How a blood is distinguished as positive or negative blood group?

Answer

If a persons blood group has Rh-factor (antigen) then it would be assigned as +ve and if does not has Rh-antigen than would be assigned as negative (–ve) blood group.

Conclusion

This +ve or –ve sign with blood group refers to the presence or absence of antigen called Rh-factor.

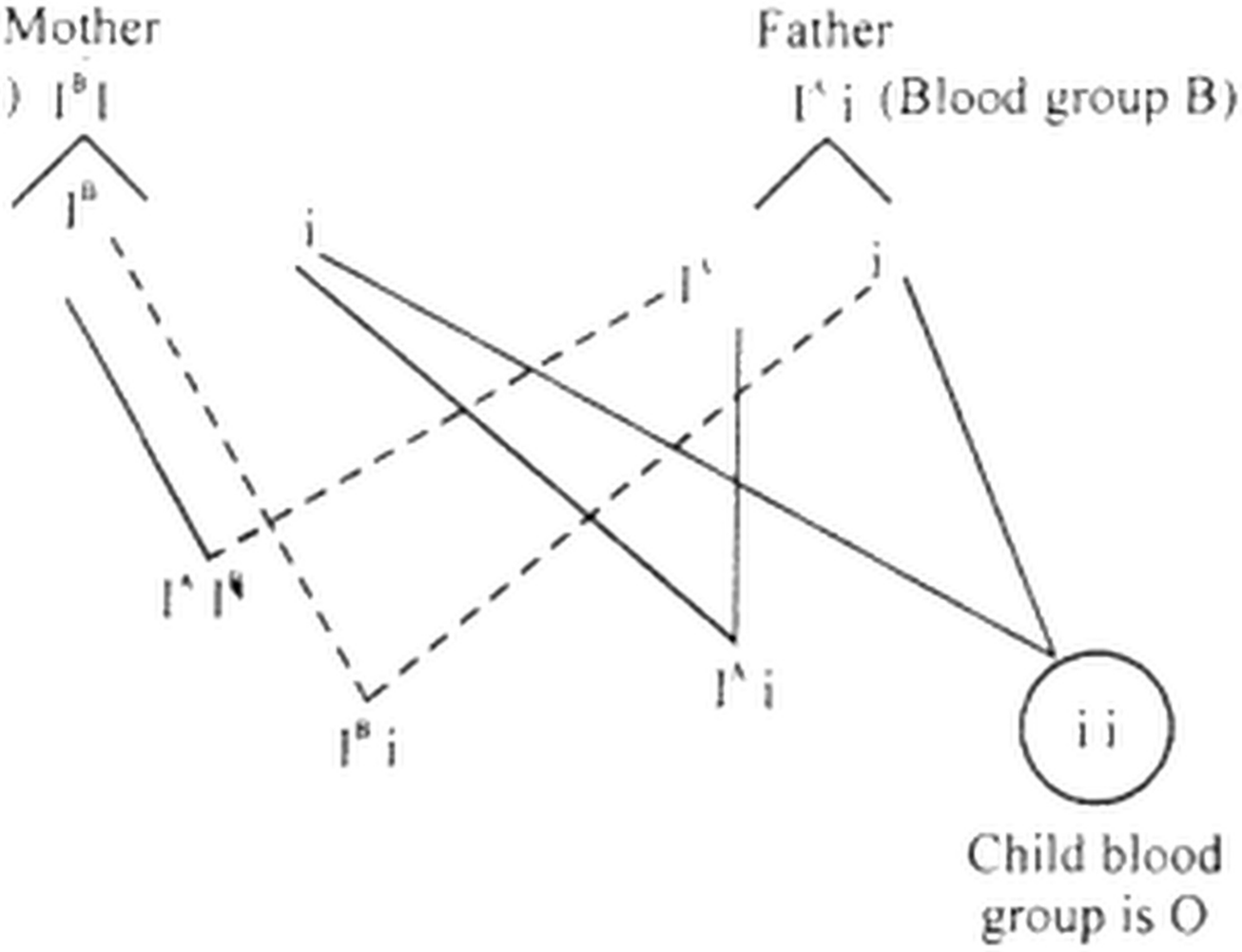
Genetic Basis

Rh – blood group system is encoded by three genes C, D and E, which occupy two tightly linked loci. Alleles of gene D occupy one locus called locus D, while genes C

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^-.

Q9. A woman with blood group B has a child with blood group O. What are the genotypes of the mother and the child? Which genotype could the father not have?

Answer



The father could not have genotype $I^A B^A$.

Q10. List any five polygenic inheritances in man.

Answer

i) Human skin colour
ii) Human weight
iii) Human height
iv) Human I.Q level
v) Human nose length

Q11. Why is human male refered as hetro-gametic?

Answer

Male are hetero gametic in humans because it forms two types of sperms. Half the sperms have X chromosome while the other half are having Y chromosomes.

Q12. If a woman who is not a carrier for genes for haemophilia is married to a man who is haemophilic, what percentage of their offspring could be expected to be haemophilic?

Answer

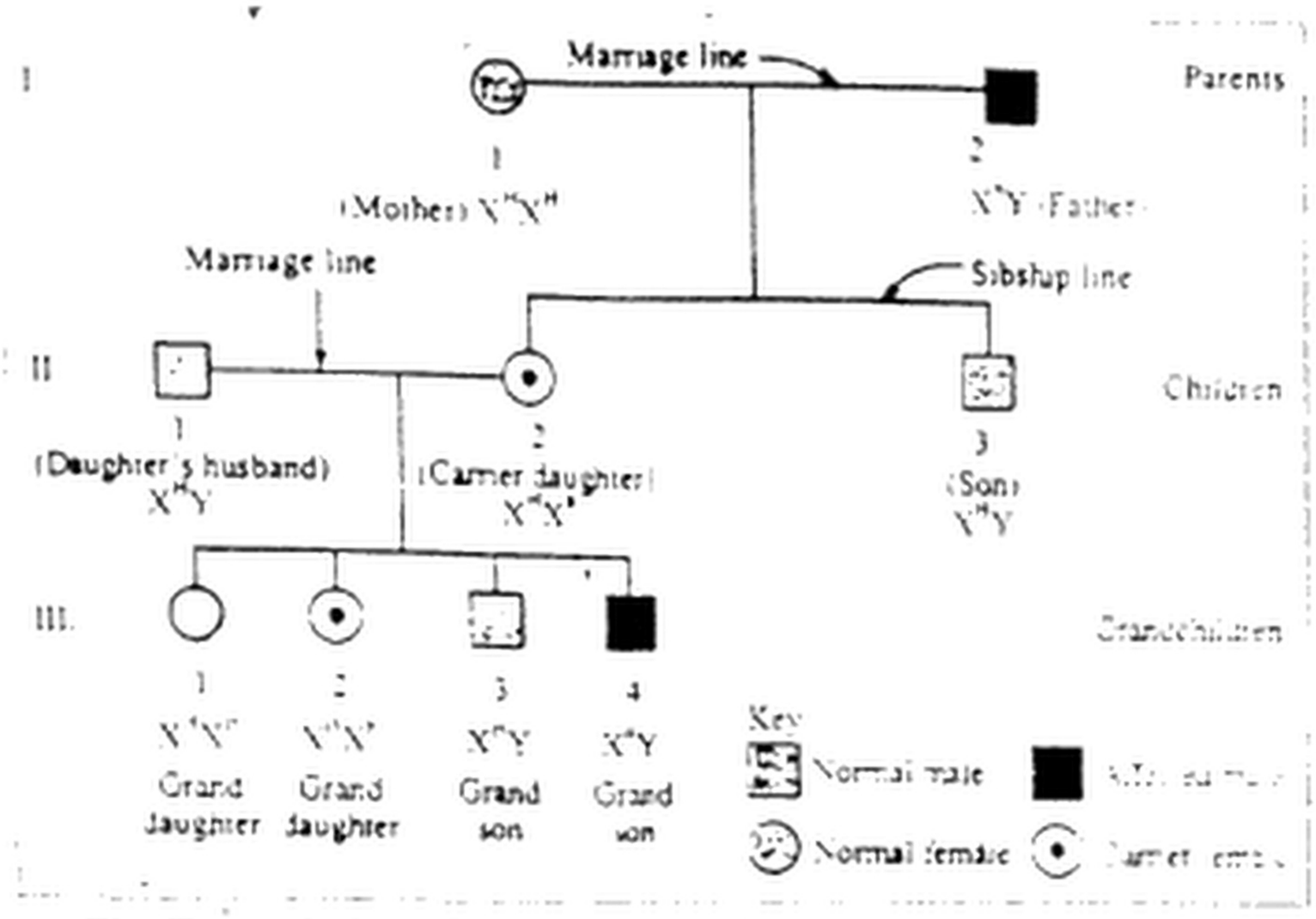


Fig. Transmission of X-linked recessive trait (haemophlia) in man

Q13.Under what circumstances is it possible for father and son to be to suffer from haemophilia?

Answer

A trait whose gene is present on X chromosome is called X – linked trait. X – linked traits are commonly referred to as sex-linked traits. A gene present only on X chromosome, having no counterpart on Y chromosome is called X – linked gene.

Pattern of Sex-linked Inheritance

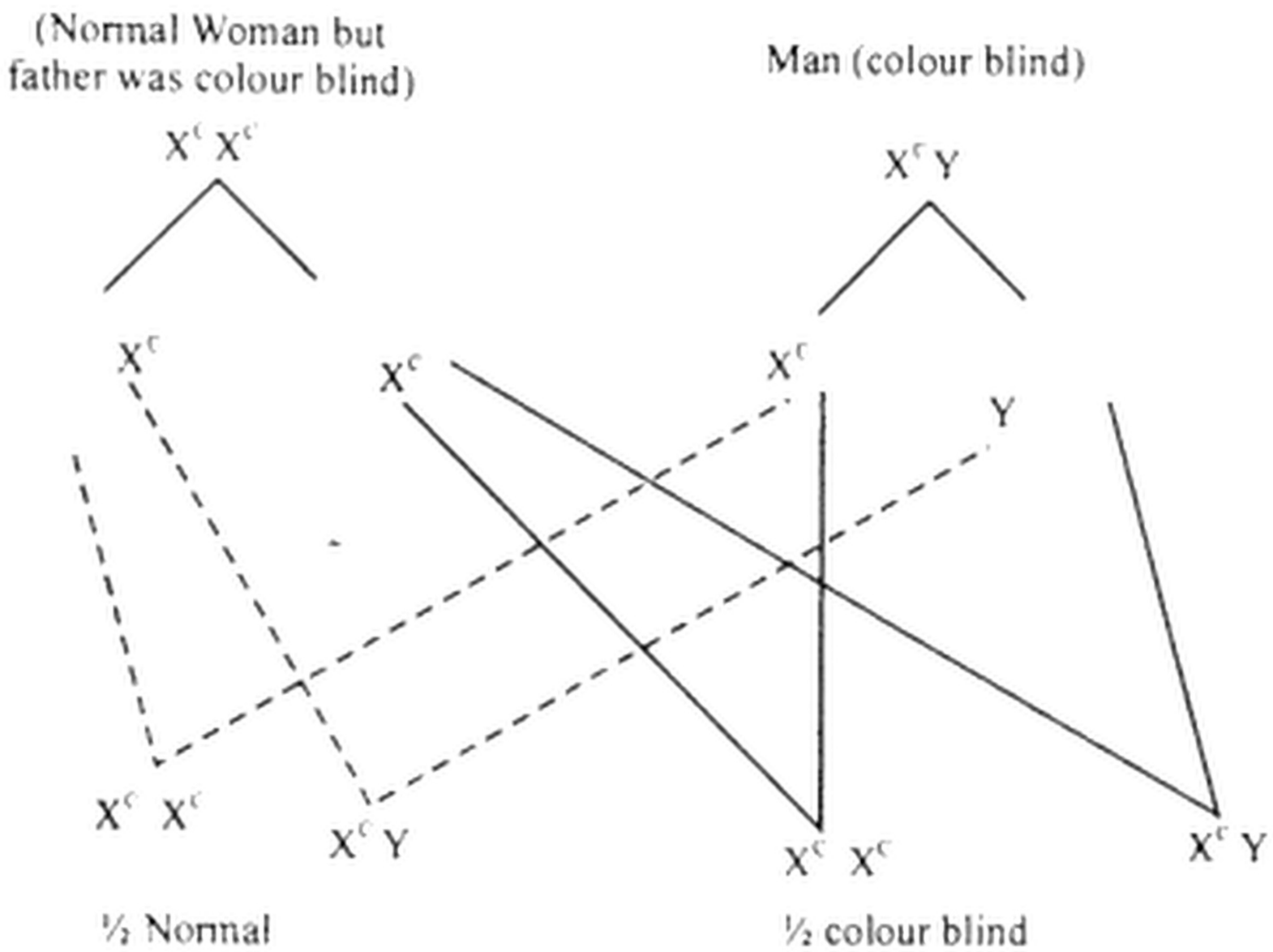
Sex-linked inheritance follows a very specific pattern. As a son inherits his X chromosome only from his mother, and a daughter gets a X chromosome from each parent, an X – linked trait passes in a crisscross fashion from maternal grandfather (P_1) through his daughter (F_1) to the grandson (F_2). It never passes direct from father to son because a son inherits only Y chromosome from father.

Haemophilia is an example of sex-linked inheritance.

So a son inherits haemophilic gene from his mother and this x linked haemophilic trait passes in a crisscross fashion from maternal grandfather (P_1) to his daughter (F_1) and to the grand son (F_2).

Q14.Red green colour blindness is caused by sex Linked recessive allele. A colour blind man marries a woman with normal vision whose father was colour blind. What is the probability that their first son will be a colour blind?

Answer



50% of their children will have normal colour vision.

Q15.Describe sex influenced trait in man.

Answer

Sex Influenced Trait

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

Q16.Describe sex limited traits in man.

Answer

Sex Limited Trait

A sex-limited trait is limited to only one sex due to anatomical differences. 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. Genes for milk yield in dairy cattle affect only cows.

Similarly 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.

Q17.Why Rh incompatibility could be dangerous to the developing foetus and mother?

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.

Erythroblastosis foetalis - Maternal - foetal Rh Incompatibility

Maternal– foetal incompatibility results when an Rh^- woman, married to an Rh^+ man conceives a child who is Rh^+. If the man's genotype is DD, all of their offspring (Dd) will be Rh^+. If the man's genotype is Dd, half of their offspring with Dd genotype will be Rh^+.

If 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. When mother's anti – Rh antibodies seep through placenta into blood circulation of foetus, they start haemolysis (break down / bursting) of RBC of foetus. As this destruction continues, the foetus becomes anaemic (weak due to deficiency of blood).

Effect of the Diseases

i) The anaemic foetus starts to release many immature erythroblasts into his blood stream. That is why this haemolytic disease of the new born is called erythroblastosis foetalis.

ii) This anaemia may lead to abortion or still birth.

iii) Even if the pregnancy continues, the liver and spleen of the foetus swell as they rapidly produce RBC.

iv) The breakdown product of RBC called bilirubin also accumulates in the foetus. Bilirubin damages his brain cells and turn his skin and whites of the eye yellow. This condition is jaundice.

v) The baby if born alive, suffer from severe haemolytic anaemia and jaundice. Such baby's blood should be immediately replaced by Rh^- blood free of anti– Rh antibodies. Later on baby will develop Rh^+ blood himself.

Degree of risk of Rh – Incompatibility

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. The anti – Rh antibdies persist in mother's blood for a long time and are persistent risk for the next Rh^+ foetus.

Treatment

Rh sensitization (make sensitive) 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

Q18. How polygenic inheritance is related to epistais.

Answer

Relationshipof Epistasis with Polygenic Inheritance

In epistasis the expression of a gene is controlled by the expression of another gene. For example, in mice there are different genes for fur colour, but there is also a gene that controls whether or not any pigment is produced at all. This is very much related to the phenomenon where one trait is affected by the expressions of many genes, the polygenic inheritance. For example, people vary greatly in traits such as height, skin colour and nose length because there are many genes that can affect the growth of a person. The only distinction between the two phenomena is that in epistasis the traits show discontinuous variations among alternative phenotype whereas, in polygenic inheritance the traits show a long range of alternative phenotypes with continuous variations.

Q19. Show that if a dihybrid cross between two heterozygous involves an epistatic gene? You do not get a ratio of 9:3:3:1.

Answer

Pigment Phenotypes in Foxgloves

Pigment phenotype in foxgloves (*Digitalis purpurea*) is an excellent example of epistasis in plants. It is determined by three separate gene loci (*M, D* and *W*). M codes for an enzyme that synthesizes anthocyanin, the purple pigment seen in these petals; *m* produces no pigment and produces the phenotype albino with yellowish spots. A plant with mm genotype will show white with yellowish spots (albino) irrespective of any genotype at other loci. Since M is dominant over m therefore a plant having *M/M* or *M/m* genotypes will show light purple colour. *D* is an enhancer gene of anthocyanin, resulting in a darker pigment; d does not enhance. Since *D* is dominant over d therefore a plant having *D/D* or *D/d* genotypes along with M/M or M/m genotypes will show dark purple colour. At the third locus, wallows pigment deposition in petals, but *W* prevents pigment deposition except in the spots and so results in the white, spotted phenotype. The plant having w/w along with other pigment producing genotype can show uniform pigmented phenotype otherwise; in the absence of w/w white with purple spotted phenotype is appeared.

Fig. Pigment phenotype in foxglove: A) Albino or white with yellow spots. B) Dark purple. C) White with purple spots. D) Dark purple

Q20. Differntiate between polygenic inheritance and multiple allelic trait.

Answer

There are two alleles per gene, which control anybody trait. But many genes have more than two alleles all them are called multiple alleles. Multiple alleles are produced by

All multiple alleles of a trait occupy the same locus. Some traits may be controlled by as many as 100 or more multiple alleles but each individual carries only two of them as each locus is twice represented in a diploid individual. For example if a trait is controlled by three multiple alleles i.e. A_1, A_2 and A_3. But every individual carries any two of them like A_1A_1, A_1A_2, A_1A_3, A_2A_2, A_2A_3, or A_3A_3.

The ABO blood group is an example of multiple alleles.

Polygenic Traits: are controlled by more genes at a time and each gene plays small or negative effect on character. These genes are present on separate loci of chromosomes.

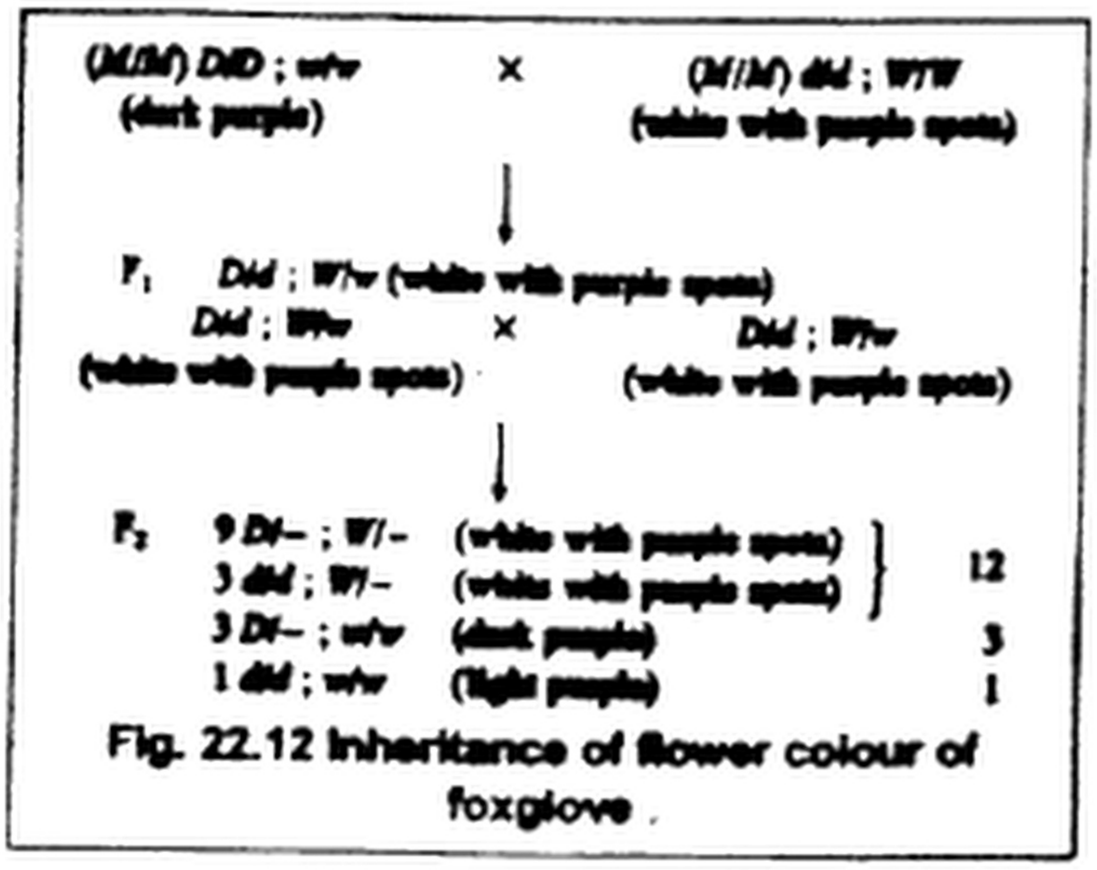


Fig. 22.12 Inheritance of flower colour of foxglove .

Q21.Why are the scientists to famous?

Answer

a) Gregor Mendel

He introduced a new theory of inheritance based on his experimental work with pea plants.

Gregor Mendel is also known as father of Genetics.

b) Carl Correns

A German botanist, who discovered the phenomenon of incomplete dorminance.

c) Karl Landsteiner

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

d) Nilson Ele

Studied the genetics of wheat grain colour.

e) Thoms H.Morgan

Studied about 85 pairs of contrasting traits in fruit fly and provided experimental evidence in support of chromosomal theory of heredity through discovery of sex Linkage in fruit fly Drosophila melanogaster.

f) Bersterin

In 1952 Bersterin explained the genetic basis of ABO blood group system.

SECTION III: Extensive Questions

Q1. Explain Mendles law of independent assortment. Express limitation in the law and its usefulness.

Answer

Mendels Law of Independent Assortment

Mendel identified the concept of independent assortment of alleles during his observations on inheritance of two traits simultaneously in dihybrid cross (a cross between two individuals which are different on two traits).

Dihybrid Cross

A dihybrid cross describes a mating experiment between two organisms that are particularly different for two traits. The offspring of such cross is called dihybrid which is heterozygous at two different genetic loci. The two of the seven characters Mendel studied were seed colour and shape. Seeds shape may be either round (dominant) or wrinkled (recessive) and colour of the seed may be either yellow (dominant) or green (recessive).

When he crossed a homozygous round yellow (RRYY) plant with homozygous wrinkled green (rryy) plant, in F_1 generation all the offspring were produced with both dominant phenotypes i.e. round yellow. In order to analyse the genotype of F_1 plants, he self-fertilized them and produced F_2 generation. In F_2 generation he observed that offspring were produced in four phenotypic combinations i.e., round yellow, round green, wrinkled yellow and wrinkled green in the ratio of 9:3:3:1 (Fig.22.2).

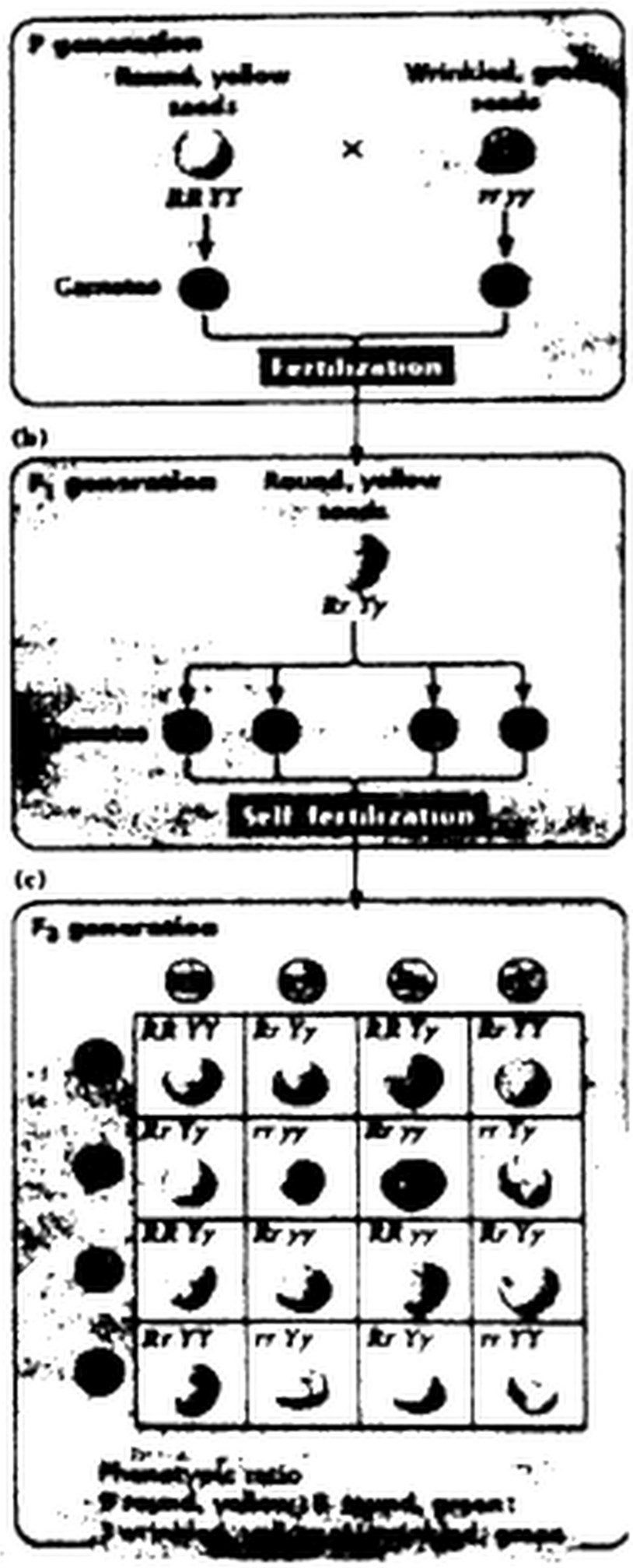


Fig. Dihybrid cross showing the inheritance of two traits

Based upon these observations, Mendel concluded that the F_1 offspring (round yellow) were dihybrid i.e., heterozygous (RrYy) for both traits. The key incidence in the experiment happened when F_1 plants self-pollinated and produce F_2 offspring. Four classes of gametes (RY, rY, Ry, and ry) would be produced by an F_1 plant in equal quantities. If the sperms of the four classes

which the alleles can combine in F_2 generation as shown in the Punnet square in figure 22.2. These combinations make up four phenotypic categories with a ratio of 9:3:3:1.

Mendel tested his seven pea characters in various dihybrid combinations and always observed as 9:3:3:1 phenotype ratio in F_2 generation. The results of Mendels dihybrid experiments are the basis of for what we now call law of independent assortment, which states that each pair of alleles assort independently of other pairs of *alleles during gamete formation* . In other words, the alleles of each pair of contrasting trait have equal probability to assort with the alleles of other pair (Fig. 22.2).

Limitations of Law of Independent Assortment

Although Mendels work forms the basis of heredity, it does not cover all situations. The fact is that Mendels work applies to diploid organisms, and not all organisms are diploid. Moréover, genes on the same chromosome could not be expected to assort independently. An offspring that inherited one trait would also inherit the other, unless crossing over occurred. Further, if genes are located on the X chromosome, the pattern of distribution in the succeeding generations is different. Males (because they have only one X chromosome - their other is Y, and does not carry many of the genes) are more likely to show recessive characteristics.

Usefulness of Law of Independent Assortment

Mendels law of independent assortment explains that if two parents have unique set of traits which are desired to express in one individual, so it is possible only because of independent assortment. Furthermore, if two parents have such traits which they do not want to be expressed in their offspring. This is also possible only because of independent assortment.

Scope of Independent Assortment in Variation

Beside mutation and crossing over (which are sources of variation), independent assortment of traits is also a major source of variations in successive generations. It is only due to the crossing over and independent assortment of the traits that the characteristics may appear in new combination in next generation which often seems necessary for adaptations in varying environment.

Q2. Describe exceptions to Mendels laws of inheritance.

Answer

Exceptions to Mendelian Inheritance

Since Mendel's time, our knowledge of the mechanisms of genetic inheritance has grown immensely. For instance, it is now understood that in how many different ways, alleles interact with their contrasting partner alleles at the same locus. These relationships between the contrasting alleles at the same locus in heterozygous state are called dominance relations. Although Mendel had observed only one form of dominance relation (complete dominance) but later on many geneticists became able to explain several exception to the Mendelian inheritance that could not be explained on the basis of complete dominance. These exceptions are said to have non-Mendelian

Q3. Explain incomplete dominance with reference to the inheritance of flower colour in 4'O Clock plant.

Answer

Incomplete Dominance

Studies of the inheritance of many traits have shown that member of a pair of alleles may not be completely dominant to other. For example red and white are common flowers in Japanese four O clock (*Mirabilis jalapa*). Each colour phenotype produces same phenotypes when these plants are self-pollinated. What flower colour might we expect in the offspring of a cross between a red-flowering plant and a white flowering one? Without knowing which is dominant, we might predict that all would have red flowers or all would have white flowers. In 1899 this cross was first made by German botanist **Carl Correns**, who found that all F_1 offspring have pink flowers. When two of these pink flowered plants are crossed, red flowered, pink-flowered and white-flowered offspring appear in a ratio of 1:2:1.

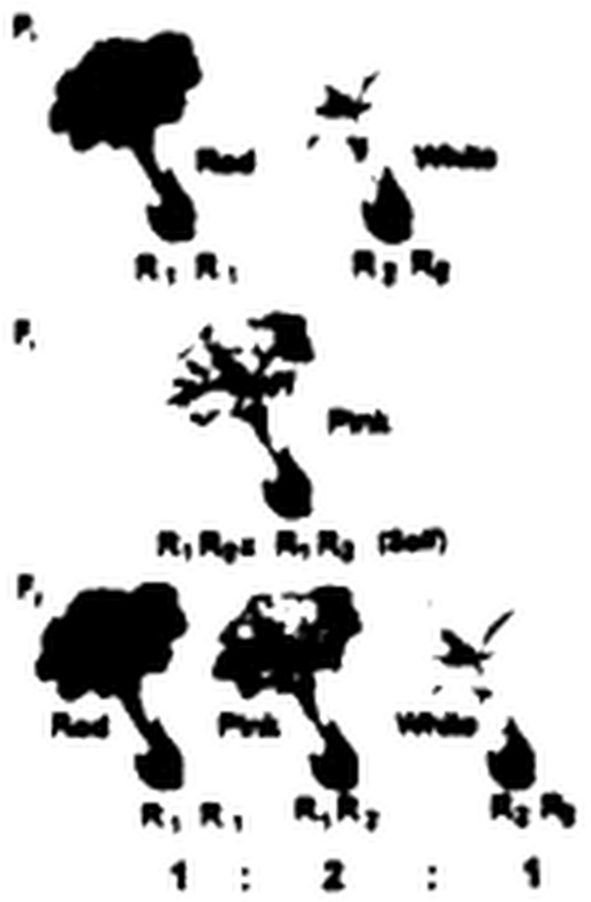


Fig. Incomplete dominance in 4-O'clock plants

The pink-flowered plants are clearly the heterozygous individuals and neither the red allele nor the white allele is completely dominant. When the heterozygous has a phenotype that is intermediate between those of its two parents, the dominance relation between the parental genes is said to be incomplete dominance.

Q4. Describe multiple alleles and state the alleles responsible for trait of ABO blood group. How blood group ABO shows that two alleles have equal dominance?

Answer

Multiple Alleles

So far we have discussed inheritance patterns involving only two alleles per gene. But many genes have more than two alleles all of them are called multiple alleles. Multiple alleles are produced by gene mutation. All the multiple alleles of a trait occupy the same locus. Some traits may be controlled by as many as 100 or more multiple alleles but each individual carries only two of them as each locus is twice represented in a diploid individual. For example, if a trait is controlled by 3 multiple alleles i.e., A_1A_2 and A_3 but every individual carries any two of them like A_1A_1, A_1A_2, A_1A_3, A_2A_2, A_2A_3 or A_3A_3. The ABO blood groups in humans are one example of multiple alleles.

Blood Group System

There are a number of different blood group systems found in humans. The International Society of Blood Transfusion has recognized up to 30 major group systems. These systems are characterized by the presence or absence of specific molecules, called antigens that are situated on the surface of the red blood cells. Most

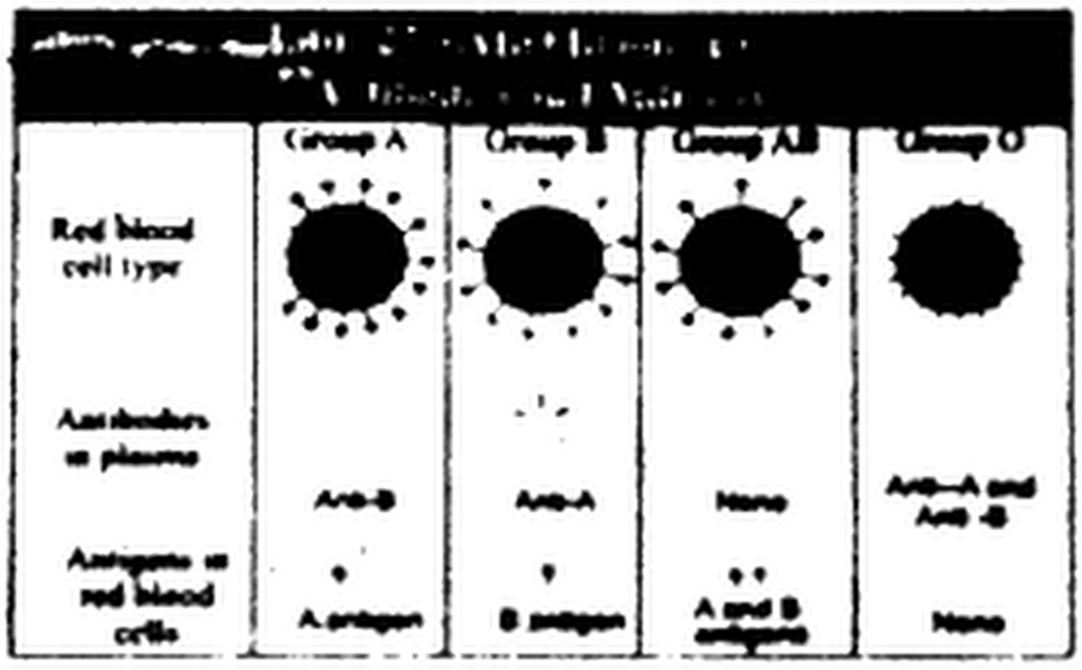
and Rh (Rhesus) system. These two systems are more significant because incompatibility between donor and recipients blood with respect to these two systems may become dangerous to life.

ABO Blood Group System

A well-known example of multiple alleles is the ABO blood group system in humans. It was discovered by Karl Landsteiner in 1901. ABO blood groups are also found in many other primates such as apes chimpanzees, bonobos, and gorillas.

Antigens of ABO system

There are several surface markers on the RBC membrane which are generally referred as antigens. ABO blood group system is based upon two such antigens i.e., A antigen and B antigen which are glyeoprotein in nature. If A antigen is present on the surface of RBC, the blood group is called type A and if B antigen is present, the blood group is called type B. If both antigens are present, the blood group is type AB and if none of these are present, the blood group is type O. (Table. 22.3).



Genetic basis of ABO system

In 1925 Berstein explained the genetic basis of ABO blood group system. Blood group is controlled by autosomal single polymorphic gene *I* (which stands for isoaemagglutinogen). The four blood types result from various combinations of three different alleles, symbolized as I^A (for the ability to make substance A) I^B (for B) and i (for neither A nor B). Their dominance relations are very interesting, alleles I^A and I^B are completely dominant over the allele i, while I^A and I^B are co-dominant to each other because each expresses equally in heterozygous (I^AI^B) state to produce AB phenotype. Therefore I^AI^A or I^Ai genotypes will produce phenotype A. Similarly I^BI^B or I^Bi produces phenotype B. The homozygous ii will produce phenotype O (Table: 22.3).

The blood groups alleles start their expression at early embryonic stage and keep on expressing themselves till death. Therefore, the blood group phenotype of a person never changes throughout life.

Table 22.4 Multiple Alleles and ABO Blood Groups

Blood Group (phenotype)	Antigen	Genotypes	Antibodies	Transfusion
A	A	I^AI^A or I^Ai	B	A and O
B	B	I^BI^B or I^Bi	A	B and O
AB	Both	I^AI^B	None	Any

Antibodies of ABO System

It has been observed that if wrong transfusion is carried out, the recipients blood start agglutination (antigen-antibody reaction) and clumping occurs. This is due to the presence of antibodies against wrong antigen. These antibodies are produced in the absence of their corresponding antigens. For example, those with type A blood have anti-B antibody in their plasma. Similarly, type B people have anti A antibody and those with type AB blood have no anti A or anti B antibodies in their plasma. Type O individuals have no antigens but have both anti A and anti B antibodies in the plasma. These antibodies of ABO system do not require any stimulus to produce their production begins from embryonic life and remain continue throughout life.

Q5. Explain Erythroblastosis foetalis. How the occurrence of Erythroblastosis foetalis can be prevented?

Answer

Erythroblastosis Foetalis

Erythroblastosis foetalis or haemolytic disease of the newborn (HDN) is an alloimmune condition that develops in a foetus, when the IgG-D (immunoglobulin-D) molecules commonly known as anti-Rh antibodies produced by the mother pass through the placenta. These antibodies attack the red blood cells in the foetal circulation, the mature red blood cells are broken down and the foetus can develop anaemia or if it is severe enough, it can cause the baby to die before birth. During this disease many erythroblasts are present in the foetal blood therefore the disease is called **erythroblastosis foetalis**.

Cause and Risk Factors

This most commonly happens when a woman with Rh-negative blood gets married to a man with Rh-positive blood and conceives a baby with Rh-positive blood (maternal foetal Rh incompatibility). If the mans genotype is DD, all of their offspring will have Dd genotype and will be Rh positive. If the mans genotype is Dd, half of their offspring with Dd genotype will be Rh positive. There is always chance of erythroblastosis foetalis whenever an Rh-positive foetus is conceived by Rh negative mother.

Red blood cells from the foetal blood can cross through the placental barrier into the mother's bloodstream during pregnancy or delivery. This causes the mother's body to make antibodies against the Rh factor.

The first Rh incompatible pregnancy may not face much problem if very few 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 the mother's blood stream and stimulate the 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 positive foetus. If the mother becomes pregnant again with an Rh-positive baby, it is possible for her antibodies to cross the placenta and attack the baby's red blood cells.

Problems and Complications in Foetus

of foetus, they start haemolysis (breakdown of blood). As this destruction continues, the foetus becomes anaemic. The anaemic foetus starts to release many immature erythroblasts into his blood stream. This anaemia may lead to abortion or still birth. Even if the pregnancy continues, the liver and spleen of the foetus swell as they rapidly produce RBCs. The breakdown product of RBC called bilirubin also accumulates in the foetus. Bilirubin damages his brain cells and turns his skin and whites of the eye yellow. This condition is jaundice. So the baby if born alive, suffer from severe haemolytic anaemia and jaundice. Such baby's blood should be immediately replaced by Rh-negative blood free of anti-Rh antibodies.

Prevention and Treatments

During a pregnant woman's first prenatal doctors visit, she should be screened for blood and Rh type. If she has Rh-negative blood, the father's blood and Rh type should be tested. If the father has Rh-positive blood, then there may be an Rh-positive foetus is being developed in the woman. In this case the Rh sensitization of Rh-negative mother can be avoided by a simple therapy. In this therapy she is given an injection of Rh antiserum (serum containing anti-Rh antibodies) during early pregnancy (1st trimester) and immediately after birth within 72 hours of delivery. This causes any of the baby's red blood cells that may have crossed into the mother's blood to be destroyed before sensitizing the mother's immune system to produce maternal anti-Rh antibodies. The injected antiserum disappears before the next pregnancy. This has to be done with each pregnancy whether it ends in a delivery or an abortion.

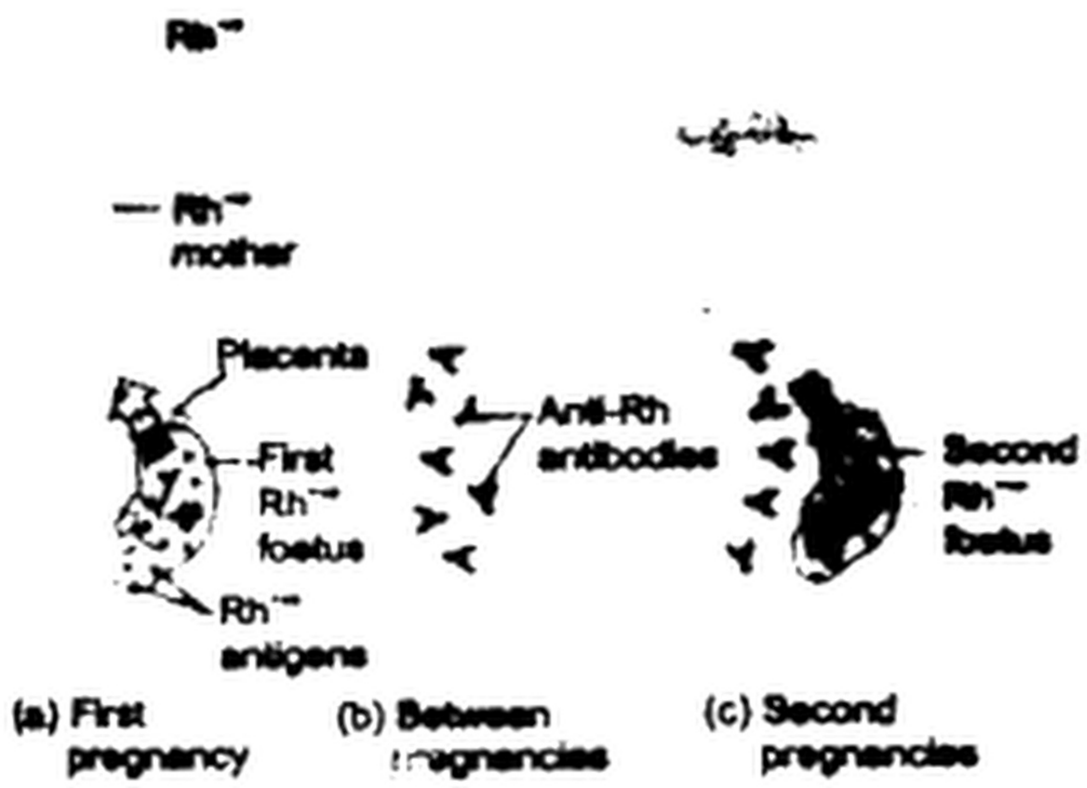


Fig. Maternal-Foetal Rh-Incompatibility

Q6. Describe polygenic inheritance citing the example of grain colours in wheat.

Polygenetic Inheritance and Epistasis

Some traits have large number of alternative phenotypes that have small and less striking difference so they have continuous variations such as height, weight intelligence and skin colour in humans; and grain colour in wheat. (Fig. 22.6) Such traits cannot be encoded by a single gene with two alleles. Even a few multiple alleles of a single gene cannot make a large number of phenotype. Such traits are encoded by alleles of two or more different gene pairs found at different loci, all influencing the same trait in an additive way. These quantitative traits are therefore called polygenic traits and their inheritance is called polygenic inheritance. All the genes that control a quantitative trait are called polygenes which have a small positive

of positive and negative effect of all individual polygenes produce quantitative phenotype of a continuous varying traits.

Wheat Grain Colour

Wheat grain colour is a good example of polygenic (multiple gene) inheritance. Wheat grains show a continuous variation in colour from white to dark red. Approximately seven different phenotypes (give in the table 22.5) are found in wheat population all over the world. Nilsson Ehle studied the genetics of wheat grain colour. When he crossed a homozygous dark red grain plant with a homozygous white grain plant, all F_1 grains had light red colour intermediate between two parental shades. It seemed as if it was a case of incomplete dominanoe. But when F_1 grains where grown to mature plants and crossed with each other, F_2 grains had exactly seven shades of colour in the following ratio:

Fig. Range of colours in wheat grain colour

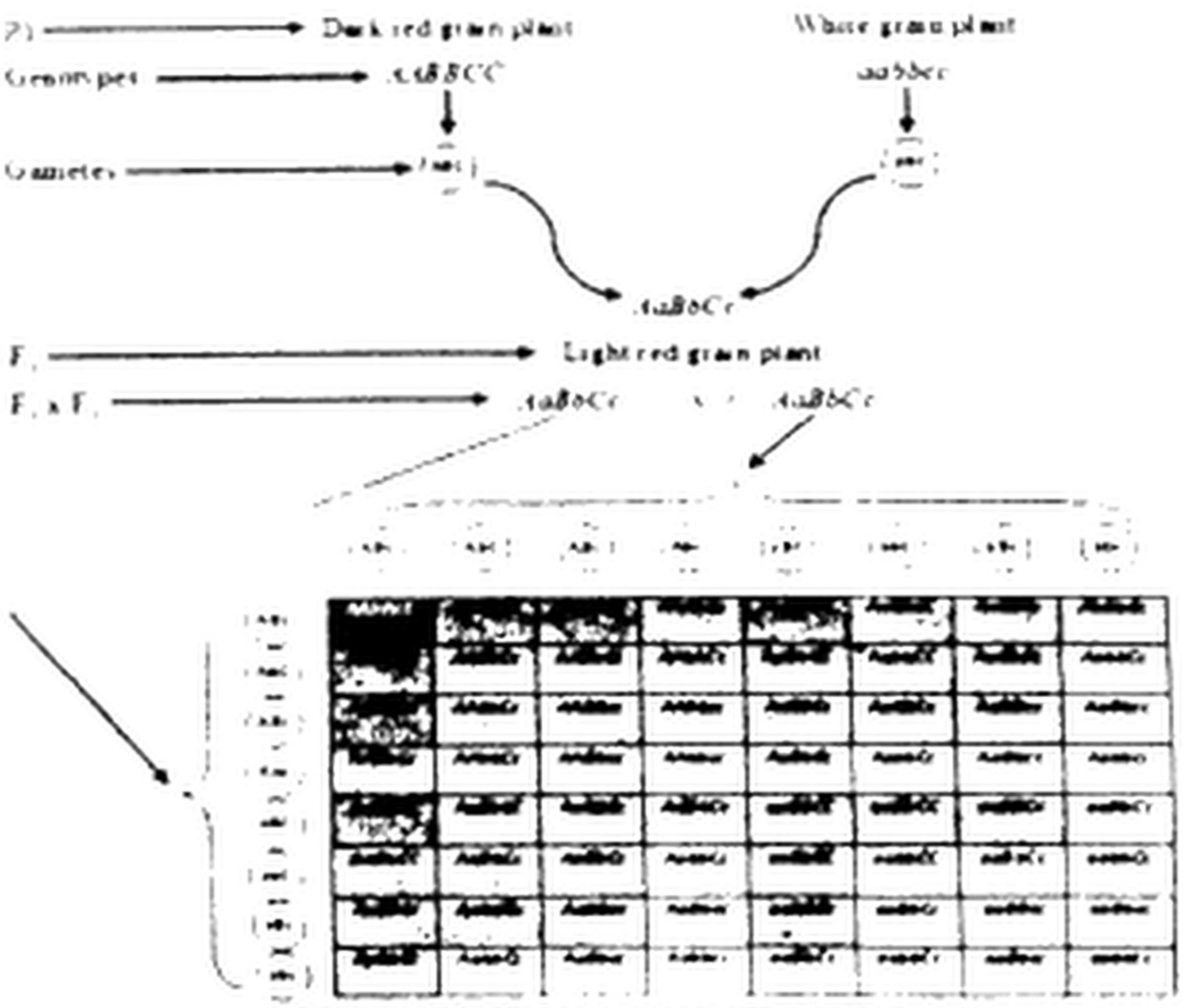


Fig. Inheritance of wheat grain colour

Table 22.6 Range of Phenotypes in Wheat Grain Colors

Pigment allele	6	5	4	3	2	1	0
Range of Phenotypes	Dark red	Moderately dark red	Red	Light red	Pink	Light Pink	White
Ratio	1	6	15	20	15	6	1

different loci contributed to the wheat grain colour. Each individual would contain six alleles for the trait. Alleles A, B and C codes for an equal amount (does) of red pigment, which is a positive effect. But none of a, b and c encode red pigment, which is a negative effect. If all the six allele code for red pigment (AABBCC), the grain is dark red; when none of the six allele encodes red pigment (aabbcc), the grain is white. When a grain has one allele for red pigment (Aabbcc aaBbcc or aabbCc) its colour is light pink; if it has two alleles for the pigment (AaBbcc or aaBbCc or AabbCc) it is pink, if it has three pigment alleles (AaBbCc or AABbcc or AabbCC), it will be light red. Similarly four alleles colour dose (AABBcc or aaBBCC or AAbbCC) will make red and five alleles colour dose (AABBCc or AABbCC or AaBBCC) will produce moderately dark red grain. Thus colour phenotypes of grains depend upon the number of pigment producing alleles (A, B, and C). Environment factors like light, water and nutrient also influence the amount of grain colour.

Q7. Describe polygenic inheritance citing the example of skin colour in man.

Answer

Inheritance of Human Skin Colour

Human skin colour is a good example of polygenic (multiple gene) inheritance. Skin colour is largely determined by the amount of melanin the skin produces. Dark-skinned individuals produce more melanin than light-skinned individuals. At least three genes regulate the amount of melanin produced.

1) Gene A is involved in the permanent survival, proliferation and migration of melanocytes.
2) Gene B encodes the enzyme tyrosinase, which is involved in the production of melanin from tyrosine.
3) Gene C is primarily responsible for determining whether pheomelanin or eumelanin is produced in humans.

Each gene has two forms, the dark-skin alleles are represented by capital case letters (A, B and C) and light-skin allelesare represented by small case letters (a, b and c). No allele is completely dominant to the other and heterozygotes exhibit an intermediate phenotype (incomplete dominance). A, B, and C act as dark-skin alleles in the genotype, because they add pigment by increasing melanin production. On the other hand a, b, and c act as light-skin alleles in the genotype because they inhibit melanin production. There are seven different shades of skin colour ranging from very light (aabbcc) to very dark (AABBCC); most individuals have the intermediate skin colour (AaBbCc). This later, genotype would be characteristic of a mulatto (an offspring of a black and a white parent). In the cross between two mulatto genotypes (AaBbCc X Aa BbCc), each parent produces eight different types of gametes and these gametes combine with each other in 64 different ways resulting in a total of seven skin colours. The skin colours can be represented by the number of capital letters, ranging from zero (no dark skin alleles) to six (all dark skin allele).

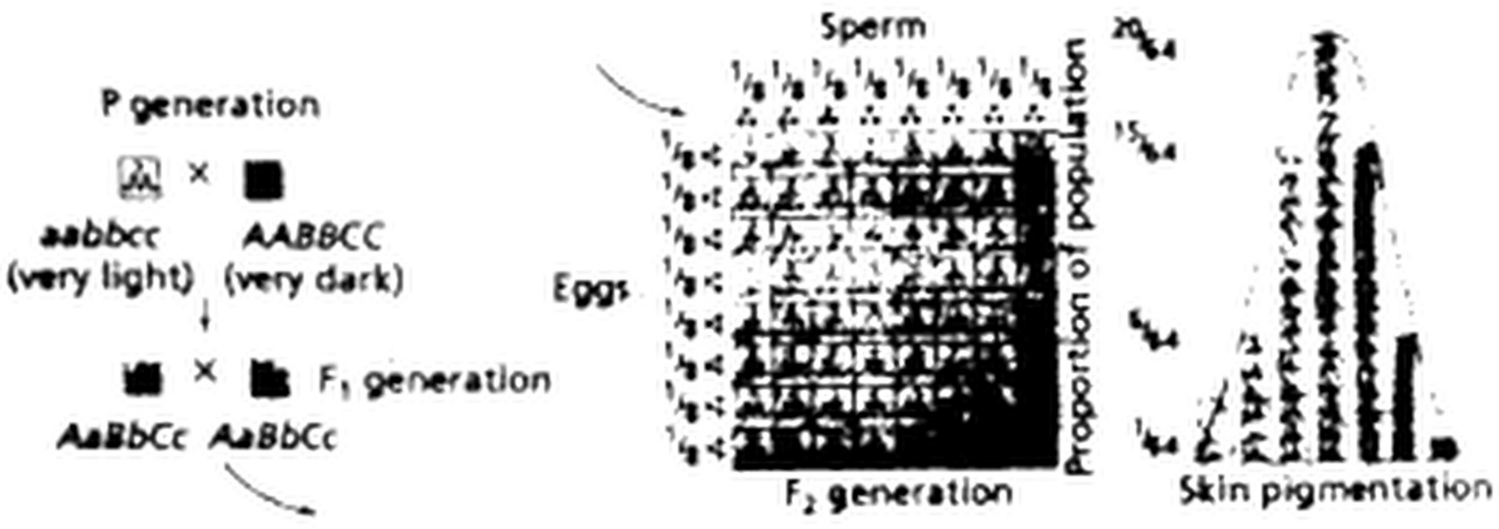


Fig. Inheritance of human skin colour

Q8. Explain modified mendelian ratios citing example of coat colour in inheritance in Labrador retriever pigment phenotype in foxgloves.

Answer

Coat Colour In the Labrador Retriever

Coat colour in Labrador retriever (a highly popular type of dog) is an excellent example of epistasis in animals. They have coats of three basic colours: yellow, black, and chocolate. The mode of inheritance of coat colour is autosomal (not related to the sex of the dog), with the information (genes) for black and chocolate at a different location in the chromosomes (locus) from the information (genes) for yellow.

Fig. Coat colour in the *Labrador retriever*

The allele for black coat colour (B) is dominant to the chocolate colour (b). Therefore, a puppy will only be chocolate if each parent contributes the chocolate alleles (bb). If one (Bb) or both (BB) parents contribute the black (dominant) gene, the puppy will be black (BB or Bb).

The gene that determines yellow coat colour is at a different location, (locus) in the DNA from the black versus chocolate gene. In order to be yellow, a Labrador must have two recessive copies of the yellow gene (yy). In this case, the yellow colour genes become epistatic and completely inactivate the black or chocolate genes, and the puppy is yellow. This means both parents contributed a yellow gene (y). However, if only one (Yy) or no (YY) yellow genes are contributed, the puppy will be either black or chocolate, determined as explained above by what is on the black/chocolate gene. Two yellow Labradors (yy) can only have yellow puppies (yy), since they both have two copies of the yellow gene and that is all they can contribute. On the other hand, some

from each parent and turn out yellow (Bbyy, BByy or bbyy) (Fig. 22.10).

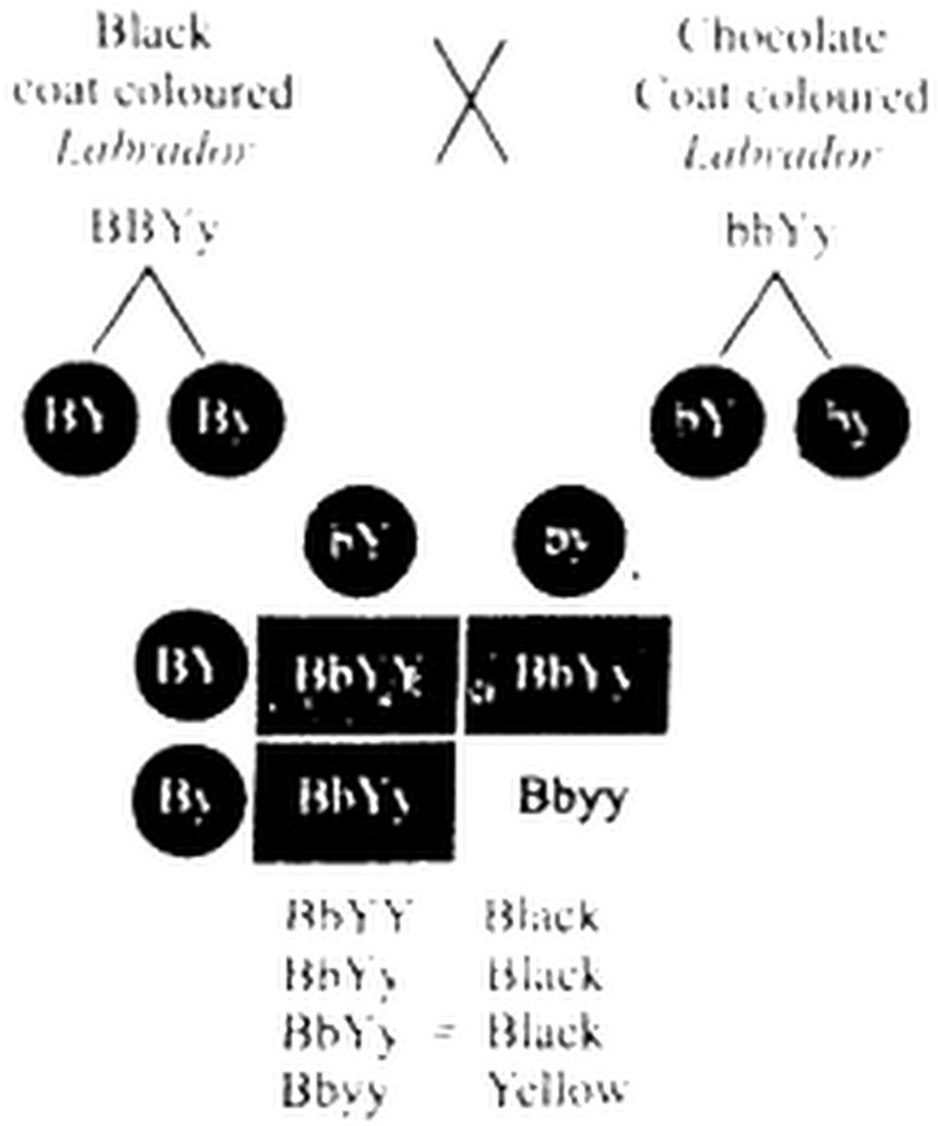


Fig. Inheritance of coat colour of *Labrador retriever*

Pigment Phenotypes in Foxgloves

Pigment phenotype in foxgloves (Digitalis purpurea) is an excellent example of epistasis in plants. It is determined by three separate gene loci (M, D and W). M codes for an enzyme that synthesizes anthocyanin, the purple pigment seen in these petals; m produces no pigment and produces the phenotype albino with yellowish spots. A plant with mm genotype will show white with yellowish spots (albino) irrespective of any genotype at other loci. Since M is dominant over m therefore a plant having M/M or M/m genotypes will show light purple colour. D is an enhancer gene of anthocyanin, resulting in a darker pigment; d does not enhance. Since D is dominant over d therefore a plant having D/D or D/d genotypes along with M/M or M/m genotypes will show dark purple colour. At the third locus, w allows pigment deposition in petals, but W prevents pigment deposition except in the spots and so results in the white, spotted phenotype. The plant having w/w along with other pigment producing genotype can show uniform pigmented phenotype otherwise; in the absence

Fig. Pigment phenotype in foxglove; A) Albino or white with yellow spots, B) Dark purple, C) White with purple spots, D) Dark purple

(*M/M*) *D/D* ; *w/w* × (*M/M*) *d/d* ; *W/W*
(dark purple) (white with purple spots)

↓

F_1 *D/d* ; *W/w* (white with purple spots)
D/d ; *W/w* × *D/d* ; *W/w*
(white with purple spots) (white with purple spots)

↓

F_2	9 *D/–* ; *W/–*	(white with purple spots)	12
	3 *d/d* ; *W/–*	(white with purple spots)	
	3 *D/–* ; *w/w*	(dark purple)	3
	1 *d/d* ; *w/w*	(light purple)	1

Fig. Inheritance of flower colour of foxglove

Q9. Explain how gene linkage encounters independent assortment and crossing over modifies the progeny by quoting the examples of wing length and width of abdomen in Drosophila melanogester.

Answer

Gene Linkage and Crossing Over

Mendel didn't know anything about the physical nature of genes or that genes are part of chromosomes, because nature of chromosomes weren't even discovered until long after his experiments were concluded.

Gene Linkage

The number of genes in a cell is far greater than the number of chromosomes; in fact each chromosome has hundreds and thousands of genes. Genes located on the same chromosome that tend to be inherited together in genetic crosses are said to be linked genes, and the phenomenon of staying together of more than one gene on the same chromosome is called gene linkage. If genes are linked on autosomes, their linkage is called autosomal linkage. Similarly, if they are linked on sex chromosome, their linkage is called sex linkage. All the linked genes found on the

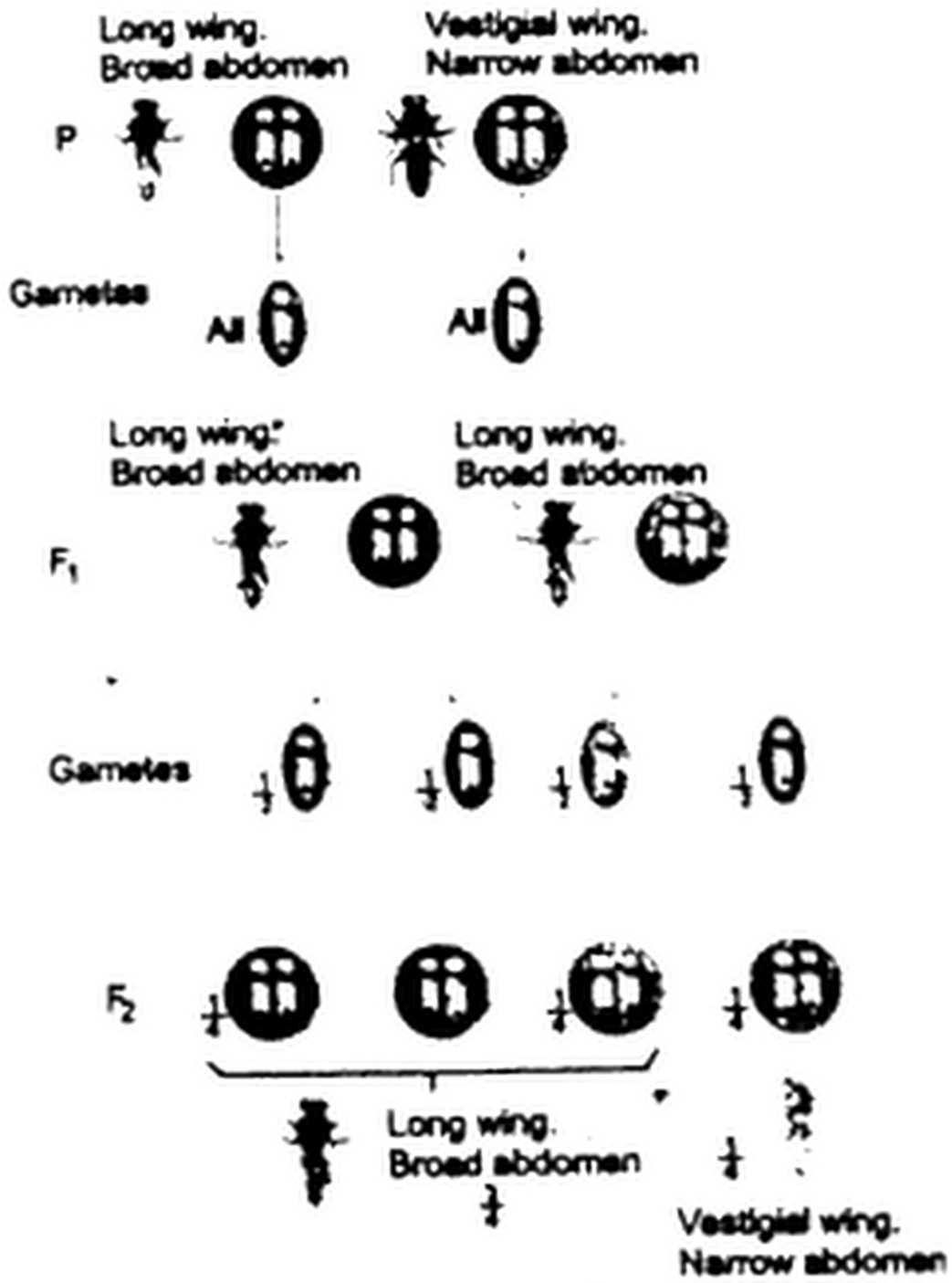


Fig. Linkage in *Drosophila*

number of linkage groups in an organism are equal to the number of homologous pair of chromosomes in that organism. As it is mentioned earlier, that linked genes tend to be inherited together (en bloc inheritance) in the offspring, so usually they do not show recombination and do not assort independently in the offspring. So the ideal Mendelian ratio of independent assortment is deviated.

Detection of Gene Linkage

Gene linkage can easily be detected by performing a test cross between two gene pairs (dihybrid test cross). In such type of test cross, a heterozygous individual for two traits (F_1) is back crossed with its recessive parent (P_1). If all four phenotypic combinations (parental and recombinants) are produced in equal 1:1:1:1 ratio, then there would be no linkage between the genes. If this ratio is deviated i.e., more parental types and less recombinant types, this indicates the incomplete or partial linkage; but if only parental types are produced, complete or tight linkage is believed. In a typical dihydrid cross, the complete or tight linkage inhibits the outcome of recombinant types and disturbs 9:3:3:1 ratio of independent assortment, as a result, only parental combinations are produced in 3:1. To see how linkage between genes affects the inheritance of two different characters, let's take an example from T.H. Morgan's experiments on Drosophila.

Morgan's Experiment

T. H. Morgan studied about 85 pair of contrasting trait in fruit fly Drosophila melanogaster. Two of them were wing length and width of the abdomen. Allele for long wings (Vg) is dominant over short or vestigial wing (vg). Similarly, allele for broad abdomen (A) is dominant over narrow abdomen (a). Morgan crossed a fly with long wings and a broad abdomen with one having vestigial wings and a narrow abdomen. The F_1 offspring all had long wings and broad abdomens. Then two of these flies were mated. In the F_2 generation about ¾ of the offspring had long wings and a broad abdomen and nearly all the remaining flies about ¼ of the total had vestigial wings and a narrow abdomen.

Gene Linkage Encounters Independent Assortment

Morgans results were very different from the results he expected based on the law of independent assortment i.e., 9:3:3:1. What had happened? From his data Morgan concluded that the genes for abdomen width and wing length were located on the same chromosome so they did no assort independently during meiosis. Instead, they inherited together. Therefore no recombinant types were produced and the standard ratio of independent assortment 9:3:3:1 is modified to 3:1.

Effect of Number of Progeny on Detection of Linkage

Gene linkage could be observed or evaluated only if the number of progeny is quite large because probability is used to determine the kinds of gametes produced and the changes of their combining. The larger the number of individuals, the greater is the likelihood that the laws of probability will hold. A small sample may not produce the results indicated by the laws of probability. Linkage can be recognized when an excess

Crossing Over

Subsequent experiments demonstrated that the process which is responsible for the recombination of linked genes is crossing over. In crossing over, an exchange of maternal and paternal chromatic parts occurs while homologous chromosomes are paired during prophase of meiosis I. The recombinant chromatids resulting from crossing over may bring alleles together in new combinations, so when they are distributed in different gametes. a wide variety of gametes are produced.

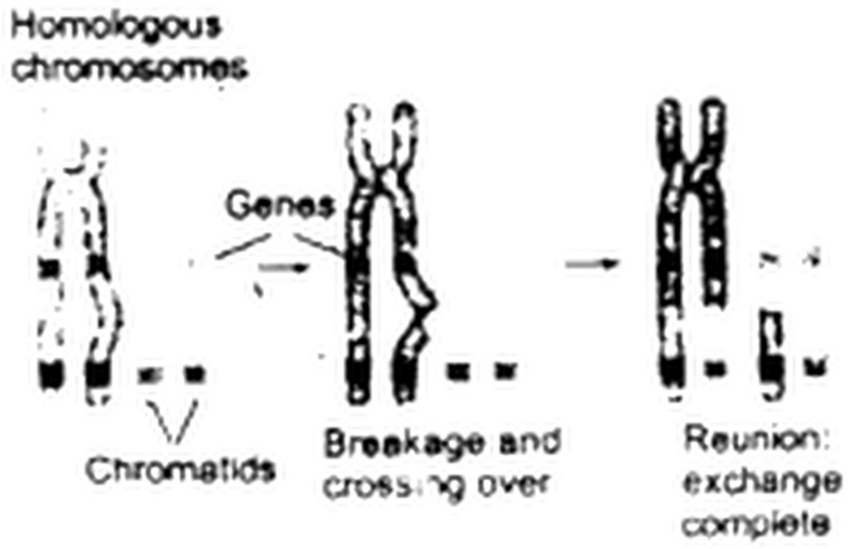


Fig. Crossing over

Morgans Experiments

Lets recall Morgans experiment in which a female fly having grey body and normal wings (dominant) was crossed by a male fly having black body and vestigial wings (recessive). Although most of the eggs had a chromosome with either the GL or gl parental genotypes for body colour and wing size, but some eggs had a recombinant chromosome with Gl or gL genotypes. Fertilization of these various classes of eggs by homozygous recessive sperms (gl) produced an offspring population in which 17% exhibited non-parental, recombinant phenotypes. These recombinants were the products of crossing over. If these two gene pairs were tightly linked, even crossing over could not separate them and parental combinations remained same in the next generation.

This observation also reflects another concept that the percentage of recombinant offspring, the recombination frequency is directly proportional to occurrence of crossing over which further depends upon distance between linked genes. Therefore, the recombination frequency is related to the distance between linked genes

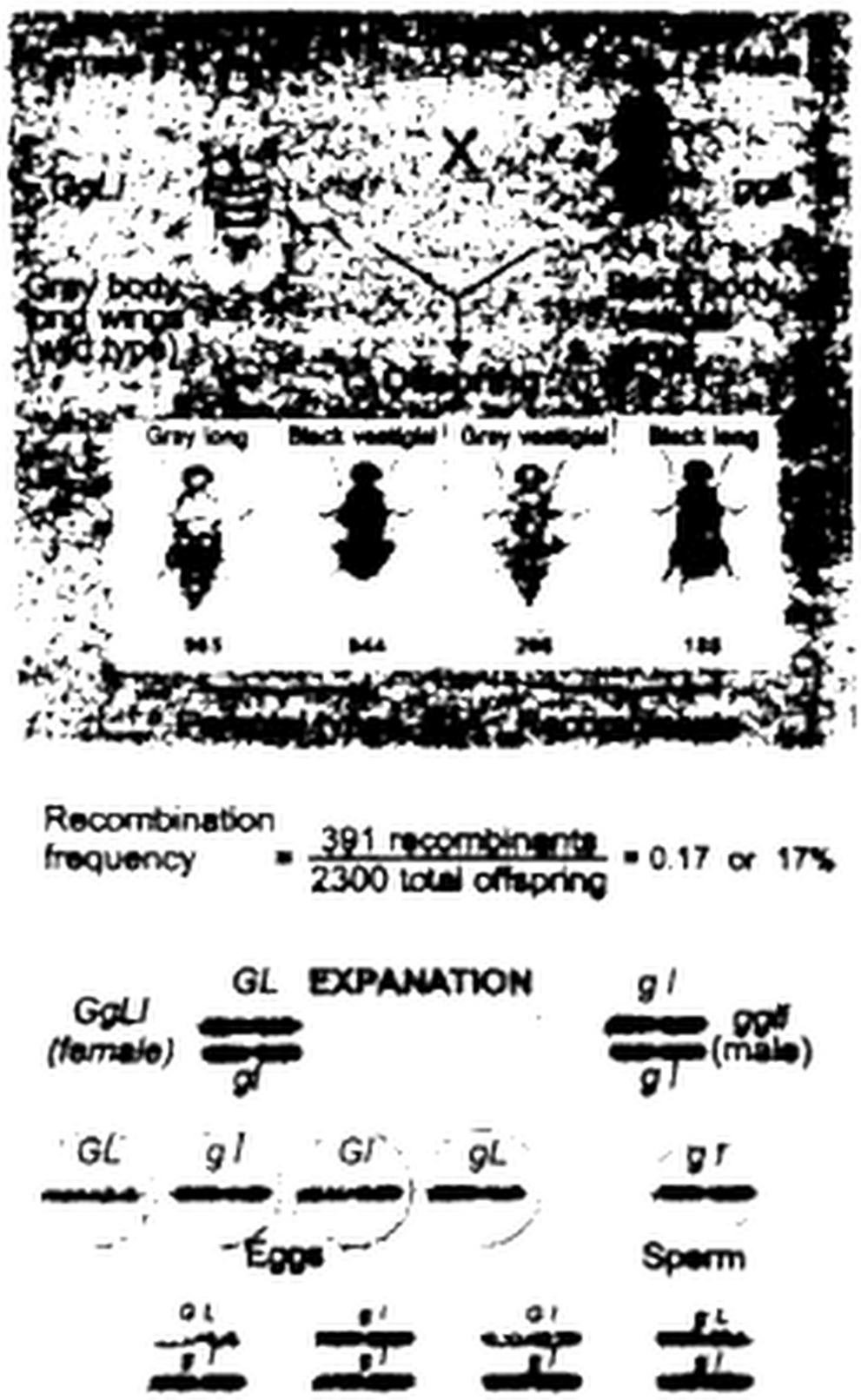


Fig: Fruit-fly experiment demonstrating the role of crossing over in inheritance

Q10. Explain mechanism of sex determination in Drosophila and man.

Answer

Sex Determination

Chromosomes in Drosophila

There are four pairs i.e., 8 chromosomes in Drosophila. Three pairs of chromosomes are identical in male and female, which are called autosomes. The fourth pair of chromosome is different in male and the female and determines genders is called sex chromosomes. In females both the sex chromosomes are identical, so these are called XX chromosomes. In males one is rod shaped and is like the sex chromosome of the female so it is also called X-chromosome, while the other chromosome is hooked shape and it is called Y-chromosome.

Chromosomes in Man

In humans there are 23 pairs of chromosomes, 22 pairs of which are autosomes, while one pair is sex chromosome. In female it is XX and XY in males. Human females have two copies of the X chromosome. All eggs cells produced by a woman contain one X chromosome. In males half of the sperms contain an X chromosome and half contain Y chromosome. The sex chromosome carried by the sperms is therefore determines the gender of the child. If a sperm carrying X-chromosome fertilizes the egg, the child will be a girl, but if a sperm carrying a Y chromosome fertilizes the egg, the child will be a boy.

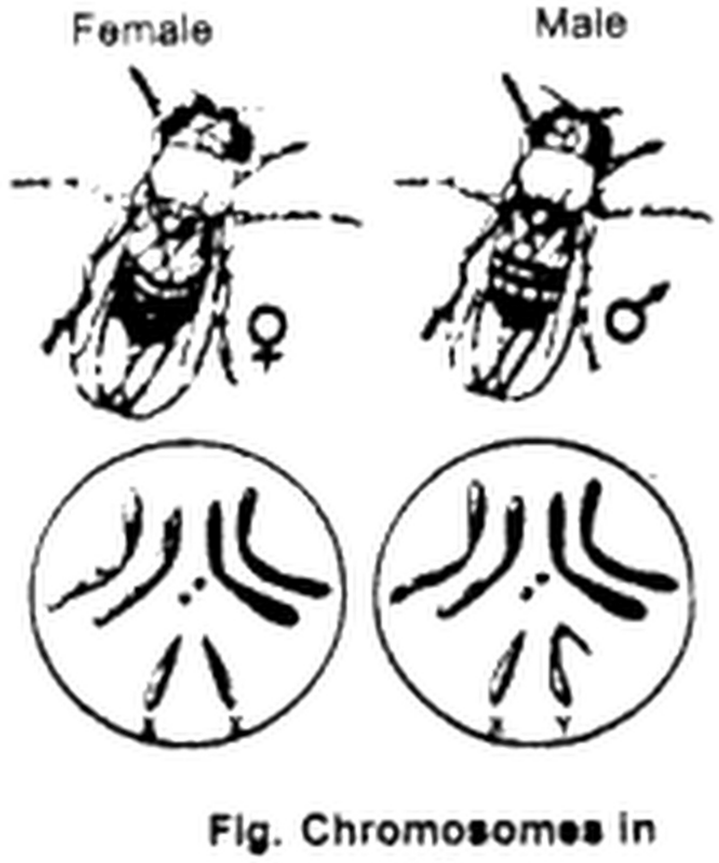


Fig. Chromosomes in *Drosophila*

Comparison of Sex Determination in Drosophila and Humans

Although both Drosophila and humans follow the same XY XY sex determining pattern, yet there is a basis technical difference between the two. Presence of SRY gene on Y chromosome is essential for triggering the development of maleness in human. Absence of Y chromosome simply leads to the female development path. XO Turners syndrome in humans produced through non-disjunction is a sterile female. But in Drosophila XO is a sterile male. Similarly XXY individual produced through non disjunction gametes in humans is a sterile male called Klinefelters syndrome, but the same XXY set of chromosomes in Drosophila produces a fertile female.

Fig. Karyotype: Human chromosomes

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

Fig. Comparison of sex determination in Drosophila and Human

There is a close genic balance between genes of different chromosomes. Drosophila has an X chromosome-autosome balance system. Its Y chromosome appears to have little influence on sex. Here actually the X chromosome is female determining and the autosomes are male determining. Sex of an individual depends more on the number of X chromosomes relative to the number of sets of autosomes. An X : A ratio of 1.00 or higher produces female whereas an X : A ratio of 0.5 or lower produces males (22.18).

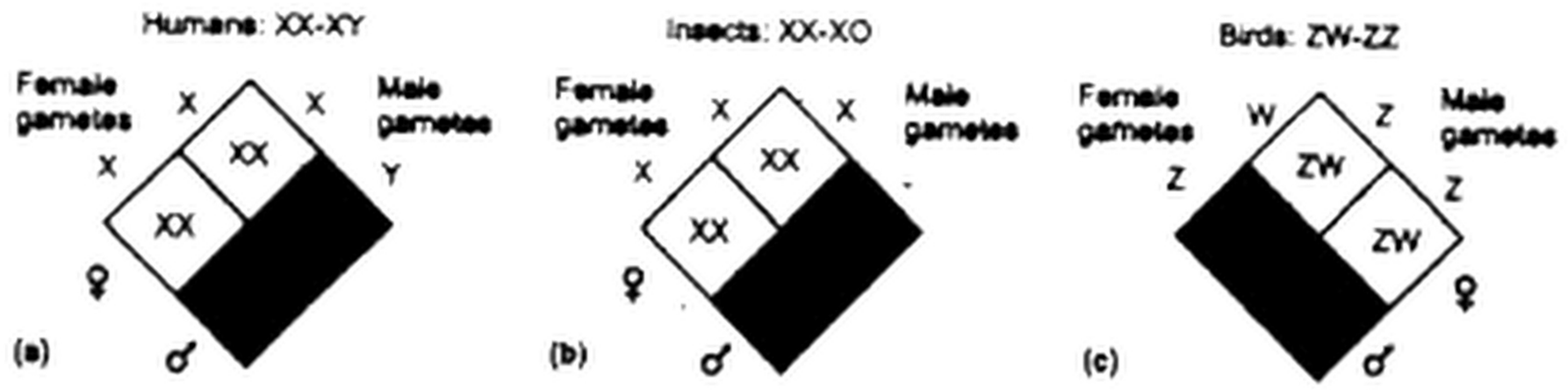


Fig. Patterns of sex determination

Q11. Describe XO-XX, XY-XX and XX- XY or ZW-ZZ sex determination system.

Answer

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

XO-XX Type

This pattern of sex determination is found in grasshopper and Protenor bug. Male is XO because it has only one X chromosome. The other sex chromosome is missing entirely. Male is heterogametic because it forms two types of sperms; half the sperms have X chromosome and the other half without any sex chromosome. A gamete without any sex chromosome is called nullo gamete.

Female is XX, because it has two X chromosomes. It is homogametic, as it forms only one type of eggs. Every egg carries an X chromosome. Sex of the offspring depends on the kind of sperm that fertilizes the egg. If an X-carring sperm fertilizes the egg, an XX female offspring is produced. If the nullo sperm fertilizes the egg, an XO male offspring is produced. Sex ratio between male and female offspring is 1:1

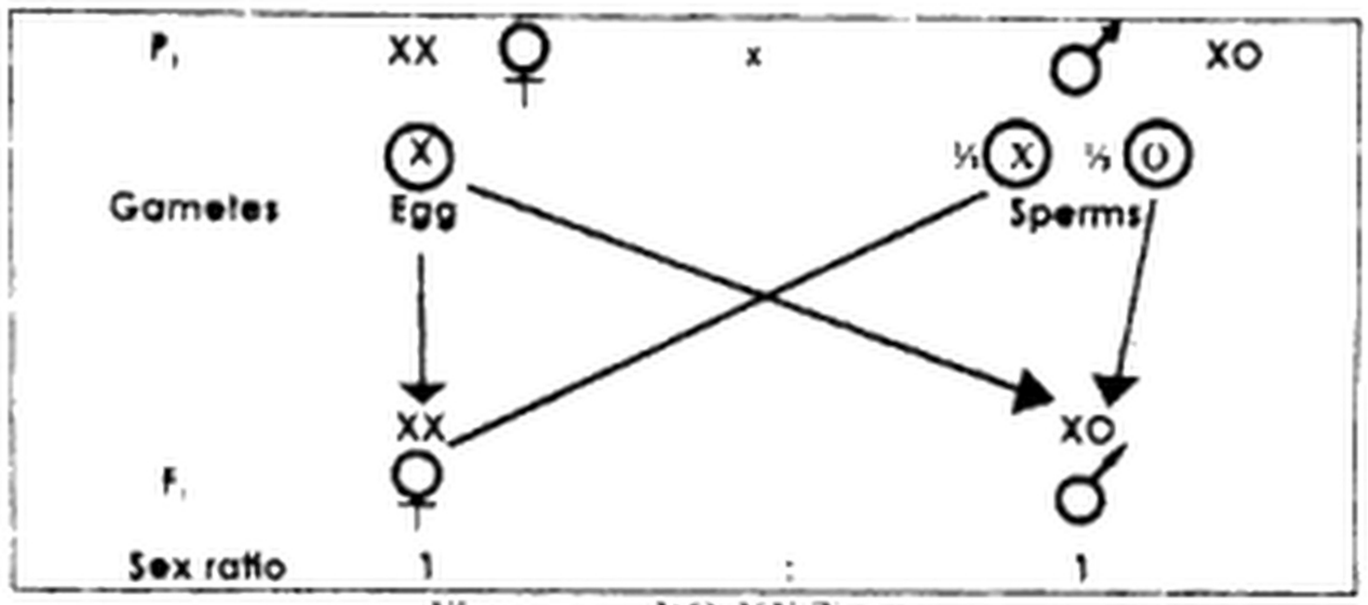


Fig: XO-XX Type

XY-XX Type

This pattern of sex determination is found in Drosophila, human and many other organisms. Male is XY and female is XX. Male being heterogametic produces two types of sex-determining sperms. Half the sperms carry X-chromosome and the other half carry Y – chromosome. Chances for both types of sperms are equal.

Female being homogametic produces only one type of eggs, each with an X chromosome. Sex of the offspring is determined by the type of sperm. If an X – carrying sperm fertilizes the egg, the zygote will be XX, and a female offspring is produced. If a Y – carrying sperm fertilizes the egg, the zygote will be XY, and a male offspring will be produced. The sex-ratio between male and female offspring is 1:1. Sex ratio indicates chances of the sex of the offspring. Chances for a son or daughter in human birth are equal.

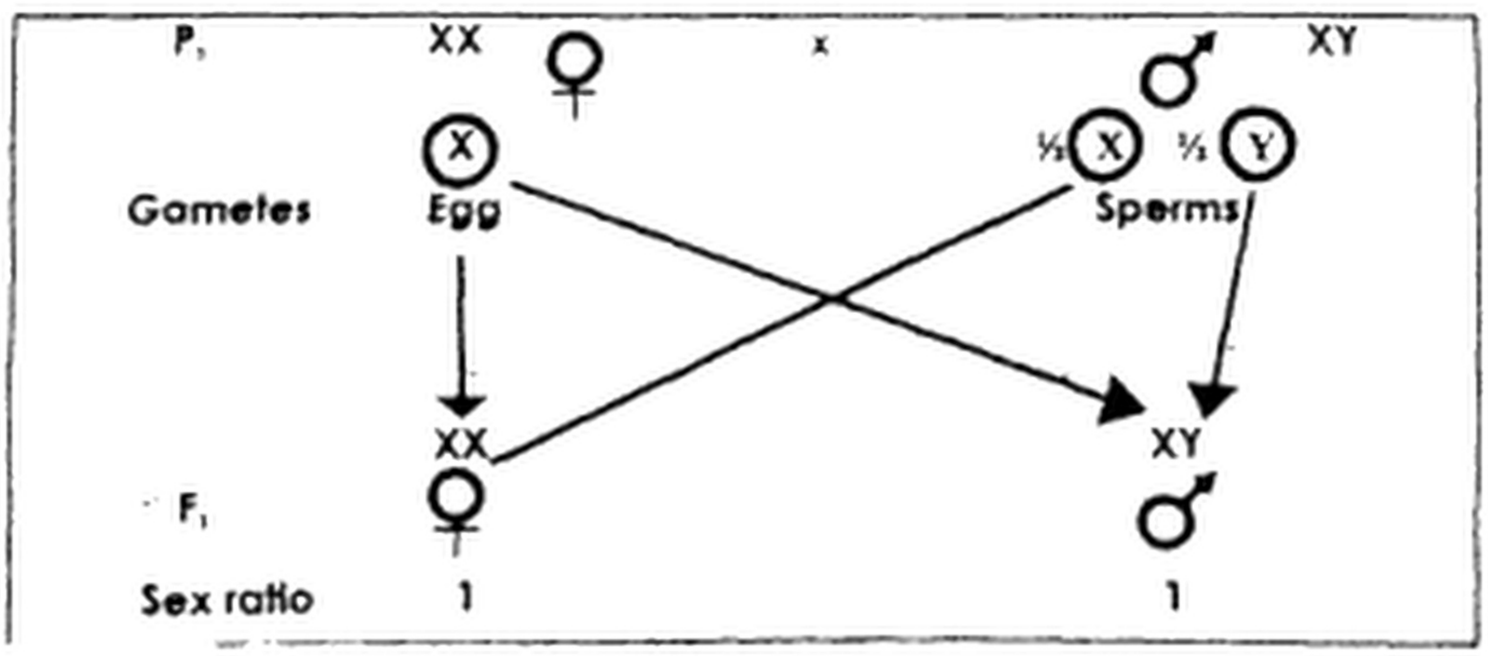


Fig. XX-XY Type

XX – XY Type or WZ – ZZ Type

This type of sex – determination pattern is common in birds, butterflies and moths. It was discovered by J. Seiler in 1914 in moths. It is the reverse of XY – XX system. Here the female is heterogametic XY but the male is homogametic XX. Female produces two kings of eggs X and Y in equal proportions. All sperms are alike, each carrying an X – chromosome. It is the kind of egg that determines the sex of offspring. When an X – carrying egg is fertilized by the sperm, a male offspring is produced, but when a Y – carrying egg is fertilized by the sperm a female offspring is produced. Sex ratio is 1:1.

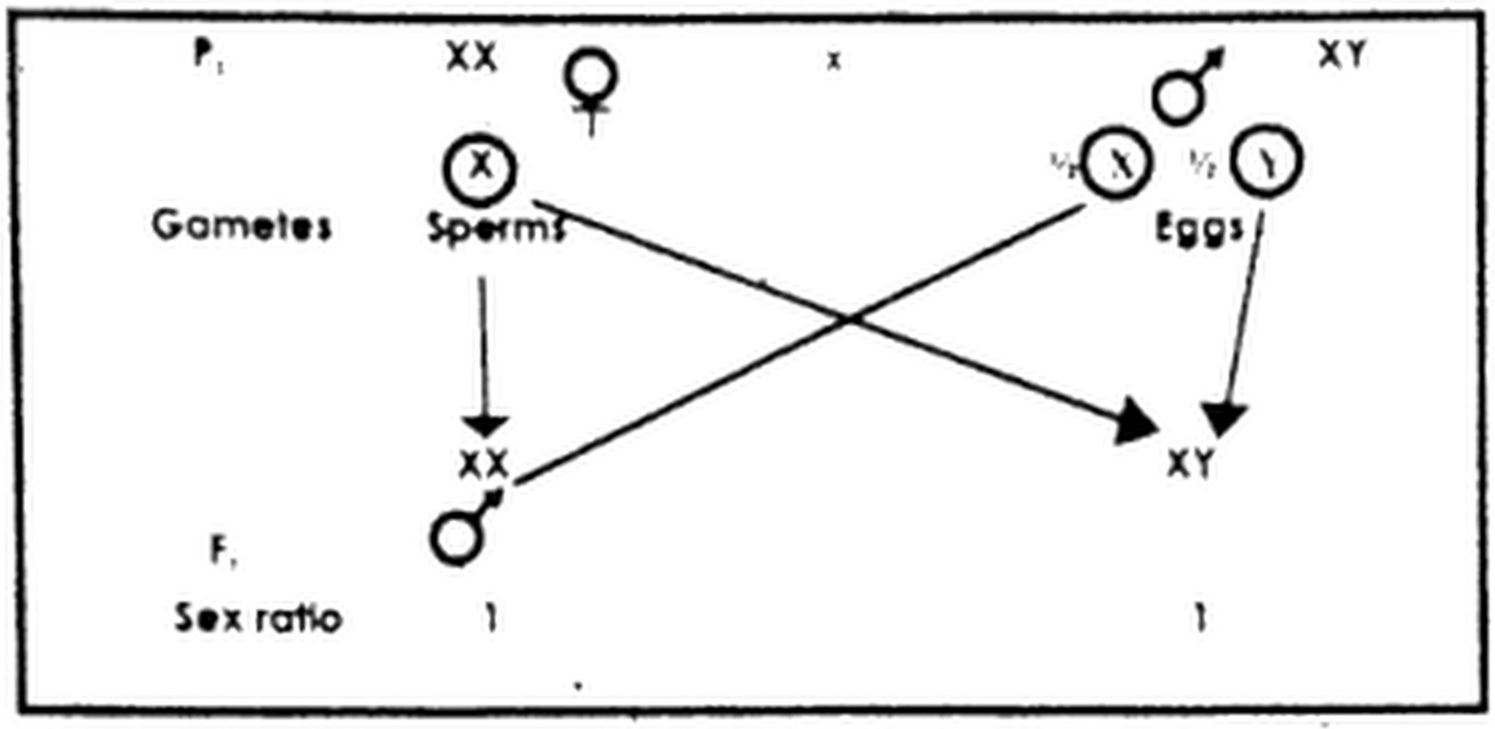


Fig. XX-XY or WZ-ZZ Type

Q12. What is sex linkage? Explain the inheritance of sex-linked traits (eye colour) in drosophila.

Answer

Sex Linkage in Drosophila

T. H. Morgan made the first demonstration of a sex linked trait in 1910. Through his experiments on Drosophila, the common fruit fly, Morgan showed that the inheritance of eye colour gene is located on the X chromosome but is not present on the Y chromosome. This meant that the recessive allele specifying white eyes is always expressed in males but could be masked by the presence of a dominant red eye colour allele in heterozygous females. Drosophila melanogaster eye colour turned out to be a most informative trait. At first, all the flies Morgan raised were wild type for eye colour; they had brick – red eye (wild type).

In 1910, a white-eyed male appeared in a laboratory bottle. Apparently the variant form arose through a spontaneous mutation in a gene controlling eye colour. Morgan established true breeding traits of white-eyed males and females. Then he did a series of reciprocal crosses represented in the following figures.

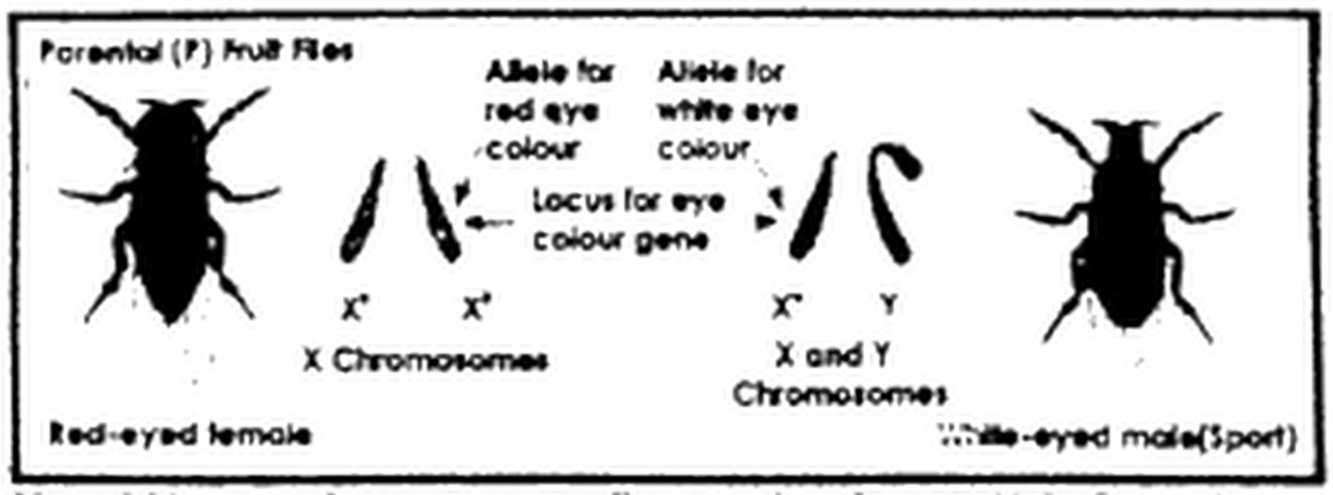


Fig X and Y = sex chromosomes, R = the dominant allele for red eye colour, w = the recessive allele for white-eye colour; X^R indicates that the X chromosomes carries the allele for red eyes; X^w indicates that the X chromosomes carries alleles for white eyes. The Y-chromosomes has no locus for eye colour genes. These are the phenotypes, sex chromosomes and alleles for eye colour in Morgan's experimental fruit flies in the P generation.

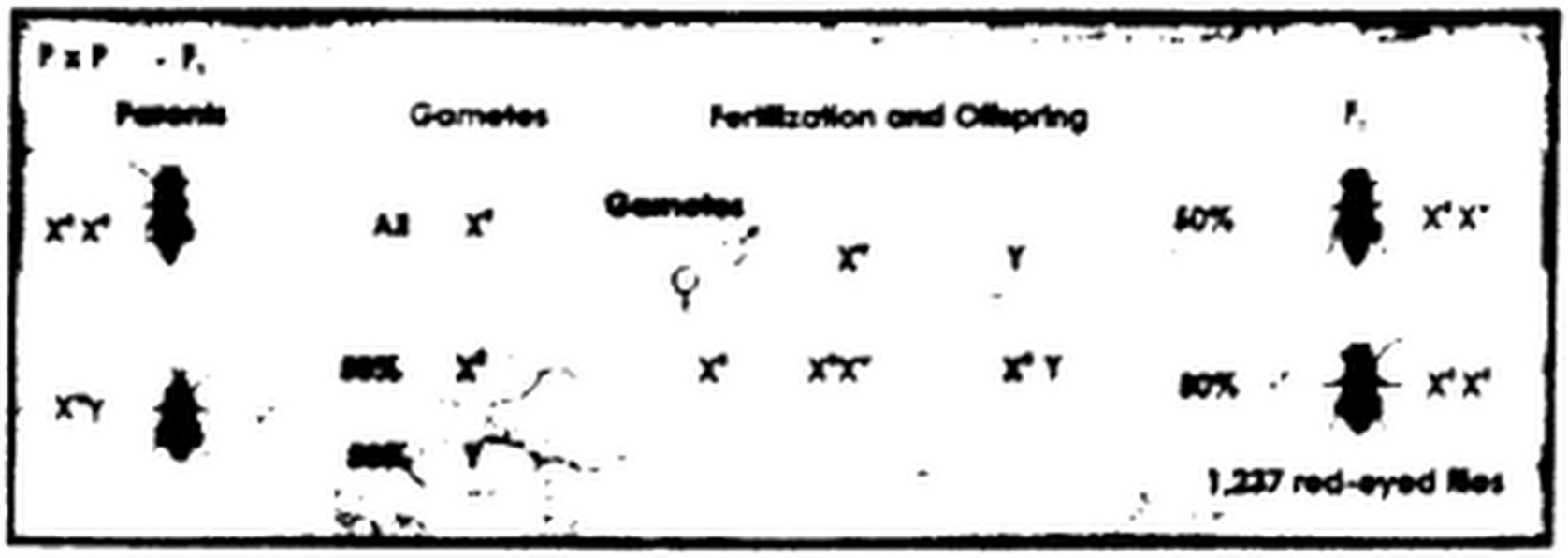


Fig. In Morgan's ?rst preliminary experiment (PxP), homozygous red-eyed females were mated with the white-eyed males. The female could produce gametes containing only X^R. The sperm of the male could contain either a Y chromosome or an X^r. A Punnett square is used to describe the offspring produce by the fertilization of parental gamets. The phenotypes and genotypes and ratios of the F_1 are also shown.

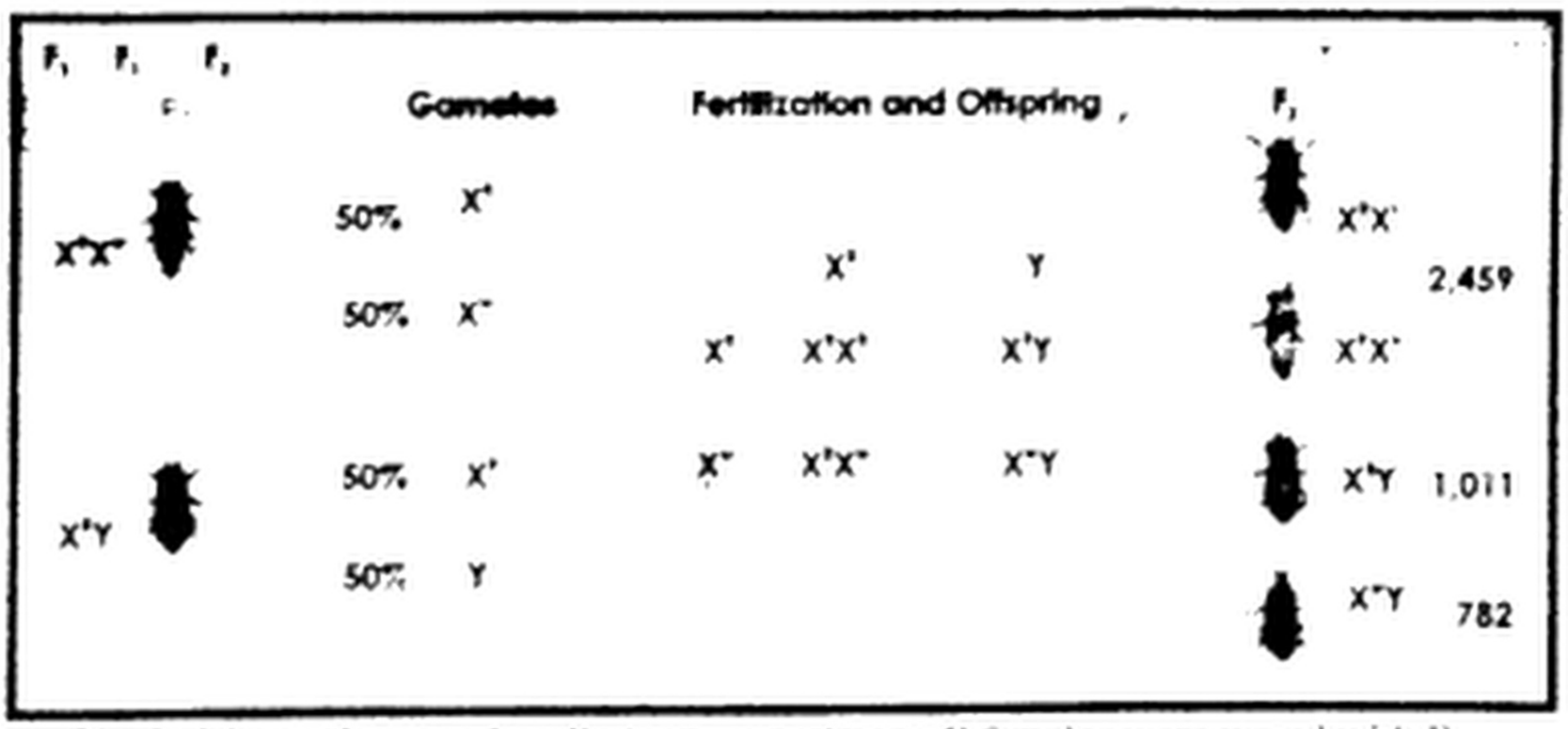


Fig. In Morgan's second preliminary experiment, F_1 females were mated with F_1 males. Morgan hypothesis explaind clearly why all the white eyed ?ies in F_2 generation were only males.

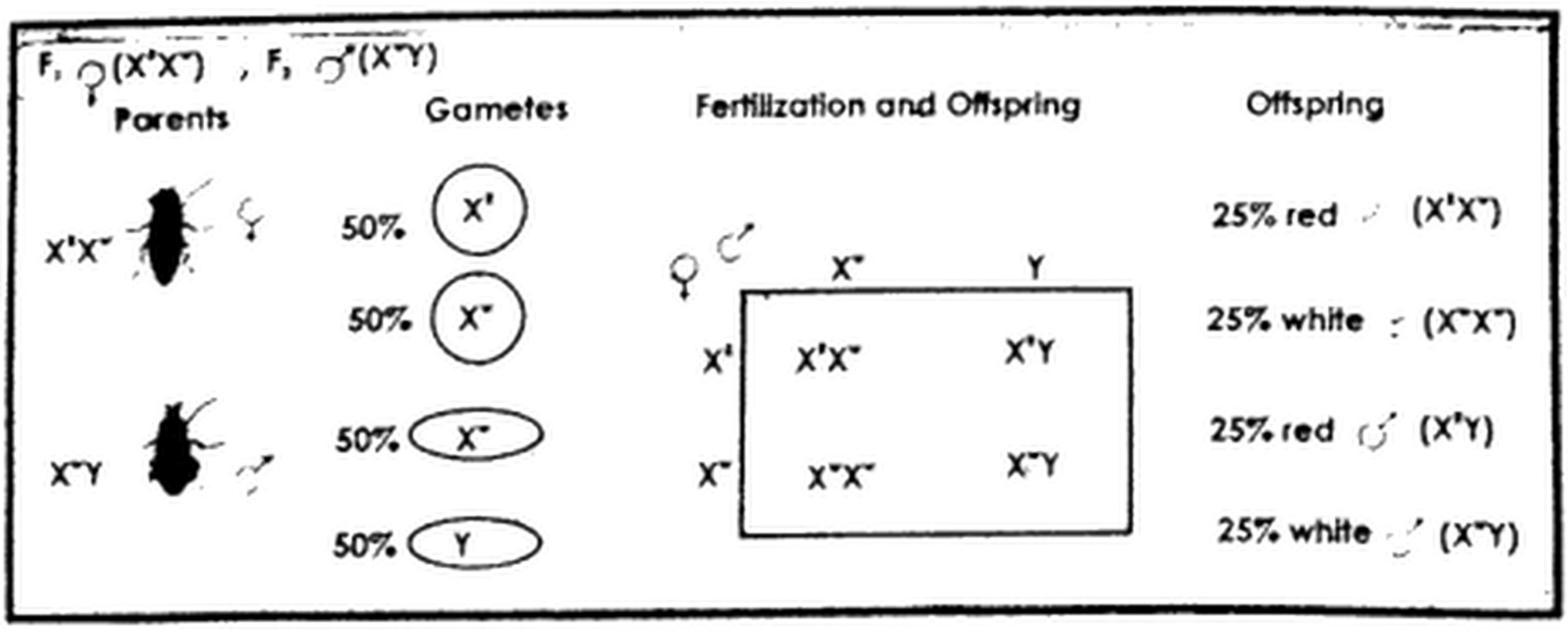


Fig:

Morgan wanted to test his hypothesis. He crossed F_1 females heterozygous for white-eyes with white-eyed male. The four combinations of offspring resulted (25 percent

confirmatory test. For this a cross was made between white eyed female (X^wX^w) with a red eyed male (X^RY). All the female's offspring's had red eyes and all male offspring had white eyes in the fig below.

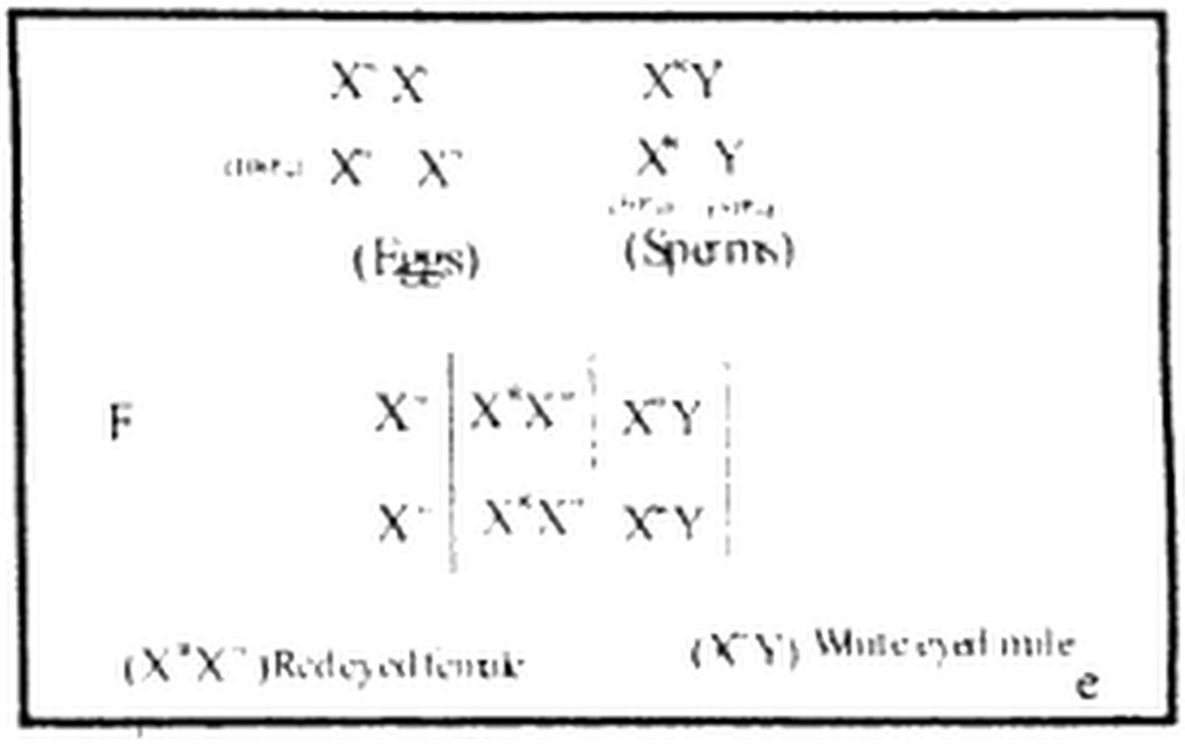


Fig:

Morgan experiments were designed to test prediction derived from his hypothesis that factor for eye colour is associated with X chromosomes. A trait whose gene is present on X-chromosome is called X-linked trait and a gene which is present on X-chromosomes with no counter part on Y-chromosome do carry some gene having no counter part on X chromosome e.g. SRY genes on Y chromosome of man determines maleness. Y-linked genes directly pass through Y chromosome from father to son.

Q13. Describe the sex-linked inheritance of male character due to Y-chromosome and the effect of holandric genes.

Answer

Y-Linked Inheritance

In mammals, Y-linkage refers to when a phenotypic trait is determined by an allele (or gene) on the Y chromosome. It is also known as holandric inheritance.

The Y-chromosome is small and does not contain many genes, therefore few traits are Y-linked, and Y-linked diseases are rare. Because the only humans which have a Y chromosome are males, the genes are simply passed from father to son, with no interchromosomal genetic recombination.

Chromosome Y deletions are a frequent genetic cause of male infertility. Another example in humans of a Y-linked trait was thought to be hairy ears (it may also be sex-limited). However, this has been discredited.

Q14. Describe the X-linked disorder with reference to pattern of inheritance.

Answer

Sex Linked Disorders In Humans

Humans have several disorders which are caused by the mutation in sex chromosomes as Drosophila has white eye colour. The inheritance patterns of some sex linked

Genetics of Haemophilia

Haemophilia is a rare X linked recessive trait. Haemophiliacs blood fails to clot properly after an injury, because it has either a reduction or malfunction or complete absence of blood clotting factors. It is a serious hereditary disease because a haemophiliac may bleed to death even from minor cuts. Haemophilia is of three types: A, B and C. Haemophilia A and B are non allelic recessive sex linked, but haemophilia C is an autosomal recessive trait. 80% haemophiliacs, suffer from haemophilia A due to abnormality of factor VIII, about 20% suffer from haemophilia B due to disturbance in factors IX, but less than 1% suffer from haemophilia C due to reduction in factor XI. Being X linked recessives, haemophilia A and B affect men more than women, but haemophilia C affected both the sexes equally because it is autosomal. Chances for a man to be affected by haemophilia A and B are double than a woman. A woman can suffer from haemophilia A or B only when she is homozygous for the recessive allele, but a man with just one recessive allele will display the trait. Haemophilia A and B zigzag from maternal grandfather through a carrier daughter to a grandson. It never passes direct from father to son. Gene for normal is H and gene for haemophilia A is h.

Fig Hairy pinna in humman

Table 22.8 Haemophilia

S. No	Types of Haemophilia	Cause	Percentage	Inheritance
1.	Haemophlia A	Abnormality of blood clotting factor VIII	80%	Recessive X linked
2.	Haemophilia B	Disturbance in blood clotting factor IX	20%	Recessive X linked
3.	Haemophia C	Reduction in blood clotting factor XI	Less than 1%	Dominant autosomal

Q15. Critically analyze the inheritance of haemophilia, colour blindness and muscular dystrophy.

Answer

I) Genetics of Colour-Blindness

Normal trichromatic colour vision is based on three different kinds of cone in the retina, each sensitive to only one of the three primary colours, red, green or blue. Each type of cone cell has specific light absorbing proteins called opsins.

The genes for red and green opsins are on X chromosome, while the gene for blue opsin is present on autosome 7. Mutations in opsin genes cause colour-blindness like dichromacy and monochromacy. A dichromate can perceive two primary colours but is unable to perceive the one whose opsin is missing due to mutation. Protanopia is red blindness; deuteronopia is green blindness, while tritanopia is blue blindness (Fig. 22.25).

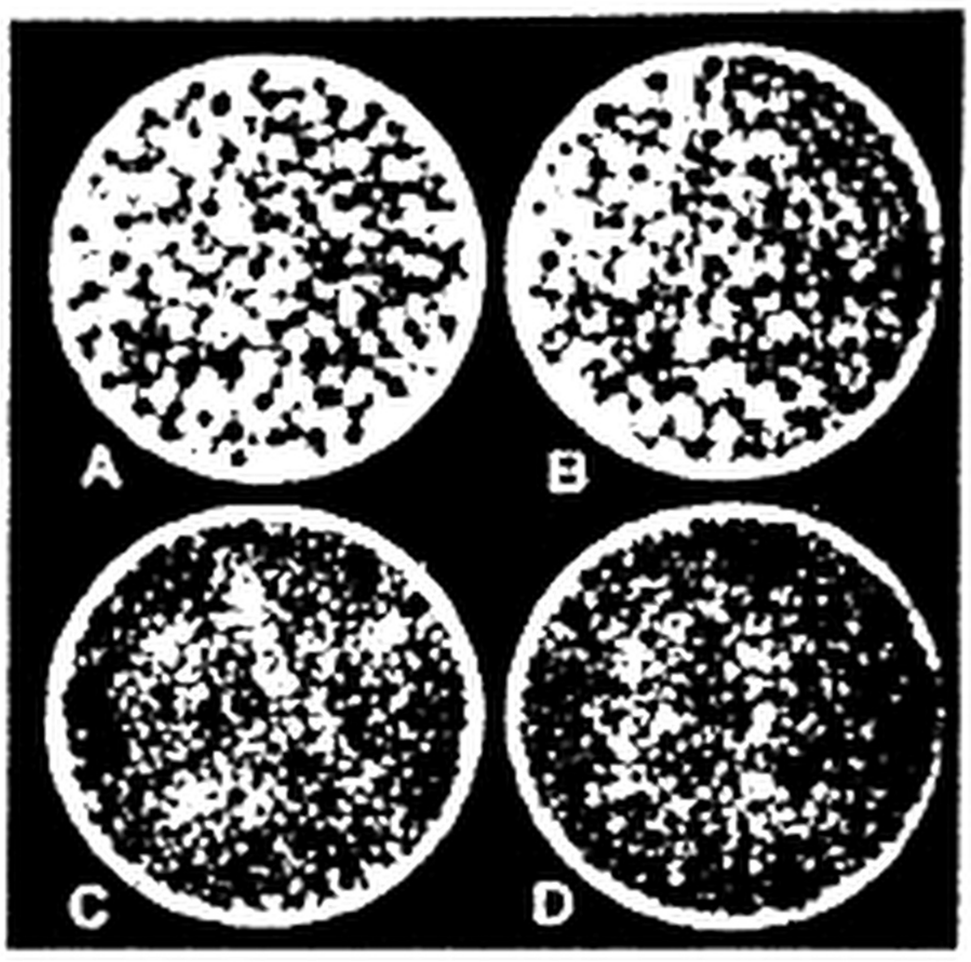


Fig. These colours may appear slightly different if you are red-green colour blind

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 opsin. They are protanomalous and deuteranomlous for red and green respectively. A monochromate can perceive only one colour. Monochromacy is true colour-blindness. Blue cone monochromacy is an X linked recessive trait in which both red and green cone cells are absent. That is why it is also called red green colour-blindness. It is a common hereditary disease.

Like any sex linked recessive trait, it also zigzags from maternal grandfather through a carrier daughter to a grandson. It never passes direct from father to son. This type of colour-blindness is more common in men than women, because chances for a male to be affected by it are double than a female.

Fig. Muscular dystrophy

ii) Genetics of Muscular Dystrophy

Muscular dystrophy, as the name implies, is characterized by a wasting away of the muscles. The most common form is Duchenne muscular dystrophy. It is a sex-linked recessive

childhood, when the child begins to have difficulty standing up and rises to a standing position in a characteristic way. He is inevitably wheelchair bound by the age of 12. Eventually, he becomes severely wasted, and normal breathing becomes difficult. Death usually occurs by age of 20, therefore, affected males are rarely fathers. The recessive allele remains in the population by passage from carrier mother to carrier daughter. The gene whose mutation causes this disorder has been mapped. Only 1% codes for a protein called dystrophin, which is present in the normal muscle but missing in Duchenne patient. The lack of dystrophin causes calcium to leak into the cell, which promotes the action of an enzyme that dissolves muscle fibres.

iii) Genetics of Haemophilia

Haemophilia is a rare X – linked recessive trait. Haemophiliac's blood fails to clot properly after an injury, because it has either a reduction or malfunction or complete absence of blood clotting factors. It is a serious hereditary disease because a haemophiliac may bleed to death even from minor cuts.

Types of Haemophilia

Haemophilia is of three types: A, B and C. Haemophilia A and B are non – allelic recessive sex – linked, but haemophilia C is an autosomal recessive trait. 80% haemophiliacs, suffer from haemophilia A due to abnormality of factor VIII, about 20% suffer from haemophilia B due to disturbance in factor IX, but less than 1% suffer from haemophilia C due to reduction in factor XI.

Degree of Effect by Haemophilia

Being X – linked recessives, haemophilia A and B affect men more than women, but haemophilia C affects both the sexes equally because it is autosomal. Chances for a man to be affected by haemophilia A&B are double than a woman. A woman can suffer from haemophilia A or B only when she is homozygous for the recessive allele, but a man with just one recessive allele will display the trait.

Pattern of Inheritance of Haemophilia

Haemophilia A and B zigzag from maternal grandfather through a carrier daughter to a grandson. It never passes direct from father to son. Gene for normal is H. Gene for haemophilia A is h. In generation I of this pedigree a man (I – 2) suffering from haemophilia A marries a normal woman (I – 1). He passes haemophilia gene to his daughter (II – 2) through

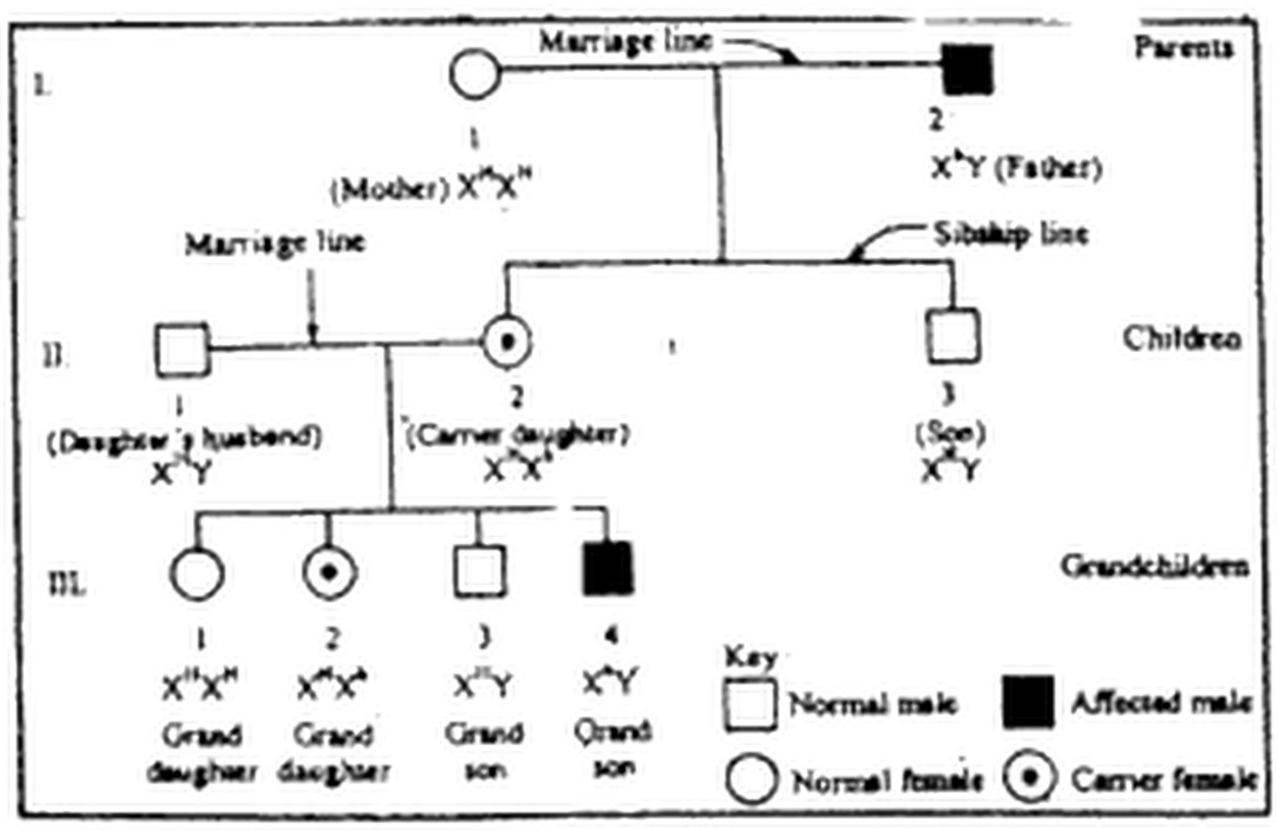


Fig. Transmission of X-linked recessive trait (haemophilia) in man.

receives only Y chromosome from him. His daughter (II – 2) also receives another X but with normal dominant allele from her mother (I – 1). The daughter looks phenotypically normal, but she is heterozygous and a carrier for the recessive gene. When she marries a normal man (II – 1) she passes her father's trait to one of her two sons (III – 4) who inherits grandfather's X from her. The single recessive allele for haemophilia expresses successfully in the hemizygous son because his Y chromosome does not carry its counterpart. The other son (III – 3) is normal as he inherits grandmother's X with normal gene. One daughter (III – 1) with both normal X is normal, but the other daughter (III – 2) is carrier like her mother.

ADDITIONAL QUESTIONS

Q16. Why Mendel is famous? What are the Mendel's experimental results? Define the two laws proposed by Mendel.

Answer

In the 1860s, Gregor Mendel introduced a new theory of inheritance based on his experimental work with pea plants. According to this Mendelian concept, inheritance of a trait depends on the passing-on of these units. For any given trait, an individual inherits one gene from each parent so that the individual has a pairing of two genes. We now understand the alternate forms of these units as alleles. If the two alleles that form the pair for a trait are identical, then the individual is said to be homozygous and if the two genes are different, then the individual is heterozygous for the trait.

Gregor Mendel

Mendel's Experimental Results

Mendel carried out breeding experiments in his monastery garden to test inheritance patterns. He selectively cross-bred common pea pants (Pisum sativum) with selected traits over several generations. After crossing two plants which differed in a single trait (tall stems vs. short stems, round peas vs. wrinkled peas, purple flowers vs. white flowers, etc), Mendel discovered that the next generation, the F_1 (first filial generation), was comprised entirely of individuals exhibiting only one of the traits. However, when this generation was interbred, its offspring, the F_2 (second filial generation), showed a 3:1 ratio-three individuals had the same trait as one parent

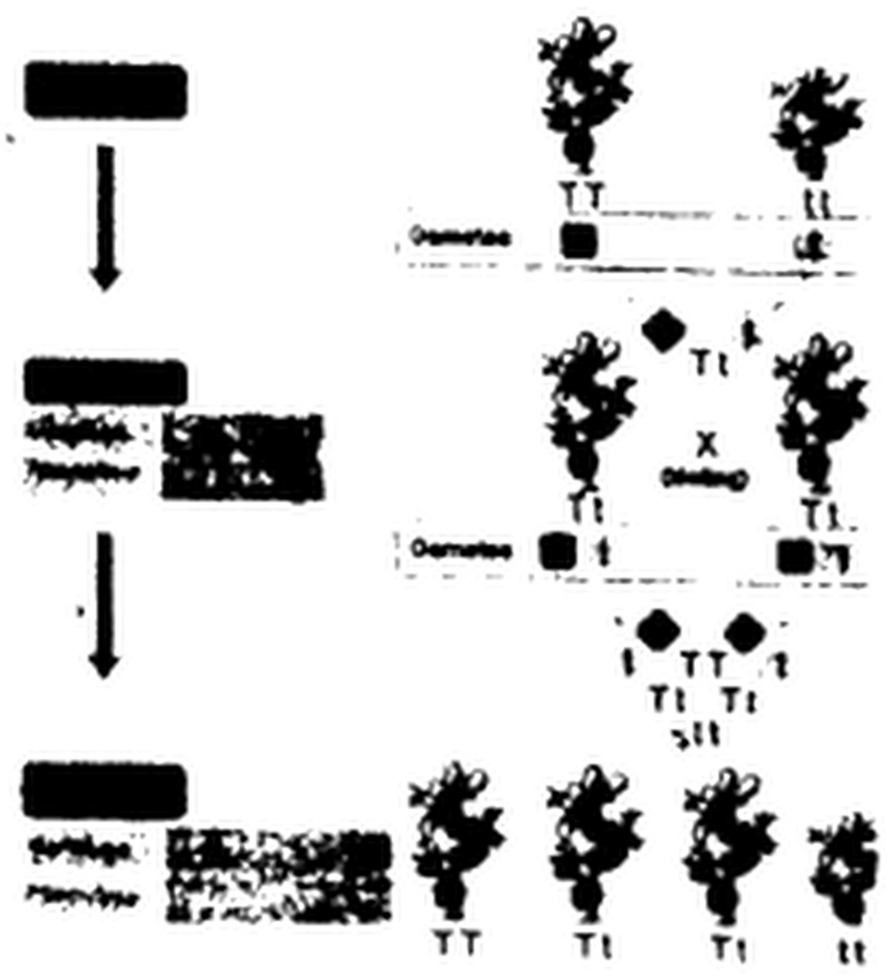


Fig. Monohybrid cross showing the inheritance of single trait

Mendel then theorized that genes can be made up of three possible pairings of heredity units, which he called factors TT, Tt and tt. The capital letter T represents the dominant factor and the small letter t represents the recessive factor. In Mendels crosses, the starting plants were homozygous TT or tt, the F_1 generation was Tt, and the F_2 generation was TT, Tt, or tt (Fig. 22.1). The interaction between these two alleles determines the physical trait that is visible to us.

Mendel's Laws of Inheritance

Mendels observations and conclusions are summarized in the following three principles or laws.

Law of Dominance

It states that when mating occurs between two parents of contrasting traits, in offspring only one trait will appear and the second trait will be masked. The trait which appears is called dominant trait whereas the trait which is masked is called recessive trait.

Law of Segregation

The law of segregation states that for any trait, each parents pairing of genes (alleles) split and one gene passes from each parent to an offspring. Which particular gene in a pair gets passed onto an offspring is completely up to chance.

Law of Independent Assortment

The law of independent assortment states that different pairs of alleles are passed onto the offspring independently from each other. Therefore, inheritance of genes at one location in a genome does not influence the inheritance of genes at another location.

Q17. a) Define probability. Differentiate sum rule from product rule.
b) Hypothesize that in a dihybrid inheritance pattern of colour and texture of pea seed, the two traits are not interdependent.

Answer

a) The chance to occur an event is called probability. The probability (P) that an event will occur is the number of favourable cases (a) divided by the total number of possible cases (n): $P = a/n$.

For example, when we toss a coin, the probability (P) of onset of head or tail (a) out of two possibilities (n) is $P = a/n$ i.e., $P = ½$.

Mendel had a firm background of mathematics. He understood that the segregation of allele pairs during gamete formation on the re-forming of pairs at fertilization obey the rules of probability. Let us see how the rules of probability apply to inheritance.

In genetics, the inheritance of a specific phenotype in a cross also has certain probability; like in monohybrid cross the probability of an offspring to inherit dominant phenotype in F_1 generation is 100% and the probability of inheritance of recessive phenotype in F_1 generation is 0%. Whereas, probability of dominant phenotype in F_2 generation is ¾ and probability of recessive phenotype in F_2 generation is ¼.

The probabilities of two or more events can be combined in two different ways, i.e.

Sum Rule

The probability of either one or the other events out of many events can be calculated by sum rule (addition rule). According to this rule the probability of either homozygous dominant (1/4) or homozygous recessive (1/4) offspring in F_2 generation of monohybrid cross will be equal to sum of their individual probabilities. i.e., P = 1/4 + 1/4 = 2/4 = 1/2.

Product Rule

The combined probability of two or more independent events can be calculated by product rule (multiplication rule). According to this rule the probability of round yellow phenotype in F_2 generation of a dihybrid cross is equal to the product of individual probabilities of round (3/4) and yellow (3/4) phenotypes. i.e., P = 3/4 x 3/4 = 9/16.

b) Hypothesize that in a dihybrid inheritance pattern of colour and texture of pea seed, the two traits are not interdependent.

It has already been observed that characters like colour of pea seed and texture of pea seed are inherited independently of each other. The explanation lies in the behaviour of the chromosomes at meiosis. Independent assortment requires that genes concerned are carried on different chromosomes. For example, the alleles of the gene for seed colour are located on one pair of chromosomes and the alleles of the gene for texture of the seed on another pair of chromosome.

Q18.a) Explain codominance. Evaluate incomplete dominance and codominance as variations of mendels research.

b) Differentiate incomplete dominance from codominance.

Answer

a) Co-dominance

Another variation on dominance relationship between alleles is called co-dominance, in which both contrasting alleles at the same locus express independently without influencing each other, so the phenotype of both the alleles become apparent. For example, the human MN blood group is determined by two co-dominant alleles (L^M and L^N) for two specific molecules located on the surface of red blood cells, the M and N molecules. A single gene locus at which two allelic variations are possible, determines the phenotypes of this blood group. Individuals homozygous for L^M allele (L^ML^M) have red blood cells with only M molecules; individuals homozygous for L^N allele (L^NL^N) have red blood cells with only N molecules; but both M and N molecules are present on the red blood cells of individuals heterozygous for M and N alleles (L^ML^N).

Table 22.1 MN Blood Groups System in Example of Co-dominance

Blood Group phenotype	Antigen	Genotype
M blood	M antigen	L^ML^M
N blood	N antigen	L^NL^N
MN blood	Both antigen	L^ML^N

Evaluate Incomplete and co-dominance as Variations of Mendels Research

Many patterns of inheritance which cannot be explained on the basis of Mendel's laws alone were discovered in plants and animals. Such patterns of inheritance are described as Non-Mendelian inheritance. Incomplete dominance is a type of interaction where both the alleles of a given trait express as a blend (mixture) as against a normal Mendelian pattern where one allele is dominant over the other. As a result of this blending, an intermediate character is expressed. Co-dominance represents a situation where two allelic genes when present together in an individual, express their traits independently instead of showing a typical dominant recessive relationship. As a result, the heterozygous progeny of the F_2 generation show a phenotype that is different from both the homozygous parents.

b)

Difference Between Incomplete Dominance and Co-dominance	
Incomplete dominance	**Co-dominance**
1. In heterozygous state, both genes blend their phenotypic effects.	1. In heterozygous state, both genes independently express their phenotypic effects.
2. The heterozygotes show an intermediate phenotype between the two parental phenotypes.	2. The heterozygotes show both parental phenotypes at a time.
3. Example: Flower colour of 4 O'clock plant.	3. Example: Human MN blood and AB blood groups.

Q19.a) What is a transfusion principle? Derive an idea to get alternatives of blood transfusion.

b) Justify why a recessive blood group allele '*i*' is more frequent in the population.

c) Justify blood donation as service to suffering humanity.

Answer

a) Transfusion Principle

When transfusions are carried out between two incompatible (different) blood groups, antigens of donor react with the antibodies (also called agglutinins) of the recipient, then the red blood cells clumps with one another. The clumping of red blood cells is known as agglutination. Therefore, the transfusions are carried out on the basis of donor's antigens and recipients antibodies. Due to these limitations the persons with type A blood group can receive blood from type A or from type O because they have anti B antibody so they cannot be given any blood carrying B antigen.

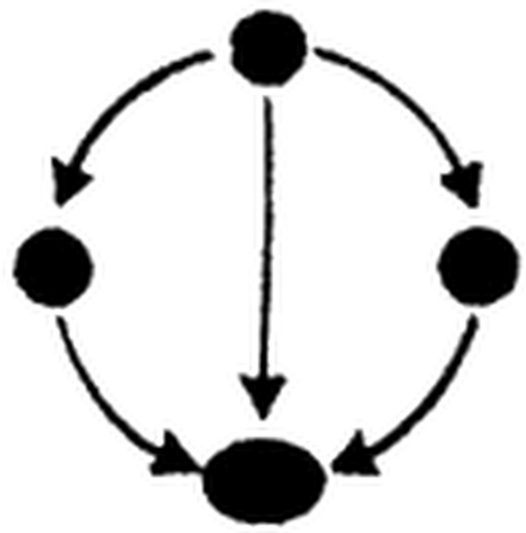


Fig. Blood Transfusion Model

blood from type B or from type O but those with type AB blood group can receive any blood since they do not have antibody to react with donor's blood, hence they are called universal recipients. The persons with type O blood group can receive blood only from type O because they have both antibodies that can react with any antigenic blood (type A, B or AB) but they can donate to any one as they do not have any antigen to interfere with recipients blood. Therefore they are called universal donor.

Derive an idea to get alternatives of blood transfusion

Some approaches are available that can decrease the need for a blood transfusion. The options currently available are:

1) Volume expanders are used to prevent or treat the shock associated with loss of body fluids. The most common volume expanders used include salt water (normal saline) and saline with some chemicals added (Ringers solution).
2) Hematopoietic growth factors encourage the bone marrow to make more red blood cells. These growth factors can be made in the laboratory and given to people with low blood cell counts.
3) Erythropoetin is a naturally occurring hormone produce by the kidneys. It stimulates the body to produce more red blood cells and is used to treat anemia. It is widely used as a transfusion alternative.
4) Aprotinin is a drug that is given prior to heart surgery to reduce the risk of bleeding and the need for transfusion.

b) Justify why a recessive blood group allele I is more frequent in the population

The cross between blood group O (ii) and O (ii) will have blood group O (ii) in all the offsprings. The cross between heterozygous I^Ai and I^Ai will produce 25% offsprings having blood group O (ii), and likewise heterozygous I^Bi and I^Bi will produce 25% offsprings having blood group O (ii). Cross between heterozygous I^Ai and I^Bi will produce 25% offsprings having blood group O (ii). That's why blood group allele i is more frequent in the population. For example, Australia 40%, Canada 39%, Iceland 47%, Ireland 47% UK 37%, USA 37%.

c) Justify blood donation as a service to suffering humanity

Blood donation is a social responsibility. The donor is donating for it as it will be used in saving lives of human beings. As The Quran says in Surah 5 verse 32 if anyone saves a life, it shall be as though he had saved the lives of all mankind. Millions of people owe their lives to people whom they will never know or meet in their lifetime. They are none other than those people, who have donated their blood freely and without any reward voluntary blood donors. Voluntary unpaid donors are the foundation of a safe blood supply which saves millions of human beings from the jaws of untimely death. Blood donation is a noble, selfless service. It gives the donor a felling of joy and contentment. Also this is an expression of love for mankind, as blood knows no caste, colour, creed, religion or race, country, continent or sex. Do you know that one unit of blood can save three lives rather than one?

Q20. a) What is a Rh blood group system? What are different types of antigens in Rh blood group system?
b) What is a Anti-Rh-antibody and transfusion principle?
c) What are genetic basis of Rh blood group system?

Answer

Rh Blood Group System

The Rh (Rhesus) blood group system (including the Rh factor) is one of the currently 30 human blood group systems. It is clinically the most important blood group system after ABO. The name of this system (Rh) is derived from Rhesus monkey, because its antigen was first discovered in it by Landsteriner in 1930s.

Antigens of Rh Blood Group System

The Rh blood group system currently consists of 50 defined blood-group antigens, among with the 5 antigens D, C, c, E, and e are the most important ones. The commonly-used terms Rh factor, Rh positive and Rh negative refer to the D antigen only. So the persons having this antigen are called Rh positive and those in which it is absent are called Rh negative. Other antigens of Rh blood group system have no significant role in blood transfusion. On the other hand the D antigen incompatibility between donor and recipient can cause problem not only during blood transfusion but it is also a relevant cause of the haemolytic disease of the newborn or erythroblastosis foetalis.

Anti Rh-Antibody and Transfusion Principle

Rh blood group system also has a mechanism of antibody production i.e., anit-Rh antibody, which is produced in Rh negative blood. Unlike ABO antibody production mechanism, the production of anti Rh antibody is different in the sense that it always requires a stimulus in the form of exposure to Rh factor for its production. An Rh negative person does not produce anti-Rh antibodies unless he is exposed to Rh antigen. Rh positive donor is totally incompatible for Rh negative recipient. If Rh negative person receives an Rh antigen through wrong Rh positive blood transfusion, he will begin to produce anti-Rh antibodies against Rh antigens. Once the mechanism of anti-Rh antibody production is stimulated, then it remains continued for whole life. Rh negative blood, clear of any anti-Rh antibody from a donor who has never been exposed to Rh antigen can be transfused to Rh positive recipient.

Genetic Basis of Rh Blood Group System

Rh blood group system is encoded by three genes C, D, and E, which 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, because it is associated with the formation of D antigen (commonly known as Rh factor). Gene D has two alleles, D and d.D is completely dominant over d. Persons having genotypes DD or Dd have D antigen (Rh factor) on their RBC and are Rh positive. Persons with genotype dd do not have Rh factor and are Rh negative.

Table Rh-Blood Groups System				
Blood group (phenotype)	**Rh-Antigen/factor**	**Genotypes**	**Anti Rh-Antibody**	**Transfusions**
Rh^{+ve}	Present	DD or Dd	Not produced	Rh^{+ve}
Rh^{-ve}	Absent	Dd	Produced (if stimulated)	Rh^{-ve}

The blood group O^{+ve} is actual universal donor because it has no antigen of ABO system and of Rh system which can interfere with recipients blood, whereas, AB^{+ve} is actual universal recipient because it has neither anti-A and anti-B nor anti-Rh antibodies, therefore, it cannot resist any donors blood.

Q21. What are the techniques employed for embryonic screening?

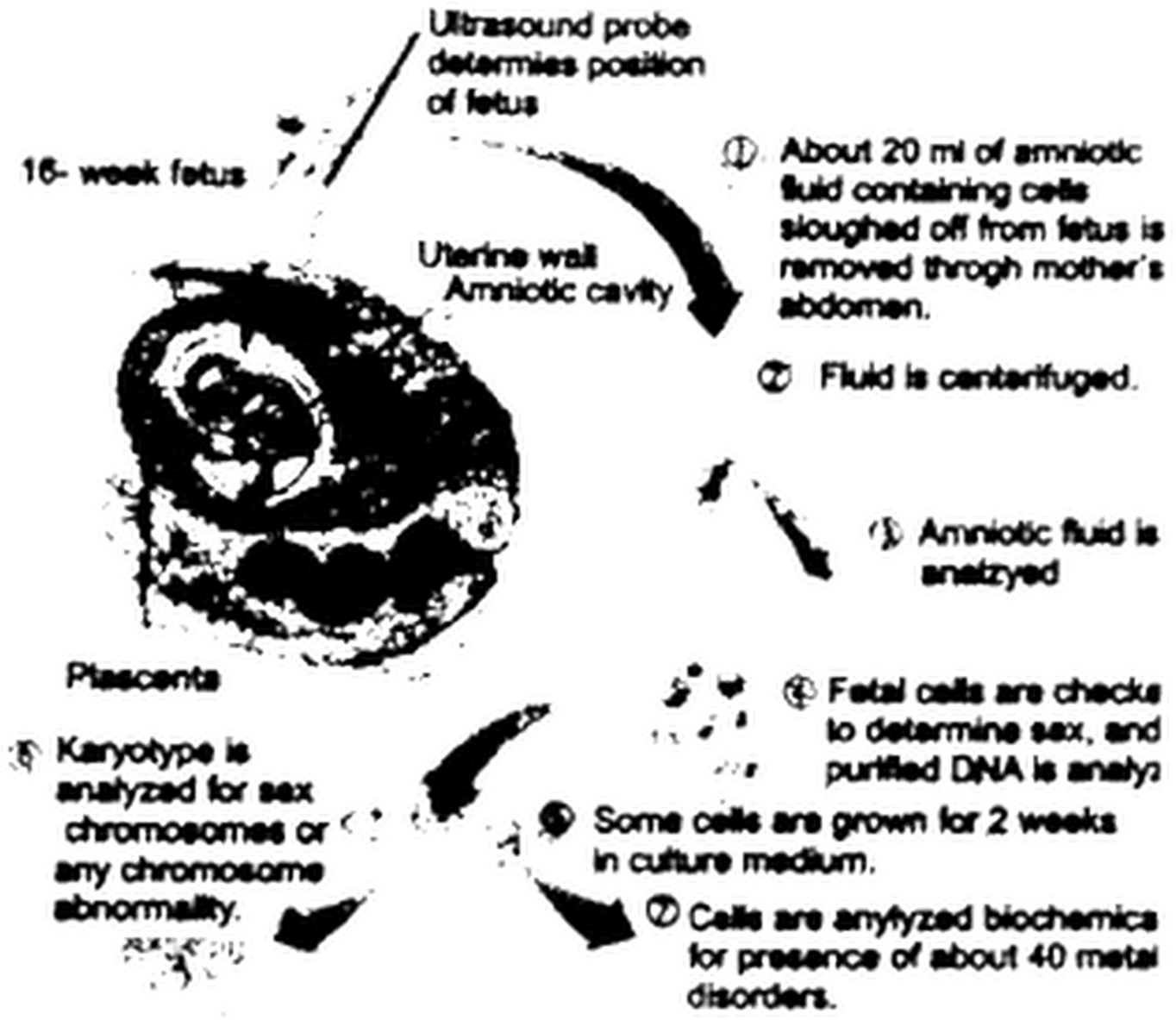
Answer

Techniques Employed for Embryonic Screening

Embryo screening checks an embryo to see if it's carrying any alleles for a genetic disease. A number of techniques are now available to test for chromosomal and gene mutations in human fetuses.

Amniocentesis

Before the start of the procedure, a local anesthetic can be given to the mother. A needle is usually inserted through the mother's abdominal wall, then through the wall of the uterus, and finally into the amniotic sac. With the aid of ultrasound-guidance, a physician punctures the sac in an area away from the foetus and extracts approximately 20ml of amniotic fluid. After the amniotic fluid is extracted, the foetal cells are separated from the sample. The cells are grown in a culture medium, then fixed and stained. Under a microscope the chromosomes are examined for abnormalities e.g., Down Syndrome (trisomy 21).



Aminocentesis

Chorionic Villi Sampling

Here a narrow tube is inserted through the cervix and a tiny sample is taken of the chorionic villi from the placenta. Then chromosomal staining and examination is carried out to test for chromosome mutations in the foetus.

Alpha Foetoprotein Screening

The concentration in the mother blood of a particular protein called alpha foetoprotein is determined. An increased maternal serum concentration of alpha protein shows Spina bifida, anencephaly. An unusually low alpha fetoprotein is associated with an increased risk of Down Syndrome. Imaging techniques allows a physician to examine a foetus directly for major anatomical abnormalities.

Ultrasound Technique

Sound waves are used to produce an image of the fetus by a simple non-invasive procedure. It is used to test for a wide range of abnormalities e.g., hydrocephaly, various types of congenital heart diseases, cleft of the lip and plate and bone abnormalities.

Q22. Explain the following types of sex linkages in man.

i) X-Linked recessive inheritance.
ii) X-Linked dominant inheritance.
iii) Y-Linked inheritance.

Answer

blindness are recessive while others like hypophosphatemic or vitamin D resistant rickets are dominant. X linked dominant is a trait which is determined by an X linked dominant gene, while X linked recessive is a trait that is determined by as X linked recessive gene. Their patterns of inheritance are very different from each other.

Experimental mating is not practically possible in humans. Mode of inheritance of human traits can be traced through pedigrees.

X-Linked Recessive Inheritance

Characteristics of X-Linked Recessive Inheritance: Females possessing one X-linked recessive mutation are considered carriers and will generally not manifest clinical symptoms of the disorder. All males possessing an X-linked recessive mutation will be affected (males have a single X-chromosome and therefore have only one copy of X-linked genes). All offspring of a carrier female have a 50% chance of inheriting the mutation. All female children of an affected father will be carriers (daughters possess their fatrher's X – chromosome). No male children of an affected father will be affected (sons do not inherit their fathers' X-chromosome). Examples: Duchenne Muscular Dystrophy, Hunter's Disease, Haemophilia A and B, Colour Blindness.

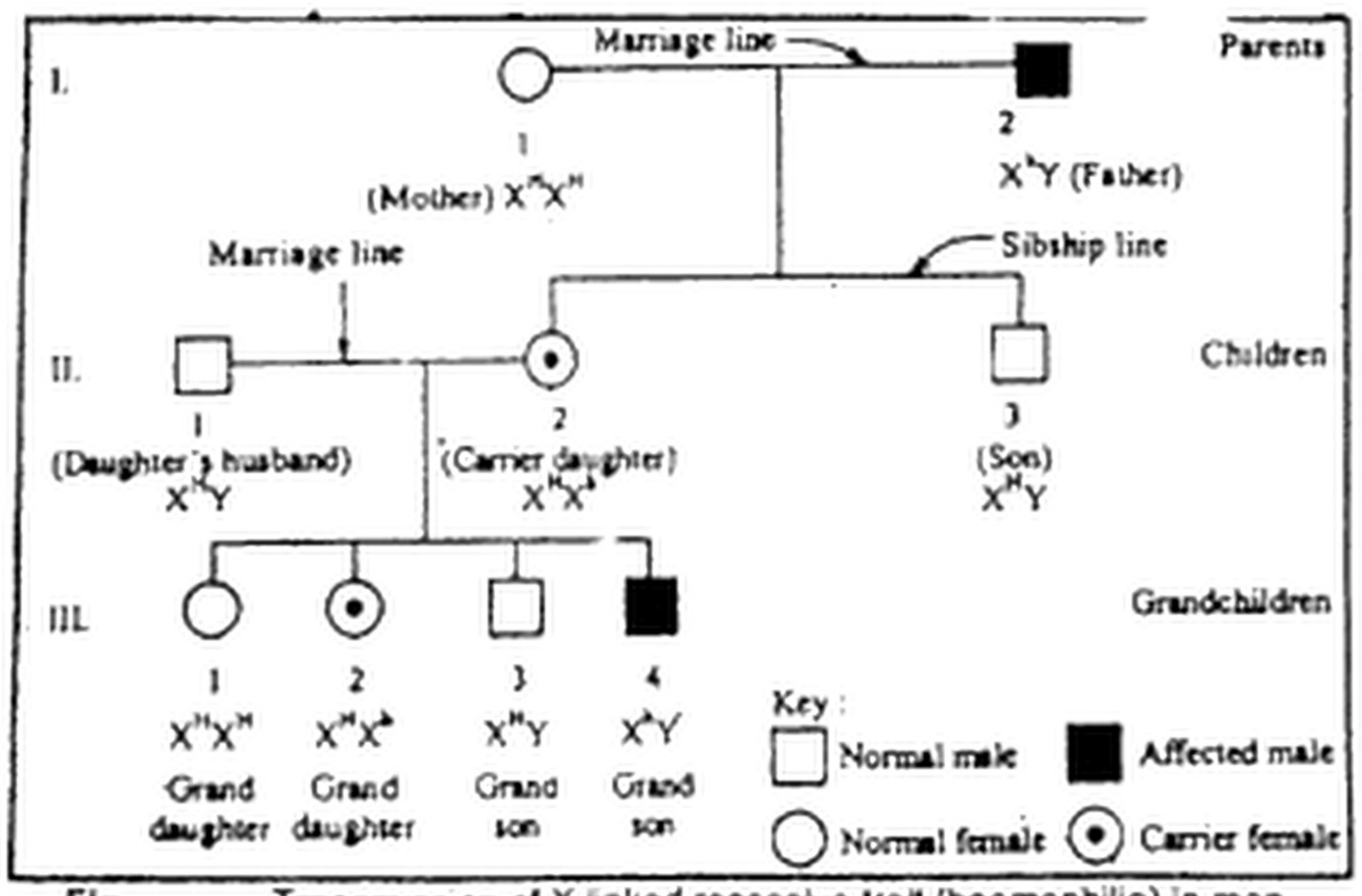


Fig., Transmission of X-linked recessive trait (haemophilia) in man.

X-Linked Dominant Inheritance

Characteristics of X-Linked Dominant Inheritance: A male or female child of an affected mother has a 50% chance of inheriting the mutation and thus being affected with the disorder. All female children of an affected father will be affected (daughters possess their father's X-chromosome). No male children of an affected father will be affected (sons do not inherit their father's X-chromosome). Example: Coffin-Lowry syndrome, Vitamin D resistant rickets, Fragile X syndrome (Fig 22.22).

Fig Hairy

Y-Linked Inheritance

(also hemizygous). The Y-linked genes are located only on the non-homologous region of Y chromosome. Their phenotypic expression is observed only in the males. They are inherited from father to son, as son only receives Y-chromosome from the father. The examples of Y-linked characters in man are: hypertrichosis (growth of hair on the rim of pinna), porcupine man (straight hair on the body) and webbing of toes. Recently two more genes have been discovered, testis determining factor (TDF) and minor histocompatibility gene (H-Y).

Q23. a) What are sex-related traits? Explain sex limited traits and sex influenced traits.

b) How genetic research is important to reduce the risks of diseases?

c) Describe how field of genetics has progressed to a more applied science.

Answer

Sex Related Traits

Sex related traits are those which are associated with maleness or femaleness. These traits are not necessarily being sex linked. These traits may be controlled by sex linked or autosomal genes. They are of two different types i.e. sex limited traits and sex influenced traits.

Sex Limited Traits

A sex limited trait is a type of sex related trait which is confined to only one sex due to anatomical differences. Such traits affect a structure or function of the body present in only males or in only females. Example: Genes for milk yield in dairy cattle affect only cows. Similarly 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 son.

Sex Influenced Trait

Sex influenced traits are also the type of sex related traits. These occur in both males and females but they are more common in one sex. Such traits are controlled by an allele that is expressed as dominant in one but recessive in the other. This difference in expression is due to hormonal difference between the sexes. The amounts of body, hair, and muscle mass and male pattern balding are sex influenced traits. For example, many more men than women are bald. It is inherited as an autosomal dominant trait in males but as an autosomal recessive trait in females. A heterozygous male is bald but a heterozygous female is not. A woman can be bald only when she is homozygous recessive.

Table 22.8 Pattern Baldness in Humans

Genotype	Phenotype in male	Phenotype in female
B_1B_1	Bald	Bald
B_1B_2	Bald	Normal hair

b) Suggest Ways to Save Lives through the Knowledge Gained through Research

Genetic research helps identify diseases and health problems that are more likely to be influenced by genetic factors as well as to assess the risk of particular diseases in an individual. These researches are known as genetic tests. When a genetic test confirms a high risk of certain condition, an expert in the field determines preventive measures to reduce the risk of that particular disease. Genetic testing is very reliable, however, it cannot tell you for sure whether you will develop a particular disease or not.

c) Describe How the Field of Genetics has Progressed to a More Applied Science

With the discovery of DNA, the field of genetics has progressed as more applied science than just a theoretical subject. Many DNA based techniques are now widely used in various fields of life. Such as DNA sequencing technique, DNA fingerprinting, PCR etc.. Genetic engineering or recombinant DNA technology is an applied field of genetics.

Q24. a) Justify the effectiveness of some of the treatments of haemophilia.

b) What are the types of haemophilia and their treatment?

Answer

a) Justify the Effectiveness of Some of the Treatments of Haemophilia

Treatment for haemophilia today is very effective. The missing clotting factor is injected into the bloodstream using a needle. Bleeding stops when enough clotting factors reach the spot that is bleeding. With an adequate quantity of treatment products and proper care, people with haemophilia can live perfectly healthy lives. Without treatment, most children with severe haemophilia will die young. An estimated 400,000 people worldwide are living with haemophilia and only 25% receive adequate treatment. The World Federation of Haemophilia is striving to close this gap.

b) Types of Haemophilia and their Treatment

Cases of mild haemophilia A, can be treated using a type of medication called desmopressin. Desmopressin is a synthetic hormone. Desmopressin works by stimulating the production of clotting agent VIII and is usually given by injection. Haemophilia B which is caused by a lack of clotting agent IX is treated with a medication called nonacogalfan which is an engineered version of clotting agent IX. Haemophilia A is treated using a synthetic version of clotting agent VIII, called octocogalfa, which is another type of genetically engineered purified protein. Persons with severe hemophilia C do not require treatment or prophylactic (preventive) therapy for daily activities. However, replacement therapy is required for dental extractions and surgery, and treatment options depend on the type of procedure. The most common type of treatment for haemophilia C is the infusion of healthy blood plasma. The main