Question 1:

Mention the advantages of selecting pea plant for experiment by Mendel.

Answer

Mendel selected pea plants to carry out his study on the inheritance of characters from parents to offspring.

He selected a pea plant because of the following features.

(a) Peas have many visible contrasting characters such as tall/dwarf plants, round/wrinkled seeds, green/yellow pod, purple/white flowers, etc.

(b) Peas have bisexual flowers and therefore undergo self pollination easily. Thus, pea plants produce offsprings with same traits generation after generation.

(c) In pea plants, cross pollination can be easily achieved by emasculation in which the stamen of the flower is removed without affecting the pistil.

(d) Pea plants have a short life span and produce many seeds in one generation.

Question 2:

Differentiate between the following –

(a) Dominance and Recessive

(b) Homozygous and Heterozygous

(c) Monohybrid and Dihybrid.

Answer

(a) Dominance and Recessive

	Dominance	**Recessive**
1.	A dominant factor or allele expresses itself in the presence or absence of a recessive trait.	A recessive trait is able to express itself only in the absence of a dominant trait.
2.	For example, tall plant, round seed, violet flower, etc. are dominant characters in a pea plant.	For example, dwarf plant, wrinkled seed, white flower, etc. are recessive traits in a pea plant.

(b) Homozygous and Heterozygous

	Homozygous	**Heterozygous**
1.	It contains two similar alleles for a particular trait.	It contains two different alleles for a particular trait.
2.	Genotype for homozygous possess either dominant or recessive, but never both the alleles. For example, RR or rr	Genotype for heterozygous possess both dominant and recessive alleles. For example, Rr
3.	It produces only one type of gamete.	It produces two different kinds of gametes.

(c) Monohybrid and Dihybrid

	Monohybrid	**Dihybrid**
1.	Monohybrid involves cross between parents, which differs in only one pair of contrasting characters.	Dihybrid involves cross between parents, which differs in two pairs of contrasting characters.
2.	For example, the cross between tall and dwarf pea plant is a monohybrid cross.	For example, the cross between pea plants having yellow wrinkled seed with those having green round seeds is a dihybrid cross.

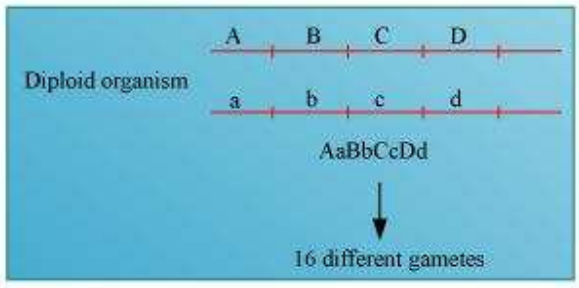
Question 3:

A diploid organism is heterozygous for 4 loci, how many types of gametes can be produced?

Answer

Locus is a fixed position on a chromosome, which is occupied by a single or more genes. Heterozygous organisms contain different alleles for an allelic pair. Hence, a diploid organism, which is heterozygous at four loci, will have four different contrasting characters at four different loci.

For example, if an organism is heterozygous at four loci with four characters, say Aa, Bb, Cc, Dd, then during meiosis, it will segregate to form 8 separate gametes.



If the genes are not linked, then the diploid organism will produce 16 different gametes. However, if the genes are linked, the gametes will reduce their number as the genes might be linked and the linked genes will be inherited together during the process of meiosis.

Question 4:

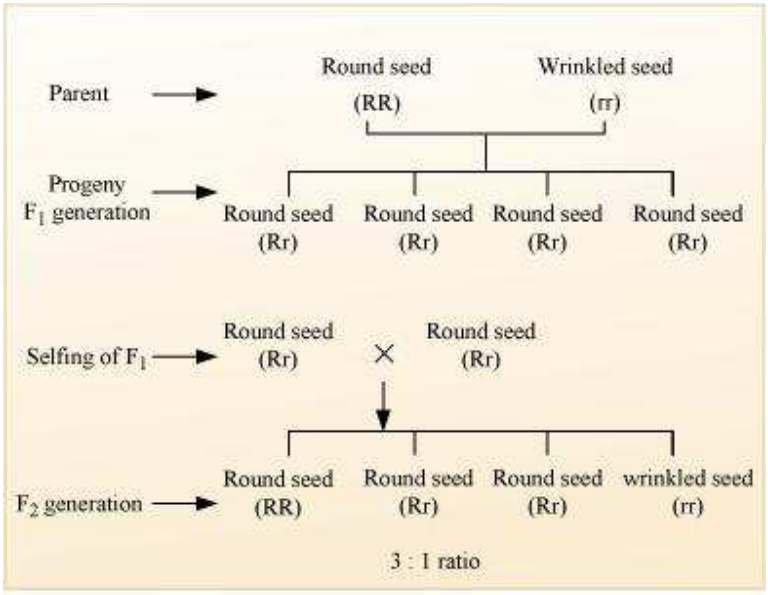
Explain the Law of Dominance using a monohybrid cross.

Answer

Mendel's law of dominance states that a dominant allele expresses itself in a monohybrid cross and suppresses the expression of recessive allele. However, this recessive allele for a character is not lost and remains hidden or masked in the progenies of F_1 generation and reappears in the next generation.

For example, when pea plants with round seeds (RR) are crossed with plants with wrinkled seeds (rr), all seeds in F_1 generation were found to be round (Rr). When these round seeds were self fertilized, both the round and wrinkled seeds appeared in F_2 generation in 3: 1 ratio. Hence, in F_1 generation, the dominant character (round

seeds) appeared and the recessive character (wrinkled seeds) got suppressed, which reappeared in F_2 generation.



A monohybrid cross between round and wrinkled pea seeds

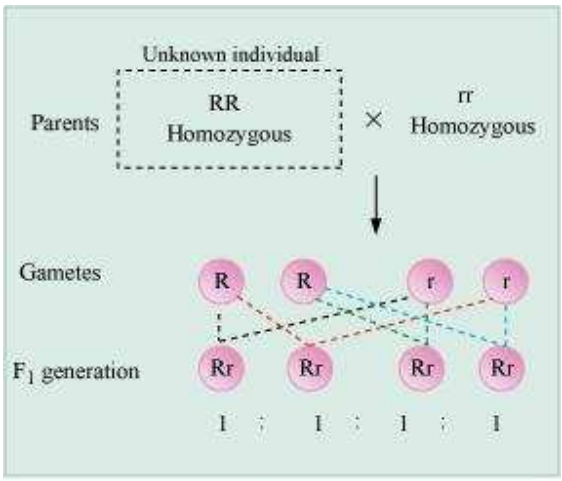
Question 5:

Define and design a test – cross?

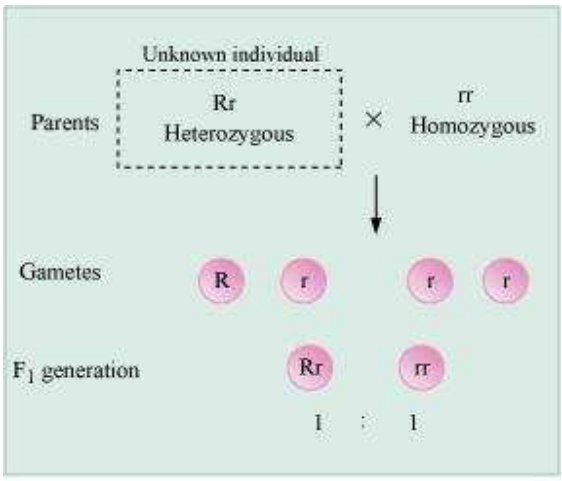
Answer

Test cross is a cross between an organism with unknown genotype and a recessive parent. It is used to determine whether the individual is homozygous or heterozygous for a trait.

If the progenies produced by a test cross show 50% dominant trait and 50% recessive trait, then the unknown individual is heterozygous for a trait. On the other hand, if the progeny produced shows dominant trait, then the unknown individual is homozygous for a trait.



Cross between homozygous (unknown) individual and homozygous recessive individual



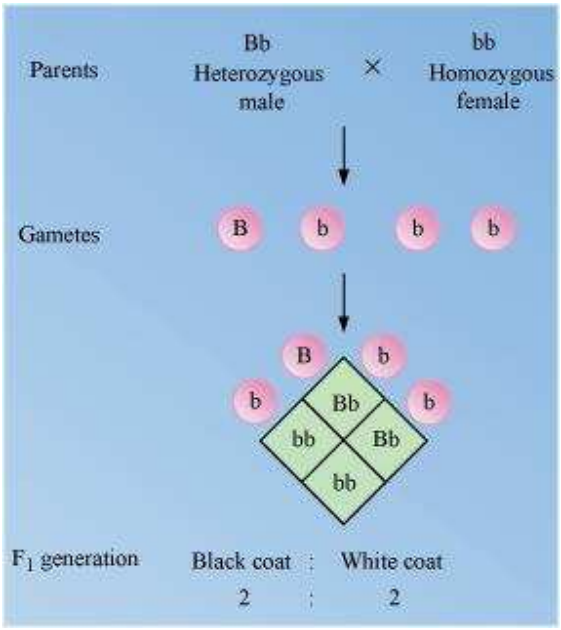
Cross between homozygous (unknown) individual and homozygous recessive individual

Question 6:

Using a Punnett square, work out the distribution of phenotypic features in the first filial generation after a cross between a homozygous female and a heterozygous male for a single locus.

Answer

In guinea pigs, heterozygous male with black coat colour (Bb) is crossed with the female having white coat colour (bb). The male will produce two types of gametes, B and b, while the female will produce only one kind of gamete, r. The genotypic and phenotypic ratio in the progenies of F_1 generation will be same i.e., 1:1.



Question 7:

When a cross in made between tall plants with yellow seeds (TtYy) and tall plant with green seed (TtYy), what proportions of phenotype in the offspring could be expected to be

(a) Tall and green.

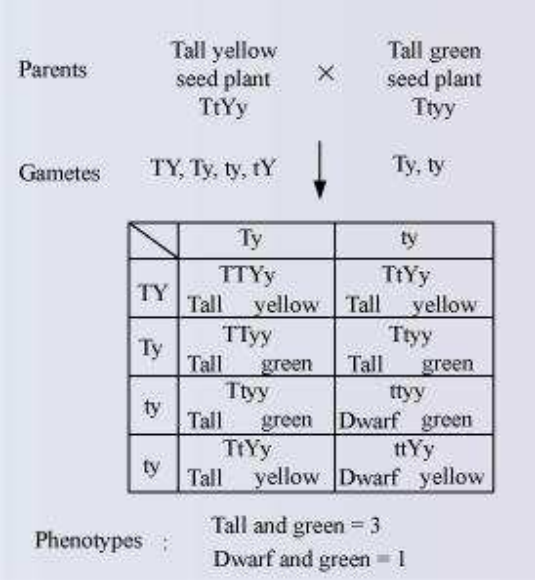
(b) Dwarf and green.

Answer

A cross between tall plant with yellow seeds and tall plant with green seeds will produce

(a) three tall and green plants

(b) one dwarf and green plant



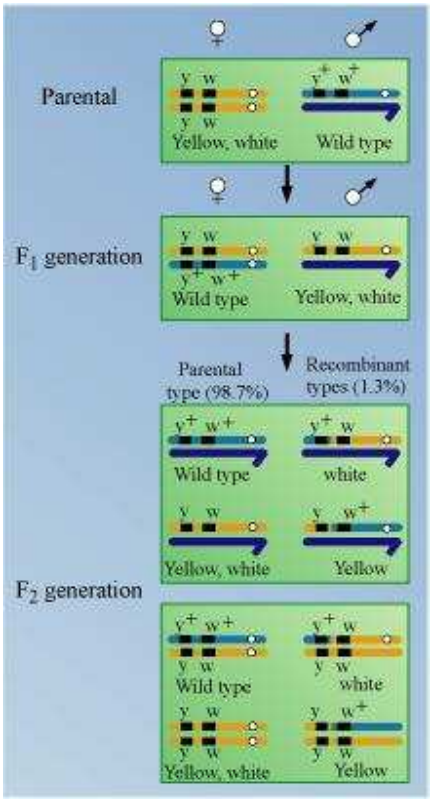
Question 8:

Two heterozygous parents are crossed. If the two loci are linked what would be the distribution of phenotypic features in F_1 generation for a dihybrid cross?

Answer

Linkage is defined as the coexistence of two or more genes in the same chromosome. If the genes are situated on the same chromosome and lie close to each other, then they are inherited together and are said to be linked genes.

For example, a cross between yellow body and white eyes and wild type parent in a *Drosophila* will produce wild type and yellow white progenies. It is because yellow bodied and white eyed genes are linked. Therefore, they are inherited together in progenies.



Question 9:

Briefly mention the contribution of T.H. Morgan in genetics.

Answer

Morgan's work is based on fruit flies (*Drosophila melanogaster*). He formulated the chromosomal theory of linkage. He defined linkage as the co-existence of two or more genes in the same chromosome and performed dihybrid crosses in *Drosophila* to show that linked genes are inherited together and are located on X-chromosome. His experiments have also proved that tightly linked genes show very low recombination while loosely linked genes show higher recombination.

Question 10:

What is pedigree analysis? Suggest how such an analysis, can be useful.

Answer

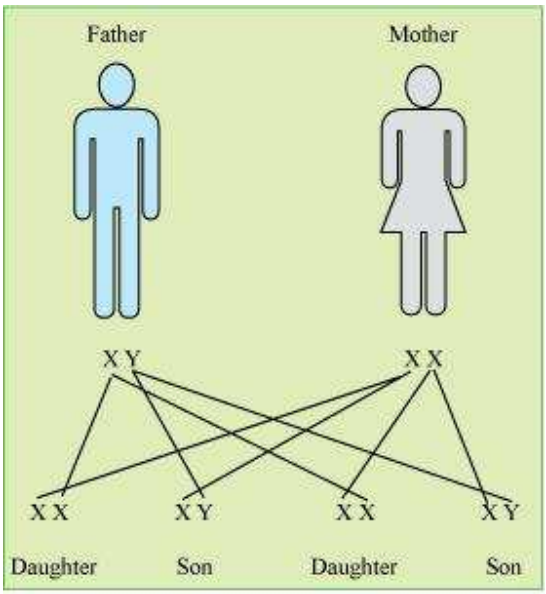
Pedigree analysis is a record of occurrence of a trait in several generations of a family. It is based on the fact that certain characteristic features are heritable in a family, for example, eye colour, skin colour, hair form and colour, and other facial characteristics. Along with these features, there are other genetic disorders such as Mendelian disorders that are inherited in a family, generation after generation. Hence, by using pedigree analysis for the study of specific traits or disorders, generation after generation, it is possible to trace the pattern of inheritance. In this analysis, the inheritance of a trait is represented as a tree, called family tree. Genetic counselors use pedigree chart for analysis of various traits and diseases in a family and predict their inheritance patterns. It is useful in preventing hemophilia, sickle cell anemia, and other genetic disorders in the future generations.

Question 11:

How is sex determined in human beings?

Answer

Human beings exhibit male heterogamy. In humans, males (XY) produce two different types of gametes, X and Y. The human female (XX) produces only one type of gametes containing X chromosomes. The sex of the baby is determined by the type of male gamete that fuses with the female gamete. If the fertilizing sperm contains X chromosome, then the baby produced will be a girl and if the fertilizing sperm contains Y chromosome, then the baby produced will be a boy. Hence, it is a matter of chance that determines the sex of a baby. There is an equal probability of the fertilizing sperm being an X or Y chromosome. Thus, it is the genetic make up of the sperm that determines the sex of the baby.



Sex determination in humans

Question 12:

A child has blood group O. If the father has blood group A and mother blood group B, work out the genotypes of the parents and the possible genotypes of the other offsprings.

Answer

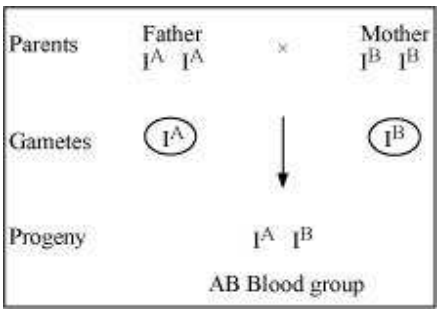
The blood group characteristic in humans is controlled by three set of alleles, namely, I^A, I^B, and i. The alleles, I^A and I^B, are equally dominant whereas allele, i, is recessive to the other alleles. The individuals with genotype, I^A I^A and I^A i, have blood group A whereas the individuals with genotype, I^B I^B and I^B i, have blood group B. The persons with genotype I^A I^B have blood group AB while those with blood group O have genotype ii.

Hence, if the father has blood group A and mother has blood group B, then the possible genotype of the parents will be

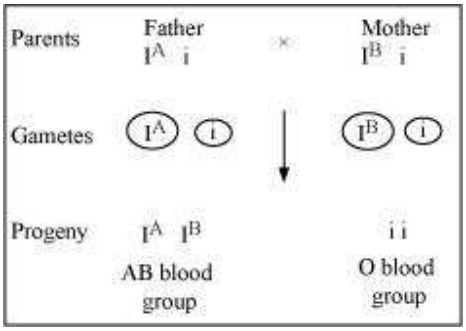
Father Mother

I^A I^A or I^A i I^B I^B or I^B i

A cross between homozygous parents will produce progeny with AB blood group.



A cross between heterozygous parents will produce progenies with AB blood group (I^AI^B) and O blood group (ii).



Question 13:

Explain the following terms with example

(a) Co-dominance

(b) Incomplete dominance

Answer

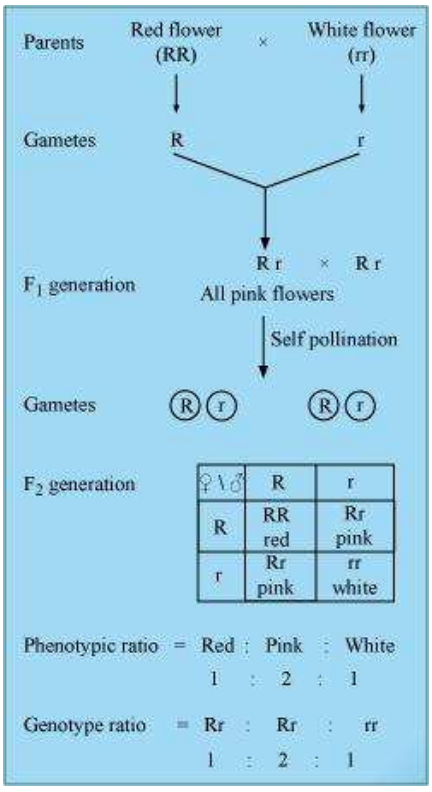
(a) Co-dominance

Co-dominance is the phenomenon in which both the alleles of a contrasting character are expressed in heterozygous condition. Both the alleles of a gene are equally dominant. ABO blood group in human beings is an example of co-dominance. The blood group character is controlled by three sets of alleles, namely, I^A, I^B, and i. The alleles, I^A and I^B, are equally dominant and are said to be co-dominant as they are expressed in AB blood group. Both these alleles do not interfere with the expression

of each other and produce their respective antigens. Hence, AB blood group is an example of co-dominance.

2. Incomplete dominance

Incomplete dominance is a phenomenon in which one allele shows incomplete dominance over the other member of the allelic pair for a character. For example, a monohybrid cross between the plants having red flowers and white flowers in *Antirrhinum* species will result in all pink flower plants in F_1 generation. The progeny obtained in F_1 generation does not resemble either of the parents and exhibits intermediate characteristics. This is because the dominant allele, R, is partially dominant over the other allele, r. Therefore, the recessive allele, r, also gets expressed in the F_1 generation resulting in the production of intermediate pink flowering progenies with Rr genotype.



Question 14:

What is point mutation? Give one example.

Answer

Point mutation is a change in a single base pair of DNA by substitution, deletion, or insertion of a single nitrogenous base. An example of point mutation is sickle cell anaemia. It involves mutation in a single base pair in the beta-globin chain of haemoglobin pigment of the blood. Glutamic acid in short arm of chromosome II gets replaced with valine at the sixth position.

Question 15:

Who had proposed the chromosomal theory of inheritance?

Answer

Sutton and Boveri proposed the chromosomal theory of inheritance in 1902. They linked the inheritance of traits to the chromosomes.

Question 16:

Mention any two autosomal genetic disorders with their symptoms.

Answer

Two autosomal genetic disorders are as follows.

1. Sickle cell Anaemia

It is an autosomal linked recessive disorder, which is caused by point mutation in the beta-globin chain of haemoglobin pigment of the blood. The disease is characterized by sickle shaped red blood cells, which are formed due to the mutant haemoglobin molecule. The disease is controlled by Hb^A and Hb^S allele. The homozygous individuals with genotype, Hb^S Hb^S, show the symptoms of this disease while the heterozygous individuals with genotype, Hb^A Hb^S, are not affected. However, they act as carriers of the disease.

Symptoms

Rapid heart rate, breathlessness, delayed growth and puberty, jaundice, weakness, fever, excessive thirst, chest pain, and decreased fertility are the major symptoms of sickle cell anaemia disease.

(b) Down's syndrome

It is an autosomal disorder that is caused by the trisomy of chromosome 21.

Symptoms

The individual is short statured with round head, open mouth, protruding tongue, short neck, slanting eyes, and broad short hands. The individual also shows retarded mental and physical growth.

CHAPTER 5

PRINCIPLES OF INHERITANCE AND VARIATION

POINTS TO REMEMBER

Allele : Various or slightly different forms of a gene, having same position on chromosomes.

Phenotype : The observable or external characteristics of an organism
Genotype : The genetic constitution of an organism.

Monohybrid cross : A cross between two individuals of species, considering the inheritance of single pair of contrasting character e.g., a cross between pure tall (TT) and Dwarf (tt).

Dihybrid cross : A cross between two individuals of a species, considering the inheritance of two pairs of contrasting traits/characters e.g., a cross between Round and Yellow (RRYY) and wrinkled and green (rryy) pea seeds

Co-dominance : When two alleles of a gene are equally dominant and express themselves even when they are together.

Multiple allelism : When a gene exists in more than two allelic forms e.g., gene for blood group exist in three allelic forms, IA, IB and i.

Aneuploidy : The phenomenon of gain or loss of one or more chromosome(s), that results due to failure of separation of homologous pair of chromosomes during meiosis.

Trisomy : The condition in which a particular chromosome is present in three copies in a diploid cell/ nucleus.

Male heterogamety : When male produces two different types of gametes/ sperms e.g., In human beings X and Y.

Mutation : The sudden heritable change in the base sequence of DNA, or structure of chromosome or a change in the number of chromosomes.

Pedigree Analysis : The analysis of the distribution and movement of trait in a series of generations of a family.

Female heterogamety : When female produces two different types of gametes/ova e.g., female bird produces Z and W gametes.

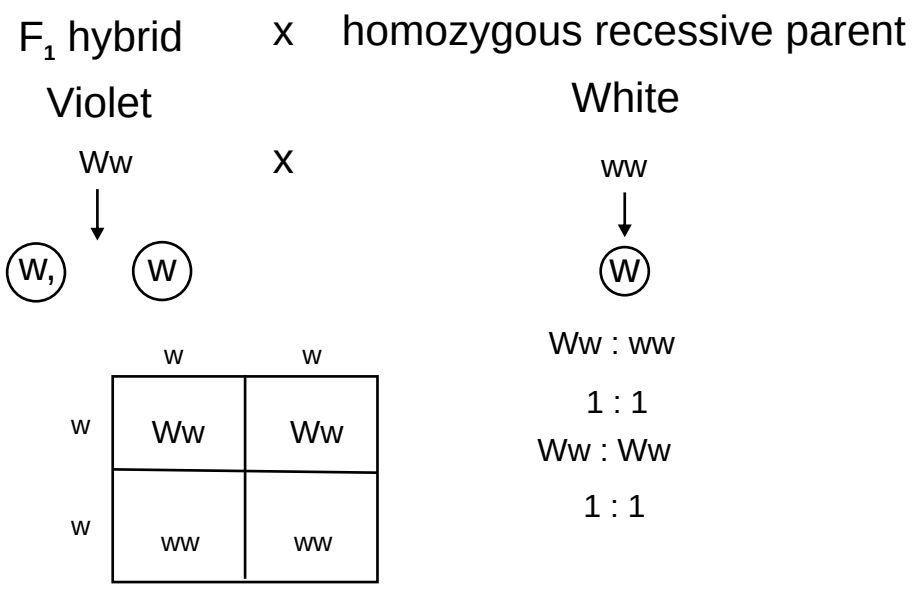
Law of Dominance : When two individuals of a species differing in a pair of contrasting characters/traits are crossed, the trait that appears in the F1 hybrid is dominant and the alternate from that remain hidden, is called recessive.

Law of Segregation : The members of allelic pair that remained together in the parent, segregate/separate during gamete formation and only one of the factors enters a gamete.

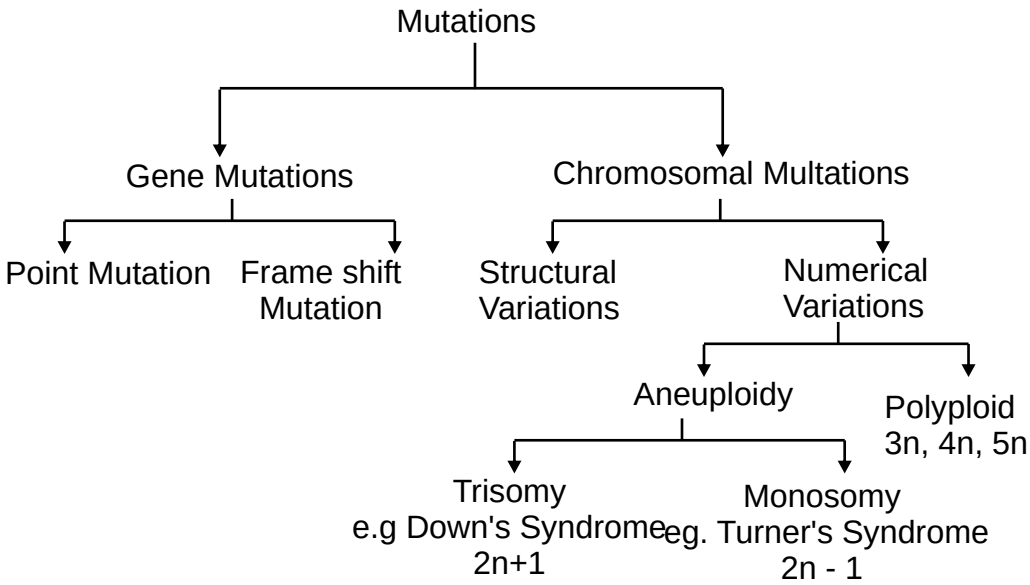
Law of Independent Assortment : In the inheritance of two pairs of contrasting characters, the factors of each pair of characters segregate independently of the factors of the other pair of characters.

Test Cross : When offspring or individual with dominant phenotype, whose genotype is not known, is crossed with an individual who is homozygous recessive for the trait.

The progeny of monohybrid test cross ratio is 1 : 1 while the dihybrid test cross ratio is 1 : 1 : 1 : 1.



Use of Test Cross : The test cross is used to find the genotype of an organism.



Incomplete dominance : It is the phenomenon where none of the two contrasting alleles is dominant but express themselves partially when resent togoether in a hybrid and somewhat intermediate.

Co-dominance : The alleles which do not show dominance recessive relationship and are able to express themselves independently when present together are called co-dominant alleles and this phenomenon is known as codominance.

Example : Human blood groups.

Blood Group	Genotype
A	I^AI^A, I^Ai
B	I^BI^B, I^Bi
AB	I^AI^B
O	ii

In human blood, there are six genotype and four phenotypes.

Chromosomal Theory of Inheritance : proposed by Suttan and Boveri. The pairing and separation of a pair of chromosomes would lead to the segregation of a pair of factors they carried. They united the knowledge of segregation with mendelian principles.

Linkage- is the tendency of genes on a chromosome to remain together.

- Linked genes occur in the same chromosome.

- They lie in linear sequence in the chromosome-There is a tendency to maintain the parental combination of genes except for accessional choosers.
- Strength of linkage between genes is inversely proportional to the distance between the two.

Recombination - is the generation of non-parental gene combinations to the offsprings.

Tightly linked genes show very low recombination frequency. Loosely linked genes show higher recombination frequency.

The frequency of recombination between gene pairs on the same chromosome is a measure of distance between genes and is used to map the position of genes on the chromosome.

Chromosomal basis of sex determination

(i) XX - XY type - female homogametic ie XX and male heterogametic ie. XY is *Drosophila,* humans.

(ii) XX - XO type All eggs bear additional X chromosome, Makes have only one X chromosome besides autosomes whereas females have a pair of X chromosomes eg grasshoppers.

(iii) ZW - ZZ type - The females are hetegametic and have one Z and one W chromosome. The males are homogametic with a pair of Z chromosomes besides autosomes eg - birds.

Pedigree Analysis

A record of inheritance of certain genetic traits for two or more generation

presented in the form of diagram or family tree is called pedigree.

Usefulness of Pedigree Analysis

1. It is useful for genetic counsellors to advice intending couples about the possibility of having children with genetic defects like haemophilia, thalassemia etc.
2. It is helpful to study certain genetic trait and find out the possibility or absence or presence of that trait in homozygous or heterozygous condition in a particular individual.

Mendelian disorders:

These are mainly determined by alternation or mutation in single genes. or

mutation in single genes.

1. **Haemophilia -** sex linked recessive disease which is transmitted from unaffected carriers female to male pregnancy. A single protein is affected that is a part of the cascade of proteins involved in the clothing of blood.

X^hY . Sufferer male

X^hX . carrier female

The heterozygous female for haemophilia may transmit the disease to her sons. The possibility of a female suffering from the disease is extremely rare (only when the mother of the female is a carrier ie X^hX and father is haemophilic ie. X^hY.

2. **Sickle - cell anaemia :** This is an autosome linked recessive trait. The defect is caused by substitution of glutamic acid by valine at the 6th position of the beta globin chain of the haemoglobin molecule. The mutant Hb molecule undergoes polymerisation under low oxygen tension causing change in shape of RBC from biconcave disc to elongated sickle like structure. The disease is controlled by a pair of allele, Hb^A and Hb^S

Hb^AHb^A . Normal Hb^SHb^S

Hb^AHb^S . Apparently unaffected/carriers sufferer

Phenylketonuria - Inborn error of metabolism autosomal recessive trait.

Affected individual lacks an enzyme that converts amino acid Phenylalanine into tyrosine. Phenylalanine is accumulated and converted into phenylpyruvic acid which accumulates in brain resulting in mental retardation.

Thalassemia - Thalassemia is autosome linked recessive disease. This disorder caused by defects in the sysnthesis of globin chain. Thalassemia is of two types - Alpha (α) Thalassemia , Beta (β) Thalassemia.

In alpha Thalassemia production of alpha globin chain is affeceted. This Thalassemia is controlled by genes HBA1 and HBA 2 located on chromosome 16th of each parent. Thalassemia occurs due to mutation or deletion of one or more of the four genes.

- In Beta thalassemia production of β-globin chain is affected this thalassemia is controlled by gene HBB located on 11th chromosome of each parent. It occurs due to one or both HBB genes.
- In Thalassemia too few globin is synthesized whereas in sickle cell anaemia there is a synthesis of incorrectly functioning globin.

Chromosomal disorders

These are caused due to absence or excess of one or more chromosomes.

Colour blindness: Colour blindness is sex-linked recessive trait in which a prism fails to distinguish red and green colour. The gene for normal vision is dominant. The normal genes and its receive alleles are carried by x-chromosome.

X^cX^c ——— Colour blind female

XX^c ——— Carrier female

X^cy ——— Colour blind male

y-chrosome of male do not carry any gene for certain vision.

Down.s syndrome . Trisomy of chromosome number 21.

Affected individual is short statured with small round head, furrowed tongue, partially open month, broad palm. Physical, psychomotor and mental development is retarded.

Klinefelter.s syndrome - extra copy of X chromosome; karyotype XXY.

Affected individual has overall masculine development with feminine characters like gynaecomastia (development of breast) and is sterile.

Turner.s syndrome - has absence of one X chromosome ie. 45 with XO. Affected females are sterile with rudimentary ovaries and lack secondary sexual characters.

PLEIOTROPY

The ability of a gene to have multiple phenotypic effects because it influences a number of characters simultaneously is known as pleiotropy. The gene having a multiple phenotypic effect because of its ability to control expression of a number of characters is called pleiotropic gene.

Eg. in Garden Pea, the gene which controls the flower colour also controls the colour of seed coat and presence of red spot in the leaf axil.

POLYGENIC INHERITANCE

It is a type of inheritance controlled by two or more genes in which the dominant alleles have cumulative effect with each dominant allele expressing a part of the trait, the full trait being shown only when all the dominant alleles are present.

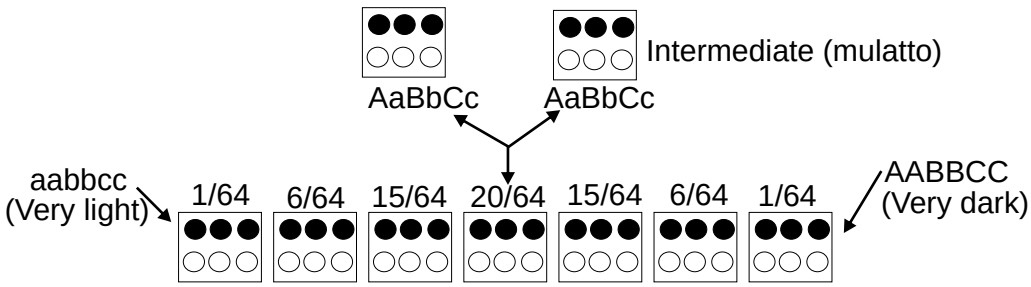
Eg. Kernel colour in wheat, skin colour in human beings, height in humans, cob length in maize etc.

In polygenic inheritance, a cross between two pure breeding parents produces an intermediate trait in F_1. In F_2 generation, apart from the two

parental types, there are several intermediates (gradiations, show a bell shaped curve). F_1 hybrid form 8 kinds of gamete in each sex giving 64 combination in F_2 having 7 genotype and phenotype.

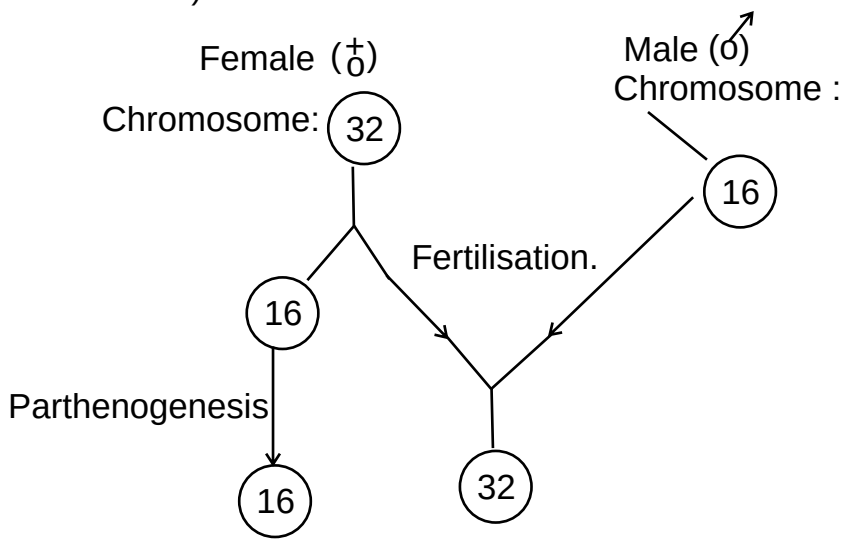
Polygenic inheritance of skin tone

3 loci : each has two possible alleles : Aa, Bb, Cc, each capital allele adds one unit of darkness, each lower case allele adds nothing Parents with intermediate tone .



Offspring can have tone darker or lighter than either parent

Sex Determination In Honey Bee - In Honey bee fertilized eggs develop into female (Queen (or) Worker) While unfertilized egg develops into male (drone) by parthenogenesis. the males have half no. of chromosomes a female. The males are haploid (16 - chromosomes) , females are diploid (32 - chromosomes).



QUESTIONS

VSA (1 MARK)

1. Give any two reasons for the selection of pea plants by Mendel for his experiments.
2. Name any one plant that shows the phenomenon of incomplete dominance during the inheritance of its flower colour.
3. Name the base change and the amino acid change, responsible for sickle cell anaemia.
4. Name the disorder with the following chromosome complement.
   (i) 22 pairs of autosomes + X X Y
   (ii) 22 pairs of autosomes + 21st chromosome + XY.
5. A haemophilic man marries a normal homozygous woman. What is the probability that their daughter will be haemophilic?
6. A test is performed to know whether the given plant is homozygous dominant or heterozygous. Name the test and phenotypic ratio of this test for a monohybrid cross.

SA-II (2 MARKS)

7. Identify the sex of organism as male or female in which the sex chromosome are found as
   (i) ZW in bird (ii) XY in Drosophila (iii) ZZ in birds. (iv) XO in grasshopper.
8. Mention two differences between Turner.s syndrome and Klinefelter.s syndome.
9. The human male never passes on the gene for haemophilia to his son. Why is it so?
10. Mention four reasons why Drosophila was chosen by Morgan for his experiments in genetics.
11. Differentiate between point mutation and frameshift mutations.

SA-I (3 MARKS)

12. A woman with O blood group marries a man with AB blood group.
    (i) work out all the possible phenotypes and genotypes of the progeny.
    (ii) Discuss the kind of dominance in the parents and the progeny in this case.

9. The gene for haemophilia is present on X chromosome. A male has only one X chromosome which he receives from his mother and Y chromosome from father. The human male passes the X chromosome to his daughters but not to the male progeny (sons).

10. (i) Very short life cycle (2-weeks)

(ii) Can be grown easily in laboratory

(iii) In single mating produce a large no. of flies.

(iv) Male and female show many hereditary variations

(v) It has only 4 pairs of chromosomes which are distinct in size and Shape.

11. **Point Mutations :** Arises due to change in a single base pair of DNA e.g., sickle cell anaemia.

Frame shift mutations : Deletion or insertion/duplication/addition of one or two bases in DNA.

SA-I (3 MARKS)

12. (i) Blood group AB has alleles as I^A, I^B and O group has ii which on cross gives the both blood groups A and B while the genotype of progeny will be IAi and IBi.

(ii) I^A and I^B are equally dominant (co-dominant). In multiple allelism, the gene I exists in 3 allelic forms, I^A, I^B and i.

13. **Cause :** Presence of an extra chromosome in male i.e., XXY.

Symptoms : Development of breast, Female type pubic hair pattern, poor beard growth, under developed testes and tall stature with feminised physique.

14. (i) Green pod colour is dominant

(ii) Green pod colour

(iii) Parents GG(green) X gg (yellow)

Gametes Ⓖ ⓖ

F1 generation Gg (Hybrid green)

Gametes Ⓖ ⓖ X Ⓖ ⓖ

F2 generation GG Gg Gg gg

Phenotypic ratio 3 : 1

Genotypic ratio 1 : 2 : 1

LA (5 MARKS)

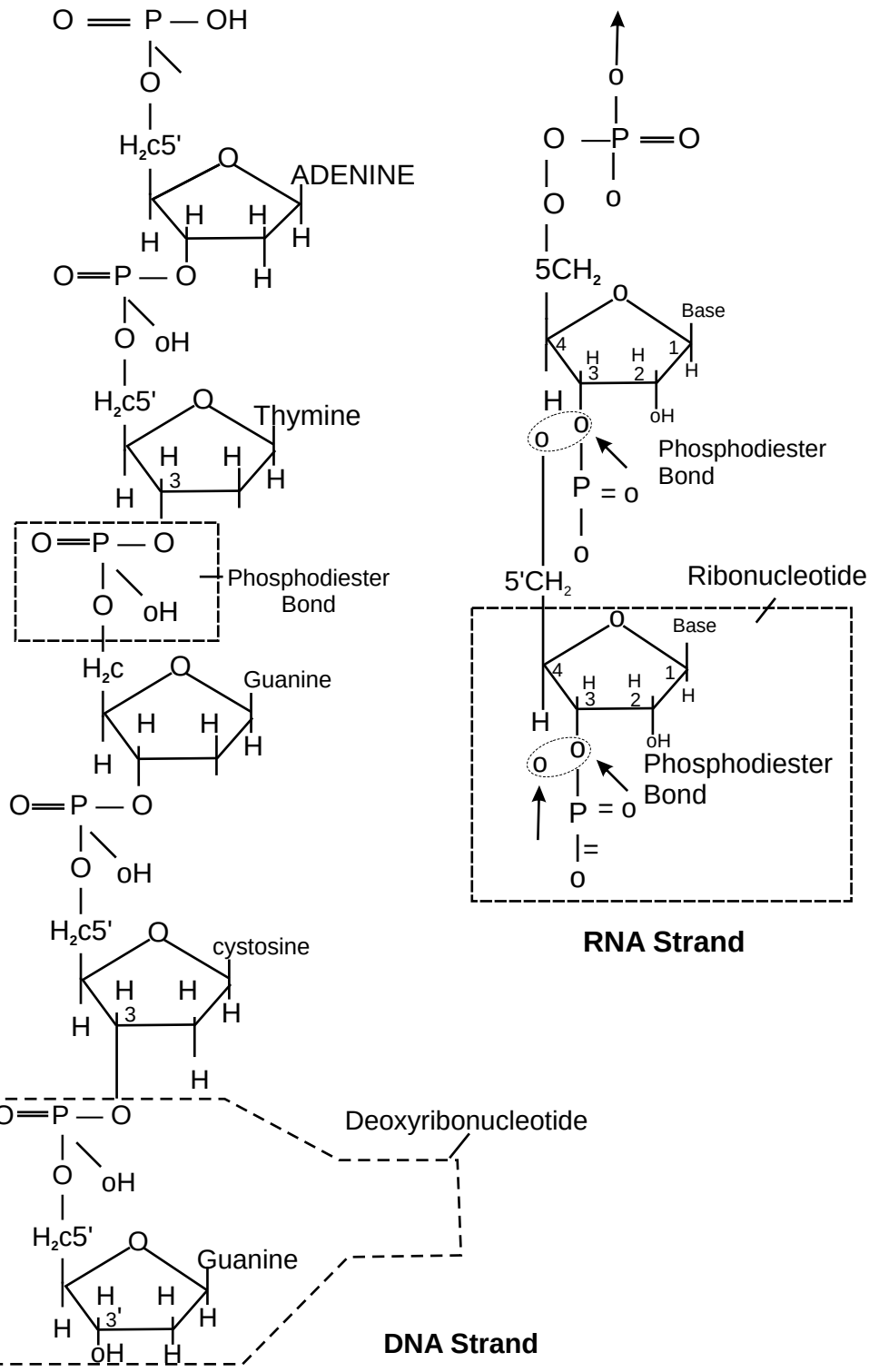
15. (i) It is a dihybrid test cross

(ii) Parent RrYy (Round Yellow) rryy (Wrinkled green)

Gametes (RY), (Ry), (rY), (ry) X ry

	Gametes ry	RY	Ry	rY	ry
F1 progeny		RrYy Round, Yellow	Rryy Round and Green	rrYy Wrinkled Yellow	rryy Wrinkled, Green
Phenotypic ratio	:	1	: 1	: 1	: 1
Genytopic ratio	:	1	: 1	: 1	: 1

(iii) It illustrates the Principle of independent assortment.


ADENINE
Thymine
Guanine
cystosine
Guanine
Phosphodiester Bond
Phosphodiester Bond
Phosphodiester Bond
Base
Base
Ribonucleotide
Deoxyribonucleotide
RNA Strand
DNA Strand