HEREDITY AND VARIATION

IMPORTANT TERMS

1. Mendel and his experiments.

(a) Mendel's experimental material.

Mendel conducted experiments on garden pea (Pisum sativum) for seven years (1856- 1863) before proposing the laws of inheritance.

- He selected garden pea for two reasons: (i) Many varieties were available with observable alternate form for a trait/character. (ii) Peas are generally self-pollinated (self-fertilised) and can be easily cross-pollinated when self-pollination is prevented.
- He selected varieties that differed with respect to seven traits with easily distinguishable contrasting forms, i.e., he selected fourteen true-breeding varities as shown in the table given below;

(b) Mendel's Approach

- Mendel's methodology during the investigation of the inheritance pattern included mathematical logic and statistical analysis of the data.
- His experiments had a large sampling size, that gave credibility to the data he collected.
- The confirmation of the inferences from his experiments on successive generations of his test plants, proved that his results pointed to the general rules of inheritance.
- Mendel first made sure that each of the fourteen varieties (seven pairs of contrasting forms) was true-breeding, by allowing successive generations to self-pollinate and eliminating any offspring that was not true-breeding.
- He studied the inheritance of each of the seven characters inidivdually by conducting monohybrid crosses and then in combination by conducting dihybrid and trihybrid crosses.
- He hybridised /cross-pollinated plants with alternate forms of a trait and used the seeds to generated the first filial (F1) or first hybrid generation.
- He allowed each F1 offspring to self-pollinate to produce the second filial(F2) generation.
- ***He also conducted test crosses.***

(c) Medel's observation.

(i) The F1 hybrids always showed one of the parental forms of the trait.

(ii) Both the parental forms of the trait (contrasting forms of the trait) appeared without any change in the F2 generation.

(iii) The two contrasting forms a trait did not show any blending either in the F1 generation or in the F2 generation.

(iv) The form of the trait that appeared in the F1 hybrids is called dominant form and it appeared in the F2 generation about three times in frequency as its alternate (recessive) form.

(d) Mendel's inferences.

- Mendel made the following inferences from his observations. (i) The characters are controlled by some 'factors' that are stably passed down without any change from are parents to the offspring, through the gametes. (ii) The factors occur in pairs. (iii)In a pair of dissimilar factors of a trail, one of them dominates the other and expresses itself in the hybrid, while the other remains hidden and is recessive.

2. Law of Dominance.

- This law explains the expression of only one of the forms of the parental trait in the F1 hybrid and both the forms in the F2 generation.
- This law states that when two individuals of a species, differing in a pair of contrasting forms of a trait are crossed, the form of the trait that appears in the F1 hybrid is dominant and the alternate form that remains hidden, is called recessive.
- When a cross was made between a true-breeding tall pea plant and a true-breeding dwarf pea plants, all the plants in the F1 generation were tall.

- When the F1 individuals were allowed to self-pollinate and an F2 generation raised, it was found that the tall plants and the dwarf plants were in the ratioof 3:1.
- When the dwarf plants were further self-pollinated, they produced only dwarf plants in successive generations, showing that they are homozygous/ true-breeding.
- When the tall plants were self pollinated, some of them produced only tall plants in the successive generations, while others produced both tall and dwarf plants, showing that they are heterozygous.

3. Law of Segregation.

- This principle states that the members of the allelic pair of a trait that remained together in the parent, segregate/ separate during gamete formation and only one of the factors entersf a gamete.
- As a result, the gametes are pure for a character. (have only one of the alleles)
  A homozygous individual produces only one type of gametes, while a heterozygous individual/hybrid produces two types of gametes in equal frequencies.
- The following example can explain the inheritance pattern.
- In this case, tallness is dominant over dwarfness and it has appeared in the F1 generation.
- The recessive character, dwarfness, has remained hidden in the F1 generation, but appeared again in the F2 generation.
- The two forms of the trait, height of stem, have appeared in the ratio of 3: 1, which is called Mendel'a monohybrid phenotypic ratio.
- It is because the two factors, T and t remained together in the F1 hybrid, but segregated from each other and entered different gametes.
- The paired condition is restored on random fertilisation.
- From the Punnett square, it can be seen that one of the tall plants is homozygous(TT) while the other two are heterozygous (Tt).
- Hence the monohybrid genotypic ratio is 1:2:1(1 true-breeding dominant : 2 hybrid or heterozygous dominant: 1 true-breeding recessive).

4. Law of Independent Assortment.

- This law states that 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.
- This law was proposed by Mendel based on the results of dihybrid crosses, where inheritance of two traits were considered simultaneously.
- The following cross between a pure-breeding plant with yellow, round seeds and another pure-breeding plant with green, wrinkled seeds, can be taken as an example to explain this law.
- In this cross, the factors for the colour of seeds and those for shape of seeds have segregated independently and each gamete has one factor for each of these two traits.

5. Test Cross.

- It is a cross devised by Mendel, where the offspring or individual with dominant phenotype whose genotype is not known, is crossed with an individual homozygous recessive for the trait.
- A monohybrid test cross is as follows:
  The F1 hybrid of a cross between pure tall plant and a pure dwarf plant is crossed with a dwarf plant, i.e., Tt x tt
- The progeny consists of tall and dwarf plants in the ratio of 1: 1.
- The Mendelian monohybrid test cross ratio is 1: 1.
- If the dominant plant had been homozygous, i.e., TT, then the progeny will have all tall plants as given below:
- A dihybrid test cross is shown below;
  An individual heterozygous for two traits is crossed with an individual homozygous recessive for those traits.
- The dihybrid test cross ratio is 1: 1: 1: 1.
- Since this type of cross is used to determine the genotype of an individual, it is called a test cross.

6. Incomplete Dominance.

- The phenotype of the hybrid offspring does not resemble any of the parents, but is intermediate between the two.
- The inheritance of flower colour in dog flower/snapdrogon (Antirrhinum majus) is an example of this phenomenon.

- When a cross was made between a true-breeding red-flowered plant and a true-breeding white-flowered plant, the F_1 hybrid was pink-flowered.
- When the F_1 individual was self-pollinated/self-fertilised, the F_2 generation consisted of red, pink and white-flowered plants as given below:
- The Phenotypic and genotypic ratios are the same, i.e., 1 Red (RR) : 2 Pink (Rr) : 1 White (rr).

7. Multiple Allelism and Codominance.

- In this phenomenon, the offspring shows resemblance to both the parents.
- A well-known example of these phenomena is the inheritance of ABO blood groups in man.
- The gene for blood group exists in three allelic forms, I^A, I^B and i.
- Any individual carries two of these three alleles.
  The allele I^A produces a glycoprotein A, found on the surface/membrane of red blood cells.
  The allele I^B produces a glycoprotein B, found on the suface/membrane of red blood cells.
  The allele i does not produce any glycoprotein.
  The allele I^A is dominant over i and the allele IB is also dominant over i.
- When the alleles I^A and I^B are together, they are equally dominant and both the glycoproteins, A and B are produced.
- The blood group is determined by the presence or absence of one or both the glycoproteins, i.e., group A has glycoprotein A, and B has glycoprotein B, group AB has both the glycoproteins while group O has neither of them.
- The inheritance of blood group character follows Mendelian pattern of inheritance.
- Since there are three alleles, six different genotypes are possible, which produce four different phenotypes, i.e., blood groups A, B, AB and O.

8. Pleiotropy

- In this phenomenon, a single gene product may produce more than one phenotypic effect.
- Even in garden peas, such a phenomenon has been observed in the following characters:
  (i) Starch synthesis/size of starch grains and the shape of seeds are controlled by one gene.
  (ii) Flower colour and seed coat colour are found to be controlled by the same gene.
- Starch is synthesised effectively by the homozygotes, BB and hence the starch grains are large and the seeds at maturity are round.
- The homozygotes, bb are less efficient in starch synthesis; hence they have small starch grains and the seeds are wrinkled.
- The heterozygotes, Bb produce round seeds, indicating that B is the dominant allele; but the starch grains are intermediate in size and hence for the starch grain size, the alleles show incompletes dominance.
- It is an example of pleiotropy as the same gene controls two traits, i.e., seed shape and size of starch grains.
  The genotypes and the phenotypes for the two traits are as follows:
  BB- Round seeds, large starch grains.
  Bb- Round seeds, intermediate sized starch grains.
  bb- Wrinkled seeds, small starch grains.
- Here, it can also be mentioned that dominance is not an autonomous feature of the gene or its product, but it depends on the production of a particular phenotype from the gene product.

9. Polygenic

- The traits studied by Mendel in garden pea have two distinct alternate forms; but there are several characters which are not so distinct in their occurrence.
- Polygenic inheritance is the phenomenon in which a character is controlled by three or more genes (multiple genes) and the graded phenotypes are due to the additive or cumulative effect of all the different genes of the trait.
- Such traits are called polygenic traits, since they are controlled by a number of genes.
- Human skin colour, height and intelligence are some examples of polygenic traits.
- Polygenic inheritance also takes into account the influence of environment; it is also called quantitative inheritance, as the character/phenotype can be quantified, like amount of pigment, intelligence, etc.

- Human skin colour is believed to be controlled by three different genes, A, B, and C, where darkness is dominant over light skin clour.
- An individual with the genotype AABBCC, all dominant alleles, will have the darkest skin colour; and the recessive individual with all recessive alleles (aabbcc), will have the lightest skin colour; an individual with three dominant alleles (like AABbcc, AAbbCc, AaBbCc, etc) have an intermediate skin colour.
- In this manner, the number of each type of alleles in the genotype is responsible for the darkness or lightness of the skin.
- The alleles of the three genes show Mendelian pattern of inheritance.
- In the following, there is mating between individuals with the drakest skin and the lightest skin produce offspring with intermediate skin colour.
- When F_1 individuals are corssed among themselves, the F_2 progeny will be as follows:

10. Rediscovery of Mendel's Laws

- Though Mendel published his work and the laws of inheritance in 1865, they remained unrecognised till1900, for the following reasons:

  (i) His work could not be widely publicised as communication was not easy.

  (ii) His concept of 'factors' as stable and discrete units that controlled the expression of traits, and that of pair of alleles which did not blend with each other, were not accepted by his contemporaries as the explaination for variation.

  (iii) Mendel's apprach of using mathematics to explain the biological phenomena was new and unacceptable to many biologists.

  (iv) Though Mendel's work suggested that factors were discrete units, he could not provide any physical proof for the existence of factors or prove what they are made of .
- In 1900, de Vries, Correns and Tchermark independently rediscovered Mendel's results on the inheritance of characters.
- By then, there had been advancements in microscopy and scientists were able to observe cell division, nucleus, chromosomes, etc.
- By 1902, chromosome movements during cell division had been worked out.

11. Chromosmal Theory of Inheritance

- Walter Sutton and Theodor Boveri independently postulated this theory in 1902.
- They found that the behaviour of chromosomes was parallel to the behaviour of mendelian factors(genes) and used the chromosome movements to explain Mendel's Laws.

  The similarities are as follows:

  (i) Both genes and chromosomes occur in pairs in normal diploid cells.

  (ii) Both of them segregate during gamete formation and only one member of each pair enters a gamete.

  (iii)Members of each pair segregate indepdently of the members of the other pair (s).
- Sutton and Boveri argued that the pairing and separation of the homologous pair of chromsomes would lead to the segregation of a pair of factors they carried.

12. Thomas Hunt Morgan and Genetics.

- T.H. Morgan worked on fruit flies(Drosophila melanogaster).

  They are found suitable for studies in genetics for the following reasons:

  (i) They could be grown on simple synthetic medium in the laboratory.

  (ii) The flies complete their life cycle in about two weeks.

  (iii) They could produce a large number flies in the progeny of a single mating.

  (iv) The male and female flies are distinct and show many types of hereditary variations that could be easily observed.
- Morgen carried out many dihybrid crosses in Drosophila, with the genes that were sex-linked, i.e., the genes are present on the X-chromosome.
- He observed that the two genes under consideration in his expriments did not segregate independently as in the case of characters studied by Mendel.

13. Linkage and Recombination

- Morgan et al observed that when the two genes in a dihybrid cross are located on the same chromosome, the proportion of parental gene combinations in the progeny was much higher than the non-parental or new combinations (also called recombination) of genes.
- They also found that the proportion of recombinants varies, even if the two genes are present on the same chromosome.
- If the linkage is stronger between two genes (tightly linked), the frequency of recombination is low, and vice versa.
- He hybridised yellow-bodies and white-eyed females with brown-bodies and red-eyed males (wild type) (Cross I) and intercrossed their F_1 progeny.
- The F_2 generation contained the following :
- The parental combinations were 98.7% and recombinants were 1.3%.
- In another cross (Cross II) between white-bodied female fly with miniature wing and a male fly with yellow-body and normal -wing, the progeny contained the following:
- The parental combinations were 62.8%, while the recombinants were 37.2%.
- It is evident that the linkage between genes for yellow-body and white-eyes is stronger than that between genes for white-body and miniature wings.
- ***Sturtevant used the frequency of recombination between the gene pairs on the same chromsome as a measure of the distance of the genes and mapped their position on the chromosome***.
- Today genetic/chromosome maps are used in the sequencing of genomes of organisms.

14. Sex-Determination.

- The concept of genetic/chromosomal basis of sex-determination came from the cytoogical observations made in a number of insects.
- ***H.Henking (1891)*** could trace a specific nuclear structure all through spermotogenesis in a few insects.
- He observed that 50% of the sperms received this struture, while the remaining 50% did not receive it.
- Henking named the structure as X body, but could not explain its significance.
- Later it was found to be a chromosome and it was named as X-chromosome.

(a) **XO-type of Sex-determination**

- A large number of insects like grasshopper show XO type of sex determination.
- It is case of male heterogamety, where 50% of the sperms bear an X-chromosome and the other 50% of the sperms do not have the X-chromosome, but only the autosomes; all the ova bear an X-chromosome.
- When an ovum is fertilised by a sperm having X-chromosome, the zygote develops into a fmale.
- When the ovum is fertilised by a sperm having no X-chromosome, the zygote develops into male

(b) **XY-type of Sex determination**

- In insects like Drosophila melanogaster and in human beings, this type of sex-determination is seen.
- The males have an X-chromosome and another small, but characteristically-shaped Y-chromosome, i.e., males have XY chormosomes alongwith other autosomes.
- The females have two X-chromosomes alongwith the other autosomes.
- It is case of male heterogamety, where the males produce two types of sperms, 50% of sperms having one X-chromosome and the other 50% with one Y-chromosome.
- The females are homogametic and produce all ova with one X-chromosome.
- Sex of an individual is decided at the time of fertilisation by the type of sperm fertilising the ovum.

(c) **ZW-type of sex-determination**

- This type of sex determination is seen in ceratain birds.
- The females have ZW chromosomes alongwith the autosomes and the males have ZZ chromosomes.
- It is a case of female heterogamety, where the females produce two types of ova, 50% with one W-chromosome.
- The males are homogametic and produce sperms all, with one Z-chromosome.
- In this case, the sex of the individual is determined by the type of ovum that is fertilised to produce the offspring.

(d) Haplodiploid sex-determination

- This type of sex-detemination is seen in honeybees; it is based on the number of sets of chromosomes, an individual reveives.
- When the ovum is fertilised by a male gamete, the diploid the zygote (2n= 32) develops into a female, i.e., a queenn or worker.
- When the ovum develops by parthenogenesis, i.e, without fertilisation, a male individual, called drone, is formed.
- The male honeybee (drone) is haploid (with n= 16 chromosomes) and forms sperms by mitosis.

The sex-determination is as follows:

The special characteristic features of this system of sex-determination are;

The male honeybees do not have a father, but have a grandfather.

They do not have sons, but can have grandsons.

15. Mutation.

Mutations are of the following types:

- Gene mutations result in alteration in the sequenes of bases of DNA or a change in the base and thereby change in the genotype and phenotype of an organism.
- A mutation that involves a single base change, is called point mutation; a classical example of point mutation is sickle-cell anaemia.
- Deletion or insertion/duplication/addition of one or two bases in th DNA results in a change in the reading frame, thereby resulting in a polypeptide with a different set of amino acids.
- Structural alterations of the chromosomes result due to loss or gain of large segment of DNA, as DNA/ genes are located on the chromosomes.
- When the members of a homologous pair of chromosomes fail to segregate during anaphase I of meiosis, aneuploidy results; there is loss or gain of one or more chromosomes.
- Failure of separation of the duplicated chromosoes into daughter nuclei results in polyplody, a phenomenon in which the cell has three, four or more sets of chromosomes.
- The physical and chemical agents/factors that bring about mutations, are called mutagens.
- ***Mutations are also responsible for variation.***

16. Pedigree Analysis.

- Inheritance pattern of traits in human beings cannot be studied by crosses as in other organisms, for the following reasons: (i) Controlled crosses cannot be perfomed. (ii) The progeny produced is very small (usually one) and taken a long time.
- Pedigree analysis is the important tool to trace the inheritance of a specific trait, abonormality or disease to study human genetics.
- In a pedigree chart, conventionally the following symbols are used:

  - Circles denote females. - Squares denote males.

  - Mating is shown by a horizontal line connecting a male symbol with a female symbol.

  -Offspring symbols are arrange from left to right in the under of birth and connected by a horizontal line below the parents and this line is connected to parent/marriage line by vertical line.

  - A solid/blackened symbol represents the individual with the trait being studied.

  - An open or clear symbol represents the absence of the trait or abnormality under study.
- The offspring are connected to a horizontal line below the parents and the line is connected to the parental line by a vertical line.

17. Genetic Disorders.

- Genetic disorders can be grouped into two categories : (a) Mendelian disorders and, (b) Chromosomal disorders

(a) Medelian disorders.

- These are mainly due to alteration or mutation in a single gene.
- These disorders may be dominant or recessive.
- The disorders are transmitted from one generation to the next following Mendel's principles of heredity; these disorders may be:

(a) autosomal as in cystic fibrosis, sickle-cell anaemia and phenylketonuria or

(b) Sex-linked as in haemophilia, colour blindness and myotonic dystrophy.

- Inheritance of Mendelian disorders can be represented in pedigree charts; the following pedgrees are given as examples for (a) an autosomal dominant (b) an autosomal recessive disorder and (c)a sex-linked.

- Some Mendelian disorders are discussed below:

(i) Haemophilia.

- It is a sex-linked, recessive disorder, whose gene is present on the X-chromosome:
- The defective allele produces a defective protein, which is part of the cascade of proteins involved in the clotting of blood.
- Clotting of blood is abnormally delayed that even a simple/small cut will result in non-stop bleeding in the affected individual.
- More males than females suffer from the disorder as they have only one X-chromosome, and recessive allele on it is expressed.
- The possibility of a female becoming haemophilic is very rare, as she has to receive the defective alleles from both the parents and be homozyogus recessive, i.e., her mother must atleast be a carrier and father haemophilic.
- Heterozygous female is a carrier and passes on the disease to some of her sons; Queen victoria was carrier of this disease and produced haemophilic descendents.

(ii) Colour blindness

- It is a sex-linked, recessive disorder with the mutant gene on the X-chromosome.
- The defect is in the red or green cones of the eye and the victim is unable to discriminate between red and green colours.
- It occurs in 8 per cent of males and only 0.4 per cent females.
- Inheritance pattern is very similar to that of haemophilia, in that more males than females suffers from the disorder.
- A colour-blind female is rare, as her mother has to be atleast a carrier and father colour blind and she must be homozygous for the recessive defective allele.
- A hetrozygous female has normal vision, but is a carrier and passes on the disorder to some of her sons.

(iii) Cystic fibrosis.

- It is caused by a recessive mutant allele on an autosome (chromosome 7).
- The gene produces a unique glycoprotein that leads to the formation of mucus of abonormally high viscosity.
- This type of mucus interferes with the functioning of many exocrine glands like sweat glands, liver, pancreas and lungs.

(iv) Sickle-cell Anaemia.

- It is caused by a mutant recessive allele on chromosome 11.
- The mutant gene causes the substitution of glutamic acid (glu) by valine (val) at the sixth position of the beta globin chain of haemoglobin; this substitution of the amino acid is the result of a single base substitution in the sixth codon of the beta globin gene, from GAG to GUG.
- The normal (Hb-A) and the defective (Hb-S) peptides are as follows:
- The defective haemoglobin undergoes polymerisation under low oxygen tension and changes the shape of RBC from biconcave cells to sickle-shaped elongated cells.
- The disease is controlled by a single pair of alleles, Hb^A and Hb^S.
- Of the three possible genotypes, only individuals homozygous for Hb^s (Hb^s Hb^s) show the disease, though heterozygous individuals (Hb^A Hb^s) are carriers.
- When both the parents are heterozygous/carrier for the gene, the disease appears in some of their children.

(v) Phenylketonuria

- It is caused by a recessive mutant allele on chromosome 12.
- The affected individuals lack an enzyme that catalyses the conversion of the amino acid phenylalanine into tyrosine.
- Consequently phenylalanine is metabolise into phenyl pyruvate and other derivatives.

- Accumulation of these chemincals in the brain results in mental retardation.
- These are also excreted in the urine as they are not absorbed by the kidney.

(vi) Thalassemia

- It is an autosomal, recessively-inerited disorder, transmitted to the offspring, when both the parents are heterozygous, i.e., carrier of the disease.
- The defect arises due to either mutation or deletion, which results in the reduced rate of synthesis of one of the globin chains of haemoglobin.
- The characteristic symptom of the disease, anaemia results due to the abnormal haemoglobin.
- Depending on the globin chain affected, thalassemia is of two types:

(a) Alpha thalassemia and (b) beta thalassemia.

(a) Alpha thalassemia

- Alpha thalassemia is controlled by two closely-linked genes, HBA-I and HBA-2 , on chromosome 16.
- Due to mutation or deletion of one or more of the four alleles, there is reduced rate of synthesis of alpha globin chai of haemoglobin; more the number of alleles affected, less will be the globin synthesis.

(b) Beta thalassemia

- Beta thalassemia is controlled by a single gene HBB on chromosome 11.
- The disease is caused due to mutation of one or both the alleles of the gene.
- There is reduced synthesis of beta globin of haemoglobin.
- Thalassemia is a quantitative problem, where sysnthesis of few globin molecules leads to anaemia, whereas sickle-cell anaemia is a qualitative problem, where synthesis of a defective globin that is non-functional causes the disease.

(b) Chromosomal Disorders.

- These are caused due to absence or excess or abnormal arrangement (structure) of one or more chormosome(s).
- Such a situation leads to serious consequences in the individual.
- The chromosomal abnormalities/disorders in an individual can be found out by karyotyping.
- Some of the chromosomal disorders are discussed below:

(i) Down's Syndrome.

It is caused by the presence of an extra copy of the chromosome 21, i.e., trisomy of 21st chromosome.
This disorder was first reported by Langdon Down (1866).

Symptons:

Short stature and small round head with a flat back.
Partialy open mouth with furrowed tongue.
Broad, flat face with slanting eyes.
Broad palm with characteristic palm crease.
Many loops on finger tips.
Small and arched palate.
Epicanthic eye fold.
Congentital heart diseases.
Physical, psychomotor and mental retardation.

(ii) Klinefelters syndrome

It is caused by the presence of an extra X-chromosome in the male, i.e., XXY the individual has 47 chromosomes.
Though the individual is a male, he shows a number of feminised characters.

Symptons:

Tall stature with feminised physique.
Breast developement (gynacecomastia)
Female type of pubic hair pattern.
Poor beard growth
Sterility.

(iii) Turner's syndrome.

This disorder is caused due to the absence of one of the X-Chromosomes in a female; the karyotype has only 45 chromosomes and they are called as XO females.

Symptons:

Short stature

Rudimentary ovaries.

Poor development of breasts

Lack of secondray sexual characters

Sterility.

VERY SHORT ANSWER TYPE QUESTIONS (1 MARK)

1. A garden pea plant (A) produced inflated yellow pods, and another plant (B) of the same species produced constricted green pods. Identify the dominant traits.

Ans. Inflated pods and green pods.

2. A garden pea plant produced axial white flowers. Another of the same species produced terminal violet flowers. Identify the dominant trains.

Ans. Axial flowers and violet flowers.

3. Name the respective pattern of inheritance where F_1 phenotype

(a) does not resemble either of the two parents and is in between the two.

(b) resembles only one of the two parents.

Ans. (a) Incomplete dominance. (b) Dominance (complete)

4. In a dihybrid cross, when would the proportion of parental gene combinations be much higher than non-parental types, as experimentally shown by Morgan and his group.

Ans. When the two genes show linkage.

5. Write the possible genotypes Mendel got when he crossed F_1 tall pea plant with a dwarf pea plant.

Ans. Tt and tt (in the ratio 1 : 1).

6. A garden pea plant produced round, green seeds. Another of the same species produced wrinkled, yellow seeds. Identify the dominant trains.

Ans. Round seeds and yellow seeds.

7. Why, in test cross, did Mendel cross a tall pea plant with a dwarf pea plant only?

Ans. Dwarfness is a recessive trait which is expressed only in homozygous condition; so he was sure of the genotype of the dwarf plant as it.

8. Name the event during cell division cycle that results in the gain or loss of chromosome.

Ans. Failure of segregation of members of homologous pairs of chromosomes (non-disjuction)

9. Mention the type of allele that expresses itself only in homozygous state in an organism.

Ans. Recessive allele.

10. Name two contrasting pod-related traits studied by Mendel in pea plant experiments.

Ans. Greenpods and yellow pods.

Inflated pods and constricted pods.

11. Mention two contrasting flower-related traits studied by Mendel in pea plant experiments.

Ans. Axial flowers and terminal flowers

Violet flowers and white flowers.

12. Name an autosomal dominant and one autosomal recessive Mendelian disorder in humans.

Ans. Autosomal dominant disorder : Brachydactyly

Autosomal recessive disorder : Sickle-cell anaemia, pheylketonuria, Thalassemia (any one)

13. Write the genotype of :

(i) an individual who is carries of sickle-cell anaemia gene, but apparently unaffected and

(ii) an individual affected with the disease.

Ans. (i) $Hb^A Hb^S$ (ii) $Hb^S Hb^S$

14. A human being suffering from Down's syndrome shows trisomy of 21 st chromosome. Mention the cause of this chromosomal abnormality.

Ans. It is due to non-disjunction (non-separation) of the 21st chromosomes during ova formation and the fertilisation of an ovum with two 21st chromosomes by a normal sperm; there is an additional copy of the 21st chromosome in such individuals.

15. Write the percentage of F_2 homozygous and heterozygous populations in a typical monohybrid cross.

Ans. Homozygous and Heterozygous populations are 50 per cent each.

16. A haemophilic son was born to normal parents. Give the genotypes of the parents.

Ans. Father: XY Mother: XX^h

17. What is heterogamety? Give an example of an organism showing it.

Ans. Heterogamety is the phenomenon in which an individual produces two types of gametes with reference to sex-chromosomes.

e.g., Human males and female fowls are heterogametic.

18. Why do certain genes tend to be inherited together in a cell at the time of cell division?

Ans. They show a phenomenon, called linkage.

19. When a tall pea plant was self-pollinated, one-fourth of the progeny were dwarf. Give the genotype of the parent and the dwarf progenies.

Ans. Parent: Tt

Dwarf progeny : tt

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

Ans. 16 types of gametes.

21. Who had proposed the chromosomal theory of the inheritance?

Ans. Sutton and Boveri.

22. What are the characteristic features of a true breeding line?

Ans. Characteristics of a true-breeding line:

23. In order to obtain the F_1 generation, Mendel pollinated a pure-breeding tall plant with a pure-breeding dwarf plant. But, for getting the F_2 generation, he simply self-pollinated the tall F_1 plants. Why?

Ans. Mendel wanted to understand the inheritance of the selected trait over a number of generations.

Since all F_1 plants are heterozygous, self-pollination was sufficient to study segregation of factors.

24. What is the genetic basis of wrinkled phenotype of pea seed?

Ans. Wrinkled seed-shape is a recessive trait; it is expressed only under the homozygous condition of the alleles.

25. What is the cross between the progeny of F_1 and the homozygous recessive parent called? How is it useful?

Ans. It is called a test cross.

It helps to determine the genotype of the F_1 individual with dominant phenotype.

26. Even if a character shows multiple allelism, an individual will only have been different if the characters he chose were located on the same chromosome? Why?

Ans. A diploid individual has only two sets of homologous chromosomes; hence only two alleles of a gene can be present on the homologous pair of chromosomes.

27. Do you think Mendel's laws of inheritance would have been different if the characters he chose were located on the same chromosome? Why?

Ans. Mendel could not have framed the law of independent assortment, if the genes for all the characters Mendel chose, were present on the same chromosome and showed the phenomenon of linkage.

28. A cross between two tall plants resulted in offspring, having few dwarf plants. What would be the genotypes of both the parents?

Ans. Both the parents must be heterozygous for tallness, i.e., Tt.

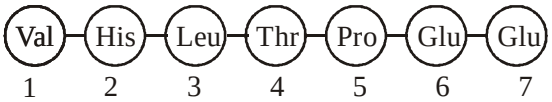
29. Two genes 'A' and 'B' are linked. In a dihybrid cross involving these two genes, the F_1 heterozygote is crossed with homozygous recessive parental type (aabb). What would be the ratio of offspring in the nest generation?

Ans. The ratio of offspring would be:

AaBb	:	Aabb	:	aaBb	:	aabb
1	:	1	:	1	:	1

SHORT ANSWER TYPE QUESTIONS (2 MARKS)

1. A relevant portion of β-chain of haemoglobin of a normal human is given below.



The codon for the sixth amino acid is GAG. The sixth codon GAG mutates to GAA as a result of mutation 'A' and to GUG as a result of mutation 'B'. Haemoglobin structure did not change as a result of mutation 'A', whereas haemoglobin structure changed because of mutation 'B', leading to sickle-shaped RBCs. Explain giving reasons how could mutation 'B' change the haemoglobin structure or bring down mutation and not mutation 'A'.

Ans. In mutation A, the change in the codon GAG to GAA does not change the amino acid glutamic acid; hence there is no change in the haemoglobin structure.

In mutation B, the codon GAG is changed to GUG, where GUG codes for valine while the original codon GAG codes for glutamic acid; hence there is a change in the haemoglobin structure and it leads to sickle-cell anaemia.

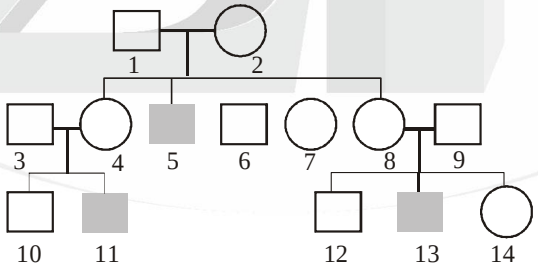
2. In a dihybrid cross, white-eyed, yellow-bodies female Drosophila crossed with red-eyed, brown-bodied male Drosophila produced in F_2 generation, 1.3 percent recombinants and 98.7 per cent progeny with parental type combinations. This observation of Morgan deviated from Mendelian F_2 phenotypic dihybrid ratio. Explain giving reasons Morgan's observations.

Ans. It is due to the phenomenon of linkage.

Linkage is defined as the phenomenon in which two or more linked genes are inherited together and their frequency of recombination in a test cross progeny is less than expected 50 per cent.

3. Haemophilia is a sex-linked inheritance condition in humans, where a simple cut causes non-stop bleeding. Study the pedigree. chart showing the inheritance of haemophilia in a family.

Answer the questions that follow:



Ans. (i) It appears in more frequencies among males than females.

It is transmitted from an unaffected carrier female to some of the male progeny.

The possibility of a female becoming haemophilic is rare as her mother has to be atleast a carrier and father haemophilic.

(ii) Since the male receives his X-chromosome from the mother, it proves that the gene is present on the X-chromosome.

So a carrier female transmits it to her son.

Since a male has only one X-chromosome, the disease appears in more males.

A female has two X-chromosomes (XX) and to be haemophilic, her father must be a haemophilic and the mother must be atleast a carrier, i.e., she must be homozygous recessive (X^hX^h).

4. Tallness of pea plant is a dominant trait, while dwarfness is the alternate recessive trait. When a pureline tall is crossed with a pureline dwarf, what fraction of tall plants in F_2 shall be heterozygous? Give reasons.

Ans. Parents : Pure tall (TT) × Pure dwarf (tt)

Gametes : T, t

F_1 generation : Tt – Tall

Selfing : Tt × Tt

Gametes : T t, T t

F_2 generation : T, t

	T	t
T	TT Tall	Tt Tall
t	Tt Tal	tt Dwarf

Phenotypic ratio : 3 Tall : 1 Dwarf Genotypic ratio : 1 TT : 2 Tt : 1 tt

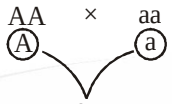
Two-thirds of the tall progeny is heterozygous.

It is because the gene for tallness (T) is dominant and expresses itself in the heterozygous condition, Tt.

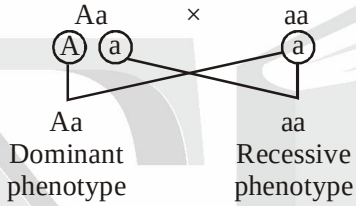
5. How does a test-cross help in identifying the genotype of the organism? Explain.

Ans. In the test corss, an individual showing dominant phenotype would be crossed to an individual with recessive phenotype for the same trait.

(i) If the individual is homozygous dominant, all the individuals in the progeny would show the dominant phenotype.



(ii) If the individual is heterozygous, the progeny will show dominant phenotype and recessive phenotype in the ratio of 1 : 1.



Thus the genotype of the individual with dominant phenotype can be ascertained.

6. When a tall pea-plant was selfed, it produced one-fourth of its progeny as dwarf. Explain with the help of a cross.

Ans. Since dwarfness (recessive trait) has appeared in the progeny, the tall pea plant must be heterozygous for tallness, i.e., its genotype is Tt

Selfing : Tall Tt × Tall Tt

Gametes : T t, T t

Progeny : T, t

	T	t
T	TT Tall	Tt Tall
t	Tt Tall	tt Dwarf

The phenotypic ratio is 3 Tall : 1 dwarf.

Since tallness is dominant over dwarfness, the homozygotes (TT) as well as the heterozygous (Tt) are tall.

The recessive trait is expressed only under homozygous condition (tt) and hence appears only in one-fourth of the propeny.

7. Why are F_2 phenotypic and genotypic ratios same in a cross between red-flowered snapdragon an white-flowered snapdragon plants? Explain with the help of a cross.

Ans. In snapdragon, the inheritance of flower colour shows incomplete dominance.

Neither of the alleles of the gene for flower colour is completely dominant over the other and the hybrid shows an intermediate phenotype.

Parents : Red-flowered (RR) × White-flowered (rr)

Gametes : R, r

F_1 generation : Rr (Pink-flowered)

Selfing : Rr × Rr

Gametes : R r, R r

F_2 generation : R × r

	T	t
T	RR Red-flowered	Rr Pink-flowered
t	Rr Pink-flowered	rr White-flowered

Phenotypic ratio:

Red-flowered	:	Pink-flowered	:	White-flowered
1		2		1

Genotypic ratio

RR 1 : Rr 2 : rr 1.

8. Look at the above diagram and answer the following questions:

(a) Write the genotypes of A, B, C, D (b) Write the phenotypes of A, B, C, D

(c) Write the phenotypic ratio of progeny (d) Write the genotypic ratio of progeny

Ans. (a) A-Tt, B-TT, C-tt, D-Tt (b) A-Tall, B-Tall, C-Dwarf, D-Tall (c) 3 Tall : 1 Dwarf (d) 1 TT : 2 Tt : 1 tt

9. A man with blood group A married a woman with B group. They have a son with AB blood group and a daughter with group O. Work out the cross and show the possibility of such inheritance.

Ans.

Man — Blood group A × Woman — Blood group B

Son — Blood group AB; Daughter — Blood group O

Since blood group O, whose genotype is ii, has appeared in the progeny, the parents are heterozygous, i.e., father has genotype I^A i (blood group A) and mother has genotype I^B i blood group B.

Parents : Man (A group) × Woman (B group)

Gametes : I^A, i × I^B, i

Progeny : I^B, i

	I^A	i
I^A	$I^A I^B$ Blood group AB	I^A i Blood group A
i	I^B i Blood group b	ii Blood group O

The progeny can have all the four phenotypes or blood groups, i.e., A, B, AB and O.

10. The male fruit-fly and female fowl are heterogametic, while the female fruit fly and male fowl are homogametic. Why are they called so?

Ans. The male fruit fly (sex-chromosomes, ZW) are heterogametic, because they produce two types of gametes, with reference to sex chromosomes; the male fruit fly produces 50% of sperms with one X-chromosome and 50% of them with one Y-chromosome alomg with the autosomes, while the female fowl produces 50% ova with one Z-chromosome and the other 50% with one W-chromosome, along with the autosomes.

The female fruit fly and male fowl are homogametic, as they produce only one type of gametes with reference to sex-chromosomes; the female fruit fly produces all ova with one X-chromosome and the male fowl produces all the sperms with one Z-chromosome, along with autosomes.

11. A plant of Antirrhinum majus with red flowers was crossed with another plant of the same species with white flowers. The plants of F_1 generation bore pink flowers. Explain the pattern of inheritance with the help of a cross.

Ans. Inheritance of flower colour in Antirrhinum majus:

Parents : Red flowers RR × White flowers rr

Gametes : R × r

F^1 generation : Rr-Pink flowers

Selfing : Rr × Rr

Gametes : R, r × R, r

F^2 generation : R, r

	R	r
R	RR Red flowers	Rr Pink flowers
r	Rr Pink flowers	rr White flowers

This character shows incomplete dominance, where neither of the two alleles of the gene is completely dominant and the F_1 hybrid is intermediate between the two.

In the F_2 generation, the parental characters appear without any change, indicating that it is not a case of blending

inheritance.

The phenotypic and genotypic ratios of the F_2 are the same, i.e.,

Red flowers	:	Pink flowers	:	White flowers
RR	:	Rr	:	rr
1	:	2	:	1

12. A woman with blood group O married a man with AB group. Show the possible blood groups of the progeny. List the alleles involved in this inheritance.

Ans. Parents : Father (AB group, I^AI^B) × Mother (O group, ii)

Gametes : I^A, I^B and i

Progeny : I^A i (A group) and I^B i (A group)

The gene for blood group exists in three allelic forms I^A, I^B and i.

13. In a particular plant species majority for the plants bear purple flowers. Very few plants bear white flowers. Very few plants bear white flowers. Very few plants bear white flowers. No intermediate colours are observed. If you are given a plant with purple flowers, how would you confirm that it is a pure-breed for that trait? Explain.

Ans. The purple flowers are dominant over white flowers and hence occur in more frequency.

A test cross must be conducted, i.e., the purple-flowered variety must be crossed to a white-flowered (recessive) variety.

Cross I

Purple-flowered (PP) × White-flowered (pp)

Gametes : P and p

Progeny : Pp-all purple-flowered

Cross II

Purple-flowered (Pp) × White-flowered (pp)

Gametes : P, p and p, p

Progeny : Pp and pp

Purple-flowered : White-flowered

1 : 1

If the progeny consists of all purple-flowered plants, the given plant is homozygous (dominant).

If the progeny consists of purple-flowered and white flowered plants in the ration of 1 : 1, the given plant is heterozygous purple-flowered.

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

Ans. Mendel selected garden pea for two reasons:

(i) Many varieties were available with observable alternate forms for a trait/character.

(ii) Peas are generally self-pollinated (self-fertilised) and can be easily cross-pollinated when self-pollination is prevented.

15. Differentiate between dominance and Recessive.

Ans.

Dominance	Recessive
Dominance refers to the phenomenon exhibited by the form of a trait, which appears in the heterozygous (hybrid) condition.	Recessive refers to the phenomenon exhibited by the form of a trait, which remains suppressed or hidden in the heterozygous (hybrid) condition.

16. 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 pea for a single locus.

Ans. **Cross I** (Where the homozygous female is recessive)

Homozygous female — Black-eyed — bb × Heterozygous male — Brown-eyed — Bb

Gametes : b

Progeny :

	b
B	Bb Brown-eyed
b	bb Brown-eyed

Brown-eyed : Blue-eyed

1 : 1

Cross II (Where the homozygous female is dominant)

Homozygous female — Black-eyed — BB × Heterozygous male — Brown-eyed — Bb

Gametes :

Progeny : b

	b
B	BB Brown-eyed
b	bb Brown-eyed

All the individuals are brown-eyed.

17. Differentiate between Homozygous and Heterozygous.

Ans.

Homozygous	Heterozygous
– It is the condition in which the members of a pair of alleles for a character are similar.	– It is the condition in which the members of a pair of alleles for a character are different.
– Individual with such a condition is pure breeding.	– Individual with such a condition is not pure breeding.

18. When a cross is made between tall plant with yellow seeds (TtYy0 and tall plant with green seeds (Ttyy), What proportion of phenotype in the offspring could be expected to be: (a) tall and green (b) dwarf and green.

Ans. Progeny : Tall, yellow — Tt Yy × Tall, green — Tt yy

Gametes : (TY) (Ty) (tY) (ty) × (Ty) (ty)

Progeny :

	TY	Ty	tY	ty
Ty	TT Yy Tall, yellow	TT yy Tall, yellow	TT Yy Tall, yellow	Tt yy Tall, green
ty	Tt Yy Tall, yellow	Tt yy Tall, green	tt Yy Dwarf, yellow	tt yy Dwarf, green

(a) Tall and green 3/8 (b) Dwarf and green 1/8

19. Two heterozygous parents are crossed. If the two loci are linked, what would be the distribution of phenotypic features in F1 generation for a dihybrid cross?

Ans. If the loci are completely linked, all the offspring will be of the parental types only.

If the loci are incompletely linked, the parental and recombinants would appear in the progeny, but the recombinants will be in a small proportion.

20. Differentiate between monohybrid and dihybrid

Ans.

Monohybrid	Dihybrid
It is the individual that is heterozygous for the alleles (of a gene) controlling one character/trait.	It is the individual that is heterozygous for the alleles (of two genes) controlling two characters/traits.

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

Ans. Morgan's contribution to Genetics:

(i) Morgan conducted experiments on Drosophila and discovered sex-linkage.

(ii) He also found that linked genes may show a phenomenon called linkage, where the recombinants in a test cross progeny are less than 50%.

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

Ans. Pedigree analysis:

It is an analysis of the distribution and movement of traits in a series of generations of a family.

It is an important method to study human genetics.

Genetic disorders occurring in a family can be identified and the movement of the trait in the future generations can be predicated.

23. What is point mutation? Give one example.

Ans. Point mutation refers to the mutation arising due to a change in a single base pair of DNA. e.g., Sickle cell anaemia arises due to a change of amino acid glutamine to valine in the sixth position of β-chain of haemoglobin.

24. How are alleles of a particular gene different from each other Explain its significance.

Ans. In a diploid organism, every gene exists as a pair of alleles.

They may be different from each other, dur to the changes one of them has undergone; the changes have modified the information this allele contains.

The modified allele is normally the recessive allele, while the unmodified allele is the dominant one.

The modified allele may produuce:

(i) the normal or less efficient enzyme or (ii) a non-functional enzyme or (iii) no enzyme at all.

According it may produce the same phonotype as the unmodified allele or the phenotype may be affected.

25. Can a child have blood group O, if his parents have blood groups 'A' and 'B' Explain.

Ans.

Man
Blood group A
×
Woman
Blood group B

Son
Blood group AB

Daughter
Blood group O

Since blood group O, whose genotype is ii, has appeared in the progeny, the parents are heterozygous, i.e., father has genotype I^A i (blood group A) and mother has genotype I^B i blood group B.

Parents : Man (A group) × Woman (B group)

I^A i I^B i

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

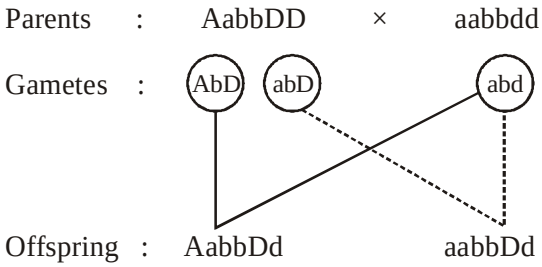
Progeny : I^B i

I^A i	$I^A I^B$ Blood group AB	I^A i Blood group A
i	I^B i Blood group b	ii Blood group O

The progeny can have all the four phenotypes or blood groups, i.e., A, B, AB and O.

26. A, B, D are three independently assorting genes with their recessive alleles a, b, d respectively. A cross was made between individual of AabbDD genotype with aabbdd. Find out the type of genotypes of the offspring produced.

Ans.



The genotypes of the offspring are AabbDd and aabbDd.

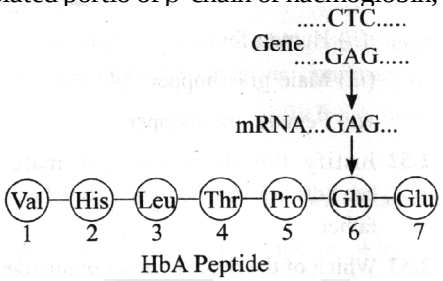
27. If a father and son are both defective in red-green colour vision, is likely that the son inherited the trait from his father? Comment.

Ans. No, the son never inherits colour blindness from his father.

The gene for colour-blindness is present on the X-chromosome, but a son receives Y-chromosome from his father. Since a male receives the X-chromosome from his mother, he inherits the disease from his mother only.

SHORT ANSWER TYPE QUESTIONS (3 MARKS)

1. Given below is the representation of amino acid composition of the relevant translated portio of β-chain of haemoglobin, related to the shape of human red blood cells.



a. Is this representation indicating a normal human or a sufferer from certain related genetic disease? Give reason in support of your answer.

b. What difference would be noticed in the phenotype of the normal and the sufferer related to this gene?

c. Who are likely to suffer more from the defect related to the gene represented- the males, the females or both males and females equally? And why?

Ans. a. The representation indicates a normal human; HbA is a normal peptide with glutamic acid at the sixth position of Beta globin chain.

b. The normal individual has biconave, disc-like RBCs, whereas, the victim has elongated-sickle-shaped RBCs.

c. Both males and females suffer equally, because it is an autosomal recessive disorder.

2. **a.** Explain the phenomena of multiple allelism and co-dominance, taking ABO blood group as an example.

b. What is the phenotype of the following : **a.** I^Ai **b.** ii

OR

Inheritance pattern of ABO blood groups in humans shows dominance, co-dominance and multiple allelism. Explain each concept with the help of blood group genotypes.

Ans. a. Multiple allelism

It is the phenomenon in which a gene exists in more than two allelic forms.

The gene for blood group character exists in three allelic forms, I^A, I^B and i, where i is recessive to both I^A and I^B.

Since an individual possesses only two alleles, multiple allelism can be studied only in a population.

b. Codominance

It is the phenomenon in which two alleles of a gene are equally dominant and express themselves in the presence of the other.

The blood group alleles I^A and I^B are codominant and the genotype $I^A I^B$ produces the blood group AB; the offspring resembles both the parents.

c. I^A i – Blood group A ; I^B i – Blood group B

3. Name the phenomenon that leads to situations like 'XO' abnormality in humans. How do humans with 'XO' abnormality suffer? Explain.

Ans. Non-disjunction is responsible for chromosomal abnormalities.

Non-disjunction is the phenomenon of failure of segregation of the members of homologous pairs of chromosomes.

XO-abnormality results from fertilisation of an ovum with X-chromosome by a sperm carrying no sex-chromosome.

The individual has underdeveloped feminie characters.

Ovaries are rudimentary.

She is sterile.

4. Explain how trisomy of 21st chromosome occurs in humans. List any four characteristic features in an individual suffering from it.

Ans. Trisomy of 21st chromosome occurs due to non-disjunction, i.e., non-separation of the homologous pair of 21st chromosomes at anaphase 1 of meiosis during ova formation and ferlilisation of such an ovum with two 21st chromosomes with a normal sperm.

Symptoms of Down's Syndrome:

(i) Partially open mouth with furrowed tongue. (ii) Broad, flat face with slanting eyes.

(iii) Broad palm with characteristic palm crease. (iv) Small and arched palate.

(v) Epicanthic eye fold. (vi) Congenital heart diseases. (vii) Mental retardation.

5. How are dominance, co-dominance and incomplete dominance patterns of inheritance different from each other?

Ans. Dominance

It is the phenomenon in which one allele of a gene expresses itself and suppresses the expression of the other (recessive) allele of the same gene, when they are present together in a hybrid.

The hybrid resembles one of the parents.

Codominance

It is the phenomenon in which two alleles of a gene are equally dominant and express themselves in the presence of the other.

The hybrid shows characters of both the parents.

Incomplete dominance

It is the phenomenon in which neither of the two alleles of a gene is completely dominant over the other.

The hybrid is intermediate between the two parents.

6. **a.** Sickle-celled anaemia in humans is a result of point mutation. Explain.

b. Write the genotypes of both the parents who have produced a sickled-celled anaemic offspring.

Ans. a. Mutation arising due to a change in a single base pair of DNA, is called point mutation.

The defect is caused by the single base substitution at the sixth codon of the beta chain of haemoglobin from GAG to GUG; this leads to substitution of glutamic acid by valine.

The mutant or defective haemoglobin molecule undergoes polymerisation under low oxygen tension causing sickle-shaped RBCs.

b. The parents must be HB^A Hb^S and Hb^A Hb^S.

7. A pea plant with purple flowers was crossed with white flowers, producing 50 plants with only purple flowers. On selfing, these plants produced 482 plants with purple flowers and 162 with white flowers. What genetic mechanism accounts for these results? Explain.

Ans. Since all the plants in the F_1 generation have purple flowers, purple flower colour is dominant.

These plants are heterozygous, produced from pure breeding/homozygous purple-flowered plants (PP) and homozygous white-flowered (pp) pea plants.

Parents : Purple-flowered PP × White-flowered pp

Gametes : (P) (P)

F_1 generation : Pp (purple-flowered)

Selfing of F_1 generation : Purple-flowered Pp × Purple-flowered Pp

Gametes : (P) (P) (P) (P)

F_2 generation :

	P	p
P	PP Purple-flowered	Pp Purple-flowered
p	Pp Purple-flowered	PP White-flowered

Purple-floweredL White-flowered are in the ratio of 3(482) : 1(162).

It is due to segregation of the factors that remained together in the hybrid.

The factors segregate from each other during gamete formation and a gemete receives only one of the two factors; a homozygous parent produces all gametes that are similar, while a heterozygous parent produces two kinds of gametes, each having one allele with equal proportion.

8. Name a disorder, give the karyotype and write the symptoms a human suffers from, as a result of monosomy of sex-chromosomes.

Ans. Turner's syndrome

The individual has 22 pairs of autosomes and only one X-chromosome, i.e., 45 chromosomes.

Symptoms:

(i) The individual is a female.

(ii) Ovaries are rudimentary.

(iii) Female secondary characters are not properly developed.

(iv) The individual is sterile.

9. Name a disorder, give the karyotype and write the symptoms which a human male suffers, as a result of an additional X-chromosome.

OR

Name the genetic disorder caused by an extra 'X' chromosome in a human male. State the diagnostic features of the individual suffering from it.

Ans. Klinefelter's syndrome

The individual has 22 pairs of autosomes and XXY sex chromosomes, i.e., 47 chromosomes.

Symptoms:

The individual is a male.

He shows development of feminine characters like development of breasts.

Body hair is sparse.

The individual is sterile.

10. Name the genetic disorder caused by trisomy of 21st chromosome in humans. Write the diagnostic features of the disorder.

Ans. Non-disjunction is responsible for chromosomal abnormalities.

Non-disjunction is the phenomenon of failure of segregation of the members of homologous pairs of chromosomes.

XO-abnormality results from fertilisation of an ovum with X-chromosome by a sperm carrying no sex-chromosome.

The individual has underdeveloped feminie characters.

Ovaries are rudimentary.

She is sterile.

11. In pea plants, let symbol 'Y' represent dominant yellow symbol 'y' the recessive green symbol 'R' the round seed shape and symbol 'r' the wrinkled seed shape. A typical Mendelian dihybrid cross was carried out in pea plants. Write the genotypes of :

a. Homozygous dominant and recessive plants. **b.** Gametes produced by both the parents.

c. F_1 offspring. **d.** Gametes produced by F_1 offspring.

Ans. a. RRYY and rryy b. RY and ry c. RrYy d. RY, Ry, rY and ry

12. During his studies on genes in Drosophila that were sex-linked, T.H. Morgan found F_2 population phenotypic ratios deviated from the expected 9 : 3 : 3 : 1. Explain the conclusion, he arrived at.

Ans. Morgan's conclusion:

The genes were located on the X-chromosome.

They found that when the two genes in a dihybrid cross were situated on the same chromosome, the proportion of parental gene combinations were much higher than the non-parental gene combinations, called recombinants.

Morgan attributed this to linkage, i.e., physical association between the genes on a chromosome.

He also found that some genes were tightly linked and showed very low recombination, while some genes were lossely linked and showed higher recombination frequency.

13. a. Why are grasshopper and Drosophila said to show male heterogamety? Explain.

b. Explain female heterogamety with the help of an example.

Ans. a. Male heterogamety

A male grasshopper produces two types of gametes with reference to sex chromosomes, i.e., 50% of them with one X-chromosome and 50% of them with no X-chromosome.

A male Drosophila produces 50% of gametes with one Y-chromosome.

Since they produce two types of gametes with reference to sex chromosome, they are said to show male heterogamety.

b. Female heterogamety

It is the phenomenon in which females of a species produce two types of gametes with reference to sex-chromosomes.

It is seen in fowls, where a female has ZW sex chromosomes and produces 50% of ova with one Z-chromosome and 50% of them with one W-chromosome.

The sex of the offspring is determined by the type of ovum fertilised.

14. Explain the sex-determination mechanism in humans. How is it different in birds?

Ans. In humans, XY type of sex determinantion is seen and both males and females have same number of chromosomes.

The males have 22 pairs of autosomes plus XY-chromosomes.

The females have 22 pairs of autosomes plus a pair of X-chromosomes.

A male produces two types of sperms, 50% of them carrying one X-chromosome and the other 50% of them carrying one Y-chromosome, apart from 22 autosomes in each of them.

A female produces ova, all containing one X-chromosome plus 22 autosomes.

The sex of the individual is determined by the type of sperm fertilising the ovum.

It the ovum is fertilised by X-carrying sperm, the zygote (XX) develops into a female.

If the ovum is fertilised by Y-carrying sperm, the zygote (XY) develops into a male.

In birds, sex determination is of ZW type.

The males are homogametic (ZZ) and females are heterogametic (ZW) and the type of ovum fertilised determines the sex of the individual.

15. Explain the mechanism of sex-determination in insects like Drosophila and grasshopper.

Ans. Sex determination in Drosophila

In Drosophila, sex determination is of XY type, and both males and females have the same number of chromosomes, i.e., 4 pairs.

The males have three pairs of autosomes and XY-chromosomes.

The males have three pairs of autosomes and XX-chromosomes.

The males are heterogametic and produce two types of sperms, 50% of them carrying one X-chromosome and the other 50% carry one Y-chromosome.

The females are homogametic and all ova contain one X-chromosome.

The sex of the insect is determined by the type of sperm fertilising the ovum; the ovum fertilised by the X-carrying sperm develops into a female, while that fertilised by the Y-carrying sperm develops into a male.

Sex determination in Grasshopper

Sex determination in grasshopper is of XO type.

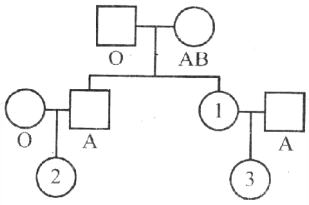
The males have one X-chromosomes other than the autosomes.

The females have two X-chromosomes (XX) other than the autosomes.

Males produce two types of sperms, 50% of them with one X-chromosome and 50% of them with no X-chromosome.

Ova fertilised by a sperm carrying X-chromosome develop into females, while those fertilised by a sperm, carrying no X-chromosome develop into males.

16. Work out a cross between true-breeding red and white-flowered dog-flower plants (snapdragon) upto F_2 progeny. Explain the results of F_1 and F_2 generations.



Ans. Parents : Red-flowered (RR) × White-flowered (rr)

Gametes : R r

F_1 generation : Rr (Pink-flowered)

Self-pollination : Rr × Rr

Gametes : R r R r

F_2 generation : R r

	R	r
R	RR Red-flowered	Rr Pink-flowered
r	Rr Pink-flowered	rr White-flowered

The F_1 individuals are pink-flowered which is a case of incomplete dominance, it is a phenomenon in which neither of the two alleles of a gene is completely dominant over the other and the F_1 hybrid is an intermediate between the two parents.

The alleles of the hybrid segregate during gamete formation and the parental characters reappear without any change; hence the phenotypic and genotypic ratios of the F_2 are the same, i.e.,

Red-flowered		Pink-flowered		White-flowered
RR	:	Rr	:	rr
1		2		1

17. Study the following pedigree chart of a family, starting with mother with AB blood group and father with O blood group.

a. Mention the blood group as well as the genotype of the offspring numbered 1 in generation II.

b. Write the possible blood groups as well as their genotypes of the offspring numbered 2 and 3 in generation III.

Ans. a. Offspring 1 may have blood group A (I^Ai) or blood group B(I^Bi)

b. Offspring 2 may have blood group A (I^Ai) or blood group I (ii)

Offspring 3 may have:

If offspring 1 is os blood group A

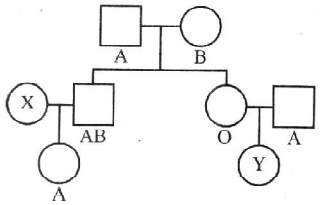
* blood group A (may be I^AI^A or I^A i) * blood group O(ii), if father is heterozygous for A

If offspring 1 is of blood group B

* blood group AB (I^AI^B) * blood group A (I^Ai) * blood group B (I^Bi), is father is heterozygous for A

* blood group O (ii), is father is heterozygous for A

18. Study the given pedigree chart showing the pattern of blood group inheritance in a family :



a. Give the genotype of the following : **i.** Parents **ii.** The individual 'X' in second generation

b. State the possible blood groups of the individual 'Y' i the third generation.

c. How does the inheritance of this blood group explain co-dominance?

Ans. a. (i) Father I^Ai and mother I^Bi

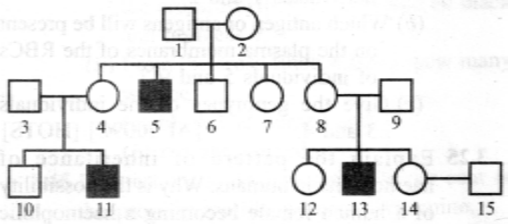
(ii) I^AI^A or I^Ai or ii

b. Y can be of blood group A or O

c. The gene for blood group trait has three alleles, I^A, I^B and i

I^A and I^B are not only dominant over i, but are also codominant, i.e., both of them express themselves in the presence of the other, in producing the phenotype, blood group AB.

19. Haemophilia is a sex-linked recessive disorder of humans. The pedigree chart given below shows the inheritance of haemophilia in one family. Study the pattern of inheritance and answer the questions given.



a. Give all the possible genotypes of the members 4, 5 and 6 in the pedigree chart.

b. A blood test shows that the individual 14 is a carrier of haemophilia. The member numbered 15 has recently married the member numbered 14. What is the probability that their first child will be a haemophilic male?

Ans. a. 4–XX^h; 5–X^hY; 6–XY

b. No. 14 is a carrier, whose genotype is XX^h.

Cross : XX^h × XY

Gametes : (X) (X^h) (X) (Y)

Progeny : X Y

	X	Y
X	XX Normal Female	XY Normal Male
X^h	XX^h Carrier Female	X^hY Haemophilic Male

The probability of the first child being a haemophilic male is 25%.

20. Recently a girl baby has been reported to suffer from haemophilia. How is it possible? Explain with the help of a cross.

OR

How is it that human females are rarely haemophilic?

Ans. The gene for haemophilia is present on the X chromosome.

The disorder is due to a recessive mutant allele; hence a female with XX sex chromosomes, must be homozygous to produce the disease.

She must receive one of the defective alleles from her haemophilic father and the other X-chromosome with is also haemophilic or at least a carrier (heterozygous for the trait, XX^h).

The cross is as follows.

Parents : Father (haemophilic) X^hY × Mother (Carrier) XX^h

Gametes : X^h Y × Y X^h

Progeny : X × Y

X^h	XX^h Carrier Female	X^hX^h Haemophilic Female
Y	XY Normal Male	X^hY Haemophilic Male

21. In one family each of the four children has a different blood group. Their mother is group A and the father is group B. Explain this pattern of inheritance with the help of a cross along with the genotypes.

Ans. Since all the blood groups (including the recessive O group) have appeared in the progeny, father must be heterozygous for B group and mother must also be heterozygous for A group.

Parents : Father B group I^Bi × Mother A group I^Ai

Gametes : I^B i × I^A i

Progeny : I^A × i

X^h	I^AI^B AB-group	I^Bi B-group
Y	I^Ai A-group	ii O-group

ABO blood group is under the control of a single gene, which exists in three allelic forms, I^A, I^B and i, i.e., it shows multiple allelism.

I^A is dominant over i and I^B is also dominant over i.

I^A and I^B are equally dominant and exhibit codominance and produce blood group AB.

Six genotypes are possible with the three alleles and they produce four phenotypes, i.e., blood groups A, B, AB and O.

22. Who proposed chromosomal theory of inheritance? Point out any two similarities in the behaviour of chromosomes and genes.

Ans. Chromosomal theory of inheritance was proposed by Sutton and Boveri.

Similarities between genes and chromosomes:

a. Both genes and chromosomes occur in pairs in normal diploid cells.

b. Both of them segregate during gamete formation and enter different gametes; i.e., one member into one gamete and the other member into another gamete.

c. Members of each pair segregate independently of the members of the other pair(s).

23. When tall pea plants were selfed, some of the offspring were dwarf. Explain with the help of a Punnet square.

Ans. Since dwarf individuals are produced in the progeny, the parent plant must be heterozygous for tallness, i.e., its genotype is Tt.

Selfing : Tall (Tt) × Tall (Tt)

Gametes : (T) (t) (T) (t)

Progeny : T t

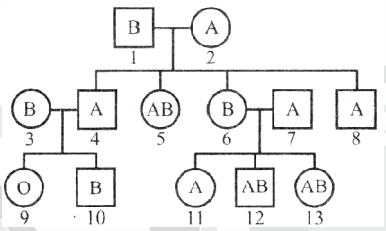
	T	t
T	TT Tall	Tt Tall
t	Tt Tall	tt Dwarf

The phenotypic ratio is 3 tall : 1 dwarf.

The genotypic ratio is 1 TT : 2Tt : 1tt

25% of the offspring are dwarf.

24. Study the pedigree chart given above, showing the inheritance pattern of blood groups in a family and answer the following questions :



a. Give the possible genotypes of the individuals 1 and 2.

b. Which antigen or antigens will be present on the plasma membranes of the RBC's on individuals 5 and 9 ?

c. Give the genotypes of the individuals 3 and 4.

Ans. a. Individual 1 – I^BI^B or I^B i

Individual 2 – I^AI^A or I^A i

(One of them at least is definitely heterozygous).

b. Individual 5-Both glycoprotein A and glycoprotein B.

Individual 9-No glycoprotein

c. Individual 3-I^B i

Individual 4-I^A i

25. Explain the pattern of inheritance of haemophilia in humans.

Why is the possibility of a human female becoming a haemophilic extremely rare? Explain.

Ans. Haemophilia is a sex-linked, recessive disorder, with the defective allele on the X-chromosome.

The defective allele from the male parent is transmitted to the female progeny.

A carrier female (heterozygous, XX^h) passes on the disease to some of her sons, while a sufferer female (homozygous, X^hX^h) passes on the disease to all her sons.

The possibility of a female becoming haemophilic is extremely rare, because she has to be homozygous (X^hX^h) recessive for the trait, i.e., her father must be a haemophilic (X^hY, unviable at later stage of life) and mother must be haemophilic or at least a carrier (XX^h).

Selfing : Father Haemophilic X^hY × Mother Carrier XX^h

Gametes : X^h Y × X X^h

Progeny : X, X^h

	X	X^h
X^h	XX^h Carrier Female	X^hX^h Haemophilic Female
Y	XY Normal Male	X^hY Haemophilic Male

26. Explain the law of Dominance using a monohybrid cross.

Ans. The doctor must have used Pedigree analysis; which refers to the analysis of distribution and movements of traits in a series of generations of a family.

Since the non-haemophilic parents give rise to a haemophilic child, the genotypes of them should be:

Father : XY (normal)

Mother : XX^h (carrier/heterozygous, non-haemophilic)

Selfing : Father XY × Mother XX^h

Gametes : X, Y × X, X^h

Progeny : X, X^h

	X	X^h
X	XX Normal Female	XX^h Carrier. non-haemophilic female
Y	XY Normal Male	X^hY Haemophilic Male

The progeny can consist of the following genotypes and phenotypes:

XX		XX^h		XY		X^hY
Normal Female	:	Carrier, non-haemophilic female	:	Normal Male	:	Haemophilic Male
1	:	1	:	1	:	1

27. Define and design a test-cross.

Ans. a. It is a recessive trait.

b. It is an autosomal trait.

c. Generation I → Aa and Aa.

Generation II → Third child - aa. Fourth child - Aa.

28. How is sex determined in human beings?

Ans. a. The trait is dominant.

b. It is autosomal.

c. Genotypes of parents in generation I : Female aa; male Aa.

Genotype of third child in generation II : Aa.

Genotype of first grandchild in generation III : Aa.

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

Ans. a. Genotype of the parents in generation I :

— Male (Father) — Aa

— Female (Mother) — Aa

Son (Generation II) — Aa

Daughter (Generation II) — aa

b. Genotype of the daughters in generation III — Aa

c. It is an autosomal trait, because if the sex-linked trait has to appear in the daughter (generation II), the father must have it; but he does not show the trait and so it is not sex-linked.

30. In peas, tallness is dominant over dwarfness and red colour of flowers is dominant over the white colour. When a tall plant bearing red flowers was pollinated with a dwarf plant bearing white flowers, the different phenotypic groups were obtained in the progeny in numbers mentioned against them :

Tall, Red = 138 ; Tal, White = 132 ; Dwarf, Red = 136 ; Dwarf, White = 128

Mention the genotypes of two parents and of the four offspring types.

Ans. a. Since a haemophilic son is born, the normal mother is heterozygous, i.e., her genotype is XX^h, father is XY.

The cross is as follows:

Cross : XX^h × XY

Parents : Father (normal) × Mother (carrier)

Gametes : X, Y × X, X^h

Progeny : X, X^h

	X	X^h
X	XX Normal Female	XX^h Carrier. non-haemophilic female
Y	XY Normal Male	X^hY Haemophilic Male

b. The genotypes of the parents must be:

Father - haemophilic (X^hY)

Mother - carrier (XX^h) or haemophilic (X^hX^h)

31. With the help of an example, differentiate between incomplete dominance and co-dominance.

Ans. Law of Dominance

Law of dominance states that dominance is a phenomenon, in which one allele of a gene expresses itself and suppresses the expression of the other allele of the same gene, when they are gene-present together in a hybrid.

When a cross was made between two individuals, one with tall stem (homozygous) and the other with dwarf stem, the F_1 individual had tall stem.

When an F_1 individual is self-pollinated, the F_2 produced tall and dwarf individuals in the ratio of 3 : 1.

Parents : Tall (TT) × Dwarf (tt)

Gametes : T × t

F_1 generation : Tt (tall)

Selfing : Tt × Tt

Gametes : T, t × T, t

F_2 generation : T × t

	T	t
T	TT Tall	Tt Tall
t	Tt Tall	tt Dwarf

The phenotypic ratio is

3 Tall : 1 Dwarf

The genotypic ratio is

1 TT : 2 Tt : 1 tt

32. How was it concluded that genes are located on chromosomes?

Ans. Test Cross

It is a cross devised by Mendel, where the offspring or an individual with a dominant phenotype, whose genotype is not known, is crossed with an individual homozygous for the trait.

A Monohybrid test cross is as follows:

The hybrid individual is crossed with a homozygous recessive individual.

		Hybrid Tall		Pure Dwarf
Gametes	:	(TT)	×	(tt)
Parents	:	(T) (t)		(t)

	t
T	Tt Tall
t	tt Dwarf

The phenotypic and genotypic ratios are the same

1 Tall (Tt) : 1 Dwarf (tt)

If the dominant individual had been homozygous, all the individuals of F_1 would have been tall (hybrid tall)

		Pure Tall		Pure Dwarf
Gametes	:	(TT)	×	(tt)
Parents	:	(T)		(t)
Progeny	:	Tt (tall)		

LONG ANSWER TYPE QUESTIONS (5 MARKS)

1. What is the inheritance pattern observed in the size of starch grains and seed shape of Pisum sativum? Work out the monohybrid cross showing the above traits. How does this pattern of inheritance deviate from that of Mendelian law of dominance.

Ans. Inheritance of size of starch grains and seed-shape in Pisum sativum

- In Pisum sativum, a single gene controls two phenotypes:

  (i) the size of starch grains

  (ii) the seed-shape.

  and such a phenomenon is called pleiotropy.
- Starch synthesis is controlled by a gene, which has two alleles, B and b.
- Strach is synthesised effectively by homozygote BB and the starch grains are large.
- Homozygotes bb, have lesser efficiency for starch synthesis and hence the starch grains produced are small.
- After maturation, BB seeds are round and bb seeds are wrinkled.
- Heterozygotes, Bb, produce round seeds, i.e., round seed-shape is dominant over wrinkled seed-shape.
- But the starch grains in them (Bb) are of intermediate size; this shows that the alleles show incomplete dominance for the size of starch grains, though they show complete dominance (round seeds) foe seed shape.

– The cross is as follows:

Parents : Round Seeds, Large grains × Wrinkled Seeds, Small grains

BB bb

Gametes : (B) (b)

F_1 generation : Bb

Selfing : Bb × Bb

Gametes : (B) (b) (B) (b)

F_2 generation : B b

B	BB Round seeds, Large grains	Bb Round seeds, Intermediate grains
b	Bb Round seeds, Intermediate size	bb Wrinkled seeds, Small grains

– The phenotypic and genotypic ration are the same and are as follows:

Round seeds, Large grains	Round seeds, Intermediate grains	Wrinkled seeds Small grains
BB	Bb	bb
1	2	1

– This has deviated from the Mendelian monohybrid phenotypic ration of 3 : 1 due to incomplete dominance of one of the traits controlled by the gene.

2. **a.** Lis the three different allelic forms of gene 'I' in humans. Explain the different phenotypic expressions, controlled by these three forms.

b. A woman with blood group 'A' marries a man with blood group 'O'. Discuss the possibilities of the inheritance of the blood groups in the following, starting with 'yes' or 'no' for each : **i.** They produce children with blood group 'A' only. **ii.** They produce children, some with 'O' blood group and some with 'A' blood group.

Ans. a. Inheritance of human blood-group character

The gene for blood group character exists in three allelic forms, I^A, I^B and i; it is a case of multiple allelism

The phenotypes and genotypes of these alleles are as follows

Blood group	Genotype(s)
A	$I^A I^A$ or I^A i
B	$I^B I^B$ or I^B i
AB	$I^A I^B$
O	ii

The alleles I^A and I^B are condominant and express themselves as blood group AB, when they are together.

Blood group O is homozygous recessive.

b. (i) Yes. When the woman is homozygous ($I^A I^A$) for blood group A, the children can be with only blood group A

Parents : Mother A - group × Father O - group

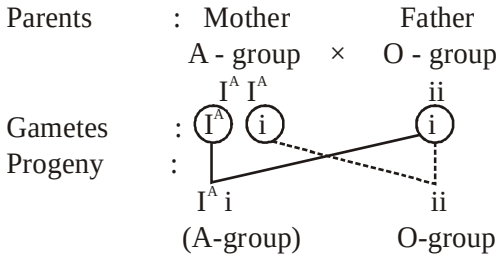
$I^A I^A$ ii

Gametes : (I^A) (i)

Progeny : I^A i (A-group)

OR

No. When the woman is heterozygous (I^A i) for blood group A, children will be of A group and O group.



(ii) Yes, when the woman is heterozygous (I^A i) for blood group A, children can be with either blood group A or O (as in the second cross of (i) above)

OR

No. When the woman is homozygous (I^A I^A) for blood group A, the children can be with only blood group A (as in the first cross of (i) of this answer).

3. **a.** Explain a monohybrid cross, taking seed coat colour as a trait in Pisum sativum. Work out the cross upto F_2 generation.

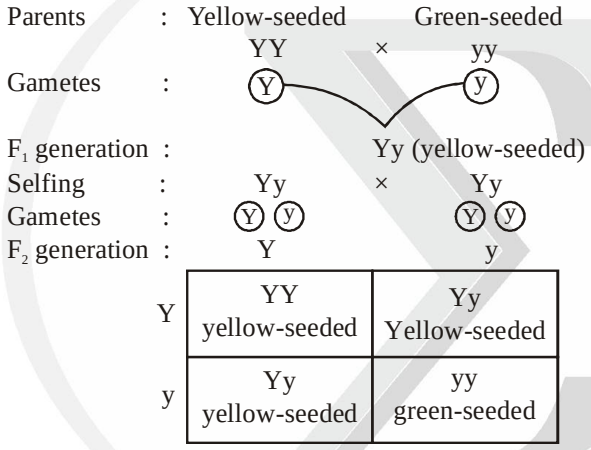
b. State the laws of inheritance that can be derived from such a cross.

c. What is the phenotypic ratio in a dihybrid cross?

Ans. a. Monohybrid cross

In Pisum sativum, yellow seeds are dominant over green seeds.

A cross between a true-breeding yellow-seeded plant and a true-breeding green-seeded plant, is as follows:



The F_2 phenotypic ratio is

3 yellow-seeded: 1 green-seeded

The F_2 genotypic ratio is

1 YY : 2 Yy : 1 yy

b. Medel's laws from the above cross.

(i) Law of dominance : When individuals differing in a pair of contrasting characters are crossed, the character that appears in the F_1 hybrids, is dominant (yellow seed coat colour) or recessive.

(ii) Law of segregation : The two factors of the trait (Y and y) that remained together in the hybrid, segregate during gametogenesis and enter different gametes.

c. The phenotypic ratio of a dihybrid cross is 9 : 3 : 3 : 1.

4. **a.** A garden pea plant bearing terminal, violet flowers, when crossed with another pea plant bearing axial, violet flowers produced axial, violet flowers and axial, white flowers in the ratio of 3 : 1.

Work out the cross showing the genotypes of the parent pea plants and their progeny.

b. Name and state the law that can be derived from this cross and not from a mohohybrid cross.

Ans. a. Dihybrid cross

Since white flowers have appeared in the progeny, both the parents must be heterozygous for flower colour, i.e., Vv.

Since all plants in the progeny have axial flowers, the parent with axial flowers, must be homozygous for the trait, i.e., TT.

Genotypes of the parents are as follows:

Terminal, violet flowers: tt Vv

Axial, violet flowers: TT Vv

Parents : Terminal Violet flowers tt Vv × Axial Violet flowers TT Vv

Gametes : (tV) (tv) (TV) (Tv)

Progeny : TV Tv

	TV	Tv
tV	Tt VV Axial, Violet flowers	Tt Vv Axial, Violet flowers
tv	Tt Vv Axial Violet flowers	Tt Vv Axial, White flowers

The phenotypic ratio:

3 Axial, violet flowers: 1 axial, white flowers is justified

b. Law of independent assortment : It states that when two pairs of traits are combined in a hybrid, segregation of one pair of traits is independent of the other pair of traits.

5. Describe the mechanism of pattern of inheritance of ABO blood groups in humans.

OR

ABO blood grouping in human population exhibits four possible phenotypes from six different genotypes. Explain the different mechanisms of inheritance involved in exhibiting the possibility of four phenotypes and six genotypes.

Ans. The gene for blood group character exists in three allelic forms, I^A, I^B, and i.

It is the phenomenon of multiple allelism, as there are more than two allelic forms of a gene.

Any individual carries two of the three alleles.

The allele I^A codes for glycoprotein A and the allele I^B codes for flycoprotein B, that are found on the surface of RBCs; the allele i does not produce any glycoprotein.

The allele I^A is dominant over i and I^B is also dominant over i.

When the alleles I^A and I^B are together, they are equally dominant and both the glycoproteins A and B are produced, making the blood group AB; this phenomenon, where both the alleles are equally dominant, is known as codominance.

The blood group is determined by the glycoprotein(s) on the RBCs.

There are six genotypes and four phenotypes as given in table.

Genotype(s)	Blood group
$I^A I^A$ or I^A i	A
$I^B I^B$ or I^B i	B
$I^A I^B$	AB
ii	O

6. a. Why is haemophilia generally observed in human males? Explain the conditions under which a human female can be haemophilic.

b. A pregnant human female was advised to undergo MTP. It was diagnosed by her doctor that the foetus she is carrying has developed from a zygote formed by an XX-egg fertilised by Y-carrying sperm. Why was she advised to undergo MTP?

Ans. a. The genes for haemophilia and colourblindness are present on the X-chromosome.

A male has only one X-chromosome and bears only one allel for the trait i.e., he is hemizygous for the trait, as

Y-chromosome does not have a corresponding allele.

A female has two X-chromosomes, received one from each of her parents; she has to be homozygous recessive, i.e., her father must be a sufferer and mother either a sufferer or a carrier, to develop the disease.

b. The zygote will be XXY and develop into a male with Klinefelter's syndrome.

Such individuals are sterile.

They show gynaecomastia and feminine characters.

To avoid such a hereditary disorder, the woman was advised M.T.P.

7. A true-breeding homozygous pea plant with green pods and axial flowers, as dominant characters, is crossed with a recessive homozygous pea plant with yellow pods and terminal flowers. Work out the cross upto F_2 generation, giving the phenotypic ratios of F_1 and F_2 generation, giving the phenotypic ratios of F_1 and F_2 generations, respectively.

Ans. a. Parents : Green pods, Axial flowers (GGAA) × Yellow pods, Terminal flowers (ggaa)

Gametes : GA ga

F_1 generation : GaAa-(Green pods, axial flowers)

Selfing of F_1 : GaAa × GgAa

Gametes :

F_2 generation :

	GA	Ga	gA	ga
GA	GGAA Green pods, axial flowers	GGAa Green pods, axial flowers	GgAA Green pods, axial flowers	GgAa Green pods, axial flowers
GA	GGAa Green pods, axial flowers	GGaa Green pods, terminal flowers	GgAa Green pods, axial flowers	Ggaa Green pods, terminal flowers
gA	GgAA Green pods, axial flowers	GgAa Green pods, axial flowers	ggAA Yellow pods, axial flowers	ggAa Yellow pods, axial flowers
ga	GgAa Green pods, axial flowers	Ggaa Green pods, terminal flowers	ggAa Yellow pods, axial flowers	ggaa Yellow pods, terminal flowers

The phenotypic ratio of F_2 generation is

Green pods, axial flowers : 9

Green pods, terminal flowers : 3

Yellow pods, axial flowers : 3

Yellow pods, terminal flowers : 1

b. Law of Independent assortment : It states that when two pairs of traits are combined in a hybrid the factors of every character segregate independently of the factors of other pair os character.

8. **a.** Do you agree to the perception in our society that the woman is responsible for the gender of the offspring? Substantiate your answer scientifically.

b. How did Morgan explain likage of genes?

Ans. a. No, this perception is wrong.

Sex determination in humans is by the XY-chromosomes

A male has 22 pairs of autosomes and a pair of sex chromosomes, XY.

A female has 22 pairs of autosomes and pair of sex chromosomes, XY.

The human female is homogamatic and produces only one type of ova, all with 22 autosomes and one X-chromosome.

The human male is heterogametic and produces two types of gametes, 50% of them with 22 autosomes and one X-chromosome and 50% with 22 autosomes and one Y-chromosome.

Sex of the offspring is determined at the time of fertilisation, by the type of sperm fertilising the ovum.

Parents : Male XY Female XX

Gametes : (X) (Y) (X)

XX (Female) XY (Male)

The probability of a male or a female child is equal, i.e., 50%

b. Linkage

Morgan worked with *Drosophila melanogaster* (fruit-fly).

He observed that when the two genes in a dihybrid cross are located on the same chromosome, the proportion of parental gene combinations was much higher than that of non-parental combinations or recombinants.

He attributed this to the physical association between the two linked genes and coined the term linkage.

Some gene pairs showed tight linkage, with very low recombination frequency and some showed loose linkage, and showed relatively, a higher recombination frequency, but much less than the expected 50% recombination frequency in a test cross.

9. a. How does a chromosomal disorder differ from a Mendelian disorder?

b. Name any two chromosomal aberration-associated disorders.

c. List the characteristics of the disorders mentioned above that helps in their diagnosis.

Ans. Differences :

Mendelian disorders	Chromosomal disorders
a. Mendelian disorders are due to alteration or mutation in a single gene.	a. Chromosomal disorders are those caused due to absence or excess number of one or more chromosome(s) or abnormal arrangement of one/more chromosome(s).
b. They are transmitted to the progeny in the same way as Mendelian principles of inheritance.	b. They are not transmitted as the afflicted individual is sterile.
c. The pattern of inheritance can be traced in a family tree by pedigree analysis, e..g, Phenylke-tonuria, Colourblindness.	c. The disease can be confirmed by karyotyping. e.g., Down's syndrome, Turner's syndrome.

b. Sickle-cell anaemia and Haemophilia.

c. **Sickle-cell anaemia**

The RBCs become elongated and sickle-shaped, as the mutant haemoglobin undergoes polymerisation under low oxygen tension.

Haemophilia

One of the proteins involved in the clotting of blood is affected; blood clotting is much delayed and there is non-stop bleeding from even a small cut.

10. Explain the causes, inheritance patterns and symptoms of any two Mendelian genetic disorders.

Ans. a. Sickle-cell anaemia

It is caused by a change of a single base pair in the gene, leading to substitution of glutamic acid by valine.

Inheritance pattern

It is transmitted from parents to the offspring, when both the partners are carriers (heterozygous) of the disease.

Symptoms

The RBCs become sickle-shaped.

b. Phenylketonuria

It is due to a defective allele on the autosome.

Inheritance pattern

It is passed on from the parent who are heterozygous for the gene to the offspring.

The affected individual lacks an enzyme that converts the amino acid phenylalanine into tyrosine; consequently, phenylalanine gets accumulated and converted into phenylpyruvate and other derivatives.

Symptoms

Accumulation of these compounds in the brain results in mental retardation; they are also excreted in the urine.

11. Write the symptoms of haemophilia and sickle-cell anaemia in humans. Explain how the inheritance pattern of the two diseases differ from each other.

Ans. Symptoms of Haemophilia

— Since a protein necessary for blood clotting is not formed, the blood does not clot and there is non-stop bleeding in case of an injury in the affected individual.

Symptoms of sickle-cell anaemia

— The RBCs become sickle-shaped.

— The oxygen transport to the tissues is impaired.

Differences:

Haemophilia	Sickle-cell anaemia
a. It is due to a defective recessive allele present on the X-chromosome, i.e., it is a sex-linked disorder.	a. It is due to point mutation, i.e., a single base pair change leading to a change in an amino acid, i.e., it is an autosomal disorder.
b. More males than females are affected.	b. Both males and females are affected equally.
c. The female parent passes on the disorder to male progeny, but father never passes it on to the male progeny.	c. The female parent on the disorder to male or female progeny in equal frequency and father also passes on the disorder to male and female progeny.

12. a. State the law of independent assortment.

b. Using Punnett square, demonstrate the law of independent assortment in a dihybrid cross involving two heterozygous parents.

OR

Let 'Y' be the genotype symbol for dominant yellow seed colour, symbol 'y' for recessive green seed colour, symbol 'R' for dominant round shape of seed and symbol 'r' for recessive wrinkled seed shape in garden pea. Using these symbols, explain Mendel's law of Independent assortment.

Ans. a. Law of Independent Assortment : This law states that when two pairs of traits are combined in hybrid, the factors of every character segregate independently of the factors of other pair of characters.

b. When two pea plants, heterozygous for flower position and flower colour (AaVv) are crossed, the results are as follows:

Parents : Axial, violet-flowered (AaVv) × Axial, violet-flowered (AaVv)

Gametes : (AV) (Av) (aV) (av) (AV) (Av) (aV) (av)

F_2 generation :

	AV	Av	aV	av
AV	AAVV Axial, violet-flowered	AAVv Axial, violet-flowered	AaVV Axial, violet-flowered	AaVv Axial, violet-flowered
Av	AAVv Axial, violet	AAvv Axial, white	AaVv Axial, violet	Aavv Axial, violet
aV	AaVV Axial, violet	AaVv Axial, violet	aaVV Terminal, violet	aaVv Terminal, violet
av	AaVv Axial, violet	Aavv Axial, violet	aaVv Terminal, violet	aavv Terminal, white

The phenotypic ratio of F_2 generation:

Axial, violet-flowered	:	Axial, white-flowered	:	Terminal, violet-flowered	:	Terminal, white-flowered
9	:	3	:	3	:	1

This phenotypic ratio is the result of independent assortment, i.e., segregation of alleles for flower position independently of the alleles for flower colour.

13. When a garden pea plant with green pods was cross-pollinated with another plant with yellow pods, 50% of the progeny bore green pods.

a. Work out the cross to illustrate this. **b.** How do you refer to this type of cross?

c. Sex-determination in human beings is an example of male heterogamity. Why is it called so?

Ans. a. The cross is as follows:

In garden Pea, green pod is dominant over yellow pod.

Since yellow pods have appeared in the progeny, the parent with green pod is heterozygous, i.e., Gg.

Parents : green pods yellow pods

Gg gg

Gametes : (G) (g) (g)

Progeny : G g

g	Gg Green pods	gg yellow pods

The phenotypic ratio and genotypic ratio are: 1 green pod (Gg): 1 yellow pod (gg)

b. Such a cross is referred to as test cross; such a cross is carried out to determine the genotype of the individual with a dominant phenotype.

c. **Heterogamety in human males**

Human males have 22 pairs of autosomes and XY-sex chromosomes

A human male is heterogametic since the produces two types of gametes with reference to sex chromosomes, i.e., 50% of them with one X-chromosome and the other 50% with one Y-Chromosome, and 22 autosomes.

14. Inheritance pattern of flower colour in garden pea plant and snapdragon differs. Why is this difference observed? Explain showing the crosses upto F_2 generation.

Ans. Inheritance of flower colour in garden pea : Inheritance of flower colour in garden pea shows true dominance and the F_1 hybrid expresses one of the parental characters, i.e., dominant trait and F_2 generation shows both dominant and recessive traits in the ratio of 3 : 1.

Parents : Purple-flowered × White-flowered

PP pp

Gametes : (P) (p)

F_1 generation : Pp (Purple-flowered)

Selfing : Pp × Pp

Gametes : (P) (P) (P) (P)

F_2 Progeny : P P

P	PP Purple-flowered	Pp Purple-flowered
P	Pp Purple-flowered	PP White-flowered

The phenotypic ratio is :

3 Purple-flowered : 1 White-flowered.

The genotypic ratio is :

1 PP : 2 Pp : 1 pp

Inheritance of flower colour in snapdragon.

Inheritance of flower colour in snapdragon shows incomplete dominance, a phenomenon in which neither of the two alleles is completely dominant over the other and the hybrid is intermediate between the two.

– In snapdragon flowers, red is homozyous dominant, white is homozyous recessive while the hybrid is intermediate, i.e., it is pink.

Parents : Red-flowered × White-flowered

RR rr

Gametes : (R) (r)

F_1 generation : Pp (Purple-flowered)

Selfing : Pp × Pp

Gametes : (R) (r) (R) (r)

F_2 Progeny : P P

P	RR Red-flowered	Rr Pink-flowered
P	Rr Pick-flowered	rr White-flowered

The phenotypic and genotypic ratios are the same.

Red-flowered		Pink-flowered		White-flowered
RR	:	Rr	:	rr
1		2		1

15. You are given a red flower-bearing pea plant and a red flower-bearing snapdragon plant. How would you find the genotypes of these two plants with respect to the colour of the flower? Explain with the help of crosses.

Comment upon the pattern of inheritance seen in these two plants.

Ans. The plants must be crossed with a white flowered (homozygous recessive) plant, i.e., a test cross is conducted.

Garden Pea : If the plant is homozygous dominant, all the plants in the progeny would bear red flowers (heterozygous) and it the plant is heterozygous, the progeny would consist of red-flowered plants and white-flowered plants in the ratio of 1 : 1.

a. Homozygous Red

Parents : Red flowered × White flowered

RR rr

Gametes : (R) (r)

Progeny : (Rr Red flowered)

b. Heterozygous Red

Parents : Red flowered × White flowered

Rr rr

Gametes : (R) (r) (r)

Progeny :

	R	r
r	Rr Red-flowered	rr White-flowered

Red flowered : White flowered, 1 : 1.

Snapdragon

In snapdragon, when a red-flowered plant is crossed to a white-flowered plant, the progeny would consist of pink-flowered plants.

So, red-flowered plants are homozygous dominant, while heterozygous condition produces pink-flowered plants.

It is because of incomplete dominance, where neither of the two alleles is completly dominant over the other and the hybrid is intermediate between the two i.e., pink-flowered.

The cross is as follows:

Parents : Red flowered × White flowered

Gametes : Rr (R) rr (r)

Progeny : Rr – Pink-flowered

16. A particular garden pea plant produces only violet flowers.

a. It is homozygous dominant for the trait or heterozygous?

b. How would you ensure the genotype? Explain wit the help of crosses.

OR

A particular garden pea plant produces only violet flowers. It may or may not be homozygous dominant for the trait. How would you ensure the genotype. Explain with crosses.

Ans. a. It must be homozygous dominant, since it produces only violet flowers.

b. The plant must be crossed with a plant bearing white (recessive) flowers.

If the progeny consists of plants, all producing violet flowers, the plant is homozygous dominant. (Cross 1)

If the progeny contains violet-flowered plants as well as white-flowered plants, the given plant is heterozygous. (Cross 2)

(i) Cross 1 : Homozygous dominant

Parents : Violet-flowered plants × White-flowered plants

Gametes : V v

Progeny : Vv – Violet-flowered

(ii) Cross 2 : Heterozygous

Parents : Violet-flowered plants × White-flowered plants

Gametes : VV (V, v) × vv (v)

Progeny : V × v

V	V v Violet-flowered
v	v v White-flowered

17. **a.** You are given tall pea plants with yellow seeds, whose genotypes are unknown. How would you find the genotype of these plants? Explain with the help of cross.

b. Identify a, b and c in the table given below :

	Pattern of inheritance	Monohybrid F_1	phenotypic expression
1.		Co-dominance	a
2.	b		The progeny resembled only one of the parents
3.	Incomplete dominance		c

Ans. a. The given plant has to be crossed with a dwarf plant with green seeds.

Tallness and yellow seeds are dominant traits, whereas dwarfness and green seeds are recessive traits.

If the progeny consists of tall plants with yellow seeds, the given plant is hozmozygous. (Cross 1)

If the progeny shows four phenotypes in the ratio of 1 : 1 : 1 : 1, the given plant is heterozygous for both the traits. (Cross 2)

Cross 1 : Homozygous pur for both the traits

Parents : Tall plants, yellow-seeded TTYY × Dwarf plants, green-seeded ttyy

Gametes : (TY) (ty)

Progeny : TtYy Tall plants, yellow-seeded

Cross 2 : Heterozygous for both the traits.

Parents : Tall plants, yellow-seeded TtYy × Dwarf plants, green-seeded ttyy

Gametes : (TY) (Ty) (tY) (ty) (ty)

Progeny :	TY	Ty	tY	ty
ty	TtTy Tall Plants, Yellow-seeded	Ttyy Tall Plants, green-seeded	ttYy Dwarf Plants, Yellow-seeded	ttYy Dwarf Plants, green-seeded

The phenotypic and genotypic ratios are 1 : 1 : 1 : 1 as shown in the table.

b. a – The progeny resembles both the parents.
b – Complete dominance.
c – The hybrid is intermediate between both the parents.

18. With the help of one example each, provide genetic explanation for the following observations :

a. F_1 generation resembles both the parents. **b.** F_1 generation does not resemble either of the parents.

Ans. a. F_1 generation resembles both the parents, if there is codominance.

Codominance is the phenomenon in which two alleles of a gene are equally dominant and both are expressed in the hybrid. e.g., AB blood group of humans, where alleles I^A and I^B are codominant.

b. F_1 generation does not resemble either of the parents, but is intermediate between the two in case is incomplete dominance.

Incomplete dominance is the phenomenon in which neither of the two alleles of a gene is completely dominant over the other and hence the hybrid shows intermediate trait.

19. a. Provide genetic explanation for the observation in which the flower colour in F_1 generation of snapdragon did not resemble either of the two parents. However parental characters reappeared when F_1 progenies were selfed.

b. State the three principles of Mendel's laws of inheritance.

Ans. a. Flower colour in snapdragon shows incomplete dominance, the phenomenon in which neither of the two alleles of a gene is completely dominant over the other and the hybrid becomes intermediate between the two.

In snapdragon, the genotype RR produces red flower colour and rr produces white flower colour; the hybrid Rr produces pink flowers.

When the F_1 hybrid is selfed, the progeny produced is as follows:

Pink-flowered Rr × Pink-flowered Rr

Gametes : (R) (r) (R) (r)

Progeny :	R	r
R	RR Red flowered	Rr Pink flowered
r	Rr Pink flowered	rr White flowered

The phenotypic and genotypic ratios are the same, i.e., 1 Red-flowered (RR): 2 Pink-flowered (Rr): 1 White-flowered (rr)

b. Medel's laws from the above cross

(i) Law of dominance : When individuals differing in a pair of contrasting characters are crossed, the character that appears in the F_1 hybrids, is dominant (yellow seed coat colour) or recessive.

(ii) Law of segregation : The two factors of the trait (Y and y) that remained together in the hybrid, segregate during gametogenesis and enter different gametes.

Law of independent assortment : It states that when two pairs of traits are combined in a hybrid, segregation of one pair of traits is independent of the other pair of traits.

20. a. Tallness of pea plant is a dominant trait; dwarfness is the alternate recessive trait. A pure tall pea plant is crossed to a dwarf one. Work out the cross to show what fraction of the tall plants in F_2 generation is heterozygous?

b. State any one law of Mendel which can be derived from this cross.

Ans. a. Round and yellow seeds

b. Rr Yy

c. (RY), (Ry), (rY), (ry)

d. 9 round, yellow seeded: 3 Round, green-seeded: 3 wrinkled, yellow seeded: 1 green, wrinkled - seeded

e. Yellow-seeds: green seeds = 3 : 1 ; Round seeds: Wrinkled seeds = 3 : 1

21. Explain the following terms with examples : **a.** Co-dominance **b.** Incomplete dominance

Ans. a. A true breeding pea plant homozygous for axial violet flowers is crossed with another pea plant with terminal white flowers (aavv). Work out the cross to show the phenotypes and genotpes of F_1 and F_2 generations along with the ratios.

Parents : Inflated, green pods FFGG × Constricted, yellow pods ffgg

Gametes : (FG) (fg)

F_1 generation : FfGg (Inflated, geen pods)

Selfing : FfGg × FfGg

Gametes : (FG) (Fg) (fG) (fg) (FG) (Fg) (fG) (fg)

F_2 generation :

	FG	Fg	fG	fg
FG	FFGG Inflated, green pods	FFGg Inflated, green pods	FfGG Inflated, green pods	FfGg Inflated, green pods
Fg	FfGg Inflated, green pods	FFgg Inflated, yellow pods	FfGg Inflated, green pods	Ffgg Inflated, yellow pods
fG	FfGg Inflated, green pods	FfGg Inflated, green pods	ffGG Constricted, green pods	ffGg Constricted, green pods
fg	Ffgg Inflated, green pods	ffGg Inflated, yellow pods	ffGg Constricted, green pods	ffgg Constricted, yellow pods

F_2 Phenotytic ratio– Inflated, green pods : Inflated, yellow pods : Constricted, green pods : Constricted, green pods

9 3 3 1

In F_1 generation all the individuals has inflated and green pods.

b. Medel's laws from the above cross

(i) Law of dominance : When individuals differing in a pair of contrasting characters are crossed, the character that appears in the F_1 hybrids, is dominant (yellow seed coat colour) or recessive.

(ii) Law of segregation : The two factors of the trait (Y and y) that remained together in the hybrid, segregate during gametogenesis and enter different gametes.

Law of independent assortment : It states that when two pairs of traits are combined in a hybrid, segregation of one pair of traits is independent of the other pair of traits.

22. Mention any two autosomal genetic disorders with their symptoms.

Ans. a. When two pea plants, heterozygous for flower position and flower colour (AaVv) are crossed, the results are as follows:

Parents : Axial, violet-flowered (AaVv) × Axial, violet-flowered (AaVv)

Gametes : (AV) (Av) (aV) (av) (AV) (Av) (aV) (av)

F_2 generation :

	AV	Av	aV	av
AV	AAVV Axial, violet-flowered	AAVv Axial, violet-flowered	AaVV Axial, violet-flowered	AaVv Axial, violet-flowered
Av	AAVv Axial, violet	AAvv Axial, white	AaVv Axial, violet	Aavv Axial, violet
aV	AaVV Axial, violet	AaVv Axial, violet	aaVV Terminal, violet	aaVv Terminal, violet
av	AaVv Axial, violet	Aavv Axial, violet	aaVv Terminal, violet	aavv Terminal, white

The phenotypic ratio of F_2 generation:

Axial, violet-flowered : Axial, white-flowered : Terminal, violet-flowered : Terminal, white-flowered

9 : 3 : 3 : 1

This phenotypic ratio is the result of independent assortment, i.e., segregation of alleles for flower position independently of the alleles for flower colour.

b. Medel's laws from the above cross: (i) Law of dominance : When individuals differing in a pair of contrasting characters are crossed, the character that appears in the F_1 hybrids, is dominant (yellow seed coat colour) or recessive. **(ii) Law of segregation :** The two factors of the trait (Y and y) that remained together in the hybrid, segregate during gametogenesis and enter different gametes.

Law of independent assortment : It states that when two pairs of traits are combined in a hybrid, segregation of one pair of traits is independent of the other pair of traits.|

23. A normal-visioned woman, whose father si color-blind, marries a normal-vision man. What would be the probability of her sons and daughters to be colour blind? Explain with the help of a pedigree chart.

Ans. Parents : Tall, green pods TTyy × Dwarf, yellow pods ttYY

Gametes : (Ty) (tY)

F_1 generation : TtYy– Tall, yellow seeds

Selfing : TtYy × TtYy

Gametes : (TY) (Ty) (tY) (ty) (TY) (Ty) (tY) (ty)

F_2 generation :

	TY	Ty	tY	ty
TY	TTYY Tall, yellow seeds	TTYy Tall, yellow seeds	TtYY Tall, yellow seeds	TtYy Tall, yellow seeds
Ty	TTYy Tall, yellow seeds	TTyy Tall, yellow seeds	TtYy Tall, yellow seeds	Ttyy Tall, green seeds
tY	TtYY Tall, yellow seeds	TtYy Tall, yellow seeds	ttYY Dwarf yellow seeds	ttYy Dwarf yellow seeds
ty	TtYy Tall, yellow seeds	Ttyy Tall, green seeds	ttYy Dwarf yellow seeds	ttyy Dwarf green seeds

a. The F_1 phenotype: Tall plants with yellow seeds. F_1 genotypes if TtYy

b. Phenotypic ratio of F_2 generation is as follows: Tall plants with yellow seeds : 9
Tall plants with green seeds : 3 Dwarf plants with yellow seeds : 3
Dwarf plants with green seeds : 1 The ratio is 9 : 3 : 3 : 1

24. Define aneuploidy. How is it different from polyploidy? Describe the individuals having the following chromosomal abnormalities : **a.** Trisomy of 21st chromosome **b.** XXX **c.** XO

Ans. a. It is called incomplete dominance.

b. Parents : Red flowers × White flowers
RR rr

Gametes : (R) (r)

F_1 generation : Rr – Pink flowers
Selfing : Rr × Rr

Gametes : (R) (r) × (R) (r)

F_2 generation :

	R	r
R	RR Red flowers	Rr Pink flowers
r	Rr Pink flowers	rr White flowers

The phenotypic and genotypic ratios in F_2 generation are same, i.e,.

Red flowers	:	Pink flowers	:	White flowers
RR		Rr		rr
1	:	2	:	1

c. The phenotypic ratio shows deviation from Mendelian monohybrid ratio of 3 : 1 to 1 : 2 : 1, since the heterozygous/ hybrid individuals show a different phenotype.

The genotypic ratio remains the same as Mendelian monohybrid ratio which is also 1 : 2 : 1.

25. What will happen : **a.** When complete sets of chromosomes are added to diploid genome?

b. When individual chromosomes are added to or deleted from the diploid genome?

c. When a part of the chromosome is lost?

d. When a part of the chromosome breaks and attaches to the homologue?

Ans. a. Parents : Tall plant × Dwarf plant
TT tt

Gametes : (T) (t)

F_1 generation : Tt (Tall plants)
Selfing : Tt × Tt

Gametes : (T) (t) × (T) (t)

F_2 generation :

	T	t
T	TT Tall plants	Tt Tall plants
t	Tt Tall plants	tt Dwarf plants

Two-thirds of all plants are heterozygous.

b. Medel's laws from the above cross

(i) Law of dominance : When individuals differing in a pair of contrasting characters are crossed, the character that appears in the F_1 hybrids, is dominant (yellow seed coat colour) or recessive.

(ii) Law of segregation : The two factors of the trait (Y and y) that remained together in the hybrid, segregate during gametogenesis and enter different gametes.