4 Principles of Inheritance and Variation

Fastrack Revision

- **Genetics** is the branch of biology which deals with inheritance and variations of characters from parents to offspring.
- Father of Genetics is **Gregor Johann Mendel.**
- **Inheritance** is the process of passing characters from parent to progeny. It is the basis of heredity.
- **Variation** is the degree by which progeny differs from their parents. Variations can be in terms of physiology, morphology and behavioural characteristics of individual belonging to same species. Variations arise due to reshuffling of chromosomes, crossing over, mutations and effect of environment.

Mendel's Law of Inheritance

- Mendel conducted hybridisation experiments on garden peas for seven years (1856-1863). On the basis of these experiments, he proposed the laws of inheritance.
- He selected the characters that has two opposing traits and concluded his hybridisation experiments on 14 true-breeding pea plant varieties. **True-breeding** means a breeding line which has undergone continuous self pollination and shows stable trait inheritance and expression for many generations.
- **Reasons for selecting garden pea plant**
  - They are easily available on large scale.
  - There are many varieties with distinct characteristics.
  - They are self-pollinated and can be cross-pollinated easily.
  - They have a short life cycle.
- **Reasons for success of Mendel**
  - He studied one character at a time.
  - He used available techniques to avoid cross-pollination by undesirable pollen grains.
  - He applied mathematics and statistics to analyse the results obtained from him.
  - Mendel selected seven contrasting characters for the experiment.

Inheritance of One Gene (Monohybrid Cross)

- Mendel crossed tall and dwarf pea plant and collected the seeds from them. Seeds were used to generate plants of first generation (F_1 or **Filial progeny**). Mendel observed that all the first generation plants were tall and none of them were dwarf. He made similar observations for the other pairs of traits. He concluded that F_1 generation resembled either one of the parents.
- He then self-pollinated the tall F_1 plants and observed that some of them were dwarf. *i.e.* 1/4th out of all.
- Similar results were obtained for other traits too. In F_2 generation, both the traits were expressed in proportion of 3:1. Dominant trait in F_2 is about thrice of the recessive form. These contrasting traits did not show any blending at either F_1 or F_2 stage.
- Based on these observations, he concluded that something was being stably passed from one generation to the other. He named it 'factors' which are now called as **'genes'.**
- Gene is the unit of inheritance. It contains information that is required to express a particular trait in an organism. Genes which code for a pair of contrasting traits are known as **'alleles'.** They are slightly different for a same gene.
- For representing traits using alphabetical symbols, capital letter is used for the trait expressed at F_1 generation and small letter is used for the other one.
- **Example: T** for tall trait, **t** for dwarf.
- **T** and **t** are alleles of each other. Pairs of alleles for height in the plants are **TT**, **Tt** and **tt**.
- **TT** and **tt** are **homozygous**. **TT** and **tt** are called **genotype** of the plant while the description terms **tall** and **dwarf** are **phenotype.** Tt represents **heterozygous.**
- **Test cross** is the cross between an individual with dominant trait and a recessive organism. It helps us to understand whether the dominant trait is homozygous or heterozygous.
- The production of gametes by the parents, formation of zygotes can be easily understood by **Punnett square.** It was given by British geneticist Reginald C. Punnett. It is a graphical representation used to calculate probability of all possible genotypes of offspring in a genetic cross.

Laws of Mendel

On the basis of his observations on monohybrid crosses, Mendel proposed two rules to consolidate his understanding of inheritance in monohybrid crosses. These rules are called **Laws of Inheritance** and are as follows:

- **Law of Dominance**
  - The dominant allele masks the effect of recessive allele. It explains the expression of only one of the parental characters in a monohybrid cross in F_1 and expression of both in F_2.
  - Characters are controlled by discrete units called **factors.**
  - In a dissimilar pair of factors, one member of pair dominate the other. **Example:** allele of tallness **(T)** is dominant over allele of dwarfness **(t).**

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- **Law of Segregation**
  - It states that every individual possess two alleles of a gene and these alleles segregate from each other during gamete formation (at the time of meiosis). Alleles do not blend and both the characters are recovered during gamete formation in F_2 generation.
  - Homozygous individuals produce one type of gametes while heterozygous individuals produce two types of gametes each having one allele with equal proportion.

Knowledge BOOSTER

The law of segregation is the most fundamental principle of heredity that has universal application with no exception.

Deviations from Mendel's Laws

On the basis of Mendel's principles, all the patterns of inheritance could not be explained which are:

- **Incomplete Dominance:** When the experiments were repeated using other traits in their plants, sometimes it was found that F_1 progeny does not resemble either of the parent but it was a mixture of two.

  Example: Snapdragon or *Antirrhinum* sp. or dog flower – inheritance of flower colour shows incomplete dominance.
- **Co-dominance:** The two alleles are able to express themselves independently when present together.
  - **Example:** ABO blood grouping in humans is controlled by gene I. It has three alleles I^A, I^B and i.
  - I^A, I^B is dominant over i. If I^A and i are present, only I^A expresses. If I^B and i are present, I^B expresses and if I^A and I^B are present, both of them express each other.
  - ABO blood grouping in human beings is also a good example of multiple alleles.

Inheritance of Two Genes (Dihybrid Cross)

Mendel also worked with two characters on pea plant. He chose colour and shape of the seed to explain the inheritance of two genes.

Y – Dominant yellow colour
y – Recessive green colour
R – Round shape of the seed
r – Wrinkled shape of the seed

- **Law of Independent Assortment**
  - It states that when two pairs of traits are combined in a hybrid, segregation of one pair of characters is independent of the other pair of characters.
  - The law was derived on the basis of Mendel's observations on dihybrid crosses in which the phenotypes—round, yellow; wrinkled, yellow; round, green and wrinkled, green appeared in the ratio 9 : 3 : 3 : 1.
- **Chromosomal Theory of Inheritance**
  - Mendel published his work on inheritance of characters in 1865 but was unrecognised till 1900.
  - In 1900, de Vries, Correns and von Tschermak worked independently and rediscovered Mendel's results on the inheritance of characters.
  - In 1902, Walter Sutton and Theodore Boveri studied the chromosomal movement during meiosis and gave some important results.
  - Genes are located at specific locations on the chromosomes.
  - Chromosomes as well as gene both occur in pairs.
  - Homologous chromosomes separate during meiosis.
  - Fertilisation restores chromosome number to diploid condition.
  - Chromosomes segregate as well as assort independently.
- **Linkage and Recombination**
  - Morgan carried out several dihybrid crosses in *Drosophila* to study genes that were sex-linked.
  - Morgan hybridised yellow-bodied, white-eyed females to brown-bodied, red-eyed males and intercrossed their F_1 progeny.
  - According to him, two genes did not segregate independently of each other and F_2 ratio deviated from 9:3:3:1. This concluded that genes are linked and this process is called **linkage.**
  - **Recombination** is the rearrangement of genetic material. The generation of non-parental gene combination during dihybrid cross is called recombination. When genes are located on same chromosome, they are tightly linked and show less linkage. This is responsible for variation.
- **Polygenic Inheritance**
  - There are many traits which are not so distinct in their occurrence and are spread across a gradient.
  - In humans, we don't have tall or short people as two distinct alternatives but a whole range of possible heights. Such traits are controlled by three or more genes and are called as polygenic traits. The phenomenon is called as polygenic inheritance.
- **Pleiotropy**
  - Sometimes, a single gene can exhibit multiple phenotypic expression. Such a gene is called a pleiotropic gene.
  - The mechanism of pleiotropy is the effect of a gene on metabolic pathways which contribute towards different phenotypes. *e.g.* disease phenylketonuria.
- **Sex Determination**
  - Different organisms have different types of sex determination.
  - Cytological observations in insects led to the development of concept of genetic or chromosomal basis of sex determination.
  - In 1891, Henking traced a specific nuclear structure all through spermatogenesis in few insects.
  - He observed that specific nuclear structure is located on 50% of sperms only. He discovered X-body but was unable to explain its significance.
  - In insects, XO type of sex determination is present. All the eggs have an additional X-chromosome besides the autosomes. Some sperms bear X-chromosome whereas some do not.
  - Eggs fertilised by sperm having an X-chromosome become females and those fertilised by sperms that do not have an X-chromosome become males.
  - **Example:** Grasshopper (males have only one X-chromosome besides autosomes and females have a pair of X-chromosomes).
- **Sex Determination in Humans**
  - XY type of sex determination is seen in human beings.

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- Males have autosomes plus XY while females have autosomes plus XX.
- *Drosophila* also has XY type of sex determination.
- Such a sex determination mechanism is an example of **male heterogamety.**

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Criss-cross inheritance is a type of sex-linked inheritance where a parent passes the traits to the grandchild of the same sex through offspring of the opposite sex, that is father passes the traits to grandson through his daughter (digynic) while the mother transfers traits to her grand daughter through her son (diandric).

Sex Determination in Birds

- ZW type of sex determination is seen in birds.
- Females have ZW and males have ZZ-chromosomes besides the autosomes.
- In birds, sex is determined by type of ovum, *i.e.* Z or W.
- In birds, females are heterogametic and such a sex determination mechanism is called **female heterogamety.**

Genetic Disorders

- Genetic disorders are caused by one or more abnormalities in autosomes or sex chromosomes of a person.
- They are Mendelian disorders and chromosomal disorders.

Mendelian Disorders

- **Autosomal Disorders:** Cystic fibrosis, sickle-cell anaemia, myotonic dystrophy, phenylketonuria, thalassemia.
- **Sex-linked Disorders:** Haemophilia, colour blindness.

I. Sickle-cell Anaemia

- Autosome linked recessive trait.
- It can be transmitted from parents to the offspring when both the parents are carrier for the gene.
- Disease is controlled by a single pair of allele, Hb^A and Hb^S.
- Hb^S Hb^S homozygous shows the diseased phenotype.
- Heterozygous individuals Hb^AHb^S show normal phenotype but they are carrier of the disease.
- The defect is caused due to substitution of Glutamic acid (Glu) by Valine (Val) at the sixth position of the beta globin chain of the haemoglobin molecule. It results from single base substitution from GAG to GUG at sixth codon of the beta globin.
- Due to this, mutant haemoglobin is formed. It undergoes polymerisation under low oxygen tension causing the change in the shape of the RBCs from biconcave to elongate sickle-like structure.

II. Thalassemia

- Autosome-linked recessive blood disease.
- Transmitted from parents to the offspring when both the partners are unaffected carrier for the gene.
- The defect could be due to either mutation or deletion which results in reduced rate of synthesis of globin chains (α and β chains) that make up haemoglobin.
- The formation of abnormal haemoglobin molecules result into anaemia.
- Thalassemia is a qualitative problem of synthesizing an incorrectly functioning globin.

III. Phenylketonuria

- Inborn error of metabolism and is an autosomal recessive trait.
- Affected individual lacks an enzyme that converts the amino acid phenylalanine into tyrosine.
- Due to this, phenylalanine gets accumulated and converted into phenyl pyruvic acid and other derivatives.
- Accumulation of phenylalanine causes mental retardation.

IV. Haemophilia

- Sex-linked recessive disease.
- A single protein that is a part of the cascade of proteins involved in the clotting of blood is affected.
- In affected individual, a simple cut will result in non-stop bleeding.
- Heterozygous female (carrier) can transmit the disease to son.
- Possibility of female becoming a haemophilic is extremely rare.

V. Colour Blindness

- Sex-linked recessive disorder.
- Defect occurs in either red or green cone of eye resulting in failure to discriminate between red and green colour.
- The defect is due to mutation in certain genes present in the X-chromosome.
- It occurs in about 8% of males and only about 0.4% of females.

Chromosomal Disorders

- It is caused due to absence or excess or abnormal arrangement of one or more chromosomes.
- **Aneuploidy:** Failure of segregation of chromatids during cell division cycle results in the gain or loss of a chromosome(s).
- **Polyploidy:** Failure of cytokinesis after telophase stage of cell division results in an increase in a whole set of chromosomes in an organism.
- Some common examples of chromosomal disorders are Down's syndrome, Turner's syndrome, Klinefelter's syndrome.

I. Down's Syndrome

- Gain of extra copy of chromosome 21 (trisomy 21).
- It was first described by Langdon Down (1866).
- Affected individual is short with small round head, furrowed tongue and partially open mouth.
- Broad palm with characteristic palm crease.
- Physical, psychomotor and mental development is retarded.

II. Turner's Syndrome

- Loss of an X-chromosome in human females *i.e.* 45 with XO.
- Such females are sterile as ovaries are rudimentary.
- Lack of other secondary sexual characters.

III. Klinefelter's Syndrome

- Presence of an additional copy of X-chromosome resulting into karyotype of 47, XXY.
- Overall masculine development though feminine development is also expressed.
- Sterile individuals.

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Practice Exercise

Multiple Choice Questions

Q 1. How many types of gametes would be produced if the genotype of a parent is AaBB?
(CBSE SQP 2021, Term-1)
a. 1 b. 2 c. 3 d. 4

Q 2. Which of the following statements indicates parallelism in genes and chromosomes?
(CBSE SQP 2021, Term-1)
(i) They occur in pairs
(ii) They segregate during gamete formation
(iii) They show linkage
(iv) Independent pairs segregate independently
a. (i) and (iii) b. (ii) and (iii)
c. (i), (ii) and (iii) d. (i), (ii) and (iv)

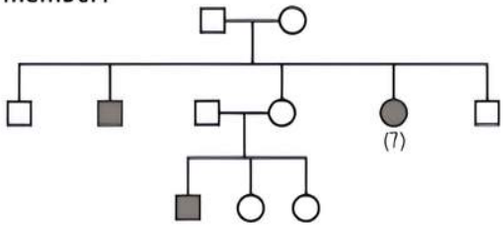
Q 3. In the F_2 generation of a Mendelian dihybrid cross, the number of phenotypes and genotypes are:
a. phenotypes – 4; genotypes – 16
b. phenotypes – 9; genotypes – 4
c. phenotypes – 4; genotypes – 8
d. phenotypes – 4; genotypes – 9

Q 4. The colour based contrasting traits in seven contrasting pairs, studied by Mendel in pea plant, were:
a. 1 b. 2 c. 3 d. 4

Q 5. Genes that code for a pair of contrasting traits are known as:
a. dominant genes b. alleles
c. linked genes d. None of these

Q 6. A recessive allele is expressed in:
a. heterozygous condition only.
b. homozygous condition only.
c. F_1 generation.
d. Both homozygous and heterozygous conditions.

Q 7. What should be the genotype of the indicated member?



a. AA b. Aa
c. XY d. aa

Q 8. In human beings, where genotype AABBCC represents dark skin colour, aabbcc represents light skin colour and AaBbCc represents intermediate skin colour; the pattern of genetic inheritance can be termed as: *(CBSE SQP 2021, Term-1)*
a. pleiotropy and codominance
b. pleiotropy and incomplete dominance
c. polygenic and qualitative inheritance
d. polygenic and quantitative inheritance

Q 9. Which of the following combination of chromosome numbers represents the correct sex determination pattern in honey bees? *(CBSE SQP 2021, Term-1)*
a. Male 32, Female 16 b. Male 16, Female 32
c. Male 31, Female 32 d. Male 32, Female 31

Q 10. Which of the following criteria must a molecule fulfil to act as a genetic material?
(CBSE SQP 2021, Term-1)
(i) It should not be able to generate its replica
(ii) It should chemically and structurally be stable
(iii) It should not allow slow mutation
(iv) It should be able to express itself in the form of Mendelian characters
a. (i) and (ii) b. (ii) and (iii)
c. (iii) and (iv) d. (ii) and (iv)

Q 11. A cross is made between tall pea plants having green pods and dwarf pea plants having yellow pods. In the F_2 generation, out of 80 plants how many are likely to be tall plants?
(CBSE SQP 2021, Term-1)
a. 15 b. 20 c. 45 d. 60

Q 12. In *Antirrhinum*, RR is phenotypically red flowers, rr is white and Rr is pink. Select the correct phenotypic ratio in F_1 generation when a cross is performed between RR × Rr: *(CBSE SQP 2021, Term-1)*
a. 1 Red : 2 Pink : 1 White b. 2 Pink : 1 White
c. 2 Red : 2 Pink d. All Pink

Q 13. What would be the genotype of the parents if the offspring have the phenotypes in 1:1 proportion?
(CBSE SQP 2021, Term-1)
a. Aa × Aa b. AA × AA
c. Aa × AA d. Aa × aa

Q 14. A couple has two daughters. What is the probability that the third child will also be a female?
(CBSE SQP 2021, Term-1)
a. 25% b. 50% c. 75% d. 100%

Q 15. Genotypic ratio of 1:2:1 is obtained in a cross between: *(CBSE SQP 2021, Term-1)*
a. AB × AB b. Ab × Ab c. Ab × ab d. ab × ab

Q 16. Person having genotype $I^A I^B$ would show the blood group as AB. This is because of:
a. pleiotropy
b. co-dominance
c. segregation
d. incomplete dominance

Q 17. ZZ/ZW type of sex determination is seen in:
a. platypus b. snails
c. cockroach d. peacock

Q 18. Choose the set of sex determining chromosomes that indicates the correct sex of the respective organism:
a. Homozygous sex chromosomes (ZZ) determine female sex in birds
b. XO type of sex chromosomes determine male sex in grasshoppers

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c. XXY condition in humans, as found in Turner syndrome, determines male sex
d. Homozygous sex chromosomes (XX) produce male sex in *Drosophila*.

Q 19. Number of autosomes present in liver cells of a human female is:
a. 22 autosomes b. 22 pairs
c. 23 autosomes d. 23 pairs

Q 20. Which of the following amino acid substitution is responsible for causing sickle-cell anaemia? *(CBSE SQP 2021, Term-1)*
a. Valine is substituted by glutamic acid in the α-globin chain at the sixth position
b. Valine is substituted by glutamic acid in the β-globin chain at the seventh position
c. Glutamic acid is substituted by valine in the α-globin chain at the sixth position
d. Glutamic acid is substituted by valine in the β-globin chain at the sixth position

Q 21. Rajesh and Mahesh have defective haemoglobin due to genetic disorders. Rajesh has too few globin molecules while Mahesh has incorrectly functioning globin molecules. Identify the disorder they are suffering from. *(CBSE SQP 2021, Term-1)*

	Rajesh	Mahesh
a.	Sickle-cell anaemia-an autosome linked recessive trait	Thalassemia-an autosome linked dominant trait
b.	Thalassemia-an autosome linked recessive blood disorder	Sickle-cell anaemia-an autosome linked recessive trait
c.	Sickle-cell anaemia-an autosome linked recessive trait	Thalassemia-an autosome linked recessive blood disorder
d.	Thalassemia-an autosome linked recessive blood disorder	Sickle-cell anaemia-an autosome linked dominant trait

Q 22. Researchers the world over are trying to transfer apomictic genes to hybrid varieties as hybrid characters in the progeny: *(CBSE 2021, Term-1)*
a. do not segregate
b. segregate
c. develop genetic variations
d. will remain unexpressed

Q 23. The cause of Klinefelter's syndrome in humans is: *(CBSE 2021, Term-1)*
a. absence of Y-chromosome
b. absence of X-chromosome
c. extra copy of an autosome
d. extra copy of an X-chromosome

Q 24. Select the incorrect pair: *(CBSE 2021, Term-1)*
a. Polygenic inheritance : Haemophilia
b. Linkage : *Drosophila*
c. Incomplete dominance : *Antirrhinum*
d. Pleiotropy : Phenylketonuria

Q 25. According to Mendel, the nature of the unit factors that control the expression of traits were: *(CBSE 2021, Term-1)*
a. stable b. blending
c. stable and discrete d. discrete

Q 26. Which of the following animals exhibit male heterogamety? *(CBSE 2021, Term-1)*
(i) Fruitfly (ii) Fowl
(iii) Human (iv) Honeybee
a. (i) and (iii) b. (ii) and (iv)
c. (ii) and (iii) d. (i) and (iv)

Q 27. The probability of all possible genotypes of offsprings in a genetic cross can be obtained with the help of: *(CBSE 2021, Term-1)*
a. Test cross b. Back cross
c. Punnett square d. Linkage cross

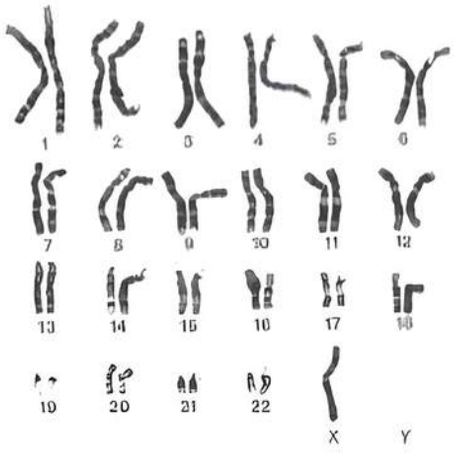
Q 28. The number of different types of gametes that would be produced from a parent with genotype AABBCc is: *(CBSE 2021, Term-1)*
a. 1 b. 2 c. 3 d. 4

Q 29. Select the important goals of HGP from the given options: *(CBSE 2021, Term-1)*
(i) Store the information for data analysis.
(ii) Clonning and amplification of human DNA.
(iii) Identify all the genes present in human DNA.
(iv) Use of DNA information to trace human history.
a. (i) and (ii) b. (ii) and (iii)
c. (i) and (iii) d. (ii) and (iv)

Q 30. In human rolling of tongue is an autosomal dominant trait (R). In a family both the parents have the trait of rolling tongue but their daughter does not show the trait, whereas the sons have the trait of rolling of tongue. *(CBSE 2021, Term-1)*
The genotypes of the family would be:

	Mother	Father	Daughter	Son
a.	Rr	Rr	rr	rr
b.	Rr	Rr	rr	RR
c.	rr	Rr	RR	rr
d.	RR	rr	Rr	Rr

Q 31. Given below is a Karyotype of a human foetus obtained for screening to find any probable genetic disorder: *(CBSE 2021, Term-1)*



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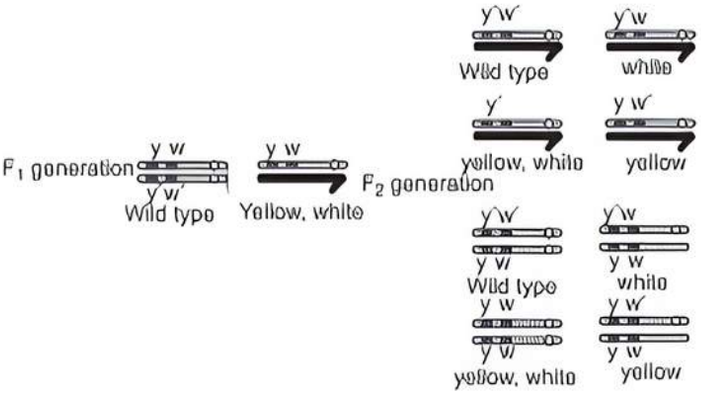
Based on the Karyotype, the chromosomal disorder detected in unborn foetus and the consequent symptoms the child may suffer from are:

a. Turner's syndrome : Sterile ovaries, short stature
b. Down's syndrome : Gynaecomastia, overall masculine stature
c. Turner's syndrome : Small round head, flat back of head
d. Down's syndrome : Furrowed tongue, short stature

Q 32. In the dihybrid cross that was conducted by Morgan involving mating between parental generation for genes yellow bodied, white eyed female *Drosophila* and wild type male *Drosophila*, upto F_2 generation is given below:



Study the result obtained of the F_2 progeny. Select the correct option from the given choices for the F_2 progeny.

a. Parental type, 1.3% : Strength of linkage high
b. Recombinant types, 1.3% : Strength of linkage low
c. Parental type, 98.7% : Strength of linkage high
d. Recombinant types, 98.7% : Strength of linkage low

Q 33. In *Pisum sativum*, the flower position may be axial (allele A) or terminal (allele a). What would be the percentage of the offspring with respect to axial flower position, if a cross is made between parents Aa × aa? *(CBSE 2021, Term-1)*

a. 25% b. 50%
c. 75% d. 100%

Q 34. Given below are the pairs of contrasting traits in *Pisum sativum* as studied by Mendel. Identify the incorrect pairs of traits: *(CBSE 2021, Term-1)*

	Character	Dominant	Recessive
a.	Stem height	Tall	Dwarf
b.	Seed shape	Round	Wrinkled
c.	Pod colour	Yellow	Green
d.	Flower position	Axial	Terminal

Q 35. A plant breeder crossed a pure bred tall plant having white flowers with a pure bred dwarf plant having blue flowers. He obtained 202 F_1 progeny and found that they are all tall having blue flowers.

Upon selfing these F_1 plants he obtained a progeny of 2160 plants. Approximately how many of these are likely to be short having blue flowers? *(CBSE 2021, Term-1)*

a. 1215 b. 405 c. 540 d. 135

Q 36. Possibility of the blood groups of the children in a family where the father is heterozygous for blood group 'A' and the mother is heterozygous for blood group 'B', would be: *(CBSE 2021, Term-1)*

a. Blood groups 'A', 'B'
b. Blood groups 'A', 'B', 'O'
c. Blood groups 'AB', 'O'
d. Blood groups 'A', 'B', 'AB', 'O'

Q 37. The correct statement with respect to thalassemia in humans is:

a. α-thalassemia is controlled by a single gene HBB.
b. The gene for α-thalassemia is located on chromosome 16.
c. β-thalassemia is controlled by two closely linked genes HBA-1 and HBA-2.
d. In β-thalassemia the production of α-globin chain is affected.

Assertion & Reason Type Questions

Directions (Q.Nos. 38-46): *Each of the following questions consists of two statements, one is Assertion (A) and the other is Reason (R). Select the correct answer to these questions from the codes a, b, c and d as given below.*

a. Both Assertion and Reason are true and Reason is the correct explanation of Assertion.
b. Both Assertion and Reason are true but Reason is not the correct explanation of Assertion.
c. Assertion is true but Reason is false.
d. Assertion is false but Reason is true.

Q 38. **Assertion (A):** In a monohybrid cross, F_1 generations indicate dominant characters.
Reason (R): Dominance occurs only in heterozygous state.

Q 39. **Assertion (A):** In thalassemia an abnormal myoglobin chain is synthesised due to a gene defect.
Reason (R): α-thalassemia is controlled by genes HBA-1 and HBA-2 on chromosome 16.

Q 40. **Assertion (A):** Pure lines are called true breed.
Reason (R): True breeds are used for cross breeding.

Q 41. **Assertion (A):** The principle of segregation given by Mendel is the principle of purity of gametes.
Reason (R): Gametes are pure for a character.

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Q 42. **Assertion (A): Co-dominant alleles lack dominant recessive relationship.**
Reason (R): Co-dominant alleles show incomplete dominance.

Q 43. **Assertion (A): Complementary genes are non-allelic genes.**
Reason (R): Complementary genes interact to produce a completely new trait.

Q 44. **Assertion (A): Quantitative inheritance is called polygenic inheritance.**
Reason (R): Several genes control the expression of a trait.

Q 45. **Assertion (A): When the two genes in a dihybrid cross are situated on the same chromosome, the proportion of parental gene combinations is much higher than non-parental type.**
Reason (R): Higher parental gene combinations can be attributed to crossing over between two genes. *(CBSE SQP 2021, Term-1)*

Q 46. **Assertion (A): Accumulation of phenylalanine in the brain results in mental retardation in phenylketonuria.**
Reason (R): The affected person lacks phenylalanine which is therefore not converted to tyrosine. *(CBSE SQP 2021, Term-1)*

Answers

1. (b) 2
According to the Law of Independent Assortment, an individual with genotype AaBB can produce gametes with either an allele 'A' or allele a for gene locus 'A'. However, for gene locus B, all gametes will carry the allele 'B' because this individual only carries one type of allele for this gene locus. Therefore, this individual will have two types of gametes, which is AB or aB.
2. (d) (i), (ii) and (iv)
The chromosome behaviour dictates the gene's segregation behaviour. They occur in pairs and they segregate during gamete formation. Also, the independent pairs segregate independently. Thus, there is a parallelism seen in genes and chromosomes.
3. (d) phenotypes = 4; genotypes = 9
4. (a) 1
5. (b) alleles
6. (b) homozygous condition only
7. (d) aa
8. (d) polygenic and quantitative inheritance
A genotype with all 'dominant' capital genes (AABBCC) has the maximum amount of melanin and very dark skin. A genotype with all 'recessive' small case genes (aabbcc) has the lowest amount of melanin and very light skin. Each 'dominant' capital gene produces one unit of colour, so that a wide range of intermediate skin colours are produced, depending on the number of 'dominant' capital genes in the genotype. Such a pattern of genetic inheritance is called polygenic and quantitative inheritance.
9. (b) Male 16, Female 32
The sex determination in honey bees is based on the number of sets of chromosomes an individual receives. An offspring formed from the union of a sperm and an egg develops as a female (queen or worker), and an unfertilised egg develops as a male (drone) by means of parthenogenesis. This means that the males have half the number of chromosomes than that of a female. The females are diploid having 32 chromosomes and males are haploid, *i.e.*, having 16 chromosomes.
10. (d) (ii) and (iv)
To act as a genetic material, a molecule should chemically and structurally be stable and it should be able to express itself in the form of Mendelian characters.
11. (d) 60
A cross is made between pure tall pea plant with green pods (TTGG) to dwarf pea plants with yellow pods (ttgg). In F_2 generation, 16 plants are formed, in this, tall green are 9, tall yellow are 3, dwarf green are 3 and dwarf yellow are 1. Tall plants are 12., green plants are 12, dwarf plants are 4, yellow plants are 4 that are formed.
Therefore, number of tall plants = 12/16 × 80 = 60.
12. (c) 2 Red : 2 Pink
When a cross is performed between RR × Rr, the resulting F_1 generation plants had a phenotypic ratio of 2 Red : 2 Pink.
13. (d) Aa × aa
If the offspring have the phenotypes in 1 : 1 proportion, then the genotype of the parents would be Aa × aa.
14. (b) 50%
Each pregnancy has a 50% chance of resulting in a boy or girl and this does not include the possibility of multiples. Having 2 daughters previously does not affect the chances of any resulting pregnancy. The chance of having third child as female remains at 50%.
15. (b) Ab × Ab
Genotypic ratio of 1 : 2 : 1 is obtained in a cross between Ab × Ab.
16. (b) co-dominance
17. (d) peacock
18. (b) XO type of sex chromosomes determine male sex in grasshoppers
19. (b) 22 pairs
20. (d) Glutamic acid is substituted by valine in the β-globin chain at the sixth position
Sickle-cell disease is caused by a single point mutation (a missense mutation) in the beta-haemoglobin gene that converts a CAG codon into GUG, which encodes the amino acid valine rather than glutamic acid in the β-globin chain at the sixth position.
21. (b) **Rajesh**-Thalassemia-an autosome linked recessive blood disorder.
Mahesh-Sickle-cell anaemia-an autosome linked recessive trait.
Through the given defect, it is clear that Rajesh is suffering from an autosome linked recessive blood disorder *i.e.*, Thalassemia and Mahesh is suffering from an autosome linked recessive trait *i.e.*, Sickle-cell anaemia.
22. (c) develop genetic variations
23. (d) extra copy of an X-chromosome
24. (a) Polygenic inheritance: Haemophilia
25. (c) stable and discrete

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26. (a) (i) and (iii)

27. (c) Punnett square
One of the easiest ways to calculate the mathematical probability of inheriting a specific trait was invented by an English Geneticist named as Reginald Punnett. His technique was thus popularly accepted as Punnett square. It is the simplest graphical way of discovering all possible combinations of genotypes that may occur in offsprings.

28. (b) 2

29. (c) (i) and (iii)

30. (b) Mother-Rr; Father-Rr; Daughter-rr; Son-RR

31. (a) Turner's syndrome: Sterile ovaries, short stature

32. (c) Parental type—98.7%; Strength of linkage high

33. (b) 50%

34. (c) Character-Pod colour; Dominant-Yellow; Recessive-Green.

35. (d) 135

$$TTWW \times ttww$$
$$\downarrow$$
$$TtWw$$
$$\downarrow \text{Self pollination}$$
$$TtWw \times TtWw \text{ (202 parts)}$$
$$2160 \text{ plants (total) in } F_2$$

TW — 9	According to ratio of dihybrid cross
Tw — 3	
tW — 3	
tw — 1	

The total number of short and blue flowered plants is: $\frac{1}{16} \times 2130 = \frac{1080}{8} = 135$.

36. (d) Blood groups 'A', 'B', 'AB', 'O'.

37. (b) The gene for α-thalassemia is located on chromosome 16.

38. (a) Both Assertion and Reason are true and Reason is the correct explanation of Assertion.
Monohybrid cross is a cross between two organisms of a species which is made to study the inheritance of a single pair of alleles or factors of a character. Dominant character is one of a pair of alleles which can express itself whether present in homozygous or heterozygous state. In F_1 generation (the generation of hybrids produced from a cross between the genetically different homozygous individuals called parents) the progenies are heterozygous dominant.

39. (d) Assertion is false but Reason is true.

40. (b) Both Assertion and Reason are true but Reason is not the correct explanation of Assertion.
Pure line is a strain of genetically pure true breeding individuals. Members of pure line are homozygous for one or more characters. In homozygous form both the factors express the same effect. These organisms are said to breed true. They are used for cross breeding in order to get the desired improvement in crops.

41. (a) Both Assertion and Reason are true and Reason is the correct explanation of Assertion.
According to principle of segregation (first law of Mendelism), the two factors of a character which remain together in an individual to not get mixed up but keep their identity distinct, separate at the time of gametogenesis or sporogenesis, get randomly distributed to different offspring as per the principle of probability. Gametes carry a single factor or allele for a trait. The two Mendelian factors present in the F_1 plants segregate during gamete formation. The principle of segregation is called the principle of purity of gametes because segregation of the two Mendelian factors of a trait results in gametes receiving only one factor out of a pair. As a result gametes are always pure for a character.

42. (c) Assertion is true but Reason is false.
Such alleles which are able to express themselves in the presence of each other are called co-dominant. They do not show dominant recessive relationship and are able to express themselves independently when present together. For example, Blood group A and B shows codominance as when present together. They show AB blood group. In incomplete dominance, one of the alleles is more pronounced for example in *Mirabilis jalapa* when red (dominant) and white (recessive) flowers crossed the progeny is pink. Therefore, co-dominance is different from incomplete dominance.

43. (b) Both Assertion and Reason are true but Reason is not the correct explanation of Assertion.

44. (a) Both Assertion and Reason are true and Reason is the correct explanation of Assertion.
Quantitative inheritance is a type of inheritance controlled by one or more genes in which the dominant alleles have cumulative effect with each dominant allele expressing a part or unit of the trait, the full trait being shown only when all the dominant alleles are present. The genes involved in quantitative inheritance are called polygenes. A polygene is defined as a gene where a dominant allele controls only a unit or partial quantitative expression of a trait.

45. (c) Assertion is true but Reason is false.
When the genes are linked, the proportion of parental gene combinations are much higher than non-parental types. When the genes for certain features (like black eyes and short wings of *Drosophila*) are located very close to each other on a chromosome, there are little chances of crossing over, giving higher parental combination. When the genes are on different locii on a chromosomes, the distance between them being considerable, there are higher chances of recombination, giving rise to mixed or non-parental features.

46. (c) Assertion is true but Reason is false.

Case Study Based Questions

Case Study 1

Inheritance of One Gene

According to Mendel, one gene controls the expression of one character only. The ability of a gene to have multiple phenotypic effect because it influences a number of characters is an exception. The gene having a multiple phenotypic effect because of its ability to control two or more characters can be seen in cotton. In cotton, a gene for the link also influences the height of plant, size of the ball, number of ovules and viability of seeds.

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Q 1. Genes with multiple phenotypic effects are known as:

a. hydrostatic genes
b. duplicate genes
c. pleiotropic genes
d. complimentary genes

Q 2. Which of the following disorder is an example of genes with multiple phenotypic effects?

a. Phenylketonuria b. Haemophilia
c. Sickle-cell anaemia d. Both a. and c.

Q 3. Which of the following is an example of gene with multiple phenotypic effect?

a. *Drosophila* white eye mutation.
b. Kernel colour in wheat.
c. Height in human beings.
d. Skin colour in human beings.

Q 4. Which of the following statements is not correct regarding genes with multiple phenotypic effect?

a. It is not essential that all the traits are equally influenced.
b. Occasionally a number of related changes are caused by a gene.
c. It occurs due to effect of the gene on two or more inter-related metabolic pathways.
d. None of the above

Q 5. Assertion (A): In garden pea, the gene which controls the flower colour also controls the colour of the seed coat and presence of red spots in the leaf axils.

Reason (R): A pleiotropic gene influences more than one trait.

a. Both Assertion and Reason are true, and Reason is the correct explanation of Assertion.
b. Both Assertion and Reason are true, but Reason is not the correct explanation of Assertion.
c. Assertion is true, but Reason is false.
d. Assertion is false but Reason is true.

Answers

1. (c) 2. (d) 3. (a) 4. (d) 5. (a)

Case Study 2

Haemophilia's a Mendelian Disorder

Haemophilia is a sex-linked disease which is also known as bleeder's disease as the patient will continue to bleed even from a minor cut since he or she does not possess the natural phenomenon of blood clotting due to absence of anti-haemophilic globulin or factor VIII and plasma thromboplastin factor IX essential for it. As a result of continuous bleeding, the patient may die of blood loss colour blindness is another type of sex-linked trait in which the eye fails to distinguish red and green colours. Vision is however, not affected and the colourblind can lead a normal life, reading, writing and driving (distinguishing traffic lights by their position).

Q 1. If a haemophilic man marries a woman whose father was haemophilic, and mother was normal then which of the following holds true for their progenies?

a. Of the total number of daughters, 50% daughters are carrier and 50% are haemophilic.
b. All the daughters are haemophilic.
c. All sons are haemophilic and all daughters are normal.
d. All sons are normal and all daughters are carriers.

Q 2. A man whose father was colourblind and mother was normal marries a woman whose father was haemophilic and mother was normal. Which of the following is true for their progenies? (Note: Percentage is from the total number of progenies).

a. 25% female progenies carry the gene for both haemophilia and colour blindness.
b. 25% male progenies carry only the gene of colour blindness.
c. 25% female progenies carry only the gene of colour blindness.
d. 25% male progenies and 25% female progenies carry the gene of haemophilia.

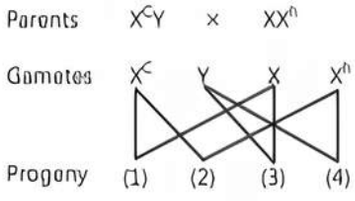
Q 3. Which of the following statements is incorrect regarding haemophilia?

a. It is a dominant disease.
b. A single protein involved in clotting of blood is affected.
c. It is recessive disease.
d. It is a Mendelian disorder.

Q 4. Anup is having colour blindness and is married to Soni who is normal. What is the chance that their son will have the disease?

a. 100% b. 50% c. 25% d. 0%

Q 5. Refer to the given cross.



Select the correct option regarding 1, 2, 3 and 4.

(a) 1.	Colourblind carrier female	(b) 1.	Colourblind people
2.	Colourblind haemophilic female	2.	Haemophilic female
3.	Normal male	3.	Normal male
4.	Haemophilic male	4.	Haemophilic male
(c) 1.	Colourblind female	(d) 1.	Colourblind carrier female
2.	Colourblind and haemophilic female	2.	Normal female
3.	Normal male	3.	Normal male
4.	Normal male	4.	Haemophilic male

Answers

1. (a) 2. (d) 3. (a) 4. (d) 5. (a)

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Case Study 3

Mendel's Law of Inheritance

Prashant wanted to find the genotype of a pea plant bearing purple coloured flowers in his kitchen garden. For this, he crossed purple flowered plant with white flowered plant. As a result, all plants which were produced had purple flower only. Upon selfing these plants, 75 purple flower plants and 25 white flower plants were produced. Now, he can determine the genotype of a purple flowered plant by crossing it with a white flowered plant.

Read the given passage carefully and give the answer of the following questions:

Q 1. Which law cannot be derived from the crosses done by Prashant?

Ans. Mendel's law of independent assortment cannot be derived from the crosses done by Prashant.

Q 2. To determine the genotype of a purple flowered plant, Prashant crossed this plant with a white flowered plant. What does this cross represent?

Ans. This cross represents a the test cross.

Q 3. In white flowered plant, allele is expressed in which condition?

Ans. In white flowered plant, allele is expressed in homozygous condition only.

OR

What does the character, *i.e.*, purple colour of the flowers that appeared in the first filial generation is called?

Ans. Such a character is called dominant character.

Case Study 4

Chromosomal Disorders

The chromosome number is fixed for all normal organisms leading to species specification whereas any abnormality In the chromosome number of an organism results into abnormal individuals. For example, In humans 46 is the fixed number of chromosomes both in male and female. In male it is '44 + XY' and in female it is '44 + XX'. Thus the human male is heterogametic, in other words produces two different types of gametes one with '22 + X' chromosomes and the other with '22 + Y' chromosomes respectively. Human female, on the other hand is homogametic *i.e.* produces only one type of gamete with '22 + X' chromosomes only.

Sometimes an error may occur during meiosis of cell cycle, where the sister chromatids fail to segregate called non-disjunction, leading to the production of abnormal gametes with altered chromosome number. On fertilisation such gametes develop into abnormal individuals.

Read the given passage carefully and give the answer of the following questions:

Q 1. State what is aneuploidy. *(CBSE 2023)*

Ans. Aneuploidy is a genetic condition in which an individual has an abnormal number of chromosomes in their cells. It may occur due to extra or less number of chromosomes.

Q 2. If during spermatogenesis, the chromatids of sex chromosomes fail to segregate during meiosis, write only the different types of gametes with altered chromosome number that could possibly be produced.

Ans. 22 + O, 22 + XY.

Q 3. A normal human sperm (22 + Y) fertilises an ovum with karyotype '22 + XX'. Name the disorder the offspring thus produced would suffer from and write any two symptoms of the disorder.

Ans. Klinefelter syndrome

Symptoms:

1. Males with feminine characteristics
2. Decreased facial and body hair.

OR

Name a best known and most common autosomal aneuploid abnormality in human and write any two symptoms. *(CBSE 2023)*

Ans. Down syndrome is the most well-known example of a chromosomal aneuploidy.

Symptoms:

1. A flattened face, especially the bridge of the nose.
2. Almond-shaped eyes that slant up.

Case Study 5

Genetic Mechanism

While studying inheritance of characters, a teacher gave the example of inheritance of attached earlobe and hypertrichosis of the ear to her students. A man with attached earlobes and extensive hair on pinna married a woman having free earlobes. The couple had four children, one son with attached earlobes and hairy pinna, one son with a free earlobes and hairy pinna and two daughters with attached earlobes. One of the daughters married a man with free earlobes and sparse hair on pinna. Teacher said if this couple would have sons, there would be equal chances for both having free or attached earlobes and sparse hair on pinna.

Read the given passage carefully and give the answer of the following questions:

Q 1. Which trait are of attached and free earlobe respective examples?

Ans. Attached and free earlobe respective examples of recessive and dominant traits.

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Q 2. What is dominant trait?

Ans. It is a character expressed by a dominant gene. Dominant gene is the one which expresses itself in the presence of its contrasting which is considered to be a suppressed gene.

Q 3. What type of trait is hypertrichosis of the ear?

Ans. Hypertrichosis of the ear is a Y-linked trait.

OR

If a female with attached earlobe married a male homozygous with free earlobe and sparse hair on pinna, then what would be the chances of daughter to have attached earlobe?

Ans. The chances of daughter to have attached earlobe is 0%.

Very Short Answer Type Questions

Q 1. Name the type of cross that would help to find the genotype of a pea plant bearing violet flowers. *(CBSE 2017)*

Ans. Test cross would help to find the genotype of a pea plant bearing violet flowers.

Q 2. British geneticist R.C. Punnett developed a graphical representation of a genetic cross called 'Punnett Square'. Mention the possible result this representation predicts of the genetic cross carried. *(CBSE 2019)*

Ans. Punnett Square for Mendelian monohybrid cross between pure line tall and dwarf plant will appear as follows:

Parent generation TT × tt

↓

F_1 generation Tt

↓

Tt × Tt

F_2 generation Selfing

	T	t
T	TT	Tt
t	Tt	tt

Result of F_2 generation phenotypic ratio of monohybrid cross is 3 : 1.

TiP

Draw well defined Punnett square from parental generation to F_2 generation.

Q 3. State a difference between a gene and an allele. *(CBSE 2016)*

Ans. Gene contains information that is required to express a particular trait whereas allele are the genes which code for a pair of contrasting traits.

Q 4. A garden pea plant (A) produced inflated yellow pod, and another plant (B) of the same species produced constricted green pods. Identify the dominant traits.

Ans. Inflated green pod is the dominant trait.

Q 5. A geneticist interested in studying variations and patterns of inheritance in living beings prefers to choose organisms for experiments with shorter life cycle. Provide a reason. *(CBSE 2015)*

Ans. This is because many generations can be obtained (in a short time) and selection of character becomes faster.

Q 6. Write the possible genotypes, Mendel got when he crossed F_1 tall pea plants with a dwarf pea plant.

Ans. Possible genotypes: Tt and tt.

Q 7. Mention any two contrasting traits with respect to seeds in pea plant that were studied by Mendel.

Ans. Round/Wrinkled, Yellow/Green traits.

Q 8. What are 'true breeding lines' that are used to study inheritance pattern of traits in plants?

Ans. True breeding lines are plants which have undergone continuous self-pollination for several generations. These are homozygous for traits.

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

Ans. Recessive allele expresses itself only in homozygous state in an organism.

Q 10. Write the percentage of F_2 homozygous and heterozygous populations in a typical monohybrid cross.

Ans. The ratio of a typical monohybrid cross is 1 : 2 : 1 where 50% are homozygous and 50% are heterozygous populations. (25% homozygous dominant, 25% homozygous recessive).

COMMON ERROR

Students usually write down the ratio, and they don't calculate the percentage. If percentage is asked then write in percentage form and if ratio then ratio is to be mentioned.

Q 11. Why, in a test cross, did Mendel cross a tall pea plant with a dwarf pea plant only?

Ans. This is to determine the genotype of the tall plant, whether it is homozygous dominant or heterozygous, as dwarfness is a recessive trait which is expressed only in homozygous condition and he was sure of genotype of dwarf plant.

Q 12. 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 genes are linked, the proportion of parental gene combinations would be much higher than non-parental types.

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Q 13. Write the types of sex determination mechanisms the following crosses show. Give an example of each type. (i) Female XX with Male XO (ii) Female ZW with Male ZZ

Ans. (i) Male heterogamety, Grasshopper
(ii) Female heterogamety, Birds.

Q 14. A male honeybee has 16 chromosomes whereas its female has 32 chromosomes. Give one reason.

Ans. Male honeybee develops from unfertilised female gamete (Parthenogenesis) and thus has 16 chromosomes whereas female develops by fertilisation and thus has 32 chromosomes.

Q 15. Why do normal red blood cells become elongated sickle shaped structures in a person suffering from sickle-cell anaemia?

Ans. Due to point mutation, glutamic acid (Glu) is replaced by valine (Val) at the sixth position of β-globin chain of haemoglobin molecule. Under oxygen stress erythrocytes lose their circular shape and undergo polymerisation to become sickle-shaped.

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

Ans. Father : 44 + XY
Mother : 44 + XX^h
Son : 44 + X^hY.
(X^h = X-chromosome with gene for haemophilia)

Q 17. Why is it that the father never passes on the gene for haemophilia to his sons? Explain.

Ans. Haemophilia is a sex-linked recessive disease and the defective gene is present on X-chromosome only and not on Y-chromosome. Father never passes X-chromosome to the son as father only contributes Y-chromosome to the son.

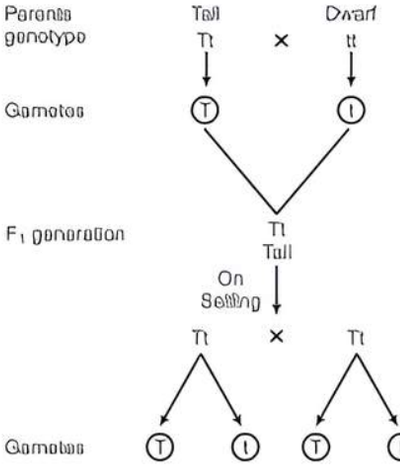
Q 18. A human being suffering from Down's syndrome shows trisomy of 21st chromosome. Mention the cause of this chromosomal abnormality.

Ans. Due to non-disjunction, 21st pair of chromosomes fails to separate during oogenesis. Therefore, the egg possesses 24 chromosomes instead of 23. When such an egg fuses with a sperm, the zygote will have three copies of chromosome 21 causing trisomy.

Short Answer Type Questions

Q 1. In a typical monohybrid cross, the F_2 population ratio is written as 3:1 for phenotype but expressed as 1:2:1 for genotype. Explain with the help of an example.

Ans. This is a case of Mendel's monohybrid cross. For example, here is a monohybrid cross between tall and dwarf pea plants.

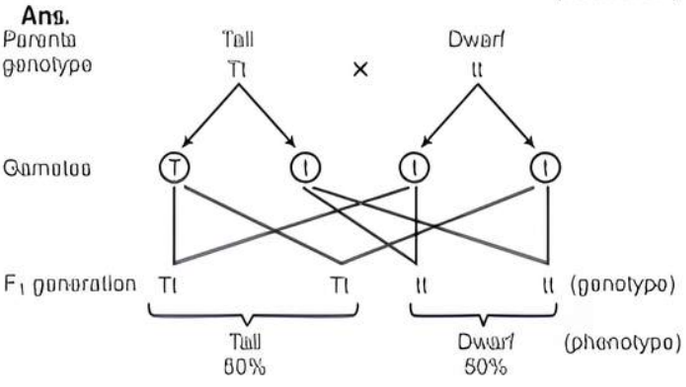


Phenotypic ratio - Tall plants : Dwarf plants
3 : 1
Genotypic ratio - TT : Tt : tt
1 : 2 : 1

Q 2. During a monohybrid cross involving a tall pea plant with a dwarf pea plant, the offspring populations were tall and dwarf in equal ratio. Work out a cross to show how it is possible.

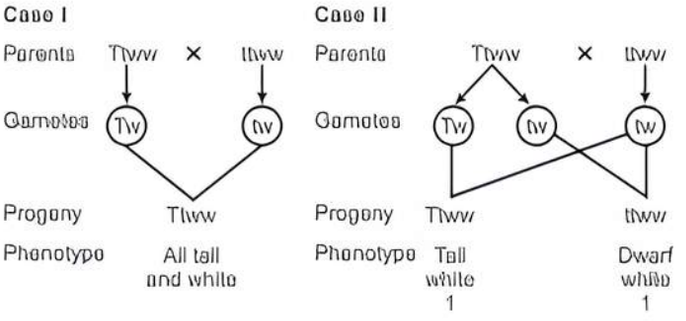
(CBSE 2015)

Ans.



Q 3. How would you find genotype of a tall pea plant bearing white flowers? Explain with the help of a cross. Name the type of cross you would use.

Ans. It can be done by a test cross. This is done by crossing the plant with homozygous recessive parent. If the ratio of progeny is 1 : 1, then the genotype of the plant is heterozygous.



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TiP

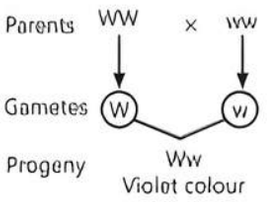
Practice making the diagrammatic cross, finding the gametes as well as the phenotypes and genotypes.

Q 4. A teacher wants his/her students to find the genotype of pea plants bearing purple coloured flowers in their school garden. Name and explain the cross that will make it possible. *(CBSE 2015)*

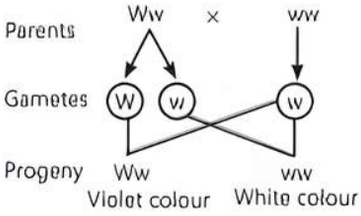
Ans. Test cross will be make it possible. It is a method devised by Mendel to determine the genotype of an organism. In this cross, the organism with unknown dominant genotype is crossed with the recessive parent.

In a monohybrid cross between violet colour flower (W) and white colour flower (w), the F_1 hybrid was violet colour flower. The test crosses are:

Case I



Case II

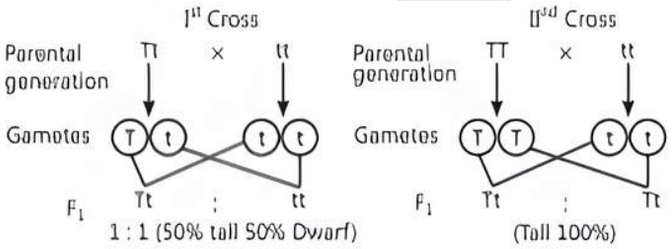


Q 5. Two independent monohybrid crosses were carried out involving a tall pea plant with a dwarf pea plant. In the first cross, the offspring population had equal number of tall and dwarf plants, whereas in the second cross, it was different. Work out the crosses, and explain giving reasons for the difference in the offspring population.

OR

Work out a cross to find the genotype of a tall pea plant. Name the type of cross.

Ans. This type of cross is called a test cross.



In the first cross, the tall parent plant is heterozygous for the trait and in second cross, the tall parent plant is homozygous for the trait, hence there respective observation.

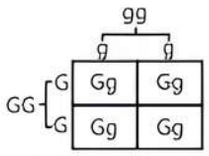
Q 6. By using Punnett square depict the genotypes and phenotypes of test crosses where green pod colour (G) is dominant over yellow pod colour (g) in garden pea with unknown genotype. *(CBSE 2023)*

Ans. Test cross is a type of genetic cross which is used to know the genotype of an unknown organism. In this, unknown parent is crossed with a homozygous recessive parent. Here, according to question:

Dominant allele = G and Dominant trait = Green pod colour

Recessive allele = g and Recessive trait = Yellow pod colour.

Condition I

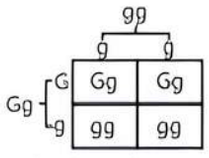


Here, Genotypic ratio: 100% heterozygous dominant *i.e.*, 1 : 0 ratio

Phenotypic ratio: All green pod colour

Conclusion: Unknown is homozygous dominant.

Condition II



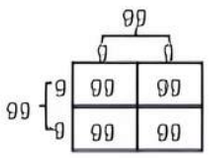
Here, Genotypic ratio: 50% heterozygous dominant 50% heterozygous dominant, *i.e.* 1 : 1 ratio

Phenotypic ratio: 50% green pod colour

50% yellow pod colour

Conclusion: Unknown is heterozygous dominant.

Condition III



Here, Genotypic ratio: 100% recessive *i.e.*, 1 : 0 ratio

Phenotypic ratio: All yellow pod colour

Conclusion: Unknown is homozygous recessive.

Q 7. In a dihybrid cross, white eyed, yellow bodied female *Drosophila* is crossed with red eyed, brown bodied male *Drosophila* produced in F_2 generation, 1.3% recombinants and 98.7% progeny with parental type combinations. This observation of Morgan deviated from Mendelian F_2 phenotypic dihybrid ratio. Explain, giving reasons, Morgan's observations.

Ans. Morgan's observations were:

(i) Morgan saw 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 type.

(ii) Morgan attributed this due to physical association or linkage of two genes and coined the term linkage to describe this physical association of genes on a chromosome and the term recombination to describe the generation of non-parental gene combinations.

Q 8. With the help of one example, explain the phenomena of co-dominance and multiple allelism in human population.

Ans. ABO blood group in human being is an example of multiple allelism and co-dominance. There are three alleles for the gene I, *i.e.*, I^A, I^B, and i, thus, exhibiting multiple allelism.

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When I^A and I^B are present together, the blood group is AB. Both A and B blood groups are expressed. This is called co-dominance.

Q 9. 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.

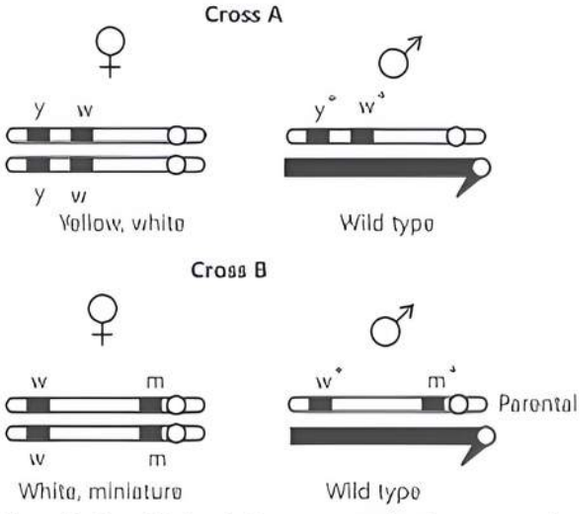
Similarities:

(i) Both genes and chromosomes occur in pairs in a diploid cell (2*n*).

(ii) Both of them separate out during gametogenesis to enter into different gametes.

(iii) Paired condition is again restored by fusion of gametes.

Q 10. Study the figures given below and answer the question.



Identify in which of the crosses is the strength of linkage between the genes higher. Give reasons in support of your answer.

Ans. The strength of linkage between the genes is higher in Cross A because the genes are closely placed. Lesser the distance between genes, greater is the strength of linkage.

Q 11. Write the scientific name of the fruit-fly. Why did Morgan prefer to work with fruit-flies for his experiments? State any three reasons.

Ans. *Drosophila melanogaster* is the scientific name of fruit-fly.

Morgan preferred to work with fruit-flies because of the followings reasons:

(i) It is grown in simple synthetic medium.

(ii) It completes the life cycle in only two weeks.

(iii) It is capable of producing large number of progeny at a time.

Q 12. Differentiate between "ZZ" and "XY" type of sex-determination mechanisms. *(CBSE 2015)*

Ans. ZZ type is seen in birds. The males are homogametic (ZZ) and females are heterogametic (ZY). Sex is determined by the type of egg getting fertilised.

XY type is seen in human beings. The males are heterogametic (XY) and females are homogametic (XX). Sex is determined by the type of sperm fertilising the ovum.

Q 13. Write the ploidy and number of chromosomes in human:

(i) melocyte and

(ii) gametes *(CBSE 2020)*

Ans. (i) In meiocytes the ploidy is 2*n* and number of chromosomes is 46. $2n = 46$

(ii) In gametes, the ploidy is *n* and the number of chromosomes is 23.

Q 14. What is aneuploidy? Name *n* chromosomal disorder in humans caused due to (i) gain of an autosome, and (ii) loss of a sex chromosome in females. *(CBSE 2020)*

Ans. Aneuploidy is a condition of having extra or missing chromosomes in a person.

(i) Down's syndrome

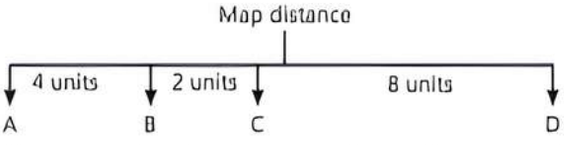
(ii) Turner's syndrome

TiP

Study the various types of chromosomal disorders with their causes and effects carefully.

Q 15. The map distance in certain organisms between gene A and B is 4 units, B and C is 2 units and between C and D is 8 units. Which one of these gene pairs will show more recombination frequency? Give reasons in support of your answer.

Ans.



The recombination frequency is directly proportional to the distance between the genes. The distance between C and D is more, *i.e.*, 8 units in the above condition, so recombination frequency will be more between them.

Q 16. Explain pleiotropy with the help of an example.

Ans. Pleiotropy is the phenomenon in which a single gene exhibits multiple phenotypic expression. The pleiotropic gene affects the metabolic pathways, resulting in different phenotypes.

For example, phenylketonuria is caused by mutation in the gene coding the enzyme phenylalanine hydroxylase.

Q 17. (i) Sickle celled anaemia in humans is a result of point mutation. Explain.

(ii) Write the genotypes of both the parents who have produced a sickle celled anaemic offspring.

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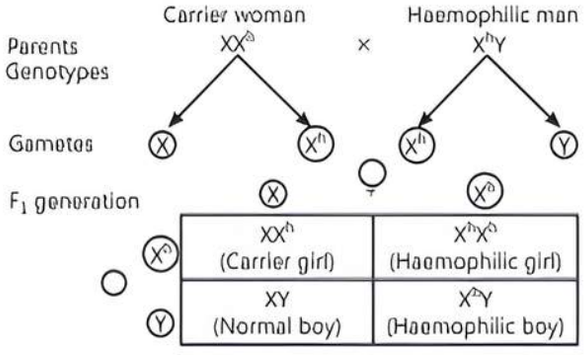
Ans. (i) In sickle cell anaemia, due to point mutation there is a substitution of a single nitrogen base at the sixth codon of the β-globin chain of haemoglobin that leads to substitution of value in place of glutamic acid.

GAG (Glutamic acid) —Mutation→ GUG (Valine)

(ii) The genotypes of both the parents would be Hb^AHb^S and Hb^AHb^S.

Q 18. Recently a baby girl has been reported to suffer from haemophilia. How is it possible? Explain with the help of a cross.

Ans. It is possible to have a haemophilic girl when the cross is between a carrier woman and a haemophilic man as shown below:



Long Answer Type-I Questions

Q 1. The F_2 progeny of a monohybrid cross showed phenotypic and genotypic ratio as 1: 2: 1, unlike that of Mendel's monohybrid F_2 ratio. With the help of a suitable example, work out a cross and explain how it is possible.

OR

Snapdragon shows incomplete dominance for flower colour. Work out a cross and explain the phenomenon. How is this inheritance different from Mendelian pattern of inheritance? Explain.

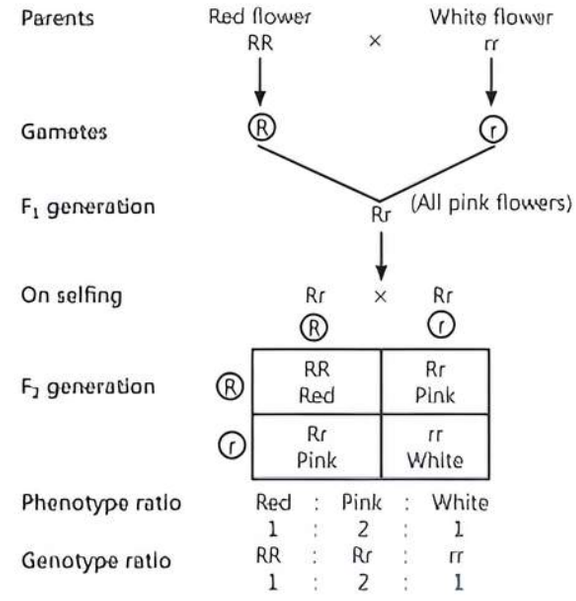
OR

In snapdragon (*Antirrhinum majus*), a plant with red flowers was crossed with a plant with white flowers. Work out all the possible genotypes and phenotypes of F_1 and F_2 generations. Comment on the pattern of inheritance in this case.

Ans. Such a cross is observed in *Mirabilis jalapa*/Four o'clock plant/*Antirrhinum majus*.

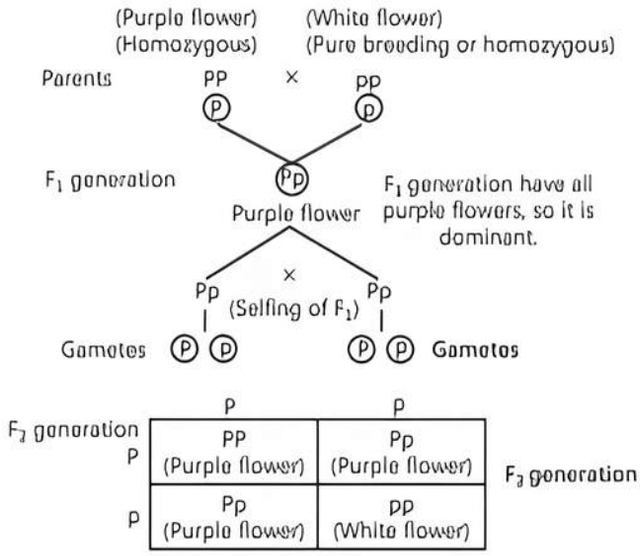
Monohybrid cross is observed in snapdragon, where one allele is incompletely dominant over the other allele.

In heterozygous condition, a single dominant gene is not sufficient to produce red colour, therefore it is a case of incomplete dominance.



Q 2. 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. The gene for purple flowers is dominant over that of white flowers. So, when two pure varieties are crossed, the F_1 generation has only purple flowers and on selfing, the flowers are produced in a 3 : 1 ratio.



This result is obtained due to segregation of the alleles at the time of gametogenesis. The alleles remain together in a zygote but during gamete formation, they segregate such that the gametes carry only one allele.

COMMON ERROR

Students do not study the crosses properly and do mistakes in dominant and recessive genes.

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Q 3. Why did T.H. Morgan select *Drosophila melanogaster* to study sex-linked genes for his lab experiments?

Ans. T.H. Morgan selected *Drosophila melanogaster* for his study because of the following reasons:

(i) It can be grown in a simple synthetic medium in laboratory.
(ii) It completes its life cycle in only two weeks.
(iii) Large number of progeny are produced at a time.
(iv) There is differentiation of sexes.
(v) Many hereditary variations can be observed.

Q 4. During his studies on genes in *Drosophila* that were sex-linked, T.H. Morgan found population phenotypic ratios deviated from expected 9 : 3 : 3 : 1. Explain the conclusion he arrived at.

Ans. The conclusions derived are:

(i) He 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 recombination of genes.
(ii) Morgan and his group found that when genes were grouped on the same chromosome, some genes are tightly linked and show less recombination.
(iii) When the genes are loosely linked, they show higher recombination.

Q 5. How are dominance, co-dominance and incomplete dominance patterns of inheritance different from each other? *(CBSE 2011)*

Ans. Dominance: It is a phenomenon in which when two contrasting alleles are present together, only one expresses itself and is called dominant whereas the other which does not express itself is called recessive.

Co-dominance: It is a phenomenon in which when two contrasting alleles are present together, both of the alleles express themselves.

Incomplete Dominance: It is a phenomenon in which when two contrasting alleles are present together, neither of the alleles is dominant over other and the phenotype formed is intermediate of the two alleles. e.g., Red flower × White flower ⟶ Pink flower

Q 6. Compare in any three ways the chromosomal theory of inheritance as proposed by Sutton and Bovery with that of experimental results on pea plant presented by Mendel. *(CBSE 2019)*

Ans. The chromosomal theory of inheritance and experimental results presented by Mendel can be compared in the following ways:

(i) In a diploid organism, the factors (genes) and chromosomes occur in pairs.
(ii) Both chromosomes as well as genes segregate at the time of gamete formation such that only one of each pair is transmitted to a gamete.
(iii) A gamete contains only one chromosome of a type and only one of the two alleles of a trait.
(iv) The paired condition of both chromosomes as well as Mendelian factors is restored during fertilisation. (Any three)

Q 7. (i) Explain linkage and recombination as put forth by T.H. Morgan based on his observations with *Drosophila melanogaster* crossing experiment.

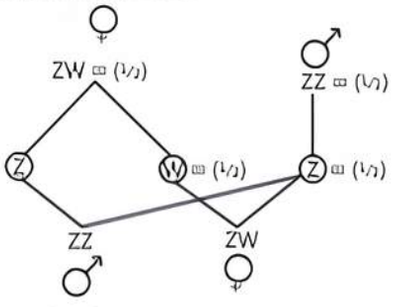
(ii) Write the basis on which Alfred Sturtevant explained gene mapping. *(CBSE 2019)*

Ans. (i) T.H. Morgan studied X-linked genes in *Drosophila* and saw that when the two genes in a dihybrid cross were situated on the same chromosome, the proportion of parental gene combinations was much higher than the non-parental type. He attributed this due to the physical association or linkage of the two genes on a chromosome and coined the term linkage and the term recombination describes the generation of non-parental gene combination.

(ii) Alfred Sturtevant explained gene mapping by using the frequency of recombination between gene pairs on the same chromosome as a measure of the distance between genes and he mapped their position on the chromosome.

Q 8. Explain the mechanism of 'sex determination' in birds. How does it differ from that of human beings? *(CBSE 2018)*

Ans. Sex determination in birds:



Birds: Female heterogamety is seen. Female produces (Z) type and (W) type of gametes = ½.

Humans: Male heterogamety is seen. Male produces (X) type and (Y) type of gametes = ½.

Q 9. Explain the phenomena of dominance, multiple allelism and co-dominance taking ABO blood group as an example.

Ans. Dominance: The alleles I^A and I^B both are dominant over allele *i* as I^A and I^B form antigens A and B, respectively, but *i* does not form any antigen.

Multiple allelism : It is the phenomenon of occurrence of a gene in more than two allelic forms on the same locus. In ABO blood group in humans, one gene I has three alleles I^A, I^B and I^O/i

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Co-dominance: It is the phenomena in which both alleles express themselves when present together. We inherit any two alleles for the blood group. When the genotype is I^AI^B, the individual has AB blood group since both I^A and I^B equally influence the formation of antigens A and B.

Q 10. Both haemophilia and thalassemia are blood related disorders in humans. Write their causes and the difference between the two. Name the category of genetic disorder they both come under. *(CBSE 2017)*

Ans. Both haemophilia and thalassemia come under Mendelian disorders.

Causes:

Haemophilia is a sex-linked recessive disorder. The gene for haemophilia is located on X chromosome. The gene passes from a carrier female to her son.

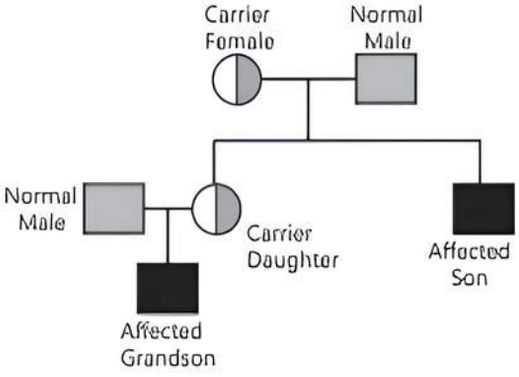
On the other hand, thalassemia is an autosomal linked recessive disease. It occurs due to either mutation or deletion resulting in reduced rate of synthesis of one of globin chains of haemoglobin.

Difference:

In haemophilia, clotting is affected, *i.e.*, there can be a non-stop bleeding even after a minor cut. But in thalassemia, anaemia is the main characteristic.

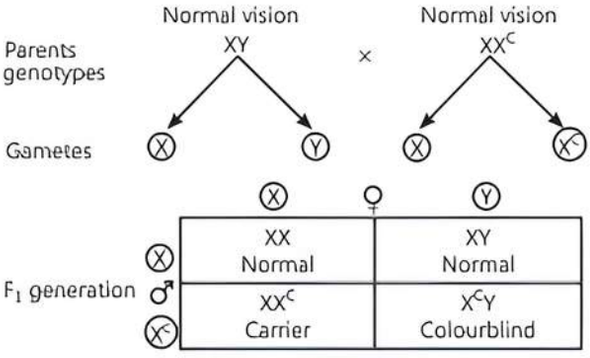
Q 11. A normal couple has their first child, who is haemophilic. Work out a cross to show how it is possible. State the possibility of the normal and the haemophilic children along with their sexes, which can be born to them. *(CBSE 2020)*

Ans. Haemophilia is a sex-linked recessive disease. Here, it is transmitted from the carrier female to the sons. From the below pedigree chart, it can be observed that the disease is being transmitted from the carrier female to her daughter (carrier) and son (affected). The carrier daughter transmits this disease to the grandson. This pattern of inheritance is called criss-cross inheritance.



Q 12. One of the twins born to parents having normal colour vision was colourblind whereas the other twin had normal vision. Work out the cross. Give two reasons how it is possible.

Ans. The cross is as shown:



It is possible in the following ways:

(i) When the mother is carrier of colourblindness gene, she will have normal vision but will pass on the gene to her children.

(ii) Another possibility is that there is a mutation on the X chromosome of one of the twins.

Q 13. 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 in humans due to Down's syndrome.

Cause: There is an additional copy of chromosome number 21 or trisomy of chromosome 21.

Symptoms:

(i) Short statured with small round head.

(ii) Partially open mouth with protruding furrowed tongue.

(iii) Palm is broad with characteristic palm crease.

(iv) Slow mental development.

Long Answer Type-II Questions

Q 1. Answer the following questions:

(i) State and explain the law of dominance as proposed by Mendel.

(ii) How would phenotypes of monohybrid F_1 and F_2 progeny showing incomplete dominance in snapdragon and co-dominance in human blood group be different from Mendelian monohybrid F_1 and F_2 progeny? Explain.

Ans. (i) The law of dominance states that when two alternative forms of a trait or character (genes or alleles) are present in an organism, only one factor expresses itself in F_1 progeny and is called dominant while the other that remains masked is called recessive. The characters are controlled by discrete units called factors. These factors occur in pairs.

	Mendelian monohybrid cross	Incomplete dominance	Co-dominance
F_1 Generation	All members resemble the parent with dominant trait.	All members do not resemble either of the two parents but show an intermediate trait.	Blood groups of all members resemble combination of dominant traits of both the parents.

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F_2 Generation	Both the parental traits reappear.	Both the parental traits and an intermediate trait appear.	Both the parental traits as well as the co-dominant trait appear.

Q 2. (i) State the law of independent assortment.

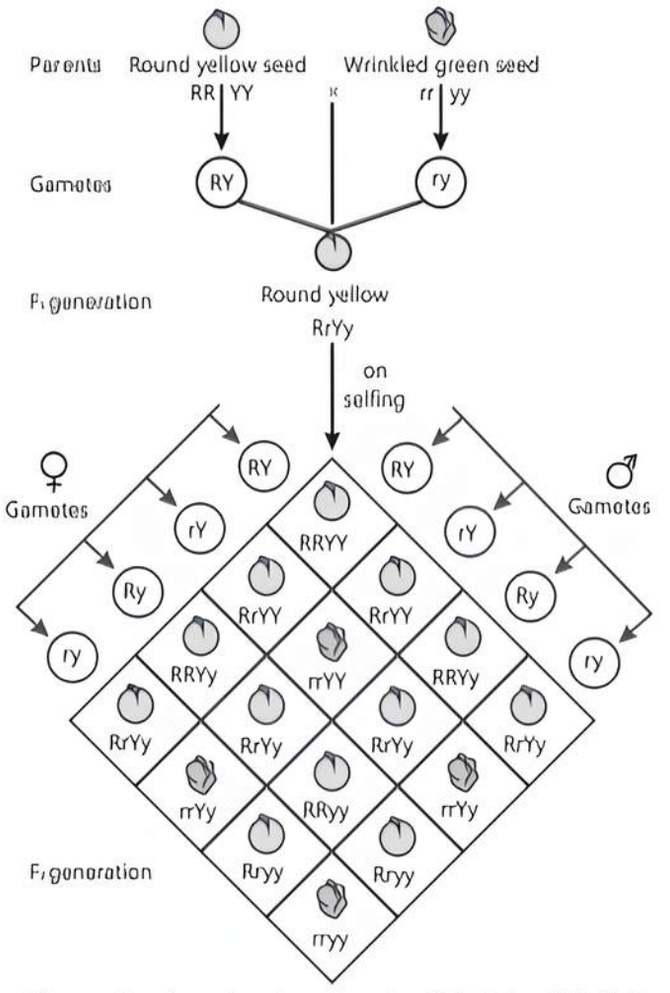
(ii) Using Punnett Square, demonstrate the law of independent assortment in a dihybrid cross involving two heterozygous parents.

OR

Describe the dihybrid cross upto F_2 generation as conducted by Gregor Mendel using pure lines of garden pea for characters seed shape and seed colour. *(CBSE 2023)*

Ans. (i) According to the law of independent assortment, the two factors of each character assort or separate out independent of the factors of other characters, at the time of gamete formation and get randomly rearranged in the offsprings, producing both parental and new combinations of characters.

(ii) **Demonstration of law of Independent Assortment:**



Phenotypic ratio: Round yellow : Round green : Wrinkled yellow : Wrinkled green

9 : 3 : 3 : 1

Genotypic ratio: 1 : 2 : 2 : 4 : 1 : 2 : 1 : 2 : 1

These are the results of a dihybrid cross where the two parents differed in two pairs of contrasting traits: seed colour and seed shape For the dihybrid cross Mendel derived the law of Independent Assortment. It states that when two pairs of traits are combined in a hybrid, segregation of one pair of character is independent of the other pair of characters.

Q 3. A homozygous tall pea plant with green seeds is crossed with a homozygous dwarf pea plant with yellow seeds.

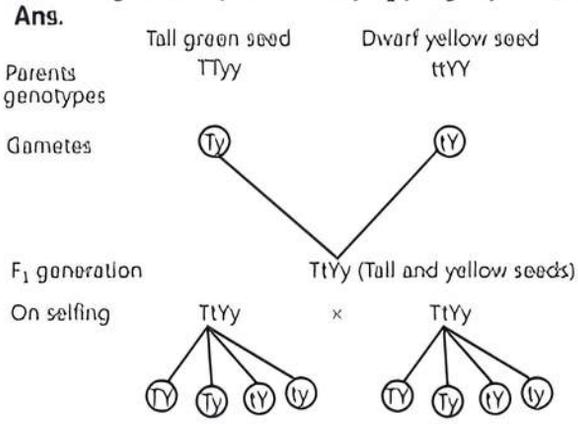
(i) Write the possible phenotype and genotype of F_1 generation.

(ii) State the laws of Mendel that are proved true by the F_1 generation.

(iii) Mention the F_2 phenotypic ratio along with their possible phenotypes.

(iv) Write the genotypes of the male and female gametes produced by F_1 progeny. *(CBSE 2023)*

Ans.



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, green 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

(i) Phenotype of F_1 – Tall plants with yellow seeds.

(ii) Genotype of F_1 – TtYy Law of dominance and Law of Independent Assortment.

(iii) Phenotypic ratio of F_2 generation:

9 :	3 :	3 :	1
Tall plant yellow seeds	Tall plant green seeds	Dwarf plant yellow seeds	Dwarf plants green seeds

(iv) The genotypes of male and female gametes produced by F_1 progeny will be Ty, Ty, tY, ty

Q 4. Describe the dihybrid cross carried on *Drosophila melanogaster* by Morgan and his group. How did they explain linkage, recombination and gene mapping on the basis of their observations?

Ans. Dihybrid cross on *Drosophila melanogaster*

(i) T.H. Morgan carried out several dihybrid crosses in *Drosophila* to study the genes that are sex-linked. He observed that when the two genes in a dihybrid

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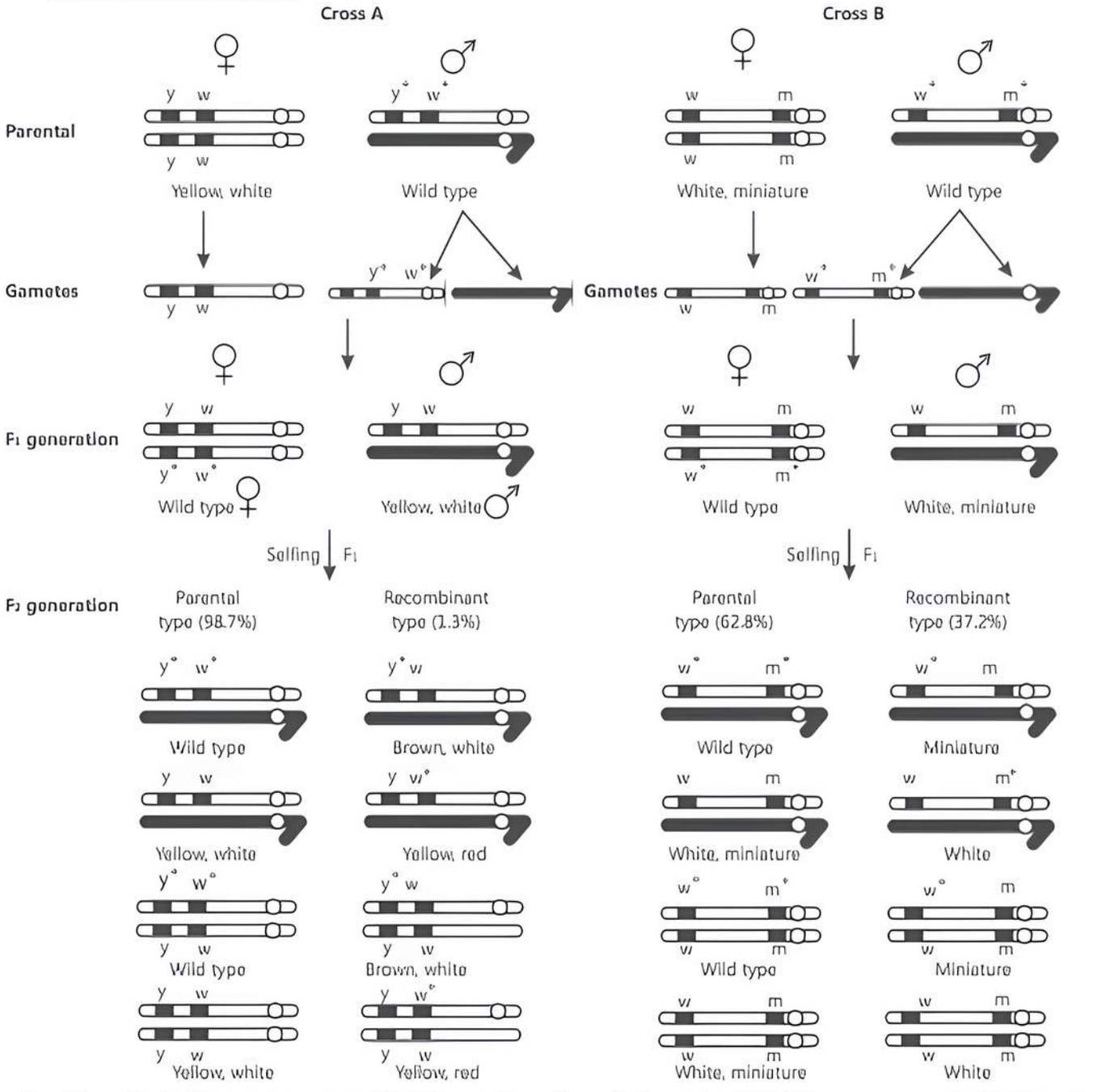
cross are located on the same chromosome, the proportion of parental gene combinations in the progeny was much higher than the non-parental or recombination of genes.

(ii) Morgan and his group found that when genes are grouped on the same chromosome, some genes are tightly linked or associated and show little recombination.



Results of two dihybrid crosses conducted by Morgan. Here, Cross A shows crossing between genes y and w; Cross B shows crossing between genes w and m. Here, dominant wild type alleles are represented with (+) sign in superscript.

(iii) When the genes are loosely linked, they show higher percentage of recombination.

(iv) Morgan hybridised yellow bodied and white eyed females with brown bodied and red eyed males (wild type) (cross-A) and inter-crossed their F_1 progeny.

(v) Alfred Sturtevant determined that genes of *Drosophila* are arranged in a linear order. He measured the distance between genes and prepared chromosome maps with the position of genes on the chromosomes based on percentage of recombinants. These are also called genetic maps.

Q 5. (i) Write the scientific name of the organism Thomas Hunt Morgan and his colleagues worked with for their experiments. Explain the correlation between linkage and recombination with respect to genes as studied by them.

(ii) How did Sturtevant explain gene mapping while working with Morgan? *(CBSE 2018)*

Ans. (i) The scientific name of the organism is *Drosophila melanogaster*.

Thomas Hunt Morgan and his colleagues observed that two genes (located closely on a chromosome) did not segregate independently of each other (F_2 ratio deviated significantly from 9 : 3 : 3 : 1).

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Tightly linked genes tend to show fewer (lesser) recombinant frequency of parental traits/show higher (more) frequency of parental type.
Loosely linked genes show higher percentage (more) of recombinant frequency of parental traits / lower frequency percentage of parental type.
Genes present on same chromosome are said to be linked and the recombinant frequency depends on their relative distance on the chromosome.

(ii) Sturtevant used the frequency of recombination between gene pairs on the same chromosome, as a measure of the distance between genes and mapped their position on the chromosome.

Q 6. Differentiate between incomplete dominance and co-dominance. Substantiate your answer with one example of each. *(CBSE 2019)*

OR

It is sometimes observed that the F_1 progeny has a phenotype that does not resemble either of the two parents and has intermediate phenotype. Explain by taking a suitable example and working out the cross upto F_2 progeny. *(CBSE 2023)*

OR

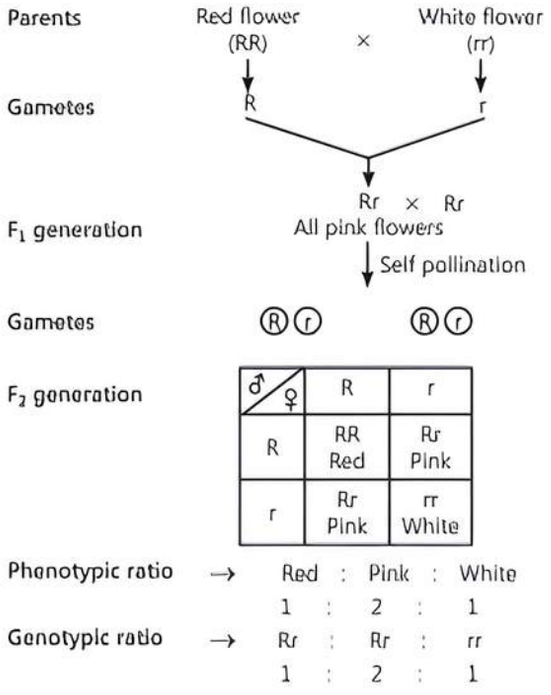
It is sometimes observed that the F_1 progeny shows a phenotype that resembles both the parents. Explain this type of inheritance using the example of A, B, O blood groups in humans.

(CBSE 2023)

Ans. Co-dominance is the phenomenon in which both the alleles of a contrasting character are expressed in the heterozygous condition. Both the alleles of a gene are equally dominant. ABO blood group in human beings is an example of I^A and I^0 co-dominance. The blood group character is controlled by three sets of alleles, namely I^A, I^B and i. The alleles, I^A and I^B, are equally dominant and are said to be co-dominant as they are expressed in the AB blood group. Both these alleles do not interfere with the expression of each other and produce their respective antigens. Hence, the AB blood group is an example of co-dominance.

ABO genotype in the offspring		ABO alleles inherited from the mother		
		A	B	O
ABO alleles inherited from the father	A	A	AB	A
	B	AB	B	B
	O	A	B	O

Incomplete dominance is a phenomenon in which one allele shows incomplete dominance over the other member of the allelic pair for a character.



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

COMMON ERROR

Students do not give explanation for both or forget to provide example in both of in complete dominance and co-dominance.

Q 7. In shorthorn cattle, the coat colours red or white are controlled by a single pair of alleles. A calf which receives the allele for red coat from its mother and the allele for white coat from its father is called a 'roan'. It has an equal number of red and white hairs in its coat.

(i) Is this an example of co-dominance or of incomplete dominance?

(ii) Give a reason for your answer.

(iii) With the help of genetic cross explain what will be the consequent phenotype of the calf when.

(a) red is dominant over white

(b) red is incompletely dominant.

(CBSE SQP 2023-24)

Ans. (i) This is an example of co-dominance.

(ii) Co-dominance is a condition in which two different alleles for a genetic trait are expressed. Individuals receive one version of a gene, called an allele, from each parent.

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(iii) (a) If pure breeding red coated cattles are represented as 'RR' and pure breeding white coated as 'rr'. If Red is dominant over White, a cross between 'RR' and 'rr' would produce red coated cattles (RR) and white coated cattle (rr) in the ratio of 3 :1.

(a) Parents: RR (Red) × rr (White)

Gametes: R r

	R	r
R	RR Red coat	Rr Red coat
r	Rr Red coat	rr White coat

F_1 generation - 3 : 1

(b) If the red and white coated cattles produce pink colour on a cross then, they exhibit incomplete dominance in the inheritance of coat colour due to which they produce pink coloured coat upon hybridisation.

If pure breeding red coated cattles are represented as 'RR' and pure breeding white coated as 'rr', then the pink coated cattles are 'Rr'.

A cross between 'RR' and 'rr' would produce pink coated cattles (Rr) and white coated cattle (rr) in the ratio of 1 : 2 : 1

Parents: RR (Red) × rr (White)

Gametes: R r

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

F_1 generation - 1 : 2 : 1

Q 8. Explain the chromosomal theory of inheritance.

Ans. Chromosomal Theory of Inheritance: The chromosomal theory of inheritance was proposed independently by Walter Sutton and Theodore Boveri in 1902. This theory has the following assumptions:

(i) Since the sperm and egg cells provide the only bridge from one generation to the other, all hereditary characters must be carried in them. The hereditary factors are carried in the nucleus.

(ii) Like the Mendelian alleles, chromosomes are also found in pairs.

(iii) The sperm and egg having haploid sets of chromosomes fuse to re-establish the diploid state.

(iv) The genes are located on the chromosomes in a linear order. As there are two chromosomes of each kind in somatic (diploid) cell, there must be two genes of each kind, one in each of the two homologous chromosomes.

(v) Homologous chromosomes synapse during meiosis and get separated to pass into different cells. This forms the basis for segregation and independent assortment. A gamete receives only one chromosome of each type and thus has only one gene for a trait. The paired condition is restored by fusion of gametes.

Q 9. Answer the following questions:

(i) Explain the mechanism of sex-determination in humans.

(ii) Differentiate between male heterogamety and female heterogamety with the help of an example of each.

Ans. (i) Sex Determination in Humans:

(a) Humans show XY type of sex determining mechanism.

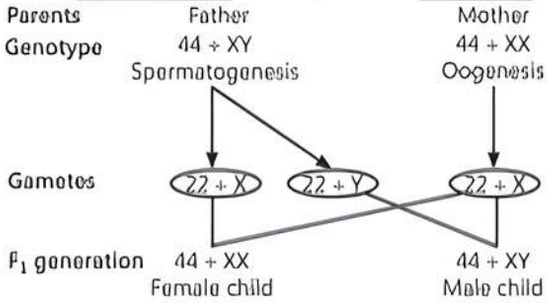
(b) Out of 23 pair of chromosomes, 22 are autosomes (same in both males and females). Females have a pair of X-chromosomes.

(c) Males have an X and a Y chromosome.

(d) During spermatogenesis, males produce two types of gametes with equal probability – sperm carrying either X or Y chromosome.

(e) During oogenesis, females produce only one type of gamete – having X chromosome.

(f) An ovum fertilised by the sperm carrying X chromosome develops into a female (XX) and an ovum fertilised by the sperm carrying Y chromosome develops into a male (XY).



(ii)

S.No.	Basis of difference	Male heterogamety	Female heterogamety
(i)	Number of gametes	Males produce two types of gametes.	Females produce two types of gametes.
(ii)	Example	Male human beings produce gametes of two types-X and Y.	Female birds produce gametes of two types-Z and W.

TiP

Understand the difference between male and female heterogamety and try to write in tabular form as far as possible.

Q 10. Thalassemia and haemophilia are both Mendelian disorders related to blood. Write the symptoms of the diseases. Explain with the help of crosses, the difference in the inheritance pattern of the two diseases.

OR

Why are thalassemia and haemophilia categorised as Mendelian disorders? Write the symptoms of these diseases. Explain their pattern of inheritance

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in humans. Write the genotypes of the normal parents producing a haemophilic son. *(CBSE 2015)*

Ans. Both thalassemia and haemophilia are caused due to alteration or mutation in a single gene and follow Mendelian pattern of inheritance. So, they are categorised as Mendelian disorders.

Symptoms:

Thalassemia: Anaemia (caused due to defective/abnormal Hb).

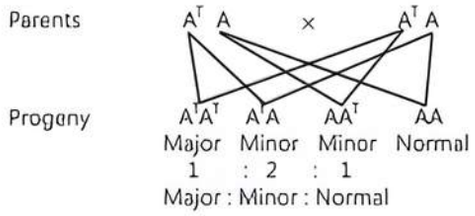
Haemophilia: Non-stop bleeding even in minor injury.

Pattern of inheritance:

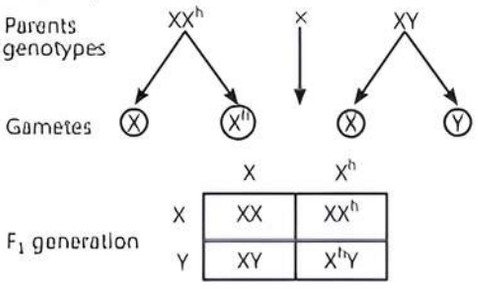
Thalassemia: Autosomal recessive inheritance pattern inherited from heterozygous/parent carrier.

Haemophilia: X-linked recessive inheritance inherited from a haemophilic father/carrier mother (females are rarely haemophilic).

Cross for Thalassemia:



Cross for Haemophilia:



Chapter Test

Multiple Choice Questions

Q 1. The allele which expresses itself in both homozygous and heterozygous condition is called:

a. dominant allele
b. recessive allele
c. incomplete dominant allele
d. inherited allele

Q 2. To determine in genotype of a tall plant of F_2 generation, Mendel crossed this plant with a dwarf plant. This cross represents a:

a. test cross
b. monohybrid cross
c. reciprocal cross
d. dihybrid cross

Q 3. ..... is an example of X-linked recessive trait.

a. Phenylketonuria
b. Haemophilia
c. Cystic fibrosis
d. Sickle-cell anaemia

Assertion and Reason Type Questions

Directions (Q.Nos. 4-5): *Each of the following questions consists of two statements, one is Assertion (A) and the other is Reason (R). Select the correct answer to these questions from the codes a, b, c and d as given below.*

a. Both Assertion and Reason are true and Reason is the correct explanation of Assertion.
b. Both Assertion and Reason are true but Reason is not the correct explanation of Assertion.
c. Assertion is true but Reason is false.
d. Both Assertion and Reason are false.

Q. 4. Assertion (A): The law of independent assortment can be studied by means of dihybrid cross.

Reason (R): The law of independent assortment is applicable only to linked genes.

Q. 5. Assertion (A): Mendelian disorders are transmitted to offspring on the same lines as in the principles of inheritance.

Reason (R): The pattern of inheritance of Mendelian disorders cannot be traced in a family by the pedigree analysis.

Case Based Questions

Case Study 1

Q 6. Mendel is known as the 'father of genetics' because of his ground breaking work on inheritance in pea plants 150 years ago. At the age of 21 Mendel while working in a monastery in Brunn (now in the Czech Republic) began a series in experiments in their botanical garden. He found out how traits are passed from one generation to another, *i.e.*, inheritance. He studied this in peas (*Pisum sativum*), because they are easy to grow and can be shown each year.

(i) Which one from those given below is the period of Mendel's hybridisation experiments?

a. 1856-1863 b. 1840-1850
c. 1857-1869 d. 1870-1877

(ii) How many pairs of true breeding varieties were selected by Mendel for his experiment on pea plant?

a. 12 b. 13 c. 14 d. 15

(iii) Among the following characters which one was not considered by Mendel in his experiments on pea?

a. Stem–Tall or Dwarf
b. Trichomes–Glandular or Non-glandular
c. Seed–Green or Yellow
d. Pod–Inflated or Constricted

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(iv) **Which is correct about traits chosen by Mendel for his experiments on pea plant?**

a. Terminal pod was dominant
b. Constricted pod was dominant
c. Green coloured pod was dominant
d. Tall plants were recessive

Case Study 2

Q 7. In a plant species that follows Mendelian inheritance yellow flower colour is dominant over white and round fruit shape is dominant over elongated. Crossing was performed between two pure lines-one having yellow-flower and round fruit and another with white flower and elongated fruits. About 20 plants survived in F_1 progeny. Plants of F_1 were allowed to self fertilise and about 960 plants survived in F_2.

Read the given passage carefully and give the answer of the following questions:

(i) **How many plants would have yellow-flower and round fruit in F_1 generation?**

(ii) **How many plants would have yellow-flower and round fruit in F_2 generation?**

(iii) **What is the ratio of phenotype and genotype when plant heterozygous for yellow-flower and round fruit is back crossed with the double recessive parent?**

(iv) **When the plant heterozygous for yellow-flower and round fruit are self crossed, then the plant with yellow-flower and elongated fruit will be represented by which genotype?**

Very Short Answer Type Questions

Q 8. **Pea flowers produce assured seed sets. Give a reason.**

Q 9. **How would you find the genotype of an organism exhibiting a dominant phenotype trait?**

Q 10. **Name one autosomal dominant and one autosomal recessive Mendelian disorder in humans.**

Short Answer Type Questions

Q 11. **Mention the advantages of selecting pea plant for experiment by Mendel.**

Q 12. **Write the chromosomal constitution and the resulting sex in each of the following syndrome.**
(i) Turner's syndrome
(ii) Klinefelter's syndrome.

Long Answer Type-I Question

Q 13. **Name a disorder, give the karyotype and write the symptoms, where a human male suffers as a result of an additional X-chromosome.**

Long Answer Type-II Question

Q 14. **Why the common Mendelian experiments cannot be carried out in human beings?**

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