

Principles of Inheritance and Variation

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.

Q1. Genes with multiple phenotypic effects are known as:

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

Q2. Which of the following disorder is an example of genes with multiple phenotypic effects?

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

Q3. 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.

Q4. 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

Q5. 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).

Q1. 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.

Q2. 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.

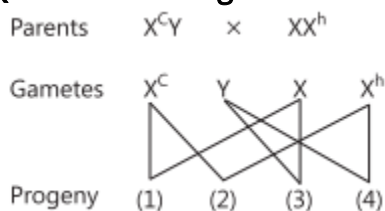
Q3. Which of the following statements is incorrect regarding haemophilia?

- 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.

Q4. 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%

Q5. 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)

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:

Q1. 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.

Q2. 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 test cross.

Q3. 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: (CBSE 2023)

Q1. State what is aneuploidy.

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.

Q2. 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. $22+0$, $22+XY$.

Q3. 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:

Q1. Which trait are of attached and free earlobe respective examples?

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

Q2. 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.

Q3. 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%.

Solutions for Questions 6 to 15 are Given Below

Case Study 6

Read the following and answer any four questions from 1(i) to 1(v) given below:

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.

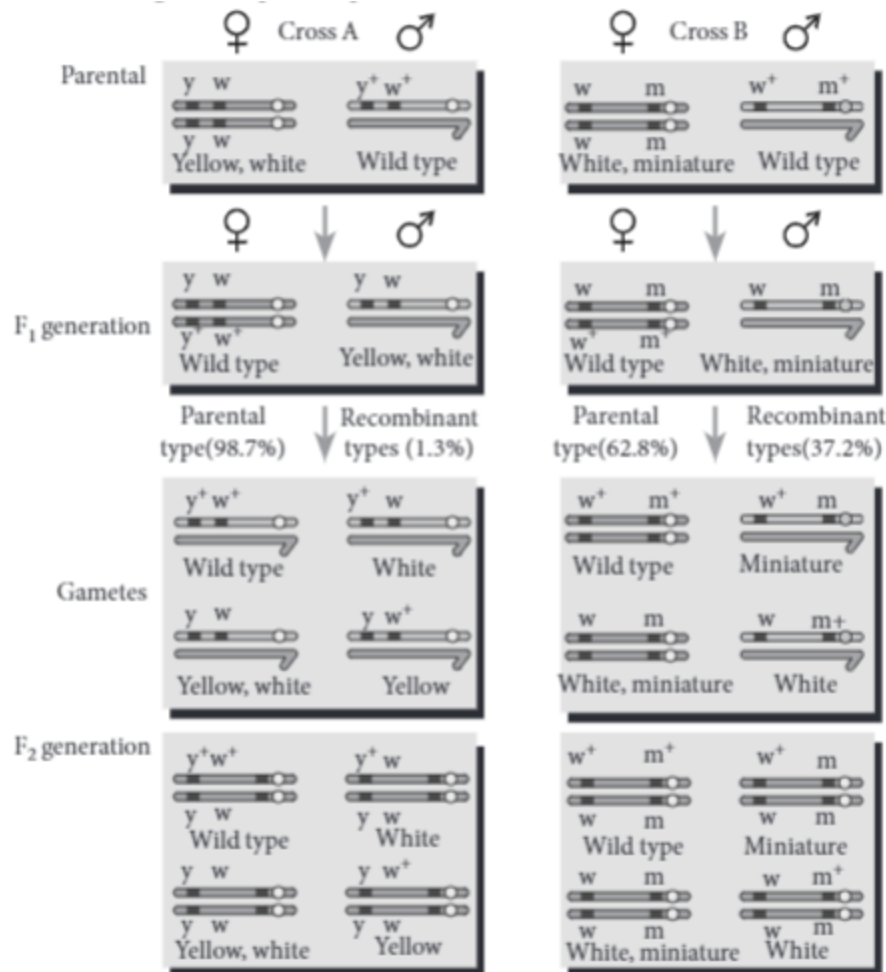
- (i) Which of the following cannot be derived from the crosses done by Prashant?
- (a) Mendel's law of segregation
 - (b) Mendel's law of dominance
 - (c) Mendel's law of independent assortment
 - (d) Both (a) and (c)
- (ii) To determine the genotype of a purple flowered plant, Prashant crossed this plant with a white flowered plant. This cross represents a
- (a) test cross
 - (b) dihybrid cross
 - (c) reciprocal cross
 - (d) trihybrid cross.
- (iii) In white flowered plant, allele is expressed in
- (a) heterozygous condition only
 - (b) homozygous condition only
 - (c) F_3 generation
 - (d) both homozygous and heterozygous condition.
- (iv) The character, *i.e.*, purple colour of the flowers that appeared in the first filial generation is called
- (a) recessive character
 - (b) dominant character
 - (c) holandric character
 - (d) lethal character.
- (v) **Assertion :** A geneticist crossed two plants and he obtained 50% purple flowered plants and 50% white flowered plants.
- Reasons :** Purple coloured flower plant might be heterozygous.
- (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.

Case Study 7

Read the following and answer any four questions from 2(i) to 2(v) given below:

During a study of inheritance of two genes, teacher asked students to perform an experiment. The students crossed white eyed, yellow bodied female *Drosophila* with a red eyed, brown bodied male *Drosophila* (i.e., wild). They observed that progenies in F₂ generation had 1.3 percent recombinants and 98.7 percent parental type combinations. The experimental cross with results is shown in the given figure. [Note: Dominant wild type alleles are represented with (+) sign in superscript.]



- (i) By conducting the given experiment, teacher can conclude that
- Genes for eye colour and body colour are linked
 - Genes for eye colour and body colour show complete linkage
 - Linked gene remain together and are inherited
- (a) A and B only (b) B only (c) A and C only (d) A, B and C
- (ii) Teacher asked to conduct an experiment on *Drosophila* because
- the male and female flies are easily distinguishable
 - it completes its life cycle in about two weeks
 - a single mating could produce a large number of progeny flies
 - all of these.

- (iii) Genes white eyed and yellow bodied located very close to one another on the same chromosome tend to be transmitted together are called
 (a) allelomorphs (b) identical genes (c) linked genes (d) recessive genes.
- (iv) Select the correct statement regarding the given experiment.
 (a) The physical distance between two genes determines strength of linkage.
 (b) The physical distance between two genes determines frequency of crossing over.
 (c) The two linked genes always segregate independently of each other.
 (d) Both (a) and (b)
- (v) **Assertion :** When yellow bodied, white eyed *Drosophila* females were hybridised with brown-bodied, red eyed males; and F_1 progeny was intercrossed, F_2 ratio deviated from 9 : 3 : 3 : 1.
Reason : When two genes in a dihybrid are on the same chromosome, the proportion of parental gene combinations are much higher than the non-parental type.
 (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.

Case Study 8

Read the following and answer any four questions from 3(i) to 3(v) given below:

Turner's syndrome is an example of monosomy. It is formed by the union of an allosome free egg and a normal 'X' containing sperm or a normal egg and an allosome free sperm. The individual has $2n = 45$ chromosomes ($44 + X0$) instead of 46. Such individuals are sterile females who have rudimentary ovaries, under developed breasts, small uterus, short stature, webbed neck and abnormal intelligence. They may not menstruate or ovulate. This disorder can be treated by giving female sex hormone to the women from the age of puberty to make them develop breasts and have menstruation. This makes them feel more normal.

- (i) Number of barr body present in a female with Turner's syndrome is
 (a) 0 (b) 1 (c) 2 (d) < 2.
- (ii) Turner's syndrome is an example of
 (a) aneuploidy (b) euploidy
 (c) polyploidy (d) autosomal abnormality.
- (iii) Turner's syndrome is a/an
 (a) autosomal recessive Mendelian disorder (b) autosomal dominant Mendelian disorder
 (c) sex linked Mendelian disorder (d) chromosomal disorder.
- (iv) Which of the following statements regarding Turner's syndrome is incorrect?
 (a) It is a case of monosomy of chromosomes.
 (b) The suffering individual is a sterile female having one 'X' chromosome missing in the cells.
 (c) The problem is due to an extra chromosome.
 (d) The individual are of short stature.
- (v) **Assertion :** Turner's syndrome is caused due to absence of any one of the X and Y sex chromosome.
Reason : Individuals suffering from Turner's syndrome show masculine as well as feminine development.
 (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.

Case Study 9

Read the following and answer any four questions from 4(i) to 4(v) given below:

According to Mendel, one gene control 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 of two or more characters can be seen in cotton. In cotton, a gene for the lint also influences the height of plant, size of the ball, number of ovules and viability of seeds.

- (i) Genes with multiple phenotypic effects are known as
 - (a) hydrostatic genes
 - (b) duplicate genes
 - (c) pleiotropic genes
 - (d) complimentary genes.
- (ii) 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)
- (iii) 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
- (iv) 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 these
- (v) **Assertion :** 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 : 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) Both assertion and reason are false.

Case Study 10

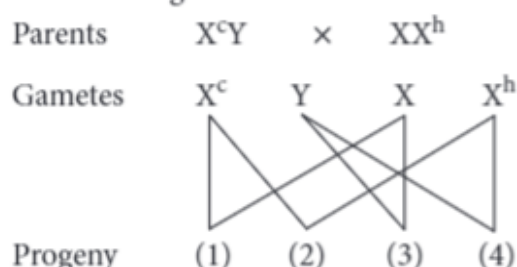
Read the following and answer any four questions from 5(i) to 5(v) given below:

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 colour blind can, lead a normal life, reading, writing and driving (distinguishing traffic lights by their position).

- (i) 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, all daughters are carriers.

- (ii) 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.]
- 25% female progenies carry the gene for both haemophilia and colourblindness.
 - 25% male progenies carry only the gene of colourblindness.
 - 25% female progenies carry only the gene of colourblindness.
 - 25% male progenies and 25% female progenies carry the gene of haemophilia.
- (iii) Which of the following statements is incorrect regarding haemophilia?
- It is a dominant disease.
 - A single protein involved in clotting of blood is affected.
 - It is recessive disease.
 - It is Mendelian disorder.
- (iv) Anup is having colourblindness and is married to Soni who is normal. What is the chance that their son will have the disease?
- 100%
 - 50%
 - 25%
 - 0%

(v) Refer to the given cross.



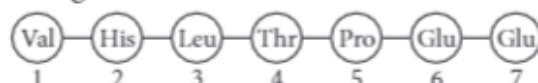
Select the correct option regarding 1, 2, 3 and 4.

- | | |
|---------------------------------------|-----------------------------------|
| (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 |

Case Study 11

Read the following and answer any four questions from 6(i) to 6(v) given below:

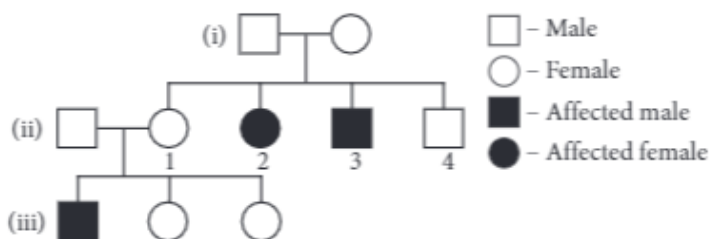
A relevant portion of β -chain of haemoglobin of a normal human is as follows



The codon for the sixth amino acid is GAG. The sixth codon GAG mutates to GAA as a result of mutation X and into GUG as a result of mutation Y.

- (i) Which of the following is incorrect statement?
- Mutation X carries no change in shape of red blood cells.
 - Mutation Y causes change in shape of red blood cell shape.
 - Both mutations X and Y causes change in shape of red blood cell shape.
 - Both (a) and (b)

- (ii) Due to mutation Y the shape of RBCs under oxygen tension will be
 (a) biconcave disc like (b) elongated and curve
 (c) circular (d) spherical.
- (iii) GUG is code for
 (a) valine (b) proline
 (c) glutamic acid (d) leucine.
- (iv) Which of the following genotype shows diseased phenotype due to mutation Y?
 (a) $Hb^S Hb^S$ (b) $Hb^A Hb^S$
 (c) $Hb^A Hb^A$ (d) Both (a) and (b)
- (v) Study the given pedigree chart for sickle-cell anaemia and select the most appropriate option for the genotypes.



- | Genotypes of parents | Genotypes of 1 st and 3 rd child in F ₁ |
|----------------------------|--|
| (a) $Hb^A Hb^S, Hb^A Hb^A$ | $Hb^A Hb^A, Hb^A Hb^S$ |
| (b) $Hb^A Hb^S, Hb^A Hb^S$ | $Hb^A Hb^A, Hb^A Hb^A$ |
| (c) $Hb^A Hb^A, Hb^A Hb^S$ | $Hb^A Hb^A, Hb^S Hb^S$ |
| (d) $Hb^A Hb^S, Hb^A Hb^S$ | $Hb^A Hb^S, Hb^S Hb^S$ |

Case Study 12

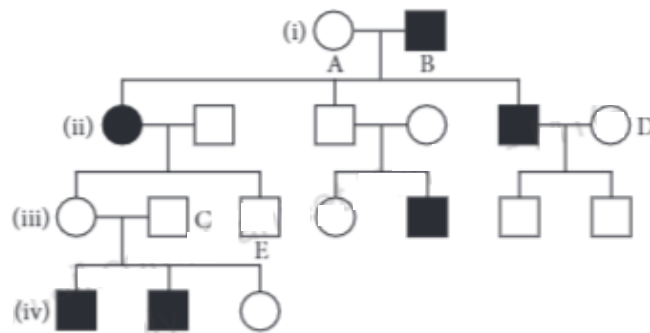
Read the following and answer any four questions from 7(i) to 7(v) given below:

Study the two cases carefully regarding the pattern of inheritance of disease.

Case	Mother	Father	Children
Case I	With disease	Normal	Sons always with diseases
Case II	With disease	Normal	Sons and daughters could show disease

- (i) Which of the following diseases is not an example of case I?
 (a) Haemophilia (b) Colour blindness
 (c) Thalassemia (d) Night blindness
- (ii) Which of the following is impossible for a case I?
 (a) Carrier mother to pass the gene to her son (b) Diseased father to pass the gene to his daughter
 (c) Diseased father to pass the gene to his son (d) Carrier mother to pass the gene to her daughter
- (iii) If inheritance pattern of disease is as case II and both parents are carrier of disease what are the chances of pregnancy resulting in an affected child?
 (a) 25% (b) 100% (c) 0% (d) 50%
- (iv) Disease that follows inheritance pattern as case I is linked with
 (a) autosomes (b) X chromosome
 (c) Y chromosome (d) chromosome 21.

(v) Study the given pedigree chart showing the inheritance pattern as case II.



What will be the genotype of individuals A, B, C, D and E respectively?

- (a) Aa, aa, aa, AA, aa (b) Aa, aa, Aa, AA, Aa
(c) Aa, Aa, aa, AA, AA (d) Aa, AA, Aa, Aa, aa

Case Study 13

Read the following and answer any four questions from 8(i) to 8(v) given below:

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 .

- (i) How many plants would have yellow flower and round fruit in F_1 generation?
(a) 20 (b) 10 (c) 5 (d) 0
- (ii) How many plants would have yellow flower and round fruit in F_2 generation?
(a) 960 (b) 540 (c) 180 (d) 60
- (iii) Which of the following is correct for the condition when plant heterozygous for yellow flower and round fruit is back crossed with the double recessive parent?
(a) 9 : 3 : 3 : 1 ratio of phenotype only (b) 9 : 3 : 3 : 1 ratio of genotype only
(c) 1 : 1 : 1 : 1 ratio of phenotype only (d) 1 : 1 : 1 : 1 ratio of phenotype and genotype
- (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 the genotype
(a) YyRr, YyRR, YYRr (b) Yyrr, YYrr, yyrr
(c) yyRr, yyRR (d) Yyrr, YYrr.
- (v) The given Punnett's square represents the pattern of inheritance in a dihybrid cross where yellow flower and round fruit (R) condition is dominant over white flower (y) and elongated fruit (r) condition.

♀ \ ♂	YR	Yr	yR	yr
YR	A	E	I	M
Yr	B	F	J	N
yR	C	G	K	D
yr	D	H	L	P

Plant 'C' will produce fruits with the genotype identical to fruits produced by the plant of

- (a) type H (b) type E (c) type K (d) type I.

Case Study 14

Read the following and answer any four questions from 9(i) to 9(v) given below:

ABO blood groups in human beings are controlled by the gene I . The gene I has three alleles. I^A , I^B and i . Since there are three different alleles six different genotypes are possible. If two persons with 'AB' blood group marry and have sufficient large number of children, their children could be classified as 'A' blood group : 'AB' blood group : B blood group in 1 : 2 : 1 ratio. Modern technique of protein electrophoresis reveals presence of both 'A' and 'B' type proteins in 'AB' blood group individuals.

- (i) How many types of phenotypes can occur in ABO blood group?
 (a) Six (b) Two (c) Three (d) Four
- (ii) ABO blood grouping in human beings cites the example of
 (a) incomplete dominance (b) co-dominance (c) multiple allelism (d) both (b) and (c).
- (iii) The presence of both 'A' and 'B' type proteins in 'AB' blood group individuals is an example of
 (a) partial dominance (b) incomplete dominance
 (c) co-dominance (d) complete dominance.
- (iv) If a man of A blood group marries a woman of AB blood group. Which type of progeny indicates that man is heterozygous?
 (a) O (b) B (c) A (d) AB
- (v) Complete the given table regarding different possibilities and their corresponding blood groups.

Genotypes	Blood groups
$I^A I^B$	(I)
$I^B i$, (II)	B
(III)	O
$I^A I^A$, (IV)	I^A

(I)	(II)	(III)	(IV)
(a) O	$I^B I^B$	$I^B i$	$I^A i$
(b) AB	$I^A i$	$I^A I^B$	$I B i$
(c) AB	$I^B I^B$	ii	$I^A i$
(d) O	$I^A I^A$	ii	$I^A i$

Case Study 15

Read the following and answer any four questions from 10(i) to 10(v) given below:

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 pinnae.

- (i) Attached and free earlobe are respective example of
 (a) dominant and recessive traits (b) recessive and recessive traits
 (c) recessive and dominant traits (d) dominant and dominant traits.

- (ii) Hypertrichosis of the ear is
- X linked trait
 - Y linked trait
 - autosomal dominant trait
 - autosomal recessive trait.
- (iii) If a female with attached earlobe married a male homozygous for free earlobe sparse hair on pinna then what would be the chances of daughter to have attached earlobe?
- 0%
 - 100%
 - 25%
 - 75%
- (iv) If a man with attached earlobe and hairy pinna married a woman with attached earlobe then what would be the chances of son to have hairy pinna?
- 50%
 - 100%
 - 75%
 - 0%
- (v) A male with attached earlobe, sparse hair on pinna married a female with attached earlobe. Which of the following is correct regarding their progenies?
- All sons have a free earlobe with hairy pinna.
 - All daughters have an attached earlobe.
 - 50% daughters have an attached earlobe whereas 50% daughters have a free earlobe.
 - 50% sons have attached earlobe with hairy pinna and 50% sons have a free earlobe.

HINTS & EXPLANATIONS

6. (i) (c) : Mendel's law of independent assortment states 'when two pairs of traits are combined in a hybrid, segregation of one pair of characters is independent of the other pair of characters'. This law can be derived by dihybrid cross but Prashant has performed monohybrid cross only, *i.e.*, one pair of traits.

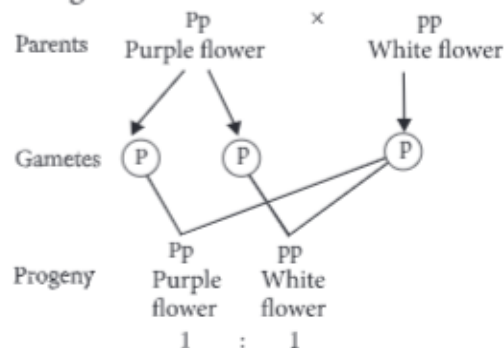
(ii) (a) : In a test cross, an organism (pea plant) showing a dominant phenotype whose genotype is to be determined is crossed with the recessive parent instead of self-crossing. The progenies of such a cross can easily be analysed to predict the genotype of the test organism. Normal test cross ratio for a monohybrid cross is 1 : 1 and for a dihybrid cross is 1 : 1 : 1 : 1.

(iii) (b) : The factor of an allelic or allelomorphic pair which is unable to express its effect in the presence of its contrasting factor in a heterozygote is called recessive factor or allele, *e.g.*, the allele 't' in hybrid tall pea plant Tt. The effect of recessive factor becomes known only

when it is present in the pure or homozygous state, *e.g.*, tt in dwarf pea plant.

(iv) (b) : In first filial generation or heterozygous individuals, out of the two factors or alleles representing the alternate traits of a character, one is dominant and expresses itself in the hybrid or F_1 generation. The other factor or allele is recessive and does not show its effect in the heterozygous individual.

(v) (a) : The given cross can be illustrated as follows:



or 50% purple flowered plant, 50% white flowered plant.

7. (i) (c) : By conducting the given cross teacher can conclude that the genes for eye colour and body colour are linked. Thus these genes were very tightly linked and showed very low recombination.

(ii) (d)

(iii) (c) : Genes located very close to one another on the same chromosome tend to be transmitted together and are called linked genes.

(iv) (a) : The physical distance between two genes determines both the strength of the linkage and the frequency of the crossing over between two genes. The strength of the linkage increases with the closeness of the two genes. On the other hand the frequency of crossing over increases with the increase in the physical distance between the two genes.

(v) (a) : In *Drosophila*, the genes for body and eye colour are located on X chromosome. When two genes in a dihybrid cross are situated on the same chromosome, the proportion of parental gene combination are higher than non-parental type. This occurs due to physical association or linkage of the two genes while non-parental gene combinations due to recombination between two genes. Thus, linkage and recombination deviates the ratio from Mendelian ratio of a dihybrid cross (9 : 3 : 3 : 1).

8. (i) (a) : Barr body is a structure consisting of a condensed X chromosome that is found in non-dividing nuclei of female mammals. The presence of Barr body is used to confirm the sex of athletes in sex determination tests. It is named after the Canadian anatomist M.L. Barr, who identified it. The number of Barr bodies is one less than total number of X chromosomes. In Turner's syndrome genotype is $45 + X0$, so, the number of Barr body is 0.

(ii) (a) : Failure of segregation of chromatids during cell division result in the gain or loss of a chromosomes called aneuploidy. For example, Turner's syndrome results due to loss of X chromosome in human females.

(iii) (d) : Turner's syndrome is a chromosomal disorder that occurs due to absence of one chromosome.

(iv) (c) : In Turner's syndrome individual lacks one X chromosome. This situation is known as monosomy.

(v) (d) : Turner's syndrome occurs due to absence of X chromosome. Individuals having a single X chromosome $22A+X0$ (45) have female sexual differentiation but ovaries are rudimentary. Other associated phenotypes of this condition are short

stature, webbed-neck, broad chest, lack of secondary sexual characteristics and sterility. Thus, any imbalance in the copies of the sex chromosomes may disrupt the genetic information necessary for normal sexual development.

9. (i) (c)

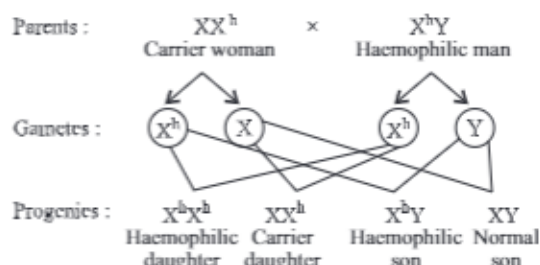
(ii) (d) : The ability of a gene to have multiple phenotypic effects because it influences a number of characters simultaneously is known as pleiotropy. In human beings pleiotropy is exhibited by syndromes, i.e., sickle cell anaemia and phenylketonuria.

(iii) (a) : Kernel colour in wheat, height in human beings and skin colour in human beings are examples of polygenic inheritance, i.e., inheritance controlled by three or more genes. In *Drosophila*, white eye mutation pleiotropic effect, it causes depigmentation in many part of the body.

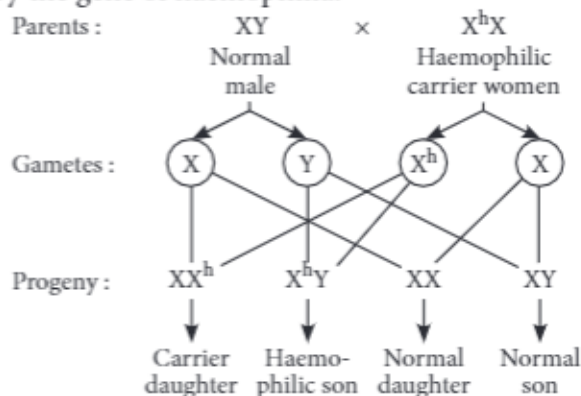
(iv) (d)

(v) (a)

10. (i) (a) : When a haemophilic man (X^hY) marries a woman whose father was haemophilic and mother was normal i.e., carrier woman (XX^h), then 50% daughters are carriers and 50% are haemophilic. This can be explained as follows :

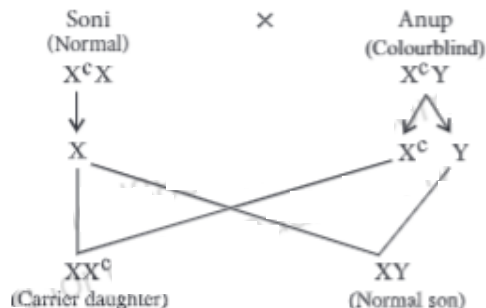


(ii) (d) : When a man whose father was colourblind and mother was normal (i.e., normal man XY) marries a woman whose father was haemophilic and mother was normal (i.e., carrier haemophilic woman X^hX), then 25% male progenies and 25% female progenies carry the gene of haemophilia.



(iii) (a) : Haemophilia is sex linked recessive Mendelian disorder.

(iv) (d) : When Anup who is colourblind (X^cY) marries Soni who is normal (XX) then 0% chances that their son will have colourblindness.



(v) (a)

11. (i) (c) : Due to mutation X, GAG mutates to GAA. But both GAG and GAA code for glutamic acid and hence there is no change in shape of RBC whereas in mutation Y, GUG is substituted by GAA that codes for valine and so the RBCs become sickle shaped.

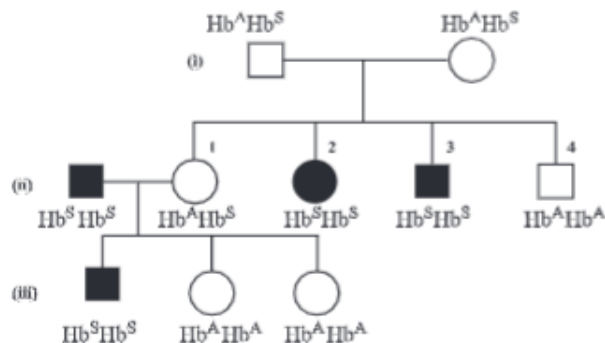
(ii) (b) : Mutation Y causes sickle cell anaemia and the mutant haemoglobin molecule undergoes polymerisation under low oxygen tension causing the change in the shape of RBC from biconcave disc to elongated sickle cell like.

(iii) (a) : Refer to answer (i).

(iv) (a) : Mutation Y causes sickle cell anaemia that is controlled by a single pair of allele, Hb^A and Hb^S . Out of three possible genotypes only homozygous individuals for Hb^S ($Hb^S Hb^S$) show the diseased

phenotype.

(v) (d) : Given pedigree chart for sickle-cell anaemia can be illustrated as :



12. (i) (c) : Thalassaemia is autosomal recessive disease. Case I represents sex linked recessive disease.

(ii) (c)

(iii) (a) : Case II shows autosomal inheritance. If both the parents are carrier, then it must be a case of autosomal recessive inheritance where the chances of having a disease child is 25% as shown :

	♀ Aa	♂ Aa
	x	
	A	a
A	AA Normal	Aa Carrier
a	Aa Carrier	aa Affected

(iv) (b)

(v) (b) : Autosomal recessive traits are the traits which are caused by recessive autosomal genes when present in homozygous condition. The given pedigree can be explained as:



As the trait appears only in homozygous recessive individuals (aa), therefore it is an autosomal recessive trait.

13. (i) (a) : In F_1 generation, all the 20 plants would be heterozygous for the trait and thus they would possess yellow flower and round fruit.

(ii) (b) : When heterozygous plants in F_1 generation undergo selfing, F_2 progeny gives 9 : 3 : 3 : 1 phenotypic ratio. Thus, in the given case, yellow flower with round

fruit are $960 \times \frac{9}{16} = 540$.

(iii) (d) : When heterozygous for yellow flower and round fruit is back crossed with double recessive parent then genotypic and phenotypic ratio will be 1 : 1 : 1 : 1.

Parents :	YrRr	x	yyrr
Progenies :	♀ YR	♂ yr	
	Yr		
	yR		
	yr		
	YrRr		Yellow round
	Yyrr		Yellow elongated
	yyRr		White elongated
	yyrr		White elongated

Phenotypic ratio = 1 yellow round : 1 yellow elongated : 1 white round : 1 white elongated

Genotypic ratio = $YyRr : Yyrr : yyRr : yyrr$

(iv) (d) : When plant YyRr is self pollinated, 9 : 3 : 3 : 1 ratio of phenotype will be observed. This can be explained as follows :

Parents : YyRr × YyRr

Progenies :

♀ \ ♂	YR	Yr	yR	yr
YR	YYRR Yellow round	YYRr Yellow round	YyRR Yellow round	YyRr Yellow round
Yr	YYRr Yellow round	YYrr Yellow elongated	YyRr Yellow round	Yyrr Yellow elongated
yR	YyRR Yellow round	YyRr Yellow round	yyRR White round	yyRr White round
yr	YyRr Yellow round	Yyrr Yellow elongated	yyRr White round	yyrr White elongated

Phenotypic ratio = 9 yellow and round : 3 yellow and elongated : 3 white and round : 1 white and elongated.

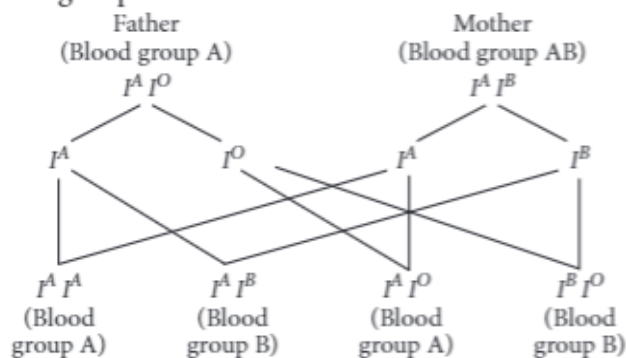
(v) (d) : Plant 'C' is formed by fusion of gametes yR and YR and hence has the genotype YyRR. Plant I is formed by fusion of gametes YR and yR and hence will have the same genotype as plant I, i.e., YyRR.

14. (i) (d) : In human beings ABO blood groups are controlled by gene *I* which has three alleles I^A , I^B and *i*. The six possible genotypes are $I^A I^A$, $I^A i$, $I^B I^B$ and *ii*. The phenotype which occur by these genotypes are A ($I^A I^A$, $I^A i$), B ($I^B I^B$, $I^B i$), AB ($I^A I^B$) and O (*ii*).

(ii) (d) : ABO blood grouping in human beings is an example of co-dominance and multiple allelism. Co-dominance is a phenomenon in which alleles do not show dominance-recessive relationship and are able to express themselves independently when present together. More than two alternate forms of a gene present on the same locus are called multiple alleles and the mode of inheritance in these alleles is called multiple allelism. Human beings have six genotypes and four blood group phenotypes - A, B, AB and O.

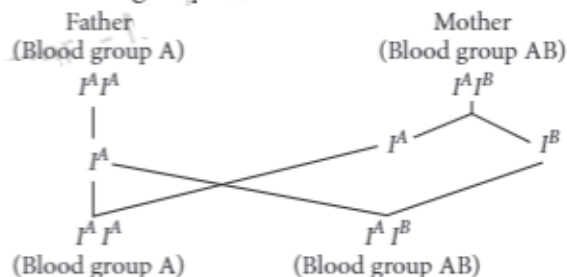
(iii) (c)

(iv) (b) : If a man with blood group A (genotype $I^A I^O$) marries a woman having blood group AB (genotype $I^A I^B$), then their offspring would have AB, B and A blood groups.



Presence of B blood group in a child is indicative that the man is heterozygous.

If the male is homozygous, then the offspring will contain blood group A, B and A.

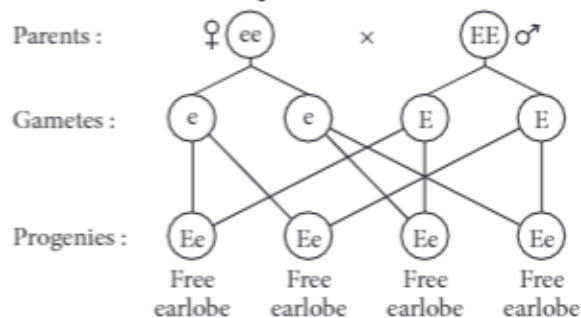


(v) (c)

15. (i) (c) : In humans, free earlobes is dominant over attached earlobes.

(ii) (b)

(iii) (a) : If a female with attached earlobes (*ee*) married a male with free lobe (*EE*) and sparse hair on pinna then chance of any progeny to have attached ear lobe is zero. It can be depicted as follows:



(iv) (b) : If a man with attached earlobe (*ee*) and hairy pinna married a woman with attached earlobes (*ee*) then 100% chances of sons to have hairy pinna as hypertrichosis or hairy pinna is Y linked feature.

(v) (b) : If a male with attached earlobe sparse hair on pinna married a female with attached earlobe then all daughters have an attached earlobe.

