UNIT-VII

Genetics and Evolution

CHAPTER 5

PRINCIPLES OF INHERITANCE AND VARIATION

Syllabus

➢ ***Heredity and variation :*** *Mendelian inheritance; deviations from Mendelism - incomplete dominance, co-dominance, multiple alleles and inheritance of blood groups, pleiotropy; elementary idea of polygenic inheritance; chromosomal theory of inheritance; chromosomes and genes; Sex determination - in humans, birds and honey bee; linkage and crossing over; sex linked inheritance - haemophilia, colour blindness; Mendelian disorders in humans - thalassemia; chromosomal disorders in humans; Down's syndrome, Turner's and Klinefelter's syndromes.*

Chapter Analysis

List of Topics		2016		2017		2018
		D	OD	D	OD	D/OD
Mendel's Law of Inheritance	• Difference between gene and allele	1 Q (1 M)				
Inheritance of one gene	• Test cross • Polygenic inheritance • Pleiotropy • Mendelian pattern of inheritance	1 Q (3 M)	1 Q (3 M) 1 Q (5 M)		1 Q (1 M)	
Inheritance of two gene	• Law of Independent Inheritance • Morgan's dihybrid cross			1 Q (5 M)		1 Q (5 M)
Sex determination	• Sex determination in honey bees, birds and humans • Fate of autosomes during gamete formation		1 Q (1 M)	1 Q (1 M)		1 Q (3 M)
Genetic disorders	• Pattern of inheritance of autosomal recessive trait • Haemophilia, Thalassemia • Down's syndrome			1 Q (3 M)	1 Q (3 M)	

- *On the basis of above analysis, it can be concluded that this is also an important chapter from the exam point of view. A major 5 marks long question is always asked from this chapter. The important topics for long question answers are Mendel's three laws, Morgan's dihybrid crosses, Polygenic, pleiotropy and Mendelian pattern of inheritance. Other important topics from exam point of view are Sex determination in human, birds and honey bees, test cross, and genetic disorders (Hemophilia, Thalassemia, Down's syndrome).*

TOPIC-1
Mendelian Laws of Inheritance and Chromosomal Theory of Inheritance

Revision Notes

Mendel's Laws of Inheritance :

➢ **Hybridization Experiments on Garden Peas (*Pisum sativum*)**

- Mendel selected 7 pairs of true breeding pea varieties.

S. No.	Characters	Dominant	Recessive
1.	**Height of the stem**	Tall (T)	Dwarf (t)
2.	**Colour of the flower**	Violet/Red (R)	White (r)
3.	**Position of the flower**	Axial (A)	Terminal (a)
4.	**Shape of pod**	Full/Inflated (I)	Constricted (i)
5.	**Colour of pod**	Green (G)	Yellow (g)
6.	**Shape of seed**	Round (R)	Wrinkled (r)
7.	**Colour of seed/cotyledons**	Yellow (Y)	Green (y)

Inheritance of One Gene

TOPIC - 1
Mendelian Laws of Inheritance and Chromosomal Theory of Inheritance .... **P. 85**

TOPIC - 2
Sex Determination and Chromosomal Disorder .... **P. 114**

- **Monohybrid Cross :**
  - A cross involving two plants differing in one pair of contrasting characters.
  - *e.g.* Mendel crossed tall and dwarf pea plants to study the inheritance of one gene.
- **Steps in Making a Cross in Pea :**
  - Selection of two pea plants with contrasting characters.
  - Removal of anthers (emasculation) of one plant to avoid self pollination. This is female parent.
  - Collection of pollen grains from the other plant (male parent) and transfer to female parent (pollination).
  - Collection of seeds and production of offspring.
  - Mendel made similar observations for other pairs of traits and proposed that factors were inherited from parent to offspring. Now they are called as genes.
  - The F_1 generation (Tt) when self pollinated, produces gametes T and t in equal proportion.
  - Mendel self-pollinated the F_2 plants.
  - He found that dwarf F_2 plants continued to generate dwarf plants in F_3 & F_4.
  - He concluded that genotype of the dwarfs was homozygous- tt.
- **Monohybrid Phenotypic Ratio :** 3 Tall : 1 Dwarf = 3 : 1
- **Monohybrid Genotypic Ratio :**

  Homozygous tall (TT) : 1, Heterozygous tall (Tt) : 2, Homozygous dwarf (tt) : 1= 1 : 2 : 1
- **Back cross and Test cross**
  - Back cross : Crossing of F_1 hybrid with either of its parent.
  - Test cross : Crossing of an F_1 hybrid with its recessive parent (Test cross ratio=1:1). It is used to find out the unknown genotype. Mendel conducted test cross to determine the F_2 genotype.

Mendel's Principles or Laws of Inheritance

1. **Principle of Dominance**
   - Characters are controlled by discrete units called factors.
   - Factors occur in pairs.
   - In a dissimilar pair of factors or contrasting alleles *i.e.,* in heterozygous condition, only one member of the pair expresses its effect in the hybrid and is called as dominant while the manifestation of the other is masked and is called as recessive.
2. **Law of Segregation**

   This law states that allelic pairs separate or segregate during gamete formation and randomly unite at fertilization, thus homozygous parent produces similar gametes. Heterozygous parent produces two kinds of gametes, each having one allele in equal proportion.

   The Concept of Dominance
   - In heterozygotes, there are dominant and recessive alleles.
   - The normal (unmodified or functioning) allele of a gene produces a normal enzyme that is needed for the transformation of a substrate.

- The modified allele is responsible for the production of
  (i) The normal/less efficient enzyme or
  (ii) A non-functional enzyme or
  (iii) No enzyme at all
- **In the first case :** The modified allele will produce the same phenotype like unmodified allele. It becomes dominant.
- **In 2nd and 3rd cases :** The phenotype is dependent only on the functioning of the unmodified allele. Here, the modified allele becomes recessive.

Non-Mendelian Inheritance

(a) Incomplete Dominance

- It is an inheritance in which heterozygous offspring shows intermediate character between two parental characteristics. *e.g.* Flower colour in snapdragon (dog flower or *Antirrhinum* sp.) and *Mirabilis jalapa* (4'O clock plant).
- Here, phenotypic and genotypic ratios are same.
- Phenotypic ratio = 1 Red : 2 Pink : 1 White
- Genotypic ratio = 1 (RR) : 2 (Rr) : 1(rr)
- This means that R was not completely dominant over r.

(b) Co-dominance

- It is the inheritance in which both alleles of a gene are expressed equally and independently in a hybrid *i.e.* both the alleles are dominant, *e.g.* ABO blood grouping in humans.
- ABO blood groups are controlled by the gene I.
- The gene (I) has three alleles I^A, I^B and i. However a person can have any two of these three alleles. I^A and I^B both are dominant alleles while *i* is a recessive allele.
- The alleles I^A and I^B produce antigen A and antigen B respectively on the RBC surface while allele *i* doesn't produce any antigen.
- When I^A and I^B are present together they both express their own types of surface antigen A and B. This is due to co-dominance.

(c) Multiple Allelism

- Here more than two alleles govern the same character.
- Since in an individual only two alleles are present, multiple alleles can be found only when population studies are made *e.g.* ABO blood grouping (3 alleles : I^A, I^B & i). Skin colour and height of humans are also examples of multiple alleles.

(d) Pleiotropy

- Pleiotropy is the phenomenon in which one gene controls many traits. For example, the gene in pea plants that controls the round and wrinkled texture of seeds also influences the phenotypic expression of starch grain size.
- So, if starch grain size is considered as the phenotype, then from this angle, the alleles show incomplete dominance.
- Therefore, dominance is not an autonomous feature of a gene or the product that it has information for. It depends as much on the gene product and the production of a particular phenotype.

Inheritance of Two Genes

➢ **Dihybrid Cross**

- A cross between two parents differing in two pairs of contrasting characters.
- Mendel made some dihybrid crosses *e.g.* Cross between pea plant with round shaped and yellow coloured seeds (RRYY) and wrinkled shaped and green coloured seeds (rryy).
- On observing the F_2, Mendel found that the yellow and green colour segregated in a 3:1 ratio.
- Round and wrinkled seed shape also segregated in a 3:1 ratio.

  Thus, the segregation of one pair of contrasting characters (Round and wrinkled shape) is independent of the segregation of another pair of contrasting character (yellow and green) colour and also that some new combinations of character appear in F_2 generation as the alleles get randomly rearranged in the offsprings at the time of fertilization.
- Dihybrid genotypic ratio: 1 : 2 : 1 : 2 : 4 : 2 : 1 : 2 : 1

  RRYY =1; RRYy =2; RrYY = 2; RrYy = 4; RRyy = 1; Rryy = 2; rrYY = 1; rrYy = 2; rryy = 1

- Dihybrid Phenotypic ratio :
  Round yellow 9 : Round green 3 : Wrinkled yellow 3 : Wrinkled green 1, *i.e.* 9 : 3 : 3 : 1
  The ratio of 9 : 3 : 3 : 1 can be derived as a combination series of 3 yellow : 1 green, with 3 round : 1 wrinkled.
  i.e. (3 : 1) (3 : 1) = 9 : 3 : 3 : 1

3. Mendel's Law of Independent Assortment :

- It states that when more than one pair of characters are involved in a cross, the segregation of one pair of contrasting characters is independent of the segregation of other pair of contrasting characters and also that new recombination of characters alongwith the parental type also appear in F_2 generation.

➢ **Non-recognition of Mendel's work**

- **Mendel's work remained unrecognized till 1900 because,**
  - **(a)** Communication was not easy.
  - **(b)** Non recognition of Mendel as a scientist.
  - **(c)** His mathematical approach was new and unacceptable.
  - **(d)** He used statistical calculations which were beyond the comprehension of the biologists of his time.
  - **(e)** Chromosomes, mitosis and meiosis were not known in Mendel's time.
  - **(f)** The concept of genes (factors) as a stable and discrete units was not accepted. (Mendel could not explain the continuous variations seen in nature).
  - **(g)** Mendel could not provide any physical proof for the existence of factors.
- In 1900, de Vries of Holland, Correns of Germany & Von Tschermak of Austria independently rediscovered Mendel's results and proclaimed his conclusions as the Mendel's Laws of inheritance.

➢ **Chromosomal Theory (1902)**

- The chromosomal Theory was proposed independently by Walter Sutton and Theodor Boveri in 1902.
- Walter Sutton & Theodor Boveri proposed that the pairing and separation of a pair of chromosomes during meiosis lead to segregation of pair of factors.
- Sutton united chromosomal segregation with Mendelian principles and called it as Chromosomal Theory of Inheritance.
- **It states that—**
  - **(a)** Chromosomes are vehicles of heredity *i.e.*, they are transmitted from parents to offspring.
  - **(b)** Two identical chromosomes form a homologous pair. Genes are present in a linear fashion on chromosomes.
  - **(c)** They segregate at the time of gamete formation.
  - **(d)** Independent pairs segregate independently of each other.
  - **(e)** Chromosomes are mutable.
  - **(f)** Sex chromosomes determine sex of an individual.

Parallelism between genes (Mendelian factors) & Chromosomes :

- Mendelian factors as well as chromosomes are transferred from generation to generation.
- The chromosomes occurs in homologous pairs. The genes also occurs in pairs (allele pairs).
- Both chromosomes and genes segregate at the time of gamete formation in such a way that gametes receive only one chromosome & similarly one allele of each pair.
- Different pairs of chromosomes segregate independently of each other. Similarly one pair of alleles segregate independently of another pair.
- Fusion of two (male & female) gametes brings about the diploid chromosome number as well as the allelic pairs in the offspring.
- Thomas Hunt Morgan proved Chromosomal Theory of Inheritance using fruit flies **(***Drosophila melanogaster***).**
- **He took fruit flies as the suitable material because**
  - **(a)** It breeds very quickly.
  - **(b)** Short generation time (life cycle : 12-14 days).
  - **(c)** Breeding can be done throughout the year.
  - **(d)** Hundreds of progenies are produced per mating.
  - **(e)** They can grow on simple synthetic medium.
  - **(f)** Male and female flies are easily distinguishable.

➢ **Linkage and Recombination**

- **Recombination :** It is the generation of non-parental gene combinations.
- **Linkage :** Physical association of two or more genes on a chromosome, Which show tendency to inherit together. They do not show independent assortment.

- Morgan et. al crossed yellow body and white eyed females with wild type brown body and red eyed males and inter-crossed F_1 offsprings. He found that the two genes did not segregate independently, resulted in deviation from normal dihybrid ratio 9 : 3 : 3 : 1 in F_2 generation because the appearance of parental combinations were higher than the non-parental and new recombinations.
- Morgan further carried out several dihybrid test crosses in *Drosophila* to study sex-linked genes.

  Cross A : Double recessive Yellow-bodied, white-eyed females YW/YW X Hybrid Brown-bodied, red-eyed males Y'W'/YW (wild type).

  Cross B : Double recessive White-eyed, miniature winged (Wm/Wm) X Hybrid Red eyed, large winged (W'm/Wm) (wild type).
- **Morgan in the above crosses found that**
  - **(a)** The two genes did not segregate independently of each other and the F_2 ratio deviated from the 9 : 3 : 3 : 1 ratio.
  - **(b)** Genes were located on the X chromosome.
  - **(c)** When two genes were situated on the same chromosome,the proportion of parental gene combinations was much higher than the non-parental type. This is due to linkage.
  - **(d)** Genes for white and yellow were very tightly linked and showed only 1.3% new recombination while white and miniature wings showed 37.2% recombination (loosely linked).
  - **(e)** Tightly linked genes show low recombination.
  - **(f)** Loosely linked genes show high recombination.

  Strength of linkage is inversely proportional to the distance between two linked genes. Thus, the linkage between y & w alleles is stronger than the linkage between w & m alleles.
- **Linkage groups :** All the genes present together on a single chromosome make up a linkage group. The total number of linkage groups in an organism is equal to its haploid number of chromosomes or number of homologous pairs in diploid organisms.
- Alfred Sturtevant used the recombination frequency between gene pairs as a measure of the distance between genes and 'mapped' their position on the chromosome.
- Recombination frequency or the cross over value (COV) can be calculated by following formula.

$$\text{COV} = \frac{\text{Number of recombinants}}{\text{Total number of offsprings}} \times 100$$

- Genetic maps are used as a starting point in the sequencing of genomes as was done in Human Genome Project.

Very Short Answer Type Questions (1 mark each)

AI Q. 1. How does a test cross help to determine the genotype of an individual ?

A [Foreign Set-I, 2016]

Ans. Individual of unknown genotype crossed with recessive parent.
All dominant in progeny—Homozygosity, dominant to recessive ratio 1 : 1 in progeny—Heterozygosity. ½ + ½
[CBSE Marking Scheme, 2016]

Detailed Answer :

Test-cross helps to determine the unknown genotype by crossing it with the recessive parent. If in the progeny all are dominant type then the individual is homozygous and if in the progeny dominant to recessive ratio is 1 : 1, the individual is heterozygous.

Commonly Made Error

- Students often get confused between 'test cross' and 'back cross'. Many of them mention only parent and offspring but they forget to write 'cross' of F_1 hybrid with recessive parent.

Q. 2. Write technical term used in human ABO blood groups for I^A, I^B and *i*

R [Delhi Set-I, Comptt. 2016]

Ans. I^A, I^B and *i* are the alleles of the gene I.
[CBSE Marking Scheme, 2016] 1

Q. 3. Give an example of a plant where the F_2 progeny of a monohybrid cross has same genotypic and phenotypic ratios.

R [Delhi Set-II & III, Comptt. 2016]

Ans. Snapdragon/*Antirrhinum majus*/ Four O' clock plants/*Mirabilis jalapa*. **1**
[CBSE Marking Scheme, 2016]

Commonly Made Error

- Students should write the scientific names properly.

Q. 4. Give an example of a polygenic trait in humans.

R [Delhi Set-II Comptt. 2016]

Ans. Skin colour/height in humans (any other suitable example)
[CBSE Marking Scheme, 2016] 1

Q. 5. In a dihybrid cross carried out by T. H. Morgan in *Drosophila* the F_2 ratio deviated from that of Mendel's dihybrid F_2 ratio. Give a reason.

A [Outside Delhi Set-I, II & III, Comptt. 2016]

Ans. Genes were linked/genes were on the same chromosome and closely associated **1**

[CBSE Marking Scheme, 2016]

Detailed Answer :

The F_2 ratio deviated from that of Mendel's dihybrid F_2 ratio (9 : 3 : 3 : 1) in an experiment performed by Morgan on *Drosophila* because of *Linkage.* The genes were linked as they were located on the same chromosome and closely associated. Therefore, they failed to segregate at the time of gamete formation resulting in greater number of parental combinations and lesser number of new recombinations in F_2 generation, thereby deviating from the normal dihybrid Mendelian ratio.

Answering Tips

- Carefully distinguish and learn the differences between the dihybrid cross done by Mendel and Morgan.

AI Q. 6. If two genes are located far apart from each other on a chromosome, what will be its effect on the frequency of recombination ?

A [CBSE SQP, 2016-17]

Ans. Frequency of recombination will be higher.

[CBSE Marking Scheme, 2016] ½ + ½

Detailed Answer :

Frequency of recombinations will be higher because of greater chances of crossing over and also because of the lesser strength of linkage as the strength of the linkage is inversely proportional to the distance between the genes located on the chromosome.

Q. 7. List any two characters of Pea plants used by Mendel in his experiments other than height of the plant and the colour of the seed.

A [Delhi Comptt. 2017 Set - II, III]

Ans. Flower colour / Flower position / Pod shape / pod colour / Seed shape **(Any two)** ½ + ½

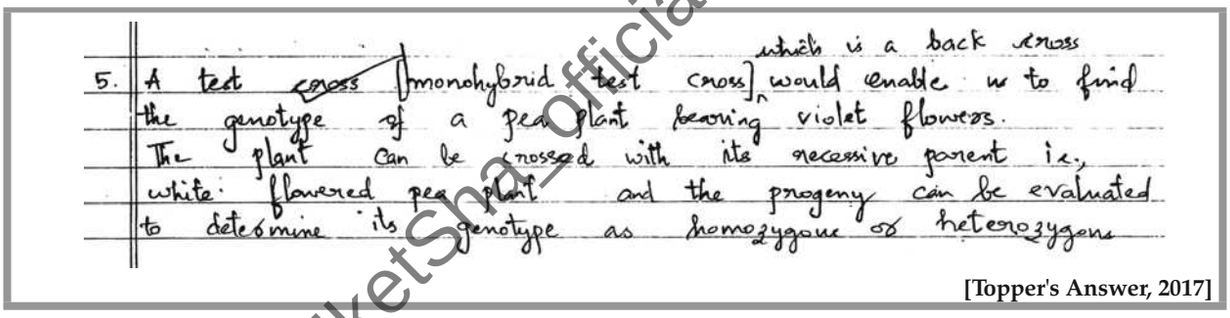
[CBSE Marking Scheme, 2017]

Q. 8. Name the type of cross that would help to find the genotype of a plant bearing violet flowers.

U [Outside Delhi - 2017, Set–I, II, III]

Ans. Crossing of an F_1 hybrid with it's recessive plant is called test cross. It is used to find out the unknown genotype. **1**

OR


5. A test cross [monohybrid test cross], which is a back cross would enable us to find the genotype of a pea plant bearing violet flowers. The plant can be crossed with its recessive parent i.e., white flowered pea plant and the progeny can be evaluated to determine its genotype as homozygous or heterozygous

[Topper's Answer, 2017]

Answering Tip

- Practice examples of test cross, back cross, monohybrid and dihybrid cross using Punette Square.

Q. 9. State a difference between a gene and an allele.

U [KVS, Delhi Set-I, 2016]

Ans. **Gene :** Contains information that is required to express a particular trait // unit of inheritance // segment of DNA called cistron // sequence of DNA coding for tRNA / rRNA / polypeptide / enzyme.

Allele : Genes which code for a pair of contrasting traits / different forms of the same gene / individual gene in a particular gene pair (for same character).

[CBSE Marking Scheme, 2016] 1

Detailed Answer :

Gene is a unit of inheritance, specific segment of DNA or a specific sequence of DNA coding for *t*-RNA/*r*-RNA, polypeptide or enzyme, that is transferred from the parent to the offspring. It controls the expression of a character.

Allele is an alternative form or forms of a single gene that lie on the same locus of homologous chromosomes. *e.g.* if a gene controls height then allele is which gives either tallness or dwarfness. **1**

Commonly Made Error

- Students often get confused between the technical terms like gene, allele etc.

Q. 10. 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. **A [Delhi Set-I, III, 2015]**

Ans. Many generations can be obtained/variations can be exhibited/selected faster. **1**

[CBSE Marking Scheme, 2015]

Detailed Answer:

A geneticist choose organism with a short life cycle because the organism will produce the offspring quickly in a short period of time. These offsprings can be studied by the geneticist and can also be further mated to produce more generation of offsprings.

Q. 11. Mention any two contrasting traits with respect to seeds in pea plant that were studied by Mendel.

U [Outside Delhi Set-I, 2014]

Ans. Round/Wrinkled, Yellow/Green. **1**

[CBSE Marking Scheme, 2014]

Detailed Answer :

Colour of seeds : Dominant - Yellow (YY), Recessive - Green (yy)

Shape of seeds : Dominant - Round (RR), Recessive - Wrinkled. (rr) **1**

Q. 12. What are true breeding lines' that are used to study inheritance pattern of traits in plants ?

R **[Delhi Set-I, 2014]**

Ans. Self pollination continuous for several generations / homozygous. ½+½

[CBSE Marking Scheme, 2014]

Detailed Answer :

True breeding lines for a trait are those plants that have been generated through repeated continuous self-pollination and have become homozygous for a particular trait. They show stability in the inheritance of trait for several future generation. **1**

Q. 13. Name the stage of cell division where segregation of an independent pair of chromosomes occurs.

R **[Delhi Set-III, 2014]**

Ans. Anaphase-1 of Meiosis - 1. **1**

[CBSE Marking Scheme, 2014]

Q. 14. How many types of phenotypes would you expect in F_2 generation in a monohybrid cross ? U **[Outside Delhi, Comptt. 2014]**

Ans. Two types in the ratio of 3 : 1. 1

Q. 15. A garden pea plant produced axial white flowers. Another of the same species produced terminal violet flowers. Identify the dominant traits.

A **[Delhi Set-I, 2012]**

Ans. Axial, violet flower. ½ +½

[CBSE Marking Scheme, 2012]

Detailed Answer :

In garden pea plant bearing axial & white flowers, axial position of the flower is a dominant trait while white flower is a recessive trait, whereas in plants having terminal violet flowers, the terminal position of the flower is a recessive trait while violet colour of the flower is a dominant trait.

Answering Tip

- Use charts to learn all the contrasting traits studied by Mendel in his pea plant experiments. Have a clear understanding of dominant and recessive trait.

Q. 16. 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. A **[Delhi Set-I, 2012]**

Ans. Inflated, green pods. ½ + ½

[CBSE Marking Scheme, 2012]

Commonly Made Error

- Students often get confused between technical terms like dominant, recessive, phenotype and genotype.

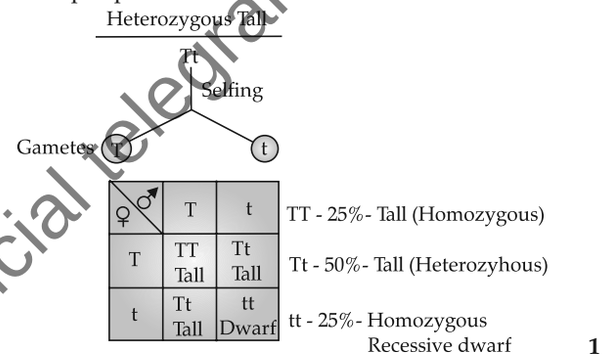
Answering Tip

- Carefully understand the technical terms used in genetics like dominant, recessive, genotype, phenotype etc.

AI **Q. 17. Write the percentage of the pea plants that would be homozygous recessive in F_2 generation when tall F_1 heterozygous pea plants are selfed.**

E & A **[Delhi Set-I, Comptt. 2012]**

Ans. 25% homozygous recessive pea plants will be obtained in F_2 generation when tall F_1 heterozygous pea plants are selfed as follows :



1

Answering Tip

- Practise a number of examples for Monohybrid cross.

Q. 18. Write the percentage of the pea plants that would be heterozygous tall in F_2 generation when tall heterozygous F_1 pea plants are selfed.

A **[Delhi Set-II, Comptt. 2012]**

Ans. 50% heterozygous tall pea plants would be obtained in F_2 generation when tall heterozygous pea plants are selfed. **1**

Q. 19. 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 ?

A **[Delhi Set-III, 2012]**

Ans. When the genes are linked *i.e.*, when genes of dihybrid cross are closely situated on the same chromosome, the proportion of parental gene combinations will be much higher than non-parental types. **1**

AI **Q. 20. Mention two contrasting flower-related traits studied by Mendel in his pea plant experiments.**

R **[Outside Delhi Comptt. 2011]**

Ans. Contrasting flower-related traits :

(i) Flower colour : violet/white or red/white.

(ii) Flower position : Axial/Terminal. ½ + ½

Q. 21. What is the phenotype of the following ?
(i) $I^A i$ & (ii) *ii* R **[Outside Delhi Set-I, 2012]**

Ans. **(i)** Blood group A (Heterozygous) because the allele I^A is dominant over allele *i*.
(ii) Blood group O (Homozygous condition)— because both alleles *ii* are recessive. **1**

Q. 22. Name the respective pattern of inheritance where F_1 phenotype.
(i) does not resemble either of the two parents and is in between the two.
(ii) resemble only one of the parents.
R **[Outside Delhi Set-II, 2012]**

Ans. **(i)** Incomplete dominance
(ii) Dominance. **½ + ½ = 1**

Answering Tip
- Understand and learn all Non-Mendelian inheritance in simple words, giving importance to operative words.

Q. 23. Write the percentage of F_2 homozygous & heterozygous populations in a typical monohybrid cross. E & A **[Foreign 2010]**

Ans. 50% homozygous and 50% heterozygous. Out of 50% homozygous population 25% is homozygous dominant and 25% homozygous recessive. **1**

Q. 24. AaBb was crossed with aabb. What would be the phenotypic ratio of the progeny ? Mention the term used to denote this kind of cross ?
U **[CBSE, SQP, 2010]**

Ans. Phenotypic ratio would be 1:1:1:1. It is test cross. **½ + ½**
[CBSE Marking Scheme, 2010]

Q 25. Mention the type of allele that expresses itself only in homozygous state in an organism.
U **[CBSE Foreign Set, 2011]**

Ans. Recessive allele. **1**
[CBSE Marking Scheme, 2010]

Short Answer Type Questions-I **(2 marks each)**

Q.1. State the Mendelian principle which can be derived from a dihybrid cross and not from monohybrid cross.
R **[CBSE, SQP, 2018]**

Ans. From the dihybrid cross, law of independent assortment can be derived which 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. **1 + 1**
[CBSE Marking Scheme, 2018]

Q. 2. How does the gene I control ABO blood groups in humans ? Write the effect the gene has on the structure of red blood cells.
U **[Delhi Set-I, II, 2014, 2011]**

Ans. **(i)** Gene I has three different alleles I^A, I^B, *i*. ½
(ii) I^A produces A type of sugar / Antigen → A group I^B produces B type of sugar / Antigen → B group ½
(iii) *i* - No sugar/Antigen – O group. ½
(iv) Structure – sugar polymers protrude from the surface of plasma membrane of RBCs. ½
[CBSE Marking Scheme, 2014]

Detailed Answer :

In humans, the ABO blood groups are controlled by a gene called I. It has three alleles, namely I^A, I^B and *i*. A person possesses any two of the three alleles. I^A and I^B are dominant over allele *i*. But I^A and I^B are **co-dominant** as they express themselves equally and independently, when present together. These three alleles yield six different combinations which give four type of blood groups. The allelic pair $I^A I^A$ or $I^A i$ yield blood group A, $I^B I^B$ or $I^B i$ the blood group B, $I^A I^B$ is blood group AB, and ii is blood group O. The plasma membrane of red blood cells has sugar polymers that protrude out from its surface and the kind of sugar is regulated by the gene 'I' of ABO blood group. The alleles I^A and I^B produce A and B types of sugar, while *i* does not produce any sugar. **2**

AI **Q. 3. Write the scientific name of the fruit-fly. Why did Morgan prefer to work with fruit-flies for his experiments ? State any three reasons.**
R **[Delhi Set-I, II, III, 2014]**

Ans. *Drosophila melanogaster* ½
Grown in simple synthetic medium, complete the life cycle in two weeks / short life cycle, single mating produce more progeny, dimorphism, many heritable variations / easy to handle.
(Any three) 1½
[CBSE Marking Scheme, 2014]

Detailed Answer :

Morgan preferred to work with fruit flies because of the following reasons:
(i) It can be grown in simple synthetic medium.
(ii) It can complete its life cycle within two weeks *i.e.*, it has short life cycle.
(iii) Single mating produce more progenies.
(iv) It shows dimorphism and many heritable variations.
(v) It is easy to handle. **2**

Q. 4. Linkage or crossing-over of genes are alternatives of each other. Justify with the help of an example.
A **[Delhi Set-III, 2014]**

Ans. In *Drosophila,* a yellow bodied white eyed female was crossed with brown bodied red eyed male, F_1 progeny produced and intercrossed. The F_2 phenotypic ratio of *Drosophila* deviated significantly from Mendel's 9 : 3 : 3 : 1, the genes for eye colour & body colour are closely located on

the 'X' chromosome showing linkage & therefore inherited together, recombinants were formed due to crossing over but at low percentage.

½ × 4 = 2

[CBSE Marking Scheme, 2014]

Detailed Answer :

Linkage is the tendency of certain genes staying together during inheritance through generations without any change or separation.

This is due to their location on the same chromosomes. The F_2 generation of *Drosophila* deviated from the Mendel's 9 : 3 : 3 : 1 ratio when eye colour was considered. Morgan found that this was due to the fact that eye colour in *Drosophila* is a sex-linked character and its gene is located on X-chromosome. He also observed that eye colour and body colour are closely located on X-chromosome and hence show linkage. The linked characters are generally inherited together. Linkage and crossing over are alternatives of each other as genes tend to remain together when they are located close to each other on the same chromosome. Crossing over between genes takes place only if they are located away from each other.

Commonly Made Error

- Students write incorrect explanation.

Answering Tip

- Understand the concept of Linkage and crossing over thoroughly.

Q. 5. In *Snapdragon*, a cross between true breeding red flower (RR) plants and true breeding white flower (rr) plants showed a progeny of plants with all pink flowers.

(i) The appearance of pink flowers is not known as blending. Why ?

(ii) What is the phenomenon known as ?

A [Delhi Set-III, 2014]

Ans. (i) R (dominant allele red colour) is not completely dominant over r (recessive allele white colour) / r maintains its originality and reappear in F_2 generation. **1**

(ii) Incomplete dominance. **1**

[CBSE Marking Scheme, 2014]

Detailed Answer :

(i) The appearance of pink flowers in snapdragon in F_1 generation is not due to blending of genes Rr which are although together in hybrid, because the parental characters i.e. red and white flowers appear again in F_2 generation without any change.

(ii) Neither of the two alleles are completely dominant over each other therefore the phenomenon is called as incomplete dominance.

Q. 6. Explain pleiotropy with the help of an example.

U [Delhi Set-I, II, 2014]

Ans. Effect of single gene on multiple phenotypic expressions. **1**

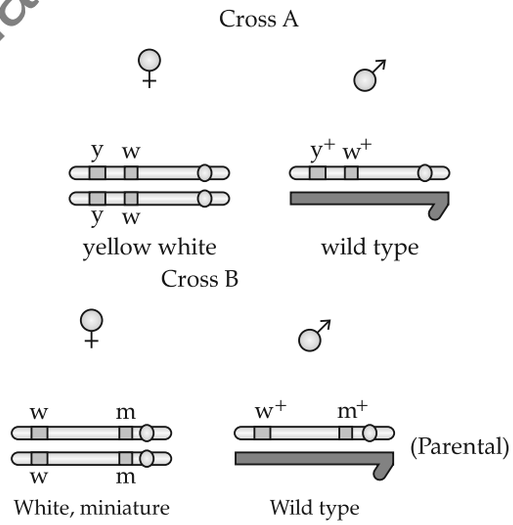
e.g. size of the starch grains produced and shape of the seeds in pea plant are controlled by a single gene // Phenylketonuria characterised by mental retardation and reduction in hair and skin pigmentation. **1**

[CBSE Marking Scheme, 2014]

Detailed Answer :

The ability of a gene to have multiple phenotypic effects because it influences more than one trait or a number of characters simultaneously is called pleiotropism and such genes are known as pleiotropic genes *e.g.* in man a gene producing the disease phenylketoneuria also produces a number of abnormal phenotypic traits such as short stature, mental retardation, widely spaced incisors, pigmented patches on skin etc. Another such example is the size of starch grains produced and shape of the seeds in pea plant are controlled by a single pleiotropic gene. In *Drosophila* the gene for vestigial wings also affects structure of reproductive organs and the bristles on the wings.

Q. 7. Study the figures given below and answer the question.



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

E & A [Delhi Set-I, II, III, 2014]

Ans. Cross A, because they are tightly linked / due to close physical association / they are closely located.

[CBSE Marking Scheme, 2014] 1 + 1= 2

Detailed Answer :

Strength of linkage between genes is higher in cross **A** than that of cross **B** because the two genes yw are located closely on the same chromosome. Whereas in case of cross B the genes w and m are located far apart on the same chromosome. Therefore, in the latter cross the chances of recombination are

higher for crossing over because lesser the distance between genes greater the strength of linkage.

Q. 8. A cross was carried out between two pea plants showing the contrasting traits of height of the plant. The result of the cross showed 50% of parental characters.

(i) Work out the cross with the help of a Punnett square.

(ii) Name the type of the cross carried out.

A **[Delhi Set-I, 2011]**

Ans. **(i)** Tt × tt. **1**

	T	t
t	Tt	tt
t	Tt	tt

(ii) Test cross. **[CBSE Marking Scheme, 2011] 1**

Answering Tip

- Practise a number of examples for test cross.

Q. 9. With the help of one example, explain the phenomena of co-dominance and multiple allelism in human population.

R **[Delhi Set-II, 2011]**

Ans. ABO blood group in human being is an example of multiple allelism.

Three alleles for the gene I *i.e.* I^A, I^B, *i.*

When I^A and I^B are present together the blood group is AB.

Both A and B are expressed and is called co-dominance.

[CBSE Marking Scheme, 2011] 2

Detailed Answer :

ABO blood group in human population is an example of multiple allelism and co-dominance. There are three alleles of gene I, *i.e.*, I^A, I^B and *i.* thus exhibiting multiple allelism. Out of these three alleles any two alleles may occur in human being, one alleles is contributed from each parent. The alleles I^A and I^B are dominant over allele *i* and produce A and B phenotypes respectively. But however when alleles I^A and I^B are present together both of them express equally and independently producing AB phenotype. This is a phenomenon called co-dominance.

Q. 10. A cross between a red flower-bearing plant and a white flower-bearing plant of *Antirrhinum* produced all plants having pink flowers. Work out a cross to explain how this is possible.

A **[Outside Delhi Set-I, II, 2013]**

Ans. A cross between a red flower bearing plant and a white flower bearing plant of *Antirrhinum* produced all plants having pink flowers. This type of cross shows that red flower colour is not completely dominant over white colour flowers *i.e.*, it is a case of incomplete dominance. Cross to explain how this is possible :

Parents — Red flower × White flower

Genotypes — RR × rr

Gametes — R, r

F_1 generation — Rr (All pink colour flowers)

2

AI **Q. 11. Differentiate between multiple allelism and pleiotropy with the help of an example each.**

U **[Delhi Set-I, Comptt. 2013]**

Ans. **Multiple alleles :** More than two alternate forms of a gene present on the same locus of a homologous pair of chromosomes in a population are called multiple alleles. They control the single trait. For example, ABO blood group in humans.

Pleiotropic genes : The gene having a multiple phenotypic effect because of its ability to control the expression of a number of characters is called pleiotropic gene. For example, skin pigmentation and phenylketonuria. **1 + 1 = 2**

Commonly Made Error

- Students often get confused between multiple alleles, polygenic inheritance and pleiotropic genes.

Answering Tip

- Carefully learn the explanation of these terms with the help of examples.

Q. 12. In a cross between two tall pea plants, some of the offsprings produced were dwarf. Show with the help of the Punnett Square how this is possible.

A **[Delhi Set-I, II, III, 2013]**

Ans. In a cross between two tall pea plants, some of the offsprings produced were dwarf. It indicates that parent pea plants were heterozygous for tallness (Tt) *i.e.*, they contain a recessive gene (t) for dwarfness from each of the parent plant.

Parent plants — Tall × Tall

Genotype — Tt × Tt

Gametes — T, t × T, t

♀ \ ♂	T	t
T	TT Tall	Tt Tall
t	Tt Tall	tt Dwarf

25% plants were dwarf. Phenotypic ratio – Tall : Dwarf :: 3 : 1 Genotypic ratio – 1 : 2 : 1

Punnett square : Punnett square is a graphic representation of the probabilities of all the possible genotypes and phenotypes of offsprings in a cross. **2**

Q. 13. List all three different allelic forms of gene I in humans. Explain different phenotypic expressions controlled by these three forms.

R [Delhi Set III. 2012]

Ans. The three different forms of gene I are I^A, I^B and *i*. ABO blood grouping is controlled by these three alleles, hence it is an example of multiple allelism. Each person possesses any two of the three I gene alleles. I^A and I^B are completely dominant over allele (*i*) while I^A and I^B are co-dominant because both of them express themselves equally and independently when present together. As there are three alleles of gene I there would be following six types of allelic combinations of genotypes resulting in four phenotypic expressions as follows :

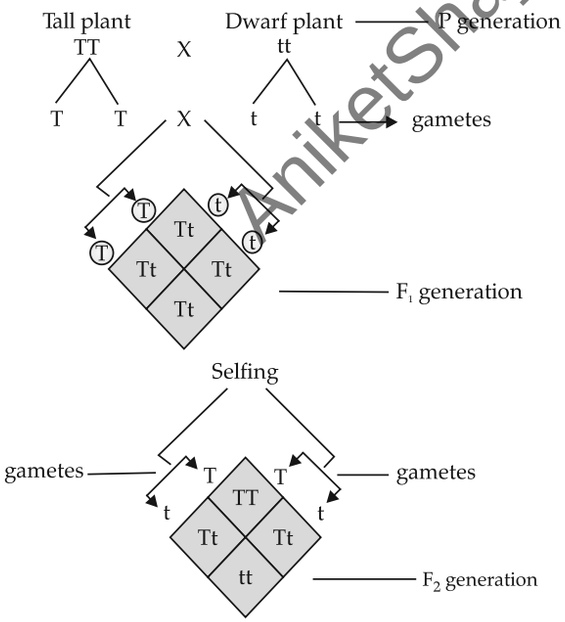
	Blood group phenotype	Genotype
(i)	A	I^AI^A, I^Ai
(ii)	B	I^BI^B, I^Bi
(iii)	AB	I^AI^B
(iv)	O	*ii*

2

Q. 14. Tallness of pea plants is a dominant trait, while dwarfness is the alternate recessive trait. When a pure-line tall is crossed with pure-line dwarf, what fraction of tall plants in F_2 shall be heterozygous ? Give reasons.

A [Delhi Comptt. 2011]

Ans. Pure line tall is crossed with pure line dwarf.



Phenotypic ratio = Tall : dwarf
3 : 1

Genotypic ratio = TT : Tt : tt
1 : 2 : 1

Two third of tall progenies are heterozygous because gene for tallness (T) is dominant and expresses itself in heterozygous condition.

A Punnet square is used to understand a typical monohybrid cross between tall and dwarf plants.

2

Q. 15. ABO blood groups is a good example of co-dominance. Justify.

U [Outside Delhi Comptt. 2017 Set – I, II, III]

Ans. **(i)** ABO blood group in humans is contributed by gene 'I' that has 3 alleles 'I^A' 'I^B' and '*i*.'

(ii) Because human beings are diploid and each person has two of the three alleles.

(iii) I^A and I^B produce two different types of sugar while allele *i* does not produce sugar on the plasma membrane of RBC.

(iv) When I^A and I^B present they produce their own type of sugar-this is called co-dominance.

½ + ½ + ½ +½

Q. 16. Explain co-dominance with the help of one example.

R [Foreign Set – II, III – 2017, (NCERT)]

Ans. When the dominant alleles of the same gene which are contributed by both parents are expressed (called co-dominance) // F_1 generation resembles both the parents :

In human blood group :

Parents	$I^A I^A$	$I^B I^B$
Gametes	I^A	I^B
F_1 –	I^AI^B	

OR

In human red blood cells, alleles I^A and I^B of gene I are both dominant, when I^A & I^B are present together in an individual both are expressed as I^A I^B, (AB blood group).

½ × 4 = 2

[CBSE Marking Scheme, 2017]

Answering Tip

- Practice cross with the help of different characteristic features in three stages:

(a) Parents
(b) F_1 generation
(c) F_2 generation

Short Answer Type Questions-II

(3 marks each)

Q. 1. True-breeding pea plants showing contrasting character for flower position were cross-bred.
(i) Mention the position of flowers in F_1 generation.
(ii) Work out the cross up to F_2 generation.
(iii) Compute the relative fraction of various genotypes in the F_2 generation ? **A [SQP, 2016-17]**

Ans. (i) Axial position — Due to Principle of Dominance

(ii)

AA × aa ..... Parental generation
(Axial) (Terminal)
A a ..... Gametes
Aa (Axial) F_1 Generation
Aa × Aa Selfing

A a A a ..... Gametes
AA Aa Aa aa F_2 generation

(iii) Phenotypic ratio—Axial : Terminal :: 3 : 1
Genotypic ratio—1 : 2 : 1
Homozygous axial (AA) = 1/4
Heterozygous axial (Aa) = ½
Homozygous terminal (aa) = 1/4 **3**

Answering Tip

- Stress on the difference between Monohybrid and Dihybrid cross, F_1 and F_2 generation, phenotypic and genotypic ratio.

Q. 2. Explain the laws that Mendel derived from his monohybrid crosses.

U **[Delhi Set. II. Comptt. 2016]**

Ans. Law of Dominance, states that factors that control characters occur in pairs and in dissimilar pair one factor dominates (expresses itself) whereas the other recessive factor does not express in the presence of the dominant one.
Law of Segregation, states that during gamete formation, the factors or alleles of a pair segregate/separate (from each other) and one gamete receives only one of the factors. **1½ + 1½**
[CBSE Marking Scheme, 2016]

Commonly Made Error

- Many students fail to explain the law correctly. They get confused with the three laws. Many of them write all the three laws as they did not understand the questions.

Answering Tip

- Understand and state Mendel's laws in simple words, giving importance to operative words.

Q. 3. Work out the monohybrid cross up to F_1 by taking a suitable example so as to show the following phenotypes.
(i) F_1 represents only one of the parents
(ii) F_1 with both the parental characteristic

A **[Outside Delhi Set-I, II, III Comptt. 2016]**

Ans. (i) Tall × Dwarf
Parent pea plant --- TT tt ½
gametes T T t t
F_1 Tt Tt Tt Tt
100% Tall (F_1 representing only one of the parents)
½ × 3 = 1½

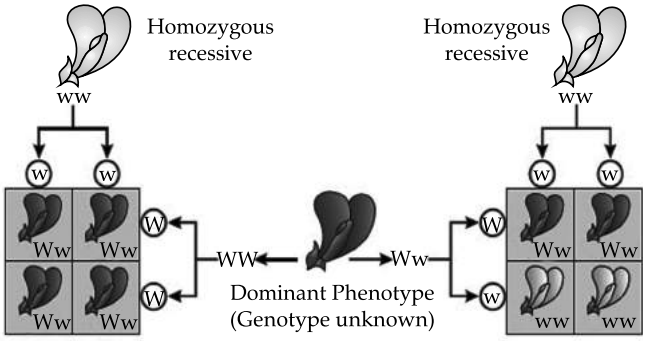
(ii) In Humans—Inheritance of blood groups.
Parent: A-Blood group B-Blood group.
Gametes — A A B B
F_1 AB AB AB AB
100% with blood group AB. **½×3=1½**

Q. 4. What is a test cross ? How can it decipher the heterozygosity of a plant ? R **[Outside Delhi Set-I, 2016]**

Ans. Test cross : A cross to analyse whether genotypes of dominant individual is homozygous or heterozygous.
On crossing with a recessive parent, if 50% of progeny have dominant trait and 50% have recessive trait then the plant is said to be heterozygous. **[CBSE Marking Scheme, 2016] 2 + 1 = 3**

Detailed Answer :

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



Test cross to know homozygous or heterozygous nature of dominant trait

If the progenies produced by a test cross show 50% dominant trait and 50% recessive trait, then the unknown individual is heterozygous for the trait. Thus on basis of this ratio 50% : 50% or 1 : 1 the heterozygosity of the plant can be deciphered. On the other hand, if the progeny produced shows only dominant trait, then the unknown individual is homozygous for a trait. **3**

OR

(23) A Test cross is a cross made between an F_1 or F_2 dominant phenotypic plant and a recessive phenotype parent in order to know the genotype of the dominant phenotyped plant.

→ There are two cases possible if a plant has dominant phenotype →
(i) It is heterozygous (say Tt).
In this case of Test cross

Tt × tt

	T	t
t	Tt	tt
t	Tt	tt

50% dominant and 50% recessive

→ If the Result of the test cross is 1:1 then it is concluded that the dominant plant is heterozygous.

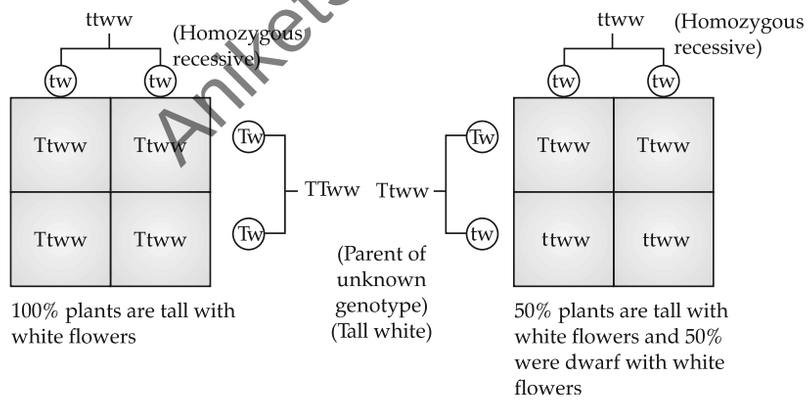
(ii) It is pure homozygous (say TT)
In this case of test cross, all the offspring obtained, will have dominant phenotype.

[Topper's Answer, 2016]

AI **Q. 5. How would you find genotypes of a tall pea plant bearing white flowers ? Explain with the help of a cross. Name the type of cross you would use.**

R **[Delhi Set-III, 2016]**

Ans.



Type of Cross is test cross.

Explanation : Tallness is a dominating trait and can be expressed in homozygous as well as heterozygous condition while recessive traits (in pea) are expressed only in homozygous condition.

So, to find out the genotype of the tall pea with white flowers we will test cross it with a dwarf pea plant bearing white flowers (as it is homozygous).

If the progeny shows 1 : 1 ratio between dominating and recessive trait *i.e.* 50% progenies are tall with white flowers and 50% are dwarf with white flowers then this proves the experimental plant is heterozygous for the dominating trait and if all the progenies *i.e.* 100% plants are tall with white flowers, this proves that the experimental plant (Tall with white flower) was homozygous for tallness and is the dominant trait.

This type of the cross is the test cross as shown above. **1+2=3**

Q. 6. Write three basic facts that are highlighted in Mendel's Law of Dominance.

R **[Delhi Comptt. 2017, Set – I]**

Ans. **(i)** Characters are controlled by discrete units called factors.

(ii) Factors occur in pairs.

(iii) In a dissimilar pair of factors, one member of a pair dominates (dominant) the other (recessive). **1 + 1 + 1 = 3**

[CBSE Marking Scheme, 2017]

Detailed Answer:

(i) Law of dominance states that characters are controlled by genes.

(ii) Genes occur in pairs.

(iii) When two alternate forms of a trait or character (genes or alleles) are present in an organism, only one factor express (dominant) itself in F_1 generation. While the other factor remains hidden (Recessive).

Q. 7. When a snapdragon plant bearing pink colour flower was selfed, it was found that, 69 plants were having red coloured flowers. What would be the number of plants bearing pink flower. Show with the help of Punnett square. Identify the principle of inheritance involved in this experiment.

U **[CBSE SQP – 2017]**

Ans. **(i)** There will be 138 pink flower bearing plants and 69 white flower bearing plants.

(ii) Pink (Rr) selfing

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

Phenotypic ratio : red : pink : white

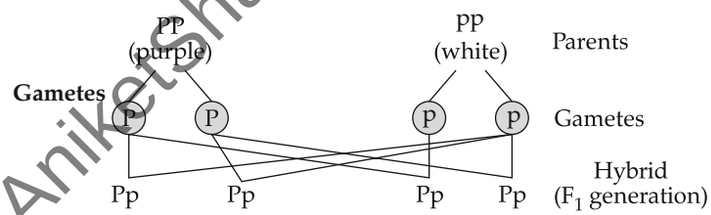
Genotypic ratio: 1 : 2 : 1

(iii) Incomplete dominance **1 + 1 + 1**

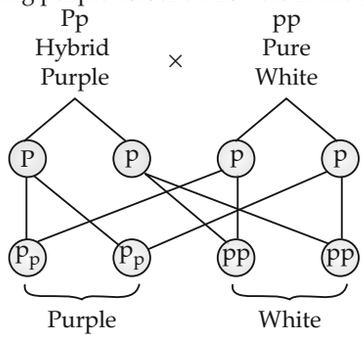
Q. 8. A teacher wants his/her students to find the genotype of pea plants bearing purple coloured flowers in their school garden. Name and explain the cross that will make it possible. A **[Delhi Set-I, 2015]**

Ans. Purple colour of flower is a dominant trait in pea plants. The genotype of such plants can be determined by test cross. Test cross determines that the dominant character is coming from homozygous dominant genotype or heterozygous genotype *e.g.* purple flower coming from PP or Pp. It can be done by crossing plants having purple coloured flowers with plants having white coloured flowers, which will always have homozygous recessive genotype.

If the progenies obtained, all have purple flowers, the genotype of purple flower would be PP as shown in the following cross :



Thus, the genotype of pea plant bearing purple coloured flowers in the school garden can be Pp or PP.



3

OR

Purple : white : : 50% : 50% or 1 : 1

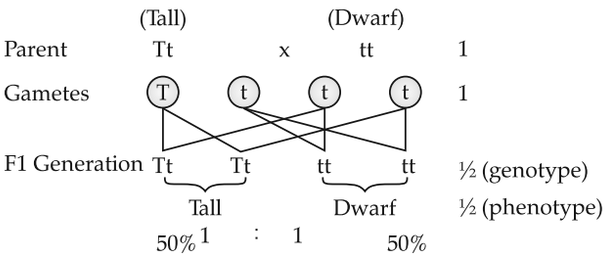
Answering Tip

- As per the scope of the syllabus the complete working of the cross should be shown by Punnett Square.

Q. 9. 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.

[Outside Delhi Set-I, 2015]

Ans.



The asked scenario is possible only when the tall pea plant is heterozygous and dwarf pea plant is homozygous.

In this case, the progeny would be 50% tall and 50% dwarf. **3**

Q. 10. (i) Write the conclusions Mendel arrived at on dominance of traits on the basis of monohybrid crosses that he carried out in pea plants.

(ii) Explain why a recessive allele is unable to express itself in a heterozygous state.

U [Delhi Set-I, 2014]

Ans. (i)

(a) Characters are controlled by discrete unit called factors. ½

(b) Factors occur in pairs. ½

(c) In a dissimilar pair of factors one member of the pair dominates / only one of the parental character is expressed in a monohybrid cross in the F_1 and both are expressed in the F_2 **1**

(ii) Due to non-functional enzyme / less efficient enzyme / no enzyme at all. **1**

[CBSE Marking Scheme, 2014]

Detailed Answer :

(i) On the basis of monohybrid crosses carried in pea plants, Mendel concluded that :

(a) The characters are controlled by discrete units called factors now known as genes or alleles.

(b) These factors/genes occur in pairs.

(c) When the factors of a pair are dissimilar only one expresses itself in F_1 generation of a monohybrid cross while the manifestation of the other is masked. The character which finds its expression in F_1 is called as dominant and the latter as recessive factor. On this basis Mendel formulated the Law of Dominance.

(ii) The recessive allele is unable to express itself in heterozygous state because this does not code for its product probably due to non-functional or less active enzymes.

Q. 11. In pea plants, the colour of the flower is either violet or white, whereas human skin colour shows many gradations. Explain giving reasons how it is possible. **A [Delhi Set-I, Comptt. 2013]**

Ans. Human skin colour, an example of polygenic inheritance and it is controlled by at least three separate genes and inheritance of these genes is called quantitative inheritance. There can be total eight allelic combinations in the gametes of a person heterozygous for all the three genes, hence 64 combinations or gradations in colour are possible. In this type of inheritance, the dominant alleles have cumulative effect where in each dominant alleles expresses only a part of the trait and the trait in its full form is expressed only when all the dominant alleles are present.

On the other hand, the colour of the flower in pea is controlled by allelic complementary genes, which independently show a complete effect. The inheritance is qualitative as here the presence of a single dominant allele expresses the trait in full form and the presence of two dominant alleles does not make any difference. **3**

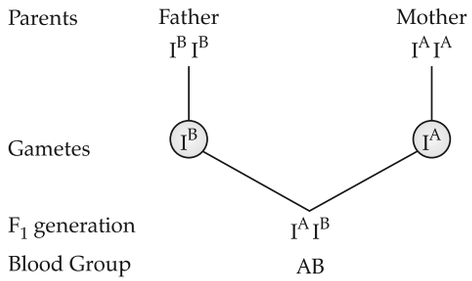
AI Q. 12. (i) Why is human ABO blood group gene considered a good example of multiple alleles?

(ii) Work out a cross up to F_1 generation only, between a mother with blood group A (homozygous) and the father with blood group B (homozygous). Explain the pattern of inheritance exhibited.

U [Delhi Set-I, II, 2013]

Ans. (i) In human ABO blood group, there are 4 possible phenotypes for this character i.e. AB, A, B, O. More than two alleles govern the human blood group. The four blood groups result from various combination of 3 different alleles.

(ii) The cross exhibits co-dominance. When the two alleles I^A and I^B are present together both the alleles express themselves equally forming the blood group AB.



This cross exhibit Mendelian pattern of inheritance which states that the two factors for a trait segregate at the time of gamete formation and again come together at the time of fertilization in the zygote and offsprings.

1½ + 1½ = 3

Q. 13. (i) Explain the phenomena of multiple allelism and co-dominance taking ABO blood group as an example.

(ii) What is the phenotype of the following :

(i) I^Ai (ii) ii

U [Outside Delhi Set-II, 2012]

Ans. (i) One gene I has three alleles I^A, I^B and i hence multiple allelism. **1**

We inherit any two of them. 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 Antigen A and B- Co-dominance. **1**

(ii) **(a)** I^A i - A blood group. ½
(b) ii - O blood group ½
[CBSE Marking Scheme, 2012]

Detailed Answer :

(i) Multiple allelism is the phenomenon of occurrence of a gene in more than two allelic forms on the same locus of homologous pair chromosome. In ABO blood group in humans, one gene I has three alleles I^A, I^B and i.

Co-dominance is the phenomena in which both alleles of a gene express themselves when present together. We inherit any two of these three 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.

(ii) **(a)** I^A — A blood group. **(b)** *ii* — O blood group. **2+1**

Q. 14. In a cross between a true-breeding red-flowered and a true-breeding white-flowered snapdragon plant, the F_1 plants produced pink flowers. Name and explain the type of inheritance.

A **[Delhi Set-I, Comptt. 2012]**

Ans.

Red		White
RR	×	rr
R		r
	Rr	F_1 generation
	Pink	

This phenomenon is called as incomplete dominance.

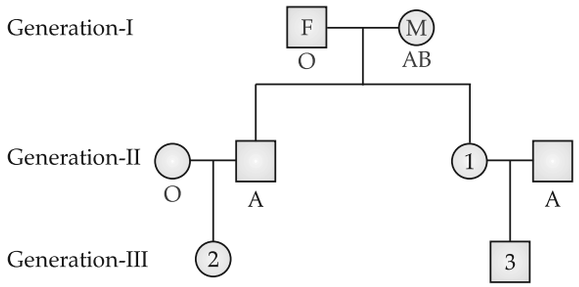
Gamete (Selfing)

↓ \ →	R	r
R	RR Red	Rr Pink
r	Rr Pink	rr White

F_2 generation 1 : 2 : 1

It shows incomplete dominance. In a cross between true-breeding red flower (RR) and true-breeding white-flowered plants (rr) in F_1 generation we get pink (Rr) flowers. When F_1 was self-pollinated, we get 1 : 2 : 1 (Red : Pink : White) ratio of flowers. Genotypic ratio was exactly as we would expect but phenotype ratio had changed from 3 : 1 (dominant : recessive) ratio to 1 : 2 : 1. This is because R was not completely dominant over r and it made it possible to distinguish Rr as pink from RR (red) and rr (white). **3**

Q. 15. Study the following pedigree chart of a family, starting with mother with AB blood group and father with O blood group.



(i) Mention the blood group as well as its genotype of the offspring numbered 1 in generation II.
(ii) Write the possible blood groups as well as their genotypes of the offsprings numbered 2 and 3 in generation III.

A **[Outside Delhi Set-I, Comptt. 2012]**

Ans. **(i)** Blood group B; Genotype – I^B i
(ii) **(a)** Offspring Numbered 2: Blood Groups can be: O/A Genotype = I^A i / ii The blood group genotype of the offspring: Mother's blood group is O: genotype is ii Father's blood group is A: genotype is I^Ai Thus, offspring can be: I^A i/ii

(b) Offspring Numbered 3: Blood Groups can be: AB/B/O. Genotype: ii/I^Bi/I^BI^A. The blood group genotype of the offspring 3 will depend on her parents blood group genotype. Mother's blood group is B: genotype is I^Bi Father's blood group is A: genotype can be I^AI^A or I^Ai Thus, offspring can be: I^Ai / ii / I^Bi / I^BI^A

The allele I^A produces glycoprotein A, found on the surface of red blood cells.

The allele I^B produces glycoprotein B, found on the membrane of red blood cells.

When the alleles I^A and I^B are together, they are equally dominant and both the glycoproteins A and B are produced.

The blood group is determined by the presence or absence of one or both the glycoprotein *i.e.* group A, B and AB while group O has neither of them. **3**

Q. 16. How are dominance, co-dominance and incomplete dominance patterns of inheritance different from each other ?

U **[Delhi Set-I, II, III, 2011]**

Ans. **Dominance :** One allele expresses itself in the hybrid heterozygous condition, other is suppressed.

Co - dominance : Both the alleles of a gene express in a heterozygous hybrid containing two dominant alleles.

Incomplete dominance : Neither of the two alleles of a gene is completely dominant over the other in heterozygous, the hybrid is Intermediate.

1 × 3 = 3
[CBSE Marking Scheme, 2011]

Commonly Made Error

- Students often get confused between terms like dominance, Co-dominance and incomplete dominance.

Q. 17. In pea plants let symbol Y represent dominant yellow; symbol y, the recessive green; symbol R, the round seed shape and symbol r, the wrinkle seed shape. A typical Mendelian dihybrid cross was carried out in pea plants. Write the genotypes of :

(i) Homozygous dominant and recessive parents.

(ii) Gametes produced by both the parents.

(iii) F_1 offspring.

(iv) Gametes produced by F_1 offspring.

A **[Outside Delhi Set-I, II, III, Comptt. 2011]**

Ans. (i) Genotype of :

(a) Homozygous dominant parent—YYRR.

(b) Homozygous recessive parent—yyrr

(ii) Gametes produced by both the parents are :

(a) YR and (ii) yr

(iii) F_1 offsprings.

Phenotype — All seeds are yellow round and Heterozygous

Genotype — YyRr.

(iv) Gametes produced by F1 offsprings

4 types – (YR) (Yr) (yR) (yr)

1 + 1 + ½ + ½

Q. 18. During his studies on genes in *Drosophila* that were sex-linked T.H.Morgan found F_2 - population phenotypic ratios deviated from expected 9 : 3 : 3 : 1. Explain the conclusion he arrived at.

A **[Delhi Set-I, 2010]**

Ans. (i) Linkage, genes on the same chromosome were either closely associated or far apart.

(ii) Higher percentage of parental combination and fewer percentage of recombinants are observed when two genes are located very close / tightly linked on the same chromosome.

(iii) Higher percentage of recombinants and fewer percentage of parental combinations are observed when two genes are located far apart / loosely / linked on the same chromosome.

1 × 3 = 3

[CBSE Marking Scheme, 2010]

AI **Q. 19. When Morgan conducted dihybrid cross on *Drosophila* like Mendel did with pea plants, the ratios deviated significantly from that of Mendel's F_2 ratio. Write the explanation Morgan and his group gave to the observations they obtained from their experiment.**

A **[Delhi Comptt. 2017, Set – II, III]**

Ans. (i) When two genes in a dihybrid cross were located on the same chromosome they did not segregate independently.

(ii) The proportion of parental gene combinations were much higher than non-parental combinations / recombinants.

(iii) Physical association of two genes was termed as linkage. **1 × 3 = 3**

[CBSE Marking Scheme, 2017]

Q. 20. Human blood group is a good example of multiple allelism and co-dominance. Justify.

A **[Foreign Set-I, 2016]**

Ans. Multiple allelism : Generally in an individual/ population only two alleles of a trait govern the character but in case of ABO blood group, three alleles I^A, I^B and i are found to govern blood group in human population.

Co-dominance : Allele I^A and I^B when present in an individual, both being dominant express their own types of sugars/antigen (no marks for the second step if two alleles are not given correctly).

[CBSE Marking Scheme, 2016] 2+1=3

Detailed Answer :

In human blood group, there are four possible phenotypes A, B, AB & O. These blood groupings are controlled by gene I. There are three instead of normally two alleles of this gene namely I^A, I^B & i which control these four blood groups. Hence it is an example of multiple allelism.

Out of these three alleles I^A and I^B are dominant over *i*. Each person in a population possesses any two of the three alleles, one from each of the two parents. When allele I^A and I^B both are present together in an individual, both being dominant express equally and independently and hence are co-dominant.

Thus, human blood is a good example of both multiple allelism as well as co-dominance.

Q.21. Write the percentage of F_2 homozygous and heterozygous population in a typical monohybrid cross. R **[CBSE Foreign, 2010]**

Ans. 50% are homozygous and 50% heterozygous. Out of the homozygous population 25% is homozygous dominant and 25% homozygous recessive. The genotypic ratio is 1 : 2 : 1 (25% homozygous dominant, 50% heterozygous dominant and 25% homozygous recessive). **3**

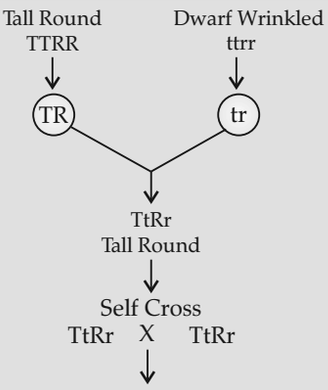
[CBSE Marking Scheme, 2010]

Long Answer Type Questions (5 marks each)

AI Q. 1. Give a genetic explanation for the following cross. When a tall pea plant with round seeds was crossed with a dwarf pea plant with wrinkled seeds then all the individual of F_1-populations were tall with round seeds. However selfing among F_1-population led to a 9 : 3 : 3 : 1 phenotypic ratio.

A [Outside Delhi Set-II, 2016]

Ans.



/ F_1 genotype

Self Cross

TtRr X TtRr

	TR	Tr	tR	tr
TR	TTRR Tall Round	TTRr Tall Round	TtRR Tall Round	TtRr Tall Round
Tr	TTRr Tall Round	TTrr Tall Wrinkled	TtRr Tall Round	Ttrr Tall Wrinkled
tR	TtRR Tall Round	TtRr Tall Round	ttRR Dwarf round	ttRr Dwarf Round
tr	TtRr Tall Round	Ttrr Tall Wrinkled	ttRr Dwarf round	ttrr Dwarf wrinkled

/ Gametes formed to develop F_2 plants

2

Phenotypic ratio —

Tall Round	Tall Wrinkled	Dwarf Round	Dwarf Wrinkled	/ F_2 Phenotype	½
9	: 3	: 3	: 1		½

Law of dominance : In a dissimilar pair of factors, one member of the pair is dominant and the other is recessive. In the given cross tall and round are dominant where as dwarf and wrinkled are recessive (explain with or without a cross). ½

Law of Segregation : Allelic pairs separate or segregate during gamete formation and the paired condition is restored during fertilisation (explain with or without a cross). ½

Law of Independent Assortment : The new combination seen in F_2 generation (Tall wrinkled) (Dwarf round) is only possible when the two gene pairs for height and seed shape assort independently of each other during gamete formation. The law 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. **1**

[CBSE Marking Scheme, 2016] 1

Detailed Answer :

Genetic explanation of the cross : If a pea plant with two pairs of contrasted characteristics like tall/dwarf and round/wrinkled seeds are crossed the F_1 progeny would have all tall plants with round seeds according to the law of dominance. This implies that tallness is dominant over dwarfness and round seed is dominant over wrinkled seeds. In F_2 progeny, there would be some tall plants with round seeds and dwarf plants with wrinkled seeds.

However, there would be some plants with recombination of characters such as tall plants with wrinkled seeds and dwarf plants with round seeds. This depicts that tall-dwarf and round/wrinkled traits are inherited independent of each other following law of independent assortment, which

states that when two traits are taken in a cross the segregation of one trait is independent to the segregation of other character.

Q. 2. (i) What is polygenic inheritance ? Explain with the help of suitable example.

(ii) How are pleiotropy and Mendelian pattern of inheritance different from polygenic pattern of inheritance? U **[Outside Delhi Set-III, 2016]**

Ans. (i) Inheritance in which traits are controlled by three or more genes, *e.g.*, human skin colour / height, the inheritance depends upon the additive / cumulative effect of alleles, more the number of dominant alleles the expression of the trait will be more distinct / prominent, more the number of recessive alleles the trait will be diluted, if member of dominant and recessive alleles are equal the effect is intermediate. **½ × 6 = 3**

Same explanation with the help of any suitable example.

(ixi)

Single gene controls multiple phenotypic expression (Pleiotropy), one gene controls one phenotypic expression (Mendelian). **1 + 1 = 2**

[CBSE Marking Scheme, 2016]

Detailed Answer :

(i) Polygenic inheritance is the inheritance of traits that are produced by the combined effect of many genes. Polygenic trait is controlled by more than one pair of non-allelic genes and shows different types of phenotypes. For example, human skin colour is an example of polygenic inheritance. It is caused by a pigment called melanin due to three pairs of polygenes (A, B and C).

(ii) Mendelian inheritance refers to the expression of monogenic traits *i.e.* gene expression is controlled by one gene. In a pair of alleles, expression of the recessive gene is always masked by the expression of a dominant gene.

Pleiotropy is the ability of a gene to have multiple phenotypic effects because it influences several characters simultaneously.

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

Q. 3. A tall pea plant bearing violet flowers is given with its unknown genotypes. Explain by working out the crosses how would you find the correct genotypes with respect to the two traits mentioned only by 'selfing' the given plants.

A **[Outside Delhi Set-II, Comptt. 2016]**

Ans. Tall plant = TT/Tt

Violet flowers = WW/Ww

Genotype of given plant could be any of the assumed four : TTWW, TTWw, TtWW, TtWw. **1**

Case 1 :

Selfing of TTWW ⟶ Gametes (TW)

♀ \ ♂	**TW**
TW	TTWW

All tall and violet, then genotype of parent is TTWW **1**

Case 2 :

Selfing of TTWw ⟶ Gametes (TW), (Tw)

♀ \ ♂	**TW**	**Tw**
TW	TTWW	TTWw
Tw	TTWw	TTww

Phenotypic ratio 3 : 1 (3 tall violet : 1 tall white) then Parent is TTWw . **1**

Case 3 :

Selfing of TtWW ⟶ Gametes (TW), (tW)

♀ \ ♂	**TW**	**tW**
TW	TTWW	TtWW
tW	TtWW	ttWW

Phenotypic ratio 3 : 1, (3 tall violet : 1 dwarf violet) then Parent is TtWW **1**

Case 4 :

Selfing of TtWw ⟶ Gametes (TW), (Tw), (tW), (tw)

♀ \ ♂	**TW**	**Tw**	**tW**	**tw**
TW	TTWW	TTWw	TtWW	TtWw
Tw	TTWw	TTww	TtWw	Ttww
tW	TtWW	TtWw	ttWW	ttWw
tw	TtWw	Ttww	ttWw	ttww

Tall violet = 9

Tall white = 3

Dwarf violet = 3

Dwarf white = 1

If phenotypic ratio 9 : 3 : 3 : 1, then parent is TtWw. **1**

Q. 4. (i) Work out a dihybrid cross upto F_2 generation between homozygous tall pea plant bearing violet flowers and dwarf pea plants bearing white flowers.

(ii) Name the law that Mendel deduced from such a dihybrid cross. A **[Outside Delhi Set-III, Comptt. 2016]**

Ans. (i) A dihybrid cross between a homozygous :

(a) Tall pea plant bearing violet flowers (Dominant). Genotype TTVV.

(b) Dwarf pea plant bearing white flowers (recessive). Genotype ttvv.

(a) Parent pea plants — Tall violet TTVV X Dwarf white ttvv

Gametes: (TV) (TV) (tv) (tv)

F_1 TtVv (Selfing) TtVv All hybrid Tall pea plants bearing violet flower

Gametes

♀ \ ♂	TV	tV	Tv	tv
TV	TTVV	TtVV	TTVv	TtVv
tV	TtVV	ttVV	TtVv	ttVv
Tv	TTVv	TtVv	TTvv	Ttvv
tv	TtVv	ttVv	Ttvv	ttvv

F_2

Phenotypic ratio : Tall violet : Tall white : dwarf violet : dwarf white

9 : 3 : 3 : 1

(ii) Law of Independent Assortment. **4 + 1 = 5**

Answering Tip

- Train students to understand and state Mendel's laws in simple words, giving importance to operative words. Stress on the difference between Monohybrid and Dihybrid cross, F_1 and F_2 generation.

AI Q. 5. How do 'Pleiotropy', 'incomplete dominance', 'co-dominance' and 'polygenic inheritance' deviate from the observation made by Mendel ? Explain with the help of example for each. A **[Delhi Set-II, Comptt. 2015]**

Ans.

No.	Mendel's Observations	Deviations
(i)	One gene responsible for a single phenotype.	**Pleiotropy :** Single gene exhibits multiple phenotypic expressions *e.g.*, Disease Phenylketonuria (a single gene codes for enzyme phenylalanine hydroxylase which manifests in form of mental retardation and reduction in hair and skin pigmentation.
(ii)	Only one of the parental characters appear in a monohybrid cross (with contrasting) character in the F_1 generation.	**Incomplete dominance :** In a monohybrid cross (with contrasting character) phenotype in the F_1 generation did not resemble either of the two parents and was in between the two *e.g.*, Dogflower/snapdragon/*Antirrhinum*.
(iii)	Only one of the parental characters appear in a monohybrid cross with contrasting characters in the F_1 generation.	**Co-dominance :** In a monohybrid cross (with contrasting character) phenotype in the F_1 generation expresses genes of both the parents *e.g.*, ABO blood group.
(iv)	All the traits have distinct alternate forms since one gene is responsible for a single phenotype.	**Polygenic inheritance :** The occurrence of traits are spread across a gradient. Such traits are controlled by three or more genes. *e.g.* Human height/skin colour.

[CBSE Marking Scheme, 2015] 5

Q. 6. (i) Dihybrid cross between two garden pea plant one homozygous tall with round seeds and the other dwarf with wrinkled seeds was carried.

(a) Write the genotype and phenotypes of the F1 progeny obtained from the cross.

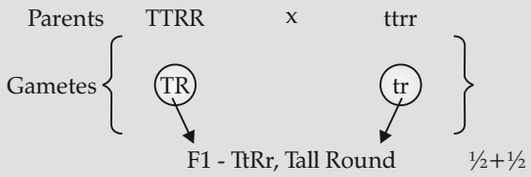
(b) Give the different types of gametes of the F1 progeny.

(c) Write the phenotypes and its ratios of the F2 generation obtained in this cross along with the explanation provided by Mendel.

(ii) How were the observations of F2 progeny of dihybrid crosses in Drosophila by Morgan different from that of Mendel carried out in pea plants ? Explain giving reasons.

A **[Delhi Set-I, Comptt. 2015]**

Ans. (i) (a)



(b) Gametes F (TR) (Tr) (tR) (tr) ½ + ½

(c) Phenotypic ratio

Tall Round : Tall Wrinkled : Dwarf Round : Dwarf Wrinkled ratio

9 : 3 : 3 : 1

Explanation : The Law of Independent Assortment states that when two pairs of traits are combined in a hybrid, segregation of one pair of character is independent of the segregation of other pair of characters.

(ii) Morgan observed the result of linkage of genes on a chromosome but Mendel did not observe phenomenon of linkage in pea plants /F_2 ratio of Morgan deviated significantly from 9 : 3 : 3 : 1 ratio (Mendelian ratio). **3**

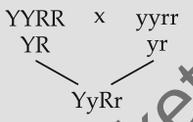
[CBSE Marking Scheme, 2015]

Q. 7. A pea plant producing yellow coloured and round seeds is given with unknown genotypes. Explain how you would find the correct genotypes of the plants with respect to the two traits mentioned. Work out the cross and name it.

A **[Outside Delhi Set-III, Comptt. 2015]**

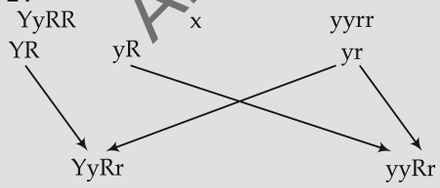
Ans. Test Cross **1**

Case 1 :



If all phenotypes is yellow and round then the genotype of parents is YYRR. (Homozygous) 1

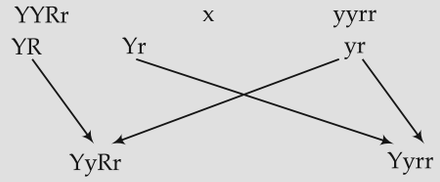
Case 2 :



Phenotypes : 1 Yellow round and 1 Green round

Then the genotype of the parent is YyRR (Heterozygous for yellowness) **1**

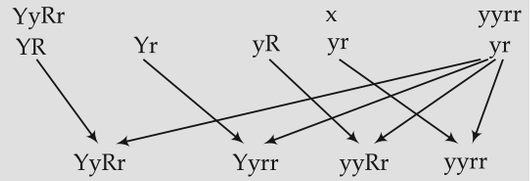
Case 3



Phenotype : 1 Yellow round and 1 Yellow wrinkled.

Then the genotype of parent is YYRr (Heterozygous for shape of seed) **1**

Case 4



Phenotype : 1 Yellow round , 1 Yellow wrinkled , 1 Green round and 1 Green wrinkled.

Then parent is YyRr

[CBSE Marking Scheme, 2015] 1

Q. 8. Explain the genetic basis of blood grouping in human population. U **[Delhi Set. III, Comptt. 2015]**

Ans. There are four types of blood groups in human population namely A, B, AB and O. They are determined by presence or absence of two types of RBC surface antigens / sugar polymer A and B. Individuals with blood group A have antigen A, while those with group B have antigen B, AB have both the antigens and 'O' persons do not have any antigen. The type of antigens and their presence or absence is controlled by gene I which has three alleles I^A, I^B and *i*. I^A produces antigen A, I^B antigen B whereas allele *i* (i°) does not form any antigen and is recessive. I^A and I^B are dominant over *i* and show dominant-recessive relationship. When I^A and I^B both are present together in a person, both express themselves equally, independently and produce the surface antigen A and B and therefore show the phenomenon of co-dominance. Such genes are called as co-dominant because human beings are diploid individuals, each person therefore have any two of these three alleles of gene I. This results into six different genotypic combination and four phenotypic expressions as follows :

Table showing genetic basis of blood groupings

Sr. No.	Blood Groups Phenotype	Possible genotypes
(i)	A	$I^A I^A$; $I^A i$
(ii)	B	$I^B I^B$; $I^B i$
(iii)	AB	$I^A I^B$
(iv)	O	*ii.*

Blood group alleles thus show both co-dominance and dominance relationship. **5**

Q. 9. Mendel published his work on inheritance of characters in 1865, but it remained unrecognized till 1900. Give three reasons for the delay in accepting his work.

U **[Outside Delhi Set-III, 2014; Delhi Set-I, 2011]**

OR

Although Mendel published his work on inheritance of characters in 1865 but for several reasons, it remained unrecognised till 1900. Explain giving three reasons why it took so long ?

[Delhi Set-I Comptt. 2016]

Ans. Work of Mendel could not be widely publicised due to poor communication / his concept of genes or factors as discrete units and which did not blend was not accepted due to continuous variation seen in nature/no proof of existence of factors and what they were made of /Mendel used mathematics to explain biological phenomenon which was new and unacceptable.

[CBSE Marking Scheme, 2016] 5

Detailed Answer :

The communication was not easy in those days and his work could not be widely publicised.

(i) His concept of genes as stable and discrete units that controlled the expression of traits and of the pair of alleles which did not 'blend' with each other was not accepted by contemporaries as an explanation for the apparently continuous variation seen in nature.

(ii) Mendel's approach of using statistical calculations to explain biological phenomena was totally new and unacceptable to many of the biologists of his time because they were beyond the comprehension of the biologists of the time.

(iii) Though Mendel's work suggested that factors (genes) were discrete units, he could not provide any physical proof for the existence of factors and what they were made of. 5

Q. 10. A cross was carried out between a pea plant heterozygous for round and yellow seeds with a pea plant having wrinkled and green seeds.

(i) Show the cross in Punnett square.

(ii) Write the phenotype of the progeny of this cross.

(iii) What is this cross known as ? State the purpose of conducting such a cross.

A **[Delhi Set-I, 2014]**

Ans. (i)

RrYy X rryy ½

Round Yellow Wrinkled Green

Gametes RY rY Ry ry ½

ry	RrYy	rrYy	Rryy	rryy	½ + ½

(ii) Round and yellow – 25% ½

Wrinkled and yellow – 25% ½

Round and green – 25% ½

Wrinkled and green – 25% ½

(iii) Test cross, to identify the genotype of unknown if it is homozygous dominant or heterozygous dominant.

[CBSE Marking Scheme, 2014] ½ + ½

Q. 11. Describe the mechanism of pattern of inheritance of ABO blood groups in humans.

U **[Delhi Set-I, 2014, Outside Delhi Set-III, 2014]**

Ans. Human blood group is determined by glycoprotein / antigen A and glycoprotein / antigen B. **1**

The alleles are I^A, I^B and i - Hence referred to as multiple allelism. **1**

The individual inherits any two of them as given below

$I^A I^A$, $I^A i$ — A group

$I^B I^B$, $I^B i$ — B group

$I^A I^B$ — AB group

i i — O group **1**

In the case of A, B and O — Law of dominance is the pattern of inheritance as I^A / I^B dominant over i. **1**

In AB group both the alleles I^A and I^B express — It is the case of Co-dominance. **1**

[CBSE Marking Scheme, 2014]

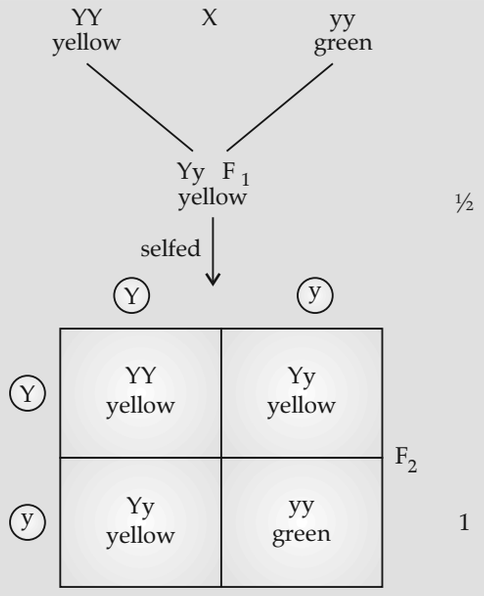
Q. 12. (i) Explain a monohybrid cross taking seed coat colour as a trait in *Pisum sativum*. Work out the cross upto F_2 generation.

(ii) State the laws of inheritance that can be derived from such a cross.

(iii) How is the phenotypic ratio of F_2 generation different in a dihybrid cross ?

U **[Outside Delhi Set-II, 2014]**

Ans. (i)



F_2 – Phenotypic ratio = 3 : 1

Genotypic ratio = 1 : 2 : 1 ½

(ii) Law of Dominance – In a contrasting pair of factors one member of the pair dominates (dominant) the other (recessive). **1**

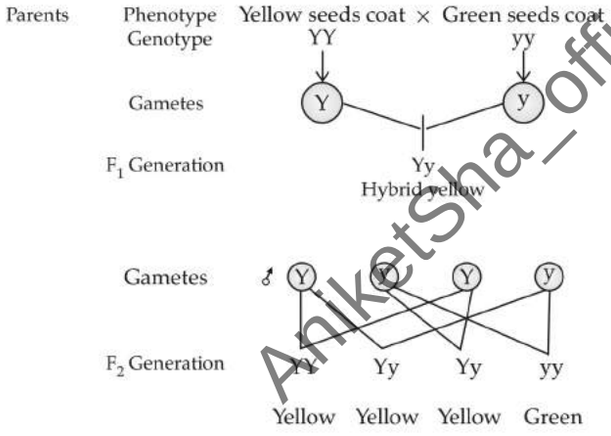
Law of Segregation : Factors or allele of pair segregate from each other so that a gamete receives only one of the two factors. **1**

(iii) Phenotypic ratio of F_2 in monohybrid cross is 3 : 1 whereas in a dihybrid cross the phenotypic ratio is 9 : 3 : 3 : 1.

[CBSE Marking Scheme, 2014] 1

Detailed Answer :

(i) Explanation of Monohybrid cross : The monohybrid cross is a cross in which the inheritance of a single pair of contrasting characters is taken into consideration. A cross is made between true breeding yellow seed coat colour variety and green seed coat colour variety of pea plants. All the hybrid offsprings obtained in F_1 generation are with yellow seed coat colour. This means the yellow seed coat colour is dominant over green seed coat colour as only this trait appeared while the masked manifestation of other trait is totally marked. When F_1 offspring hybrids were allowed for selfing the yellow seed coat plants and green seed coat plants appeared in the ratio of 3 : 1. The green seed coat plants are homozygous (yy) as they produced only green seed coat plants in successive generations when self pollinated. 1/3 of yellow seed coat plants also are homozygous (YY) because they produced only yellow seeded plants on selfing while 2/3 of yellow seeded plants are heterozygous (Yy) as they produced both yellow and green seeded plants on selfing. Thus the genotypic ratio is 1 : 2 : 1.



(ii) The following laws of inheritance can be derived from the monohybrid cross.

(a) Principle of dominance : During hybridization out of the two contrasting alleles / characters only one makes it appearance in the hybrid in F_1 generation while the manifestation of the other is masked. Then the character which appears in the hybrid or the allele which expresses its effect in the individual / hybrid is called as dominant while the other, which does not show its effect in the hybrid / heterozygous individual is called as recessive. This is universal phenomenon in all types of crosses and is called as the Principle of Dominance.

(b) Law of Segregation : This is the second law of Mendel. This law states that if hybrids of F_1 individuals are allowed for selfing the two expressions of a character separate out and appear in F_2 generation in a definite proportion of 3 : 1. This is called as monohybrid ratio or segregation ratio. The two alleles for a character in the hybrid individual do not blend or influence each other but separate during gamete formation and therefore gamete receive only one of the two factors but never both. The gametes are pure for a given character. This is known as the Law of Purity of Gametes.

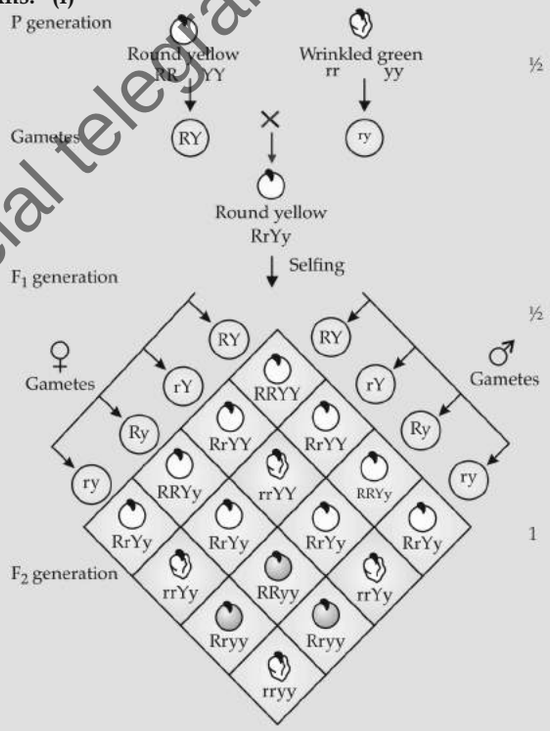
(iii) Refer to CBSE Marking Scheme (Answer).

Q. 13. (i) Explain Mendel's Law of Independent Assortment by taking a suitable example.

(ii) How did Morgan show the deviation in inheritance pattern in *Drosophila* with respect to this law ?

[Outside Delhi Set-III, 2013]

Ans. (i)



Phenotypic ratio : round yellow : round green : wrinkled yellow : wrinkled green

9 3 3 1 **1**

Law of Independent Assortment : It states that when two pairs of contrasting traits are combined in a hybrid, segregation of contrasting one pair of character is independent of the other pair of characters. **1**

(ii) Both parental type and recombinant types are observed to show that genes for the colour and genes for the shape of seeds segregate independently during gamete formation. **1**

[CBSE Marking Scheme, 2013]

Detailed Answer :

(i) Law of Independent Assortment : It states that when two individuals differing in two pairs of contrasting characters are crossed, the segregation of one pair of character into 3 : 1 ratio is independent of the segregation of another pair of contrasting characters into 3 : 1 ratio and also some non-parental new recombination of character also appear in the ratio of 9 : 3 : 3 : 1 in F_2 generation as shown in the above cross. This is because the alleles of different characters are located on different pairs of homologous chromosomes and that they are independent from one another in their segregation during gamete formation.

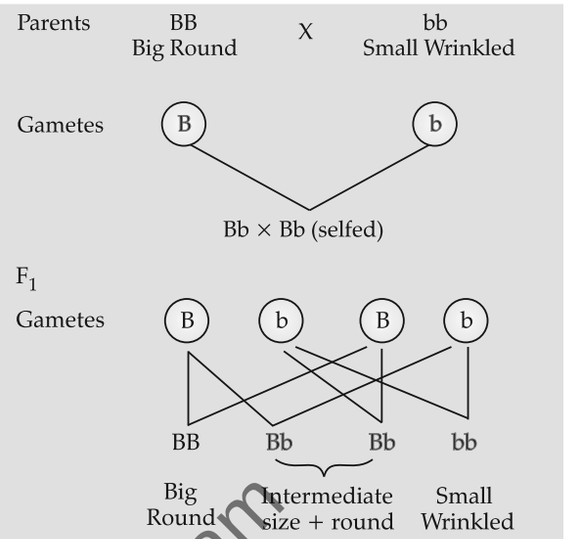
(ii) Morgan however observed the deviation in inheritance pattern in *Drosophila* with regard to this law. He observed that in *Drosophila* the F_2 ratio in a dihybrid cross deviates significantly from 9 : 3 : 3 : 1 ratio. This is because the genes were linked. They are located on the same chromosome and therefore inherited together. Because of this the parental types observed in F_2 generation were greater than the new recombinations. Linkage was not observed by Mendel because the characters which he had chosen were not linked as their genes are located on different chromosome and when on the same chromosome they were quite apart.

Q. 14. What is the inheritance pattern observed in the size of starch grains and seed shape of *Pisum sativum* ? Work out the monohybrid cross showing the above traits. How does this pattern of inheritance deviate from that of Mendelian Law of Dominance ?

A **[Delhi Set-II, 2012]**

Ans. A single gene controls the size of the starch grain and the seed shape.

The trait of size of starch grain shows incomplete dominance. Hence in heterozygous condition the starch grain are of intermediate size.



(Deviation from Mendelian Law of Dominance) : The trait of seed shape follows Law of Dominance and the hybrid will show only dominant trait. **5**

[CBSE Marking Scheme, 2012]

Detailed Answer :

The starch synthesis in pea plant is controlled by a single gene. This gene in pea plant shows some degree of pleiotropy as it controls the shape of the seed and in addition the size of starch grain too. This gene has two alleles B and b. The BB homozygotes produce large starch grains as compared to those produced by bb homozygotes. Mature homozygous BB seeds were round while bb seeds were wrinkled. The heterozygotes Bb form round seeds but the starch grains were of intermediate size. Thus if the size of starch grain is considered the Bb seeds show the phenomenon of incomplete dominance, but if seed shape is considered then allele B and b show dominant-recessive relationship. Thus in this case the pattern of inheritance deviates from the Mendelian Law of Inheritance in that here the dominance of an allele is not absolute but depends upon the product and the particular phenotype that it forms.

Q. 15. (i) Explain Polygenic and Multiple allelism with the help of suitable examples.

(ii) "Phenylketonuria is a good example that explains Pleiotropy." Justify. A **[Outside Delhi - 2017 Set - II]**

Ans. **(i)** Traits that are generally controlled by three or more genes, the phenotype reflects the contribution of each allele / effect of each allele is additive. ½ + ½

eg. Human skin colour, controlled by three genes (A, B, C). ½ + ½

In multiple allelism more than two alleles, govern the same character / phenotype. ½ + ½

eg. Human blood group (ABO system), controlled by three different alleles (I^A, I^B, i). ½ + ½

(ii) In pleiotropy a single gene can exhibit multiple phenotypic expressions, in phenylketonuria single mutated gene express mental retardation and reduction in hair and skin pigmentation. ½ + ½

[CBSE Marking Scheme, 2017]

Detailed Answer :

(i) In case of polygenic inheritance, traits are controlled by three or more genes (multiple genes). The phenotype is produced as a result of participation of several genes. For example, human skin colour is controlled by three genes (A, B, C).

In case of multiple allelism, more then two alleles are present for a character. For example, human blood group system (ABO). In this case, more than two *i.e.,* three alleles are governing the same character.

(ii) Pleiotropy is the phenomenon in which a single gene exhibit multiple phenotypic expression. In phenyketonuria, a single gene mutation leads to multiple phenotypic expression *i.e.,* hair and skin pigmentation and mental problems.

OR

Ans.

a) Polygenic inheritance is the inheritance of characters which are controlled by 3 or more genes. Such characters do not have two alternative forms but are spread across a gradient. Such characters are called polygenic traits or quantitative traits. They also take into account the influence of environment conditions. In such inheritance, the phenotype reflects the contribution of each allele ie, the effect of each allele is additive. Hence these genes are called additive or cumulative gene

eg:- Skin colour in humans is a polygenic trait. Consider that 3 genes A, B and C consider the skin colours whose dominant forms are A, B, C while recessive forms are a, b, c.
A person with all dominant alleles (genotype AABBCC) will have the darkest skin colour - negro. A person with all recessive alleles ie, aabbcc will have lightest skin colour - white. A person with 3 dominant

(AaBbCc)
and 3 recessive alleles will have intermediate skin colour - mulatto.
A cross between AABBCC and aabbcc will result in a progeny generation with 7 phenotypes of phenotypic ratio 1:6:15:20:15:6:1 corresponding to black, very dark, dark, mulatto, light, very light, white phenotypes. Thus the number of dominant and recessive alleles determines the skin colour.

Multiple allelism is the phenomenon in which more than 2 alleles are present for a character in a population
eg:- ABO Blood grouping in man.
The blood group is determined by the type of sugar polymer protruding from the surface of plasma membrane of red blood cells. The type of sugar is controlled by 'I' gene. It has 3 alleles - I^A, I^B and i. I^A and I^B produce slightly different forms of sugars while 'i' do not produce any sugar. Being diploid an individual can have only 2 of the 3 alleles. I^A and I^B are completely dominant over 'i' i.e., when

I^A and I^B i are present together, only I^A expresses since 'i' do not produce any gene. Similarly I^B is expressed in presence of i. But when both I^A and I^B are present together, both of them expresses. Thus blood cell has both type of sugars. Thus this is an example ~~for total~~ of codominance – where both alleles expresses equally and the phenotype resembles both the parents. In total, there are six genotypes and 4 phenotypes.

Genotype	Phenotype (Blood group)
I^A i	A
I^A I^A	A
I^B i	B
I^B I^B	B
I^A I^B	AB
ii	O

The polygenic inheritance differs from multiple alleles in the fact that the different polygenes may be located on different loci on same or different homologous chromosomes while in multiple alleles, the alleles are present on the same loci on homologous chromosomes.

b) Pleiotropy refers to the multiple phenotypic effect of a gene. It arises due to the effect of a gene on metabolic pathway controlling different phenotypes.
Phenylketonuria is caused due to the mutation of a gene causing the lack of an enzyme phenylalanine hydroxylase. This results in the accumulation of the enzyme and its derivatives in brain. This is manifested in the form of phenotypic effects such as retardation of mental development, or reduced skin and hair pigmentation. Thus the gene seems to affect different phenotypes. Hence this is an example of pleiotropy.

[Topper's Answer, 2017]

Answering Tip

- Learn main laws, principles and key-words/ acronyms with proper understanding.

Q. 16. (i) Work out a dihybrid cross upto F_2 generation between pea plants bearing violet coloured axial flowers and white coloured terminal flowers. Give their phenotypic ratio.

(ii) State the Mendel's law of inheritance that was derived from such a cross.

A **[Outside Delhi Comptt. 2017, Set - III]**

Ans. (i)

Parents : V V A A (Violet, axial) × v v a a = (1) (White, terminal)

gametes : VA, va

F_1 VvAa (Violet axial) 1

	VA	vA	Va	va
VA	VVAA Violet axial	VvAA Violet axial	VVAa Violet axial	VvAa Violet axial
vA	VvAA Violet axial	vvAA White axial	VvAa Violet axial	vvAa White axial
Va	VVAa Violet axial	VvAa Violet axial	VVaa Violet terminal	Vvaa Violet terminal
va	VvAa Violet axial	vvAa White axial	Vvaa Violet terminal	vvaa White terminal

1

Phenotypes – violet axial : white axial : violet terminal : white terminal

Phenotype ratio – 9 : 3 : 3 : 1 1

(ii) Law of Independent Assortment : When two pairs are combined in a hybrid segregation of one pair of characters is independent of the other pair of characters. 1

[CBSE Marking Scheme, 2017]

Q. 17. State and explain the "law of independent assortment" in a typical Mendelian dihybrid cross. U **[Delhi 2017, Set – I, II, III]**

OR

Work out a typical Mendelian dihybrid cross and state the law that he derived from it.

[Delhi Set-II, 2014]

OR

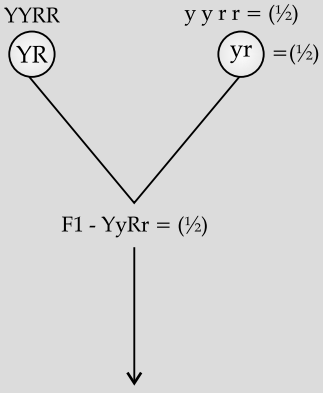
(i) State the Law of Independent Assortment.

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

[Outside Delhi Set-I, 2010]

Ans. (i) Law of Independent Assortment : When two pair of traits are combined in a hybrid, inheritance of one pair of characters is independent of the other pair of characters / when two pairs of contrasting characters or genes or traits are inherited together in a dihybrid cross (in a pea plant) the inheritance of one pair of character is independent of inheritance of the other character in the progeny. **1**

Explanation : Mendel took homozygous pea plant producing yellow and round seeds, crossed them with homozygous pea plant producing green and wrinkled seeds / shown in a flow chart of a dihybrid cross given.



	YR	Yr	yR	yr (=½)
YR	YYRR	YYRr	YyRR	YyRr
Yr	YYRr	YYrr	YyRr	Yyrr
yR	YyRR	YyRr	yyRR	yyRr
yr	YyRr	Yyrr	yyRr	yyrr

(½) (½)

Phenotypes – Yellow : Yellow : Green : Green round wrinkled round wrinkled

Phenotype ratio – 9 : 3 : 3 : 1

(Four different types of phenotypes in correct ratio) ½ + ½

(Formation of new phenotypes along with parental phenotypes is possible because inheritance of two pairs of contrasting traits or genes in the progeny is independent of each other) **4 + 1 = 5**

[CBSE Marking Scheme, 2017]

Q. 18. (i) How are polygenic inheritance and multiple allelism different ? Explain with the help of an example each.

(ii) List the criteria a chemical molecule must fullfil to be able to act a genetic material.

A **[Delhi Comptt. 2017, Set - II, III]**

Ans. (i)

Polygenic Inheritance	Multiple Allelism
Controlled by three or more genes	More than two alleles govern the same character
Example : A - B - C gene control human skin colour	**Example :** ABO blood grouping in humans = 2

2

(ii) (a) It should be able to generate its replica / replication.

(b) It should be chemically and structurally stable.

(c) It should provide the scope for slow changes / mutation that are required for evolution.

(d) It should be able to express itself in the form of a Mendelian characters.

(Any three) 1 × 3=3

[CBSE Marking Scheme, 2017]

Q. 19. Skin colour in humans does not have distinct alternate forms but shows a whole range of possible variations in skin colour. Explain the pattern of inheritance of such a triait. What is this type of inheritance known as ? Provide another example of exhibiting such an inheritance pattern.

A **[Outside Delhi Comptt. 2017, Set - II]**

Ans. Skin colour is controlled by three genes; A, B, C dominant genes and a, b, c the recessive genes; the effect of each type of allele is additive; more dominant allele, darker the skin colour; more the recessive allele, lighter the skin colour; when three dominant alleles and three recessive alleles are present in an individual the skin colour is intermediate. **½ × 6**

(i) Polygenic inheritance **1**

(ii) Human Height / or any other correct example. **1**

[CBSE Marking Scheme, 2017]

Detailed Answer :

Human skin colour is caused by pigment called melanin. The quantity of melanin is due to three pairs of polygenes (A, B and C). If black or very dark (AA, BB, CC) and very light (aa, bb, cc) individual marry, the offspring show intermediate colour (Aa, Bb, Cc). Such type of inheritance is called polygenic inheritance because a phenotype is controlled by one or more genes and show the cumulative effect.

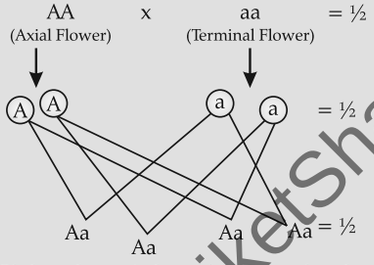
Another example is : In cotton, a gene for the lint also influences the height of plant, size of ball, number of ovules and viability of seeds.

Q. 20. (i) A pea plant bearing axial flowers is crossed with a pea plant bearing terminal flowers. The cross is carried out to find the genotype of pea plant bearing axial flowers. Work out the cross to show the conclusions you arrive at.

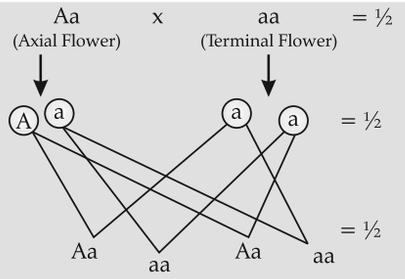
(ii) State the Mendel's law of inheritance that is universally acceptable.

A **[Outside Delhi - 2017, Set-II]**

Ans. (i) If the plants is homozygous for the dominant trait



(ii) If the plants is heterozygous for the dominant trait



(50% plants are with Axial flower and 50 % plants with terminal flower)

Conclusion : If all progeny show axial flowers (dominant) the plant is homozygous (AA), If 50% of progeny show Axial flower (Dominant) and 50% Terminal flower (Recessive) the plant is heterozygous. ½ + ½

(ii) Law of Segregation : allelic pair segregate (separates) during gamete formation (do not loose their identity). ½ + ½

[CBSE Marking Scheme, 2017] 5

AI **Q. 21. 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 ?**

A **[Foreign Set - I, II, III , 2017]**

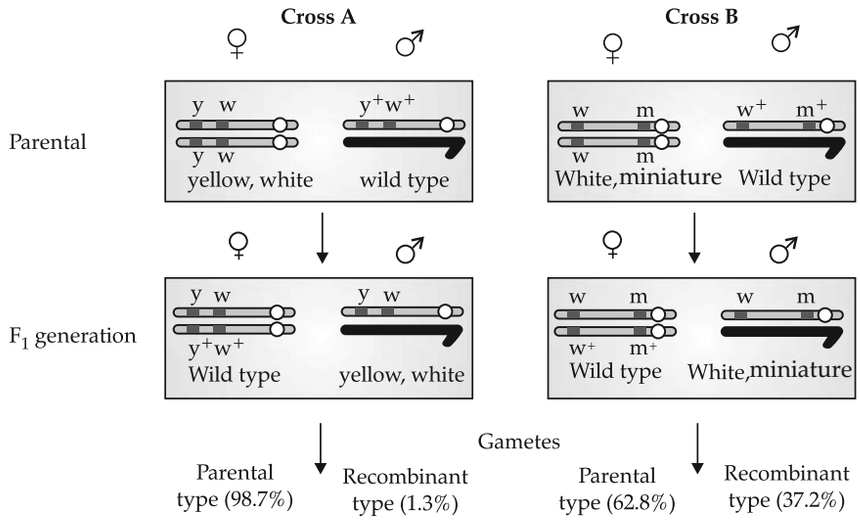
Ans. According to Morgan and his group, if genes are tightly linked they showed very low recombination. **1**

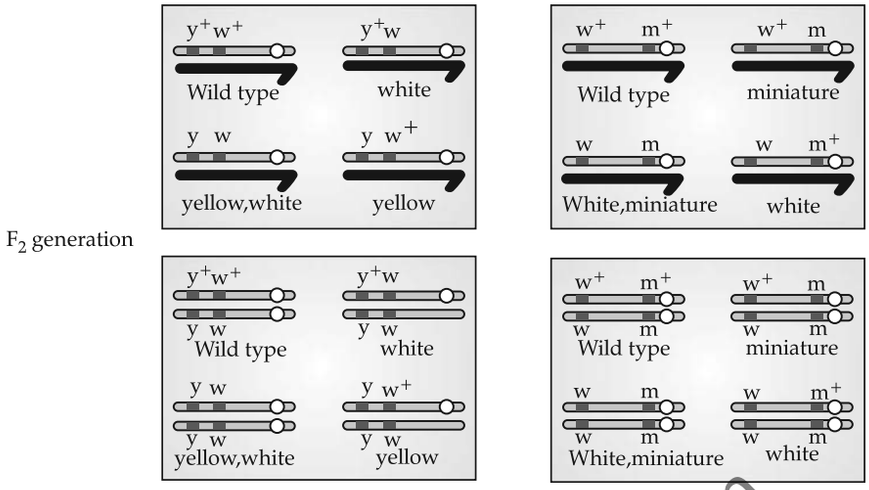
(shown in cross A) **1**

If genes were loosely linked they showed very high recombination. **1**

(shown in cross B) **1**

The group used the frequency of recombination between gene pairs on the same chromosome as a measure of distance between genes and 'mapped' their position on the chromosome. **1**





Q. 22. In a dihybrid cross, white eyed, yellow bodied female *Drosophila* was crossed with red eyed, brown bodied male *Drosophila*. The cross produced 1.3 percent recombinants and 98.7 progeny with parental type combinations in the F_2 generation. Analyze the above observation and compare with the Mendelian dihybrid cross.

A **[CBSE SQP, 2018]**

Ans. Morgan observed that the two genes did not segregate independently of each other and the F_2 ratio deviated very significantly from the 9:3:3:1 ratio. **1**

He attributed this to physical association or linkage of two genes, coined the term linkage and the term recombination to describe the generation of non-parental gene combinations. **1**

Morgan and his group found that even when the genes are grouped on the same chromosome, some genes are very tightly linked (show very low recombination) while others were loosely linked (showed higher recombination). **1**

In the Mendelian dihybrid cross, the phenotypes round, yellow; wrinkled, yellow; round, green and wrinkled, green appeared in the ratio 9:3:3:1. OR cross (given below). **1**

Wrinkled, yellow and round, green is possible because the distance between two genes are more. Therefore, recombination of parental type is possible. **1**

For flow chart: Refer- LAQ/Q. 17

[CBSE Marking Scheme, 2018]

Q. 23. (a) Write the scientific name of the organism Thomas Hunt Morgan and his colleagues worked with for their experiments the correlation between linkage and recombination with respect to genes as studied by them.

(b) How did Sturtevant explain gene mapping while working with Morgan

R **[Outside Delhi/Delhi, 2018]**

Ans. (a) *Drosophila melanogaster* **1**

They 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). =½ Tightly linked genes tend to show fewer (lesser) recombinant frequency of parental traits / show higher (more) frequency of parental type. ½

Loosely linked genes show higher percentage (more) of recombinant frequency of parental traits / lower frequency percentage of parental type ½

Genes present on same chromosome are said to be linked and the recombinant frequency depends on their relative distance on the chromosome. ½

(b) He 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. **1+1**

[3+2=5 Marks]

[CBSE Marking Scheme, 2018]

Detailed Answer:

(a) Thomas Hunt Morgan and his colleagues used fruit fly or *Drosophila melanogaster* to study linkage.

Morgan carried out several dihydric crosses in *Drosophila* to study sex-linked genes. In one such experiment, he crossed yellow-bodied, white-eyed females with brown-bodied, red-eyed males (wild type). He found that the :

Two genes did not segregate independently of each other and the F_2 ratio deviated from the 9:3:3:1 ratio, (expected when the two genes are independent).

Morgan attributed this due to the physical association or linkage of the two genes and coined the term linkage. He stated that higher the linkage between two, lesser are the chances of recombination.

(b) Alfred Sturtevant used the recombination frequency between gene pairs as a measure of physical distance between genes and 'mapped' their position on the chromosome. This process

of mapping, which are used today for genome sequencing projects as in Human Genome Project.

3+2

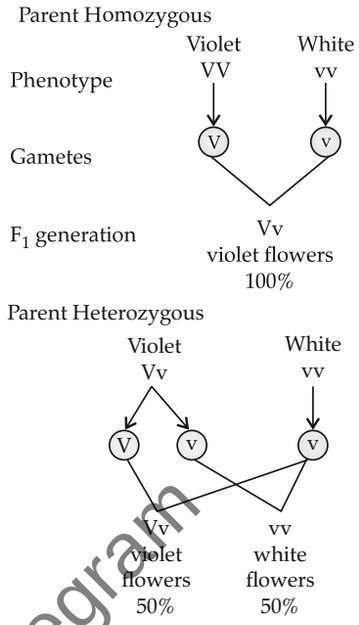
Q. 24. A particular garden pea plant produces only violet flowers

(i) Is it homozygous dominant for the trait or heterozygous ?

(ii) How would you ensure its genotype ? Explain with the help of crosses. R **[Outside Delhi 2009]**

Ans. (i) The plant should be homozygous dominant as it produces violet flowers only.

(ii) Its genotype can be ensured by performing the test cross. In this cross if all the F_1 plants obtained (100%) are with violet flowers then it is homozygous & if the violet & white flowers appear in 1:1 ratio then the plant is heterozygous (Vv). Test crosses can be shown as follows :



5

TOPIC-2
Sex Determination and Chromosomal Disorder

Revision Notes

➢ **Sex determination**

- The method by which the distinction between male and female is established in a species is called sex determination.
- Sex of an individual is finalized at the time of zygote formation.

➢ **Autosomes and Sex chromosomes (allosomes)**

- Autosomes are chromosomes other than sex chromosomes. They contain genes which determine somatic characteristics.
- Number of autosomes is same in males and females.
- Sex chromosomes (X & Y) are the chromosomes which are involved in sex determination.
- **Henking (1891)** studied spermatogenesis in some insects and observed that 50 % of sperm received a nuclear structure after spermatogenesis, whereas other 50 % sperm did not receive it.
- Henking called this structure as the **X body** (later it was called as **X-chromosome**).

➢ **Mechanism of Sex Determination**

(i) Chromosomal sex determination : It is based on heterogamety i.e., occurrence of two types of gametes in one of the two sexes. It is of following types :

(a) XX-XO mechanism :

Here, male is heterogametic *i.e.* XO besides autosomes (Gametes with X and gametes without X) and female is homogametic *i.e.* XX (all gametes are with X chromosomes) *e.g.* Many insects such as grasshopper.

(b) XX-XY mechanism :

Male is heterogametic (X & Y) and female is homogametic (X only). *e.g.* Human and *Drosophila.*

(c) ZZ-ZW mechanism :

Male is homogametic (ZZ) and female is heterogametic (Z & W). *e.g.* Birds.

(d) ZO-ZZ mechanism : Females have only Z-chromosomes besides autosomes and males have a pair of Z-chromosomes *e.g.* in cockroaches.

XX-XO & XX-XY mechanisms show male heterogamety. ZZ-ZW mechanism shows female heterogamety.

Females have only Z chromosome besides autosomes and males have a pair of Z chromosome as seen in cockroaches.

➢ **Sex Determination in Humans (*XX-XY* type)**
- Human has 23 pairs of chromosomes (22 pairs are autosomes and 1 pair is sex chromosomes).
- A pair of X-chromosomes (XX) is present in the female, whereas X and Y chromosomes are present in male.
- During spermatogenesis males produce 2 types of gametes *i.e.*, 50 % with X-chromosome and 50 % with Y-chromosome.
- Females produce only ovum with an X-chromosome.
- There is an equal probability of fertilization of the ovum with the sperm carrying either X or Y chromosome.
- The sperm determines whether the offspring will be male or female.

(ii) Environmental Sex-determination : Determination of sex depends upon the environmental condition. The environmental factors like temperature etc. determine whether the zygote will develop into male or female. *e.g.* turtles and crocodile.

(iii) Genetic balance mechanism of sex determination : Sex of the individual is decided by the ratio of X-chromosome and autosome as is found in ***Drosophila***.

(iv) Cytoplasmic Sex-determination : Cytoplasmic or fertility factor called as F^+ factor located in plasmid determines the sex as in found is some bacteria.

➢ **Mutation**
- It is a sudden heritable change in DNA sequences resulting in changes in the genotype and the phenotype of an organism. The term mutation was given by Hugo de vries (1901).
- It is caused either by loss or gain or change in a single base pair of DNA.
- **Frame-shift mutation :** Loss (deletions) or gain (insertion/ duplication) in DNA segment so that the whole frame of codons is changed.
- **Point mutation :** Mutation due to change in a single base pair of DNA. *e.g.* sickle cell anaemia.
- Mutation results in Chromosomal abnormalities (aberrations).
- Chromosomal aberrations are seen in **cancer cells**.
- **Mutagens** (agents which induce mutation) include,
  - **(a) Physical mutagens:** UV radiation, α, β, γ rays, X-ray, etc.
  - **(b) Chemical mutagens:** Mustard gas, phenol, formalin, etc.

➢ **Pedigree Analysis**
- The representation or chart showing family history is called **family tree (pedigree).**
- Thus, analysis of traits in several generations of a family is called **pedigree analysis.**
- In humans, control crosses are not possible.
- So, the study of family history about inheritance is used.
- In human genetics, pedigree study is utilized to trace the inheritance of a specific trait, abnormality or disease.

➢ **Genetic Disorders**
- There are two types of genetic disorders namely, Mendelian disorders and Chromosomal disorders.

(1) Mendelian Disorders
- It is caused by alteration or mutation in the single gene.
- The pattern of inheritance of Mendelian disorders can be traced in a family by the pedigree analysis.
  e.g. Haemophilia, Cystic fibrosis, Sickle-cell anaemia, Colour blindness, Phenylketonuria, Thalassemia, etc.
- Mendelian disorders may be dominant or recessive.
- By pedigree analysis one can easily understand whether the trait is dominant or recessive.

➢ **Pedigree Analysis of Autosomal Dominant Trait** *e.g.* – Myotonic dystrophy

(a) Myotonic Dystrophy
- It is an autosomal dominant disorder which is characterized by increasing contractility of muscles with decreasing relaxation. This leads to atrophy of muscles particularly of face and neck. Hypogonadism, balding and cardiac irregularities may also be caused due to this disorder.

➢ **Pedigree Analysis of Autosomal Recessive Trait** *e.g.* – Sickle cell anaemia

(a) Sickle-cell Anaemia
- This is an autosome linked recessive trait.
- It can be transmitted from parents to the offspring when both the partners are carrier for the gene (or heterozygous).
- The disease is controlled by a pair of allele, Hb^A and Hb^S.
- Homozygous dominant (Hb^AHb^A) : normal Heterozygous (Hb^AHb^S): carrier; sickle cell trait Homozygous recessive (Hb^SHb^S) : affected

- The defect is caused by the substitution of Glutamic acid (Glu) by Valine (Val) at the sixth position of the β-globin chain of the haemoglobin (Hb).
- This is due to the single base substitution at the sixth codon of the β-globin gene from GAG to GUG.
- The mutant Hb molecule undergoes polymerization under low oxygen tension causing the change in shape of the RBC from biconcave disc to elongated sickle like structure.

(b) Haemophilia (Royal disease)

- Sex linked recessive disease.
- In this, a protein involved in the blood clotting is affected.
- A simple cut results in non-stop bleeding.
- The heterozygous female (carrier) for haemophilia may transmit the disease to sons.
- The possibility of a female becoming a haemophilic is very rare because mother has to be at least carrier and father should be haemophilic (unavailable in the later stage of life).
- Queen Victoria was a carrier of the disease. So her family pedigree shows a number of haemophilic descendents.

(c) Phenylketonuria

- An inborn error of metabolism.
- Autosomal recessive trait.
- The affected individual lacks an enzyme (phenylalanine hydroxylase) that converts the amino acid phenylalanine into tyrosine.
- As a result, phenylalanine accumulates and converts into phenyl pyruvic acid and other derivatives.
- They accumulate in brain resulting in mental retardation.
- These are also excreted through urine because of poor absorption by kidney.

(2) Chromosomal Disorders

- They are caused due to absence or excess or abnormal arrangement of one or more chromosomes.
- These are of two types namely,
  (a) Aneuploidy **(b)** Euploidy.

(a) Aneuploidy

- The gain or loss of chromosomes due to failure of segregation of chromatids during cell division.
  It includes,
  (a) Nullisomy (2n-2): A complete homologous pair is lost from diploid set.
  (b) Monosomy (2n-1): One chromosome is lost from diploid set.
  (c) Trisomy (2n+1): One chromosome is added to diploid set, so that one chromosome occurs in triplicate.
  (d) Tetrasomy (2n+2): 2 chromosomes are added to diploid set, so that a chromosome is found in quadripulate.

(b) Polyploidy (Euploidy)

- It is an increase in number of chromosomes sets beyond diploid X condition (2n).
- This is often seen in plants.
- On the basis of number of chromosome sets, the polyploids are of following types : triploids (3n), tetraploids (4n), pentaploids (5n), hexaploids (6n), etc.
  (a) Autopolyploidy : It is an increase in number of the same genome. *e.g.* AAA (autotriploid), AAAA (autotetraploid), etc.
  (b) Allopolyploidy : It is the increase in number of sets of chromosome due to coming together of diploid genomes of two or more than two individuals of different species. *e.g.* AABB, AABBDD. Bread wheat is allohexaploid (AABBDD). Triticale is the man made cereal formed by hybridization between durum wheat and rye. It is allohexaploid.
- **Autoallopolyploidy :** It is a kind of polyploidy where the genomes of two species come together in which one has double set of chromosomes. *e.g. Helianthus tuberosus* which is autoallohexaploid.
- **Chromosomal aberrations :** These are the changes in morphology and structure of chromosome resulting in the change in number and sequence of genes on them without any change in ploidy. They are of following types :
  1. **Deletion :** It is the loss of a terminal segment of a chromosome or from within the a chromosome (interstitial segment) followed by reunion of its remaining parts.
  2. **Inversion :** It is a change in a chromosome architecture due to breaking up, rotation through 180° of a segment and its reunion so that sequence of genes is reversed in the inverted region.

3. **Duplication :** It is a change in chromosome structure in which a part of chromosome breaks up and unites with another homologous chromosome. This process repeats the chromosome segments because the same block of genes is present more than once in a haploid component.
4. **Translocation :** It is a change in chromosome architecture which is due to breaking up of segment of chromosome and its union with another non-homologous chromosome. It may also be due to mutual exchange of chromosomal segments between non-homologous chromosomes.

➢ **Examples for Chromosomal Disorders**

(a) Down's Syndrome (Mongolism) :

- It is the presence of an additional copy of chromosome number 21 (trisomy of 21).
- Genetic constitution : 45 A + XX or 45 A + XY (*i.e.* 47 chromosomes).
- **Features :**
  - **(a)** They are short statured with small round head.
  - **(b)** Broad flat face.
  - **(c)** Furrowed big tongue and partially open mouth.
  - **(d)** Many "loops" on finger tips.
  - **(e)** Palm is broad with characteristic palm crease.
  - **(f)** Retarded physical, psychomotor & mental development.
  - **(g)** Congenital heart disease.

(b) Klinefelter's Syndrome :

- It is the presence of an additional copy of X-chromosome in male.
- Genetic constitution: 44 A + XXY (*i.e.* 47 chromosomes).
- **Features :**
  - **(a)** Overall masculine development however the feminine development is also expressed. *e.g.* development of breast (Gynaecomastia).
  - **(b)** Sterile.
  - **(c)** Mentally retarded.

(c) Turner's Syndrome :

- This is due to the absence of one of the X chromosomes in female.
- Genetic constitution: 44 A + XO (*i.e.* 45 chromosomes).
- **Features :**
  - **(a)** Sterile, Ovaries are rudimentary.
  - **(b)** Lack of other secondary sexual characters.
  - **(c)** Dwarf.
  - **(d)** Mentally retarded.

Very Short Answer Type Questions (1 mark each)

Q. 1. A male honeybee has 16 chromosomes whereas its female has 32 chromosomes. Give one reason.

A **[Outside Delhi Set-I, 2016]**

Ans. Male honey bee develops from unfertilized female gamete/unfertilised egg/ Parthenogenesis of female gamete (16 chromosomes), female develops by fertilization/fertilised egg. (32 chromosomes).

[CBSE Marking Scheme, 2016] ½ + ½

Detailed Answer :

Male honey bees are borne from the unfertilised eggs by the process known as parthenogenesis whereas female honeybees are borne from fertilised egg. Since, unfertilised egg carries only half the number of chromosomes as compared to fertilised egg, male honeybees have half the number of chromosomes (n) as compared to female honey bee (2n).

Answering Tip

- Carefully understand the process of sex determination in honey bees, *Drosophila* and humans. Don't get confused between the three.

Commonly Made Error

- Mostly students did not understand the concept of haploid and diploid.

Q. 2. Give an example of a human disorder that is caused due to a single gene mutation.

A **[Delhi Set-II, 2016]**

Ans. Sickle cell anaemia/Thalassemia/Phenylketonuria.

[CBSE Marking Scheme, 2016] 1

Commonly Made Error

- Spelling error is commonly seen. Learn spellings of disorder carefully.

Q. 3. What is point mutation? Give one example.

R **[(KVS, NCERT) Foreign Set-I,2016]**

Ans. Arising due to change in a single base pair of DNA, sickle cell anaemia. **1**

[CBSE Marking Scheme, 2016]

Detailed Answer :

Point mutation is a gene mutation, which arises due to a change in a single base pair of DNA. *e.g.* sickle cell anaemia.

Q. 4. What is a Mutagen? Name a physical factor that can be a Mutagen. R **[Foreign Set-II, 2016]**

Ans. All the physical and chemical factors that induce mutation, UV radiation/X rays. **1**

[CBSE Marking Scheme, 2016]

Detailed Answer :

All the physical and chemical factors that induce mutation are called mutagens.

The physical factors which can be a mutagen are the ionizing radiations like X-rays, gamma rays & the non-ionizing radiations like ultraviolet rays.

Q. 5. Mention two causes of frame-shift Mutation.

R **[Foreign Set-III, 2016]**

Ans. Insertion, deletion of three bases/one codon or multiple of three bases/multiple codon (hence one or more amino acid) (reading frame remains unaltered from that point onwards) **1**

[CBSE Marking Scheme, 2016]

Detailed Answer :

Frame-shift mutations are caused by addition or deletion of nitrogenous bases in the DNA or mRNA. These mutations are so called because these shift the reading frame of codons from the site of change onwards. Deletion involves the loss of one or more nucleotides while addition or insertion involves the addition of one or more extra nucleotides in the DNA molecule resulting in shift in reading the frame of codons.

Commonly Made Error

- Students often write definition instead of causes of frameshift mutation.

Q. 6. Give an example of a sex-linked recessive disorder in humans. A **[Delhi Set-II, Comptt. 2016]**

Ans. Colour blindness. **1**

Q. 7. Write the chromosomal basis of sex determination in birds. R **[Outside Delhi Set-I, Comptt. 2016]**

Ans. ♂ (Male) ZZ

♀ (Female)ZW/Heterogamety **1**

[CBSE Marking Scheme, 2016]

Commonly Made Error

- Students often get confused between the combination of sex chromosomes in male and female.

Answering Tip

- Make sure, you are thorough about the concept of sex chromosomes in various animals.

Q. 8. Give an example of an organism that exhibits haplodiploid sex-determination system.

A **[Outside Delhi Set-II, Comptt. 2016]**

Ans. Honey bees.

[CBSE Marking Scheme, 2016] 1

Q. 9. Give one example of organism exhibiting female heterogamety.

A **[Outside Delhi, Set-III, Comptt. 2016]**

Ans. In many birds (ZZ / ZW)

Male / female/heterogamety **1**

[CBSE Marking Scheme, 2016]

Detailed Answer :

Birds show female heterogamety as the female birds produce two different type of ova *i.e.* (A+Z) and (A + W) whereas male produces only one type of sperms. **1**

AI **Q. 10. Indiscriminate diagnostic practices using X-rays etc., should be avoided. Give one reason.**

A **[Delhi, Set-III, 2015]**

Ans. Indiscriminate diagnostic practices using X-rays, gamma rays etc. are ionizing radiations which usually produce breaks in the chromosomes and chromatids and abnormal mitosis in the irradiated cells. They cause abnormal functioning of the cells, mutations resulting in the development of various types of cancers specially blood cancer or leukemia etc. **1**

Q. 11. State the chromosomal defects in individuals with Turner's syndrome. U **[Delhi Set-I, Comptt. 2015]**

Ans. Monosomy of sex chromosome/XO condition/ Absence of one X chromosome (in female). **1**

[CBSE Marking Scheme, 2015]

Q. 12. Write the chromosomal defect in individuals affected with Klinefelter's syndrome.

R **[Outside Delhi Set-I, Comptt. 2015]**

Ans. Male - Additional copy of X chromosome / XXY. **1**

[CBSE Marking Scheme 2015]

Q. 13. On what basis is the skin colour in humans considered polygenic ?

R **[Outside Delhi Set-III, Comptt. 2015]**

Ans. Controlled by more than one gene, cumulative and additive effect of genes. **1**

[CBSE Marking Scheme, 2015]

Q. 14. How many chromosomes do drones of honeybee possess ? Name the type of cell division involved in the production of sperms by them.

U **[Outside Delhi Set-I, 2015]**

Ans. 16, Mitosis. **[CBSE Marking Scheme, 2015]** ½+½

AI **Q. 15. Give an example of a chromosomal disorder caused due to non-disjunction of autosomes.**

A **[CBSE SQP, 2015]**

Ans. Down's syndrome. **1**
[CBSE Marking Scheme, 2015]

Q. 16. Why do normal red blood cells become elongated sickle shaped structures in a person suffering from sickle cell anaemia?

U **[Delhi Set-II, III, 2014]**

Ans. The mutant haemoglobin molecule (substitution of Glutamic acid by valine) undergoes polymerization, under low oxygen tension causing the change. ½ + ½
[CBSE Marking Scheme, 2014]

Detailed Answer :

The defect is caused by the substitution of Glutamic acid (Glu) by Valine (Val) at the sixth position of the beta globin chain of the haemoglobin molecule. The substitution of amino acid in the globin protein results due to the single base substitution at the sixth codon of the beta globin gene from GAG to GUG. The mutant haemoglobin molecule undergoes polymerisation under low oxygen tension causing the change in the shape of the RBC from biconcave disc to elongated sickle like structure. **1**

Q. 17. Name one autosomal dominant and one autosomal recessive Mendelian disorder in humans.

R **[Outside Delhi Set-I, 2010]**

Ans. Autosomal dominant — Myotonic dystrophy ½
Autosomal recessive — Phenylketonuria / sickle cell anaemia / cystic fibrosis / Thalassaemia. ½
[CBSE Marking Scheme, 2010]

Q. 18. The son of a haemophilic man does not get this genetic disorder. Mention the reason.

A **[Delhi Set-I, Comptt. 2010]**

Ans. The son gets Y chromosome from the father and X chromosome from the mother. Therefore, as the gene for haemophilia is located on X chromosome, a son cannot get the disease from his father. **1**

Q. 19. What is the difference in the amino-acid sequence in the B-chain of haemoglobin in a normal person and a sickle-cell anaemia person ? U **[Delhi Set-III, Comptt. 2010]**

Ans. In a person suffering from sickle-cell anaemia, the amino acid glutamine present at the sixth position, is replaced by the valine. **1**

Q. 20. What is the cause of Down's syndrome in humans ?

R **[Outside Delhi Set-I, Comptt. 2010]**

Ans. This syndrome develops due to trisomy of chromosome 21. The non-disjunction of the 21 chromosome during meiosis causes the trisomy of 21st chromosome and results in Down's syndrome. **1**

Q. 21. A colour blind boy is born to a couple with a normal colour vision. Write the genotype of the parents.

R **[Outside Delhi Comptt. - 2017, Set - I, II, III]**

Ans. Father - XY, Mother - XX^C ½ + ½
[CBSE Marking Scheme, 2017]

AI **Q. 22. Mention the combination (s) of sex chromosomes in a male and a female bird.**

A **[Foreign Set - I, 2017]**

Ans. Male - ZZ, Female - ZW ½ + ½
[CBSE Marking Scheme, 2017]

Detailed Answer :

The type of sex chromosomes in a female bird is ZW and in case of male bird is ZZ.

Q. 23. State the fate of a pair of autosomes during gamete formation. A **[Delhi 2017, Set - I, II, III]**

Ans. Segregate / separate **1**
[CBSE Marking Scheme, 2017]

Detailed Answer :

During gamete formation, a diploid germinal cell changes to a haploid germ cell. Hence, a pair of autosomes get segregated by means of meiotic division to produce haploid gametes.

Q. 24. Name the disorder caused due to the absence of one of the X-chromosomes in a human female.

A **[Delhi Comptt. 2017, Set - I, II]**

Ans. Turner's syndrome
[CBSE Marking Scheme 2017] 1

Detailed Answer :

A syndrome that occur due to monosomy is called turner's syndrome. It occurs due to union of an allosome free egg (22 + 0) and a normal X sperm or a normal egg and an allosome free sperm (22 + 0). The individual has $2n = 45$ chromosomes (44 + XO) instead of 46.

Q.25. Write the sex of a human having XXY chromosomes with 22 pairs of autosomes. Name the disorder this human suffers from.

R **[CBSE, Comptt Set 1, 2018]**

Ans. Male, Klinefelter's syndrome ½ + ½
[CBSE Marking Scheme, 2018]

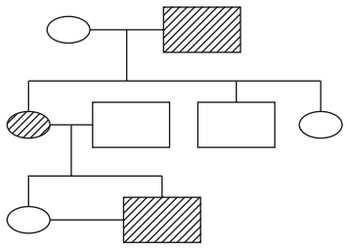
Commonly Made Error

- Students get confused between the genetic constitution of Down's syndrome, Klinefelter's syndrome and Turner's syndrome. Students often write opposite answers.

Answering Tip

- Understand all three disorders- Down's syndrome, Klinefelter's syndrome and Turner's syndrome, separately with relevant examples. Emphasize on operative terms.

Q. 26. Observe the pedigree chart and answer the following questions :



(i) Identify whether the trait is sex-linked or autosomal.

(ii) Give an example of a disease in human beings which shows such a pattern of inheritance.

R **[CBSE SQP 2015] (KVS)**

Ans. **(i)** Sex-linked.

(ii) Haemophilia/colour blindness. ½+½

[CBSE Marking Scheme, 2015]

Commonly Made Error

- Students are unable to understand the pedigree chart. It seems they are unaware about the symbols used in it.

Answering Tip

- Learn symbols used in pedigree chart to analyse it.

Q. 27. The prophase I stage of meiosis plays a vital role in r-DNA formation. Justify with reason.

A **[CBSE SQP, 2018]**

Ans. The prophase I stage of meiosis plays vital role in r-DNA formation because crossing over occurs at this stage, which helps in recombination.

[CBSE Marking Scheme, 2018]

AI **Q. 28. Write the genotype of (i) an individual who is carrier of sickle cell anaemia gene but apparently unaffected and (ii) an individual affected with the disease.**

U **[Outside Delhi, 2016]**

Ans. **(i)** Hb^A, Hb^S

(ii) Hb^S Hb^S ½ + ½

Short Answer Type Questions-I (2 marks each)

Q. 1. Differentiate between 'ZZ' and 'XY' type of sex-determination mechanisms. U **[Delhi Set-III, 2015]**

Ans. ZZ type of sex determination mechanism is found in birds, reptiles and fishes. In this type, the females have heteromorphic sex chromosomes (ZW), while males have homomorphic sex-chromosomes (ZZ). Females are heterogametic *i.e.* produce two dissimilar types of eggs while males produce only one type of sperms. The egg determines the sex of the individual.

XY type of sex determination mechanism is found in human beings. In this type, the male individuals have heteromorphic sex chromosomes (XY) and are therefore heterogametic *i.e.* producing two types of sperms are with X and the other carrying Y chromosome. The females have homomorphic sex chromosome (XX) and homogametic *i.e.* produce only one type of eggs. The sex of the offspring is determined by type of sperm taking part in fertilization. **2**

Q. 2. What happens when chromatids fail to segregate during cell division cycle ? Explain your answer with an example.

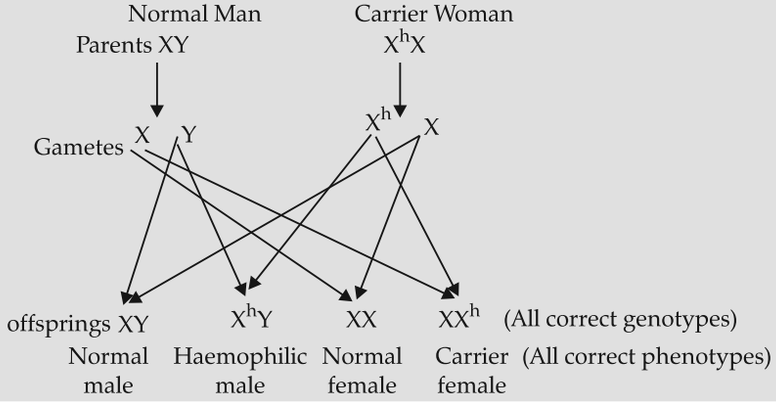
A **[Outside Delhi Set - I, II, III (Comptt. 2017)]**

Ans. Failure of segregation of chromatids during cell division cycle results in the gain or loss of chromosome / called aneuploidy *e.g.,* Downs' syndrome results in the gain of extra copy of chromosome 21 / Turner's syndrome results due to loss of an X-chromosome in human female. **1 + 1**

AI **Q. 3. Is haemophilia in humans a sex linked or autosomal disorder ? Work out a cross in support of your answer.**

A **[Outside Delhi Set-I, Comptt. 2016]**

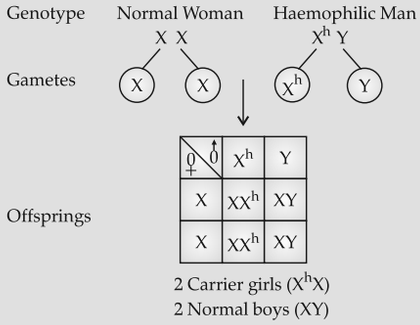
Ans. Haemophilia in humans is a sex linked disorder.



(any one incorrect genotype or phenotype-no marks)

//

Cross showing haemophilic father and normal mother



[CBSE Marking Scheme, 2016] 2

Answering Tip

- Graphic representations should be in the proper sequence. Write correct genotype and phenotype in the cross, otherwise this may deduct your marks.

Q. 4. Which chromosomes carry the mutant genes causing thalassaemia in humans ? What are the problems caused by these mutant genes ?

R **[Delhi Set-II, Comptt. 2015]**

Ans. 11^{th} and 16^{th} chromosomes carry the mutant gene causing thalassaemia.

These cause the formation of abnormal haemoglobin molecules resulting into anaemia.

[CBSE Marking Scheme, 2015] 1+1

Q. 5. Why is the possibility of human female suffering from haemophilia rare ? Explain.

R **[Delhi Set-II, 2014]**

Ans. $X X^h \times X^h Y$ ½

$X^h X^h$

haemophilic female. ½

Rare because mother should be atleast carrier and father haemophilic (non viable at later stage). **1**

[CBSE Marking Scheme, 2014]

Detailed Answer :

Haemophilic females can be produced only in the homozygous condition *i.e.* when the genes for haemophilia are present on both of the X chromosomes (X^hX^h). Marriage between a haemophilic man (X^hY) and carrier woman (X^hX) will produce haemophilic female (X^hX^h) but however this condition is lethal and therefore haemophilic female dies in the foetus state. Thus, the possibility of human female suffering from haemophilia is rare.

Commonly Made Error

- Many students fail to draw the correct cross. They forget to mention the correct genotype and phenotype in the cross, which may deduct their marks.

Answering Tip

- Practice drawing different crosses with proper understanding. Make sure you label all the genotypes and phenotypes correctly.

Q. 6. A haemophilic father can never pass the gene for haemophilia to his son. Explain.

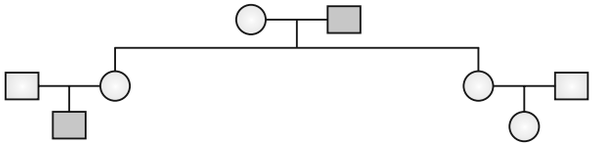
R **[CBSE, Comptt, Set 1, 2018]**

Ans. It is a sex linked recessive disorder in which X-chromosome has the haemophilic gene. Son inherits a Y chromosome from father and gene for haemophilia is not present on Y chromosome.

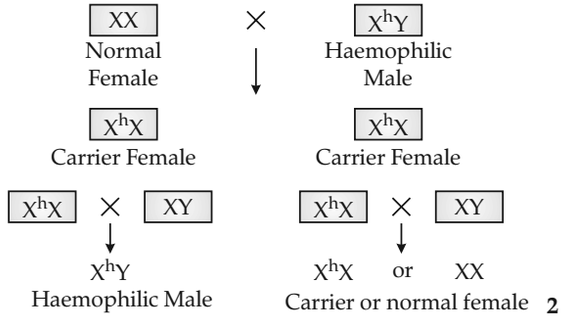
1+1

[CBSE Marking Scheme, 2018]

Q. 7. This is the pedigree of a family tracing the movement of the gene for haemophilia. Explain the pattern of inheritance of the disease in the family. E & A **[Delhi Set-I, Comptt. 2013]**



Ans. In the given pedigree of a family, a normal female is crossed with a haemophilic male. The offspring generated are carrier females as haemophilia is an X-linked disease and female has two X chromosomes. These carrier females when crossed with normal males produced carrier females and haemophilic males.



2

Q. 8. A non-haemophilic couple was informed by their doctor that there is possibility of a haemophilic

child be born to them. Draw a checker board and find out the percentage of possibility of such child in the progeny.

A **[CBSE SQP, 2013, 2012]**

Ans.

♀ / ♂	X	Y
X	X X	XY
X^h	X X^h	X^hY

Phenotypes : 50% daughter normal (XX)

50% daughter carrier (XX^h)

50% son normal (XY)

50% son haemophilic (X^hY) **2**

[CBSE Marking Scheme, 2010]

AI **Q. 9. (i) Why are grasshopper and *Drosophila* said to show male heterogamety ? Explain.**

(ii) Explain female heterogamety with the help of an example.

U **[Outside Delhi Set-I, 2010]**

Ans. **(i)** In grasshopper, males have one X only (XO type), in *Drosophila* males have one X and one Y (XY type) - Males in both cases produce 2 different kinds of gametes so heterogametic. ½ + ½

(ii) In birds female has ZW, produce two kinds of gametes and so heterogametic. ½ + ½

OR

(i) Male heterogamety, Grasshopper. ½ + ½

(ii) Female heterogamety, Birds. ½ + ½

[CBSE Marking Scheme, 2010]

Detailed Answer :

(i) In grasshoppers, there is XX—XO type of sex determination. Males have only one sex chromosome and therefore produce two types of male gametes : one with X chromosome (A + X) and the other without sex chromosome (A + O), thus shows heterogamety.

In *Drosophila,* there are two sex chromosomes (XY) in males which are heteromorphic and therefore two types of male gametes : one with X chromosome (A + X) and another with Y chromosome (A + Y).

(ii) Production of dissimilar types of gametes by females is called as female heterogametic. It is usually found when the sex chromosomes are heteromorphic. In female bird, the sex chromosomes are ZW and produce two types gametes (A + Z) and (A + W), thus showing heterogamety.

Q. 10. Explain the cause of chromosomal disorders. Describe the effect of such disorders with the help of an example each involving (i) autosomes, and (ii) sex chromosomes.

R **[Foreign Set - I, II, 2017]**

Ans. Gain or loss of a chromosome (due to non disjunction) **1**

(i) Down Syndrome - Additional copy of 21st chromosome / trisomy of 21. ½ + ½

(ii) Klinefelter's Syndrome - presence of an additional copy of X chromosome leading to XXY. Turner's Syndrome - absence of one of the X chromosome *i.e.,* 44 with XO ½ + ½

[CBSE Marking Scheme, 2017]

Answering Tip

- Learn the genetic constitution and features of all chromosomal disorders (Down's syndrome, Klinefelter's syndrome and Turner's syndrome) thoroughly.

Q. 11. During a cytological study conducted on the chromosomes of the insects, it was observed that only 50% of the sperms had a specific structure after spermatogenesis. Name the struture and write its significance in sex determination of insects. R **[CBSE SQP, 2017]**

Ans. X body / X factor / X chromosome.

In insects the sex chromosome consists of XX female; XO-Males **1 + 1**

[CBSE Marking Scheme, 2017]

Detailed Answer :

The specific structure is X chromosome or X factor or X body.

In insects, all eggs bear additional X chromosome. Male have only one X chromosome besides autosomes where as females have a pair of X chromosomes *e.g.,* grasshopper.

Q. 12. Why is pedigree analysis done in the study of human genetics ? State the conclusions that can be drawn from it. U **[Delhi Set-II, 2014]**

Ans. **(i)** Control crosses are not possible in case of humans beings. **1**

(ii) Analysis of traits in several generations of family / To trace pattern of inheritance / whether the trait is dominant or recessive / sex linked or not. **1**

[CBSE Marking Scheme, 2014]

Detailed Answer :

(i) This is because control crosses are not possible in case of humans beings.

(ii) **(a)** Traits can be analysed for several generations.

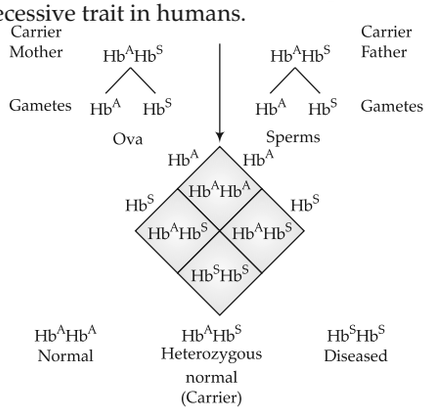
(b) Pattern of inheritance can be traced.

(c) To trace whether the trait is dominant or recessive or sex-linked or not. **2**

Short Answer Type Questions-II (3 marks each)

Q. 1. Give an example of an autosomal recessive trait in humans. Explain its pattern of inheritance with the help of a cross. A **[Delhi Set-I, 2016]**

Ans. Sickle cell anaemia is an example of autosomal recessive trait in humans.

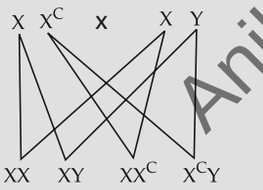


The disease is controlled by a single pair of allele Hb^A and Hb^S. The disease is only expressed if both the copies are defective *i.e.* only when the autosomal recessive genes are present in homozygous condition (Hb^SHb^S). People with a single defective copy of the gene are clinically normal, however they act as carrier and can pass on the defective gene to their next generations in the ratio of 1 : 2 : 1 as shown in the above cross. **2+1**

Q. 2. A couple with normal vision bear a colour blind child. Workout a cross to show how it is possible and mention the sex of the affected child.

A **[Delhi Set-II, 2016]**

Ans.

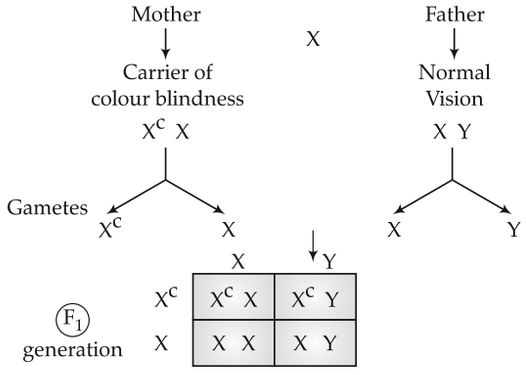


½ × 5 = 2½

Affected child is male. **½**

[CBSE Marking Scheme, 2016]

Detailed Answer :



X^C X : X^C Y : X X : X Y

Carrier daugher : Affected son : Normal daughter : Normal son

1 : 1 : 1 : 1

Thus, affected child is male (son).

Q. 3. Write the type and location of the gene causing thalassaemia in humans. State the cause and symptoms of the disease. How is sickle cell anaemia different from this disease ?

R **[Delhi Set-III, 2014]**

Ans. **(i)** Autosomal, recessive gene, gene for alpha thalassemia is on chromosome 16, for Beta thalassemia it is on chromosome 11. **1**

(ii) Cause of symptoms—Mutation or deletion of the gene / genes, resulting in reduced rate of synthesis of one of the globin chains / alpha or beta chains). **½ + ½**

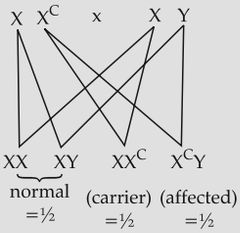
(iii) Thalassaemia is a quantitative problem of too few globin molecules of haemoglobin, while sickle-cell is a qualitative problem of synthesizing an incorrectly functioning globin. **1**

[CBSE Marking Scheme, 2014]

Q. 4. One of the twins born to parents having normal colour vision was colour blind whereas the other twin had normal vision. Work out the cross. Give two reasons how it is possible.

A **[Foreign Set - III, 2017]**

Ans.



½

Genes that lead to colour blindness located on X-chromosome. **½**

Gene is recessive and is suppressed in heterozygous mother (female) but expressed in male in single dose. **½**

[CBSE Marking Scheme, 2017] 2 + ½ + ½ = 3

AI **Q. 5. 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.** U **[Outside Delhi 2017, Set - I, II, III]**

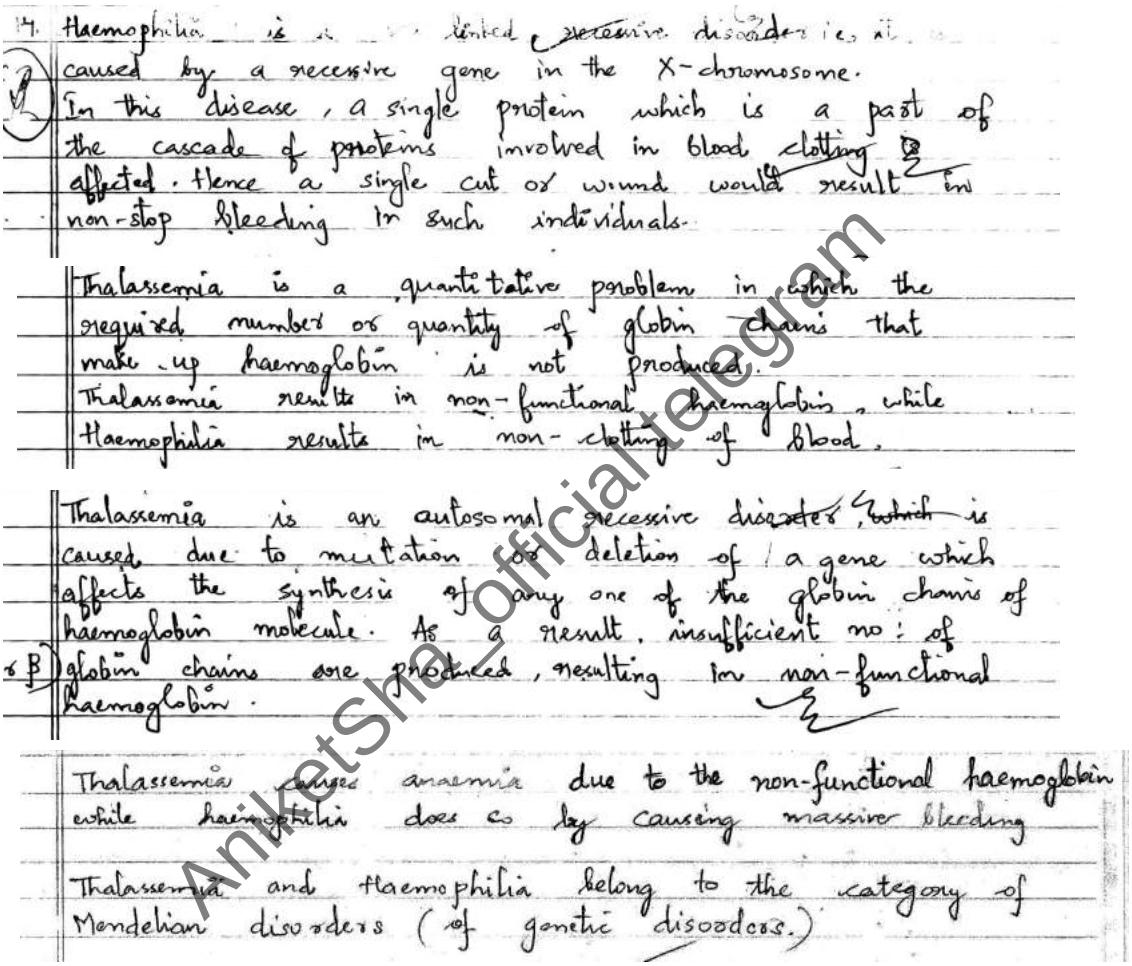
Ans.

Haemophilia	Thalassemia
Single protein involved in the clotting of blood is affected	Defects in the synthesis of globin leading to formation of abnormal haemeoglobin
Sex linked recessive disorder	Autosomal recessive disorder
Blood does not clot	Results in anaemia

Mendelian disorder ½ × 6=3

[CBSE Marking Scheme, 2017]

OR



Haemophilia is a sex linked recessive disorder i.e. it is caused by a recessive gene in the X-chromosome. In this disease, a single protein which is a part of the cascade of proteins involved in blood clotting is affected. Hence a single cut or wound would result in non-stop bleeding in such individuals.

Thalassemia is a quantitative problem in which the required number or quantity of globin chains that make up haemoglobin is not produced. Thalassemia results in non-functional haemoglobin, while Haemophilia results in non-clotting of blood.

Thalassemia is an autosomal recessive disorder, which is caused due to mutation or deletion of a gene which affects the synthesis of any one of the globin chains of haemoglobin molecule. As a result, insufficient no. of α & β globin chains are produced, resulting in non-functional haemoglobin.

Thalassemia causes anaemia due to the non-functional haemoglobin while haemophilia does so by causing massive bleeding

Thalassemia and Haemophilia belong to the category of Mendelian disorders (of genetic disorders.)

[Topper's Answer 2017]

Answering Tip

- Learn the differences in tabular form for better retention and understanding.

Q. 6. Explain with the help of an example each– Male and female heterogamety mechanisms of sex determination. R **[Delhi Comptt. Set - III]**

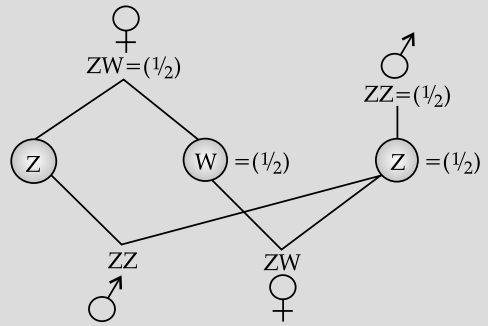
Ans. Male produces 2 different types of gametes
XO - *e.g.* grasshopper.
XY - *e.g.* human, it is the type of sperm fertilising the egg that determine the sex of the offspring. **1 + ½**

Female produces 2 different types of gametes.
ZW - *eg.* : Birds, it is the type of egg getting fertilised with the sperm that determine the sex of the chick. **1 + ½**

[CBSE Marking Scheme, 2017]

Q.7 Explain the mechanism of 'sex determination' in birds. How does it differ from that of human beings? U **[Outside Delhi/Delhi, 2018]**

Ans.



Birds : Female heterogamety / female produces (Z)

types and (W) type of gametes ½
Humans : Male heterogamety / male produces (X) types and (Y) types of gametes ½
[2+1=3 marks]
[CBSE Marking Scheme, 2018]

Answering Tip

- Sex determination according to type of chromosomes (XX/XY) and according to haplo-diploidy should be discussed taking suitable examples.

AI **Q. 8. Identify 'a', 'b', 'c', 'd', 'e' & 'f' in the table given below :**

S. No.	Syndrome	Cause	Characteristics of affected individual	Sex Male/ Female/ Both
1.	Down's	Trisomy of 21	'a' (i) (ii)	'b'
2.	'c'	XXY	Overall masculine development	'd'
3.	Turner's	44 with OX	'e' (i) (ii)	'f'

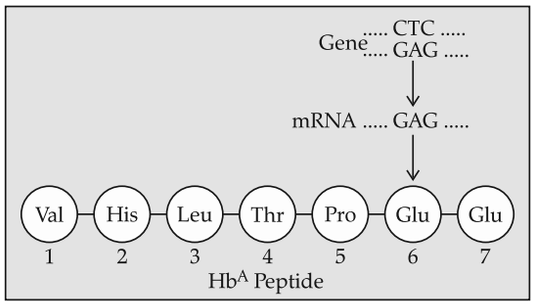
R **[Outside Delhi Set-II, 2014]**

Ans. **(a)** Short statured / small round head / furrowed tongue / partially open mouth / palm is broad / physical development retarded / psychomotor development retarded / mental development retarded. ½
(b) both / male and female ½
(c) Klinefelter's syndrome ½
(d) male ½
(e) sterile ovaries / rudimentary ovaries, lack of secondary sexual characters. ½
(f) female. **[CBSE Marking Scheme, 2014]** ½

Q. 9. Why are human females rarely haemophilic ? Explain. How do haemophilic patients suffer ?
U **[Outside Delhi Set-I, II, III, 2013]**

Ans. Haemophilia is a X-linked genetic disorder, which means that it shows criss cross inheritance. Like most recessive sex-linked X chromosome disorders, haemophilia is more likely to occur in males than females. This is because females have two X chromosomes (XX), while males have only one (XY), so the defective gene is guaranteed to manifest in any male who carries it. A female having two defective copies of the gene is very rare.
Haemophilia impairs the body's ability to control blood clotting and coagulation. **2 + 1**

Q. 10. Given below is the representation of amino acid composition of the relevant translated portion of b-chain of haemoglobin, related to the shape of human red blood cells.



(i) Is this representation indicating a normal human or a sufferer from certain genetic disease ? Give reason in support of your answer.
(ii) What difference would be noticed in the phenotype of the normal and the sufferer related to this gene ?
(iii) Who are likely to suffer more from the defect related to the gene represented – the males, the females or both males and females equally ? And why ? A **[Delhi Set-I, 2012]**

Ans. **(i)** This representation (Hb^A peptide) indicates a normal human, because the glutamic acid in the sixth position is not substituted by Valine. ½ + ½
(ii) The sufferer's RBCs become elongated and sickle shaped as compared to the normal biconcave RBCs. **1**
(ii) Both males and females are likely to suffer from the disease equally, as this is not a sex linked disease. It is an autosomal linked recessive trait. ½ + ½
[CBSE Marking Scheme, 2012]

Detailed Answer :

(i) This micrograph representation (Hb^A peptide) is of amino acid composition of a part of b-chain of haemoglobin molecule of a normal human because the sixth codon of b-globin mRNA is GAG and therefore there is glutamic acid at the sixth position of b-globin chain.

(ii) The sufferer's RBCs get elongated and becomes sickle shaped because this gene, if simulated cause the haemoglobin molecule to undergo polymerization due to oxygen tension resulting in the change of shape of RBCs from concave disc to elongated sickle like structure. Such RBCs can not pass through narrow blood capillaries, therefore they tend to slow down the blood flow, clot, degenerate and thus causing sickle cell anaemia.

(iii) The males and females both are likely to suffer from this disorder or defect because this is not a sex linked disorder. It is an autosomal linked recessive disorder due to single base substitution resulting in the change at the sixth codon of beta globin chain from GAG to GUG resulting in the substitution of glutamic acid to valine.

Q. 11. (i) Sickle cell anaemia in human is a result of point mutation. Explain.

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

U **[Delhi Set-I, II, III, 2011]**

Ans. (i) Mutation arising due to change in single base pair of DNA, the defect is caused by the substitution of Glutamic acid (Glu) by Valine (Val) at the sixth position of the beta globin chain of the haemoglobin molecule. **2**

(ii) Father - $Hb^A Hb^S$, Mother - $Hb^A Hb^S$ ½ + ½

(Both parents are heterozygous)

[CBSE Marking Scheme, 2011]

Q. 12. (i) Name the genetic disorder in a human female having 44 + XO karyotype. Mention the diagnostic features of the disorder.

(ii) Explain the cause of such chromosomal disorder.

R **[Outside Delhi Comptt. 2011]**

Ans. (i) (a) Turner's syndrome.

(b) 44 with XO chromosomes — such females are sterile as ovaries are rudimentary. Other features include lack of other secondary sexual characters, short stature and under developed feminine characters.

(ii) Such a disorder is caused due to the absence of one of X chromosomes. **2 + 1 = 3**

Q. 13. If there is a history of haemophilia in the family, the chances of male members becoming haemophilic are more than that of the female.

(i) Why is it so ?

(ii) Write the symptoms of the disease.

A **[Outside Delhi Set-I, Comptt. 2015]**

Ans. (i) Defective gene is on X chromosome, in case the carrier female (mother) passes X^h to the son he suffers, if she passes X^h to the daughter, she has the other X (from father) to make it heterozygous so the daughters escape as carriers.

(ii) The blood does not clot in the affected person after an injury or a small cut. **2+1=3**

[CBSE Marking Scheme, 2015]

Detailed Answer :

(i) Haemophilia is a sex linked recessive trait. The gene for this trait is located on X chromosome. There are greater chances of males getting haemophilic because the males have only one X chromosome and haemophilic gene located on this chromosome (X^hY) expresses itself. Human males are hemizygous. Therefore, X^hY would by haemophilic while X^hX would not be haemophilic, she will be a carrier. If carrier mother X^hX passes X^h to son he would be haemophilic but if she passes it to daughter and she has the other normal X from father, she escapes as carrier.

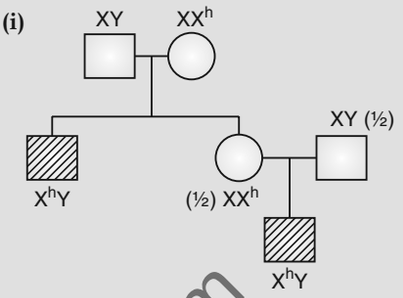
(ii) In haemophilic person, the blood does not clot after injury due to the absence of blood clotting factor.

AI **Q. 14. A cross between a normal couple resulted in a son who was haemophilic and a normal daughter. In course of time, when the daughter was married to a normal man, to their surprise, the grandson was also haemophilic.**

(i) Represent this cross in the form of a pedigree chart. Give the genotypes of the daughter and her husband.

(ii) Write the conclusion you draw from the inheritance pattern of this disease. E & A **[Delhi Set-II, 2014]**

Ans. (i)



1

Construction of pedigree chart **1**

(ii) Sex - linked recessive inheritance pattern **1**

[CBSE Marking Scheme, 2014]

Detailed Answer:

(i) Genotype of daughter is XX^h

Genotype of her husband is XY

(ii) Conclusion drawn from the inheritance pattern :

(a) XX^h females are carriers of haemophilia. They are not suffering from haemophilia but can pass on the gene X^h to offspring.

(b) X^hY males suffer from haemophilia. They seldom reach reproductive age.

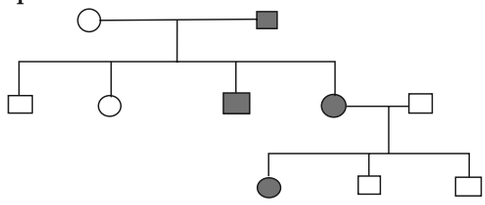
(c) XX females are normal.

(d) XY males are normal too.

(e) X^hX^h females die in embryonic stage.

(f) Inheritance of haemophilia follows a criss-cross pattern.

Q. 15. Study the given pedigree chart and answer the questions that follow:



(a) Is the trait recessive or dominant?

(b) Is the trait sex-linked or autosomal?

(c) Give the genotypes of the parents shown in generation I and their third child shown in generation II and the first grandchild shown in generation III. E & A **[CBSE SQP, 2018]**

Ans. (a) Dominant ½

(b) Autosomal. ½

(c) Genotype of parents in generation I – Female – aa and Male – Aa. ½ + ½

Genotype of third child in generation II - Aa.

Genotype of first grandchild in generation III - Aa. ½ + ½

[CBSE Marking Scheme, 2018]

Q. 16. Haemophilia is a sex linked recessive disorder of humans. The pedigree chart given below shows the inheritance of Haemophilia in one family. Study the pattern of inheritance and answer the questions given.

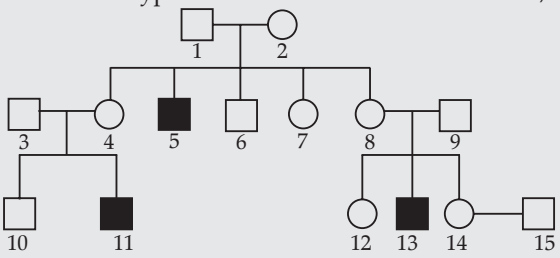
(a) Give all the possible genotypes of the members 4, 5 and 6 in the pedigree chart.

(b) A blood test shows that the individual 14 is a carrier of haemophilia. The member numbered 15 has recently married the member numbered 14. What is the probability that their first child will be a haemophilic male? Show with the help of Punnett square. C **[CBSE SQP, 2018]**

Ans. (a) Genotypes of member 4 – XX or XX^h ½

Genotypes of member 5 – X^hY ½

Genotypes of member 6 – XY ½



(b) The probability of first child to be a haemophilic male is 25%.

1 mark for punnetts square + ½ for probability

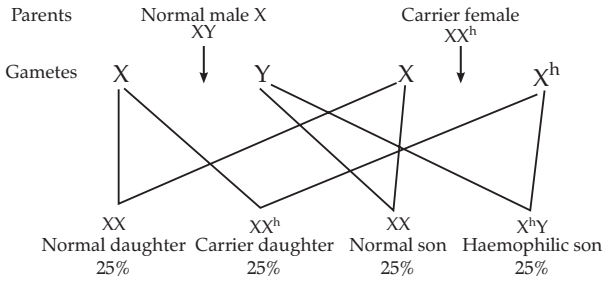
[CBSE Marking Scheme, 2018]

Detailed Answer:

(a) Same as in marking scheme.

(b) Probability of first child to be hemophilic male is 25 %.

Cross :



Q. 17. A haemophilic son was born to normal parents. Give the genotypes of parents and son.

C **[Delhi Comptt. 2009]**

Ans. Father (A + XY)

Mother (A + XX^h)

Son (A + X^hY)

(A = Autosomes, which are 44 in number & XY = Sex chromosomes). **3**

AI **Q. 18. Why is that the father never passes on the genes for haemophilia to his son ?**

R **[Outside Delhi Set-III, 2012]**

Ans. Haemophilia is a sex linked trait. The gene for it is located on X chromosome only. Since, father contributes only Y chromosome to the son, he never passes haemophilic gene to his son. **3**

Q. 19. Why do normal RBCs become elongated sickle shaped structures in a person suffering from sickle shaped anaemia. R **[Foreign, 2014]**

Ans. Sickle cell anaemia is caused due to point mutation because of which in β-globulin chain of haemoglobin molecule, the glutamic acid (Glu) is replaced by valine. This results in oxygen stress. Under this condition the RBCs loose their circular shape, polymerise and become elongated and sickle shaped. **3**

Long Answer Type Questions

(5 marks each)

Q. 1. Aneuploidy of chromosomes in human beings results in certain disorders. Draw out the possibilities of the karyotype in common disorders of this kind in human beings and its consequences in individuals. R **[CBSE, SQP, 2018]**

Ans. Down's syndrome, Turner's syndrome, Klinefelter's syndrome are the common examples of Aneuploidy of chromosomes in human beings.

Down's syndrome results in the gain of extra copy of chromosome 21- trisomy. ½ + ½

Turner's syndrome results due to loss of an X chromosome in human females- XO monosomy. ½

Klinefelter's Syndrome is caused due to the presence of an additional copy of X-chromosome resulting into XXY condition. ½

Down's Syndrome: The affected individual is:

(a) Short statured with small round head.

(b) Furrowed tongue and partially open mouth.

(c) Palm is broad with characteristic palm crease.

(d) Physical, psychomotor and mental development is retarded. **(Any two)** ½ + ½

Klinefelter's Syndrome: The affected individual is:

(a) A male with development of breast i.e. Gynecomastia.

(b) Such individuals are sterile. ½ + ½

Turner's Syndrome: The affected individual shows following characters:

(a) Females are sterile as ovaries are rudimentary.

(b) Lack of other secondary sexual characters. ½ + ½

[CBSE Marking Scheme, 2018]

Q. 2. (i) A couple with blood group 'A' and 'B' respectively have a child with blood group 'O'. Work out a cross to show how it is possible and the probable blood groups that can be expected in their other off-springs.

(ii) Explain the genetic basis of blood groups in human population.

A **[Outside Delhi Set-I, Comptt. 2016]**

Ans. **(i)** Father = I^Ai
Mother = I^Bi
Child = ii

Father = I^Ai X I^Bi (Mother)

	I^A	i
I^B	I^AI^B Blood Group AB	I^Bi Blood Group B
I	I^Ai Blood Group A	ii Blood Group O

Phenotypes of all offsprings = AB, B, A and O blood group

(ii) Genetic basis of blood group :
Three alleles of one gene/multiple alleles/gene I^A, I^B, i
A and B are co-dominant/expressed together.

I^AI^A, I^A i	A
I^BI^B, I^B i	B
I^AI^B	AB
ii	O

3+2

[CBSE Marking Scheme, 2016]

Commonly Made Error

- Most of the students just show the cross between woman having blood group 'B' and man with blood group 'A'. Correct alleles were not given. Many of them forget to mention the occurrence of all the four blood groups in the progeny. Very few students use the correct format to represent the type of alleles. Instead of writing as I^A, I^B and i, they write 'alleles A, B and O'. Some students show the possible combinations of alleles in the progeny but forget to mention the corresponding blood groups.

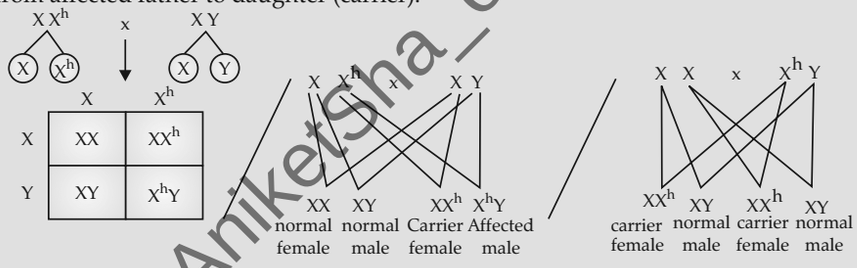
Answering Tip

- Discuss simple problems along with the explanation of Punnett square. Teach the concept of multiple allele with examples of blood groups and with alleles involved.

AI Q. 3. 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.

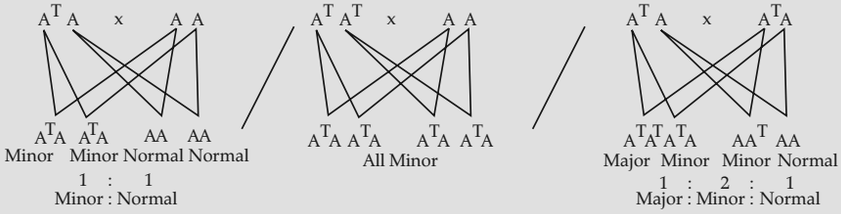
U [Foreign Set-III, 2016]

Ans. Thalassemia- Anaemia
Haemophilia-Non stop bleeding
Haemophilia- Sex linked recessive disorder, is generally passed on from (carrier) mother to some of her sons/ from affected father to daughter (carrier).



(Note : Any one cross, one mark to be given if the entire diagram (cross) is correct)

Thalassemia - Autosome linked recessive blood disease, inheritance is like Mendelian inheritance pattern.



[CBSE Marking Scheme, 2016] 5

Detailed Answer :

Difference in inheritance pattern :

Haemophilia : It is a sex-linked (X-linked) recessive disorder, inherited from haemophilic father (X^hY) or carrier mother (X^hX). Females are haemophilic only in homozygous double recessive state (X^hX^h) but such females die before birth.

Thalassemia : It is caused by haemolytic anaemia. It shows autosomal recessive pattern of inheritance and is controlled by two pairs of alleles (HBA1 & HBA2). The effect is more pronounced when the defective gene occur in homozygous state, causing thalassemia major. In heterozygous state the adverse effect of thalassemia is minor. The trait (Thalassemia) is inherited as autosomal recessive. This is found equally in both males and females. However the defective alleles for thalassemia in both males & females unlike haemophilia expresses itself only when it is in homozygous condition. The heterozygotes for recessive trait remain unnoticed but act as heterozygous carriers.

Q. 4. (i) State the cause and symptoms of colour blindness in humans.

(ii) Statistical data has shown that 8% of the human males are colour-blind whereas only 0.4% of females are colour-blind. Explain giving reasons how is it so. A **[Foreign Set-II, 2015]**

Ans. (i) Cause-sex-linked recessive disorder.
Symptoms-failure to discriminate between red and green colour.

(ii) Since males have only one X chromosome gene for colour blindness, if present in any one parent will always be expressed, whereas in female it will be expressed only if it is present on both the X chromosomes, when both parents are carrying gene for colour blindness. **5**

[CBSE Marking Scheme, 2015]

Detailed Answer :

(i) The colour blindness is due to recessive sex linked disorder. In this disorder of vision the patient is unable to distinguish between red and green colour. The gene for colour blindness is located on the X chromosome.

(ii) The colour blindness is found in about 8% of the males and only 0.4% of the females. The greater prevalence of the colour blindness in males is due to the presence of only one X chromosome and the hemizygous (X & Y) expression of the allele for colour blindness i.e. if gene for colour blindness is present on X-chromosome of male then it will always express while in case of females the incidence of disease of colour blindness is possible only in homozygous condition (X^CX^C) i.e. if both the X-chromosomes carry the allele (X^CX^C) for colour blindness. Occurrence of allele for colour blindness on one of the X chromosome makes the female a carrier X^CX.

Q. 5. (i) Why are thalassemia and haemophilia categorized as Mendelian disorders ? Write the symptoms of these diseases. Explain their pattern of inheritance in humans.

(ii) Write the genotypes of the normal parents producing a haemophilic son.

U **[Outside Delhi Set-I, 2015]**

Ans. (i) Both are caused due to alteration/mutation, in a single gene and follow Mendelian pattern of inheritance. ½ × **2**
Symptoms
Thalassemia-anaemia (caused due to defective/abnormal Hb).
Haemophilia-non stop bleeding even in minor injury. ½ × **2**
Pattern of inheritance
Thalassemia-autosomal recessive inheritance pattern, inherited from heterozygous/parent carrier. ½ × **2**
Haemophilia-X linked recessive inheritance, inherited from a haemophilic father/carrier mother (females are rarely haemophilic). ½ × **2**

(ii) X^hX-Mother ½
XY-Father **[CBSE Marking Scheme, 2015]** ½

Detailed Answer :

(i) Thalassemia and haemophilia are categorised as Mendelian disorders because they occur by mutation in a single gene. Their mode of inheritance follows the principles of Mendelian genetics.
Mendelian disorders can be :
(a) Autosomal dominant (muscular dystrophy)
(b) Autosomal recessive (thalassemia)
(c) Sex linked (haemophilia)
Symptoms of thalassemia :
Thalassemia minor results only in mild anaemia, characterised by low haemoglobin level.
Thalassemia major is also known as Cooley's anaemia. In this disease, affected infants are normal but as they reach 6 to 9 months of age, they develop severe anaemia, skeletal deformities, jaundice, fatigue etc.
Symptoms of Haemophilia : Persons suffering from this disease does not develop a proper blood clotting mechanism. A haemophilic patient suffers from non-stop bleeding even on a simple cut, which may lead to death.
Patterns of inheritance of Thalassemia :
Pairs of alleles Hb^A and Hb^T controls the expression of this disease.
Conditions for Thalasemia :
Hb^A and Hb^A: Normal
Hb^A and Hb^T: Carrier
Hb^T and Hb^T : Diseased
Let us assume that parents are carriers of beta-thalassemia.
Parents:
$Hb^AHb^T \times Hb^AHb^T$
Offsprings:

Hb^AHb^A	Hb^AHb^T	Hb^AHb^T	Hb^THb^T
(Normal)	(Carrier)	(Carrier)	(Severe affected -Thalassemic)

Patterns of inheritance of Haemophilia :
It is X-linked genetic disorder. Compared to females, males have higher chances of getting affected because females have XX chromosomes while males have only one X with Y chromosomes. Thus, for a female to get affected by haemophilia, she has to have the mutant gene on both the X-chromosomes while males can be affected if they carry it on the single X-chromosome.
Conditions for haemophilia :
XX, XX - Normal
X^hY - Haemophilic
X^hX - Carrier
X^hX^h-Haemophilic
Let us assume that a carrier female (X^hX) is married to a normal male :
Parents : XY (**Male**) X^hX (**Female**)
Offspring :

X^hX	XX	X^hY	XY
(Carrier female)	(Normal female)	(Haemophilic Male)	(Normal Male)

(ii) When normal male marries a carrier female (she is considered normal as she contains the mutant gene on one of her X-chromosomes), they can produce a haemophilic son. So, the genotype of the parents would be XY and X^hX.
Parents : XY (Father) × X^hX (Mother)
Offspring :

X^hX	XX	X^hY	XY
(Carrier female)	(Normal female)	(Haemophilic Male)	(Normal Male)

5

Answering Tip

- Illustrate by showing different examples of crosses between haemophilic and normal. Cite some examples of sex-linked inheritance. Also explain why sex-linked characters are common in males. Ask students to carry out crosses with different combinations of sex linked diseases to write the genotype and phenotype correctly.

AI **Q. 6. (i) How does a chromosomal disorder differ from a Mendelian disorder ?**
(ii) Name any two chromosomal aberrations associated disorders.
(iii) List the characteristics of the disorders mentioned above that help in their diagnosis.
U **[Delhi Set-I, 2014]**

Ans. (i) Mendelian disorders are mainly determined by alteration or mutation in the single gene, chromosomal disorders are due to absence / excess / abnormal arrangement of one or more chromosomes. ½ + ½
(ii) Turner's syndrome, Klinefelter's syndrome, Down's syndrome. (**Name any two**) **1+1=2**
(iii) Turner's syndrome : Such a disorder is caused due to the absence of one of the X chromosomes *i.e.* 44 with XO, Such females are sterile as ovaries are rudimentary besides other features including lack of other secondary sexual characters.
Klinefelter's syndrome : This genetic disorder is also caused due to the presence of an additional copy of X-chromosome resulting into a karyotype of 47, XXY. Such an individual has overall masculine development, however, the feminine development (development of breast i.e. Gynaecomastia) is also expressed. Such individuals are sterile.
Down's syndrome : The cause of this genetic disorder is the presence of an additional copy of the chromosome number 21 (trisomy of 21) This disorder was first described by Langdon Down (1866).
The affected individual is short statured with small round head, furrowed tongue and partially open mouth. Palm is broad with characteristic palm crease. Physical, psychomotor and mental development is retarded. **[CBSE Marking Scheme, 2014] 2**

Q. 7. Why is thalassaemia categorized as a Mendelian disorder ? Write the symptoms and explain the causes of the disease. How does it differ from sickle-cell anaemia ? U **[Delhi Set-III, Comptt. 2013]**

Ans. Thalassaemia is categorized as a Mendelian disorder because it is determined by single allelic mutation. It involves the genes HBA1 and HBA2, inherited in a Mendelian recessive fashion.
Symptoms : People with thalassaemia make less haemoglobin and less circulating red blood cells than normal human beings, which results in mild or severe anaemia.
Cause : Both α and β-thalassaemias are often inherited in an autosomal recessive fashion, although this is not always the case. Thalassaemias are a group of disorders caused by defects in the synthesis of globin polypeptide. Absence or reduced synthesis of one of the globin chains results in an excess of the other. In this situation, free globin chains, which are insoluble, accumulate inside the red blood cells and form precipitates which damage the cells, causing cell lysis, resulting in anaemia. There are two main types of thalassaemias in which synthesis of α or β globin is defective.
In thalassaemia, patients have a deficiency of either α or β globin. But patients with sickle-cell anaemia have a specific mutant form of globin, causing production of abnormal red blood cells. Sickle- cell anaemia is caused by the mutant recessive allele on chromosome 11. **1 + 1 + 2 + 1 = 5**

Q. 8. (i) Why is haemophilia generally observed in human males ? Explain the condition under which a human female can be haemophilic.
(ii) A pregnant human female was advised to undergo M.T.P. It was diagnosed by her doctor that the foetus she is carrying has developed from a zygote formed by an XX-egg fertilized by Y-carrying sperm. Why was she advised to undergo M.T.P. ?
A **[Delhi Set-I, 2011]**

Ans. (i) Haemophilia is caused due to the recessive gene on X chromosome. **½ + ½ = 1**
Y has no allele for this / if a male is X^hY then he is haemophilic / if male inherits X^h from the mother he is haemophilic (with the genotype X^hY). **1**
If female inherits X^hX^h, one from the carrier mother and one from her haemophilic father then she can be haemophilic. **1**
(ii) Embryo has (trisomy of sex chromosome) XXY / Klinefelter's syndrome. **1**
Advised MTP since child will have the following problems :
Male with feminine traits / like gynecomastia / under developed testes / sterile. **1**
[CBSE Marking Scheme, 2011]

Know the Terms

- **Alleles or allelomorphs :** A pair of Mendelian factors or genes located on the same locus of two homologous chromosomes of an individual which control the expression of a trait or character are called alleles or allelomorphs.
- **Back cross :** Cross between hybrid and one of its parent.
- **Clone :** The group of organisms produced by asexual reproduction. They are morphologically and genetically similar to one another as well as their parents (The individuals of a clone are called Ramet).
- **Cross :** Deliberate mating of two parental types of organisms of the same species.
- **Diploid :** An individual or cell containing two complete sets of chromosomes.
- **Diplotene chromosomes :** Lampbrush Chromosomes.
- **Dominant factor or allele :** It is one of a pair of alleles which can express itself whether present in homozygous or heterozygous state. e.g. T (tallness in pea), R (round seed in pea), A (axial flower in pea).
- **Dysgenics :** Study of undesirable traits of human race and the genes that cause them.

- **F_1 generation :** Hybrids Produced from a cross between the genetically different individuals called parents. e.g. Tt individuals are produced in F_1 generation from a cross between TT and tt parents.
- **F_2 generation :** It is the generation of individuals which arises as a result of interbreeding or selfing amongst individuals of F_1 generation.
- **Father of genetics :** Gregor Johann Mendel
- **Felix Bernstein :** Discovered multiple alleles, co-dominance and dominant recessive relationship in determination of human blood groups.
- **Gene locus :** A particular portion or region of the chromosomes representing a single gene is called gene locus.
- **Gene pool :** Aggregate of all the genes and their alleles present in an interbreeding population is known as gene pool.
- **Genetics (Gk. Genesis – origin) :** It is a branch of biology that deals with the study of principles and mechanism of heredity and variations. The term genetics was coined by Bateson (1906).
- **Genome :** It is a complete set of chromosomes where every gene/chromosome is represented singly as in a gamete.
- **Genotype : (Gk. Geno- race; typos – image).** It is the genetic constitution of individual with regard to one or more characters irrespective that whether the genes are expressed or not, for e.g. genotype of hybrid tall pea plant is Tt, pure tall TT and pure dwarf tt.
- **Haploid (Monoploid) :** An individual or cell containing a single complete set of chromosomes.
- **Heredity (L. hereditas – heirship or inheritance) :** It is the sum of all biological processes by which a particular characteristics are passed on from parents to their offspring, either through asexual reproduction or sexual reproduction.
- **Heterozygote (heterozygous) :** It is an individual which contain the two contrasting factors of a character or two different alleles of a gene on the same locus of its homologous chromosomes. It is not pure and is called hybrid for that character. e.g. Tt.
- **Homozygote (homozygous) :** It is an individual which contains identical alleles of a gene or factor of a character on its homologous chromosomes. e.g. TT or tt.
- **Hybrid :** The organisms produced after crossing two genetically different individuals is called hybrid. Also called heterozygote or heterozygous individuals.
- **Inheritance :** Transmission of characters from parents to progeny.
- **Ishihara cards :** Cards used for checking colour blindness.
- **Mendelian factor or gene :** It is a unit of inheritance which passes from one generation to the next through the gamete and controls the expression of a character in the organisms.
- **Phenotype :** (Gk. Pheno – to appear, typos – image): It is observable or measurable distinctive structural or functional characteristic of an individual. e.g. phenotypic tall pea plant can be genotypically TT ot Tt.
- **Punnett square (Checker board) :** A grid that enables to calculate the results of simple genetic crosses.
- **Recessive factors or allele :** 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. 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.
- **Reciprocal cross :** Cross which involves two types of individual where the male of one type is crossed with female of the second type and vice versa.
- **Test cross :** Cross to know whether an individual is homozygous or heterozygous for dominant characters. The F_1 individual is crossed with one of its recessive parent.
- **Trait :** A phenotypic characteristic of an inherited character.
- **Variation :** Tendency of differences in various traits of individuals of a progeny from one another and their parents.
- **W. Johannsen :** Coined the term pure line (1903), gene (1909), genotype and phenotype.
- **Wild and mutant alleles (wild and mutant phenotype) :** Wild allele is one which is originally present in the population and is dominant, usually widespread. Mutant allele is less common and is believed to be formed through mutation of wild allele.
- **Wild type :** The species variety most commonly found in the natural population.

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