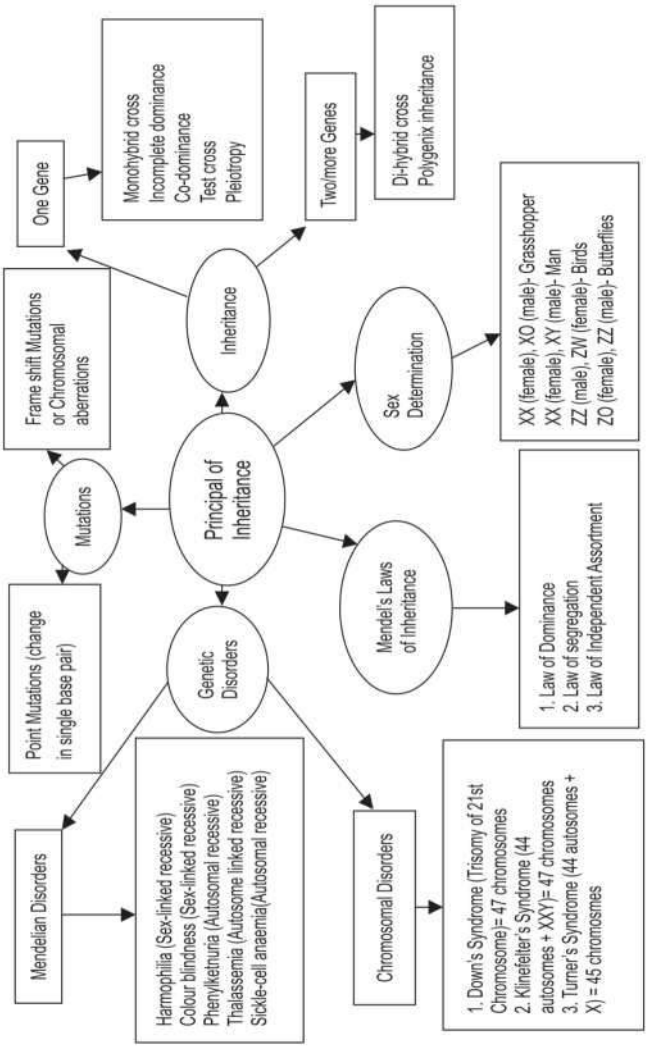
Chapter - 4 Principles of Inheritance and Variation



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

Phenotype : The observable or external characteristics of an organism.

Genotype : The genetic constitution of an organism.

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

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

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

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

Male heterogamety : When male produces two different types of gametes/ sperms e.g. In human beings X and Y. Sperm with X chromosome or sperm with Y chromosome

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

Gene- It is a segment of DNA called cistron and the unit of in heritance which is carried from parent by a gamete.

Genome- The entire genetic set of a prokaryote or virus or the haploid genetic set of a eukaryote.

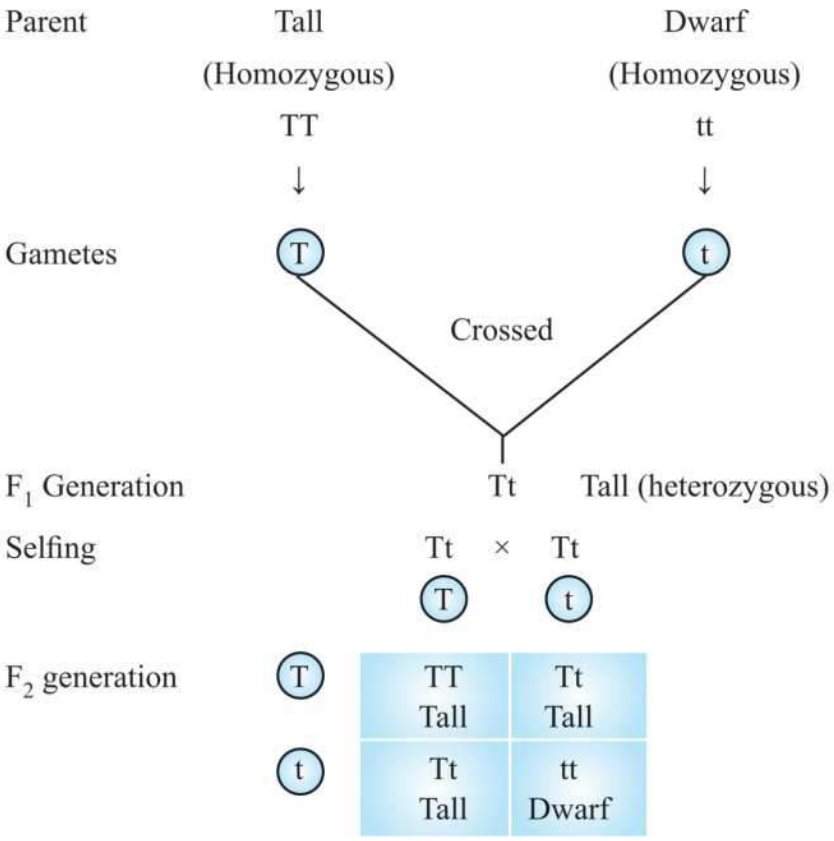
Gene Pool- An aggregate of all genes and their alleles, present in an interbreeding population.

Test Cross- A cross between an individual of unknown genotype and recessive parent. It is used to test whether an individual is homozygous (pure) or heterozygous (hybrid). It is also used as test for linkage.

Heredity- Also called inheritance is the process of transmission of genetic characters (traits) from parents to their offsprings.

Gregor Johann Mendel Conducted controlled breeding experiment on garden pea

(*Pisum sativum*) with a single trait. It is called monohybrid cross.



Tall homozygous (TT) – 25% Pure

Tall heterozygous (Tt) – 50% Hybrid.

Dwarf homozygous (tt) – 25% Pure.

Law of Dominance : When two individuals of a species differing in a pair of contrasting characters/traits are crossed, the trait that appears in the F_1 generation is dominant and the alternate form that remain hidden, is called recessive.

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

Law of Independent Assortment : In the inheritance of two pairs of contrasting characters (dihybrid cross) the factors of each pair of characters segregate independently of the factors of the other pair of characters.

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

Test cross is done to determine whether the individual parent exhibiting dominant traits is homozygous or heterozygous.

Flower colour is → Violet (Dominant phenotype, Genotype is unknown)

Genotype may be WW or Ww

Example :

Violet	×	White	
VV		vv	homozygous recessive

Case 1

	v	v
V	Vv Violet	Vv Violet
V	Vv Violet	Vv Violet

Here, all flowers are violet

If all the offsprings show dominate trait, it indicate that individual under test is homozygous (WW) for dominant trait.

Violet	×	White	
VV		vv	homozygous recessive

Case 2

	v	v
V	Vv Violet	Vv Violet
v	vv White	vv White

50% flowers are violet

50% flowers are white

ratio Ww : ww

Incomplete Dominance : In this, both alleles of a gene at a locus, are partially expressed.

Example : In Mirabilis jalapa.

	Red RR	x	White rr

F1

	R	R
r	Rr Pink	Rr Pink
r	Rr Pink	Rr Pink

Parents	Pink Rr	×	Pink Rr
Gametes			

F2 generation

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

Phonotypic ratio	Red	:	Pink	:	White
	1	:	2	:	1
	RR	:	Rr	:	rr
Genotypic ratio	1	:	2	:	1

In incomplete dominance, phenotypic ratio is equal to the genotypic ratio.

Multiple Allelism : It is a phenomenon in which a single character is governed by more than two alleles.

Example :

- ABO blood groups are controlled by gene I
- 'I' has three alleles—I^A, I^B, and *i*

I^A and I^B alleles produce slightly different form of sugar present on plasma membrane of red blood cells.

- In allele 'i' do not produce any sugar.
- In any diploid individual only two alleles can be found. So multiple alleles can be detected only in a population.

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

There are 3 different alleles, 6 different genotypes control 4 different type of Phenotypes :

Blood Group	Genotype	Blood Type
A	I^AI^A, $I^A\underline{i}$	A
B	I^BI^B, $I^B\underline{i}$	B
AB	I^AI^B,	both A & B
O	$\underline{ii}$	O

In humans, blood group AB shows co-dominance as both the alleles I^A and I^B express themselves fully in presence of each other.

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

- **Linkage-** is the tendency of genes on a chromosome to remain together.
- Linked genes occur on the same chromosome.
- They lie in linear sequence on the chromosome - There is a tendency to maintain the parental combination of genes except for accessional choosers.
- Strength of linkage between genes is inversely proportional to the distance between the two.

Recombination : is the generation of non-parental gene combinations to the offsprings.Tightly linked genes show very low recombination frequency. Loosely linked genes show higher recombination frequency.

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

Linkage and Recombination.

T. H. Morgan carried out several dihybrid crosses in *Drosophila melanogastor.* Two of them are given below :

Cross–I : Yellow-bodied and white eyed females crossed with brown-bodied, red eyed males (wild type)

- F_1 Progeny intercrossed and F_2 generation ratio deviated from 9 : 3 : 3 : 1 (two genes didn't segregate independently)
- The Parental combinations were 98.7% and recombinats were 1.3%

Conclusion : The two genes (body colour and eye-colour) are tightly linked ; results in less crossing over and less no. of non-parental progeny.

Cross-II : White bodied female with miniature wings and yellow-bodied male with normal wings (wild type) were crossed.

- The parental combinatios were 62.8% while the recombinants were 37.2%.

Conclusion : The two genes (body colour & wing's size) are loosely-linked ; results in more crossing over and more no. of non-parental progeny.

The seven character : Mendel Studied in garden pea, had their genes located on different (non-homologous) ehromusomes or far apart on the same chromosome that they got separated by crossing over. So he was lucky that he could not note linkage and propose law of independent assortment.

Chromosomal basis of sex Determination :

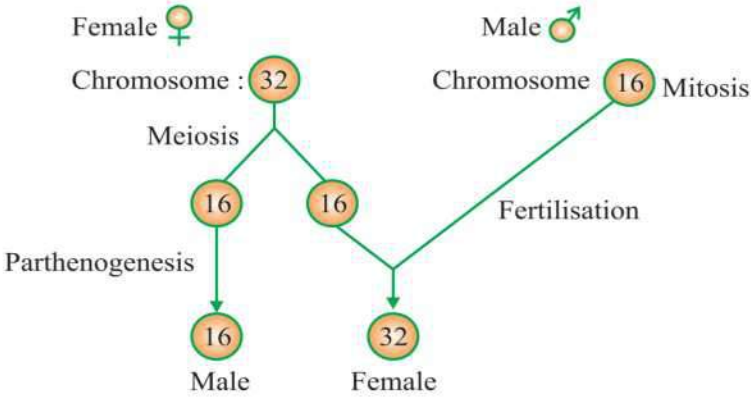
(i) XX-XY type - Female homogametic i.e. XX and male heterogametic i.e. XY in *Drosophila*, humans.

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

(iii) ZW-ZZ type—The females are heterogametic and have one Z and one W chromosome. The males are homogametic with a pair of Z chromosomes besides autosomes e.g., birds.

(iv) ZO-ZZ type—Females are heterogametic and produce 2 types of eggs - (A + Z) and (A + O). The males are homogametic with all the sperms having (A + Z) e.g. moths and butterflies.
here A = autosome

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



There are three types of individuals :

1. Queen — diploid
   - developed from fertilized egg
   - functional female
2. Worker — diploid
   - developed from fertilized egg
   - non-functional female
3. Drone (male) — haploid
   - developed from unfertilized egg parthenogenetically
   - functional Male.

Pedigree Analysis

A record of inheritance of certain genetic traits for two or more generation presented in the form of diagram or family tree is called pedigree.

Usefulness of Pedigree Analysis

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

Mendelian disorders

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

1. **Haemophilia :** Sex linked recessive disease which is transmitted from unaffected carriers female to male progeny. A single protein is affected which is a part of the cascade of proteins involved in the clotting of blood.

   $X^h Y$ = affected male

   $X^h X$ = carrier female

   The heterozygous female for haemophila may transmits the disease to her sons. The possibility of a female suffering from the disease is extremely rare (only when the mother of the female is a carrier is $X^h X$ and father is haemophilic i.e. $X^h Y$.

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

Hb^A Hb^A. Normal Hb^S Hb^S sufferer

Hb^A Hb^S. Apparently unaffected/carriers

Example Hb^AHb^S (Carrier) × Hb^A Hb^A (Normal)

	Hb^A	Hb^A
Hb^A	Hb^AHb^A	Hb^AHb^A
Hb^S	Hb^AHb^S	Hb^AHb^S

Phenylketonuria : Inbron error of metabolism, autosomal recessive trait. Affected individual lacks an enzyme that converts amino acid phenylalanine into tyrosine. Phenylalanine is accumulated and converted into phenylpyruvic acid which accumulates in brain resulting in mental retardation.

Thalassemia : Thalassemia is autosome linked recessive disease. This disorder caused by defers in the synthesis of globin chain. Thalassemia is of three types—Alpha (α) Thalassemia, Beta (β) Thalassemia and delta(s).

- In alpha Thalassemia production of alpha globin chain is affected. This Thalassemia is controlled by genes HBA1 and HBA2 located on chromosome 16th of each parent. Thalassemia occurs due to mutation or deletion of one or more of the four genes.
- In Beta Thalassemia production of (β-globin chain is affected, this thalassemia is controlled by gene HBB located on 11th chromosome of each parent. It occurs due to one or both HBB genes
- In Thalassemia very few globin is synthesized and is quantitative problem whereas in sickle cell anaemia there is a synthesis of incorrectly functioning globin and is a qualitative disorder.

Delta Thalassemia- It is caused due to defective allele of HBD gene present on chromosome II that form delta chain of heamoglobius. The effect of this thalassemia is minor as the adults have about 3% harmoglobius consisting of a and of chains.

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

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

X^c X^c — Colour blind female

X X^c — Carrier female

X^c Y — Colour blind male

Y Chromosome of male do not carry any gene for certain vision.

Inheritance Pattern in Colour Blindness

Father		Mother		Son		Daughter	
Pheno-type	**Geno-type**	**Pheno-type**	**Geno-type**	**Pheno-type**	**Geno-type**	**Pheno-type**	**Geno-type**
Normal	XY	Carrier	X^CX	Normal	XY	Normal	XX
				Colour-blind	X^CY	Carrier	X^CX
Normal	XY	Colour-blind	X^CX^C	Colour-blind	X^CY	Carrier	X^CX
Colour-blind	X^CY	Normal	XX	Normal	XY	Carrier	X^CX
Colour-blind	X^CY	Carrier	X^CX	Colour-blind	X^CY	Colour-blind	X^CX^C
				Normal	XY	Carrier	X^CX

Inheritance Pattern in Haemophilia

Father		Mother		Son		Daughter	
Pheno-type	Geno-type	Pheno-type	Geno-type	Pheno-type	Geno-type	Phen-type	Geno-type
Normal	XY	Haemophilic	X^hX^h	Haemophilic	X^hY	Carrier	X^hX
Normal	XY	Carrier	X^hX	Normal	XY	Normal	XX
				Haemophilic	X^hY	Carrier	X^hX
Haemop-hilic	X^hY	Carrier	X^hX	Normal	XY	Carrier	X^hX
				Haemophilic	X^hY	Haemop-hilic	X^hX^h
Haemop-hilic	X^hY	Normal	XX	Normal	XY	Carrier	X^hX

Chromosomal Disorder :

1. **Down's syndrome :** Trisomy of chromosomes number 21 (2n + 1) Affected individual is short statured with small round head, furrowed tongue, partially open month, broad palm. Physical, psychomotor and mentaly development is retarded.
2. **Klinefelter's syndrome :** extra copy of X chromosome ; karyotype XXY. Affected individual has overall masculine development with feminine characters like gynaecomastia (development of breast) and is sterile

   44 autosomes + xxy = 47 chromosomes
3. **Turner's syndrome :** has absence of one X chromosome i.e. 45 with XO. Affected females are sterile with rudimentary ovaries and lack secondary sexual characters.

   44 autosomes +xo= 45 chromosomes

Pleiotropy

The ability of a gene to have multiple phenotypic effects because it influences a number of characters simultaneously is known as pleiotropy. The gene having a multiple phenotypic effect because of its ability to control expression of a number of characters is called pleiotropic gene. E.g. in Garden Pea, the gene which controls the flower colour also controls the colour of seedcoat and presence of red spot in the leaf axis.

The disorder phenylketonuria shows pleiotropy.

Polygenic Inheritance

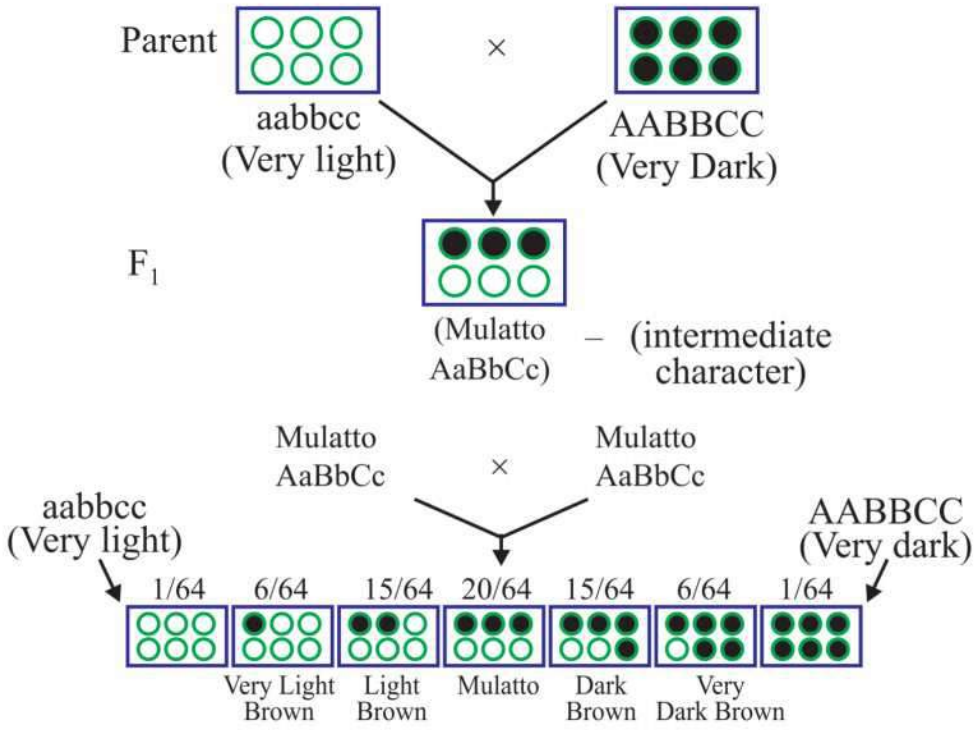
It is a type of inheritance controlled by three or more genes in which the dominant alleles have cumulative effect with each dominant allele expressing a part of the trait, the full being shown only when all the dominant alleles are present.

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

In polygenic inheritance, a cross between two pure breeding parents produces an intermediate trait in F_1. In F_2 generation, apart from the two parental types, there are several intermediates (gradiations, show a bell shaped curve). F_1 hybrid form 8 kinds of gametes in each sex giving 64 combination in F_2 having 7 phenotypes.

Polygenic inheritance skin tone

3 loci : each has two possible alleles : Aa, Bb, Cc, each capital allele adds one unit of darkness, each lower case allele adds nothing. Parents produce F_1 offsprings with intermediate tone.



Offspring can have tone darker or lighter than either parent

Questions

VSA **(I Mark)**

1. Name the base change and the amino acid change, responsible for sickle cell anaemia.
2. Name the disorder with the following chromosome complement.
   (i) 22 pairs of autosomes + X X Y
   (ii) 22 pairs of autosomes + 21st chromosome + XY.
3. A test is performed to know whether the given plant is homozygous dominant or heterozygous. Name the test and phenotypic ratio of this test for a monohybrid cross.
4. Write the number of chromosomes body cells of honey bee workers and drone have.

Multiple Choice Question

5. When red blood corpuscles containing both A and B antigens are mixed I a person's blood serum, they agglutinate. The blood group of the person is
   a) O
   b) AB
   c) B
   d) A
6. The offspring produced from a marriage have only 0 or A blood groups. Of the genotypes given below, the possible genotypes of the parents would be
   a) ii and ii
   b) I^AI^A and ii
   c) l^Ai and ii or l^Ai and l^Ai
   d) I^AI^A and I^Ai
7. The gene of sickle cell anaemia is inherited by
   a) Haemoglobin
   b) Autosomes and sex chromosomes
   c) Sex chromosomes
   d) Autosomes

8. More men suffer from colour blindness than women because

a) women are more resistant to disease than men

b) the male sex hormone testosterone causes the disease

c) the colour-blind gene is carried on the 'Y' chromosome

d) men are hemizygous and one defective gene is enough to make them Colour blind

9. if a cross between two individuals produces offsprings with 50% dominant character P and 50% recessive character p the genotypes of parents are

a) Pp x Pp

b) Pp xpp

c) PP x x pp

d) PP x Pp

Assertion-Reason Questions

10. **Assertion:** Aneuploidy is the phenomenon of gain or loss of one or More chromosome(s).

Reason: Sometimes homologous chromosomes are unable to separate

a) Both assertion and reason are true, and reason is the correct explanation of assertion.

b) Both assertion and reason are true, but reason is not the correct explanation of assertion

c) Assertion is true but reason is false.

d) Both assertion and reason are false.

11. Assertion: Genes of body-colour and eye-colour of Drosophila melanogaster are tightly linked. Reason: Genes of body-colour and wing-size of Drosophila melanogaster are loosely linked.

a) Both assertion and reason are true, and reason is the correct explanation of assertion.

b) Both assertion and reason are true, but reason is not the correct explanation of assertion.

c) Assertion is true but reason is false.

d) Both assertion and reason are false.

SA-I **(2 Marks)**

12. Identify the sex of organism as male or female in which the sex chromosome are found as (i) ZW in bird (ii) XY in Drosophila (iii) ZZ in birds, (iv) XO in grasshopper.

13. The human male never passes on the gene for haemophilia to his son. Why is it so ?

14. Mention four reasons why Drosophila was chosen by Morgan for his experiments in genetics.

15. Differentiate between point mutation and frameshift mutations.

SA-II **(3 Marks)**

16. A woman with O blood group marries a man with AB blood group

(i) Work out all the possible phenotypes and genotypes of the progeny.

(ii) Discuss the kind of dominance in the parents and the progeny in this case.

17. Give reasons for success of Mendel.

18. In Mendel's breeding experiment on garden pea, the offspring of F_2 generation are obtained in the ratio of 25% pure yellow pod, 50% hybrid green pods and 25% green pods State (i) which pod colour is dominant (ii) The Phenotypes of the individuals of F_1 generation, (iii) Workout the cross.

LA **(5 Marks)**

19. A dihybrid heterozygous round, yellow seeded garden pea (*Pisum sativum*) was crossed with a double recessive plant.

(i) What type of cross is this?

(ii) Work out the genotype and phenotype of the progeny.

(iii) What principle of Mendel is illustrated through the result of this cross?

Case Based Questions

20. Read the following and answer the questions given below:

TURNER'S SYNDROME

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

(a) What organ does turner syndrome affect?

(b) What is the couse of turner syndrome?

(c) What are clinical features of turner syndrome?

OR

Who suffer from turner syndrome? It is an example of which plaid?

21. Read the following and answer the questions given below:

In case of honey bee, the male is haploid while the female is diploid. Similar conditions are found in some other insects like ants and wasps. Male insects are haploid because they develop parthenogenetically from unfertilised eggs. The phenomenon is called arrhenotoky. Meiosis does not occur in the formation of sperms. Females grow from fertilized egg and are hence diploid.

Queen bee picks up all the sperms from drone during nuptial flight and stores them in seminal receptacle. When the queen visits drone cells it lays eggs but seminal receptacles fails to emit the sperms. The male honey bee develops parthenogenetically from these unfertilised eggs. However in all other cells, i.e., cells of workers, the female lays eggs and sperms are emitted properly from its seminal receptacle, upon these eggs leading to their fertilization. Hence except drones other honey bees (worker and queen) are diploid.381718.

(i) The 2n number of chromosomes for honey bee is 32. How many chromosomes will be present in the cells of drone?

(a) 64 (b) 32
(c) 16 (d) 8

(ii) The purpose of queen and drone for performing nuptial flight is

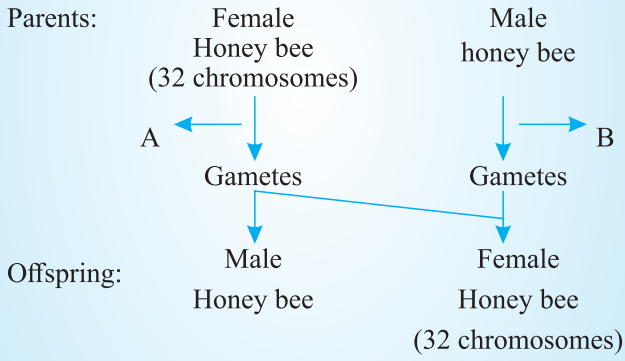
(a) to estabilish a new life (b) to perform copulation
(c) to collect pollen and necter (d) all of these

(iii) What type of cell division is involved in spermatogenesis in honey bee?

(a) Meiosis (b) Endomitosis
(c) Mitosis (d) None of these

(iv) Which of the following factors is responsible for the fertilized eggs to develop into queen or worker?

(a) Amount of temperature for incubation of eggs
(b) Type of nutrition given to the larvae
(c) Type of sperm performing fertilization of eggs
(d) All of these



(a) What forms the basis of sex determination in honey bees? What term is given to this method of sex

(b) Identify the type of cell division at 'A' and 'B' occuring during gamete formation in the female and malehoney bees respectively.

(c) Name the scientist who conducted the cytological studies during gametogenesis in insects. What did heobserve?

OR

(c) How many chromosomes does a male honey bee possesses? Give reason for the difference in the number of chromosomes between males and females of honey bees.

Answers

VSA (1 Mark)

1. GAG changes GUG, Glutamic acid is substituted by valine.
2. (i) Klinefetter's Syndrome (ii) Down's syndrome
3. Test cross 1 : 1.
4. Honey bee workers : 32 and Drones : 16 chromosomes

Answer of Multiple-Choice Question :

5. (a) 6. (c) 7. (d) 8. (d) 9. (b)

Answer of Assertion-Reason Question :

10. (a) 11. (b)

SA-I (2 Marks)

12. (i) Female (ii) Male (iii) Female (iv) Male
13. The gene for haemophilia is present on X chromosome. A male has only one X chromosome which he receives from his mother and Y chromosome from father. The human male passes the X chromosome to his daughters but not to the male progeny (sons).
14. (i) Very short life cycle (2-weeks)
    (ii) Can be grown easily in laboratory
    (iii) In single mating produce a large no. of flies.
    (iv) Male and female show many hereditary variations
    (v) It has only 4 pairs of chromosomes which are distinct in size and shape.
15. **Point Mutations :** Arises due to change in a single base pair of DNA e.g., sickle cell anaemia.

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

SA-II (3 Marks)

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

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

10. (i) He used large samples for his experiments.

(ii) He selected only pure breeding varieties.

(iii) He choose the character which had distinctive contrasts.

(iv) He selected pea plant which can be cross-bred as well as self bred.

(v) Use of statistical methods and law of probability.

(i) Green pod colour is dominant

11. (ii) Green pod colour

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

Gametes (G) (g)

F1 generation Gg (Hybrid green)

Gametes (G) (g) X (G) (g)

F2 generation GG Gg Gg gg

Phenotypic ratio 3 : 1

Genotypic ratio 1 : 2 : 1

LA (5 Marks)

12. (i) It is a dihybrid test cross

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

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

Gametes	RY	Ry	rY	ry
F1 progeny	RrYy Round, Yellow	Rryy Round and green	rrYy wrinkled, yellow	rryy wrinkled, green.

Phenotypic ratio	1	:	1	:	1	:	1
Genotypic ratio	1	:	1	:	1	:	1

F_2 Progeny

Phenotypic ratio	Round yellow	:	Round green	:	Wrinkled yellow	:	Wrinkled green
Genotypic ratio	9	:	3	:	3	:	1

(iii) Principle of Independent Assortment.

20. (a) Tumer syndrome affect veins, heart, eyes bones and kidneys.
    (b) Loss of 'x' chromosome 44 + x0
    © Short neck, underdeveloped breast, no menstral cycle, rudimentary overlies.
    (d) It is an example of monosomy in females.
21. No. of sets of chromosome and received by an individual called as haplodiploid system of sex determination.
    (a) A – Meiosis
    (b) B – Mitosis
    (c) Henking. He observe a specific nuclear structure all through Spermatogenesis in few Insects.

OR

A male honey bee develops pantheon genetically C without fertlisation /from an ovum. Hence haploid. A female honey bee develops from fertilized eg. so diploid (32 chromosome.)

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