CHAPTER 5

PRINCIPLES OF INHERITANCE AND VARIATION

POINTS TO REMEMBER

Allele : Various or slightly different forms of a gene, having same position on 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) and wrinkled and green (rryy) pea seeds

Incomplete dominance : When one of the two alleles of a gene is incompletely dominant over the other allele.

Co-dominance : When two alleles of a gene are equally dominant and express themselves even when they are together.

Multiple allelism : When a gene exists in more than two allelic forms e.g., gene for blood group exist in three allelic forms, I^A, I^B and i.

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.

Mutation : The sudden heritable change in the base sequence of DNA, or structure of chromosome or a change in the number of chromosomes.

Pedigree Analysis : The analysis of the distribution and movement of trait in a series of generations of a family.

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

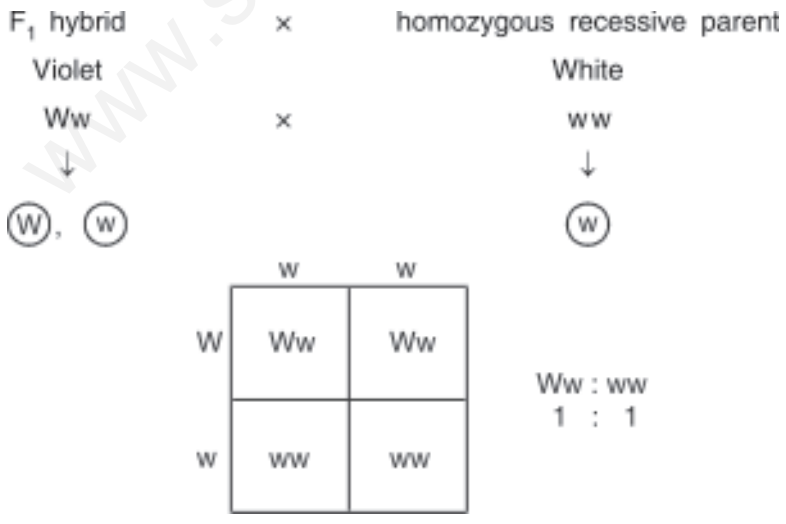
Law of Dominance : When two individuals of a species differing in a pair of contrasting characters/traits are crossed, the trait that appears in the F_1 hybrid is dominant and the alternate from that remain hidden, is called recessive.

Law of Segregation : 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.

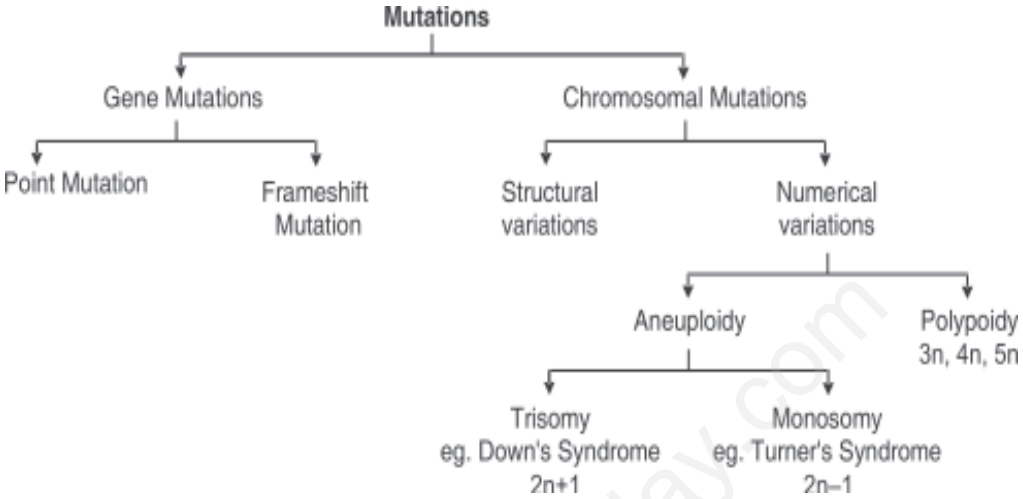
Law of Independent Assortment : In the inheritance of two pairs of contrasting characters, the factors of each pair of characters segregate independently of the factors of the other pair of characters.

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

The progeny of monohybrid test cross ratio is 1 : 1 while the dihybrid test cross ratio is 1 : 1 : 1 : 1.



Use of Test Cross : The test cross is used to find the genotype of an organism.



Incomplete dominance : It is the phenomenon where none of the two contrasting alleles is dominant but express themselves partially when present togoether in a hybrid and somewhat intermediate.

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 co-dominance. Example : Human blood groups.

Blood Group	Genotype
A	I^AI^A, I^Ai
B	I^BI^B, I^Bi
AB	I^AI^B
O	ii

In human blood, there are six genotype and four phenotypes.

Chromosonal Theory of Inhertance : proposed by Suttan 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 in the same chromosome

– They lie in linear sequence in 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.

Chromosomal basis of sex determination

(i) XX - XY type - female homogametic ie XX and male heterogametic ie. XY is *Drosophila*, humans.

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

(iii) ZW - ZZ type - The females are hetegametic and have one Z and one W chromosome. The males are homogametic with a pair of Z chromosomes besides autosomes eg - birds.

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.

Mendelian disorders

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

1. **Haemophilia –** sex linked recessive disease which is transmitted from unaffected carriers female to male pregnancy. A single protein is affected that is a part of the cascade of proteins involved in the clothing of blood.

   X^h Y – Sufferer male

   X^h X – carrier female

   The heterozygous female for haemophilia may transmit 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 ie X^hX and father is haemophilic ie. X^hY.

2. **Sickle - cell anaemia :** This is an autosome linked recessive trait. The defect is caused by substitution of glutamic acid by valine at the 6^{th} position of the beta globin chain of the haemoglobin molecule. The mutant Hb molecule undergoes polymerisation under low oxygen tension causing 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^A Hb^s – Apparently unaffected, carriers Hb^s Hb^s – sufferer

   Phenylketonuria – Inborn 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.

Chromosomal disorders

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

Down's syndrome – Trisomy of chromosome number 21.

Affected individual is short statured with small round head, furrowed tongue, partially open month, broad palm. Physical, psychomotor and mental development is retarded.

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.

Turner's syndrome – has absence of one X chromosome ie. 45 with XO. Affected females are sterile with rudimentary ovaries and lack secondary sexual characters.

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.

Eg. in Garden Pea, the gene which controls the flower colour also controls the colour of seed coat and presence of red spot in the leaf axil.

POLYGENIC INHERITANCE

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

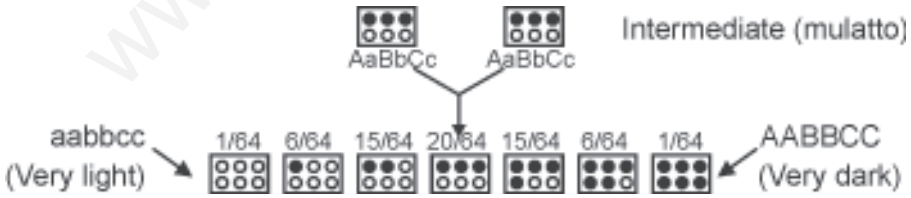
Eg. Kernel colour in wheat, skin colour in human beings, height in humans, cob length in maize etc.

In polygenic inheritance, a cross between two pure breeding parents produces an intermediate trait in F1. In F2 generation, apart from the two parental types, there are several intermediates (gradiations, show a bell shaped curve). F1 hybrid form 8 kinds of gamete in each sex giving 64 combination in F2 having 7 genotype and phenotype.

Polygenic inheritance of 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 with intermediate tone



Offspring can have tone darker or lighter than either parent

QUESTIONS

VSA (1 MARK)

1. Give any two reasons for the selection of pea plants by Mendel for his experiments.
2. Name any one plant that shows the phenomenon of incomplete dominance during the inheritance of its flower colour.
3. Name the base change and the amino acid change, responsible for sickle cell anaemia.
4. Name the disorder with the following chromosome complement.
   (i) 22 pairs of autosomes + X X Y
   (ii) 22 pairs of autosomes + 21st chromosome + XY.
5. A haemophilic man marries a normal homozygous woman. What is the probability that their daughter will be haemophilic?
6. 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.

SA-II (2 MARKS)

7. 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.
8. Mention two differences between Turner's syndrome and Klinefelter's syndome.
9. The human male never passes on the gene for haemophilia to his son. Why is it so?
10. Mention four reasons why *Drosophila* was chosen by Morgan for his experiments in genetics.
11. Differentiate between point mutation and frameshift mutations.

SA-I (3 MARKS)

12. 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.

13. Explain the cause of Klinefelter's syndrome. Give any four symptoms shown by sufferer of this syndrome.

14. In Mendel's breeding experiment on garden pea, the offspring of F2 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 F1 generation. (iii) Workout the cross.

LA (5 MARKS)

15. 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?

ANSWERS

VSA (1 MARK)

1. (i) Many varieties with contrasting forms of characters

   (ii) Can easily be cross pollinated as well as self pollinated.

2. Dog flower (Snapdragon or Antirrhinum sp.)

3. GAG changes as GUG, Glutamic acid is substituted by valine.

4. (i) Klinefelter's Syndrome (ii) Down's syndrome

5. Their daughter can never be haemophilic. (0%).

6. Test cross 1 : 1.

SA-II (2 MARKS)

7. (i) Female; (ii) Male; (iii) Female (iv) Male

8. **Turner's Syndrome :** The individual is female and it has 45 chromosomes i.e., one X chromosome is less.

   Klinefelter's Syndome : The individual is male and has 47 chromosomes i.e., one extra X chromosome.

9. 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).

10. (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.

11. **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-I (3 MARKS)

12. (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.

13. **Cause :** Presence of an extra chromosome in male i.e., XXY.

    Symptoms : Development of breast, Female type pubic hair pattern, poor beard growth, under developed testes and tall stature with feminised physique.

14. (i) Green pod colour is dominant

    (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)

15. (i) It is a dihybrid test cross

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

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

	Gametes	RY	Ry	rY	ry
F_1 progeny	ry	RrYy Round, Yellow	Rryy Round and Green	rrYy Wrinkled Yellow	rryy Wrinkled, Green
Phenotypic ratio	:	1	: 1	: 1	: 1
Genytopic ratio	:	1	: 1	: 1	: 1

(iii) It illustrates the Principle of independent assortment.