HEREDITY AND VARIATION

'Like begets like' is an important and universal phenomenon of life. It means that living beings produce offsprings of their own kind. The branch of science which deals with the study of hereditary traits and variations is termed as genetics. Inheritance is the process by which characters of traits are passed from parent to their progeny, i.e. from one generation to the next. It is the basis of heredity. Gene is the unit of inheritance. Variation is the degree by which progeny differs from their parents.

1. Genetic Terminology

A. **Gene or Factor** is a structure located on chromosomes which transfers a hereditary character from one generation to another generation. It is the unit of DNA.

B. The alternative forms of genes for a single trait or character are called **alleles**, e.g. colour of flower (red and white),stem height (tall and dwarf).

C. **Homozygous** A diploid condition, in which both alleles are same, e.g. TT, tt.

D. **Heterozygous** A diploid condition, in which both alleles are different, e.g. Tt.

E. **Monohybrid Cross**A cross in which only one pair of character is taken.

F. **Dihybrid Cross**A cross in which two pairs of characters are taken.

G. **Reciprocal Cross** A cross where sexes of parents are reversed.

H. **Back Cross**A cross between homozygous dominant parent and heterozygous F_1 - hybrid is known as back cross.

I. **Test Cross**A cross between homozygous recessive parent and heterozygous F_1 - hybrid is known as test cross. By this cross, it can be identified that a plant is homozygous or heterozygous.

2. Mendel's Experiments

Mendel conducted his experiments on garden pea for seven years to investigate inheritance pattern. He had applied statistical analysis and mathematical logic to his experiments.

The following contrasting traits were studied by Mendel in pea.

Character	Dominant	Recessive

Stem height	Tall	Dwarf
Flower position	Axial	Terminal
Pod colour	Green	Yellow
Pod shape	Inflated	Constricted
Seed shape	Round	Wrinkled
Flower colour	Purple	White
Seed coat colour	Yellow	Green

3. Laws of Mendel

On the basis of his observations, Mendel proposed three general rules to consolidate his understanding of inheritance in monohybrid cross. These laws are given below

A. **Law of Dominance** It states following rules

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

(ii) Factors occur in pairs.

(iii) In a pair of dissimilar factors, one member of the pair is dominant while the other is recessive.

B. **Law of Segregation** It states that the parents contain two alleles during gamete formation. The factors or alleles of a pair segregate from each other, such that a gamete receives only one of the two factors. The following table shows the phenotypic and genotypic ratio(s) of a monohybrid cross for better understanding of the above two laws

Generations/Ratio	F_1	F_2
Phenotypic	1	3 : 1
Genotypic	1	1 : 2 : 1

C. **Law of Independent Assortment**It states that when two pairs of traits are combined in a hybrid, segregation of one pair of characters is independent of the other pair of characters at the time of gamete formation. It also gets randomly rearranged in the offspring producing both parental and new combinations of characters. The following table explains the law of independent assortment on the basis of ratio(s) obtained through dihybrid cross.

Generations/Ratio	F1	F2
Phenotypic	1	9 : 3 : 3 : 1
Genotypic	1	1 : 2 : 2 : 4 : 1 : 2 : 1 : 2 : 1

4. Deviations from Mendel's Laws

All patterns of inheritance could not be explained on the basis of Mendel's principles which are as follows

(i) **Incomplete Dominance**It is a phenomenon in which phenotype of the F_1 -hybrid offspring does not resemble any of the parent but is an intermediate between the expression of two alleles in their homozygous state. Both phenotypic and genotypic ratios is 1 : 2 : 1, e.g. flower colour in Mirabilis jalapa.

(ii) **Codominance**It is a phenomenon in which two alleles express themselves independently when present together in an organism, e.g. ABO blood group in humans.

(iii) **Multiple Alleles** More than two alternative forms (allele) of a gene occupying the same locus on a chromosome in a population are known as multiple alleles, e.g. ABO blood grouping.

Human Blood Groups, their Genotype and Transfusion

Blood groups	Antigen in RBC	Antibody in plasma	Can give blood to	Can take blood from	Gene type
O (universal donor)	None	a,b	O, A, B, AB	O	$I^{\circ} I^{\circ}$
A	A	b	A, AB	O, A	$I^A I^A$ or $I^A I^{\circ}$
B	B	a	B, AB	O, B	$I^B I^B$ or $I^B I^{\circ}$
AB (universal receiver)	A or B	None	AB	O, A, B, AB	$I^A I^B$

(iv) **Polygenic Inheritance** The inheritance of trait controlled by multiple genes where each gene contributes a little to the phenotype, e.g. human skin colour.

(v) **Pleiotropy**It is the phenomenon in which a single gene may produce multiple or more than one phenotypic effects, e.g. sickle-cell anaemia, white eye in Drosophila, Phenylketonuria (PKU).

5. Chromosomal Theory of Inheritance

It was proposed by Boveri and Sutton in 1902. It is the fundamental unifying theory of genetics which identifies chromosomes as the carriers of genetic material.

6. Crossing Over

The exchange of genetic material between non-sister chromatids of homologous chromosomes is known as crossing over.

Linkage and Recombination The physical association of two genes is called linkage while the generation of non-parental gene combinations is called recombination.

7. Sex-Determination

The establishment of sex through differential development in an individual at the time of zygote formation, is called sex-determination. There are following types of sex-determination mechanisms observed in various organisms

Male Heterogamety

■ XX-XO type is seen in insects like grasshopper, etc. It is an example of male heterogamety where males have only one X-chromosome (called XO), whereas females have two X-chromosomes (XX).

■ XX-XY type is seen in insects like Drosophila melanogaster and humans. Males have XY type chromosomes while females have XX type (homomorphic) chromosomes.

Female Heterogamety

■ ZZ-ZW type is seen in birds, fowls and fishes. It is an example of female heterogamety because female produces two different types of gametes.

■ ZZ-ZO type female is heterogametic (ZO) and male is homogametic (ZZ), e.g. in butterflies and moths.

Haplo-Diploidy is a sex-determination mechanism in which males develop from unfertilised eggs and are haploid. Females develop from fertilised eggs and are diploid. It is common in insects like honeybee, wasps, etc.

Note • On the basis of type of allosomes present in the gamete, the parents can be of two types

(i) Homogametic produces similar gametes.

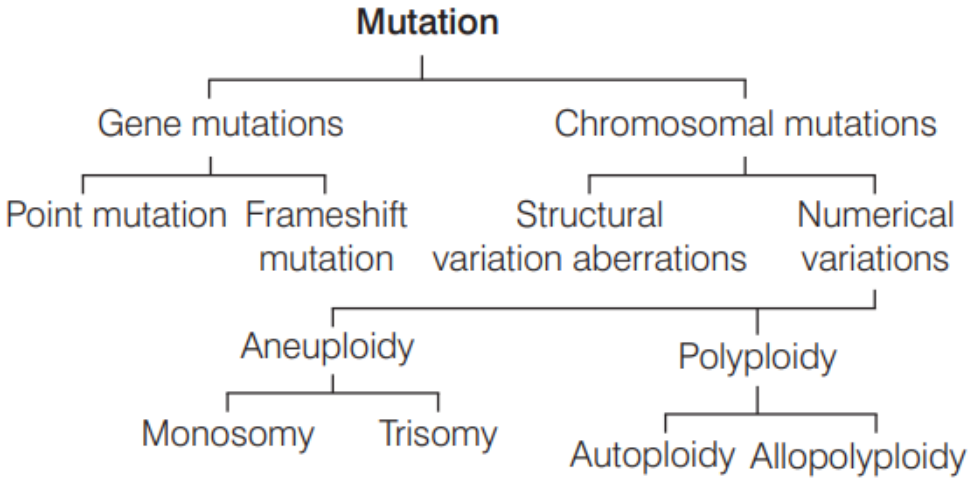
(ii) Heterogametic produces different gametes.

8. Sex-Determination in Human

Males have two types of gametes, i.e. X and Y, while females have only one type of gamete, i.e. XX. If an ovum gets fertilised with a sperm carrying X-chromosome, the zygote develops into a female (XX). If an ovum fertilises with a sperm carrying Y-chromosome then it will develop into a male (XY). Hence, the type of sperm, which fertilises the ovum determines the sex of a child.

9. Mutation

It is a sudden, stable and inheritable change in genetic material or DNA sequences of an organism. The organism which undergoes mutation is called mutant. The factors, i.e. chemical and physical which induce mutations are called mutagens, e.g. UV-radiations, etc. The mutations are of following types



- Point mutation arises due to change in single base pair of DNA.
- Frameshift mutation refers to deletions and insertions of base pairs in DNA.
- The loss or gain of a segment of DNA, results in structural alteration in chromosomes because genes are located on the chromosomes.
- When the members of a homologous pair of chromosomes fail to segregate during meiosis, aneuploidy occurs, i.e. loss or gain of one or more chromosomes. Due to this, there may be monosomy, i.e. lack of one chromosome of normal complement or trisomy, i.e. three instead of the normal two chromosomes.
- Polyploidy occurs when there is failure of cytokinesis after telophase stage of cell division. It results in an increase in a whole set of chromosomes in an organism.

10. Pedigree Analysis

The analysis of traits in several generations of a human family in the form of a family tree or a diagram is known as pedigree analysis.

11. Genetic Disorders

The disorders or illness which are caused by one or more abnormalities in autosomes or sex chromosomes of a person are called autosomal disorders and sex-linked disorders, respectively. It can be divided into the following

A. **Mendelian Disorders** These are determined by alteration or mutation in a single gene. These disorders can be recessive or dominant. Some diseases are described below

(i) **Colour Blindness** Person suffering from this disorder is not able to distinguish between red and green colour. Gene controlling colour blindness is located on X-chromosome and is recessive.

(ii) **Haemophilia** (bleeder's disease) Person suffering from this disorder is not able to form blood clots when hurt or wounded. The continuous bleeding causes death of the person. This disorder is caused due to X-linked recessive gene.

(iii) **Sickle-cell Anaemia** It is an autosomal linked recessive trait that can be transmitted from parents to the offsprings, when both the partners are carrier for the gene (heterozygous). In this, the substitution of valine by glutamic acid changes the shape of the RBCs to a sickle like structure.

(iv) **Phenylketonuria** (PKU) is an inborn error metabolism, which is inherited as an autosomal recessive trait. The affected individual lacks the enzyme that converts phenylalanine to tyrosine. Its gene is associated with the 12th chromosome.

(v) **Thalassemia** is an autosomal recessive disease, which occurs due to either mutation or deletion of genes. It results in reduced rate of synthesis of one of the globin chains (α or β) of haemoglobin.

B. **Chromosomal Disorders** These are caused by the absence, excess or abnormal arrangement of one or more chromosomes. Some of the disorders are described below

(i) **Down’s Syndrome or Mongolism** It is caused by the presence of an extra copy of chromosome number 21 in autosomes, i.e. trisomy. An individual suffering from Down's syndrome is short statured, have furrowed tongue, slow motor and mental development.

(ii) **Turner's Syndrome**It is caused by the absence of one X-chromosome in females. The females have 45 (44+X) chromosomes. Females are sterile with less developed uterus, short height and webbed neck are the symptoms.

(iii) **Klinefelter's Syndrome** It is due to the presence of an extra copy of X-chromosome in males like 44+XXY, 44+XXXY, etc. The affected individuals are phenotypically males. The symptoms include underdeveloped testes, presence of breast-like structures, little growth of hair on face and body, sterility (no sperm formation), mental retardedness, etc.