



PRINCIPLES OF INHERITANCE AND VARIATIONS

I. Mendel's law of inheritance

1. Genetics: The branch of biology which deals with study of heredity and variations is called as a genetics
2. Artificial selection: The selection of plants and animals with desirable characters is called as artificial selection

II. Inheritance of one gene

1. **Hybridization:** The process of combining different varieties of organisms to create a hybrid is called hybridization.
2. **Gene:** is the determinant character of an organism. It is the fundamental physical unit of heredity. A gene is a nucleotide sequence specifies sequence of amino acids in a polypeptide.
3. **Allele:** An alternative form a gene is called allele.
4. **Locus:** The specific place on a chromosome where a gene is located is called as **locus**.
5. **Homozygous:** A diploid organism in which same alleles occur at a given locus of a pair of homologous chromosomes is called as **homozygous**.
6. **Heterozygous:** A diploid organism in which two different alleles occur at a given locus of a pair of homologous chromosomes is called **heterozygous**.
7. **Dominance allele/factor:** factor: It is one of the alleles of a pair alleles, which can express when it is present in heterozygous condition also is called as **dominant allele**.
8. **Recessive allele factor:** It is one of the alleles of a pair alleles which is unable to express it is present in heterozygous condition is called as recessive allele.
9. The genetic constitution of an organism is called **Genotype**.
10. The physical manifestation of a genetic trait (physical appearance) that results from a specific genotype and its interaction with the environment is called **Phenotype**.
11. **Cross:** A mating between two individuals leading to fusion of gametes.
12. **Monohybrid cross:** A cross between two organisms with different trait of a character.
13. **Segregation:** The separation of the two alleles of each gene and their distribution to separate sex cell during meiosis.
14. **Punnett square:** It is a graphical representation to calculate the probability of all possible genotypes of offspring in a genetic cross. It was developed by British geneticist, **Reginald c Punnett**.
15. **Test cross:** A cross conducted between F1 X double recessive parent.
16. **Back cross:** A cross between an individual member of F1 generation and either of its parents.
17. **Law of Dominance:** In a pair of contrasting traits of a character, one is dominant traits and the other is recessive traits. The traits that is expressed in the F1 generation is called dominant traits and the factor for this traits is dominant factor. The other trait which is not expressed in FI generation is called recessive traits and the factor for this is recessive factor.
18. **Law of segregation:** During the formation of gametes due to meiosis the paired unit factors separate or segregated randomly with equal probability, so that each gamete receives only one of these factors.

III. In complete dominance (or) Partial dominance

1. **Incomplete dominance:** The condition in which both alleles of the heterozygote interact influencing the phenotype expressing incomplete phenotype of the parents is called incomplete dominance.
e.g: snap dragon
2. **Co-dominance:** The condition in which the alleles of the heterozygote show individual expression in the phenotype is called co-dominance. e.g: Blood group.



IV. Inheritance of two genes

1. **Dihybrid cross:** The cross involving two pairs of contrasting traits of two characters is called Dihybrid cross.
2. **Law of Independent Assortment:** When two more pairs of contrasting traits are brought together in a cross, the inheritance of one pair of traits is independent of other pair of traits.
3. **Chromosomal Theory of Inheritance** was proposed by Sutton and Boveri in 1909.
4. Chromosomal Theory of Inheritance is a fundamental theory of genetics which identifies chromosomes are carriers of genetic material.
5. The behaviour of genes and chromosomes are parallel to each other.
6. **Homologous chromosomes** are pairs that are similar in length, gene procession and centromere location.
7. **Heterologous chromosomes:** chromosomes pair for different length, with different genes at corresponding loci are called Heterologous chromosomes.
8. **Linkage:** The physical association between genes on chromosomes is called **linkage**.
9. **Recombination:** The exchange of genetic material between non-sister chromatids of homologous chromosomes is called as recombination.
10. **Polygenic inheritance/Quantitative inheritance/Galton's inheritance/ Cumulative inheritance/ multiple factor inheritance:** This type of inheritance is controlled by two or more gene in which each dominant alleles have cumulative effect with each dominant allele expressing a part /unit of the trait.
11. **Pleiotropy:** The genes that control more than one phenotype characters are called **pleiotropy**.

V. Sex determination:

1. **Sex determination:** The property or quality by which organisms are classified as female or male on the basis of their reproductive organs and functions.
2. **Autosomes:** The chromosomes which have no role in the sex determination are called Autosomes.
3. **Allosomes /Sex Chromosomes / Idiochromosomes :** The Chromosomes which are responsible for the sex determination in unisexual organisms is called allosomes.
4. **Heterogamy:** sexual reproduction involving fusion of unlike gametes often differing in size, structure and physiology.
5. **Homogamy:** sexual reproduction involving fusion of same type of gametes in size, structure and physiology.

VI. Mutation

1. **Mutation** - It is a phenomenon which results in alteration of DNA sequences and consequently result in change in the genotype and the phenotype of an organism.
2. **Chromatid:** A Chromatid is one copy of a duplicated Chromosomes, which generally is joined to the other copy by a centromere.
3. **Frame-shift mutation:** A frameshift mutation is a genetic mutation caused by a deletion or insertion in a DNA sequence that shifts the way the sequence is read.
4. **Mutagens:** An agent which causes mutation is called mutagen.

VII. Genetic disorders

1. **Pedigree:** Study of a family history of an individual.
2. **Pedigree analysis:** The study of Pedigree for a few generations for the analysis of transmission of certain exceptional phenotype/disorders and the possibility of their occurrence in a particular generation is known as Pedigree analysis.
3. **Mendelian disorder/hereditary disorder:** Disorder may be due to autosomal or allosomal genes or due to dominant or recessive genes are called mendelian disorders.
4. **Chromosomal disorder/ Syndromes:** Genetic disorder due to Chromosomal aberrations or ploidy changed Chromosomal disorder.



5. **Haemophilia:** It is a sex-linked recessive disease, in which a single protein that is a part of the cascade of proteins involved in the clotting of blood is affected. Due to this in an affected individual a simple cut will result in non stop bleeding.
6. **Cystic fibrosis:** it is an autosomal recessive genetic disorder that affect most critically the lungs and also the pancreas, liver and intestine, It is characterized by abnormal transport of chloride and sodium across an epithelium leading to thick, viscous secretions.
7. **Sex linked genes/characters:** Genes which are present on allosomes are considered as sex linked genes.
8. **Sickle cell anaemia (SCA) or Sickle cell disease (SCD) or Drepanocytosis:** It is an autosomal linked recessive disease, characterized by RBC that assume an abnormal, rigid sickle shape. The defect is caused by the substitution of Glutamic acid (Glu) by Valine (Val) at the 6 po the 6th position of the β globulin chain of the haemoglobin molecule.
9. **Colour blindness (or) Colour vision deficiency:** It is a X-linked recessive gene disease. It is the inability or decreased ability to see colour or perceive colour differences, under normal lighting conditions.
10. **Phenylketonuria (PKU)** is an autosomal recessive metabolic genetic disorder characterized by a mutation in the gene for the hepatic enzyme phenylalanine hydroxylase (PAH) rendering it non functional. this enzyme is necessary to metabolize the amino acid phenylalanine (Phe) to the amino acid tyrosine When PAH activity is reduced, phenylalanine accumulates and converted into phenylpyruvate (also known as phenyl ketone), which can be detected in the urine.
11. **Thalassemia:** It is autosomal recessive blood disorder, characterized by the weakening and destruction of red blood cells people with Thalassemia make less haemoglobin and fewer circulating red blood cells than normal, which results in mild or severe anaemia.
12. **Aneuploidy:** Aneuploidy is a type of chromosomal abnormality, in this an extra or missing chromosome is a common.
13. **Polyploidy:** Containing more than two paired (homologous) sets of chromosomes are called polyploidy.
14. **Trisomy ($2n+1$):** it is a type of polysomy in which there are three instances of a particular chromosomes instead of the normal two. It is a type of aneuploidy.
15. **Monosomy ($2n-1$):** It is a form of aneuploidy with the presence of only one chromosome (instead of the typical two in humans) from a pair.
16. **Down's syndrome (DS):** Also known as trisomy 21, is a genetic disorder caused by the presence of all or part of a third copy of chromosomes 21. It is typically associated with a delay in cognitive ability (mental retardation) and physical growth and a particular set of facial characteristics.
17. **Kleinfelter's syndrome: Also 45 XXX or XXX syndromes,** is a genetic disorder in which there is at least one extra X chromosome to a standard human male karyotype for a total of 47 chromosome rather than the 46 found in genetically normal humans. Sympyoms include hypogonadism, gynacomastism etc.
18. **Karyotype:** A karyotype is the number and appearance of chromosomes in the nucleus of a eukaryotic cell. The term is also used for the complete chromosomes in a species or an individual organism.
19. **Hypogonadism:** It is a medical term which describes a diminished functional activity of the gonads-the testes and ovaries in males and females respectively that may result in diminished sex hormone biosynthesis and impaired gametes production and or / regulation.
20. **Gynecomastia:** It is the benign enlargement of breast tissue in males.
21. **Turner's syndrome or Ullrich - Turner syndrome or Gonadal dysgenesis or 45X.** It is a chromosomal abnormality in which all part of one of the sex chromosomes is absent in some cases the chromosome is missing in some cells but not others a condition referred to **mosaicism** or **Turner**.