

GENETICS

Classical Genetics

1. Germinal variations affect the reproductive cells. These are also called as Fluctuating variations, *Sports or Saltations*. Germinal variations are heritable. *Genome* denotes to the haploid set of chromosomes in a cell. *Test cross* is the back cross of F1 generation with the Recessive parent. Alleles are the alternate forms of a gene. *Locus* is the position of a gene.
2. Correns and Tschermak rediscovered Mendelism. Bateson and Punnett discovered Complementary action of genes. Davenport discovered Polygenic inheritance (Skin color) in man. Waldeyer coined the term Chromosome. Balbiani discovered Polytene chromosomes in *Drosophila*. Muller discovered mutagenic property of X-rays. Hershey and Chase conducted experiments in viral replication. Jacob and Monod proposed the Operon concept. Alec Jeffrey introduced DNA finger printing. Kari Mullis discovered Polymerase chain reaction (PCR).
3. *Codominance* is the condition in which two dominant genes express together. Eg. AB blood group in man, Coat color in Cattle etc. Incomplete dominance is the partial expression of two dominant genes to produce a mixture of two characters. Eg. Sickle cell anemia, Pink flower in *Mirabilis*. *Polygenes* (Multiple genes) are groups of genes controlling the same character. Eg. Skin color in man. *Multiple alleles* are alternate forms of a gene expressing the same character. Eg. Blood groups in man, Coat color in Rabbit etc. *Epistatic gene* suppresses the character of a gene (Hypostatic Gene). Eg. Coat color in Dogs, Coat color in Mice. *Pleiotropism* is the condition in which a gene produces multiple effects. Eg. Gene for sickle cell anemia, Eye color in *Drosophila*. *Lethal genes* are harmful genes destroying the possessor. Eg. Thalassemia and Huntington's chorea in man, leaf color in Snapdragon (Homozygous dominant). Some times homozygous recessive genes become lethal. Eg. Congenital Ichthyosis and Amaurotic idiocy in man.
4. *Pedigree analysis* is the method to trace back the ancestral characters by taking one or more characters from an individual. In the Pedigree tree, Squares represents males and Circles females. Shaded squares or circles indicate dominant traits and open squares or circles denote recessive traits.
5. Hofmeister first reported Chromosomes in 1849. Sutton and Boveri in 1902 independently proposed the Chromosome theory of Heredity. Chromosomes may be Metacentric (centromere in the middle), Submetacentric (centromere towards one end), Acrocentric (centromere near the end) and Telocentric (centromere at the tip). *Giant chromosomes* are found in Dipteran insects. Polytene chromosome (Salivary gland of *Drosophila*) and Lamp brush chromosome (Amphibian Oocytes) are giant chromosomes. *SAT chromosome* possesses a swollen part (Satellite body) at the tip of the chromosome.
6. Somatic genes in the Y chromosomes are called Y linked or *Holandric genes*. Eg. Gene producing Hypertrichosis (hair in the ear pinna) in man. *Sex limited characters* express only in one sex. It will not express in the other sex even if the gene is present. Eg. Feather pattern in Poultry, premature baldness in human males, milk production in cattles. *Sex influenced characters* behave like Dominant in one sex and Recessive in the other sex. Phenotype will be different even though the genotype is same. Eg. Horn in Sheep, baldness in man, Hare lip etc.
7. *Mutation* is a sudden heritable change in organisms leading to variations. Mutation was observed by Hugo de Vries in *Oenothera lamarckiana* (Evening Prim rose). *Gene mutations* (Point mutations) affect the genes. It may be Deletion (removal of bases), Addition or Insertion and Substitution (replacement of one base by another one). Substitution may be Transition (Purine-Purine or Pyrimidine-Pyrimidine change) or Transversion (Purine-Pyrimidine change)

or vice versa). Transition is caused by Tautomerization (shifting of protons), Deamination (removal of amino group) or through base analogues (5-Bromo Uracil).

8. Gene mutations change the arrangement of genetic code in the DNA. So these are called **Frame shift mutations**. **Mis-sense mutation** changes the amino acid sequence in the protein. **Non sense mutations** results in termination codons. **Silent mutations** do not alter the amino acid sequence in the protein. **Cladogenic agents** (X-ray, Gamma rays) are chromosome breaking agents. **Cri-du-chat syndrome** is caused due to a deletion in the short arm of 5th chromosome. **Philadelphia chromosome** is the small 22nd chromosome in man produced due to the translocation between the chromosomes 9 and 22. It is seen in patients with **Chronic myeloid leukemia (CML)**.
9. **Colchicine** is called as Mitotic Poison since it destroys spindle fibers and arrest mitosis. It is used to induce Polyploidy in plants. **Triticale** is the first man made Cereal produced by crossing Wheat and Rye and then inducing polyploidy.
10. **Mutagens** are agents causing mutations. Physical mutagens are X-ray, alpha, beta, gamma rays, UV rays etc. Chemical mutagens include Nitrous acid, Ethyl methane sulphonate (EMS), Acridine orange etc. **Sharbati Sonora** is a variety of Wheat produced by induced mutation.

Molecular genetics

11. **Cistron** is the smallest unit of DNA that transcribes a messenger RNA. **Recon** is the unit of recombination and **Muton** is the part of DNA that undergoes mutation.
12. **B-DNA** is the usual form of right handed DNA with 10 base pairs in one turn. A-DNA (11 base pairs), C-DNA (9 base pairs) and D-DNA (8 base pairs) are forms of DNA produced in experimental conditions. **Z-DNA** is the left handed DNA discovered by Rich, Nordheim and Wang in 1984. It has 12 base pairs per turn. **Palindrome DNA** has inverted repetition of base sequences. **H-DNA** contains inverted repetition of base sequences to form Triple helix DNA.
13. Watson, Crick, Wilkins and Rosalind Franklin are associated with the Double Helical model of DNA. There are 10 base pairs per turn in the DNA. The base pairs are separated by a distance of 3.4 Angstrom so the total length of the turn is 34 Angstroms. **Hydrogen bonds** in the DNA stabilize the topology and antiparallel nature of DNA. **Phospho diester bonds** maintain the polarity (3'-5' and 5'-3') of the DNA strands.
14. In **Semi-conservative** replication of DNA (proposed by Matthew, Mehelson and Stahl), two daughter DNA are formed each having a new and old strands. In **Conservative replication**, the parent helix remains intact. In **Dispersive replication**, all the four strands of two daughter DNA are mixture of parental and daughter DNA. **Rolling circle model** of DNA replication is proposed for the replication of Circular DNA of bacteria.
15. **Helicase** is the enzyme that separate DNA strands during replication. **Topoisomerase** (Relaxing enzyme) reduces tension in the DNA during unwinding. DNA Polymerases I, II and III are DNA synthesizing enzymes. DNA polymerase III is the most active enzyme. Bacterial DNA Polymerases are Pol I and Pol II (DNA repair) and Pol III (Chromosome replication). **Ligase** (Molecular glue) joins DNA segments after synthesis. **Telomerase** is the enzyme containing RNA and protein. It terminates DNA replication. **Reverse Transcriptase** is the bacterial Polymerase that synthesise DNA from RNA. **Primase** (RNA polymerase) is used to synthesize Primer RNA during replication.

16. DNA replication occurs in the 5'-3' direction because the DNA polymerase III can act only in this direction. "**Leading strand**" synthesis occurs in the 5'-3' direction. "**Lagging strand**" synthesis (Discontinuous synthesis) occurs as DNA segments called "**Okasaki Fragments**". This is because the second strand of DNA has 3'-5' direction and Polymerase III can act only in the 5'-3' direction.
17. Beadle and Tatum conducted experiments in *Neurospora crassa* (Pink bread mold) to show the relationship between genes and enzymes. Wild type Neurospora (Prototroph) grow in Minimal medium containing few salts and minerals. Mutant forms (Auxotroph) grow only when amino acids like Ornithine, Citrulline and Arginine are added to the culture. Beadle and Tatum conducted tetrad analysis in Neurospora and proposed the "One Gene One Enzyme" hypothesis.
18. **RNA Splicing** involves the removal of Introns (non coding sequences) from the m-RNA. Then the Exons (coding sequences) are joined together. **RNA Editing** is the modification of the m-RNA before translation. **SnRNPs or Spurps** are ribonucleoproteins binds to the m-RNA during RNA splicing. **Spliceosomes** are formed by the interaction of SnRNPs with other proteins. **RNA capping** is the process by which a guanine nucleotide (with methyl group) is added to the 5' end of RNA after splicing. RNA cap determines the site of translation. **PolyA tailing** is the process by which a long tail of Adenine residue is added to the 3' end of m-RNA during splicing. **Ribozymes** are RNA molecules act as enzymes. RNase P is a Ribozyme.
19. **Recombinant DNA technology** involves manipulation of DNA which involves DNA cloning, DNA profiling etc. **R-DNA** or Recombinant DNA contains a bacterial plasmid and a desired gene sequence of another organism. **Restriction enzymes** are "Molecular Scissors" that cut double stranded DNA at specific sites producing 'Staggered ends'. These enzymes are present in bacteria to destroy foreign DNA such as Viral DNA. The enzyme will not destroy the own DNA of bacteria because it is kept in the methylated form. Most widely used restriction enzymes are Eco R1, Bam 1, Hind III etc.