Genetics & Evolution

Inheritance and Variation

Mendel's Law of Inheritance

(1) **Mendelism** means experiments performed by Mendel on genetics.

(2) Mendel's experiment involved 4 steps as selection, hybridization, selfing and calculations. His results led to the formation of laws of genetics later.

(3) Mendel performed monohybrid and dihybrid crosses and gave three principles of inheritance.

(4) Mendel's three principles of inheritance are:

(i) Law of dominance

(ii) Law of segregation or law of purity of gametes

(iii) Law of independent assortment

(4) **Law of Dominance –** The dominant characters are expressed when factors are in heterozygous condition.

(5) The recessive characters are only expressed in homozygous conditions. The characters never blend in heterozygous condition. A recessive character that was not expressed in heterozygous condition may be expressed again when it becomes homozygous.

(6) **Law/Principle of segregation** states that when a pair of contrasting factor or gene is brought together in a hybrid, these factors do not blend or mix up but simply associate themselves and remain together and separate at the time of gamete formation.

(7) **Principle of independent assortment** states that genes of different characters located in different pairs of chromosomes are independent of one another in this segregation during gamete formation.

(8) **Test Cross:** A cross between F1 hybrid (Aa) and its homozygous recessive parent (aa) is called Test Cross. This cross is called test cross because it helps to find out whether the given dominant phenotype is homozygous or heterozygous.

(9) **Monohybrid cross –** When we consider the inheritance of one character at a time in a cross, this is called monohybrid cross.

(10) **Dihybrid Cross –** A cross made to study the inheritance of two pairs of contrasting traits.

Exceptions of Conclusions of Mendel

Incomplete Dominance

(1) When neither of the alleles of a character is completely dominant over the other and the F1 hybrid is intermediate between the two parents, the phenomenon is called incomplete dominance.

(2) Incomplete dominance was first discovered by Correns in Mirabilis jalapa. The plant is called as 4'O clock plant or 'Gul-e-Bans'. Homozygous red (RR) flowered variety of the plant was crossed with white (rr) flowered variety. F1 offspring had pink flowers (Rr). This is called incomplete dominance.

(3) Incomplete dominance is also known to occur in snapdragon. The phenotypic ratio and genotypic ratio in F2 generation in case of incomplete dominance is 1:2:1.

Co-dominance

(1) In co-dominance both the gene expressed for a particular character in F1 hybrid progeny. There is no blending of characters, whereas both the characters are expressed equally.

(2) Co-dominance is seen in animals for coat colour. When a black parent is crossed with white parent, a roan color in F1 progeny is produced.

Sex determination

(1) Fixing the sex of an individual as it begins life is called sex determination. The various genetically controlled sex-determination mechanisms have been classified into following categories

(2) Chromosomal theory of sex determination: The X-chromosome was first observed by German biologist, Henking in 1891 during the spermatogenesis in male bug and was described as X-body. The chromosome theory of sex determination was worked out by E.B. Wilson and Stevens (1902-1905).

(3) They named the X and Y chromosomes as sex-chromosomes or allosomes and other chromosomes of the cell as autosomes.

(4) Sex chromosomes carry genes for sex. X-chromosomes carries female determining genes and Y-chromosomes has male determining genes.

(5) The number of X and Y chromosomes determines the female or male sex of the individual, Autosomes carry genes for the somatic characters. These do not have any relation with the sex.

Sex Determination by chromosomes:

Those chromosomes which are involved in the determination of sex of an individual are called sex chromosomes while the other chromosomes are called autosomes.

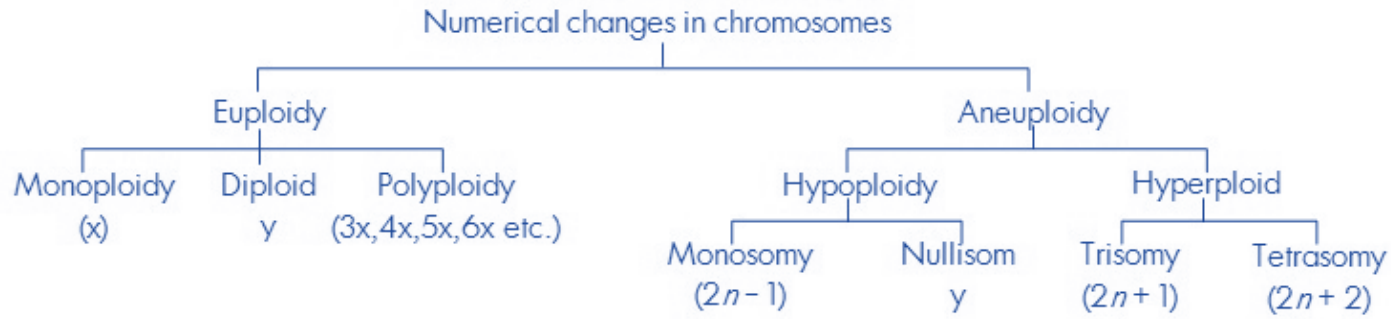
1) **XX – XY type:** In most insects including fruit fly Drosophila and mammals including human beings the females possess two homomorphic sex chromosomes, named XX. The males contain two heteromorphic sex chromosomes, i.e., XY. Hence the males produce two types of gametes / sperms, either with X-chromosome or with Y-chromosome, so they are called Heterogamety.

2) **ZZ – ZW type:** In birds and some reptiles, the males are represented as ZZ (homogamety) and females are ZW (heterogamety).

3) **XX – XO type:** In round worms and some insects, the females have two sex chromosomes, XX, while the males have only one sex chromosomes X. There is no second sex chromosome. Therefore, the males are designated as XO. The females are homogametic because they produce only one type of eggs. The males are heterogametic with half the male gametes carrying X-chromosome while the other half being devoid of it.

Numerical aberrations of chromosomes:

Each species has a characteristic number of chromosome. Variations or numerical changes in chromosomes (Heteroploidy) can be mainly of two types:



(1) **Turner's syndrome:** Such persons are monosomic for sex chromosomes *i.e.* possess only one X and no Y chromosome (XO). In other words they have chromosome number $2n - 1 = 45$. They are phenotypic females but are sterile because they have under developed reproductive organs. They are dwarf about 4 feet 10 inches and are flat chested with wide spread nipples of mammary glands which never enlarge like those in normal woman. They develop as normal female in childhood but at adolescence their ovaries remain under developed. They lack female hormone estrogen. About one out of every 5,000 female births results in Turner's syndrome.

(2) **Klinefelter's syndrome:** Since 1942, this abnormality of sex is known to geneticists and physicians. It occurs due to Trisomy of sex chromosomes which results in (XXY) sex chromosomes. Total chromosomes in such persons are $2n + 1 = 47$ in place of 46. Klinefelter (1942) found that testes in such male remain under developed in adulthood. They develop secondary sex characters of female like large breasts and loss of facial hair. Characters of male develop due to Y chromosome and those like female due to XX chromosomes. About one male child out of every 5,000 born, develops Klinefelter's syndrome.

Molecular Basis of Inheritance

DNA

(1) DNA is a long polymer of deoxyribonucleotides.
(2) The length of the DNA depends on the number of nucleotide pairs present in it.
(3) Bacteriophage lambda has 48,502 base pairs.

Central dogma of molecular biology

(1) Crick proposed the Central dogma in molecular biology
(2) It states that the genetic information flows from DNA à RNA à Protein.
(3) In some viruses like retroviruses, the flow of information is in reverse direction, which is from RNA à DNA à mRNA à Protein.

Structure of polynucleotide chain:

(1) A nucleotide has three components-
    (a) A nitrogen base
    (b) A pentose sugar (ribose in RNA and deoxyribose in DNA)
    (c) A phosphoric acid.
(2) There are two types of nitrogen bases:
    (a) Purines (Adenine and Guanine)
    (b) Pyrimidines (Cytosine, Uracil and Thymine)
(3) Adenine, Guanine and Cytosine are common in RNA and DNA.
(4) Uracil is present in RNA and in DNA in place of Uracil, Thymine is present.
(5) In RNA, Pentose sugar is ribose and in DNA, it is Deoxyribose.
(6) Based on the nature of pentose sugar, two types of nucleosides are formed - ribonucleoside and deoxyribonucleotides.
(7) Two nucleotides are joined by 3'-5' Phosphodiester linkage to form dinucleotide.
(8) More than two nucleotides join to form polynucleotide chain.
(9) The two strands of DNA (called DNA duplex) are antiparallel and complementary, i.i., one in 5'->3' direction and the other in 3"->5" direction.

History of DNA

(1) DNA is an acidic substance in the nucleus.
(2) It was first identified by Friedrich Meischer in 1869. He named it as 'Nuclein"
(3) In 1953 double helix structure of DNA was given by James Watson and Francis Crick, based on X-ray diffraction data produced Maurice Wilkins and Rosalind Franklin.

Packaging of DNA Helix

(1) The basic unit into which DNA is packed in the chromatin of eukaryotes.
(2) Nucleosome is the basic repeating structural (and functional) unit of chromatin, which contains nine histone proteins.
(3) Distance between two conjugative base pairs is 0.34nm
(4) The length of the DNA in a typical mammalian cell will be 6.6 X109 bp X 0.34 X10-9 /bp, it comes about 2.2 meters.
(5) The length of DNA is more than the dimension of a typical nucleus (10-6m)

DNA Replication

(1) DNA is the only molecule capable of self duplication so it is termed as a living molecule.
(2) All living beings have the capacity to reproduce because of DNA.
(3) DNA replication takes place in S-phase of the cell cycle. At the time of cell division, it divides in equal parts in the daughter cells.
(4) **Delbruck** suggested three methods of DNA replication i.e.
    (i) Dispersive
    (ii) Conservative
    (iii) Semi-conservative
(5) The process of DNA replication takes a few minutes in prokaryotes and a few hours in eukaryotes.

RNA

(1) RNA is the first genetic material.
(2) RNA is a non hereditary nucleic acid except in some viruses (retroviruses).
(3) RNA used to act as a genetic material as well as catalyst.
(4) It is a polymer of ribonucleotide and is made up of pentose ribose sugar, phosphoric acid and nitrogenous base (A,U,G,C).
(5) RNA may be of two types – genetic and non-genetic.

Genetic Code

(1) Term genetic code was given by **George Gamow (1954).** He was the first to propose the triplet code (one codon consists of three nitrogen bases).
(2) The relationship between the sequence of amino acids in a polypeptide chain and nucleotide sequence of DNA or mRNA is called genetic code.
(3) There occur 20 types of amino acids which participate in protein synthesis. DNA contains information for the synthesis of any types of polypeptide chain. In the process of transcription, information transfers from DNA to m-RNA in the form of complementary N_2-base sequence.
(4) A **codon** is the nucleotide sequence in m-RNA which codes for particular amino acid; whereas the **genetic code**is the sequence of nucleotides in **m-RNA** molecule, which contains information for the synthesis of polypeptide chain.

(5) 61 out of 64 codons code for only 20 amino acids.

(6) The main problem of genetic code was to determine the exact number of nucleotide in a codon which codes for one amino acid.

Characteristics of genetic code

(1) **Triplet in nature**

(a) A codon is composed of three adjacent nitrogen bases which specify one amino acid in polypeptide chain.

(b) For example- In m-RNA if there are total 90 N_2– bases. Then this m-RNA determines 30 amino acids in polypeptide chain.

(2) **Univerality**

(a) The genetic code is applicable universally.

(b) The same genetic code is present in all kinds of living organism including viruses, bacteria, unicellular and multicellular organisms. In all these organisms, triplet code for specific amino acid.

(3) **Non-ambiguous**

(a) Genetic code is non ambiguous i.e. one codon specifies only one amino acid and not any other.

(b) In this case one codon never code two different amino acids. **Exception** GUG codon which code both valine and methionine amino acid.

(4) **Non-overlapping**

(a) A nitrogen base is a constituent of only one codon.

(5) **Comma less**

(a) There is no punctuation (comma) between the adjacent codon i.e. each codon is immediately followed by the next codon.

(b) If a nucleotide is deleted or added, the whole genetic code read differently.

(c) A polynucleotide chain having 50 amino acids shall be specialized by a linear sequence of 150 nucleotides. If a nucleotide is added in the middle of this sequence, the first 25 amino acids of polypeptide will be same but next 25 amino acids will be different.

(6) **Degeneracy of genetic code**

(a) Only two amino acids – tryptophan and methionine are specified by single codon.
UGG for tryptophan, AUG for methionine

(b) All the other amino acids are specified or coded by 2 to 6 codons.

(c) Leucine, serine and arginine are coded or specified by 6-codons.

(d) Degeneracy of genetic code is related to third position (3'-end of triplet codon) of codon. The third base is described as 'Wobble base'.

Genomics and Human Genome project:

(1) The term genome has been introduced by **Winkler** in 1920 and the genomics is relatively new, coined by**Thomas Rodericks** in 1986.

(2) Genomics is the subdiscipline of genetics devoted to the mapping, sequencing and functional analysis of genomes. Genomics is subdivided into following types:

(a) **Structural genomics:** It is the study of genome structure deals with the complete nucleotide sequences of the organisms.

(b) **Functional genomics:** It is the study of genome function which includes transcriptome and proteome. Transcriptome is a complete set of RNAs transcribed from a genome while proteome is a complete set of proteins encoded by a genome and aims the determination of the structure and function of all the proteins in living organisms.

(3) The human genome project, sometimes called "biology's moon shot", was launched on october 1, 1990 for sequencing the entire human genome of 2.75 billion (2.75×10^9 or 2750000 bp or 2750000 kilobase pairs or 2750 megabase pairs) nucleotide pairs.

(4) Two important scientist associated with human genome are **Francis Collins**, director of the Human Genome Project and **J. Craig Venter**, founding president of Celera genomics.

(5) The complete sequencing of the first human chromosome, small chromosome 22, was published in December 1999.

Genome of Model organisms

S. No.	Organism	No. of base pair	No. of genes
(1)	Bacteriophage	10 thousand	–
(2)	E. coli	4.7 million	4000
(3)	Saccharomyces cerevisiae	12 million	6000
(4)	Caenorhabditis elegans	97 million	18,000
(5)	Drosophila melanogaster	180 million	13,000
(6)	Human	3 billion	30,000
(7)	Lily	106 billion	–

DNA finger printing

(1) **Alec Jeffreys et al** (1985) developed the procedure of genetic analysis and forensic medicine, called DNA finger printing.

(2) It is individual specific DNA identification which is made possible by the finding that no two people are likely to have the same number of copies of repetitive DNA sequences of the regions.

(3) It is also known as DNA profiling.

(4) The chromosomes of every human cell contain scattered through their DNA short, highly repeated 15 nucleotide segments called “mini-satellites” or variable-number Tandem Repeat (VNTR).

Technique for DNA fingerprinting

(1) Only a small amount of tissues like blood or semen or skin cells or the hair root follicle is needed for DNA fingerprinting.
(2) Typically DNA content of about 100,000 cells or about 1 microgram is sufficient.
(3) The procedure of DNA fingerprinting involves the following major steps:
    (i) DNA is isolated from the cells in a high-speed refrigerated centrifuge.
    (ii) If the sample of DNA is very small, DNA can be amplified by Polymerase Chain Reaction (PCR).
    (iii) DNA is then cut up into fragments of different length using restriction enzymes.
    (iv) The fragments are separated according to size using gel electrophoresis through an agarose gel. The smaller fragments move faster down the gel than the larger ones.
    (v) Double stranded DNA is then split into single stranded DNA using alkaline chemicals.
    (vi) These separated DNA sequences are transferred to a nylon or nitrocellulose sheet placed over the gel. This is called ‘Southern Blotting’ (after **Edward Southern**, who first developed this method in 1975).
    (vii) The nylon sheet is then immersed in a bath and probes or makers that are radioactive synthetic DNA segments of known sequences are added. The probes target a specific nucleotide sequence which is complementary to VNTR sequences and hybridizes them.
    (viii) Finally, X-ray film is exposed to the nylon sheet containing radioactive probes. Dark bands develop at the probe sites which resemble the bar codes used by grocery store scanners to identify items.

Applications of DNA fingerprinting

This technique is now used to:

(i) Identify criminals in forensic laboratories.
(ii) Settle paternity disputes.
(iii) Verify whether a hopeful immigrant is, as he or she claims, really a close relative of already an established resident.
(iv) Identify racial groups to rewrite biological evolution.

Evolution

(i) Ancient theories of origin of life:

(a) Theory of special creation.

(b) Theory of spontaneous generation or Abiogenesis.

(c) Biogenesis

(d) Cosmozoic theory

(e) Theory of sudden creation from inorganic material.

(f) Naturalistic theory

(ii) **Oparin's Modern Theory:**

(a) Oparin (1924) proposed that “life could have originated from non-living organic molecules.”

(b) He believed in Biochemical origin of life. Haldane (1929) also stated similar views. Oparin greatly expanded his ideas and presented them as a book “The origin of life” in 1936.

(c) According to this theory, the Earth originated about 4,500 million years ago. When the earth was cooling down, it had a reduced atmosphere. In this primitive atmosphere nitrogen, hydrogen, ammonia, methane, carbon mono-oxide and water were present. Energy was available in the form of electric discharges by lightening and ultraviolet rays. As soon as the earth crust was formed, it was very much folded. Torrential rains poured over the earth for centuries and were deposited in deep places.

(d) **Miller's Experiment:** An American scientist (Biologist) Stanley Miller (1953) performed an experiment under support Oparin's theory of origin of life. He believed that basic compounds which are essential for life can be synthesised in the laboratory by creation in the laboratory, on a small scale, the conditions which must have existed at the time of origin of life on earth.

(e) Miller took a flask and filled it with methane, ammonia and hydrogen in proportion of 2:1:2 respectively at 0°C. This proportion of gases probably existed in the environment at time of origin of life. This flask was connected with a smaller flask, that was filled with water, with the help of glass tubes. In the bigger flask, two electrodes of tungsten were fitted. Then a current of 60,000 volts was passes, through gases containing bigger flask for seven days. At the end of seven days, when the vapours condensed, a red substance was found in the U-tube. When this red substance was analyzed, it was found to contain amino acids, Glycine and nitrogenous bases which are found in the nucleus of a cell.

(f) The entire process of the origin of life, as proposed by Oparin, can be summarised as under –

(i) **The Chemical Evolution:**

(1) Step 1: Formation of simple molecules

(2) Step 2: Formation of Simple organic compounds

(3) Step 3: Formation of complex organic compounds

(4) Step 4: Formation of nucleic acids and nucleoproteins

(ii) **Organic Evolution:**

(1) Step 5: Formation of Coacervates

(2) Step 6: Formation of Primitive cell

(3) Step 7: Origin of autotrophism

(4) Step 8: Origin of Eukaryotic cells

Evidences of Organic Evolution

The following are the evidences in favour of Organic Evolution:

(i) Evidences from Classification

(ii) Evidences from Comparative Anatomy

(a) Analogy and Homology

(b) Vestigeal organs

(iii) Evidences from Physiology

(iv) Evidences from Serology

(v) Evidences from Embryology

(vi) Evidences from Palaeontology

(vii) Evidences from geographic distribution

(viii) Evidences from Genetics

(i) **Evidences from Classification:** All the known living animals and plants have been classified into various species, genera, families, order, classes, phyla and kingdoms. The classification of a particular animal is attempted only after its extensive study.

(ii) **Evidences from Comparative Anatomy:** In all the living animals, the basic substance of life is Protoplasm. If the species had been created separately, then there should be no relationship in the various organs and systems of animals. But on the contrary, we see that large number of animals although unlike in appearance show most of the systems and organs made on the same plan. The resemblance is very close in the members of the same group.

(iii) **Evidences from Physiology**: Various types of chemical tests exhibit many basic similarities in physiological and chemical properties that show a physiological relationship among animals.

(iv) **Evidences from Serology:** This is a method by which the reactions of blood serum are observed. From the blood are also extracted the crystals of Oxyhaemoglobin. The structure differs in different vertebrates, but in a definite order. The reaction is nearly identical in man and anthropoid monkeys, but slightly less identical with other mammals.

(v) **Evidences from Embryology:** With the exception of a few, every multi-cellular animal originates from a zygote. The development from zygote to adult shows many similarities in various organisms. The development is termed as ontogeny

(vi) **Evidences from Palaeontology:** The study of fossils and their interpretation forms one of the great evidences of evolution. An Italian scientist, Leonardo da Vinci, was the first person to recognize their importance and said they were either remains of organisms of their impressions on some sort of clay or rock.

Important living fossils

1. Peripatus (Arthropoda)	**2. Limulus (Arthropoda)**
3. Nautilus (Mollusca)	4. Neopilina (Mollusca)
5. Lingula (Brachiopoda)	6. Latimeria (Coelacanth fish)
7. Sphenodon (Reptilia)	8. Didelphis (Opossum)

(vii) **Evidences from geographic distribution:** If the study of horizontal distribution of animals on the face of this earth is made, it would be seen that animals are not evenly distributed. Two identical places with the same climate and vegetation may not have same sort of animal fauna

(viii) **Evidences from Genetics:** Johan Gregor Mendel in 1866 published his work on experimental breeing. He bred two individuals differing in certain well-defined characters, and observed the ratio in which various contrasting parental characters appeared in successive generations.

(c) **Connecting links:** Intermediate or intergrading forms between two groups of organisms:

Organism	Connecting link between
1. Viruses	Living and nonliving
2. Euglena (Protozoa)	Plants and animals
3. Proterospongia (Protozoa)	Protozoa and Porifera
4. Peripatus (Arthropoda)	Annelida and Arthropoda
5. Neopilina (Mollusca)	Annelida and Mollusca
6. Balanoglossus (Chordata)	Nonchordata and Chordata
7. Dipnoi (Lungfish)	Pisces and Amphibia
8. Archaeopteryx (Aves)	Reptiles and Birds
9. Prototheria (Mammalia)	Reptiles and Mammals

Theories of organic evolution

(i) **Lamarckism:** Lamarck (1744 –1829) was one of the most brilliant stars on the horizon of the history of evolution. He was the first naturalist to put forward a general theory of evolution in his famous book. Philosophic Zoologique published in 1809. His evolutionary theory may be summarised in the form of following laws:

(a) The internal forces of life tend to increase the size of an organism.

(b) The necessity in animals to produce new structures.

(c) The effect of use and disuse.

(d) Inheritance of acquired characters.

(iii) **Darwinism:** Charles Robert Darwin was undoubtedly the first naturalist who put the idea of organic evolution on sound footing. His statements and theories were based upon practical experiences and large number of proofs which he collected directly from the nature.

His main ideas about the evolution are given below –

(a) Over – production of offspring

(b) Limited supply of food and shelter

(c) Struggle for existence:

(i) Intra –specific

(ii) Inter –specific

(iii) Environment

(d) Survival of the fittest

(e) Universal occurrence of variations

(f) Inheritance

(g) Natural selection

Difference between Darwinism and Neo–Darwinism

Darwinism (Natural Selection)	Neo–Darwinism
(1) It is the original theory given by Charles Darwin (1859) to explain the origin of new species.	(1) Neo–Darwin is a modification of the original theory of Darwin to remove its short–comings.
(2) According to this theory accumulation of continuous variations causes changes in individuals to form new species.	(2) Instead of continuous variations, mutations are believed to help form new species.
(3) It believes in the selection of individuals on the basis of accumulation of variation.	(3) Variations accumulate in the gene pool and not in the individuals.
(4) Darwinism does not believe in isolation.	(4) Neo–Darwinism incorporates isolation as an essential component of evolution.
(5) It can explain the origin of new characters.	(5) The theory can explain the occurrence of unchanged forms over millions of years.
(6) Darwinism cannot explain the persistence of certain forms in the unchanged condition.	(6) Normally only those modifications are transferred to next generation which influence germ cells or where somatic cells give rise to germ cells.