CHAPTER

5 Genetics and Evolution

Study of heredity and variation is called **genetics**.

- Term genetics was given by - **Bateson**.
- Father of genetics - **Gregor Johann Mendel**.
- Father of experimental genetics - **Thomas Hunt Morgan**.
- Father of human genetics - **Archibald Garrod**.

Some Terms in Genetics

Gene : It is segment of DNA. It is basic unit of heredity.

Back cross : It is cross which is performed between hybrid and one of its parents.

Test cross : Test cross is crossing of offspring with unknown dominant phenotype with the individual homozygous recessive for the trait.

Monohybrid cross : It is a cross between two organisms of a species which is made to study the inheritance of a single pair of alleles or factors of a character.

Monohybrid ratio : Monohybrid ratio is usually 3 : 1 (phenotypic ratio) or 1 : 2 : 1 (genotype ratio) in which 25% of the individuals carry the recessive trait, 25% pure dominant and 50% have hybrid dominant trait.

Dihybrid cross : It is a cross between two organisms of a species which is made to study the inheritance of two pairs of factors or alleles of two genes.

Dihybrid ratio : Dihybrid ratio is 9 : 3 : 3 : 1 (phenotypic ratio) where 9/16 first recessive and second dominant and 1/16 carry both the recessive traits.

- Mendel conducted cross hybridization experiments on Garden Pea plant (*Pisum sativum*). The first was the **Principle of segregation**, which claimed that each trait was specified by paired hereditary determinants (alleles of genes) that separate from each other during gamete formation. This law is also called **Law of purity of gametes or Law of splitting of hybrids.**
- **Gregor Mendel** was the first individual to apply a modern scientific approach to the study of heredity. Mendel proposed two basic principles of trasmission genetics.
- Mendel's second basic conclusion was the **Principle of independent assortment**, which stated that the segregation of one pair of genes-controlling a given trait - was not influenced by the segregation of other gene pairs. The chormosome theory provided a physical basis for the principle of independent assortment. Genes located on different chromosomes move to gametes independently of each other during meiosis.

Human Blood Groups and Multiple Allele

- The system of blood groups in humans was discovered by Karl Landsteiner in 1900 s.
- There are four phenotypes of Blood namely **A, B, AB** and **O** produced by three different alleles $\mathbf{I^A}$, $\mathbf{I^B}$ **and i** of a gene.
- The allele $\mathbf{I^A}$ and $\mathbf{I^B}$ are equally dominant and do not interfere with expression of each other hence the allele $\mathbf{I^A\ I^B}$ are said to be co-dominant because both are expressed in the phenotype **AB.**
- **Linkage** is the phenomenon of certain genes staying together during inheritance through generations without any change or separation due to their being present on the same chromosomes.
- Linkage in the genes can be identified by test cross.
- The rearrangements of linked genes due to crossing over is known as **recombination**. Recombination also occurs due to chance separation of chromosomes during gametogenesis and their random coming together during fertilization.

Sex Determination

- **Henking** discovered X body in spermatogenesis of few insects and it was given name of X chromosome. Due to involvment of X and Y chromosomes in determination of sex, they were called **sex chromosomes.**
- Rest of the chromosomes which determine other metabolic character of the body are called **autosomes**.

Mutation

- Phenomenon that results in alteration of DNA sequence and consequently results in change in genotype and phenotype of an organism is called **mutation**.
- **Mutagens** are various chemical and physical factors that induce mutations, *e.g.*, UV radiations, carcinogenic chemicals like nicotine, nitric oxide (NO).

Genetic Disorder

- A genetic disorder is a disease that is caused by an abnormality in an individual's DNA.

Haemophilia

- A protein involved in clotting of blood is affected in an affected individual; if person gets a cut, will result in non-stop bleeding.
- Females are heterozygous and carriers of haemophilia.

Sickle Cell Anaemia

- It is due to inheritance of defective allele coding for β-globin. It results in the transformation of Hb^A into Hb^S in which glutamic acid is replaced by valine at 6th position in each of two β-chains of haemoglobin.
- It is an excellent example of single mutation.

Phenylketonuria

- Affected individual lacks enzyme phenylalanine hydroxylase that converts amino acid phenylalanine to tyrosine.
- It is characterized by severe mental retardation, hypopigmentation of skin & hair, eczema, etc.

Chromosomal Mutation

Name of disorder or syndrome	Chromosomal Alteration involved	Symptoms or Associated Traits
Down's Syndrome	Trisomy 21	Characteristic facial features, short stature, heart defects, respiratory infections, mental retardation.
Klinefelter's Syndrome	XXX (or XXYY, XXXY, etc)	Sterility, small testes, feminine body contours, normal intelligence or mental retardation.
Turner's Syndrome	Monosomy X (XO)	Short stature, sex organs do not mature, no secondary sex characteristics, normal intelligence.
Cri du chat Syndrome	Deletion in Chromosome 5	Mental retardation, small head, unusual cry.

Deoxyribonucleic Acid

- DNA is a long chain polymer of deoxyribonucleotides.
- Nucleotide is made up of 3 chemical groups →

$$\underbrace{\text{A nitrogen base moiety + pentose sugar}}_{\text{Nucleoside}}$$

+ phosphate group

- Nitrogenous base are of two types – **purines** (9 membered double rings with nitrogen at 1, 3, 7 & 9^{th} positions) and **pyrimidines** (6 membered rings with nitrogen at 1 and 3^{rd} position).
- Purines are of **two types** – adenine (A) and guanine (G) and pyrimidines are of **three types** – thymine (T), cytosine (C) and uracil (U).
- A characteristics that differentiate DNA from RNA is that DNA contains all of the nitrogen bases except uracil and RNA contains all of the nitrogen bases except thymine.
- **Wilkins-Franklin** carried out X ray diffraction (X-ray crystallography) on the basis of which **Watson and Crick** suggested secondary structure of DNA in 1953.
- 2 DNA strands are organized in antiparallel and complementary arrangement [*i.e.*, 2 strands run in opposite orientation (one in 5' – 3' and allies in 3' – 5')].
- Adenine pairs with thymine with 2 hydrogen bonds and guanine pairs with cytosine with three hydrogen bonds.
- The helix is generally right handed *i.e.* its turn run clockwise looking along the helix axis. The pitch of helix is **3.4 nm** (1 nm = 10^{-9}m) and there are **10bp** in each turn.
- The concept of central dogma in molecular biology was proposed by **Francis Crick** (1958). It proposes unidirectional or one way flow of information from DNA to RNA & then to protein.

$$\text{DNA} \xrightarrow{\text{Transcription}} \text{mRNA} \xrightarrow{\text{Translation}} \text{Polypeptide (protein)}$$

Evolution

- It is the formation of newer types of organisms from the pre-existing ones through modification. Evolution is therefore, often called descent with modification.
- Earth originated 4600 million years ago.
- Life appeared 3.7 billion years ago. This is indicated by the discovery of microfossils of cyanobacteria like organisms.
- **Theory of spontaneous generation (Abiogenesis/ autogenesis)**
  - Given by **Aristotle**.
  - Life originated from non-living things in spontaneous manner.
- **Theory of Biogenesis**
  - Life originated from pre-existing life.
  - Given by **Francesco Redi**.
  - **Oparin – Haldane** theory was supported experimentally by **Miller-Urey** experiment in 1953.

    Discharge Apparatus – a large flask containing mixture of CH_4, NH_3, H_2 and H_2O with electric source and boiling of water for a week.
  - Miller observed dark condensed liquid which was analysed. Analysis reports concluded, that it was a mixture of amino acids like alanine, glycine, glutamic acid, aspartic acid, valine and leucine and number of other organic compounds like HCN, aldehyde and cyanocompounds.

Theories of Evolution

Darwinism

- Darwinism is the term coined for the explanation offered by **Darwin** for the origin of species by **Natural selection** in 1858.
- Darwinism or theory of natural selection is a theory of organic evolution which states that new species evolve over a long period of time through accumulation of small variations which provide the organism with structural and functional superiority over others in their survival and differential reproduction.

Genetic Equilibrium (Hardy Weinberg Law)

- Law states that "both gene frequency and genotype frequency" will remain constant from generation to generation in an infinitely large interbreeding population in which mating is random and no selection, migration or mutation occurs.
- **Hardy Weinberg formula or binomial expression is given as**
  $p^2 + 2pq + q^2 = 1$ for two alleles A and a.
- **Genetic drift** refers to chance elimination of gene(s) of certain traits when a section of population migrates or dies of natural calamity. It eliminates certain alleles and fixes other alleles.
- Genetic drift in a new colony is called **founder effect**.
- Natural calamity like earthquake greatly reduces the size of population, killing the individuals randomly. The genetic pool of surviving population decreases. This condition of reduced genetic variability is called **bottleneck effect**.

Vestigial Organs

- The useless and functionless degenerate structures which were large and functional in some other animals.
- Examples : Vermiform appendix, coccyx, pinna muscle, wisdom tooth in humans, rudiments of hindlimbs in python.

Adaptive radiation

- It is a special evolutionary pattern, characterized by a rapid increase in number of kinds of closely related species.
- A **good example of adaptive radiation** is found among the **Finches of Galapagos Islands**. Another example is **Australian Marsupials**.

Biological evolution

- It is the process of change over time, in the heritable characteristics, or traits of a population organisms.
- Many experts believe that *Australopithecus garhi* or a similar species gave rise to the genus *Homo*.
- Early hominids–members of the genus *Homo*–lived contemporaneously with *Australopithecines* for perhaps a half million years.
- The oldest fossil remains of a member of the genus *Homo* were discovered in Tanzania. It was named *H. habilis*.
- *Homo erectus* is the only other known extinct species of the genus *Homo*.
- *H. erectus* was replaced in tropical regions by *Homo sapiens* about 200,000 years ago.

EXERCISE

1. Mutation rates are affected by
   (a) temperature
   (b) X-rays
   (c) gamma and beta radiation
   (d) All of the above
2. A dihybrid condition is
   (a) tt Rr (b) Tt rr
   (c) tt rr (d) Tt Rr
3. In case of incomplete dominance the monohybrid ratio of phenotypes in F_2 generation is
   (a) 1 : 2 : 1 (b) 3 : 1 : 1
   (c) 9 : 3 : 3 : 1 (d) 2 : 3 : 1
4. The number of autosomes in human female ovum is
   (a) 11 (b) 12
   (c) 22 (d) 23
5. Which one of the following represents a test cross ?
   (a) Ww × WW (b) Ww × Ww
   (c) Ww × ww (d) WW × WW
6. How many different kinds of gametes will be produced by a plant having the genotype AABbCC?
   (a) Nine (b) Two
   (c) Three (d) Four
7. How many pairs of contrasting characters in pea pod were chosen by Mendel?
   (a) 2 (b) 3
   (c) 4 (d) 7
8. From a cross AABb x aaBb, the genotypes AaBB : AaBb : Aabb : aabb will be obtained in the following ratio
   (a) 1: 1 : 1 : 1 (b) 1: 2 : 1 : 0
   (c) 0: 3 : 1 : 0 (d) 1: 1 : 1 : 0
9. A nucleotide is formed of
   (a) purine, pyrimidine and phosphate
   (b) purine, sugar and phosphate
   (c) nitrogenous bases, sugar and phosphate
   (d) pyrimidine, sugar and phosphate
10. The process of translation is
    (a) ribosome synthesis
    (b) protein synthesis
    (c) DNA synthesis
    (d) RNA synthesis

11. Which one of the following pairs of nitrogenous bases of nucleic acids, is wrongly matched with the category mentioned against it?
    (a) Thymine, Uracil - Pyrimidines
    (b) Uracil, Cytosine - Pyrimidines
    (c) Guanine, Adenine - Purines
    (d) Adenine, Thymine - Purines
12. Ligase helps in
    (a) removal of few genes
    (b) translation
    (c) inserting few genes in DNA
    (d) bringing transversion in chromosomes
13. A nucleoside differs from a nucleotide in not having
    (a) phosphate
    (b) sugar
    (c) nitrogen base
    (d) phosphate and sugar
14. Nucleotide arrangement in DNA can be seen by
    (a) X-ray crystallography
    (b) Electron microscope
    (c) Ultracentrifuge
    (d) Light microscope
15. Scientific name of man is
    (a) *Canis familiaris* (b) *Homo habilis*
    (c) *Homo erectus* (d) *Homo sapiens*
16. The primitive atmosphere of earth contained water vapours, hydrogen, ammonia and
    (a) CO_2 (b) O_2
    (c) N_2 (d) CH_4
17. The book 'Origin of Species' is written by
    (a) Aristotle (b) Darwin
    (c) Watson (d) Lamarck
18. Which was absent in the atmosphere at the time of origin of life ?
    (a) NH_3 (b) H_2
    (c) O_2 (d) CH_4
19. Homologous organs are
    (a) wings of insects and bat
    (b) gills of fish and lungs of rabbit
    (c) pectoral fins of fish and fore limbs of horse
    (d) wings of grosshopper and crow
20. The kind of evolution in which two species of different geneology come to resemble one another closely, is termed as
    (a) progressive evolution
    (b) convergent evolution
    (c) parallel evolution
    (d) retrogressive evolution
21. Which one is linked to evolution?
    (a) Extinction (b) Competition
    (c) Variation (d) Reproduction
22. Genetic variation in a population arises due to
    (a) Mutations only
    (b) Recombination only
    (c) Mutations as well as recombination
    (d) Reproductive isolation and selection
23. The first organisms were
    (a) Chemoautotrophs
    (b) Chemoheterotrophs
    (c) Autotrophs
    (d) Eukaryotes
24. What was the most significant trend in evolution of modern man (*Homo sapiens*) from his ancestors ?
    (a) Upright posture
    (b) Shortening of jaws
    (c) Binocular vision
    (d) lncreasing brain capacity
25. The factors which represent the contrasting pairs of characters are called
    (a) dominant and recessive
    (b) alleles
    (c) homologous pairs
    (d) determinants
26. Who proved that DNA is basic genetic material?
    (a) Griffith
    (b) Watson
    (c) Boveri and Sutton
    (d) Hershey and Chase
27. The two strands of DNA are held together by
    (a) peptide bonds (b) phosphodiester bonds
    (c) hydrogen bonds (d) S – S bonds
28. According to special creation theory the earth is about
    (a) 4000 yrs old (b) 4.5 M yrs old
    (c) 4.5 B yrs old (d) 10000 yrs old
29. Which of the following is not a part of Darwin's theory of evolution?
    (a) Genetic drift
    (b) Natural selection
    (c) Survival of the fittest
    (d) Struggle for existence
30. In Down's syndrome of a male child, the sex complement is
    (a) XO (b) XY
    (c) XX (d) XXY

ANSWER KEY											
1	(d)	**6**	(b)	**11**	(d)	**16**	(d)	**21**	(c)	**26**	(d)
2	(d)	**7**	(d)	**12**	(c)	**17**	(b)	**22**	(c)	**27**	(c)
3	(a)	**8**	(b)	**13**	(a)	**18**	(c)	**23**	(b)	**28**	(a)
4	(c)	**9**	(c)	**14**	(a)	**19**	(c)	**24**	(d)	**29**	(a)
5	(c)	**10**	(b)	**15**	(d)	**20**	(b)	**25**	(b)	**30**	(b)

HINTS AND SOLUTIONS

2. (d) Dihybrid condition involves simultaneous inheritance of two pairs of mendelian factors or genes.
4. (c) In human female ovum, number of autosome are 22 pairs.
7. (d) Mendel in his experiment considered total 7 characters (3 characters of seed i.e., seed shape, seed colour, cotyledon colour, 2 characters of pod i.e., plant height and position of pods on the stem).
8. (b) AABb × aaBb

AB	Ab	AB	Ab	
aB	AaBB	AaBb	AaBB	AaBb
ab	AaBb	Aabb	AaBb	Aabb
aB	AaBB	AaBb	AaBB	AaBb
ab	AaBb	Aabb	AaBb	Aabb

AaBB : AaBb: Aabb: aabb
4 : 8 : 4 : 0
AaBB = 1; AaBb = 2; Aabb = 1; aabb = 0

9. (c) Nucleotide is a unit of DNA, which is formed of nitrogenous bases (purines & pyrimidines), sugar (pentose) & phosphate.
10. (b) Protein synthesis occurs over ribosomes which are also referred to as protein factories.
11. (d) Purine is an organic nitrogenous base sparingly soluble in water, that gives rise to a group of biologically important derivatives, notably adenine and guanine, which occur in nucleotides and nucleic acids (DNA and RNA).
12. (c) Ligase helps in inserting few genes in DNA.
13. (a) In a nucleotide, purine or pyrimidine nitrogenous base is joined by deoxyribose pentose sugar (D), which is further linked with phosphate (P) group to form nucleotides.
14. (a) In 1953 Wilkins obtained very fine X-ray crystallographic pictures of DNA from which Watson and Crick developed the double helix model of DNA.
16. (d) The primitive atmosphere of the earth contained : H_2O, H_2, NH_3 & CH_4. The ratio was (2 : 2 : 1) of : NH_3, CH_4 and hydrogen.
17. (b) 'Origin of Species' book was written by Charles Robert Darwin. During his world journey (1831–1936) as a naturalist in H.M.S. Beagle ship, he visitied many parts of the world. On the basis of the observations of this journey, he wrote the book thereby describing his theory of natural selection.
18. (c) Oxygen gas was not present due to reducing atmosphere, it only came to existence after the evolution of photosynthesis process.
19. (c) Organs that are similar in fundamental structure but different in functions are "homologous organs", Richard Owen, introduced the term homologous. Pectoral fins of fish and fore limbs of horse similar in structure but different in functions are homologous organs.
Rest of the organs compared in the question are analogous organs.
20. (b) Progressive evolution is development of organisms with more elaborate and specialized structures from those having less elaborate features e.g. amphibians from reptiles. Retrogressive or degenerative evolution is development of simpler forms from more complex ones. Such evolution has occurred in case of vestigial organs, parasitic forms, and in reduction of overspecialized structures such as wings in flightless birds. Parallel evolution is formation of similar traits in related groups of organisms independently due to similar requirement e.g. running of two toed deer and one toed horse. Evolution of wings in insects and birds serve as example of convergent evolution.
21. (c) Organic evolution is process of cumulative change of living populations and in the descendant populations of organisms. According to Darwin, variations are differences among individuals. Variations which help the individual to fit in its surroundings are passed on to the next generation while others disappear. Thus, variation plays an important role in evolution.
22. (c) Crossing over leads to recombination of genetic material on the two chromosomes. Mutation results in alternation of DNA sequences and consequently results in change in the genotype and the phenotype of an organism. In addition to recombination, mutation is another phenomenon that leads to variation in DNA.
23. (b) Chemoheterotrophs were first organisms. They were prokaryotic like bacteria, anaerobes, as molecular oxygen was absent. They obtained energy by fermentation of some of the organic molecules present in the broth. Thus they absorbed organic molecules from outside for body building and energy.
    - Chemoautotrophs - Organisms that are capable to synthesize organic molecules from inorganic molecules.
      E.g., Nitrifying bacteria, sulphur reducing bacteria etc.
    - Autotrophs are photosynthesizing plants / organisms.
24. (d) The most significant trend in evolution of modern man (*Homo sapiens*) from his ancestors is development of brain capacity.