SECTION-A (Multiple choice questions)

Q. 1-Answer

(i) **a** (ii) **b** **(iii) a** **(iv) c** **(v) a** (vi) **a** **(vii) c**

(viii) b **(ix) d** **(x) a**

SECTION –B (Descriptive type questions)

Q. 2- Answer:

Sex Determination in Humans: (XX/XY system)

The discovery that human females are XX and that human males are XY suggested that sex might be determined by the number of X chromosomes or by the presence or absence of a Y chromosome. As we now know, the second hypothesis is correct. In humans and other placental mammals, maleness is due to a dominant effect of the Y chromosome (Fig-1). The evidence for this fact comes from the study of individuals with an abnormal number of sex chromosomes. XO animals develop as females, and XXY animals develop as males. The dominant effect of the Y chromosome is manifested early in development, when it directs the primordial gonads to develop into testes. Once the testes have formed, they secrete testosterone, a hormone that stimulates the development of male secondary sexual characteristics.

Researchers have shown that the **testis-determining factor (TDF)** is the product of a gene called ***SRY*** (for ***s*ex-determining *r*egion *Y***), which is located just outside the pseudoautosomal region in the short arm of the Y chromosome. The discovery of *SRY* was made possible by the identification of unusual individuals whose sex was inconsistent with their chromosome constitution—XX males and XY females (Fig-2). Some of the XX males were found to carry a

small piece of the Y chromosome inserted into one of the X chromosomes. This piece evidently carried a gene responsible for maleness. Some of the XY females were found to carry an incomplete Y chromosome. The part of the Y chromosome that was missing corresponded to the piece that was present in the XX males; its absence in the XY females apparently prevented them from developing testes. These complementary lines of evidence showed that a particular segment of the Y chromosome was needed for male development. Molecular analyses subsequently identified the *SRY* gene in this male-determining segment. Additional research has shown that an *SRY* gene is present on the Y chromosome of the mouse, and that—like the human *SRY* gene—it triggers male development.

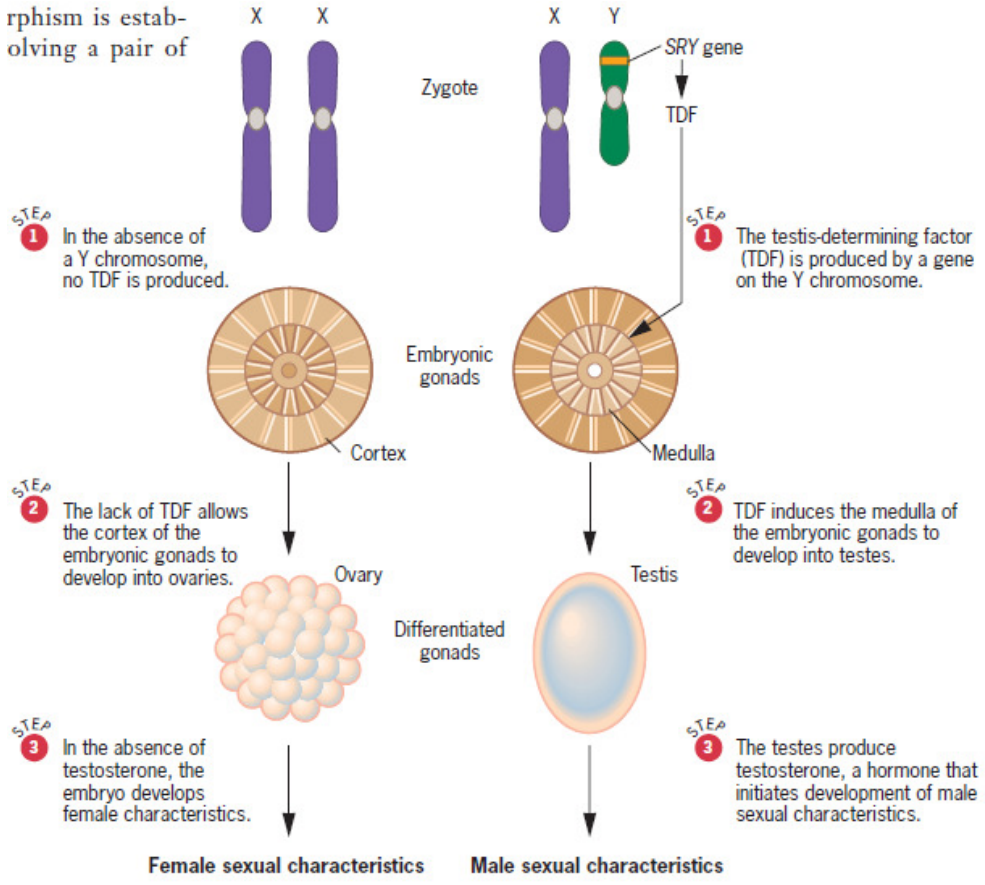

Fig-1: The process of sex determination in humans. Male sexual development depends on the production of the testisdetermining factor (TDF) by a gene on the Y chromosome. In the absence of this factor, the embryo develops as a female.

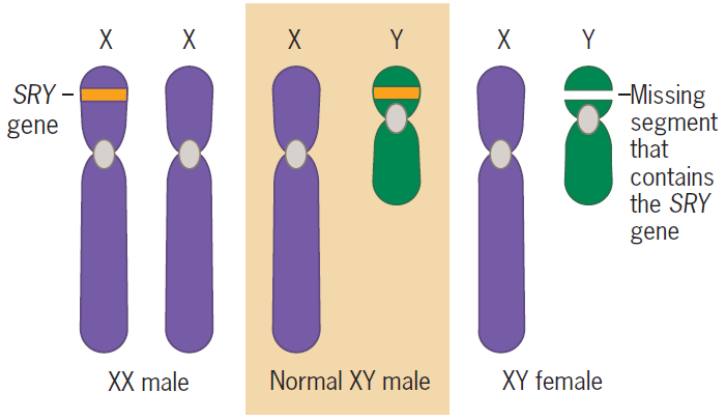

Fig-2: Evidence localizing the gene for the testis-determining factor (TDF) to the short arm of the Y chromosome in normal males. The TDF is the product of the *SRY* gene. In XX males, a small region containing this gene has been inserted into one of the X chromosomes, and in XY females, it has been deleted from the Y chromosome.

Sex Determination in *Drosophila:* (XX/XY system)

The Y chromosome in *Drosophila*—unlike that in humans—plays no role in sex determination. Instead, the sex of the fly is determined by the ratio of X chromosomes to autosomes. This mechanism was first demonstrated by Bridges in 1921 through an analysis of flies with unusual chromosome constitutions.

Normal diploid flies have a pair of sex chromosomes, either XX or XY, and three pairs of autosomes, usually denoted AA; here, each A represents one haploid set of autosomes. In complex experiments, Bridges contrived flies with abnormal numbers of chromosomes (**Table 1**). He observed that whenever the ratio of X's to A's was 1.0 or greater, the fly was female, and whenever it was 0.5 or less, the fly was male. Flies with an X:A ratio between 0.5 and 1.0 developed characteristics of both sexes; thus, Bridges called them *intersexes*. In none of these

flies did the Y chromosome have any effect on the sexual phenotype. It was, however, required for male fertility.

Table-1:

Ratio of X Chromosomes to Autosomes and the Corresponding Phenotype in *Drosophila*

X Chromosomes (X) and Sets of Autosomes (A)	X:A Ratio	Phenotype
1X 2A	0.5	Male
2X 2A	1.0	Female
3X 2A	1.5	Metafemale
4X 3A	1.33	Metafemale
4X 4A	1.0	Tetraploid female
3X 3A	1.0	Triploid female
3X 4A	0.75	Intersex
2X 3A	0.67	Intersex
2X 4A	0.5	Tetraploid male
1X 3A	0.33	Metamale

Q. 3- Answer:

Multiple allelism:

The Mendelian concept that genes exist in no more than two allelic states had to be modified when genes with three, four, or more alleles were discovered. This phenomenon where a gene controls a character through more than two alleles is called multiple allelism. A classic example of a gene with **multiple alleles** is the one that controls coat color in rabbits. Another example of multiple alleles comes from the study of human blood types. The A, B, AB, and O blood types, like the M, N, and MN blood types, are identified by testing a blood sample with different sera.

Mechanism of linkage: Sturtevant based his chromosome mapping procedure on the principle that genes on the same chromosome should be inherited together. Because such genes are

physically attached to the same structure, they should travel as a unit through meiosis. This phenomenon is called **linkage. (***Genes located close together on the same chromosome are called* ***linked genes*** *and belong to the same* ***linkage group.*** *Linked genes travel together during meiosis, eventually arriving at the same destination (the same gamete), and are not expected to assort independently*). However, linkage is not absolute. The experimental data demonstrated that genes on the same chromosome could be separated as they went through meiosis and that new combinations of genes could be formed. This phenomenon is called **recombination.**

Early Evidence for Linkage and Recombination:

Some of the first evidence for linkage came from experiments performed by W. Bateson and R. C. Punnett **(Fig-1).** These researchers crossed varieties of sweet peas that differed in two traits, flower color and pollen length. Plants with red flowers and long pollen grains were crossed to plants with white flowers and short pollen grains. All the F1 plants had red flowers and long pollen grains, indicating that the alleles for these two phenotypes were dominant. When the F1 plants were self-fertilized, Bateson and Punnett observed a peculiar distribution of phenotypes among the offspring. Instead of the 9:3:3:1 ratio expected for two independently assorting genes, they obtained a ratio of 24.3:1.1:1:7.1. Among the 803 F2 plants that were examined, the classes that resembled the original parents (called the parental classes) are significantly overrepresented and the two other (nonparental) classes are significantly underrepresented.

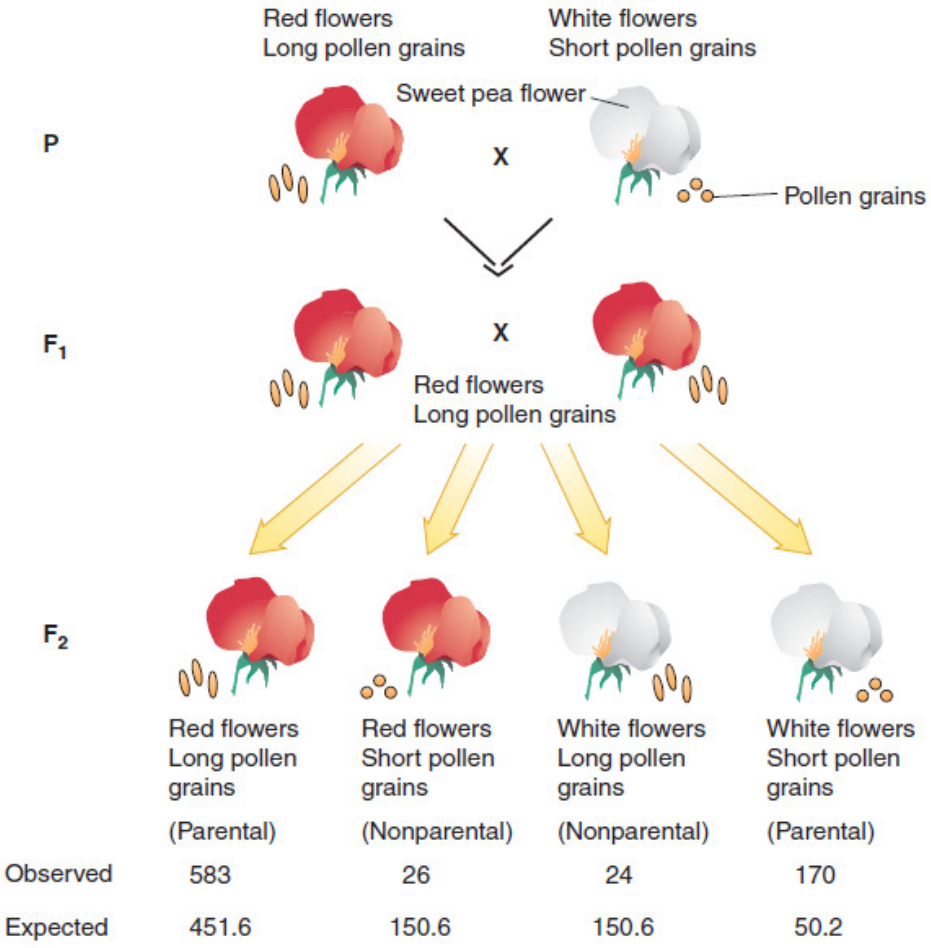

Fig-1: Bateson and Punnett's experiment with sweet peas. The results in the F2 indicate that the genes for flower color and pollen length do not assort independently.

Bateson and Punnett devised a complicated explanation for their results, but it turned out to be wrong. The correct explanation for the lack of independent assortment in the data is that the genes for flower color and pollen length are located on the same chromosome—that is, they are linked. This explanation is diagrammed in **–Fig-2**. The alleles of the flower color gene are *R* (red) and *r* (white), and the alleles of the pollen length gene are *L* (long) and *l* (short); the *R* and *L* alleles are dominant. (Note here that for historical reasons, the allele symbols are derived from the dominant rather than the recessive phenotypes.) Because the flower color and pollen length

genes are linked, we expect the doubly heterozygous F1 plants to produce two kinds of gametes, *R L* and *r l*. However, once in a while a crossover will occur between the two genes and their alleles will be recombined, producing two other kinds of gametes, *R l* and *r L*. The frequency of these two types of recombinant gametes should, of course, depend on the frequency of crossing over between the two genes.

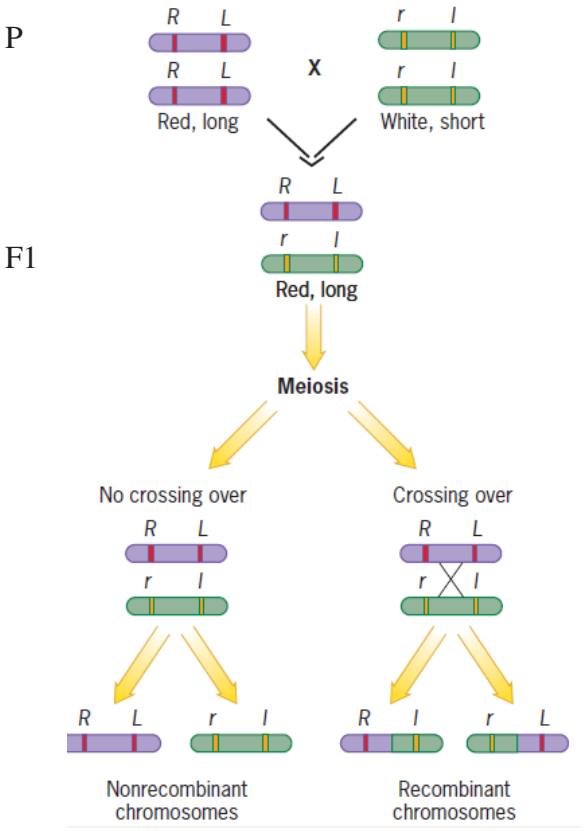

Fig-2: Hypothesis of linkage between the genes for flower color and pollen length in sweet peas. In the F1 plants the two dominant alleles, *R* and *L*, of the genes are situated on the same chromosome; their recessive alleles, *r* and *l*, are situated on the homologous chromosome.

Q. 4- Answer:

Karyotype is a term used for the chromosome constitution of a cell or an individual; chromosomes arranged in order of length and according to position of centromere; also, the

abbreviated formula for the chromosome constitution, such as 47, XX + 21 for human trisomy-21. **The human karyotype:**

Diploid human cells contain 46 chromosomes—44 autosomes and two sex chromosomes, which are XX in females and XY in males. At mitotic metaphase, each of the 46 chromosomes consists of two identical sister chromatids. When stained appropriately, each of the duplicated chromosomes can be recognized by its size, shape, and banding pattern. For cytological analysis, well-stained metaphase spreads are photographed, and then each of the chromosome images is cut out of the picture, matched with its partner to form homologous pairs, and arranged from largest to smallest on a chart (Fig-1).

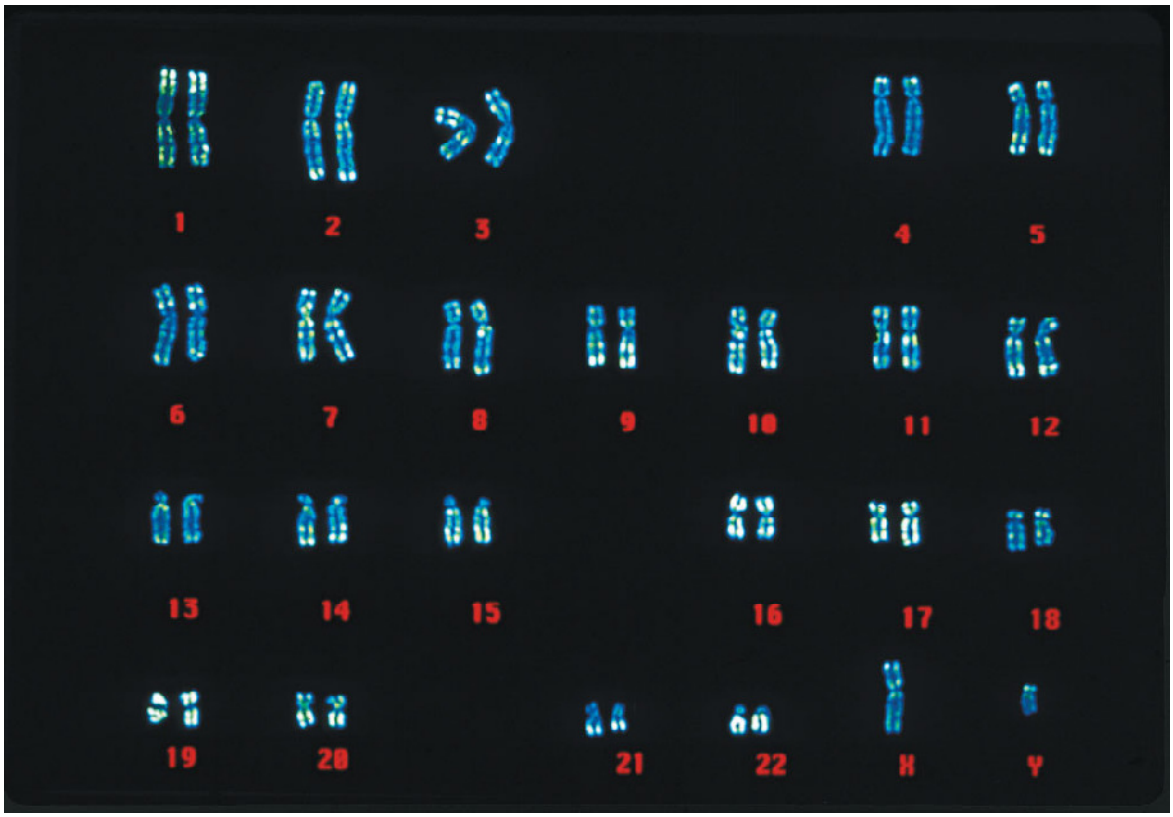

Fig-1: The karyotype of a human male stained to reveal bands on each of the chromosomes. The autosomes are numbered from 1 to 22. The X and Y are the sex chromosomes.

The largest autosome is number 1, and the smallest is number 21. (For historical reasons, the second smallest chromosome has been designated number 22.) The X chromosome is intermediate in size, and the Y chromosome is about the same size as chromosome 22. This chart of chromosome cutouts is called a **karyotype** (from the Greek word meaning "kernel," a reference to the contents of the nucleus). A skilled researcher can use a karyotype to identify abnormalities in chromosome number and structure.

Different banding pattern:

Preparation and staining techniques have been developed to help distinguish among chromosomes of similar size and shape. Before the banding and painting techniques were available, it was difficult to distinguish one human chromosome from another. Cytogeneticists could only arrange the chromosomes into groups according to size, classifying the largest as group A, the next largest as group B, and so forth. Although they could recognize seven different groups, within these groups it was nearly impossible to identify a particular chromosome. Today—as a result of the banding and painting techniques—we can routinely identify each of the chromosomes. The banding and painting techniques also make it possible to distinguish each arm of a chromosome and to investigate specific regions within them. For instance, chromosomes may be treated with enzymes that partly digest them; staining with a special dye called Giemsa reveals **G bands**, which distinguish areas of DNA that are rich in adenine–thymine base pairs. **Q bands** are revealed by staining chromosomes with quinacrine mustard and viewing the chromosomes under UV light. Other techniques reveal **C bands,** which are regions of DNA occupied by centromeric heterochromatin, and **R bands**, which are rich in guanine – cytosine base pairs.

Q. 5- Answer:

Cry of the cat syndrome (*cri-du-chat* syndrome): *(Due to deletion)*

A missing chromosome segment is referred to either as a **deletion** or as a **deficiency.** Large deletions can be detected cytologically by studying the banding patterns in stained chromosomes, but small ones cannot. In a diploid organism, the deletion of a chromosome segment makes part of the genome hypoploid. This hypoploidy may be associated with a phenotypic effect, especially if the deletion is large. A classic example is the ***cri-du-chat*** **syndrome** (from the French words for "cry of the cat") in humans. This condition is caused by a deletion in the short arm of chromosome 5. The size of the deletion varies. Individuals heterozygous for the deletion and a normal chromosome have the karyotype 46 del(5)(p14), where the terms in parentheses indicate that bands in region 14 of the short arm (p) of one of the chromosomes 5 is missing. These individuals may be severely impaired, mentally as well as physically; their plaintive, cat like crying during infancy gives the syndrome its name.

Cystic fibrosis: *(due to gene mutation)*

Cystic fibrosis is characterized by the accumulation of mucus in the lungs, pancreas, and liver, and the subsequent malfunction of these organs. It is the most common inherited disease in humans of northern European descent. It is inherited as an autosomal recessive mutation. Approximately 70 percent of the cases of CF result from a specific mutant allele of the *CF* gene, present on the long arm of chromosome 7. This mutant allele, *CFΔF508,* contains a three-base deletion that eliminates a phenylalanine residue at position 508 in the polypeptide product.

The polypeptide product of *CF* gene called the *cystic fibrosis transmembrane conductance regulator,* or *CFTR protein,* forms ion channels through the membranes of cells that line the respiratory tract, pancreas, sweat glands, intestine, and other organs and regulates the

flow of salts and water in and out of these cells. Because the mutant CFTR protein does not function properly in CF patients, salt accumulates in epithelial cells and mucus builds up on the surfaces of these cells.

The presence of mucus on the lining of the respiratory tract leads to chronic, progressive infections by *Pseudomonas aeruginosa, Staphylococcus aureus,* and related bacteria. These infections, in turn, frequently result in respiratory failure and death. However, the mutations in the *CF* gene are pleiotropic; they cause a number of distinct phenotypic effects. Malfunctions of the pancreas, liver, bones, and intestinal tract are common in individuals with CF.

Q. 6- Answer:

Industrial melanism: The industrial melanism in the peppered moth, ***Biston betuleria***, provides a well-studied example of directional selection from nature. Until 1848 in Europe (England), the vast majority of peppered moths had light colouration, which effectively camouflaged them against the light-coloured trees and lichens which they rested upon. However, because of widespread pollution during the Industrial Revolution in England, many of the lichens died out, and the trees that peppered moths rested on became blackened by soot, causing most of the light-coloured moths, to die off from predation. At the same time, the dark-coloured, or *melanic*, moths, ***Biston betuleria carbonaria***, flourished because of their ability to hide on the darkened trees. Resistance of insects to DDT and bacteria (*E. coli*) to drug, like chloramphenicol, are also a example of progressive natural selection.

Based upon the different organism-environment relationship, following four types of natural selection have been recognized:

1. Stabilizing selection: Operates in constant environment and prefer average characters and eliminate extreme characters. Example: Sparrows, Red Checkered Moth, etc.

2. Directional selection: It operates when environment is changing in one direction. It favors non-average or specialized phenotypes and eliminates the normal or average individuals. Example: Industrial melanism, drug resistance etc.

3. Disruptive selection: It acts to break up a previously homogenous population into several different adaptive forms. It indicated that extreme values have the highest fitness and the intermediate or mean values are relatively disadvantageous. It operates when a population previously adapted to a non-homogenous environment is subjected to divergent selection pressures in different parts of its distributional areas. Example: Sunflower population and Mimetic butterflies.

4. Cyclic selection: It operates when environment is not stable between generation or between seasons, the optimum phenotype and also the optimum genotype may show fluctuation because of selection operating in one direction in one generation or season, and in opposite direction for the next. This type of selection is called cyclic selection. It helps in maintaining genetic differences in a population and fixes all the allels of the gene pool.

Q. 7- Answer:

Recapitulation theory or Biogenetic law given by Ernst Haeckel, says that "ontogeny repeats phylogeny". Ontogeny is the life history of the individual starting from ovum and phylogeny is the sequence of adult ancestors which must have incurred in the evolution of the group of this individual.

Evidences of organic evolution from embryology:

A comparative study of embryology of different groups of animals reveals certain features that provide evidences for organic evolution. There are striking similarities among the different species of invertebrates that have a sequence of embryonic developments. These similarities are as follows:

(i) Gill clefts and notochord appear on the embryonic development of all vertebrates from fishes to mammals.

(ii) The notochord is replaced by the vertebral column in all adult vertebrates. (iii) Lungs in adult amphibians, reptiles and mammals replace gills.

The embryonic evidences can be taken into consideration on the basis of the following facts -

(i) Similarly in early development: In all the multicellular animals the fertilized egg (zygote) under goes cleavage to produce morula (solid mass of cells), blastula (a single layered hollow structure) & gastrula.

(ii) Resemblance in vertebrate embryos: The embryos of all vertebrates resemble one another.

(iii) Heart of a mammal or bird is initially two - chambered as in fished and finally becomes four-chambered.

(iv) Tadpole of frog resembles to fish shows "ontogeny repeats phylogeny".

Embryological evidences

Q. 8- Answer: Describe about the modern synthetic theory of evolution.

In the last few decades, a number of new facts have been added to the knowledge of evolution and theory of natural selection is reanalyzed in the light of genetics, Mendelism, population genetics and Biological species concept. This has given rise to 'Modern Synthetic Theory'.

The modern theory of origin of species or evolution is known as modern synthetic theory of evolution or neo-Darwinism. It is the merger of Darwinian selection, and genetic theory. The concept evolve after the book published by Julian Huxley entitled, "Evolution: The Modern Synthesis". Dobzhansky reviewed the Darwin concept of evolution by natural selection in

Mendelian population. In his book "Genetics and Origin of Species", he presented the chromosomal study of drosophila population and interrelation among its different species. Stebbins provided an account of "Variation and Evolution in Plants". Mayr established that modern synthetic theory is applicable to animals too. It presents a combination of mutations, variations, heredity, isolation and natural selection. According to modern synthetic theory, origin of species can be explained on the basis of:

1. Genetic variability in population

2. Significance of genetic variability

3. Natural selection, and

4. Isolation