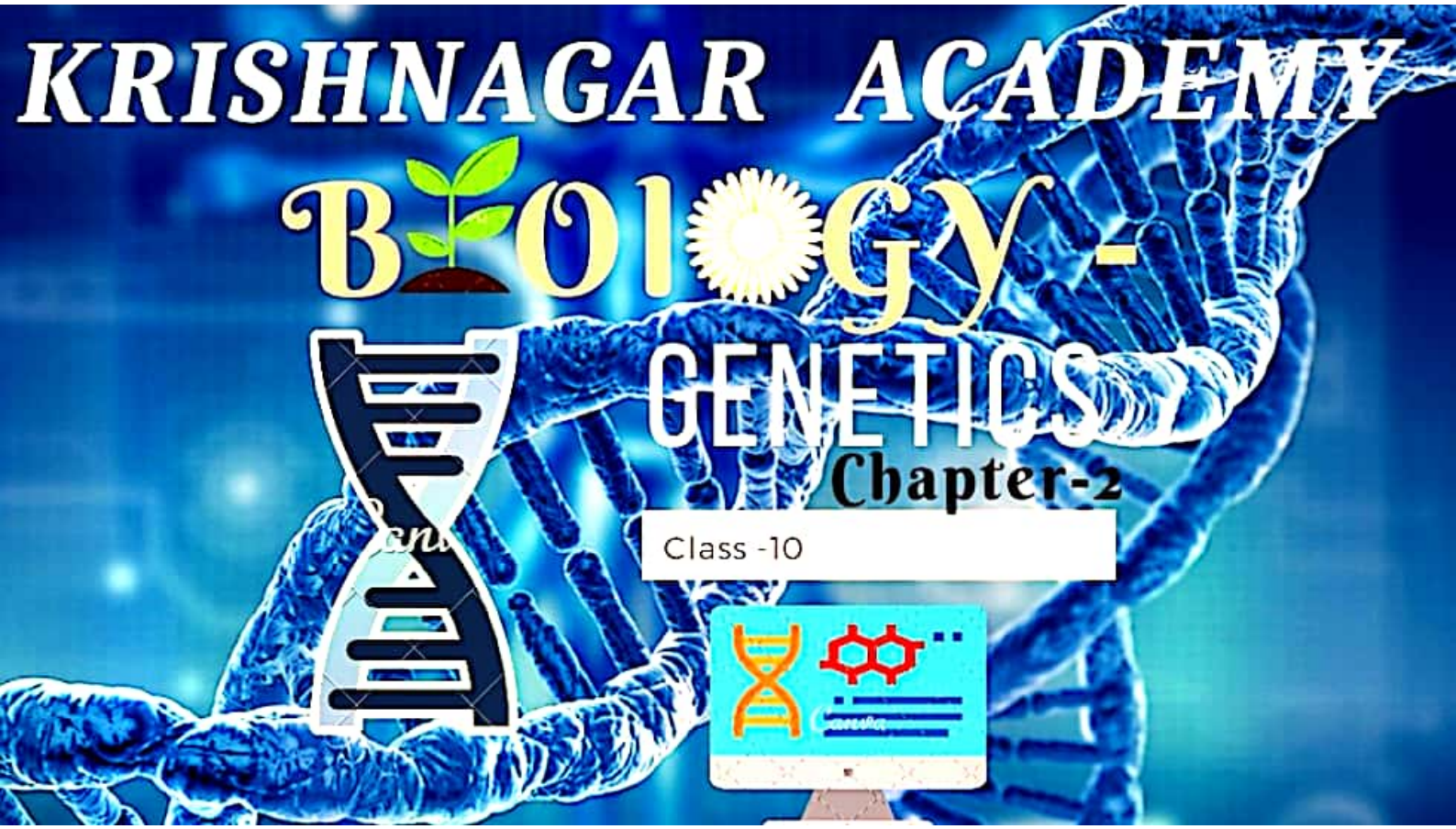


SYLLABUS

Structure of chromosome.

- *Basic structure of chromosome with elementary understanding of terms such as chromatin, chromatid, gene structure of DNA and centromere.*

Genetics : Mendel's laws of inheritance and sex linked inheritance of diseases.

- *Monohybrid cross, dihybrid cross.*
- *The following terms to be covered : gene, allele, heterozygous, homozygous, dominant, recessive, mutation, variation, phenotype, genotype.*
- *Sex determination in human beings.*
- *Sex linked inheritance of diseases to include haemophilia and colour blindness (only criss cross inheritance).*

GENETICS

'Like begets like'. An organism produces a young one of its own kind. But it is observed that no two individuals of the same species, in a sexually reproducing population, possess all similar characteristics with the exception of identical twins.

All members of a species inherit some common basic characteristics and all offsprings in a family share some common parental characteristics. The inheritance of both species and parental characteristics is called as "Heredity".

Individual characteristics are different in different individuals and hence help in identifying one individual from all others including his/her own real brothers and sisters. These are called variations. Variations may be acquired or somatogenic which are due to environmental factors or blastogenic which are due to inheritance of somewhat variable genetic material from parents.

The branch of biology which deals with the mechanism of the phenomenon of heredity and variations is called genetics.

The characters from one generation to next generation are transferred by thread like structures present in the nucleus. These are called chromosomes. Chromosomes exist as chromatin reticulum in the nucleus during interphase.

Chromosomes carry units of hereditary factors called genes. Higher organisms are diploid, *i.e.*, their chromosomes occur in pairs called homologous chromosomes. In a homologous chromosome each chromosome comes from either of the parents. Homologous chromosomes are similar in shape, size and structure and carry similar sequence of genes for a particular character.

Haploid cell like gametes carry unpaired chromosomes. This condition is called haploid (n).

In a diploid organism the characters may be present in alternative forms. For example, one homologous chromosome may carry the gene for tallness and on the other homologous chromosome the gene may be for short height. These two alternative forms of a gene are called alleles.

Chromatin reticulum converts into chromosome during cell division. When viewed under light microscope these chromosome show differences in their size and shape. These chromosomes are arranged in their decreasing size. The size, shape and structure of chromosomes are different for different organisms and is called the karyotype of that organisms.

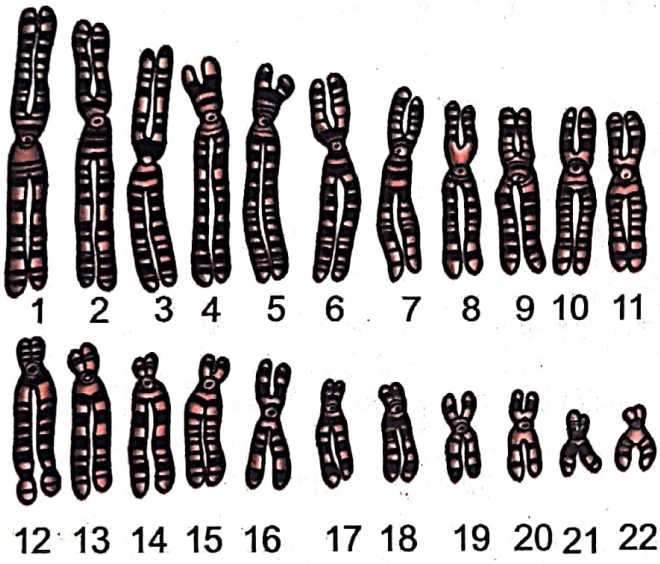


Fig. 1. Human karyotype.

Fig. 2. X and Y-chromosomes.

Chromosomes in an organism are of two different types.

(a) Autosomes : Chromosomes which control the various characters of the organisms due to genes present on them, but have no genes for the expression of sexual characteristics.

(b) Sex Chromosomes : Chromosomes which control the expression of sex or determine sex of an individual are called sex chromosomes. These are 'X' and ' Y'. 'Y' is the smallest chromosome.

Since, females carry similar sex chromosomes they are homomorphic or isomorphic. The males contain two different chromosomes X and Y and are thus heteromorphic.

SEX DETERMINATION

In human beings males carry XY sex chromosome. These chromosomes separate during meiosis for sperm formation. Sperms are of two types (22 + X) and (22 + Y) and are produced in equal proportion. The males are thus heterogametic. The human females have (44 + XX) condition and produce similar ova carrying 22 + X chromosomes. The females are thus homogametic.

The sex of the child is determined at the time of fertilization and is decided by the kind of sperm which fertilizes the ova, which is a matter of chance. Female parent does not decide the sex of the child.

(a) Ova (22 + X) + Sperm (22 + X) = 44 + XX, *i.e.*, female child.

(b) Ova (22 + X) + Sperm (22 + Y) = 44 + XY, *i.e.*, male child.

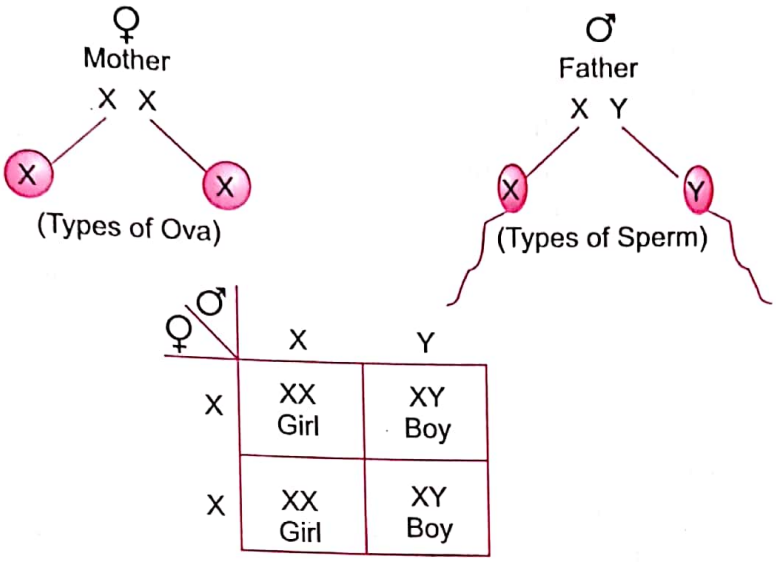


Fig. 3. Mechanism of sex-determination in humans.

GENES

The physical as well as physiological characteristics of organisms are determined by the genes which are present in the chromosomes in a linear order. Genes express themselves by producing enzymes which are proteins, *i.e.*, each gene synthesizes a particular protein which acts as an enzyme and is responsible for the appearance of a particular character.

A gene is a segment of DNA of a chromosome which determines the appearance of a particular character. Each gene occupies a specific position in a specific chromosome. This position is known as locus. A single gene may occur in alternative forms known as alleles. Alleles occupy the same locus on homologous chromosomes. The alleles may be related as dominant or recessive.

Dominant : The allele which expresses itself both in homozygous and heterozygous condition is a dominant allele.

Recessive : The allele which expresses itself only in homozygous condition is a recessive allele.

CHROMOSOMES

Chromosomes in eukaryotic organisms remain uncondensed in the form of chromatin fibres during interphase. These chromatin fibres condense during cell division forming chromosomes. Chromosomes are hereditary vehicle because they store hereditary information and transmit them from parents to offspring.

Chromosome

Nuclear Chromosome	Extranuclear Chromosome
Contain DNA + Histone	Formed of double stranded circular DNA
Present inside the nucleus	Present in mitochondria and plastids

The number of chromosome in the somatic cells of higher animals and plants is known as diploid, *i.e.*, the chromosomes are paired and is represented by 2n. In gametes and lower organisms the chromosomes are unpaired and the condition is called haploid. The number of chromosomes is constant for a particular species.

DNA is coiled around a core of eight histone proteins to form a bead like structure called nucleosome. These nucleosomes coil in a spiral manner to form a solenoid. The solenoid coils further to form a supersolenoid. By further condensation, the supersolenoid form chromosome.

Structure of Chromosome : Each chromosome is made of two identical halves called sister chromatids. The two chromatids remain connected with each other at a point called centromere. The position of centromere in a chromosome remains fixed and is characteristic for a given chromosome. The centromere provides attachment to the spindle fibre during cell division. The free ends (tips) of the chromosome are called telomeres. They prevent the chromosomes from sticking with each other.

Nucleosome

Histone	DNA
Basic Protein Eight histone molecules form octamer which forms core of nucleosome	Deoxyribonucleic acid, a long polymer of nucleotide or polynucleotide

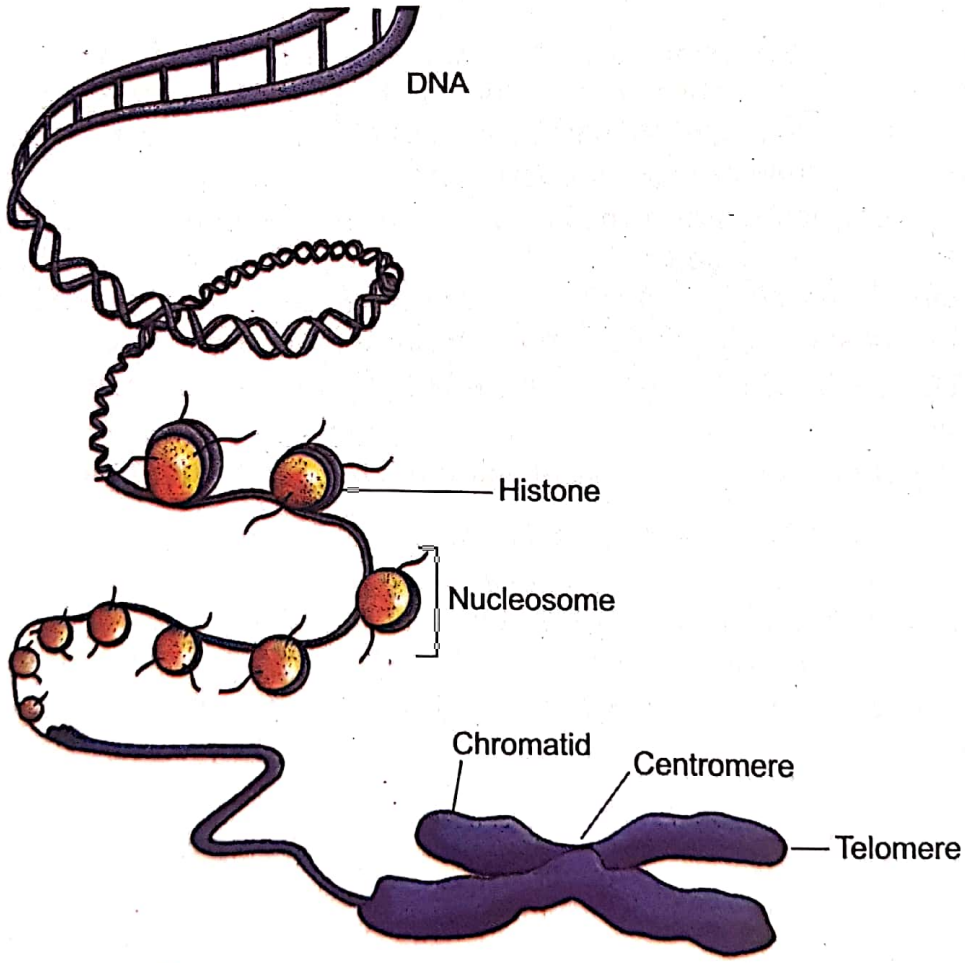


Fig. 4. Levels of organisation of chromosome.

STRUCTURE OF DNA

DNA is a polymer of nucleotides. Its molecular structure was proposed by Watson and Crick for which they were awarded Nobel Prize in the year 1953.

DNA (Deoxyribonucleic acid) is made up of three different types of molecules :

(a) Nitrogenous bases

(b) Pentose sugar-deoxyribose

(c) Phosphoric acid (H_3PO_4) : represented by (P)

(a) Nitrogenous bases are of two types.

Pyrimidine : These are single-ringed compounds and are of the following types in DNA :

(i) Cytosine

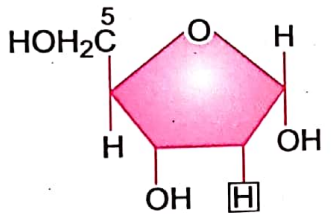
(ii) Thymine

Purine : These are double-ringed compounds and are of the following types :

(i) Adenine

(ii) Guanine

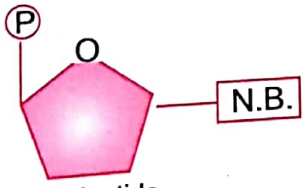
(b) Pentose sugar : It is a five-carbon sugar named as deoxyribose.



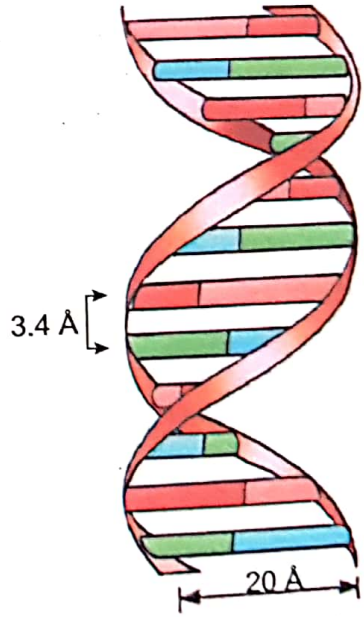
Pentose sugar-deoxyribose

Nitrogen base + Sugar = Nucleoside

Nucleoside + H_3PO_4 = Nucleotide



A nucleotide

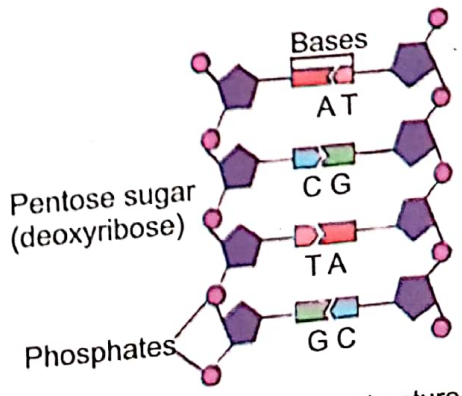


(a) Helical structure

Each DNA is a double helical structure 20Å in diameter. The two strands of DNA are antiparallel and are linked together by complementary base pairing in which one purine is attached to a pyrimidine by hydrogen bond. Adenine always pairs with thymine by two hydrogen bonds; similarly cytosine always pairs with guanine by three hydrogen bonds.

A = T

C ≡ G



(b) Molecular structure

During S-phase of interphase, DNA forms its identical copies. This process is called replication of DNA.

IMPORTANT TERMS AND DEFINITIONS

1. **Allele:** It is one of a number of alternative forms of the same gene responsible for determining contrasting characteristics. For example, A or a.
2. **Character:** Any feature that is transmitted from the parent to the offspring.
   Hereditary characters: Characteristics that are transmitted from the parents to the offspring.
3. **Chromosomes** are the carriers of heredity.
4. **Criss-cross Inheritance:** The transmission of a gene from mother to son or father to daughter. For example, X-chromosome linkage.
5. **Cloning:** It is the process of producing genetically identical individual. Organisms like bacteria, insects or some plants reproduce asexually to form exact individual of itself.
6. **Dominant Character:** It is one out of a pair of contrasting characters (alleles) which is fully expressed in an offspring either in homozygous or heterozygous form.
7. **Dihybrid Cross:** It is the cross which is carried out by taking two pairs of contrasting characters at a time.
8. **Dihybrid Ratio:** The ratio obtained in a dihybrid cross. Its phenotype ratio is 9 : 3 : 3 : 1 (1 : 2 : 1 : 2 : 4 : 2 : 1 : 2 : 1 = 9 types of genotypes).
9. **F_1 Generation:** First filial generation that is produced by crossing two parents (homozygous or heterozygous).
10. **F_2 Generation:** Second filial generation that is produced by crossing two individuals of F_1 generation.
11. **Genetics:** It is the branch of biology that deals with the study of transmission of characters from the parents to the offspring.
12. **Gene:** Gene are the basic unit of heredity and these are found on chromosomes. Genes are the DNA segments that carry genetic information.
    DNA is the chemical basis of gene. **Watson** and **Crick** have given the **model of DNA.** The change of character due to several conditions carried by genes is called **mutation**. Linkage is the tendency of genes to stay together in hereditary transmission. Pedigree helps in understanding the nature of traits and their inheritance.
    Every gene has alternative forms for a character producing different effect. These alternative forms are called affects.
13. **Genome:** It is the whole complement of DNA of an organism.
14. **Genotype:** The characters which are not visible from outside in an organism.
15. **Heredity:** The transmission of genetically based characteristics from the parents to the offspring.

16. **Homologous Chromosomes:** A pair of corresponding chromosomes of the same size and shape, one from each parent.
17. **Homozygous:** It is the condition in which both the alleles are identical or same for a particular character.
18. **Heterozygous:** It is the condition in which both the alleles are different for a particular character.
19. **Law of Dominance:** Out of a pair of contrasting characters (alleles) present together in an offspring, one dominates over the other.
20. **Law of Segregation (First Law of Mendel):** The contrasting factors or characters (alleles) separate during gamete formation. At the time of zygote formation, the gametes combine randomly but alleles or characters do not blend and separate during gamete formation.
21. **Law of Independent Assortment (Second Law of Mendel):** The inheritance of each contrasting character (allele) is independent of other contrasting character present together.
22. **Linkage:** The state in which two genes are linked because they are located on the same chromosome, and are often inherited together.
23. **Monohybrid Cross:** It is a cross which is carried out by taking only one pair of contrasting character at a time.
24. **Monohybrid Ratio:** The ratio obtained in a monohybrid cross. The monohybrid phenotype ratio is 3 : 1 and genotype ratio is 1 : 2 : 1.
25. **Mutation:** The sudden change in one or more genes, which change the hereditary materials of an organism and ultimately change the character of an organism. It may be:
    (*i*) **Gene Mutation:** Changes in the DNA,. Sickle-cell anemia is caused due to gene mutation.
    (*ii*) **Chromosomal Mutation:** Changes in the number or arrangement on the number of the chromosome. Chromosomal mutation is seen in Down's syndrome.
26. **Phenotype:** The characters which are visible from outside in an organism.
27. **Recessive Character:** It is one out of a pair of contrasting characters (alleles) which remains masked in an offspring.
28. **Sex-Linked Inheritance:** Sex-linked inheritance is the appearance of trait by an allele on X chromosome or Y chromosome. Haemophilia and colour blindness are the two sex-linked inherited diseases. These defects are due to recessive genes which occur on the X chromosome.
29. **Traits:** A quality or characteristic that can belong to only a person.
30. **Variation:** The differences shown amongst individuals of a species or offspring of the same parent due to their different genetic makeup.

Genetics Hometask

Class - 10.

1. Define : a) Karyotype
b) Allele.
c) Genes.
d) Heredity.

2. Differentiate : a) Chromosome & chromatin
b) Diploid & haploid.
c) Autosome & Allosome.
d) Dominant & recessive
e) Homogametic & heterogametic

3. Describe in brief : a) Structure of DNA.
b) Organisation of chromosome.