CHROMOSOMES & GENES

The chromosomes are capable of self–reproduction and maintaining morphological and physiological properties through successive generations. They are capable of transmitting the contained hereditary material to the next generation. Hence these are known as '**hereditary vehicles**'.

(1) **Discovery of chromosomes**

Hofmeister (1848) : First observed chromosomes in microsporocytes (microspore mother cells) of *Tradescantia*.

Flemming (1879) : Observed splitting of chromosomes during cell division and coined the term, 'chromatin'.

Roux (1883) : He believed the chromosomes take part in inheritance.

W.Waldeyer (1888) : He coined the term 'chromosome'.

Benden and Boveri (1887) : They found a fixed number of chromosomes in each species.

(2) **Kinds of chromosomes**

(i) **Viral chromosomes :** In viruses and bacteriophages a single molecule of DNA or RNA represents the viral chromosome.

(ii) **Bacterial chromosomes :** In bacteria and cyanobacteria, the hereditary matter is organized into a single large, circular molecule of double stranded DNA, which is loosely packed in the nuclear zone. It is known as bacterial chromosome or *nucleoid*.

(iii) **Eukaryotic chromosomes :** Chromosomes of eukaryotic cells are specific individualized bodies, formed of deoxyribonucleo proteins (DNA + Proteins).

(3) **Chromosomal theory of inheritance:** It was proposed independently by *Sutton* and *Boveri* in 1902. The chromosome theory of inheritance proposes that chromosomes are vehicles of hereditary information and expression as Mendelian factors or genes.

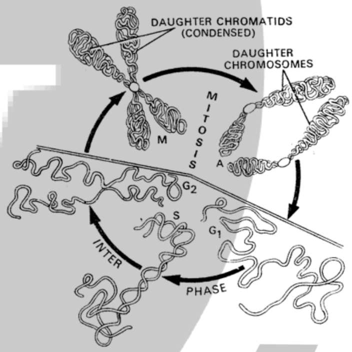


Fig : Diagram showing chromosome

(i) Bridge between one generation to the next are sperm and ovum.

(ii) Both sperm and ovum contribute equally in heredity. Sperm provides only nucleus for fertilization. Therefore, heredity must be based in nuclear material.

(iii) Nucleus possesses chromosomes. Therefore, chromosomes must carry hereditary characters.

(iv) Chromosomes, like hereditary factors are particulate structures, which maintain their number, structure and individuality in organisms from generation to generation.

(4) **Chromosomes number :** Chromosome number is n = 2 in Mucor hiemalis, 2n=4 in plant *Haplopappus gracilis*. Chromosome number is 14 (n=7) in Pea, 20 in Maize, 46 in human beings. Maximum number of chromosomes is known for Adder's Tongue Fern (*Ophioglossum reticulatum,* 2n = 1262) and *Aulocantha* (2n=1600). Number of chromosomes is not related to complexity or size of organism *e.g.*, Domestic Fowl and Dog both possess 78 chromosomes. Study of chromosome structure is performed at metaphase and study of chromosome shape at anaphase.

(5) **Chromosome cycle and cell cycle :** Chromosomes exhibit cyclic change in shape and size during cell cycle. In the non-dividing interphase nucleus, the chromosomes form an interwoven network of fine twisted but uncoiled threads of chromatin, and are invisible. During cell division the chromatin threads condense into compact structures by helical coiling.

(6) **Chromosome structure :** Different regions (structures) recognized in chromosomes are as under.

(i) **Pellicle** : It is the outer thin but doubtful covering or sheath of the chromosome.

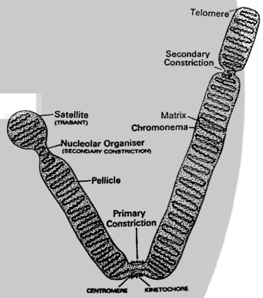
(ii) **Matrix :** Matrix or ground substance of the chromosome is made up of proteins, small quantities of RNA and lipid. It has one or two chromonemata (singular - chromonema) depending upon the state of chromosome.

(iii) **Chromonemata :** They are coiled threads which form the bulk of chromosomes. A chromosome may have one (anaphase) or two (prophase and metaphase) chromonemata. There are three view points about the constitution of chromonema and chromosome.

(iv) **A Primary Constriction and Centromere (kinetchore) :** A part of the chromosome is marked by a constriction. It is comparatively narrow than the remaining chromosome. It is known as primary constriction. The primary constriction divides the chromosome into two arms. It shows a faintly positive Feulgen reaction, indicating presence of DNA of repetitive type. This DNA is called centromeric heterochromatin.

(v) **Centromere :** Centromere or kinetochore lies in the region of primary constriction. The microtubules of the chromosomal spindle fibres are attached to the centromere. Therefore, centromere is associated with the chromosomal movement during cell division. Kinetochore is the outermost covering of centromere.

(vi) Secondary constriction or nucleolar organizer : **Sometimes one or both the arms of a chromosome are marked by a constriction other than the primary constriction. During interphase this area is associated with the nucleolus and is found to participate in the formation of *nucleolus.* It is, therefore, known as nucleolar organizer region or the secondary constriction.**



Nucleolar organizer region (NOR) : In certain chromosomes, the secondary constriction is (In human beings 13, 14, 15, 20 and 21 chromosome are nucleolar organizer) intimately associated with the nucleolus during interphase. It contains genes coding for **18S** and **28S** ribosomal **RNA** and is responsible for the formation of nucleolus. Therefore, it is known as *nucleolar organizer region* (*NOR*).

(vii) **Telomeres :** The tips of the chromosomes are rounded and sealed and are called telomeres which play role in Biological clock. The terminal part of a chromosome beyond secondary constriction is called *satellite.* The chromosome with satellite is known as *sat chromosome*, which have repeated base sequence.

(viii) **Chromatids :** At metaphase stage a chromosome consists of two chromatids joined at the common centromere. In the beginning of anaphase when centromere divides, the two chromatids acquire independent centromere and each one changes into a chromosome.

(7) **Types of chromosomes based on number of centromeres :** Depending upon the number of centromeres, the chromosomes may be:

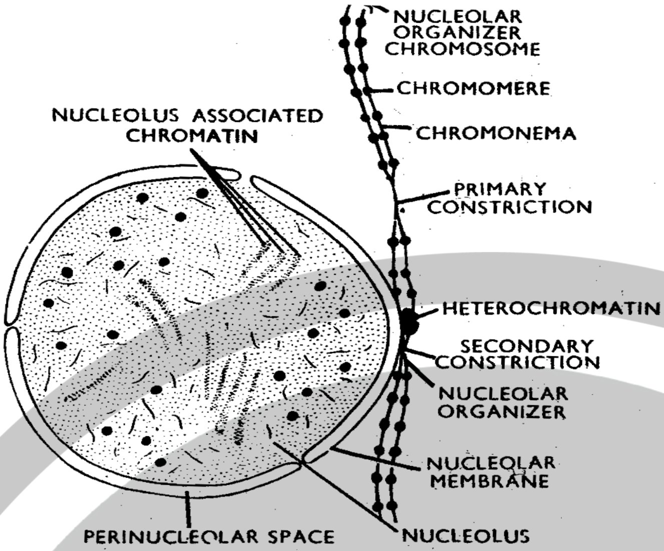


Fig : Nucleolus organizer or secondary constriction and its association with necleolus

(i) Monocentric with one centromere.

(ii) Dicentric with two centromeres, one in each chromatid.

(iii) Polycentric with more than two centromeres.

(iv) Acentric without centromere. Such chromosomes represent freshly broken segments of chromosomes, which do not survive for long.

(v) Diffused or non-located with indistinct centromere diffused throughout the length of chromosome. The microtubules of spindle fibres are attached to chromosome arms at many points. The diffused centromeres are found in insects, some algae and some groups of plants (*e.g. Luzula*).

(8) **Types of chromosomes based on position of centromere :** Based on the location of centromere the chromosomes are categorised as follows:

(i) **Telocentric :** These are rod-shaped chromosomes with centromere occupying a terminal position. One arm is very long and the other is absent.

(ii) **Acrocentric :** These are rod-shaped chromosomes having subterminal centromere. One arm is very long and the other is very small.

(iii) **Submetacentric :** These are **J** or **L** shaped chromosomes with centromere slightly away from the mid-point so that the two arms are unequal.

(iv) **Metacentric :** These are **V**-shaped chromosomes in which centromere lies in the middle of chromosomes so that the two arms are almost equal.

(9) **Molecular organisation of chromosome :** Broadly speaking there are two types of models stating the relative position of DNA and proteins in the chromosomes.

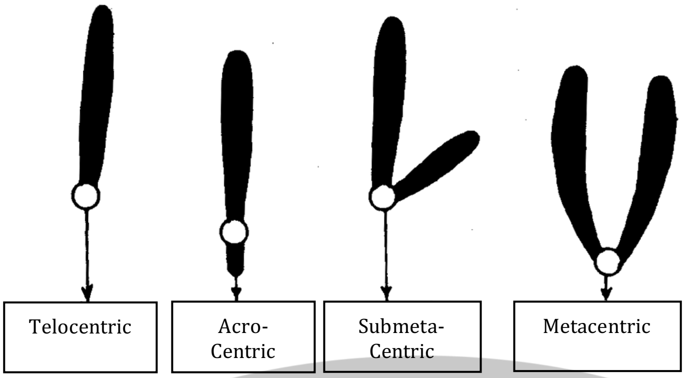


Fig : Types of chromosomes

Multiple strand models : According to several workers (Steffensen 1952, Ris 1960) a chromosome is thought to be composed of several DNA protein fibrils, chromatids are made up by several DNA protein fibrils and atleast two chromatids form the chromosome.

Single strand models : According to Taylor, Du prow etc. The chromosome is made up of a single DNA- protein fibril. There are some popular single strand models.

(i) **Folded fiber model :** Chromosomes are made up of very fine fibrils 2 *nm* - 4 *nm* in thickness. As the diameter of DNA molecule is also 2 *nm* (20Å). So it is considered that a single fibril is a DNA molecule. It is also seen that chromosome is about a hundred times ticker than DNA whereas the length of DNA in chromosome is several hundred times that of the length of chromosome. So it is considered that long DNA molecule is present in folding manner which forms a famous model of chromosome called folded fibre model which given by *E.J. Dupraw* (1965).

(ii) **Nucleosome model :** The most accepted model of chromosome or chromatin structure is the 'nucleosome model' proposed by Kornberg and Thomas (1974). Nucleosomes are also called *core particles or Nu-bodies*. The name nucleosome was given by *P. Outdet* etal. The nucleosome is a oblate particle of 55Å height and 110Å diameter. Woodcock (1973) observed the structure of chromatin under electron microscope. He termed each beaded structure on chromosome as nucleosome. Nucleosome is quasicylindrical structure made up of histones and DNA.

(a) **Structural proteins (histones) :** Histones are main structural protein found in eukaryotic cells. These are low molecular weight proteins with high proportion of positively charged basic amino acids arginine and lysine.

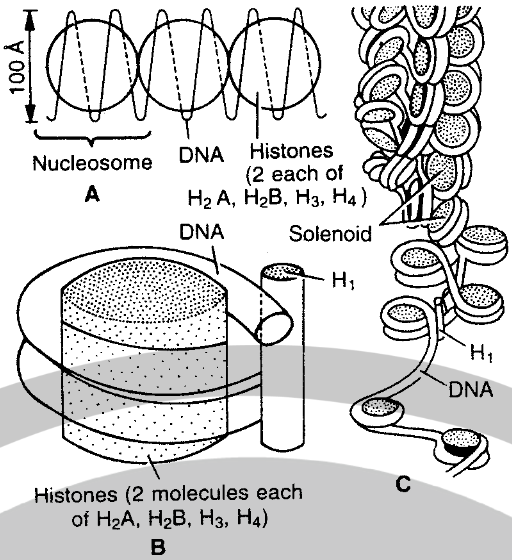


Fig : A-C Nucleosome : A Units of nucleosome, B Nucleosome model C Solenoid model

Types of histones : These are <u>five different types of histones</u> that fall into two categories.

Nucleosomal histones : These are small proteins responsible for coiling DNA into nucleosome. These are H_2A, H_2B, H_3 and H_4 (two molecule of each four histone protein form a octamer structure). These form the inner core of nucleosome.

***H*$_1$-histones or linker histon protein :** These are large (about 200 amino acids) and are tissue specific. These are present once per 200 base pairs. These are loosely associated with DNA. H_1 histones are responsible for packing of nucleosomes into 30 *nm* fiber.

Functions of histones : Histones in eukaryotic chromosomes serve some functions.

- These either serve as structural elements and help in coiling and packing of long DNA molecules.
- Transcription is possible only by dissolution of histones in response to certain molecular signals.

(b) **DNA in nucleosome :** Nucleosome is made of core of eight molecules of histones wrapped by double helical DNA with $1\frac{3}{4}$ turns making a repeating unit. Every $1\frac{3}{4}$ turn of DNA have 146 base pairs. When H_1 protein is added the nucleotide number becomes 200. DNA which joins two nucleosome is called linkar DNA or spacer DNA.

(iii) **Solenoid model :** In this model the nucleosomal bead represents the first degree of coiling of DNA. It is further coiled to form a structure called solenoid (having six nucleosome per turn). It represents the second degree of coiling. The diameter of solenoid is 300Å. The solenoid is further coiled to form a supersolenoid of 2000-4000Å diameter. This represent the third degree of coiling. The supersolenoid is perhaps the unit fiber or chromonema identified under light microscopy. The solenoid model was given by Fincy and Klug 1976. A Klug was awarded by noble prize in 1982 for his work on chromosome.

(iv) **Dangier-String or Radial Loop Model :** (Laemmli, 1977). Each chromosome has one or two interconnected scaffolds made of nonhistone chromosomal proteins. The scaffold bears a large number of lateral loops all over it. Both exit and entry of a lateral loop lie near each other. Each lateral loop is 30 *nm* thick fibre similar to chromatin fibre. It develops through solenoid coiling of nucleosome chain with about six nucleosomes per turn. The loops undergo folding during compaction of chromatin to form chromosome.

(10) **Giant chromosomes :** These chromosomes are of two types.

(i) **Polytene chromosome :** Polytene chromosome was described by *Kollar* (1882) and first reported by *Balbiani* (1881). They are found in salivary glands of insects (*Drosophila*) and called as salivary gland chromosomes. These are reported in endosperm cells of embryosac by *Malik* and *Singh* (1979). Length of this chromosome may be upto 2000μm. The chromosome is formed by somatic pairs between homologous chromosomes and repeated replication or endomitosis of chromonemata. These are attached to chromocentre. It has pericentromeric heterochromatin. Polytene chromosomes show a large number of various sized intensity bands when stained. The lighter area between dark bands are called interbands. They have puffs bearing *Balbiani rings*. Balbiani rings produce a number of m-RNA, which may remain stored temporarily in the puffs, are temporary structures.

(ii)

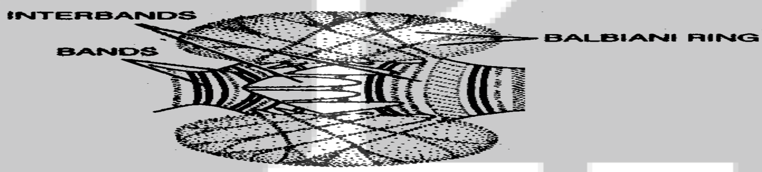


Fig : Polytene chromosome showing

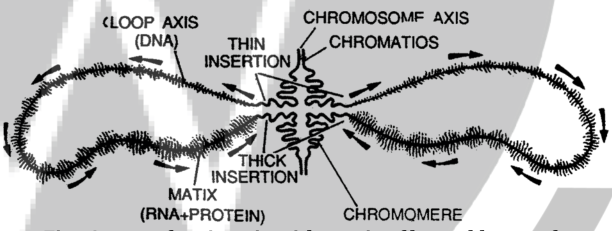


Fig : A part of main axis with a pair of lateral loops of a lampbrush chromosome showing synthesis of RNA

Lampbrush chromosomes : They are very much elongated special type of synapsed or diplotence chromosome bivalents already undergone crossing over and first observed by *Flemming* (1882). The structure of lampbrush chromosome was described by *Ruckert* (1892). They are found in *oocyte, spermatocytes* of many animals. It is also reported in *Acetabularia* (unicellular alga) by *Spring et.al. in* 1975*. In urodele oocyte the length of lampbrush chromosome is upto* 5900*μm*. These are found in pairs consisting of homologous chromosomes jointed at chiasmata (meiotic prophase–I). The chromosome has double main axis due to two elongated chromatids. Each chromosome has rows of large number of chromatid giving out lateral loops, which are uncoiled parts of chromomere with one–many transcriptional units and are involved in rapid transcription of mRNA meant for synthesis of yolk and other substances required for growth and development of meiocytes. Some mRNA produced by lampbrush chromosome is also stored as informosomes *i.e.*, mRNA coated by protein for producing biochemicals during the early development of embryo. Length of loop may vary between 5-100 μm.