Chromosome number:

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- Chromosomes vary both in number and structure among organisms and the number of chromosomes is characteristic of every species. Benden and Bovery in 1887 reported that the number of chromosomes in each species is constant. W.S. Sutton and T. Boveri in 1902 suggested that chromosomes are the physical structures which acted as messengers of heredity.

Chromosome number:

There are normally two copies of each chromosome present in every somatic cell. The number of unique chromosomes (N) in such a cell is known as its **haploid number,** and the total number of chromosomes (2N) is its diploid number. The suffix ‘ploid’ refers to chromosome ‘sets’.

The haploid set of the chromosome is also known as **the genome.** Structurally, eukaryotes possess large linear chromosomes unlike prokaryotes which have circular chromosomes. In Eukaryotes other than the nucleus chromosomes are present in mitochondria and chloroplast too.

The number of chromosomes in each somatic cell is same for all members of a given species. The organism with lowest number of chromosome is the nematode, *Ascaris megalocephalusunivalens* which has only two chromosomes in the somatic cells (2n=2).

Autosomes and sex chromosomes:

In a diploid cell, there are two of each kind of chromosome (termed homologus chromosomes) except the sex chromosomes. In humans one of the sex has two of the same kind of sex chromosomes and the other has one of each kind. In humans there are 23 pairs of **homologous chromosomes** (2n=46).

The human female has 44 non sex chromosomes, termed autosomes and one pair of homomorphic sex chromosomes given the designation XX. The human male has 44 autosomes and one pair of heteromorphic sex chromosomes, one X and one Y chromosome.

Human Chromosome: The human genome is 3 x 109 base pairs of DNA and the smallest human chromosome is several times larger than the entire yeast genome; and the extended length of DNA that makes up the human genome is about 1 m long. The human genome is distributed among 24 chromosomes (22 autosomes and the 2 sex chromosomes), each containing between 45 and 280 Mb of DNA. The sex chromosomes are denoted by X and Y and they contain genes which determine the sex of an individual i.e., XX for female and XY for male.

The rest are known as autosomes. The haploid human genome contains about 23,000 protein-coding genes, which are far fewer than had been expected before sequencing. In fact, only about 1.5% of the genome codes for proteins, while the rest consists of non-coding genes, regulatory sequences, introns, and noncoding DNA. Chromosomes are stained with A-T (G bands) and G-C (R bands) base pair specific dyes **(Figure 1)**. When they are stained, the mitotic chromosomes have a banded structure that unambiguously identifies each chromosome of a karyotype.

Each band contains millions of DNA nucleotide pairs which do not correspond to any functional structure. G-banding is obtained with Giemsa stain yielding a series of lightly and darkly stained bands. The dark regions tend to be heterochromatic and AT rich. The light regions tend to be euchromatic and GC rich. R-banding is the reverse of G-banding where the dark regions are euchromatic and the bright regions are heterochromatic.

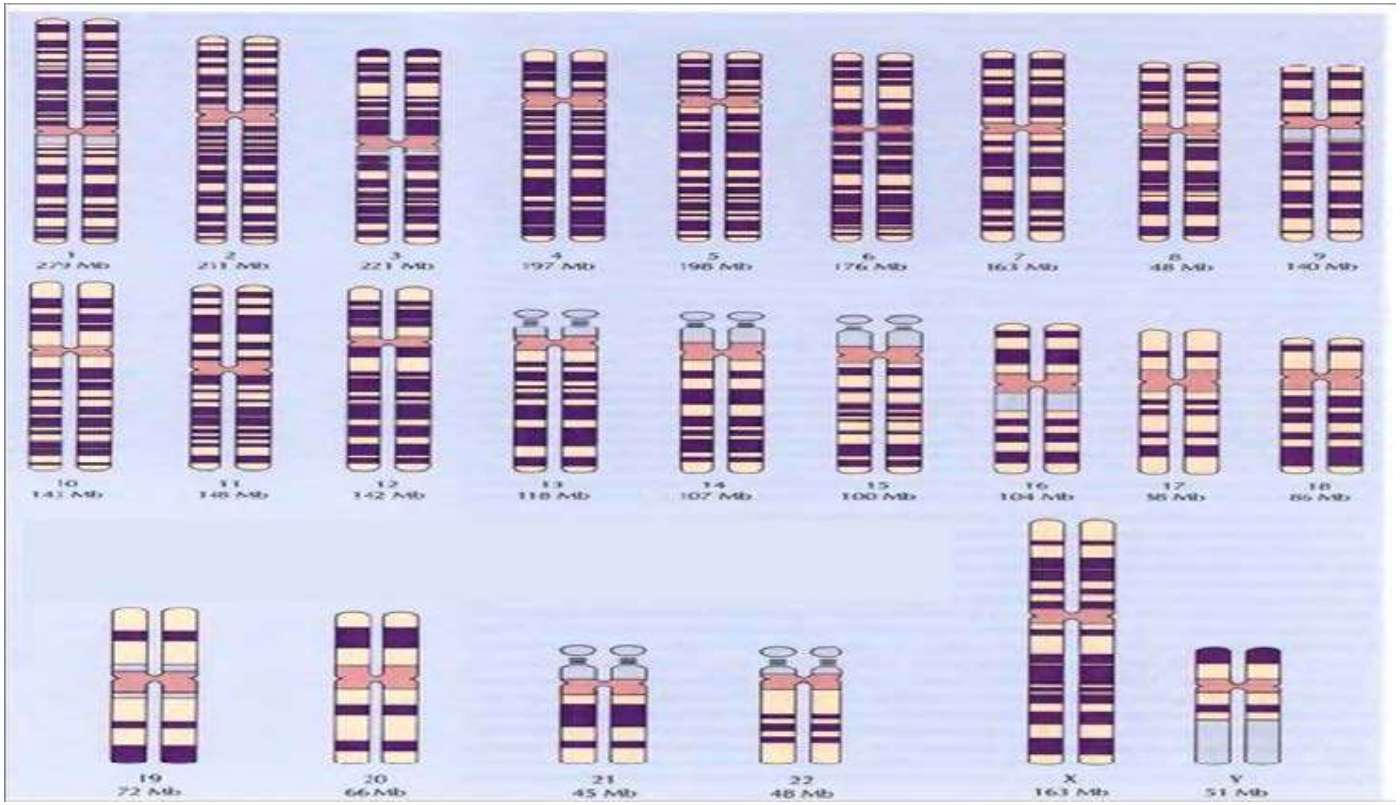


Figure 1: Human metaphase chromosome showing the banding pattern obtained after cytogenetic staining. This figure has been adapted from "The Cell, A Molecular Approach" by Geoffrey M. Cooper, 4th Ed. 2007.

Types of chromosomes

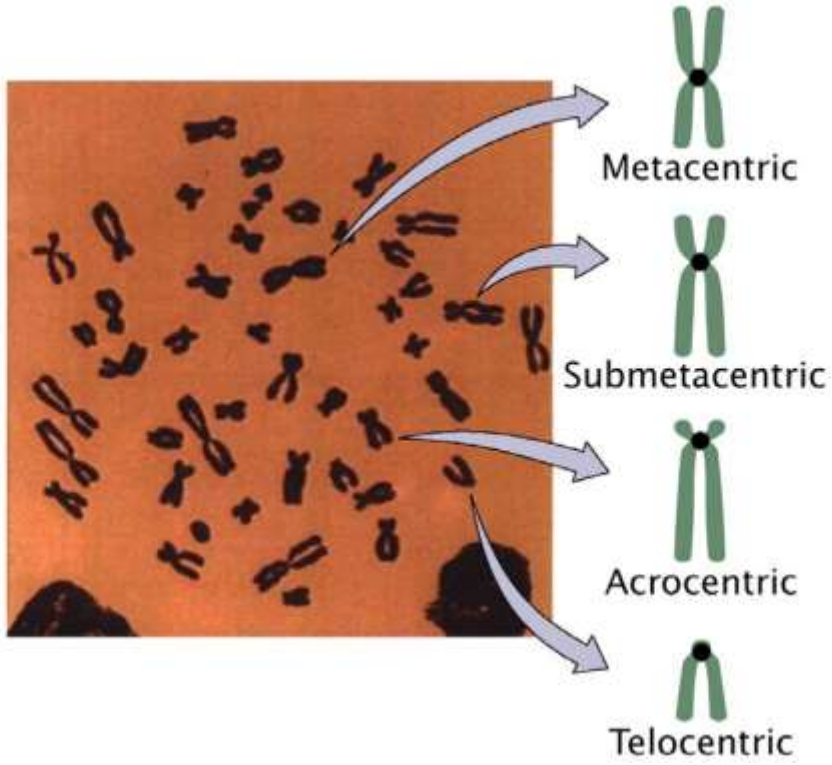
There are four types of chromosomes based upon the position of the centromere **(Figure 2)**.

1) **Metacentric**: In this type of chromosome the centromere occurs in the centre and all the four chromatids are of equal length.

2) **Submetacentric**: In this type of chromosome the centromere is a little away from the centre and therefore chromatids of one side are slightly longer than the other side.

3) **Acrocentric**: In this type of chromosome the centromere is located closer to one end of chromatid therefore the chromatids on opposite side are very long. A small round structure, attached by a very thin thread is observed on the side of shorter chromatid. The small round structure that is a part of the chromatid is termed as satellite. The thin strands at the satellite region are termed as Nucleolar Organiser Region.

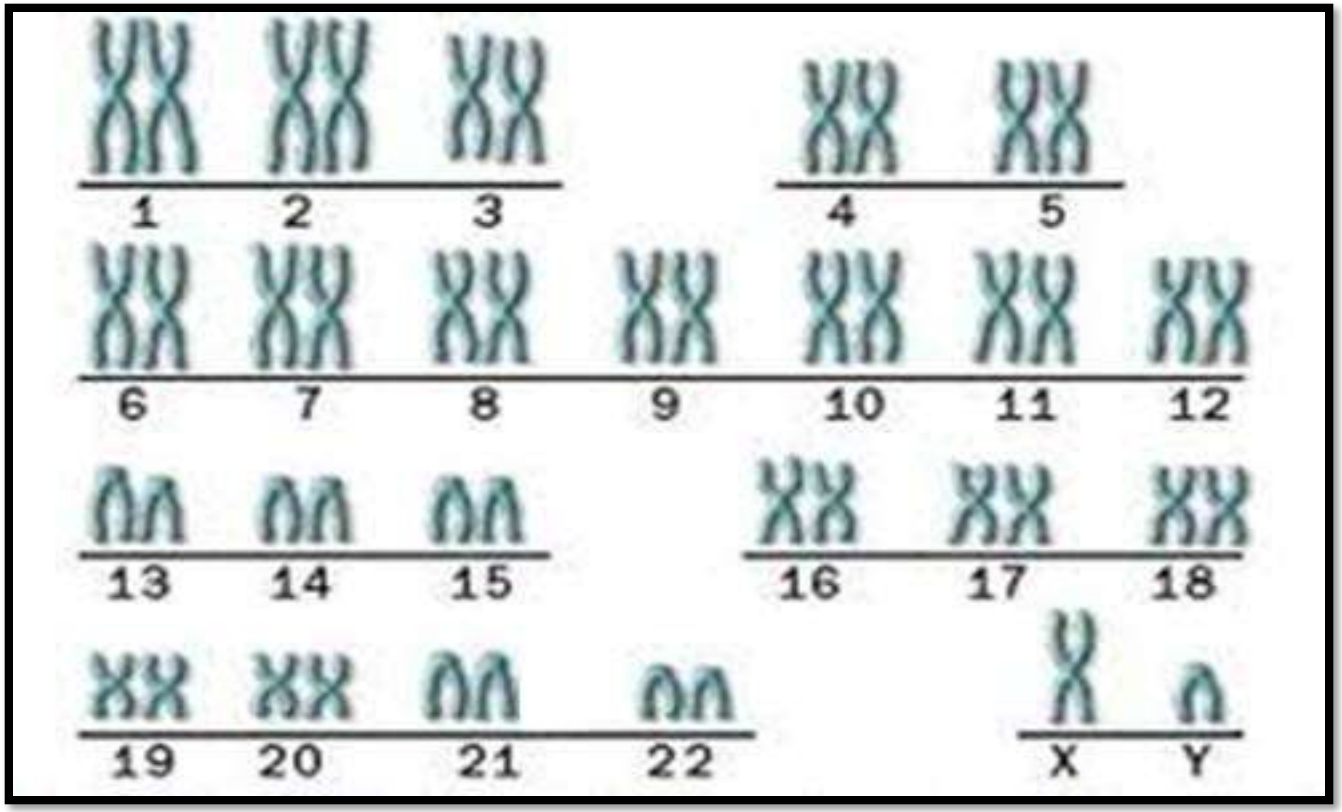
4) **Telocentric**: In this type of chromosome the centromere is placed at one end of the chromatid and hence only one arm. Such telocentric chromosomes are not seen in human cells.



Human Chromosome Karyotype

Eukaryotic species have several chromosomes and are detected only during mitosis or meiosis. They are best observed during the metaphase stage of cell division as they are found in the most condensed state. Thus each eukaryotic species is characterized by a **karyotype** which is the numerical description (number and size) of chromosomes in the normal diploid cell. For example, the *Homo sapiens* possess 46 chromosome i,e., 23 pairs **(Figure 3)**.

The karyotype is important because genetic research can correlate changes in the karyotype with changes in the phenotype of the individual. For example, Down's syndrome is caused by duplication of the human chromosome number 21. Insertions, deletions and changes in chromosome number can be detected by the skilled cytogeneticist, but correlating these with specific phenotypes is difficult.


1 2 3
4 5
6 7 8 9 10 11 12
13 14 15
16 17 18
19 20 21 22
X Y

Giant chromosomes:

Some cells at certain particular stage of their life cycle contain large nuclei with giant or
large sized chromosomes. Polytene and lampbrush chromosomes are examples of giant chromosomes.

Polytene Chromosome

Giant chromosomes were first time observed by E.G. Balbiani in the year 1881 in nuclei
of certain secretory cells (salivary glands) of Chironomas larvae (Diptera). However he could not conclude them to be chromosomes. They were conclusively reported for the first time in insect cells (*Drosophila*) by Theophilus Painter of the University of Texas in the year 1933. Since they were discovered in the salivary glands of insects they were termed as salivary gland chromosomes. The anme polytene chromosome was proposed by Kollar due to the occurrence of many chromonemata (DNA) in them.

Cells in the larval salivary gland of *Drosophila*, mosquito *and Chironema* contain chromosomes with high DNA content. However they may also occur in malphigian tubules, rectum, gut, foot pads, fat bodies, ovarian nurse cells etc. Polyteney of giant chromosomes happens by replication of the chromosomal DNA several times without nuclear division (endomitosis) and the resulting daughter chromatids do not separate but remain aligned side by side. During endomitosis the nuclear envelope does not rupture and no spindle formation takes place. The polytene chromosomes are visible during interphase and prophase of mitosis.

They are about 100 times thicker contain 1000 to 2000 chromosomes, than the chromosomes found in most other cells of the organism. When stained and viewed under compound microscope at 40X magnification they display about 5000 bands. In them the chromomere or the more tightly coiled regions alternate with regions where the DNA fibres are folded loosely. A series of dark transverse bands alternates with clear zones of inter bands.

Such individual bands can be correlated with particular genes (Figure 1). About 85% of the DNA in polytene chromosomes is in bands and rest 15% is in inter bands. The cross banding pattern of each polytene chromosome is a constant characteristic within a species and helps in chromosome mapping during cytogenetic studies. In *Drosophila melanogaster* there are about 5000 bands and 5000 interbands per genome. These chromosomes are not inert cellular objects but dynamic structures in

which certain regions become “puffed out” due to active DNA transcription at particular stages of development. These chromosome puffs are also termed Balbiani rings. Puffs may apprear and disappear depending on the production of specific proteins which needs to be secreted in large amounts in the larval saliva. Another peculiarity of the polytene chromosomes is that the paternal and maternal chromosomes remain associated side by side and the phenomenon is termed somatic pairing.

Both polyteney and polyploidy have excess DNA per nucleus, but in the later the new chromosomes are separate from each other. A polytene chromosome of *Drosophila* salivary glands has about 100 DNA molecules which are arranged side by side and which arise from 10 rounds of DNA replication (2_{10}=1024). *Chironimus* has 16000 DNA molecules in their polytene chromosomes.

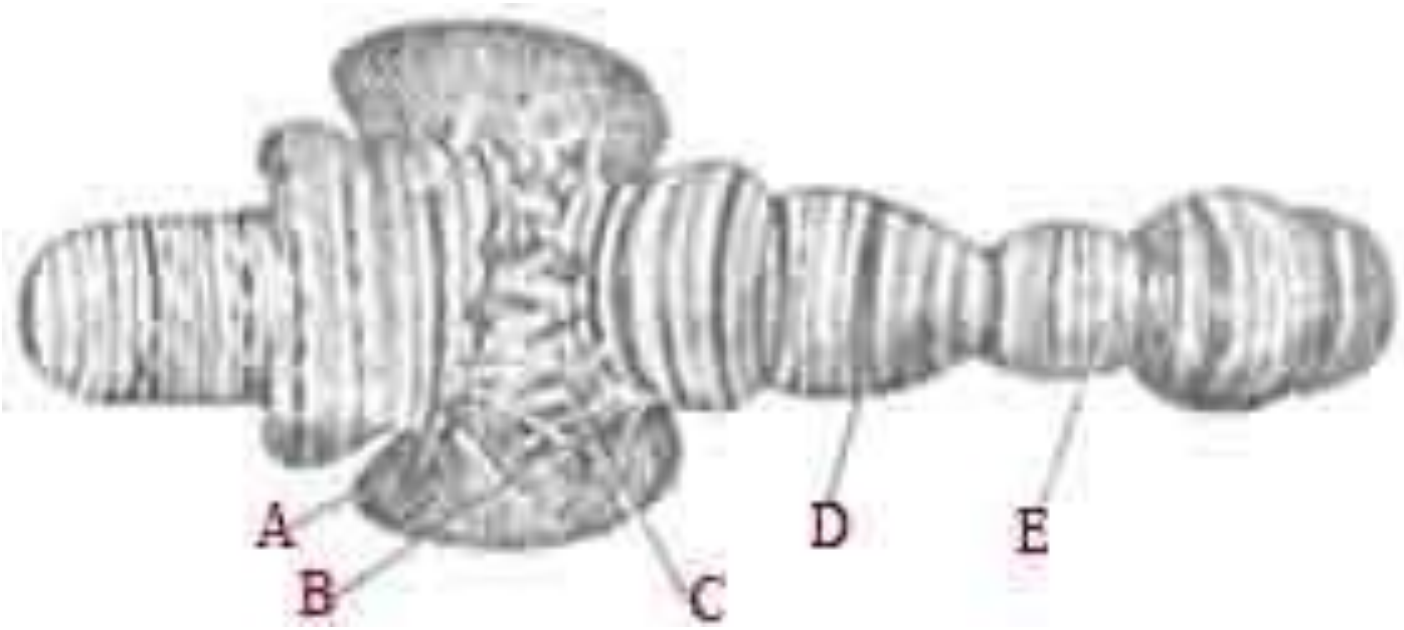


Figure 1: The structure of Drosophilla polytene chromosome. A: mRNA; B-Chromosome puff; C: Chromonemata; D: Dark band; E: Interband. The figure has been adapted from the site http://www.microbiologyprocedure.com/genetics/chromosomes/special-types-of-chromosomes.htm.

Lampbrush chromosome

Lampbrush chromosomes were first observed by Flemming in 1882 in sections of
Salamander oocytes and later described by Ruckert in the year 1892. They appeared like brushes used for cleaning lamps, hence the name lampbrush chromosome. They are transitory structures and can be observed during the diplotene stage of prophase I in meiosis in the oocytes of all animal species both vertebrates and invertebrates. They have been described in Sepia (Mollusca), Echinaster (Echinodermata) and in several species of insects, shark, amphibians, reptiles, birds and mammals (humans). Lampbrush chromosomes have also been found in spermatocytes of several species, giant nucleus of Acetabularia and even in plants. Generally they are smaller in invertebrates than vertebrates. They are observed in oocytes because oocytes are high in DNA content.

Lampbrush chromosomes are functional for studying chromosome organization and genome function during meiotic prophase. Additionally lampbrush chromosomes are widely used for construction of detail cytological maps of individual chromosomes.

They are of exceptionally large sizes and present in bivalent form. They are formed due to the active synthesis of mRNA molecules for future use by the egg cells, when no synthesis of mRNA molecule is possible during the mitotic cell division. Lampbrush chromosomes are clearly visible in the light microscope they are organized into a series of chromomeres with large chromatin symmetrical loops extending laterally **(Figure 2)**.

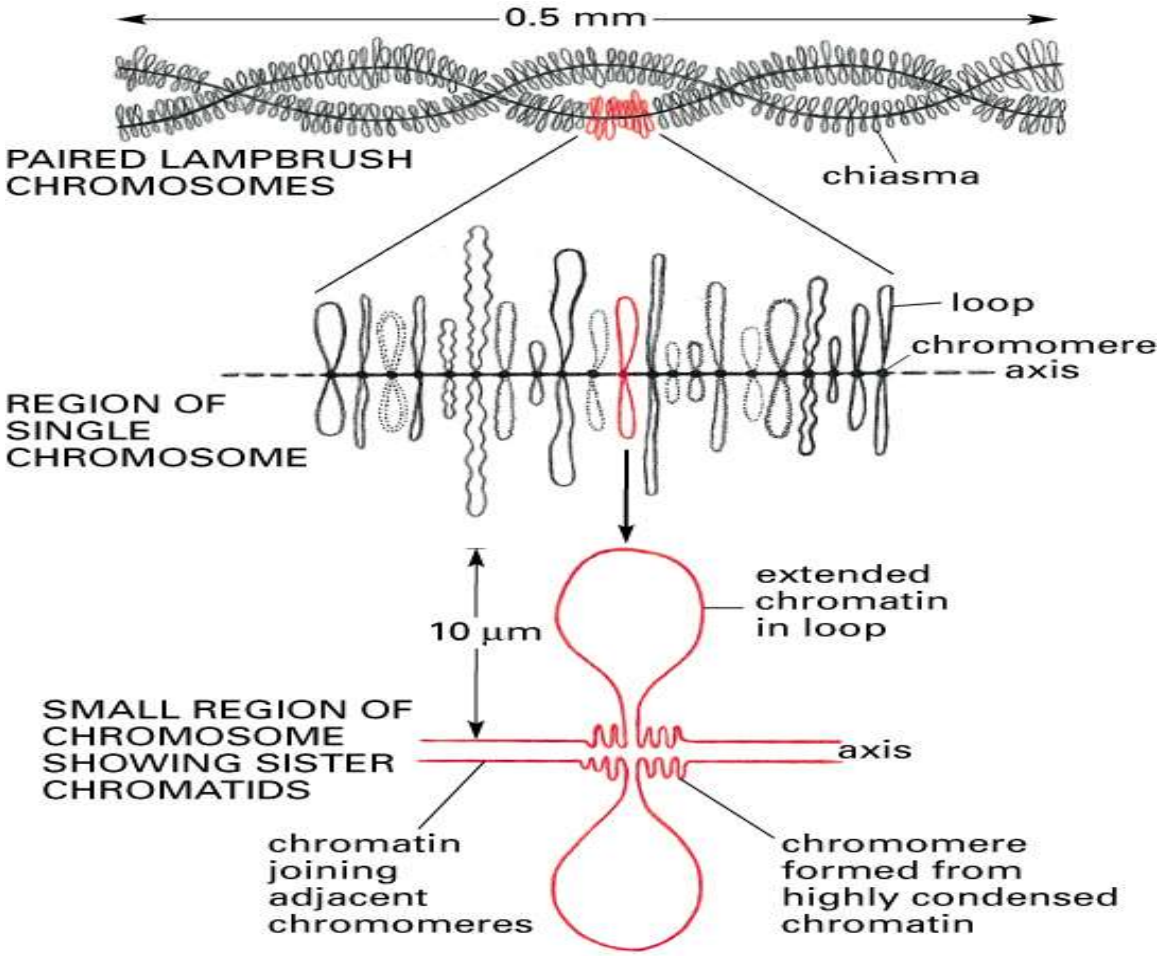


Figure 2: Lampbrush chromosome. This figure has been adapted from the molecular biology of the cell, by Bruce Alberts, 4th Ed. 2008.

Each loop appears at a constant position in the chromosome (10,000 loops per chromosome set or haploid set). Each loop has an axis made up of DNA unfolded from the chromosome and is transcriptionally highly active. Wherein several transcription units with polarized RNP-matrix coats the DNA axis of the loop. The majority of the DNA, however, is not in loops but remains highly condensed in the chromomeres on the axis and lacks expression of genes.

The loops perform intense transcription of heterogenous RNA (precursors of mRNA molecules for ribosomal and histone proteins). Thus each lateral loop is covered by an assymetrical matrix of RNA transcripts; thicker at one end of the loop than other. The number of pairs of loops gradually increases during meiosis till it reaches maximum at diplotene.

This stage may persist for months or years as oocytes build up supply of mRNA required for further development. As meiosis proceeds further number of loops gradually decrease and loops ultimately disappear due to reabsorption into the chromosome or disintegration.

Certain hypothesis regarding loops are that they may be static or dynamic with new loop material spinning out of one side of a chromosome and returning to a condensed on the other side. This is called spinning out or retraction hypothesis. This hypothesis has been rejected recently through DNA-RNA hybridization studies. The other hypothesis is known as the Master and Slave hypothesis which suggested that each loop pairs and thus chromomere is associated with the activity of many copies of specific genes.

There is a master copy at each chromomere and information is transferred to the slave copies which are matched against it to ensure that all are identical. The master copy does not take part in RNA synthesis, but the slave copy is involved in transcription. Large number of duplicate genes ensures higher level of transcription.

Question