

CHROMOSOMES

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Etymology

- Originated from the **Greek** word (*chroma*, colour) and (*soma*, body).

DEFINITION

“A **chromosome** is an organized structure of DNA and protein found in cells. It is a single piece of coiled DNA containing many genes, regulatory and other nucleotide sequences. Chromosomes also contain DNA-bound proteins, which serve to package the DNA and control its functions”.

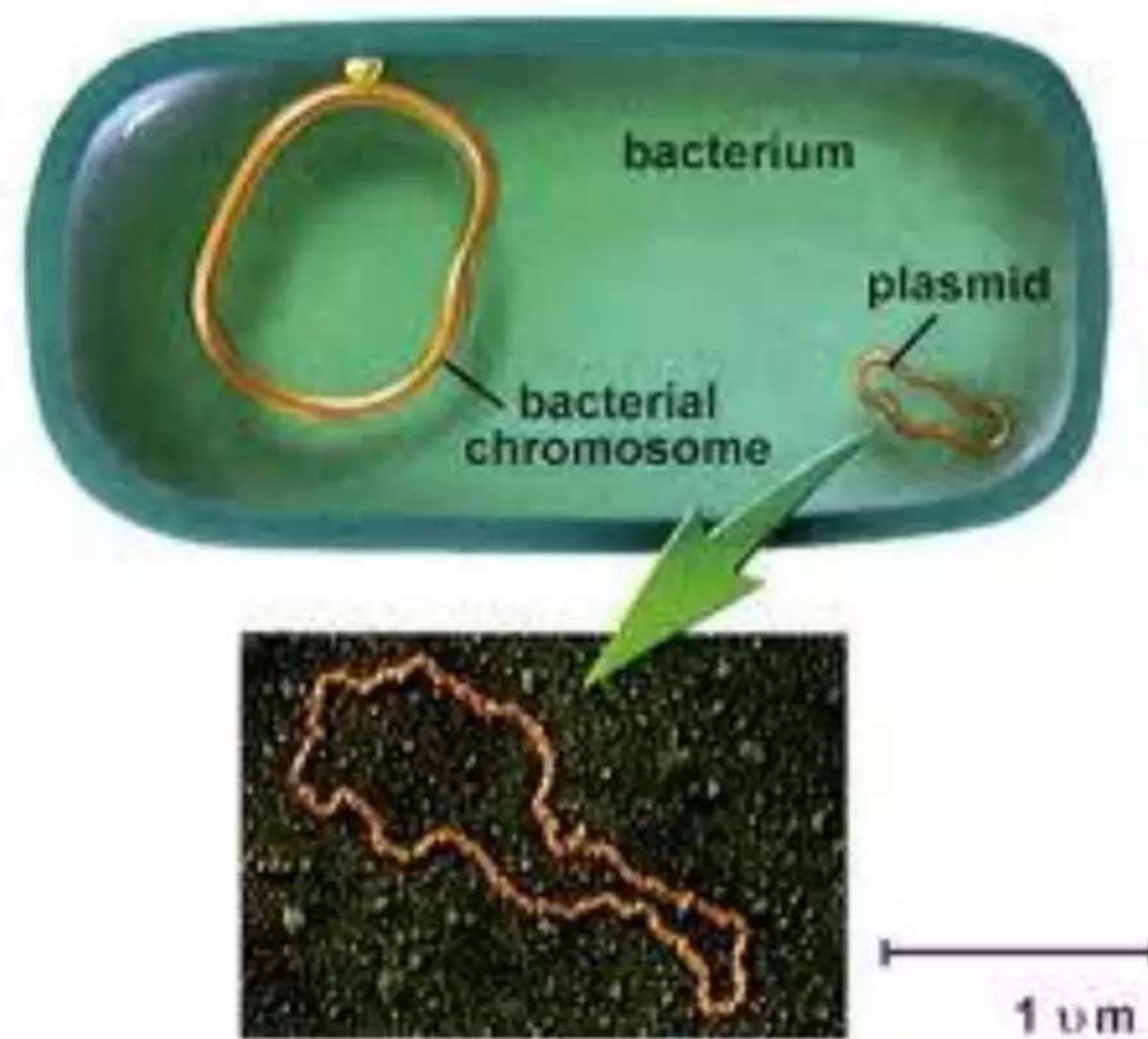
Discovery

- First described by **Strausberger** in **1875**.
- The term “**Chromosome**”, however was first used by **Waldeyer** in **1888**.

Prokaryotic & Eukaryotic Chromosomes

Prokaryotic Chromosomes

- Genomes are simple and the organization of DNA is also different.
- Genomes are contained in single chromosomes, which are usually circular DNA molecules.
- DNA is associated with proteins but no histones present.
- DNA packaging is different from and is not well understood.
- Variation in genome length bearing genes.
- **RNA viruses** having smallest genome.



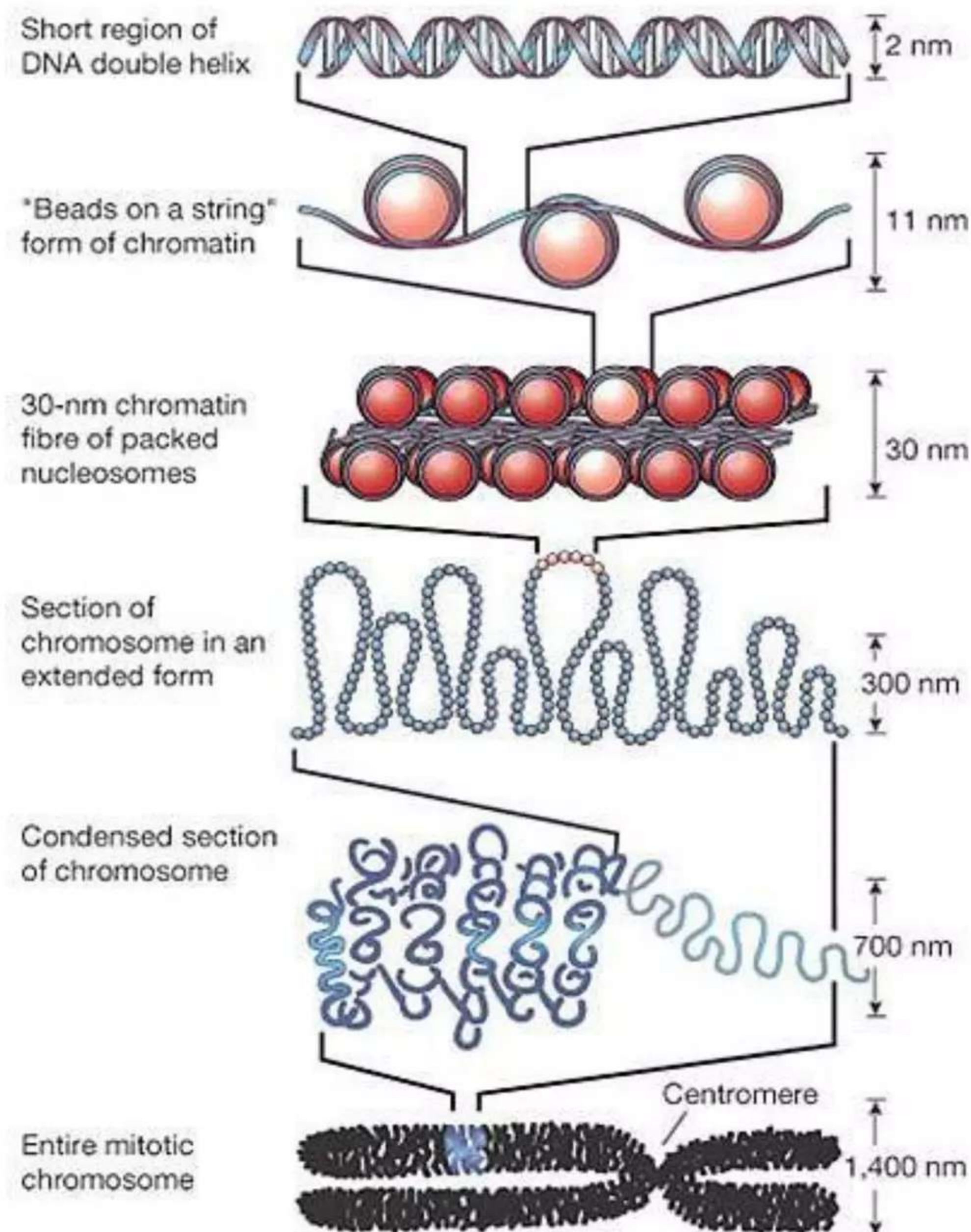
Eukaryotic Chromosomes

- Genomes are composed of multiple chromosomes, each containing a linear molecule of DNA.
- **Histones** are unique feature of eukaryotic cells.
- The packaging of DNA is due to histones.

Chemical Composition

- Composed of thin chromatin threads called **Chromatin Fibers**.
- Interphase chromatin consists of about **30 - 40%** DNA, **50 - 65%** protein and **1-10%** RNA.
- Metaphase chromosomes contain **15 - 20%** DNA, **10 - 15%** RNA and **65 - 75 %** protein.
- DNA consists of nucleotides while the primary protein component is histones.

- Histones strongly attracted to the negatively charged phosphate groups of the DNA.
- There are 5 major types of Histones.
- Thousand of other types of proteins involved in DNA replication and gene expression.
- The degree to which DNA is condensed is expressed as its packing ratio.
- **Packing ratio** - the length of DNA divided by the length into which it is packaged.
- To achieve the overall packing ratio, DNA is not packaged directly into final structure of chromatin.
- Chromatin is composed of repeating 200 base pair unit, which were called **nucleosome**.
- **Nucleosome** was described by **Roger Kornberg** in **1974**.
- **146** base pairs of DNA wrapped **1.75** times around a histone core.
- One molecule **H1** is bound to the DNA as it enters and exits each nucleosome core particle.
- This forms a chromatin subunit known as **Chromatosome**; consist of **166** base pairs of DNA.
- The histones cores act as magnetic forms for the coiling of DNA.
- Nucleosomes wraps up into higher order coils called super coils.



Structure of Chromosome

- Typically a chromosome is made of two chromatids, a centromere and a secondary constriction.
- **Sister chromatids** are two identical copies of the chromosome connected by a centromere.
- The two chromatids of one homologous chromosome with respect to those of the other homologue are called **Nonsister chromatids**.

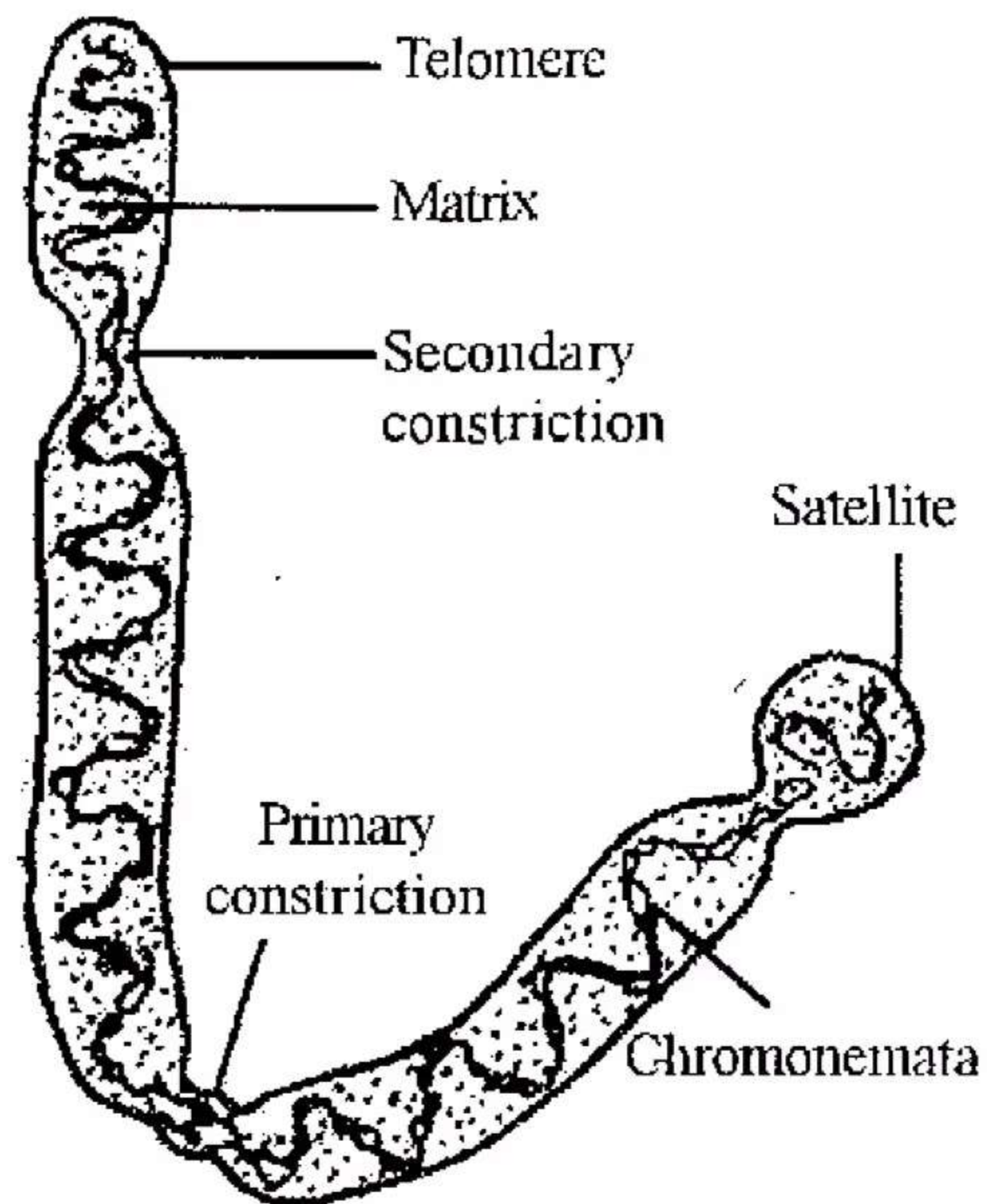
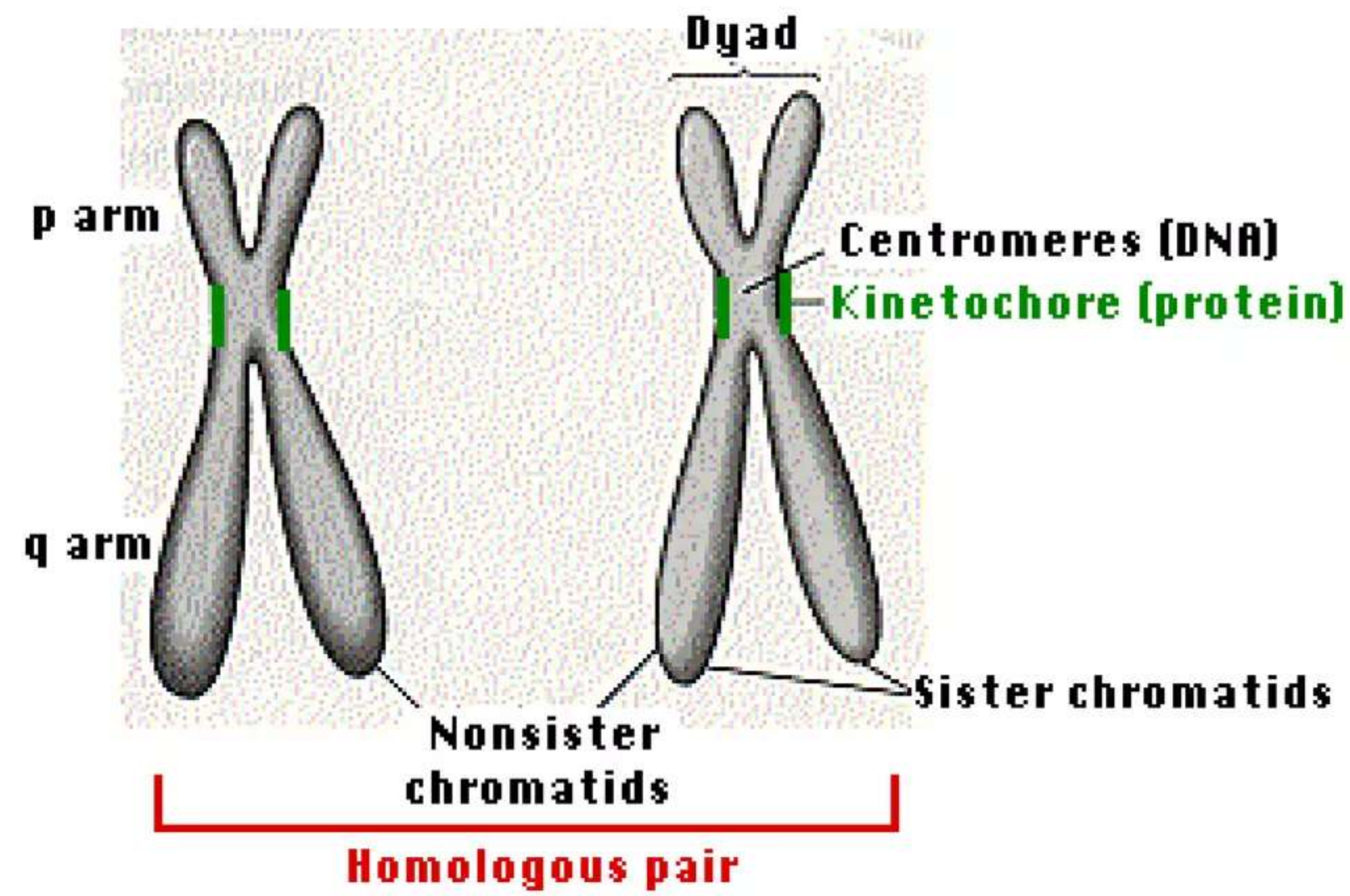


Fig. 3.1 Structure of chromosome

Centromere

- The region where two sister chromatids of a chromosome appear to be joined during cell division is called **Centromere**.
- Also termed as **Primary constriction**.
- **Darkly-stained** region.
- In humans, the centromere contains 1–10 million base pairs of DNA.

- First part of chromosomes to be seen moving towards the opposite poles during anaphase.

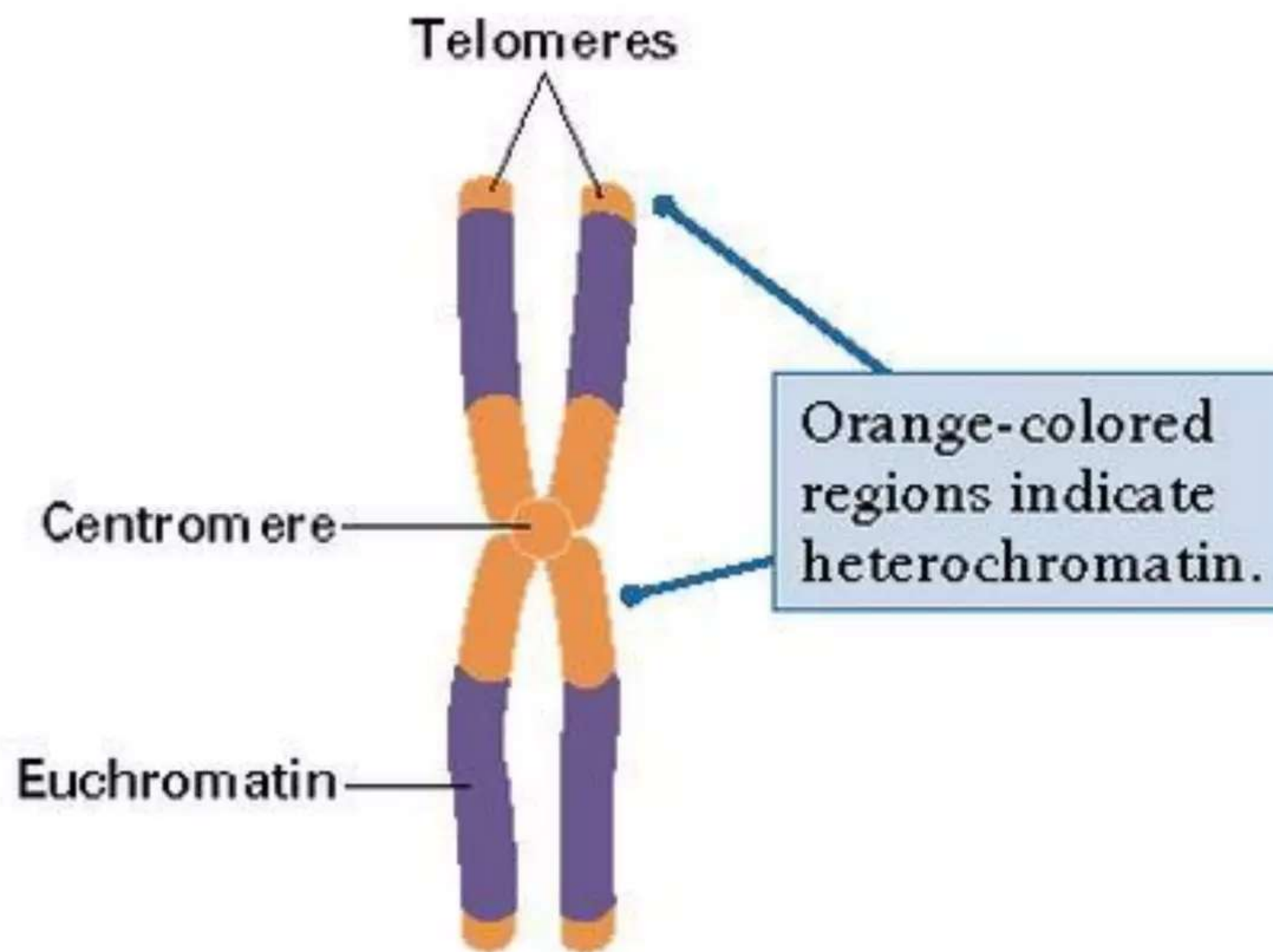
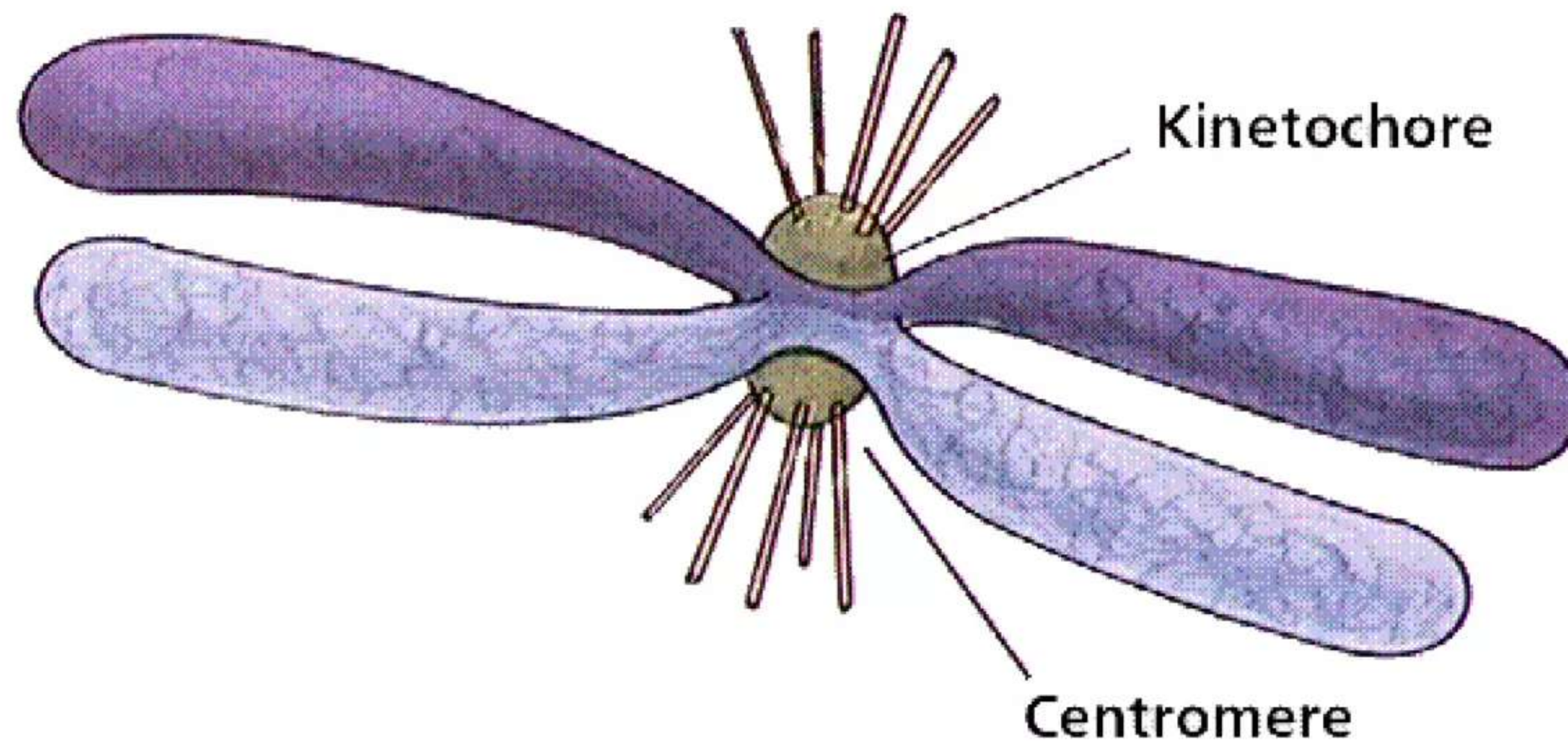
Kinetochores

- The **kinetochore** is the protein structure on chromosomes where the spindle fibers attach during cell division to pull the chromosomes apart.
- DNA sequence within these regions is called **CEN DNA** of about **120 bp**.
- Sub-domains, **CDE-I**, **CDE-II** and **CDE-III**.
- A complex of three proteins called **Cbf-III** binds to normal CDE-III regions but can not bind to a CDE-III region with a point mutation.

Telomeres

- Derived from the Greek nouns **telos** "end" and **meros** "part"
- A **telomere** is a region of repetitive DNA sequence at the end of a chromosome, which protects the end of the chromosome from deterioration or from fusion with neighbouring chromosomes.
- Required for the replication and stability of the chromosome.
- **McClintock** noticed that if two chromosomes were broken in a cell, the end of one could attach to the other and vice versa.

Species	Repeated Sequence
<i>Arabidopsis</i>	TTTAGGG
Human	TTAGGG
Slime Mold	TAGGG



Regions of Chromosomes

- Several different chemical techniques are used to identify certain chromosomal regions by staining them so that they form **chromosomal bands**.
- There are 2 types of regions in chromosomes.

Euchromatin

- Light staining regions.

- Lightly packed form of chromatin and is often under active transcription.
- Found in both cells with nuclei and cells without nuclei.

Heterochromatin

- Darkly staining and tightly coiled throughout the cell cycle and that is, for the most part, genetically inactive.

Constitutive Heterochromatin

- It is fixed and irreversible in form and function.
- It does not reverse to the euchromatic stage.
- Chromosomes 1, 9, 16 and the Y chromosome contain regions of constitutive heterochromatin.

Facultative Heterochromatin

- It has the faculty to return to the normal euchromatic state.
- Consists of euchromatin that takes on the staining and compactness characteristics of heterochromatin during some phase of development.
- The inactive X chromosome is made up of facultative heterochromatin.

Types of Chromosomes

Autosomes & Allosomes

Autosomes

- Chromosomes that are not directly concerned with reproduction and sex determination are called autosomes.
- These are identical in both the two sexes in man.

- They have loci occupied by autosomal genes.
- The term "**autosome**" was coined by **T.H. Montgomery** in **1904**.

Allosomes/Heterosome

- These chromosomes are directly associated with reproduction and differ from autosomes in size, form and behaviour.
- Usually there is a single pair of allosomes in mammals termed as 'X' and 'Y' chromosomes.
- In bugs of Heteroptera like locusts, the female has two X chromosomes while the male has one X. The Y chromosome is absent in these species.

Types of Chromosomes Based on the position of Centromere

1. Metacentric Chromosomes

- The two arms are equal in length.
- Appears in 'V' shape.

2. Submetacentric Chromosomes

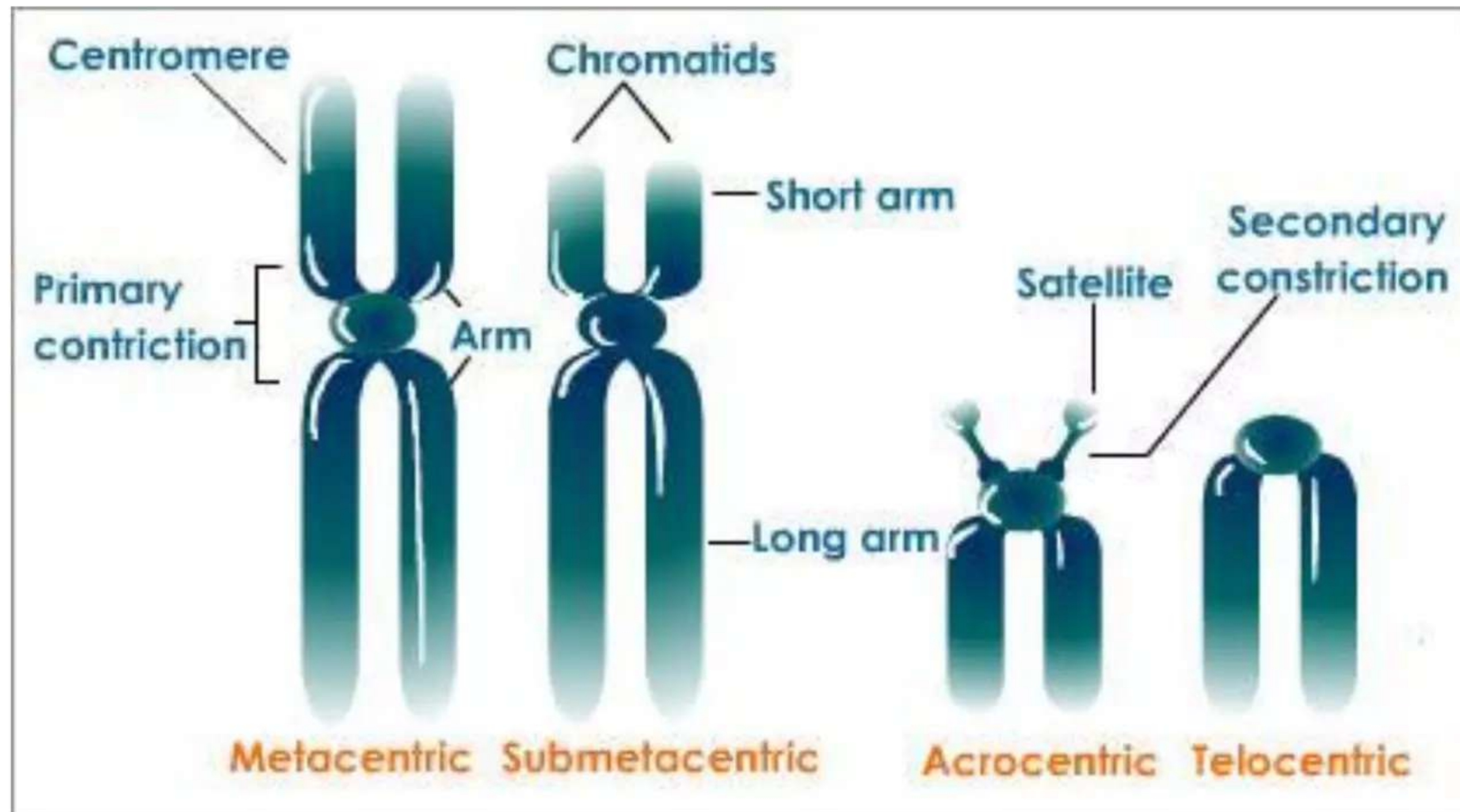
- Also called as **Heterobrachal**.
- Chromatids of one side are slightly longer than the other side.
- Resemble the letter 'L'.

3. Acrocentric Chromosomes

- Centromere is located closer to one end of chromatid.
- The small round structure is termed as **satellite**.
- Resemble the letter 'j'.

4. Telocentric Chromosomes

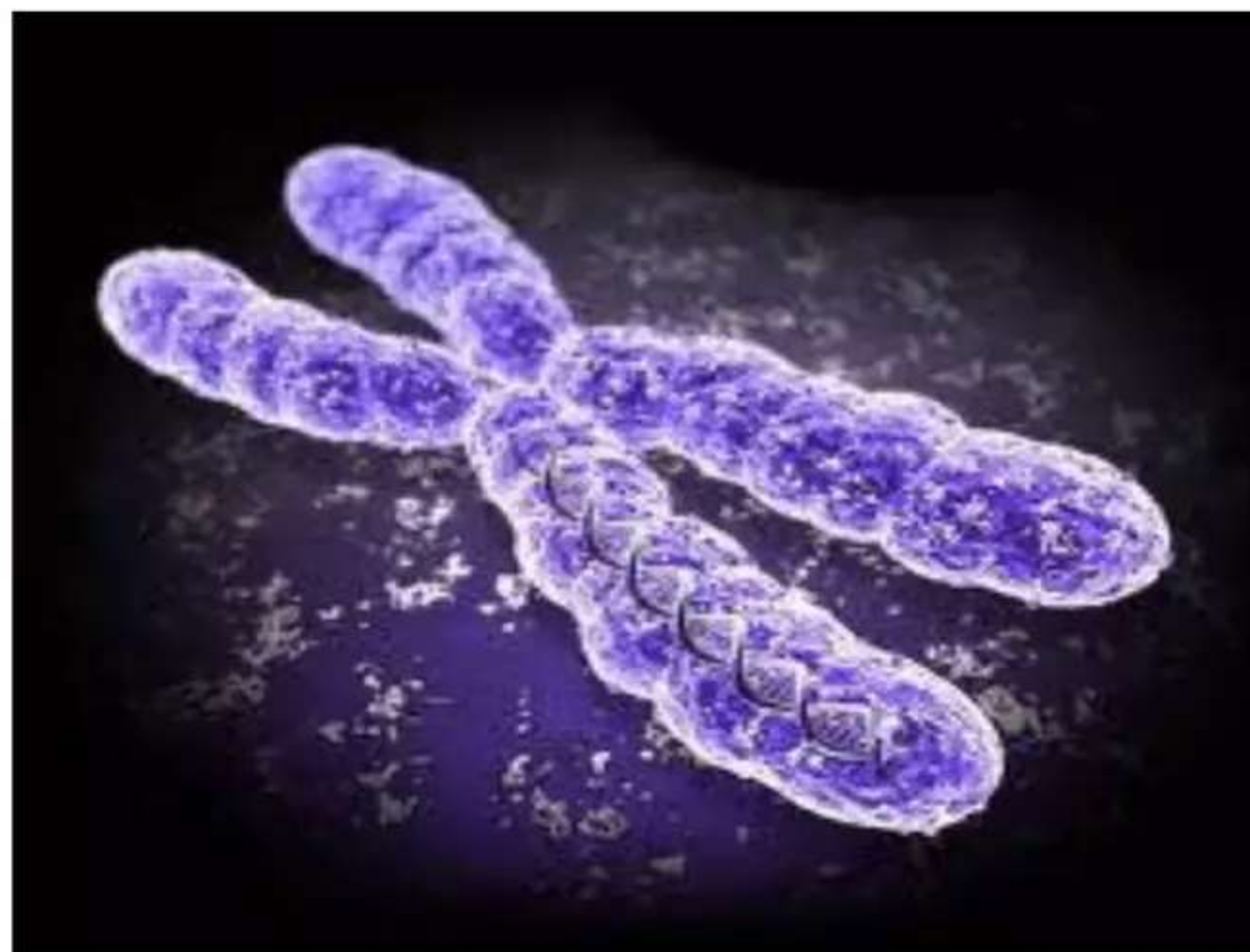
- Also called as **monarchial** type.
- Centromere at the end of chromosomes.
- Not seen in human cells.
- Resemble the letter 'i'.



Types of Chromosomes Based on the number of Centromere

Based on the number of Centromere, chromosomes are grouped into five types.

- Chromosomes without centromere are called **Acentric Chromosomes**.

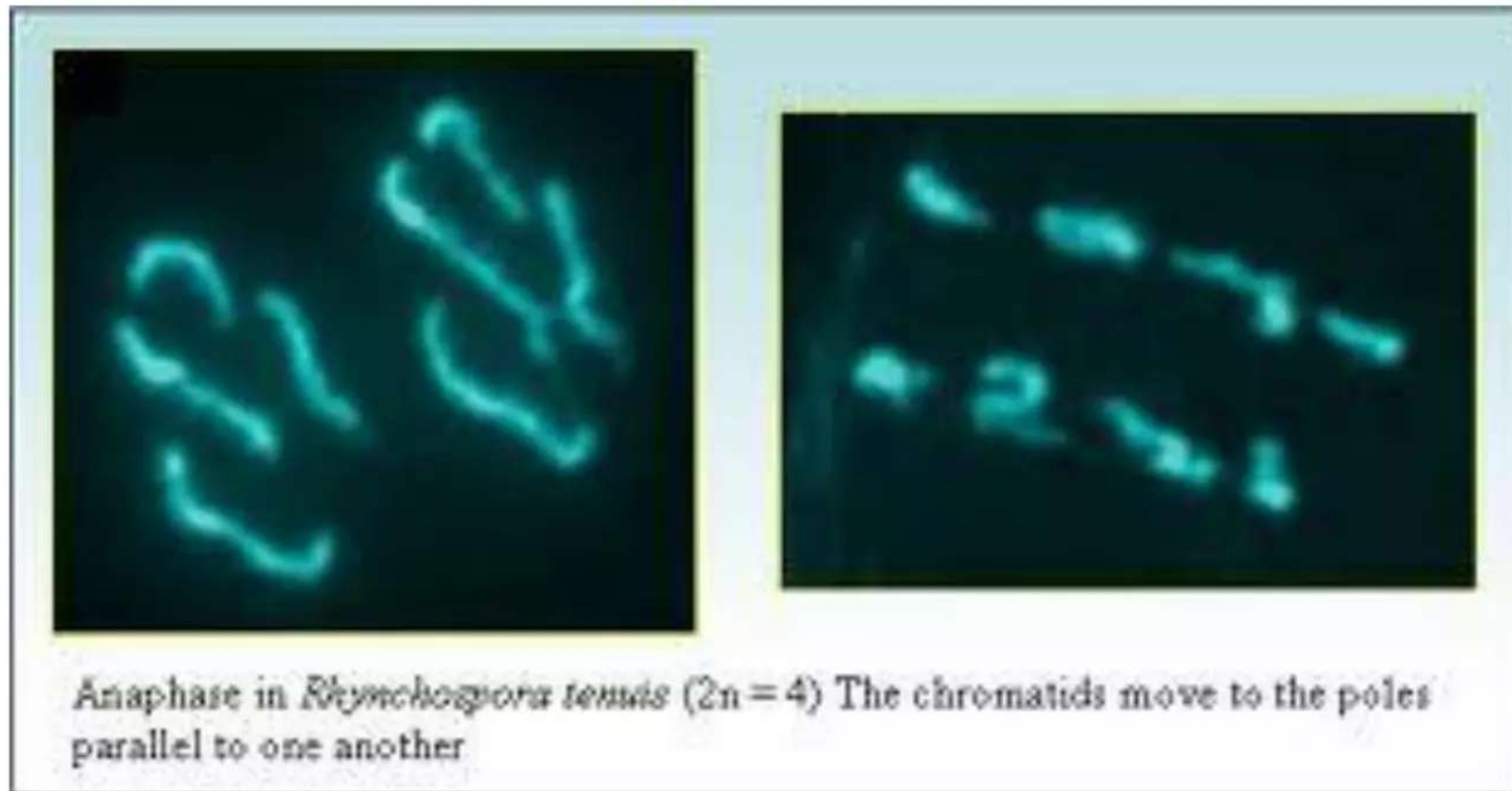


- Chromosomes with one centromere and called **Monocentric Chromosomes.**
- Chromosomes having two centromeres are called **Dicentric Chromosomes.**
- Chromosomes having more than two centromeres are called **Polycentric Chromosomes.**

Chromosomes Diffuse

- Also known as **Holocentric Chromosomes.**
- The entire length of the chromosome acts as the centromere.

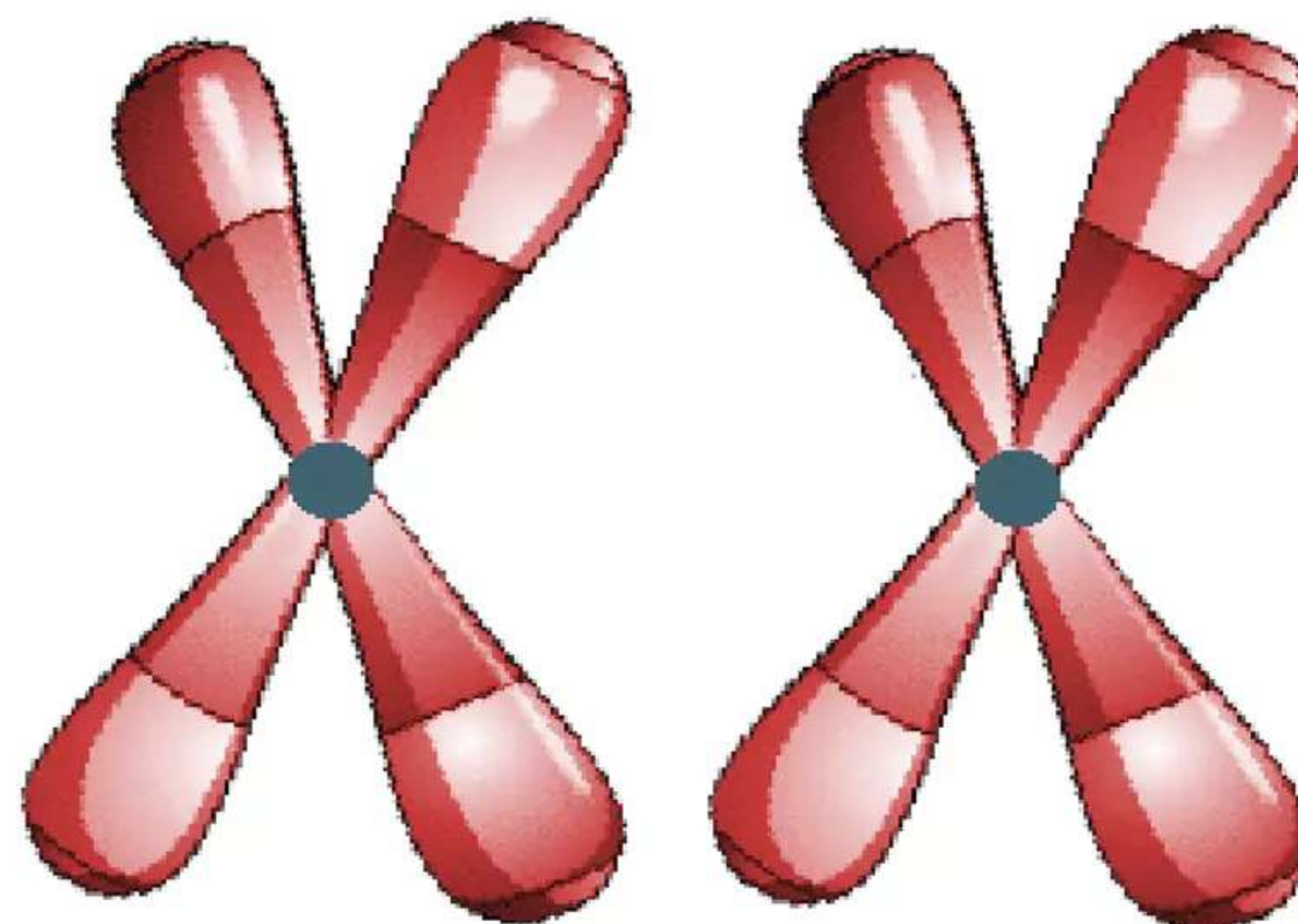




Homologous and Non Homologous Chromosomes

Homologous chromosomes

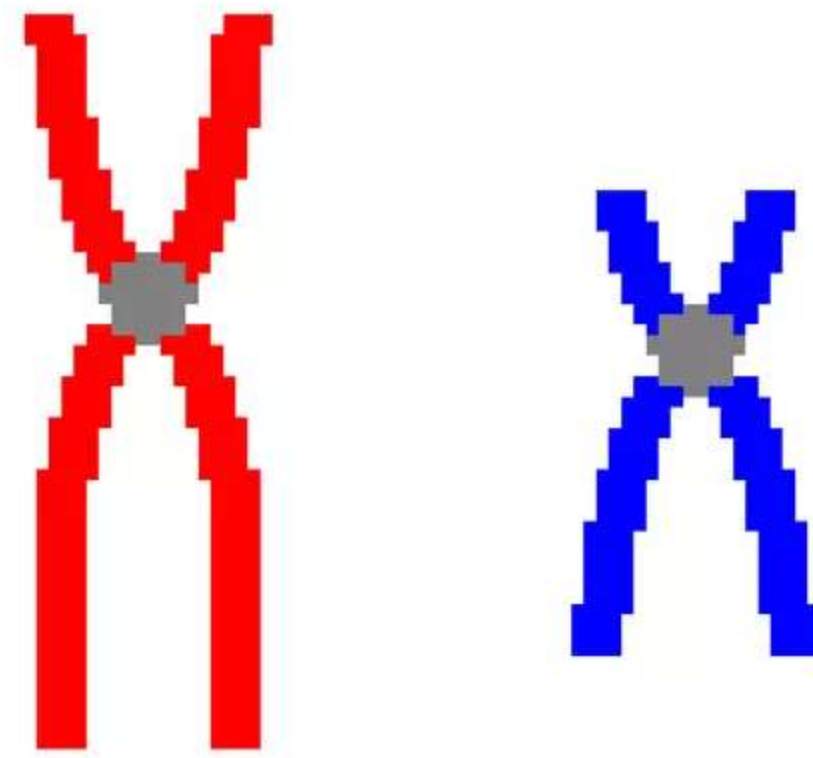
- Chromosome pairs of the same length, centromere position, and staining pattern, with genes for the same characteristics at corresponding loci.
- The pair (synapse) during meiosis.
- Each pair contains genes for the same biological features, such as eye color, at the same locations (loci) on the chromosome.



homologous pair

Non Homologous chromosomes

- Chromosomes that are not members of the same pair.
- Each chromosome of the pair is obtained from the each parent in diploids and contains all the gene pool of that organism.

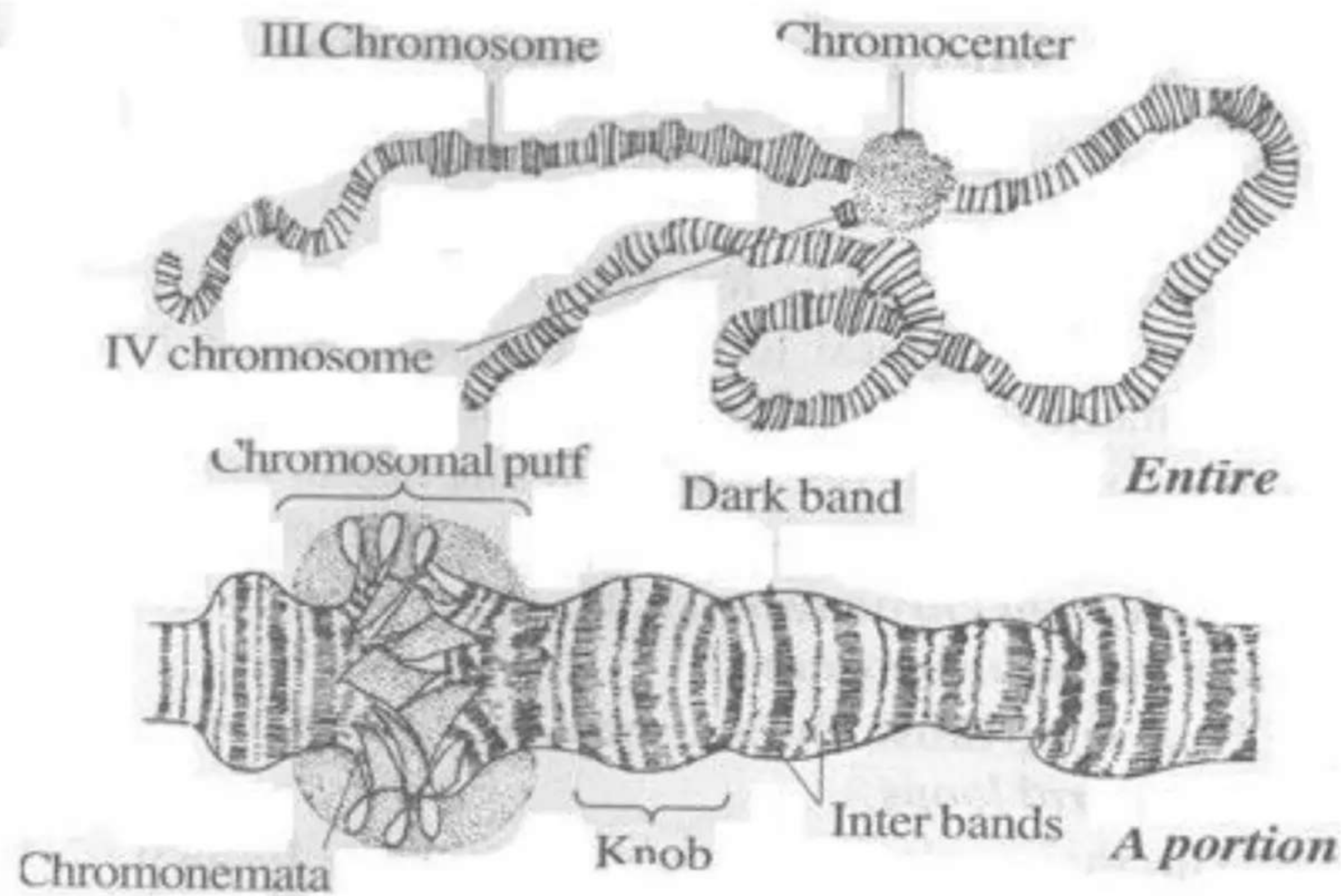


Special Types of Chromosomes

Polytene chromosomes

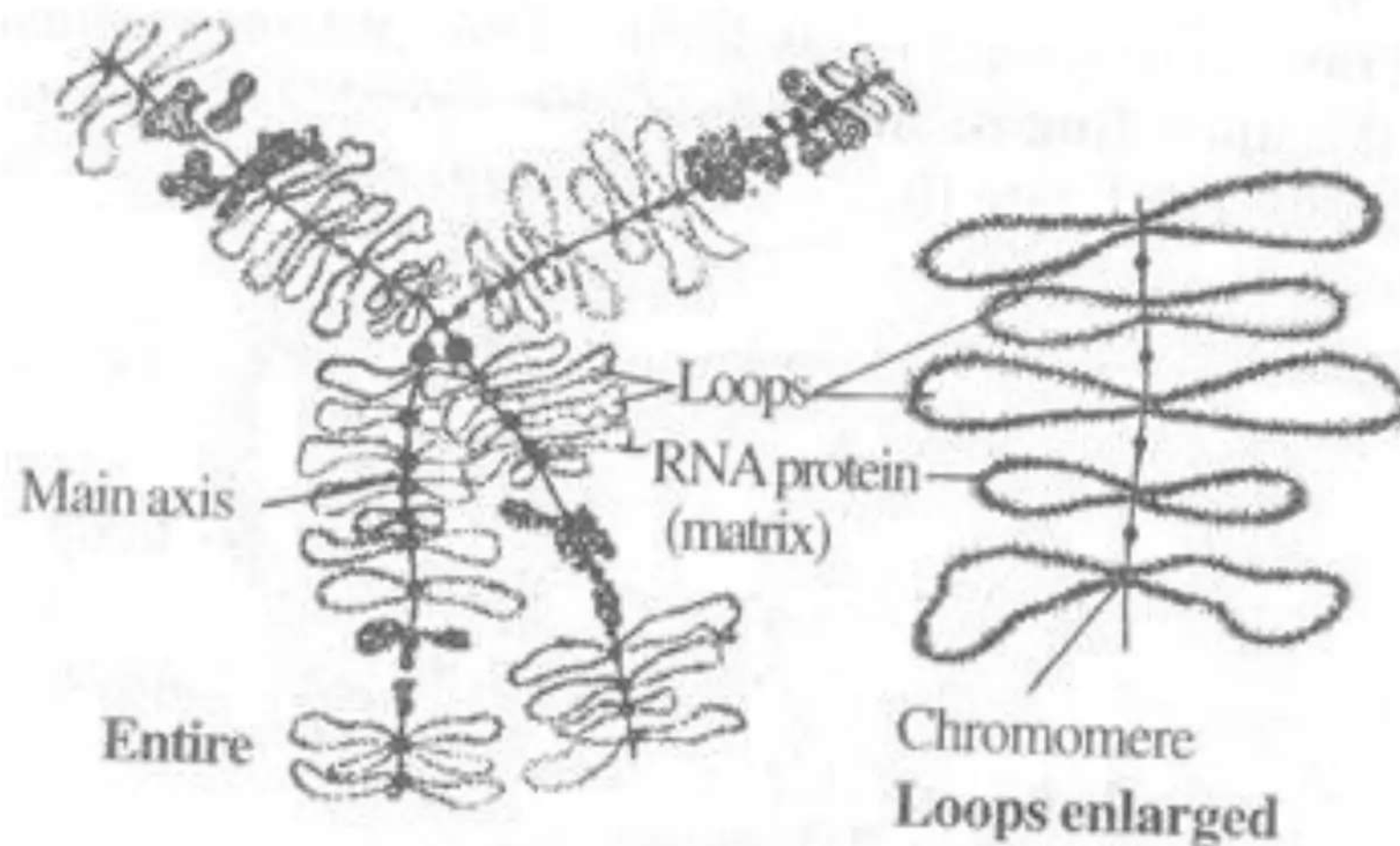
- The nuclei of the salivary gland cells of the larvae of dipterans like *Drosophila* have unusually long and wide chromosomes, 100 or 200 times in size of the chromosomes in meiosis and mitosis of the same species.
- Salivary gland cells do not divide after the glands are formed, yet their chromosomes replicate several times (a process called **endomitosis**) and become exceptionally giant-sized.
- They are discovered by **Balbiani (1881)** and named by **Koller**.
- The endomitosis process result in the production of 2X chromosomes, where X gives the number of multiplication cycle.

- They have alternating dark and light bands. The dark bands are disc-shaped structures occupying the whole diameter of chromosome. They contain euchromatin
- The light bands are fibrillar and composed of heterochromatin.



Lampbrush Chromosome

- Discovered by **Ruckert** in **1892**.
- Formed during the diplotene stage in the nuclei of oocytes during the active synthesis of mRNA molecules for the future use.
- It contains a main axis whose chromonemal fibres (DNA molecule) gives out lateral loops throughout its length.

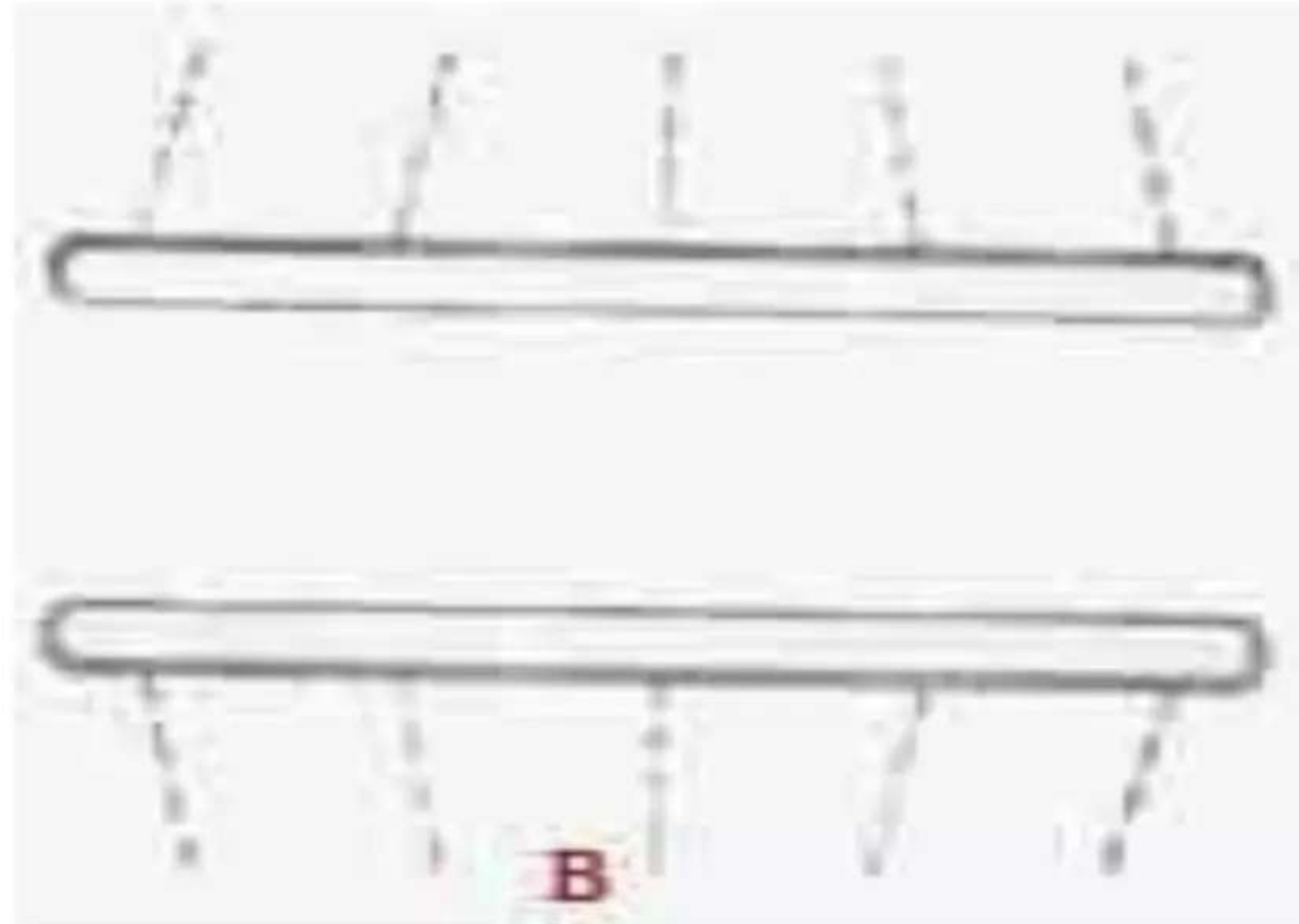


B Chromosomes

- Many organisms have a special chromosome in addition to the autosomes which are called B chromosomes.
- Also termed as **supernumery** or **accessory chromosomes** or **accessory fragments**.
- Smaller than autosomes and the number varies from 0 to 30 /cell.
- In some animals they may be derivatives of sex chromosomes.
- May have negative affects on the cell.

Holokinetic Chromosome

- In insects of the order Hemiptera and in some monocotyledonous plants the kinetic activity is distributed over the entire chromosome.
- The term **diffuse centromere** has been used as an alternative.
- In **1966 Flach** observed this type of centromere in some primitive Dicotyledons along with pseudoscorpion and *Ascaris*.



Number of Chromosomes

- Normally all the individuals of a species have the same number of chromosomes.
- Closely related species usually have similar chromosome number.
- Presence of a whole set of chromosomes is called **Euploidy**.
- It includes haploids, diploids, triploids, tetraploids etc.
- The condition in which the chromosomes sets are present in multiples of “n” is **Polyploidy**.

Chromosome Size

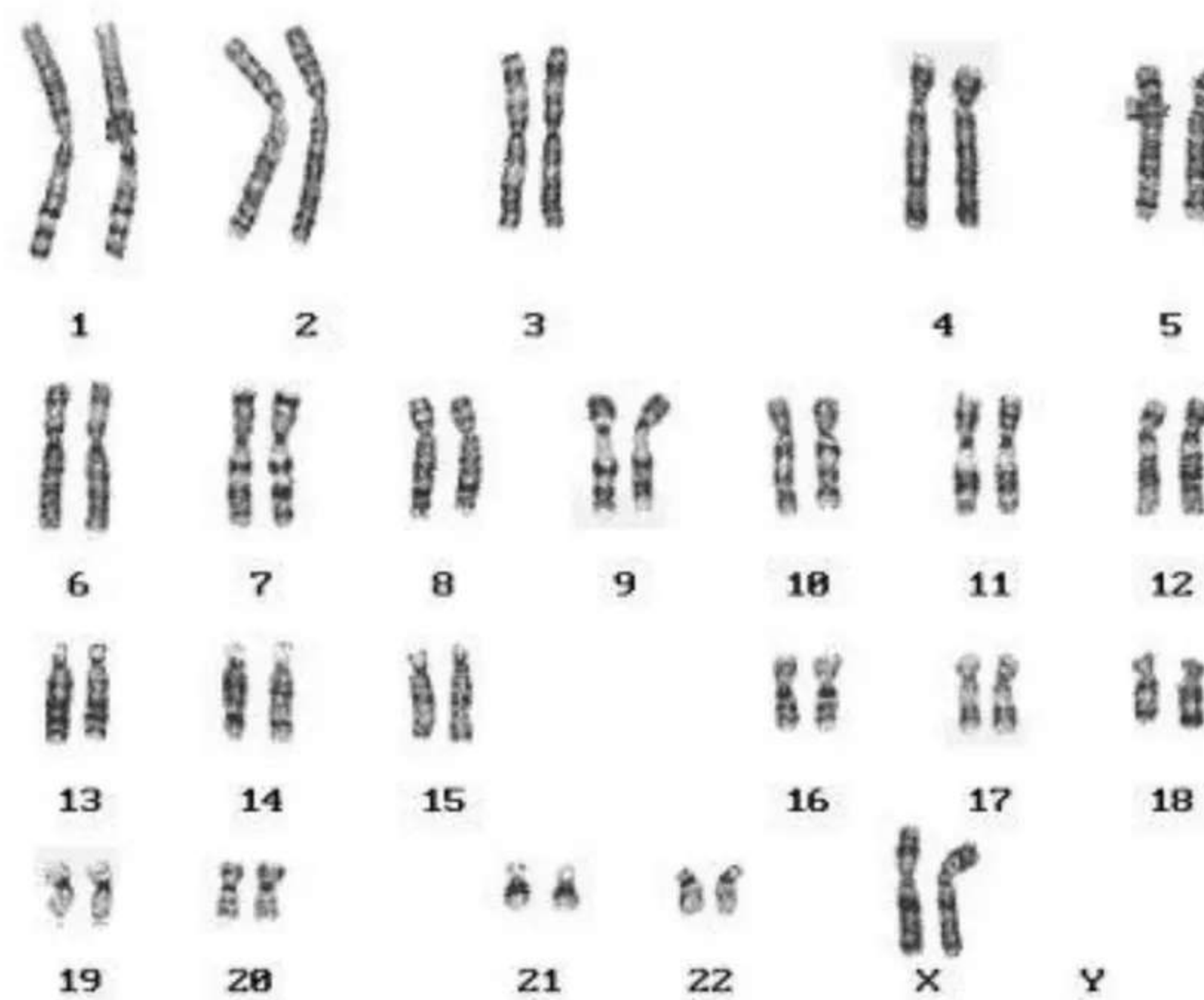
- The size of chromosomes shows a remarkable variation depending upon the stages of the cell division.
- Longest and thinnest in Interphase.
- Progressive decrease in their length with an increase in thickness in **prophase**.
- Most easily observed during **metaphase** when they are very thick, quite short and well spread in the cell.

- Chromosomes are smallest in **anaphase**.

Therefore, chromosomes measurements are generally taken during mitotic metaphase.

Karyotype

- A **karyotype** is the number and appearance of chromosomes in the nucleus of a eukaryotic cell.
- It describes the number of chromosomes, and what they look like under a light microscope.
- Attention is paid to their length, the position of the centromeres, banding pattern, any differences between the sex chromosomes, and any other physical characteristics.
- The study of whole sets of chromosomes is sometimes known as *karyology*.
- Karyotypes can be used for various purposes; such as, to study chromosomal aberrations, cellular functions, taxonomic relationships, and to gather information about past evolutionary events.



Karyotype of Human Female

Functions of Chromosomes

- In charge of all the processes.
- **“Packaging material”** that binds DNA and protein together.
- Protein synthesis steps are the responsibility of genes.
- Very important roles in the development of an individual.
- They are the '**vehicles of heredity**'.
- DNA provides the genetic information for various cellular functions essential for survival, growth, development etc.
- Chromosomes protect the genetic material (DNA) from being damaged during cell division.
- Essential for the process of cell division and are responsible for the replication, division and creation of daughter cells.
- Centromeres perform an important function in chromosome movement during cell division.