# CHROMOSOMES: STRUCTURE AND FUNCTION

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## Introduction:

Chromosomes are the carriers of heredity and may be considered as stainable thread like nuclear components having special organization, individuality and function. They were first demonstrated in eukaryotic cells by Strasburger (1875) and named as "Chromosomes" by Waldeyer (1888). Chromosomes are not visible in the interphase nucleus or metabolically active nucleus but appear as distinct entities during cell division.

Eukaryotic chromosomes which carry genes for somatic characteristics are called autosomes while those which control sex expression and differentiation are called sex chromosomes. Some other specialized chromosomes and supernumerary chromosomes are found in some eukaryotic cells/organisms.

## Chromosome Number:

(i) In higher organisms, each somatic cell has a diploid (2n) number of chromosomes, one set contributed each by male and female parents.

– Homologous chromosomes are two copies of a chromosome having identical gene loci and contributed one each by the male and female parents.

(ii) A genome is a set of chromosomes corresponding to the haploid set of a species.

(iii) Chromosome number varies widely in

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different plant and animal species.

- Smallest somatic chromosome number in the plant Kingdom: *Haplopappus gracilis* (Asteraceae) [2n = 4]
- Highest Chromosome Number in plants: *Ophioglossum reticulatum* [2n = 1260]

(iv) Somatic chromosome number generally remains constant among individuals of the same species.

- However, in some species somatic cells of the same individual exhibit variable diploid chromosome numbers (2n, 4n, 8n, etc.) due to endomitosis which leads to endopolyploid cells.

(v) In polyploid individuals, ancestral (basic) chromosome number is represented by x. For example, in common wheat (*Triticum aestivum*) 2n = 42, n = 21 and x = 7 showing that common wheat is a hexaploid (2n = 6x = 42).

## Chromosome size:

(i) Size of the mitotic metaphase chromosomes is taken to be the standard size.

(ii) This size varies in various plant and animal species.

Size range: Length : 0.5μ – 32μ
Diameter : 0.2μ – 3.0μ

(iii) Giant Chromosomes of dipteran salivary glands are permanently in metaphase stage and are easily visible in the interphase nucleus (Size: Length – 300μ; Diameter – 10μ)

(iv) Plant chromosomes are generally longer than those of animals.

- Longest metaphase chromosomes in plants are reported in *Trillium* (32μ long).

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(v) Size, shape and number of metaphase chromosomes constitute the Karyotype which is a distinctive attribute of each species.

— Asymmetrical karyotype refers to the chromosomes of varying sizes and centromeric positions in a set.

## Chromosome Morphology:

(i) Morphology of Chromosomes changes with the stage of cell division.

(ii) Mitotic metaphase is easily available by arresting the division cycle with chemical agents. So this is the most suitable stage for the study of chromosome morphology.

(iii) Chromosomes are normally measured at mitotic metaphase and Chromosome shape is usually observed at anaphase.

(iv) The sheath surrounding the chromosome is called pellicle which encloses an achromatic matrix.

— Both the matrix and the pellicle are non-genetic materials and appear only at metaphase when the nucleolus disappears.

(v) During prophase, the chromosomal material becomes visible as very slender coiled threads called chromonemata, which form the gene-bearing portions of the chromosomes.

(vi) Mitotic metaphase chromosomes are seen to be made up of two longitudinal halves called chromatids which remain connected together at the centromere which leaves a constriction called primary constriction.

(vii) While a chromatid is a half chromosome, the chromonemata are structures of sub-chromatid nature, and there can be more than one chromonemata in a chromatid.

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(viii) Corresponding to different positions of the centromere, chromosomes are called—

Acrocentric/Telocentric – Terminal centromere

Sub-metacentric – Sub-terminal centromere

Metacentric – Median centromere

Sub-telocentric – Centromere slightly inner from the telomere

Diffuse centrome – Centromeric property distributed throughout the length of the chromosome

(ix) Besides centromere, which produces a primary constriction in chromosomes, secondary constrictions can also be observed in some chromosomes.

– Such a secondary constriction, if present in the distal region of a chromosome arm, would pinch off a small fragment called satellite (= Trabant).

– The satellite remains attached to the rest of the body of the chromosome by a thread of chromatin.

– Secondary constrictions can be used as useful markers.

– Chromosomes having a satellite are marker chromosomes and are called SAT-chromosomes.

(x) Acentric chromosomes are of very rare occurrence.

– Dicentric chromosomes may be produced as a result of translocation, paracentric inversion, etc.

(xi) The chromosome extremities or terminal regions at either end of a chromosome are called telomeres.

– Chromosomes cannot fuse at the telomeric ends suggesting that a telomere has a polarity which prevents other chromosome segments from joining with it.

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- Detailed studies of telomeres at the molecular level have been made during some recent decades.

(xii) The term Karyotype represents a group of characteristics of somatic chromosomes of a species, viz., length of chromosomes, centromeric position, presence of secondary constriction, arm lengths, etc. This is usually depicted by a diagram in which chromosomes are arranged in a descending order of length (longest chromosomes on the left and smallest on the right) called idiogram.

- Symmetrical and asymmetrical karyotypes have been shown to represent evolutionary trends in angiosperms (Stebbins, 1971).

(xiii) Development of chromosome banding techniques during late 1960s and early 1970s proved very useful for the preparation of karyotypes in which linear differentiation due to distinct banding patterns allow identification of chromosomes with similar morphology.

- Banding technique allows detection of GC or AT rich regions or the regions with repetitive DNA.

## Euchromatin and Heterochromatin:

(i) On the basis of stainability with basic dyes (e.g., Acetocarmine, Feulgen, etc.) during various stages of the cell cycle, chromatin is subdivided into two main classes: Euchromatin and Heterochromatin.

(ii) Euchromatin region takes light stain in the interphase nucleus and comparatively deep stain during cell division.

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(iii) On the other hand, heterochromatin takes deep stain during interphase and prophase and light stain during metaphase.

(iv) Changes of stainability can be correlated with the changes of condensation and decondensation of chromosomes during various stages of cell division.

—Obviously, condensed stain of chromatin takes deeper stain while decondensed/extended form takes light stain.

(v) Heterochromatin has been found to be located at some specific chromosomal regions such as the centromere, chromomeres, nucleolar organizing region, satellite, etc.

(vi) Certain heterochromatic regions of chromosomes, particularly those proximal to centromeres are constant and are called <u>constitutive heterochromatic</u> regions serving as chromosome markers.

There are other heterochromatic regions called <u>facultative heterochromatic</u> and represented by whole sex chromosomes which become heterochromatic only at certain stages. In plants, <u>accesory chromosomes (B chromosomes)</u> in general and among dioecious genera like <u>Melandrium</u> and <u>Rumex</u>, one or both sex chromosomes may undergo partial or complete heterochromatization.

(vii) <u>DNA replication</u> in heterochromatic region occurs at a time different than that in euchromatic regions.

(viii) Genes in heterochromatic regions are inactive.

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## Ultrastructure of Chromosomes:

(i) Chromosome is a nucleoprotein complex. But how the DNA-protein complex builds up the chromosome structure has been variously interpreted by different workers. Initially, it was under speculation and several models were proposed from time to time to explain the DNA-protein association and its packing in the chromosome.

(ii) Two broad categories of models- single-stranded and multistranded have been postulated by various workers.

### (a) Single-stranded Models

1. Taylor (1957) proposed a single-stranded model and explained that chromosome is made of a long protein backbone from which DNA coils branch off like the legs of a centipede.

- Hence, this model was popularly known as Taylor's centipede model.

2. Taylor and Preese (1958) proposed that there are two protein spines instead of one.

- DNA chains stretch between them like a zigzag stair. In effect, DNA molecules are kept in position by the protein linkers.

3. Ris (1967) postulated a modified single-stranded model.

- According to this model DNA double helix binds with histone protein to form nucleoprotein fibrils.
- Folding of these fibrils takes place because of $Ca^{2+}$ bridge to form basic fibrils.
- Basic fibrils undergo further folding to form the chromosome.

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4. Du Braw (1965) proposed a "Folded Fibre Model" to describe the structure of Chromosomes. — According to this model chromosomes are made of chromatin fibres. Each chromatin fibre contains only one DNA double helix which is spirally coiled and coated with histone and non-histone proteins.

(b) Multi-stranded Models

(i) A number of multi-stranded models have been proposed to explain chromosome structure.

(ii) According to some of these models, each chromosome consist of two chromatids which is divisible into two half chromatids. Each half chromatid is again composed of two quarter chromatids. Each quarter chromatid is composed of four chromatin fibres. Each chromatin fibre is made up of two strands. Each strand consist of a single DNA molecule plus the associated histone and non-histone proteins.

(iii) Thus one chromatid is made of 4×2×2×2 = 32 DNA double strands. Hence a Chromosome with two chromatids is composed of 32×2 = 64 DNA molecules.

(iv) These 64 DNA double helices are arranged in a parallel manner and twisted together like the strands of a rope.

The above two groups of single-stranded and multi-stranded hypotheses are complementary and not exclusive to each other. Both of these conditions have been found in several species of plants and animals.

Chemical Composition of Chromosomes:

(i) Major Chemical components of

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Chromosomes are DNA, RNA, histone proteins and non-histone proteins.

Calcium is also present in the chromosomes.

(ii) Relative proportions of different components vary in different organisms. However, there are more proteins than DNA in the chromatin matter of all species.

(iii) Quantitative measurements of DNA have been made in a large number of cases. Quantity of DNA varies greatly in the cells of different organisms.

(iv) There are five fractions of histones ($H_1$, $H_2A$, $H_2B$, $H_3$ and $H_4$).

$H_1$ histone is most easily removed and so is least tightly bound. $H_3$ and $H_4$ are extremely conserved, having the same structure in different species.

(v) Histones play a structural role than a regulatory role.

(vi) Non-histones display higher but still limited diversity.

— Number of non-histones can vary from 12 to more than 20. Heterogeneity of these proteins suggested that these proteins are not conserved in evolution as histones did.

— Specific non-histone proteins exercise positive control on the activity of specific genes.

## Molecular Archetecture of Chromosomes:

(i) Kornberg and Thomas (1974) proposed an attractive Nucleosome Model for the basic chromatin structure involving DNA and histones.

(ii) They suggested that DNA interacts with a tetramer ($H3_2 - H4_2$) and two molecules of an oligomer ($H_2A - H_2B$) so that a tetramer

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involving two molecules each of the histones $H_3$ and $H_4$, is associated with two molecules each of the histones $H_2A$ and $H_2B$ and with 200 base pairs of DNA. This makes a repeating unit. One molecule of $H_1$ is also associated with each repeating unit.

— They also proposed that the tetramer makes the core of the unit and oligomers determine the space thus giving a flexible structure.

(iii) This model is supported from biochemical and electron microscopic results.

(iv) Oudet et al. (1975) proposed the term nucleosome for the repeating units which were observed as beads on strings.

Nucleosome (diameter: 12.5 nm) = 200 base pairs + 2 molecules each of $H_2A$, $H_2B$, $H_3$ and $H_4$

## Special Types of Chromosomes:

Lampbrush chromosomes of the amphibian oocytes, Giant chromosomes of salivary gland cells of dipteran insects and B chromosomes of some plants are such special types of chromosomes.

### (a) Lampbrush Chromosomes:

(i) Found in a variety of primary oocyte nuclei in vertebrates (mainly amphibians) and some invertebrates.

(ii) Pairs of loops in these chromosomes give them the characteristic lampbrush appearance.

(iii) They are found during the prolonged diplotene stage of first meiotic division in the primary oocytes of amphibians, and in the spermatocyte nuclei of *Drosophila*.

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(iv) There is enormous increase in the length of the chromosome, usually up to 1 mm.

(v) Chromosomes seem to have a chromomeric pattern with loops projecting in pairs from the majority of chromomeres.

— One to nine loops may arise from a single chromomere.

(vi) Size of loops varies with an average of 9.5 μ in inter-chromomeric fibres.

(vii) Frequently the loops exhibit a thin axis (which probably consists of one DNA double helix) from which fibres project which are covered with a loop matrix consisting of RNA and protein.

(viii) Number of pairs of loops gradually increases in meiosis till it reaches maximum in diplotene.

— As meiosis proceeds further, number of loops gradually decreases and the loops ultimately disappear due to disintegration.

(ix) Loops mainly consist of RNA.

(b) **Salivary Gland Chromosomes/Giant Chromosomes/Polytene Chromosomes:**

(i) Found in salivary gland cells of dipteran insects

(ii) First observed by Balbiani (1881)

(iii) They may reach a size up to 200 times or more the size of corresponding chromosomes at meiosis or in nuclei of ordinary mitotic cells. Hence they are called giant chromosomes.

(iv) They are somatically paired.

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As a result, number of these giant chromosomes in the salivary gland cells always appears to be half that in the normal somatic cells.

(v) They have a distinct pattern of transverse banding which consists of alternate chromatic and achromatic regions.

(vi) These bands occasionally form reversible puffs known as chromosome puffs or Balbiani rings, which are associated with differential gene activation.

(vii) These chromosomes represent each a bundle of fibrils having arisen by repeated cycles of endo-reduplication of single chromatids.

— That is why they are also called polytene chromosomes.

— Number of chromonemata (fibrils) per chromosome may reach up to 2000 in extreme cases. Some workers have placed this figure as high as 16,000.

(viii) Giant chromosomes in Drosophila melanogaster are found in the form of five long and one short strands radiating from a single roughly amorphous mass known as chromocentre.

— One long strand corresponds to the X chromosome and the remaining four long strands are the arms of II and III chromosomes.

— The centromeres of all these chromosomes fuse to form the chromocentre.

(C) B-Chromosomes :

(i) They are supernumerary chromosomes found in an organism as extra

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chromosomes over and above the standard diploid (or polyploid) chromosome complement.

(ii) They are found in the natural populations of many plant and some anima species.

— B-chromosomes have been reported in over 1000 species of bryophytes, ferns, gymnosperms and angiosperms.

— More commonly occur in outbreeders.

— Common in several grasses, e.g., *Agrostis*, *Avena*, *Bromus*, *Dactylis*, *Sorghum*, *Zea*, etc.

(iii) Supernumerary chromosomes are not found in all the individuals of a species and may not be found in all the cells of an individual.

(iv) B-chromosomes are not homologou with any of the basic A chromosomes, and thei inheritance is non-Mendelian, sometimes due to non-disjunction.

(v) They are usually smaller in size than A chromosomes and have their own unique pattern of heterochromatin distribution.

(vi) In general, they are genetically inert, but may rarely organize nucleoli and carry functional genetic material.

(vii) If present in higher numbers, they suppress vigour and fertility of the individual.

(viii) Origin and functions of B-chromosomes are largely obscure.

(ix) They usually do not pair with normal chromosomes during meiosis, though they may pair with each other when present in even number.

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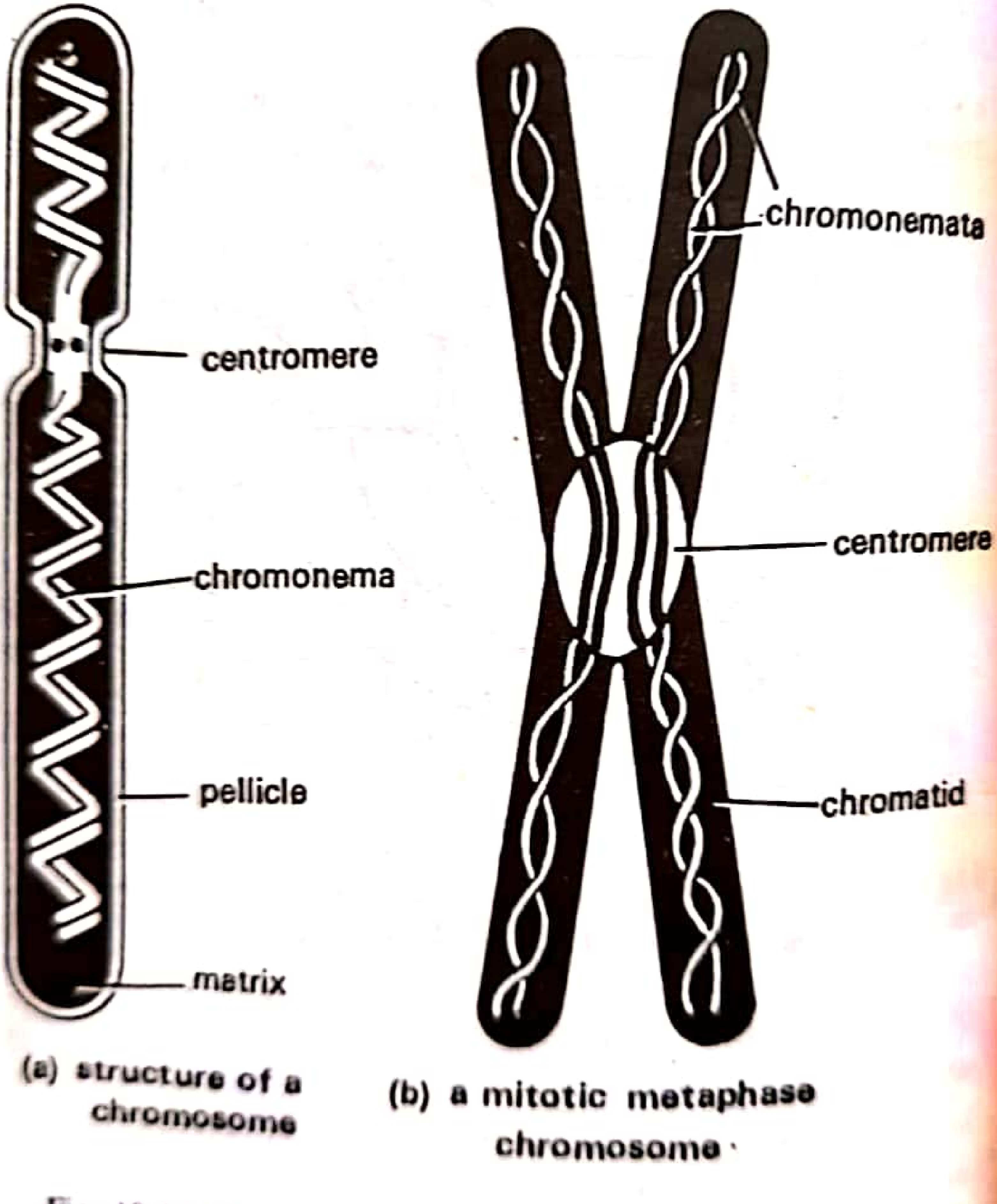
(x) Most significant effect of B-chromosomes is on seed and pollen fertility. Flowering time is generally delayed by B-chromosomes and several characters (plant height, weight and tiller number) are adversely affected.

(figs. below ↓)

—x—



(a) structure of a chromosome

(b) a mitotic metaphase chromosome

Fig. 12.12. Structure of a chromosome and chromatids.

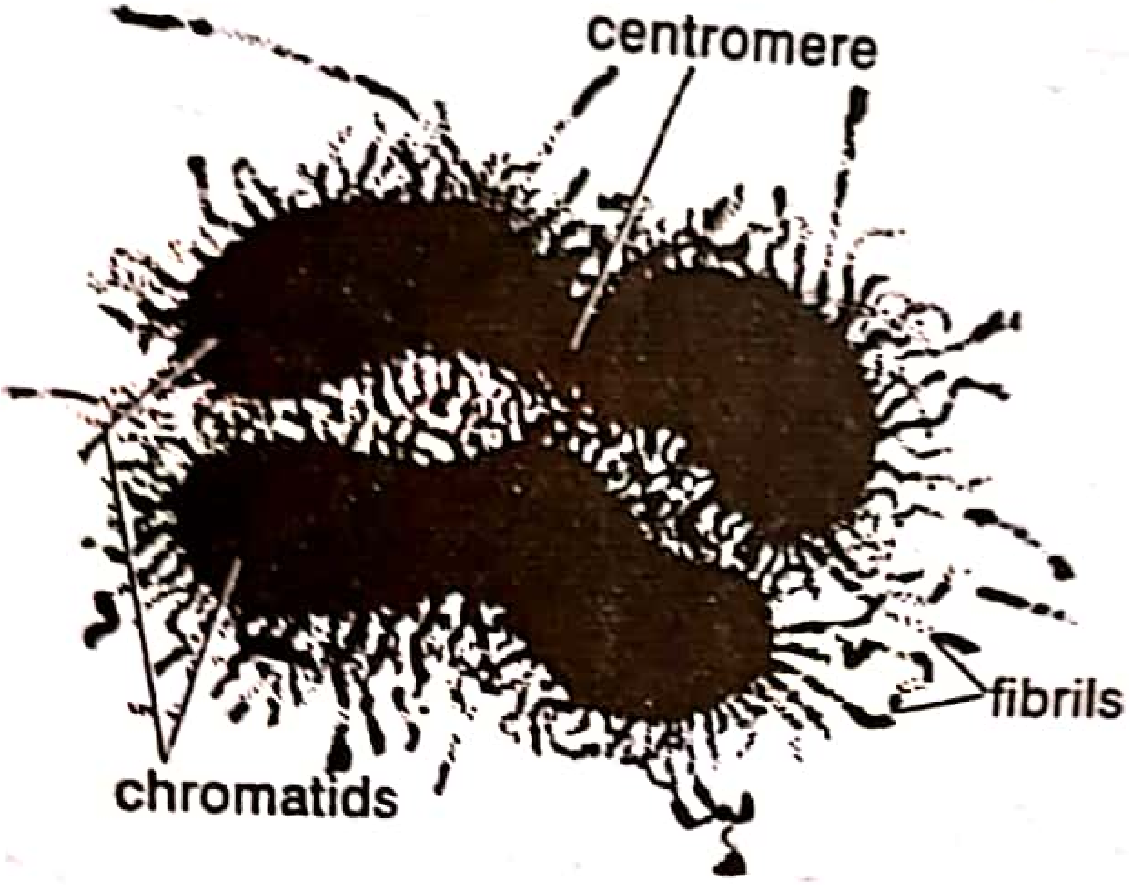
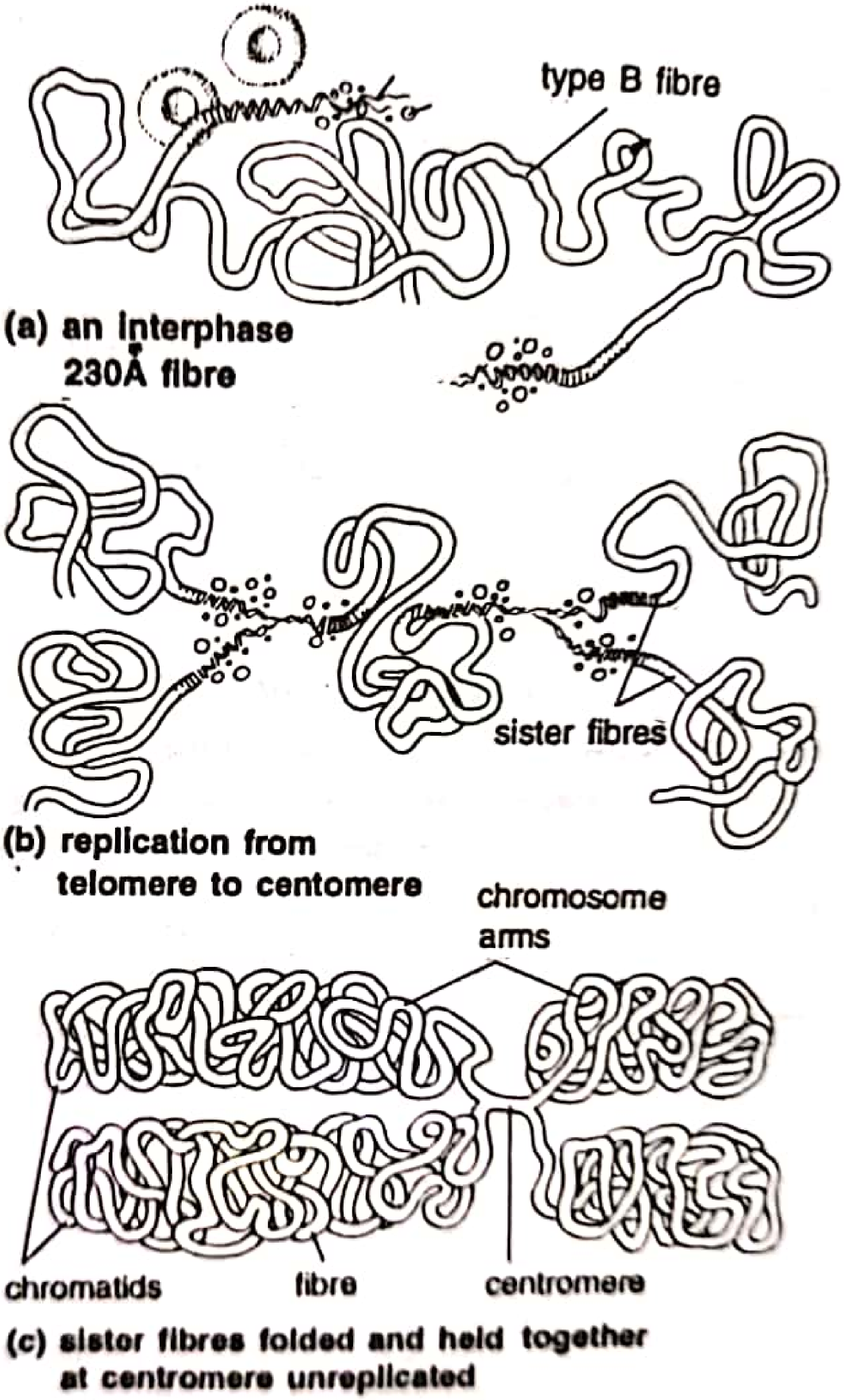


Fig. 12.17. Ultrastructure of chromosome (drawn from an electron micrograph. Du Praw : *Cell and Molecular Biology*, 1969).



(a) an interphase 230Å fibre

(b) replication from telomere to centomere

(c) sister fibres folded and held together at centromere unreplicated

Fig. 12.18. Structural organization in a chromosome according to folded fibre model (redrawn from Du Praw : *Cell and Molecular Biology*).

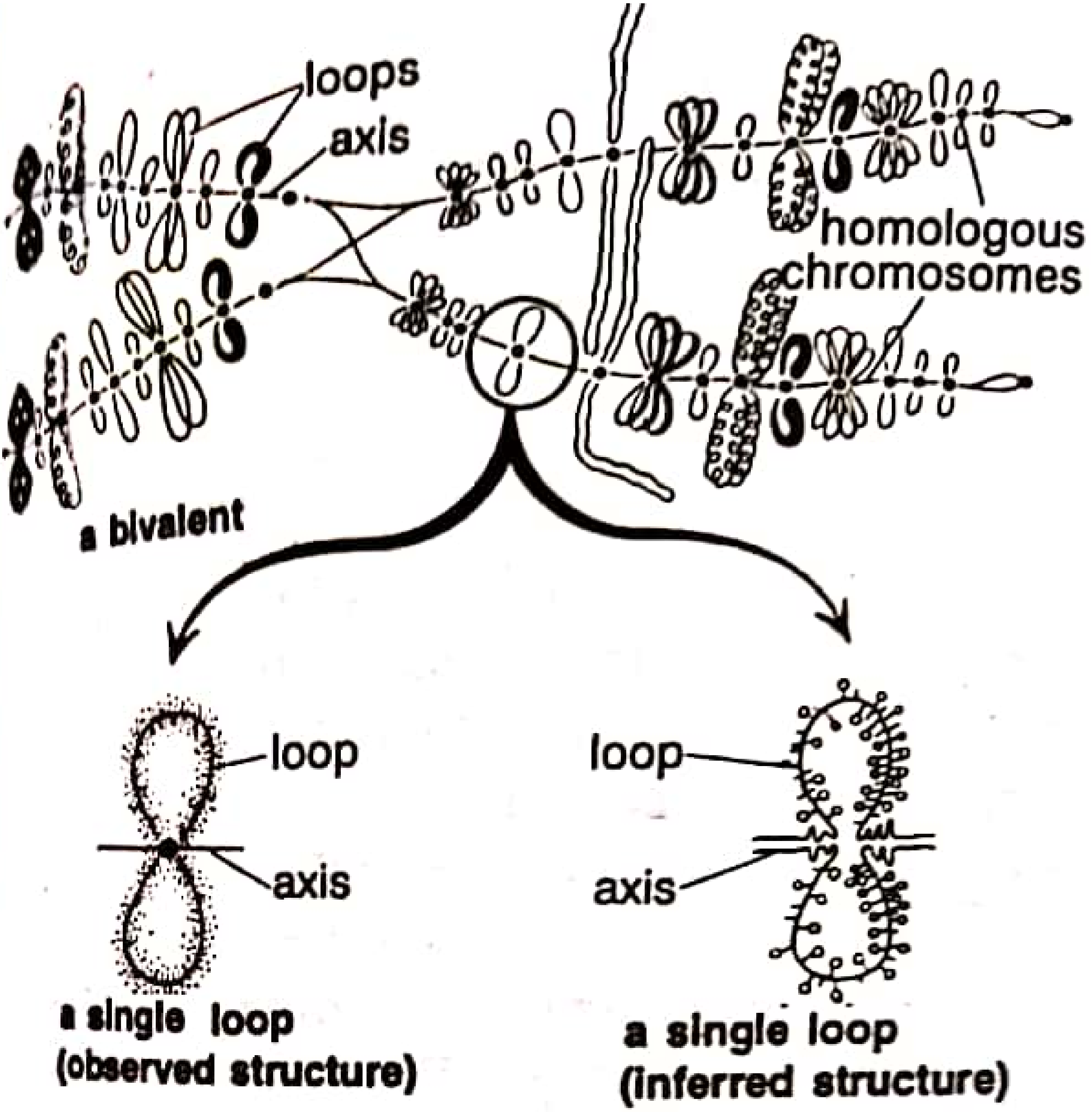
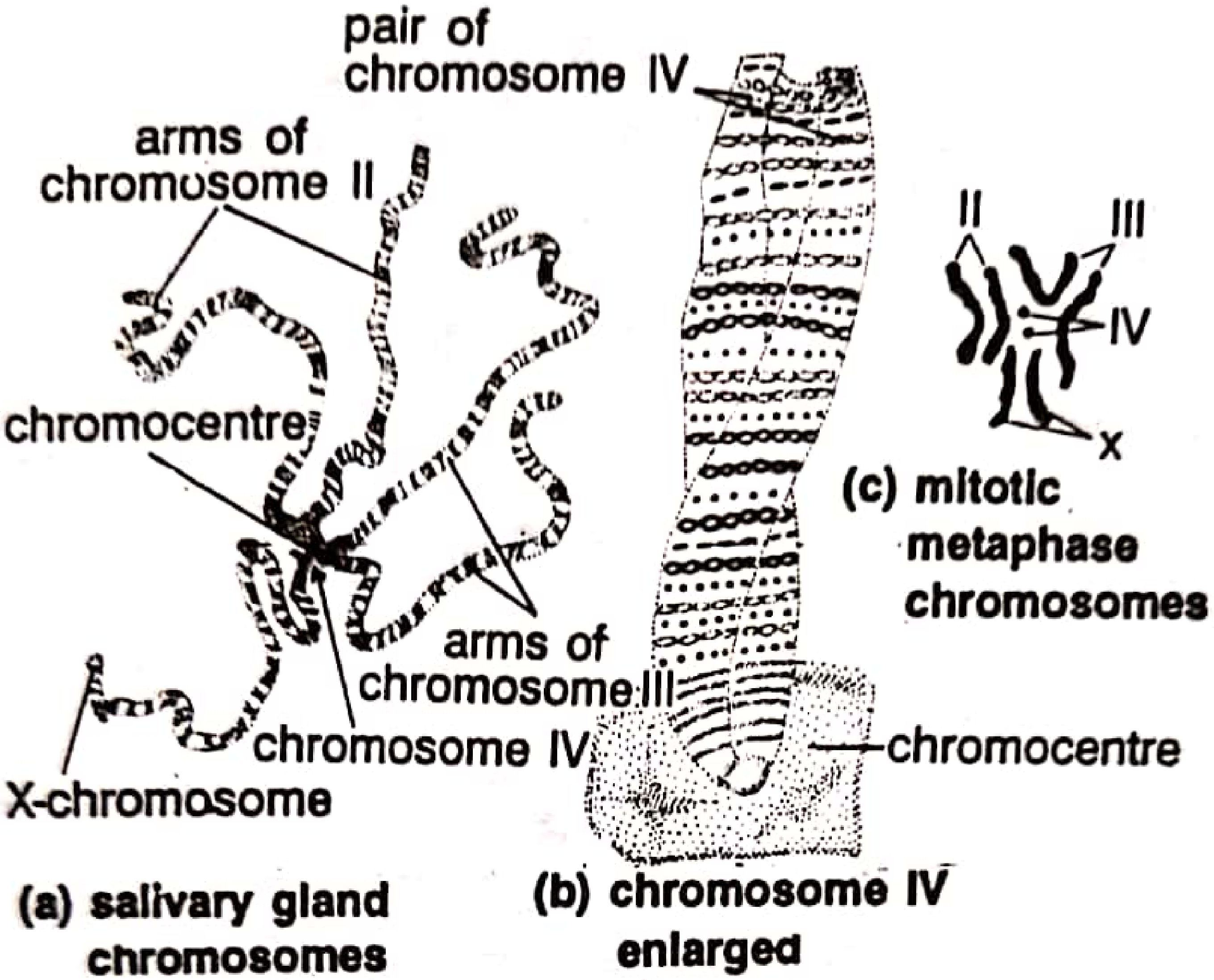


Fig. 12.21. Lampbrush chromosomes showing details of tructures (redrawn from Lewis & John : *Chromosome Marker*, 1963).



(a) salivary gland chromosomes

(b) chromosome IV enlarged

(c) mitotic metaphase chromosomes

Fig. 12.22. Salivary gland chromosome (a,b) and mitotic chromosomes (c) of *Drosophila melanogaster* (redrawn from Swanson : *Cytology and Cytogenetics*, 1957).

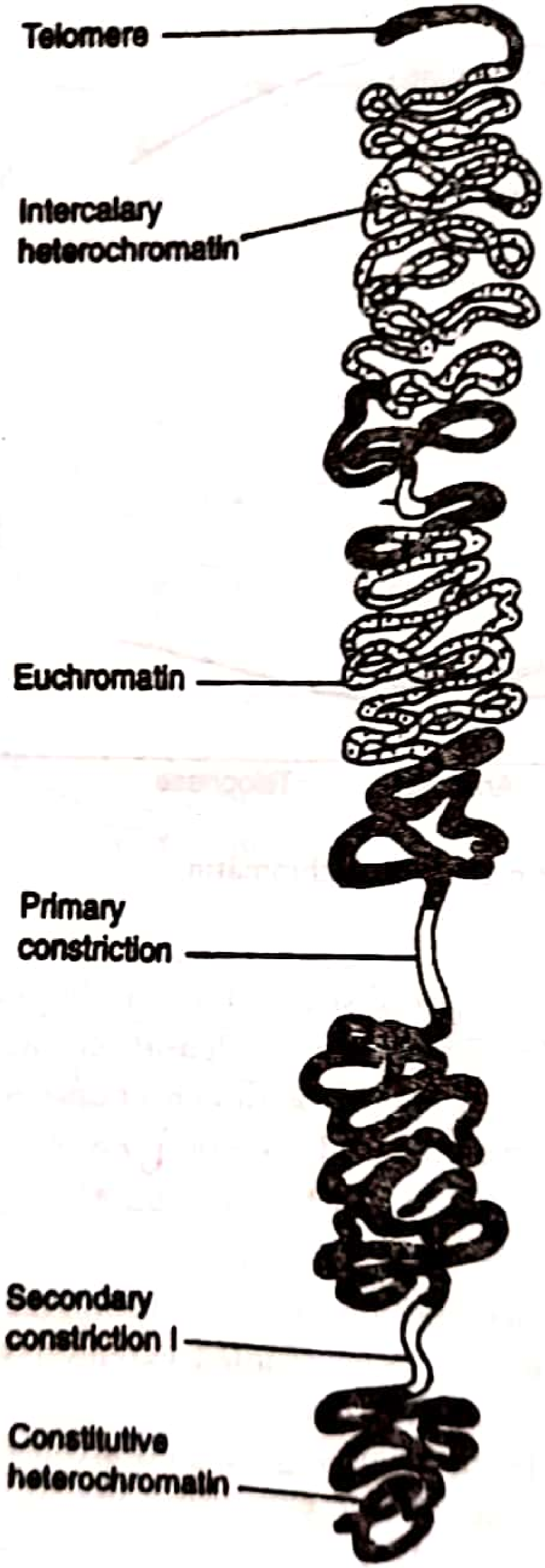


Fig. 13.9: Showing distribution of heterochromatin.