Polytene Chromosome

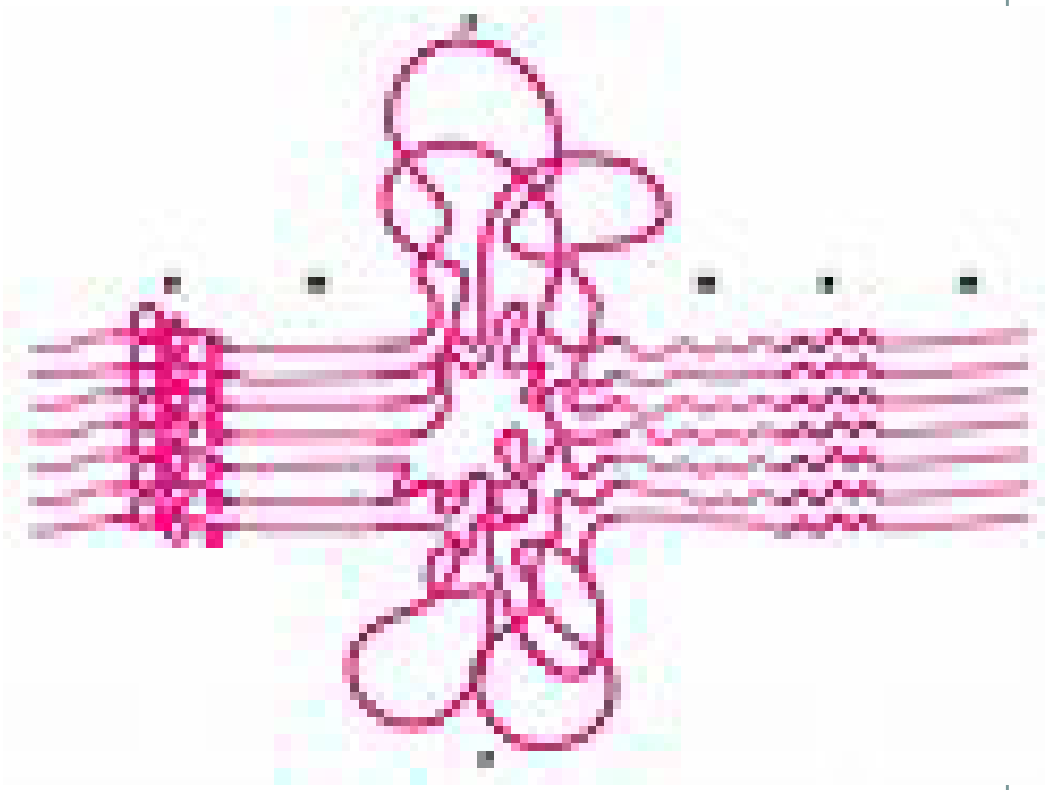
Specialized chromosome found in fly larvae. It is actually a pair
homologues, which is unusual (chromatin is the norm)

Polytene Chromosome

They are composed of several strands of DNA side by side produced when the DNA undergoes many rounds of replication without separating.

The strands present in bands undergo local uncoiling, called a **puff**. Puffs are areas of high transcriptional activity.

Polytene Puff

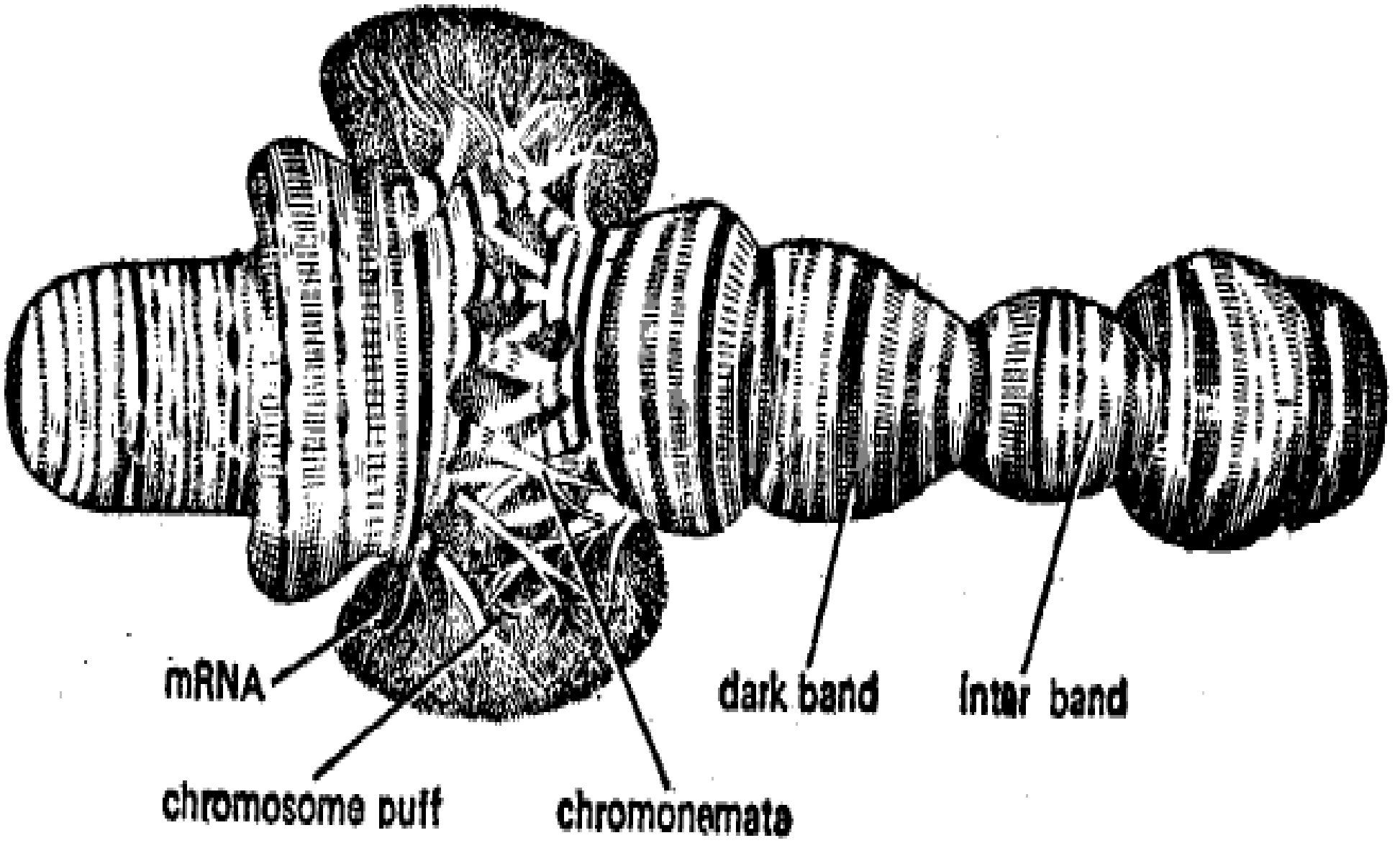


Polytene chromosomes

- Also called as Giant chromosomes / Salivary gland chromosomes
- First reported by Balbiani in 1881
- The nuclei of the salivary gland cells of the larvae of *Drosophila* have **unusually long and wide chromosomes, 100 or 200 times in size of the normal chromosomes**.
- The **salivary gland cells do not divide** after the glands are formed
- But their chromosomes replicate several times **(a process called endomitosis)** and become exceptionally giant – sized to be called polytene chromosomes.

- The polytene chromosomes of the salivary gland cells of *D. melanogaster* contain **1000 to 2000 chromosomes, which are formed by nine or ten consecutive multiplication cycles and remain associated parallel to each other.**
- Further, the polytene chromosomes have **alternating dark and light bands along their length**.
- The dark bands are comparable with the chromomeres of a simple chromosomes and are disc-shaped structures occupying the whole diameter of chromosome.

- They contain **euchromatin**.
- The light bands or inter bands are fibrillar and composed of heterochromatin.
- The swollen regions are known as chromosome **"puffs" or Balbiani rings**
- They are the regions of genetic activity
- Such puffs **change location** as development proceed, at specific locations.
- The presence of a specific puff is related with the appearance of a specific protein



Significance of Puffs

- They represent the **site of DNA synthesis** – gene transcription
- Transcription also occurs in the bands, but to a very small extent
- The ribonucleoprotein is found accumulated in the region of puff

Lampbrush Chromosome

Chromosome characteristic of vertebrate oocytes.

They are found during the diplotene stage of Prophase I in meiosis.

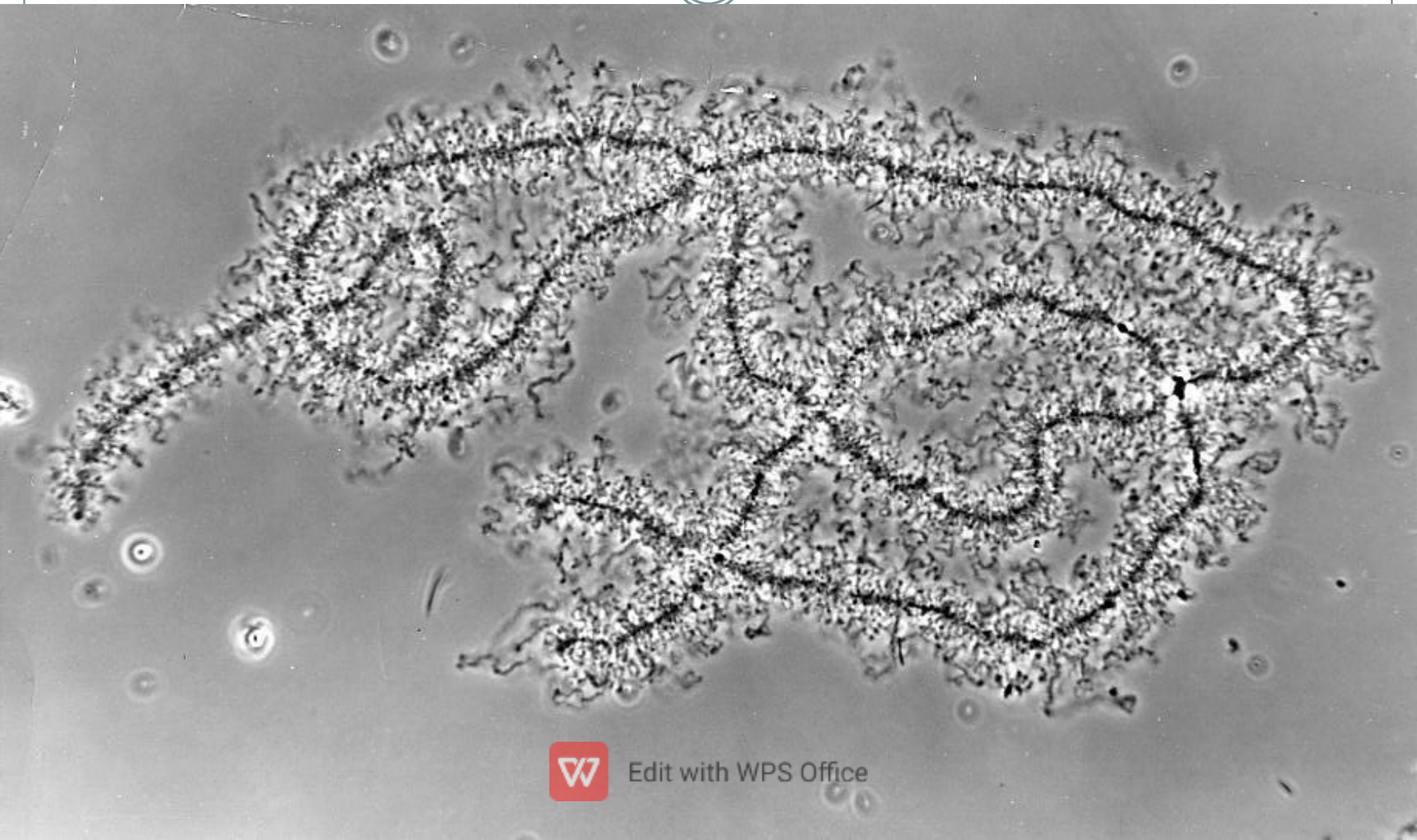
They are synapsed homologue pairs that do not condense like regular chromosomes.

Lampbrush Chromosome

They are composed of a center strand (two strands of DNA) with lateral loops (one strand of DNA).

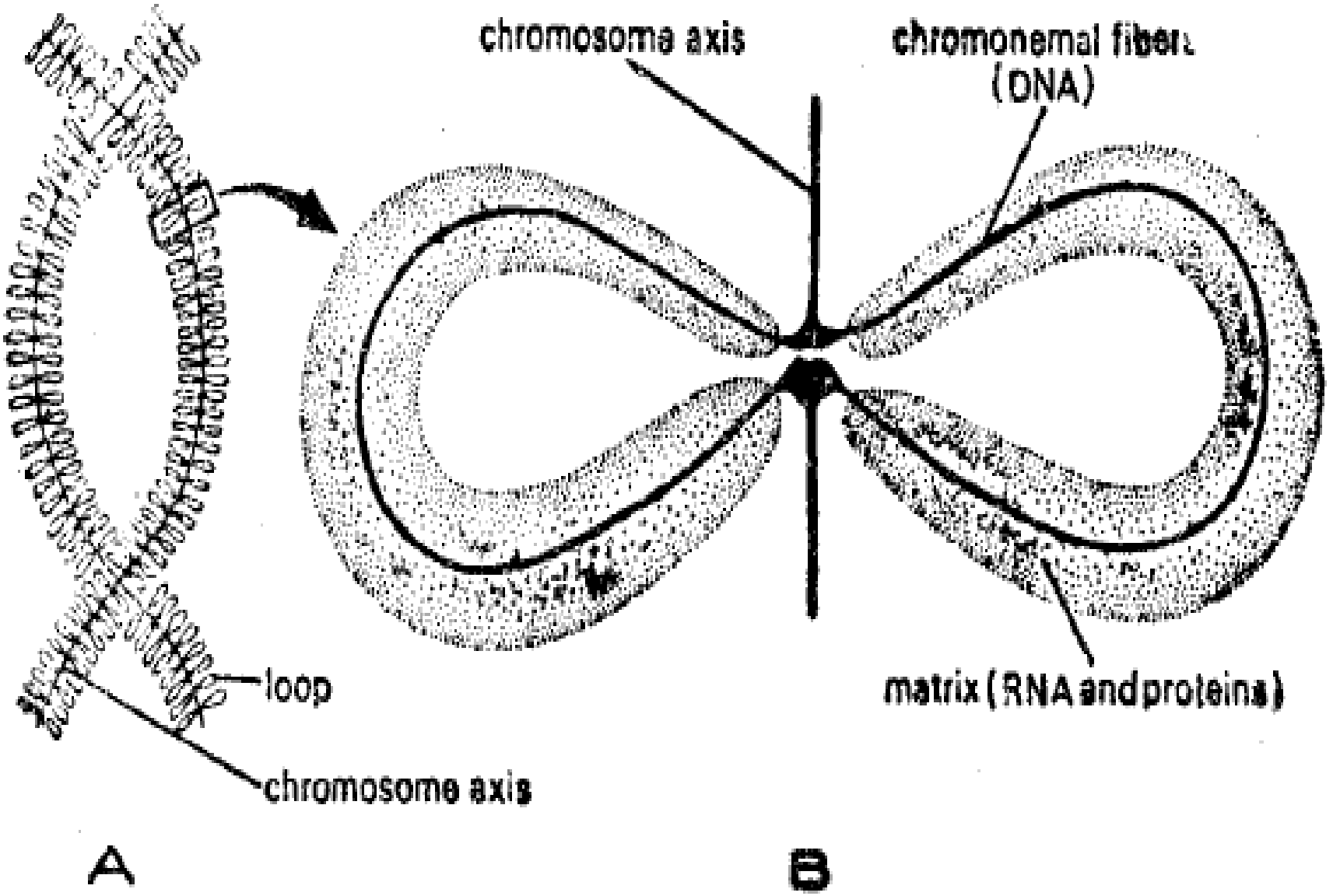
The lateral loops are transcriptionally active.

Lampbrush Chromosome



2. Lampbrush Chromosomes

- In diplotene stage of meiosis, the yolk-rich oocyte of vertebrates contain the nuclei with many lampbrush -shaped chromosomes of exceptionally large size.
- Lampbrush chromosomes discovered by Rucker in 1892
- They have remarkable length
- Sometimes larger than polytene chromosomes



A- At low magnification, B- A loop magnified (after Robertis, *et al.*, 1970).

- The large number of loops – projected in pair from the chromomere
- One to nine loops may arise from a single chromomere
- The chromomeres are connected by inter chromomere fibres
- The chromosomal axis, the chromomere and the loop axis are all made up of DNA- genetically active

Types of chromosomes based on Essentiality – A and B chromosomes

- **A – chromosome** :
  Normal member of chromosome complement of a species which are essential for normal growth and development
- **B- Chromosome**:Chromosome found in addition to normal chromosome

Other chromosomes

- Other chromosomes

- B Chromosomes
- Ring Chromosomes
- Isochromosomes

- In certain animals and plants, one or more chromosomes are found different from the basic structure of the normal chromosomes and these chromosomes are abnormal in size and shape. These chromosomes are called unusual of chromosomes.
- Some of the common examples of this type are **B-chromosomes** and Double minutes.

Unusual of Chromosomes

- The B-chromosomes have **other names as accessory and supernumerary chromosomes.**
- They are common in plants and they reduce their viability.
- These chromosomes are certain extra chromosomes that are found in certain individuals in a group like in case of maize.

Unusual of chromosoemes

- The **double minutes** are present in the cancer cells which show resistence against the drugs that are intaken by the cancer patient.
- These are chromosome like structures that are unstable.
- These chromosomes have telomeres that are formal and do not have any centromere.

3. B-Chromosomes

- Also called as **accessory chromosome**
- **Super numerary chromosome**
- It is an **extra chromosomes** not necessary for an individual
- Usually **acro or telocentric** in nature
- They do not pair with normal chromosomes during meiosis
- Recorded by **Longley and Randolph in Maize (1927)**
- Now reported in 42 families of Angiosperms including 163 genera and 475 species
- Much **smaller than normal chromosome**

Classified in two different ways

1. On the basis of their stability
2. On the basis of their size

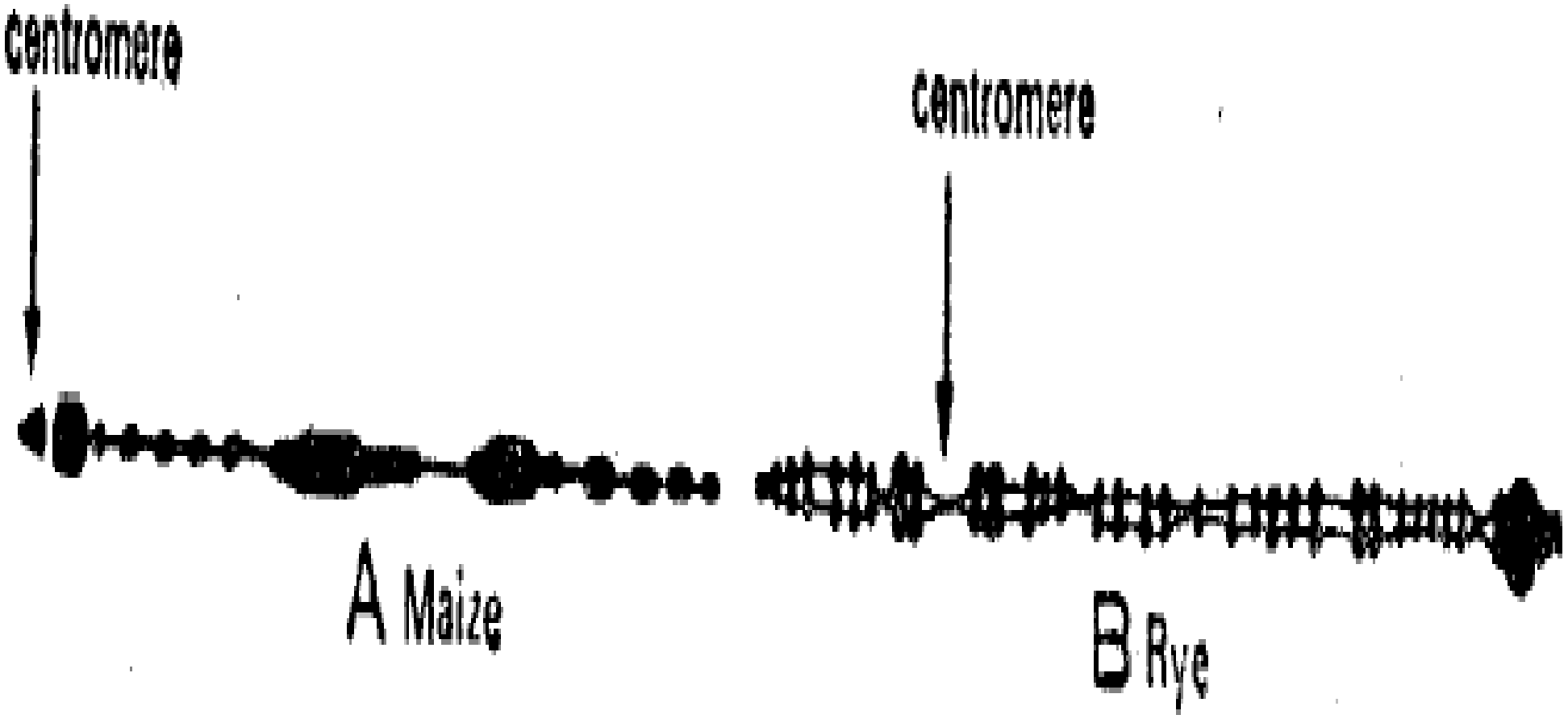
On the basis of their stability

1. **Stable** – mitotically stable and all cells will have the same number
2. **Unstable** - mitotically unstable and each cell will have different number of B chromosome within the same individual

On the basis of their size

1. Standard type – 1/4th length of normal chromosome, median centromere and uniform thickness
2. Small type – smaller size but not smaller than 1/2 of the standard B chromosomes
3. Very small type- small dot like structures, smaller than standard B chromosomes
4. Large type- chromosomes of double the size- large accessory chromosomes

The origin of the B-chromosomes is uncertain. In some animals they may be derivatives of sex chromosomes.



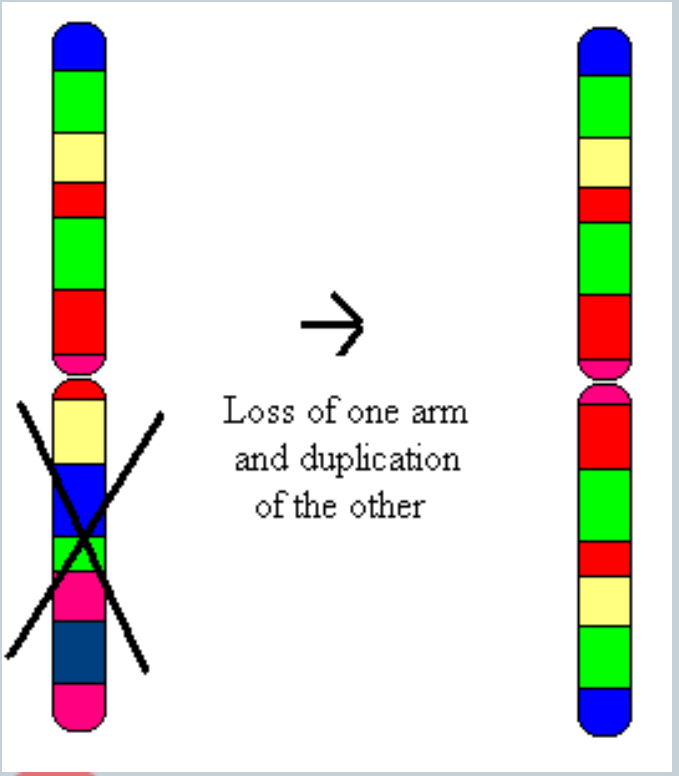
Meiotic behaviour of B chromosomes

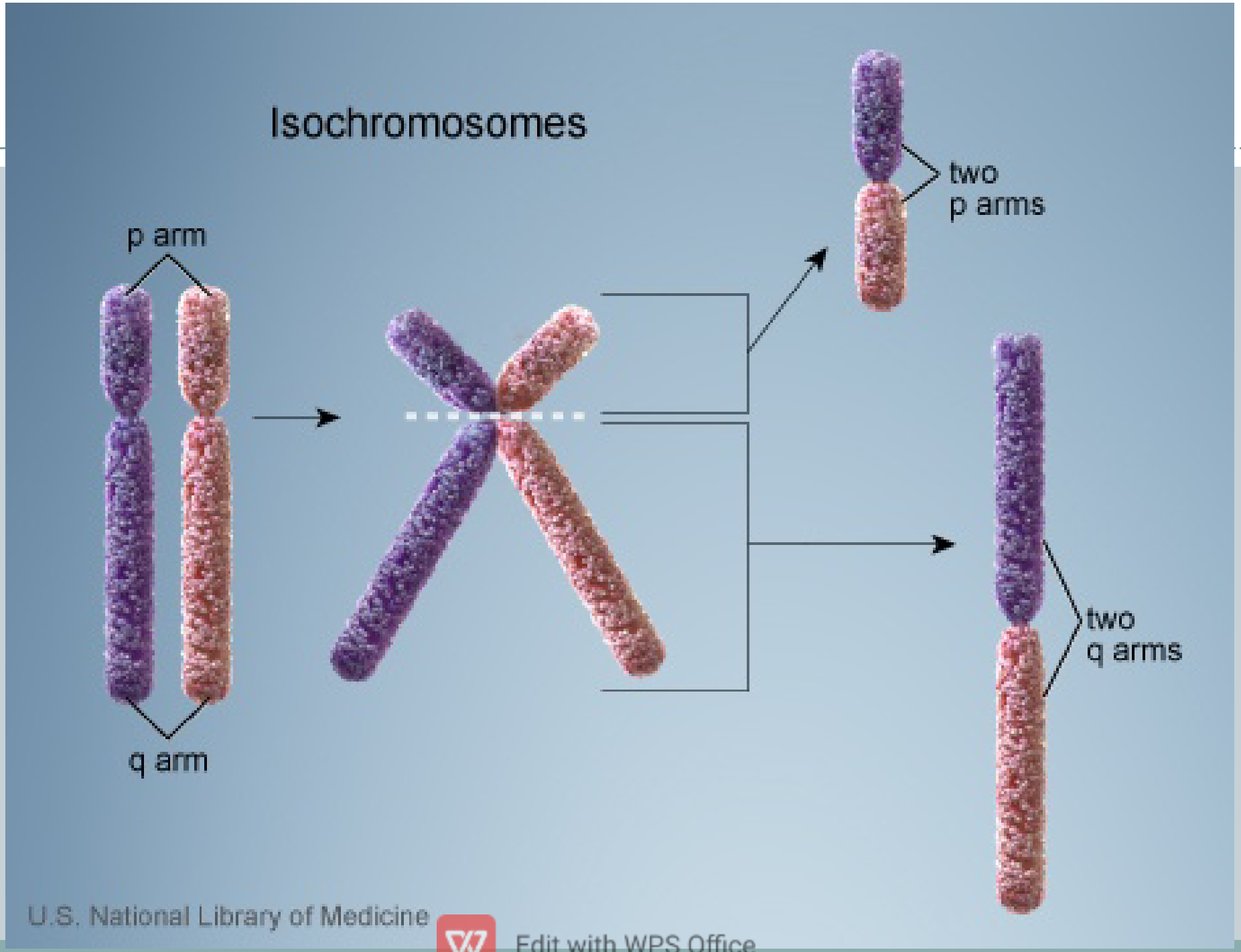
- They do not pair with A chromosomes
- Low degree of pairing is observed in B chromosomes
- Long B will pair with Long B and short B will pair with Short B
- When single B chromosome is present, they remain univalent during Pachytene
- When two B chromosomes are present they pair at pachytene

4. ISO Chromosomes

- An isochromosome is a chromosome in which both arms are identical.
- Both the arms are similar in morphology and gene content
- Originate by misdivision of centromere (vertical or transverse manner
- It is thought to arise when a centromere divides in the wrong plane, yielding two daughter chromosomes, each of which carries the information of one arm only but present twice.

- An **isochromosome** is a chromosome that has lost one of its arms and replaced it with an exact copy of the other arm





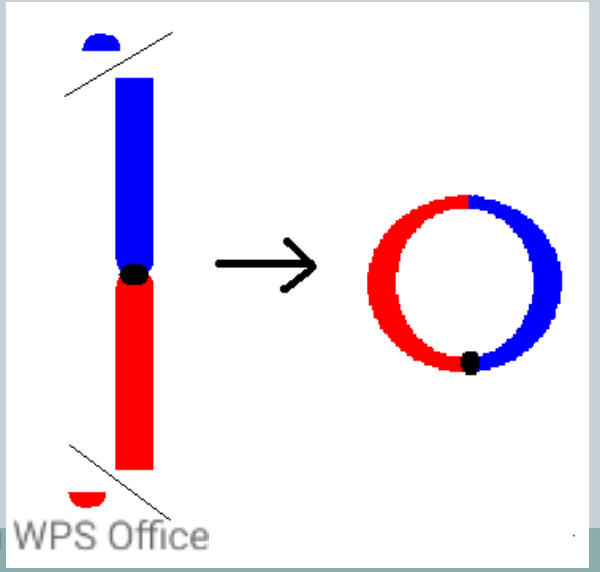
- The isochromosomes are formed during **mitosis and meiosis.**
- If a gamete having a isochromosome is fertilized by a normal gamete, the zygote will possess an unbalanced karyotype.
- In *Drosophila,* the misdivision of centromere of telocentric X chromosome changes that into an “attached-X” isochromosome,
- In man X- isochromosome causes the disease called **gonadal dysgenesis.**

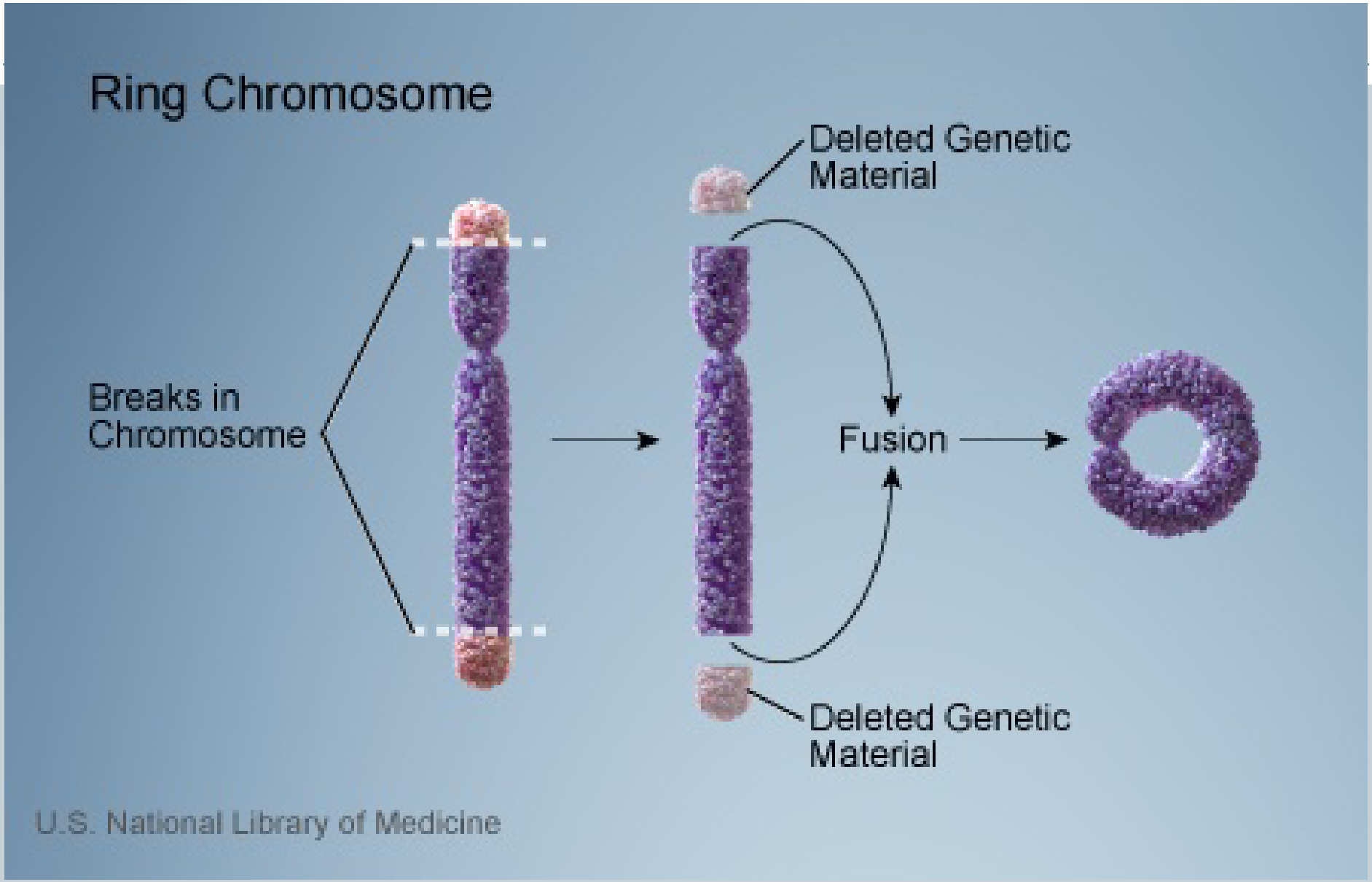
Ring chromosomes

- A **ring chromosome** is a chromosome whose arms have fused together to form a ring.
- A ring chromosome is denoted by the **symbol r.**
- Ring chromosomes may form in cells following genetic damage by mutagens like radiation, they may also arise spontaneously during development.
- **Ring chromosomes found in prokaryotes**
- Ring chromosomes otherwise called as genophores

Formation of ring

- Normally, the ends of a chromosome are lost, enabling the arms to fuse together. However, ring formation can also occur with only one end being lost.
- In rare cases, the telomeres at the ends of a chromosome fuse without any disappearing of material





- Ring chromosomes were Also reported in humans and drosophila
- Ring chromosomes thoroughly studied in maize by Mc Clintock
- Ring chromosomes are meiotically unstable

Hypothesis of Sutton & Boveri- chromosome theory of inheritance (Physical Basis of Heredity)

"Genes must be carried on the chromosomes"

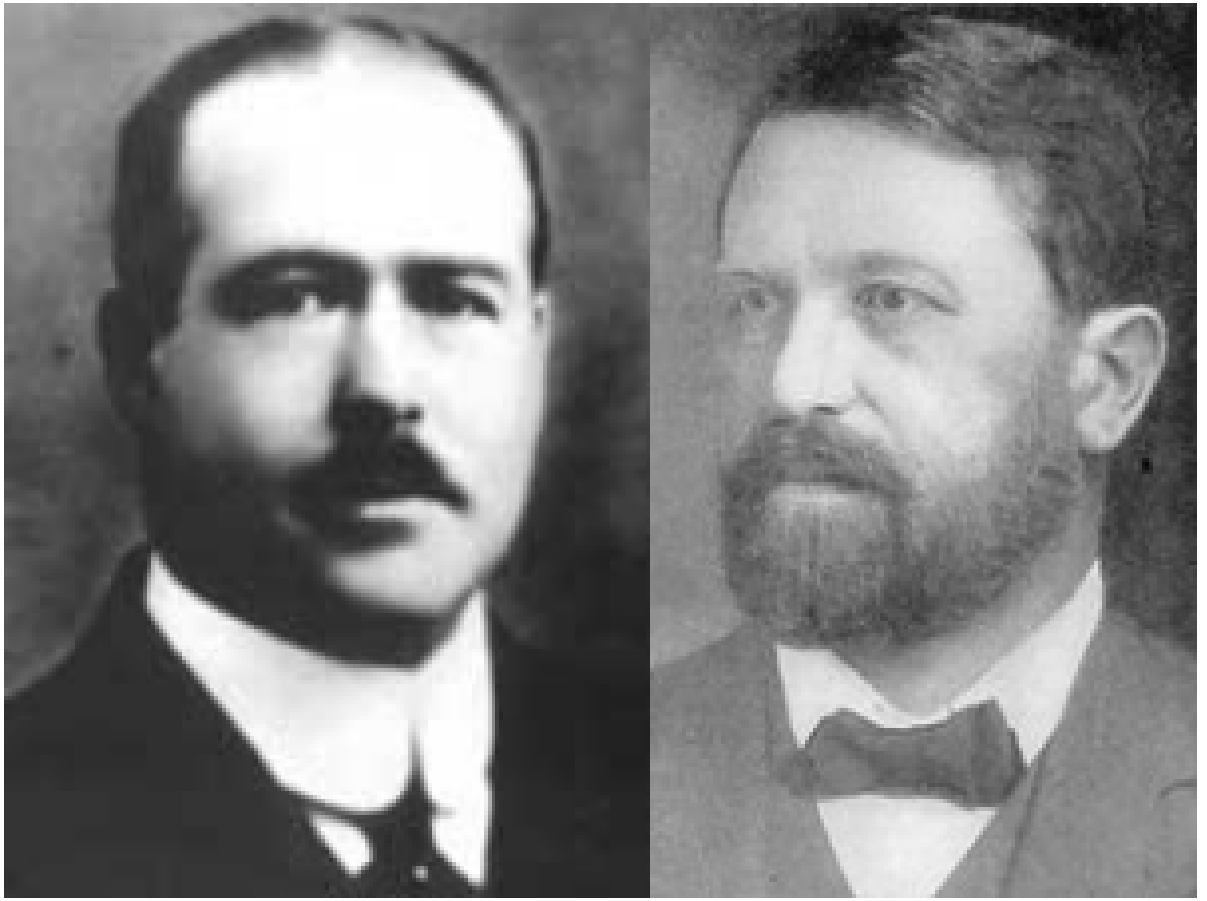
Sutton - American Biologist
Boveri - German Cytologist (1902)

They studied **cell structure and function** and concluded

Mendelian factors must be present in the chromosome and they assigned **genes to chromosomes**

because the **behavior of chromosomes** at meiosis and fertilization resembled in a very striking way, the **behavior of genes.**

This was observed in several breeding experiments.



Sutton (Left) and **Bovery** (right) independently developed the chromosome theory of inheritance in 1902.

Chromosomal theory of inheritance

- Fundamental unifying theory of genetics which identifies chromosomes as the carriers of genetic material.
- It correctly explains the mechanism underlying the laws of Mendelian inheritance by identifying chromosomes with the paired factors (particles) required by Mendel's laws.
- It also states that chromosomes are linear structures with genes located at specific sites called loci along them.
- It states simply that chromosomes, which are seen in all dividing cells and pass from one generation to the next, are the basis for all genetic inheritance.

Sutton & Bovery(1902) chromosome theory of inheritance

They proposed that

- The heredity determined characters are controlled by the heredity determinants, namely the genes,

  They occupy a definite loci on the chromosome and are transmitted from parent to offsprings

- In diploids, these genes exist as pairs of alleles and each gamete produced during meiosis will receive only one of the two alleles.
- Fusion of gametes during fertilization is usually at random
- Allelic gene pairs controlling different characters are transmitted independently
- When an organism receives different alleles for a particular character during fertilization, only one of these is usually expressed (the dominant) and the other (the recessive) but maintained and later transmitted, though not expressed phenotypically in F1.

Proof for the theory of Sutton and Boveri

Mc Clung, an American Zoologist was the first to give a definite suggestion that "Chromosomes determine the character".

Male grasshoppers differ from females in the absence of one chromosome.

- The female had even number of chromosomes, all being in pairs.

  All the eggs produced by the XX female are alike with single X chromosome each.

- The male had odd number of chromosomes, one of the chromosomes being always without a partner (XO)

  Two types of sperms are produced by the XO male in equal numbers. One type with the odd X chromosome and the other without it.

- Since the eggs are all alike and the two kinds of sperms are equal in number, the ratio of 1 female : 1 male was observed in the offsprings.

- The eggs that are fertilized by
  the sperms with X chromosome developed into females and
  the sperms lacking X chromosome developed into males.

Thus, the X chromosome determining the Sex was proved

Explanation for the results

- In some females, the XX chromosomes fail to disjoin at Meiosis and this primary disjunction leads to the production of eggs either with XX chromosomes or with no X chromosome
- Four types of Zygotes were produced when the eggs are fertilized by normal sperms

Egg	Sperm	Zygote	Progenies
X X	X	X X X	Super females (usually die)
X X	Y	X X Y	White eyed female (fertile)
No X	X	X	Red eyed male (sterile)
No X	Y	Y	Always die

the gene for colour of the eye in Drosophila
is located on the X chromosome.

This experiment proved that
specific genes are borne on specific chromosomes.

Chromosomal theory of Heredity – Proof by T.H.Morgan

- In 1910, T.H. Morgan presented a more direct evidence supporting the chromosome theory of heredity.
- He found that the pattern of transmission of white eye gene was identical with that of the X chromosome of Drosophila.
- This prompted Morgan to postulate that the gene for white eye was located in the X chromosome.

Morgan Chromosomal Theory – Experiment

Direct cross - Experiment in Drosophila

- In a cross between a red-eyed female and a white-eyed male, the F1 f les of both sexes are red-eyed.
- Of the F2 offsprings, all the females are red-eyed, whereas half the m a l e s a r e r e d - e y e d a n d t h e o t h e r half are white-eyed.
- The F2 shows a segregation of 3 red : 1 white, but strangely enough, the white-eyed flies are always male.

Reciprocal cross

- In the reciprocal cross between a white-eyed female and a red-eyed male, the F1 females are red-eyed and F1 males are white eyed.
- In the F2 generation, one half of the females and males are red- eyed and the other half white eyed.

Contd.., Morgan Experiment

- The different results from the reciprocal crosses can be explained only on the assumption that **the gene for colour of the eyes is located on the X chromosome**.
- Morgan thus showed that the distinctive pattern of inheritance of sex-linked genes parallels the transmission of the X chromosome.
- The work of Morgan and Bridges f irmly established the fact that **specific genes are borne on specific chromosomes.**

Morgon's chromosomal theory of inheritance

- Based on the genetic and cytological studies on Drosophila,
- Morgan postulated that,

  "genes are arranged in a linear order along the length of the chromosome, each gene having a fixed place on the chromosome, and its allele in corresponding position on the homologous chromosomes".