Identification of Chromosomal Aberrations in Polytene Chromosomes of *Drosophila/Chironomid* Larvae – Deletion, Inversion (Paracentric and Pericentric), and Ring Chromosome (from photographs)

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Objective

To identify different types of chromosomal aberrations, such as deletion, paracentric inversion, pericentric inversion, and ring chromosome, in polytene chromosomes of *Drosophila* or *Chironomid* larvae using photographs.

Principle

Polytene chromosomes are giant chromosomes formed by repeated rounds of DNA replication without cell division (endo-replication), resulting in many chromatids aligned side by side.

These chromosomes exhibit a characteristic pattern of alternating dark bands and light interbands, which serve as landmarks for mapping genes and detecting structural changes.

Structural chromosomal aberrations disturb the normal banding pattern, pairing, and continuity of polytene chromosomes and can be recognised as visible alterations in band arrangement.

Deletions, inversions and ring chromosomes in polytene chromosomes are therefore excellent cytological indicators of mutagenic effects and chromosomal rearrangements.

Materials Required

- Photographs of polytene chromosomes of *Drosophila* or *Chironomid* larvae
- Reference diagrams or charts of normal polytene chromosomes

Brief Theory of Aberrations

Deletion

- Loss of a segment of a chromosome, leading to missing bands in the polytene chromosome map.
- In heterozygous condition, the normal homolog cannot pair with the missing segment, producing an unpaired “loop” or gap in the region of deletion.

Inversion

An inversion is a rearrangement in which a chromosomal segment is cut out, rotated 180° and reinserted into the same chromosome.

- Paracentric inversion: Inverted segment does not include the centromere; both breakpoints lie on one arm.
- Pericentric inversion: Inverted segment includes the centromere; one breakpoint on each arm.

In polytene chromosomes, inversions are recognised by altered order and orientation of bands and formation of inversion loops when homologous chromosomes attempt to pair.

Ring chromosome

- Formed when both ends of a chromosome break and the broken ends fuse, producing a ring.
- In polytene chromosomes, ring structures may be seen instead of a normal linear chromosome arm or entire chromosome.

Procedure

- Observe the provided photographs of polytene chromosomes carefully.
- Identify the normal banding pattern of chromosomes.
- Compare the observed banding pattern with reference diagrams.
- Look for abnormalities such as missing segments, reversed segments, or circular chromosomes.
- Identify the type of chromosomal aberration present.
- Record the observations and justify the identification.

Observations

1. Deletion

<u>Identification Features</u>

- A **segment of chromosome is missing**.
- In polytene chromosomes, the homologous chromosome shows **loop formation** where the deleted region is absent in one chromosome but present in the other.

<u>Reason</u>

Deletion occurs due to **loss of a chromosomal segment**, resulting in missing genetic material.

2. Paracentric Inversion

<u>Identification Features</u>

- A **segment of chromosome is reversed in orientation**.
- The inverted segment **does not include the centromere**.
- In polytene chromosomes, an **inversion loop** is formed during pairing.

<u>Reason</u>

This occurs when a chromosomal segment breaks and reinserts in **reverse order on the same arm without involving the centromere**.

3. Pericentric Inversion

Identification Features

- The inverted segment **includes the centromere**.
- The inversion loop observed during pairing is often **larger and includes the centromere region**.

Reason

This inversion involves **both arms of the chromosome**, with the centromere included in the reversed segment.

4. Ring Chromosome

Identification Features

- Chromosome forms a **circular or ring-like structure**.
- Occurs due to **fusion of chromosomal ends after breakage**.

Reason

Both ends of a chromosome break and **join together to form a ring**, often leading to loss of terminal segments.

Discussion

Chromosomal aberrations are important structural mutations that may alter gene arrangement and expression. In polytene chromosomes, these aberrations can be easily recognised due to the **distinct banding pattern and pairing of homologous chromosomes**. Observing these aberrations helps in understanding **chromosome structure, gene mapping, and evolutionary genetics**.

Conclusion

Different chromosomal aberrations such as **deletion, paracentric inversion, pericentric inversion, and ring chromosome** were identified from photographs of polytene chromosomes of *Drosophila/Chironomid* larvae based on their characteristic structural features.

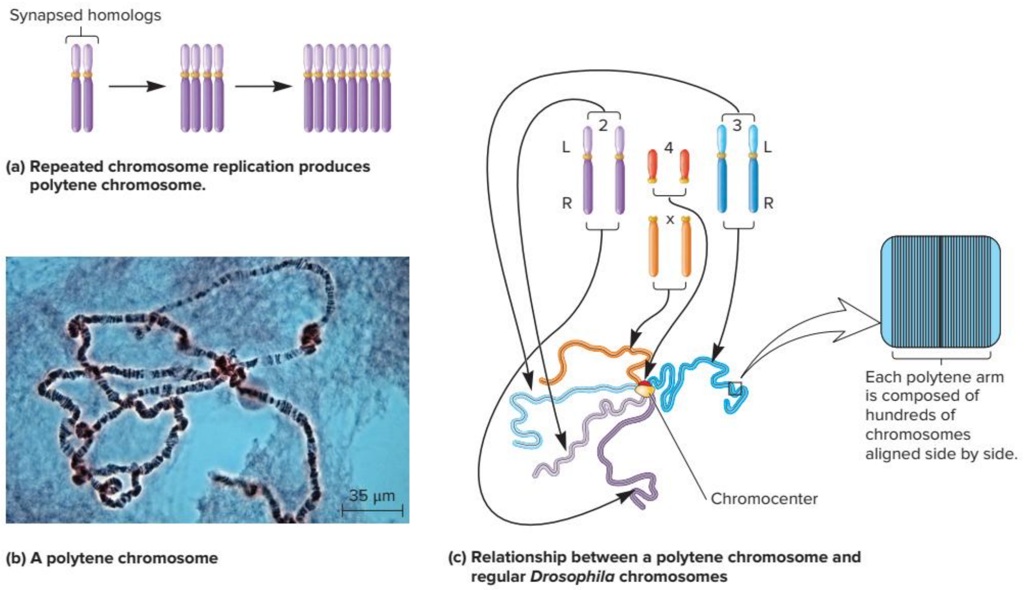
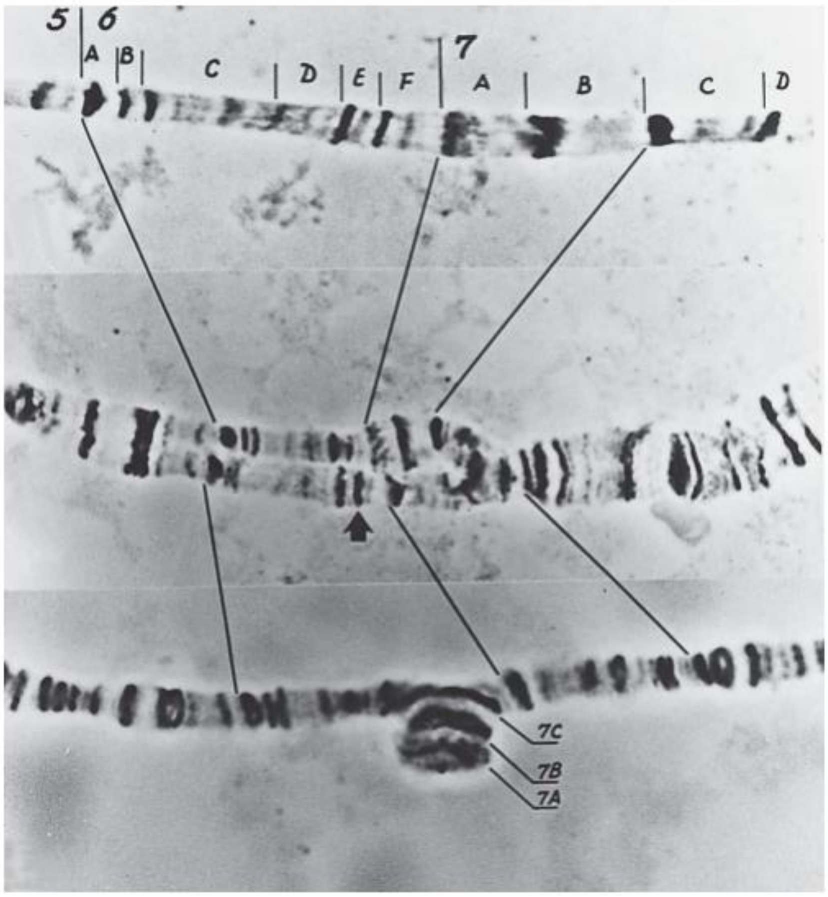


FIGURE 8.19 Polytene chromosomes in *Drosophila*. **(a)** A schematic illustration of the formation of a polytene chromosome. Homologous chromosomes synapse and undergo several rounds of replication without separating from each other. This results in a bundle of chromosomes that are parallel to each other. Note: This replication does not occur in highly condensed, heterochromatic DNA near the centromere. **(b)** A photograph of a polytene chromosome. **(c)** This drawing shows the relationship between the four pairs of chromosomes and the formation of a polytene chromosome in a salivary gland cell of *Drosophila*. The heterochromatic regions of the chromosomes aggregate at the chromocenter, and the arms of the chromosomes project outward. In chromosomes with two arms, the short arm is labeled L and the long arm is labeled R.

Source: Brooker, R.J. 2015. Genetics: Analysis and Principles. 6th Edition. Mc Graw Hill Education, NY



■ **FIGURE 6.18** Polytene chromosomes showing (*a*) the normal structure of regions 6 and 7 in the middle of the *Drosophila* X chromosome, (*b*) a heterozygote with a deletion of region 6F-7C in one of the chromosomes (arrow), and (*c*) an X chromosome showing a reverse tandem duplication of region 6F-7C. In (*b*) the prominent bands in regions 7A and 7C are present in the upper chromosome but absent in the lower one, indicating that the lower chromosome has undergone a deletion. In (*c*) the duplicated sequence reads 7C, 7B, 7A, 7A, 7B, 7C from left to right.

Source: Snustad, D.P., Simmons, M. J. 2012. Principles of Genetics, 6th Edition. John Wiley and Sons. NJ

Fig: Paracentric inversion [Singh, G., Singh, A.K., 2020. Non-random Distribution of Heterozygous Inversions in a Natural Population of *Drosophila malerkotliana*. Journal of Scientific Research. 64(1): 85-89.

Fig: Chromosomal fusion leading to a ring chromosome.[Sharma, S., Shrivastava, P., 2021. Polytene chromosome aberrations based genotoxicity appraisal by implementing insect genome. International Journal of Applied Research. 7(4): 100-106.