Faculty Science

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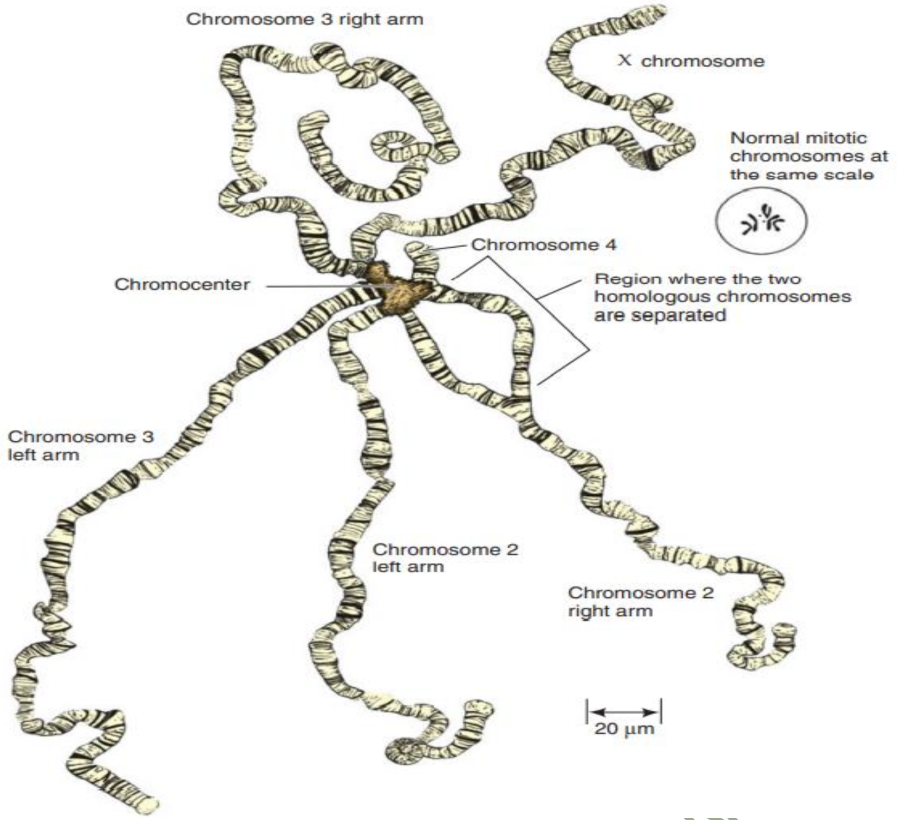
B.Sc II Paper-II(Cytology, Genetics, Evolution& Ecology)

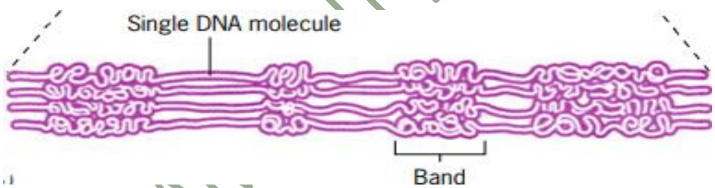
Unit-I Topic- Polytene Chromosome, Lampbrush Chromosome and B Chromosome

Polytene chromosome

The larval salivary glands of Drosophila species and other dipteran insects contain enlarged interphase chromosomes that are visible in light microscope. When fixed and stained with a dye that stains DNA, these are known as **polytene chromosome** (many threads) **or salivary gland chromosome**.
Polytene chromosomes were first observed by **E. G. Balbiani** in 1881. Later, in the 1930s**, Theophilus Painter** and colleagues recognized that the large size of polytene chromosomes provided geneticists unique opportunities to study chromosome structure and gene organization. Drosophila cells contain eight chromosomes (two sets of four chromosomes each). In the salivary glands, the homologous chromosomes synapse with each other and undergo repeated rounds of chromosome replication without separating from each other approximately ten rounds. The repeated rounds of chromosomal replication produce a bundle of chromosomes that lie together in a parallel fashion in the absence of cytoplasmic division. This type of division is called endomitosis. During this process, the four types of chromosomes aggregate to form an enormous polytene chromosome with several arms. The central point where the chromosomes aggregate is known as the **chromocenter**. Each of the four types of chromosome is attached to the chromocenter near its centromere. The X and Y chromosomes and chromosome 4 are telocentric, and chromosomes 2 and 3 are metacentric. Therefore, the X and Y and chromosome 4 have a single arm projecting from the chromocenter, whereas chromosomes 2 and 3 have two arms. The polytene chromosomes of D. melanogaster forms an alternating series of **bands and interbands**. Each member of the polytene set consists of a visible series of **bands or chromomeres**. These bands vary in size. The bands contain most of the mass of DNA and stain intensely with appropriate reagents. The regions between them stain more lightly and are called interbands. There are about 5,000 bands in the D. melanogaster set. The chromocenter consists largely of heterochromatin (In the male it includes the entire Y chromosome).



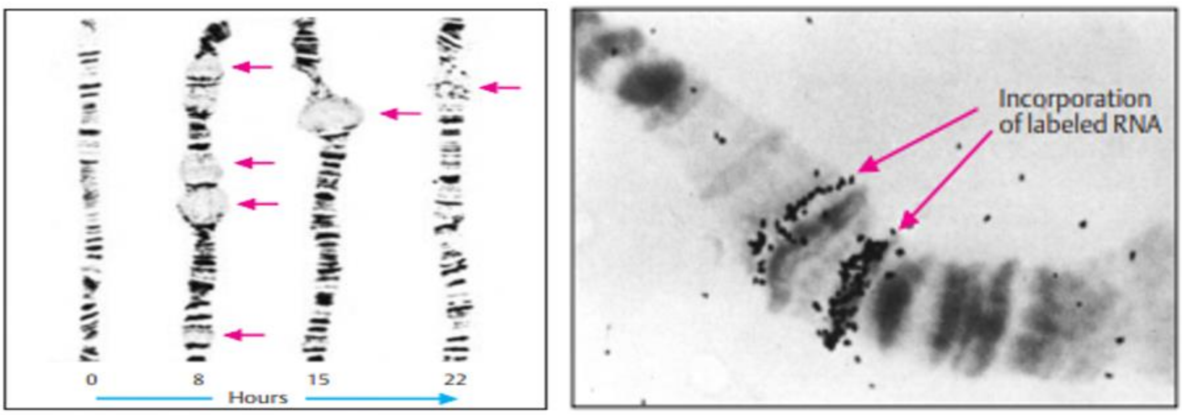
The entire set of polytene chromosomes in one Drosophila salivary cell



A part of polytene chromosome showing bands and interbands

The remaining 75% of the genome is organized into alternating bands and interbands in the polytene chromosomes. A polytene chromosome of Drosophila salivary glands has about 1000 DNA molecules which are arranged side by side and which arise from 10 rounds of DNA replication (2^{10}=1024). The number of doublings is different in the various tissues of the D.melanogaster larva. Chironomus has 16000 DNA molecules in their polytene chromosomes. The degree of polyteny is the number of haploid chromosomes contained in the giant chromosome. The banding pattern is characteristic for each strain of Drosophila. The constant number and linear arrangement of the bands form a cytological map of the chromosomes. Rearrangements—such as deletions, inversions, or duplications—result in alterations of the order of bands. The linear array of bands can be equated with the linear array of genes. Thus, genetic rearrangements, as seen in a linkage map, can be correlated with structural rearrangements of the cytological map. Ultimately, a particular mutation can be located in a particular band. The total number of genic loci in D. melanogaster exceeds the number of bands, so there are probably multiple genes in most or all bands. Using in situ hybridization, it is possible to determine directly the band within which a particular sequence lies. The relative positions of these genes on the giant chromosomes agreed with those predicted on the basis of genetic maps prepared from recombination frequencies, thus providing visual confirmation of the validity of the entire mapping procedure.

Comparisons of banding patterns among polytene chromosomes of different species have provided an unparalleled opportunity for the investigation of evolutionary changes at the chromosome level. These chromosomes are not inert cellular objects but dynamic structures in which certain regions become "puffed out" at particular stages of development. These puffs are the sites where DNA is being transcribed at a very high level, providing one of the best systems available for the direct visualization of gene expression. These chromosome puffs are also termed **Balbiani rings**. Puffs may appear and disappear depending on the production of specific proteins which needs to be secreted in large amounts in the larval saliva. Chromosome IV of the insect C. tentans has three Balbiani rings in the salivary gland. Another peculiarity of the polytene chromosomes is that the paternal and maternal chromosomes remain associated side by side and the phenomenon is termed as somatic pairing. Both polyteney and polyploidy have excess DNA per nucleus, but in the later the new chromosomes are separate from each other.



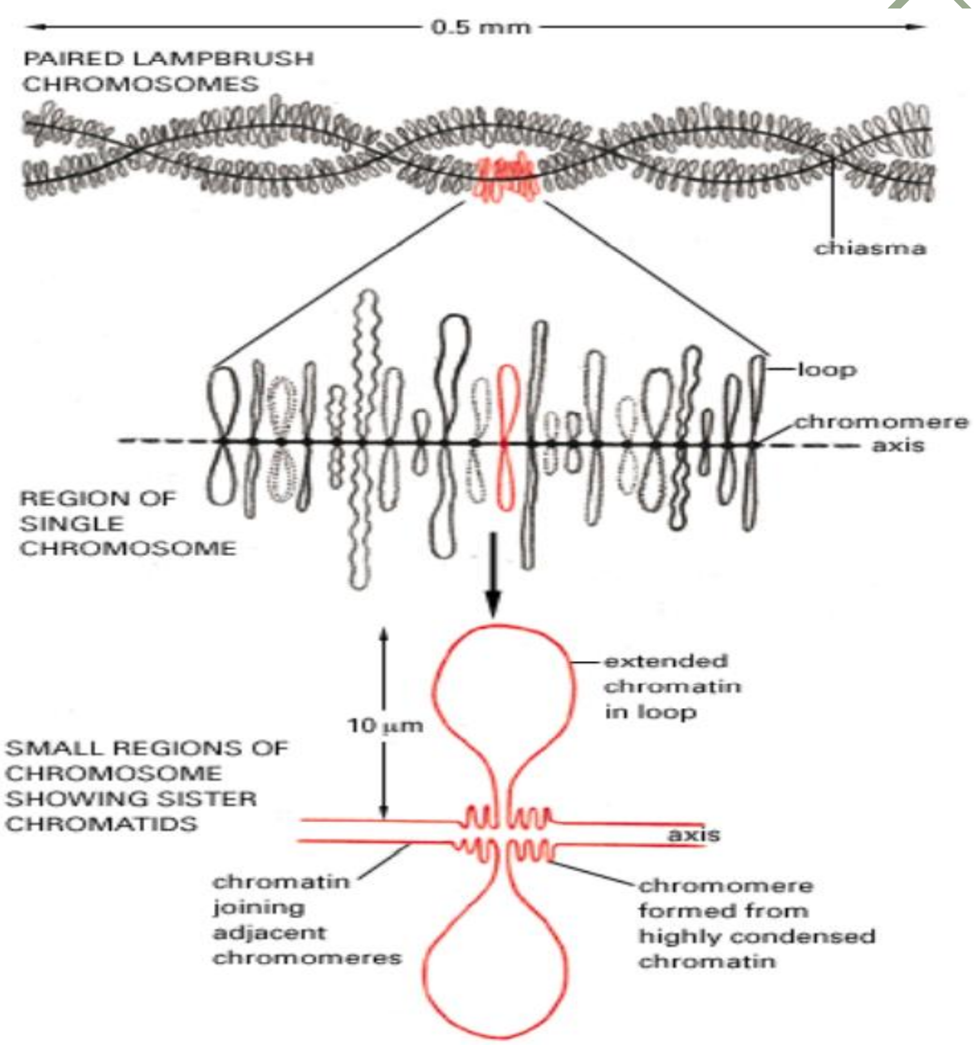
1. Formation of puffs (arrows)

2. Evidence of gene activity

B. Functional stages in polytene chromosomes

Lampbrush chromosome

The lampbrush chromosomes were first observed by **Walther Flemming** in 1882 in growing amphibian oocytes (immature eggs). They occur in the diplotene stage of meiotic prophase during oogenesis in oocytes of vertebrates and some invertebrates. These were identified in shark and described by **Ruckert** in 1982. These are unusual lampbrush chromosomes (the **largest chromosomes known**) are clearly visible even in the light microscope, where they are seen to be organized into a series of large chromatin loops emanating from a linear chromosomal axis. A given loop always contains the same DNA sequence, and it remains extended in the same manner as the oocyte grows. Lampbrush chromosomes are formed during an unusually extended meiosis, which can last up to several months. During this period, the chromosomes exist in a stretched-out form. At a later point during meiosis, the chromosomes revert to their usual compact size. The extended state provides a unique opportunity to see the structure of the chromosome.



A model for the structure of a lampbrush chromosome

A lampbrush chromosome is a meiotic bivalent in which the two pairs of sister chromatids are held together at chiasmata. The sister chromatids remain connected along their lengths and each homolog appears, therefore,

as a single fiber. Each sister chromatid pair forms a series of ellipsoidal chromomeres, 1 to 2 µm in diameter, which are connected by a very fine thread. This thread contains the two sister duplexes of DNA and runs continuously along the chromosome, through the chromomeres.

The lampbrush chromosomes are about 30 times less compacted along their axis than their somatic counterparts. The total length of the entire lampbrush chromosome set is 5 to 6 µm and is organized into about 5,000 chromomeres. The lampbrush chromosomes take their name from the **lateral loops** that extrude from the chromomeres at certain positions. The arrangement of fibers around the chromosome axis resembles the cleaning fibers of a lampbrush. The loops extend in pairs, one from each sister chromatid. The loops are continuous with the axial thread, representing chromosomal material extruded from its more compact organization in the chromomere. The loops are surrounded by a matrix of ribonucleoproteins that contain nascent RNA chains. Often, a transcription unit can be defined by the increase in the length of the RNP moving around the loop. The loop is an extruded segment of DNA that is being actively transcribed. In some cases, researchers have identified loops corresponding to particular genes. For these cases, the structure of the transcribed gene and the nature of the product can allow for a rare situation wherein gene expression can be directly visualized and studied in situ.

B Chromosome

B chromosomes are additional chromosomes possessed by some individuals in a population, but not all. The normal members of chromosome complements are known as A chromosomes. B chromosomes are common in plants and fungi, insects and animals. B chromosomes are also known as **supernumerary, accessory, dispensable or lineage-specific chromosomes**. These chromosomes are not essential for the life of a species, and are lacking in the most of the individuals. Thus a population would consist of individuals with 0, 1, 2, 3 (etc.) B chromosomes. B chromosomes are distinct from marker chromosomes or additional copies of normal chromosomes as they occur in trisomies.

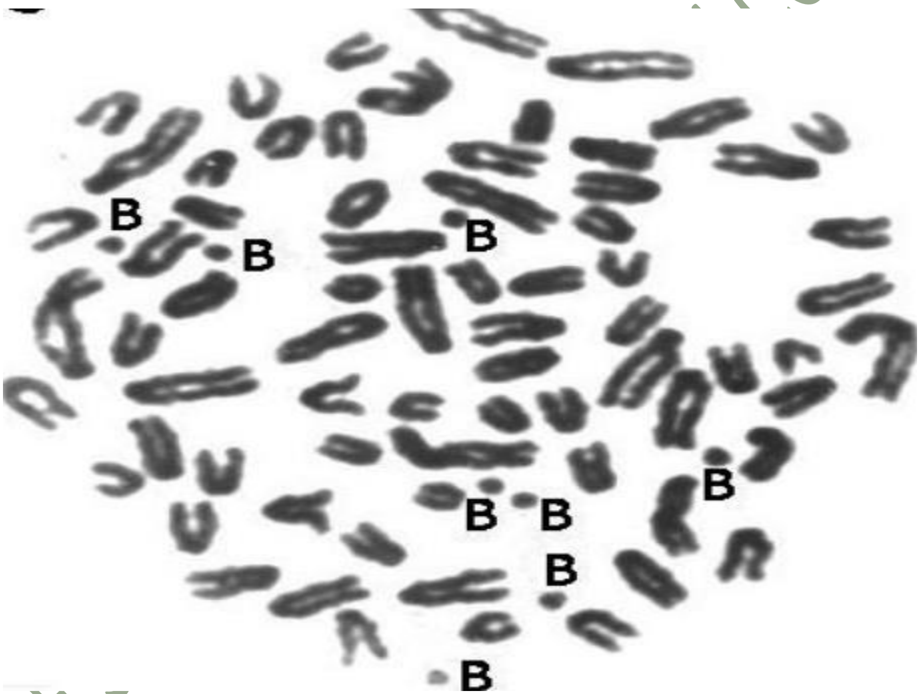
The evolutionary origin of supernumerary chromosomes is obscure, but presumably they must have been derived from heterochromatic segments of normal chromosomes in the remote past. Next generation sequencing has shown that the B chromosomes from rye are amalgamations of the rye A chromosomes. **Function**: B chromosomes are mainly or entirely heterochromatic (i.e. largely non-coding), but some contain sizeable euchromatic segments (E.g. such as the B chromosomes of maize). In some cases, B chromosomes act as selfish genetic elements. In other cases, B chromosomes provide some positive adaptive advantage. For instance, the British grasshopper Myrmeleotettix maculatus has two structural types of B chromosomes: metacentrics and submetacentrics. The supernumeraries, which have a satellite DNA, occur in warm dry environments and are scarce or absent in humid, cooler localities.

There is evidence of deleterious effects of supernumeraries on pollen fertility and favourable effects or associations with particular habitats are also known in a number of species. B chromosomes may play a positive role on normal A chromosomes in some circumstances. In wheat, an allopolyploid, the B chromosomes suppress homologous pairing which reduces multiple pairing between homologous chromosomes. Bivalent pairing is ensured by a gene on chromosome 5 of the B genome Ph locus. The B chromosomes also have the following effects on A chromosomes:

1.Increases asymmetry chiasma distribution

2.Increases crossing over and recombination frequencies: increases variation

3.Cause increased unpaired chromosomes: infertility



B Chromosomes with metaphase A Chromosomes

B chromosomes have tendency to accumulate in meiotic cell products resulting in an increase of B number over generations, thereby acting as selfish genetic elements. However this effect is counterbalanced for selection against infertility.
In fungi- Chromosome polymorphisms are very common among fungi. Different isolates of the same species often have a different chromosome number, with some of these additional chromosomes being unnecessary for normal growth in culture. The extra chromosomes are known as conditionally dispensable, or

supernumerary, because they are dispensable for certain situations, but may confer a selective advantage under different environments.

Supernumerary chromosomes do not carry genes that are necessary for basic fungal growth, but may have some functional significance. For example, it has been discovered that the supernumerary chromosome of the pea pathogen Haematonectria haematococca carries genes that are important to the disease-causing capacity of the fungus. This supernumerary DNA was found to code for a group of enzymes that metabolize toxins, known as **phytoalexins**, that are secreted by the plant's immune system. It is possible that these supernumerary elements originated in horizontal gene transfer events because sequence analysis often indicates that they have a different evolutionary history from essential chromosomal DNA. The wheat infecting fungal pathogen Zymoseptoria tritici contains 8 dispensable B-chromosomes (the largest number of dispensable chromosomes observed in fungi).

Reference

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2. Krebs J.E., Golldstein E. S., Kilpatrick S. T. (2018) in "Lewin's Genes XII", 12th edition. Jones & Bartlett Learning USA.

3. https://en.wikipedia.org/wiki/B_chromosome