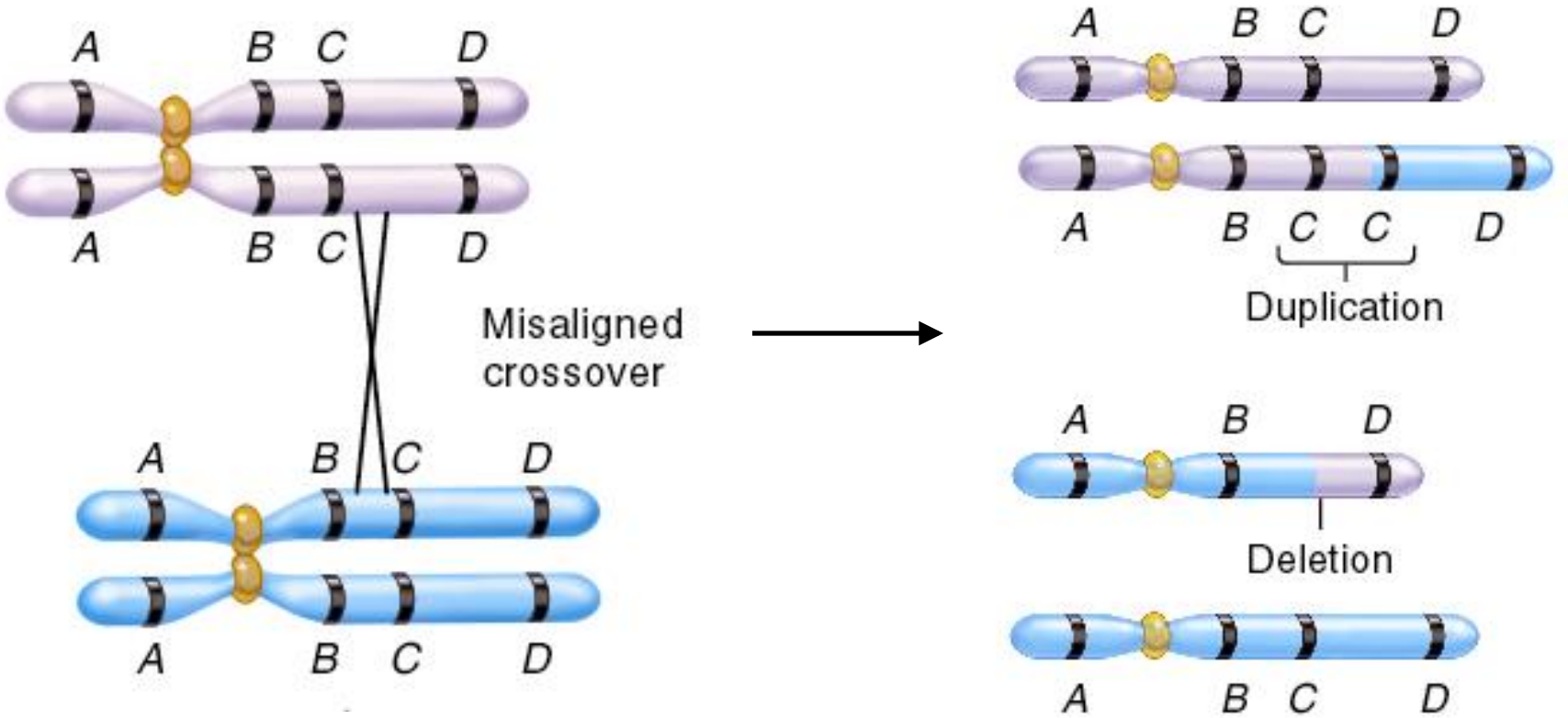
Gene Duplications and the Bar Eye Phenotype

Please Read for Lab Four

Duplications

- A chromosomal duplication is usually caused by abnormal events during recombination

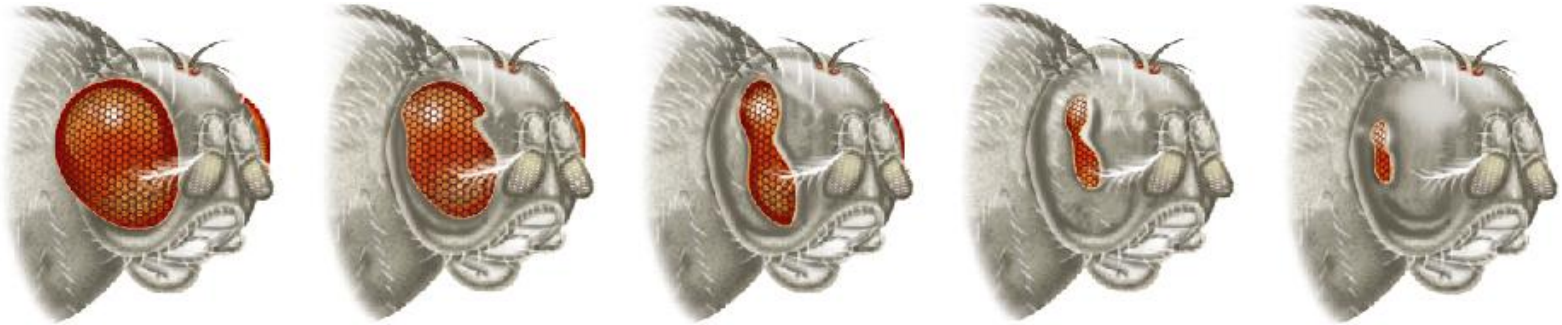


Duplications

- Like deletions, the phenotypic consequences of duplications tend to be correlated to size
  - Duplications are more likely to have phenotypic effects if they involve a large piece of the chromosome
- However, duplications tend to have less harmful effects than deletions of comparable size
- In humans, relatively few well-defined syndromes are caused by small chromosomal duplications

Bridges' Experiment Investigating the Bar-Eye Phenotype in *Drosophila*

- Bar eyes is a trait in which flies have a reduced number of facets
- Ultra-bar (or double-bar) is a trait in which flies have even fewer facets than the *bar* homozygote
- Both traits are X-linked and show gene dosage



	Normal	Bar heterozygote	Bar homozygote	Ultra-bar heterozygote	Ultra-bar homozygote
Approx. number of facets	810	350	70	45	25

Bridges' Experiment Investigating the Bar-Eye Phenotype in *Drosophila*

- Calvin Bridges in the 1930s investigated the bar/ultra-bar phenomenon at the cytological level

- The cells of the salivary gland of *Drosophila* have gigantic chromosomes, termed polytene chromosomes
  - The banding patterns on these chromosomes is easily seen
    - It is thus possible to detect the duplication or deletion of single genes

The Hypothesis

– Information concerning the nature of the bar and ultra-bar phenotypes may be revealed by a cytological examination of polytene chromosomes

Testing the Hypothesis

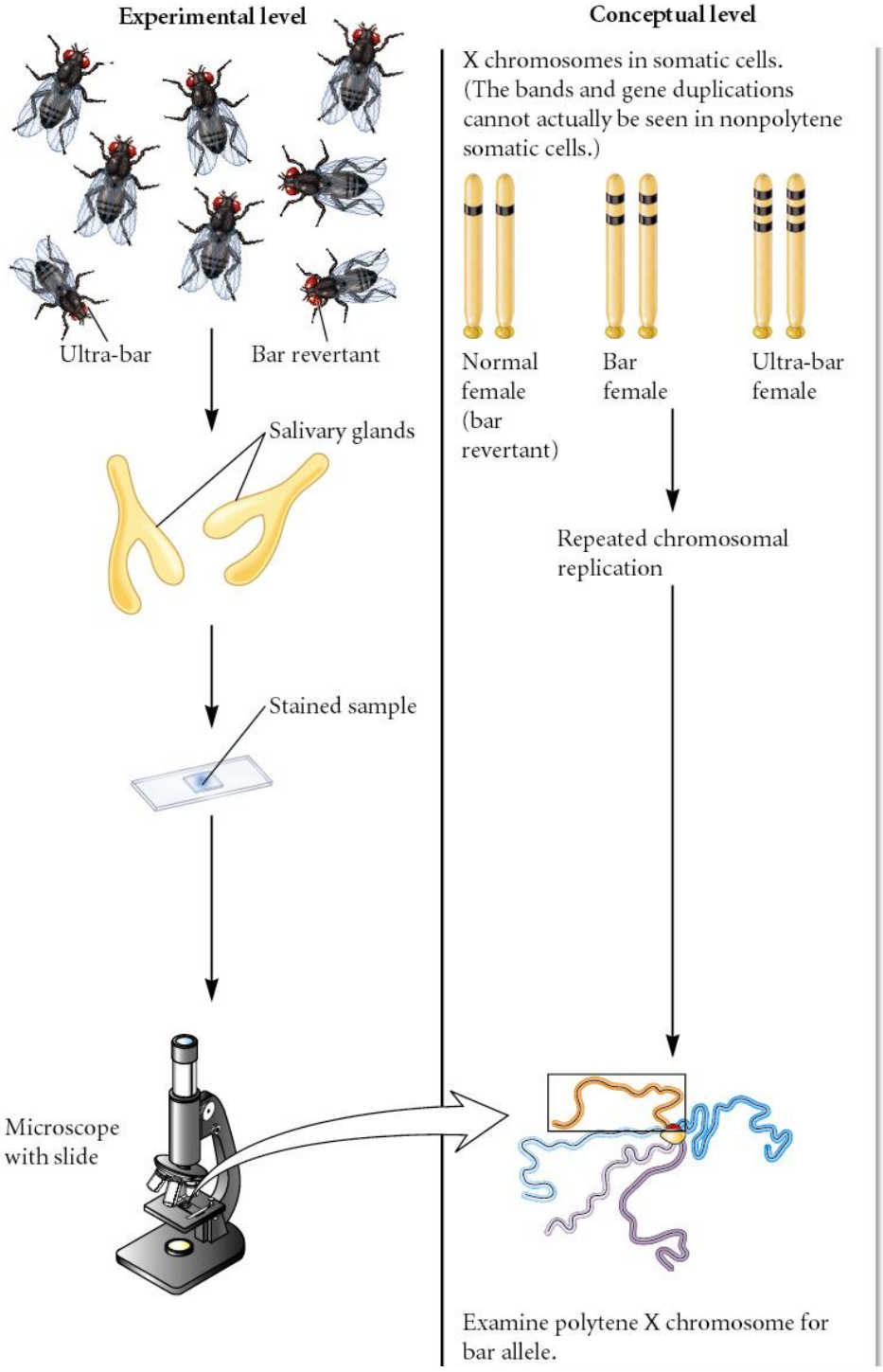
- Refer to next slides

1. Within the strain of homozygous bar-eyed flies, identify rare flies that have normal eyes (bar revertants) or ultra-bar eyes.

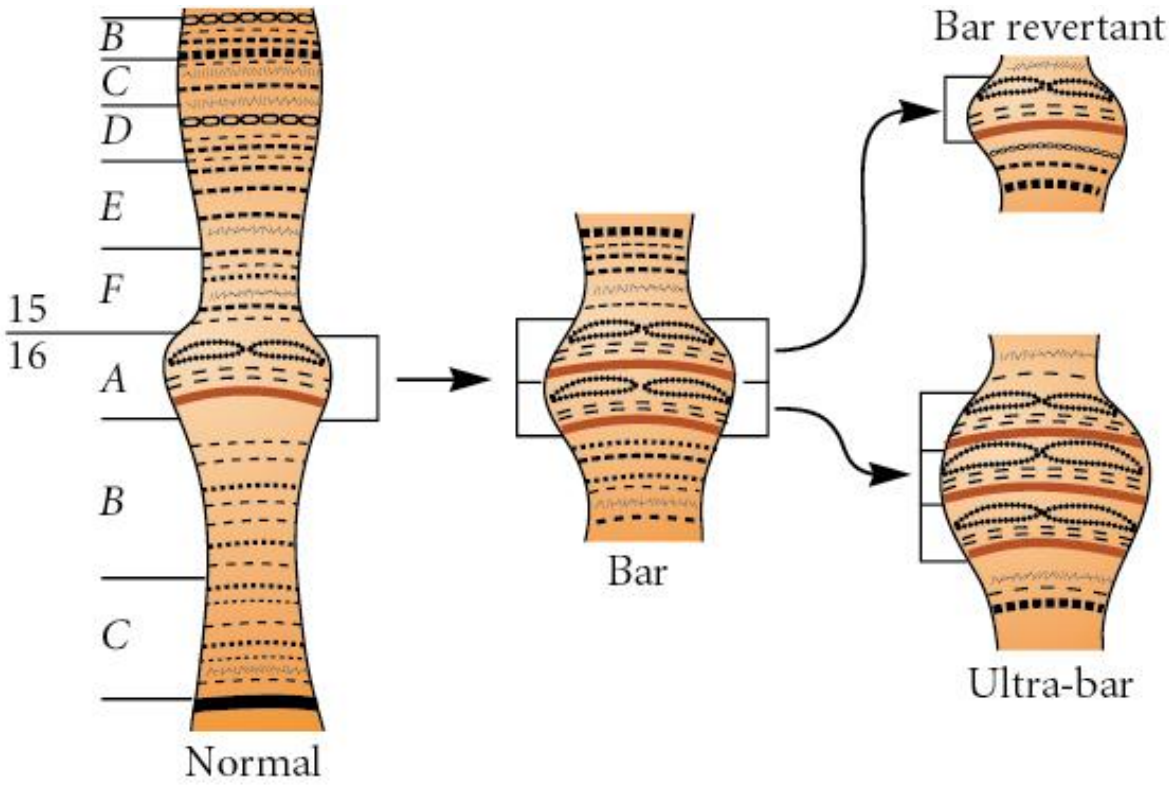
2. Dissect the salivary glands from the larva of a normal strain, a homozygous bar strain, a bar-revertant strain, and an ultra-bar strain.

3. Prepare the salivary cells for the microscopic examination of the polytene chromosomes. Note: The microscopic examination of chromosomes is described in chapter 3. It involves gently breaking open the cells, staining the chromosomes with dyes, and squashing the preparation on a microscope slide underneath a coverslip.

4. View the banding patterns of the polytene chromosomes under the microscope.

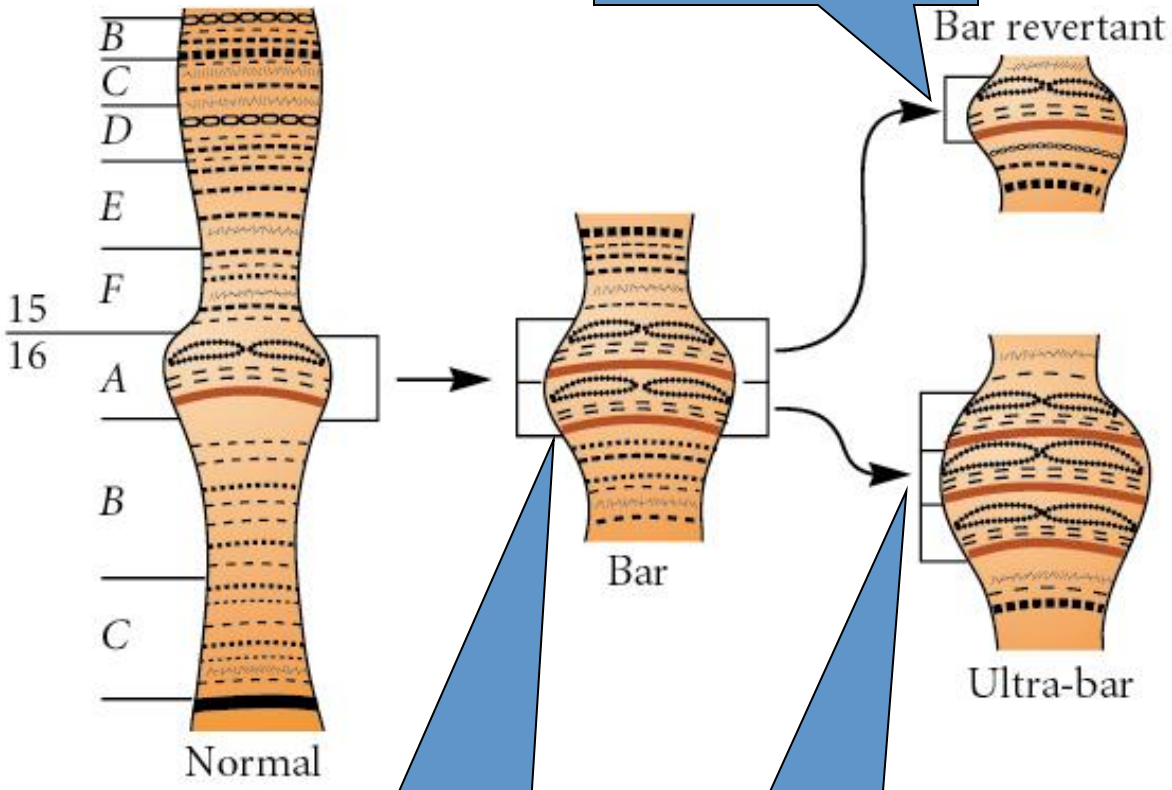


The Data



- This is a drawing of a short segment of a polytene chromosome that corresponds to the region of the X chromosome where the bar allele is located. This *bar* allele is found within the region designated *16A*

Interpreting the Data



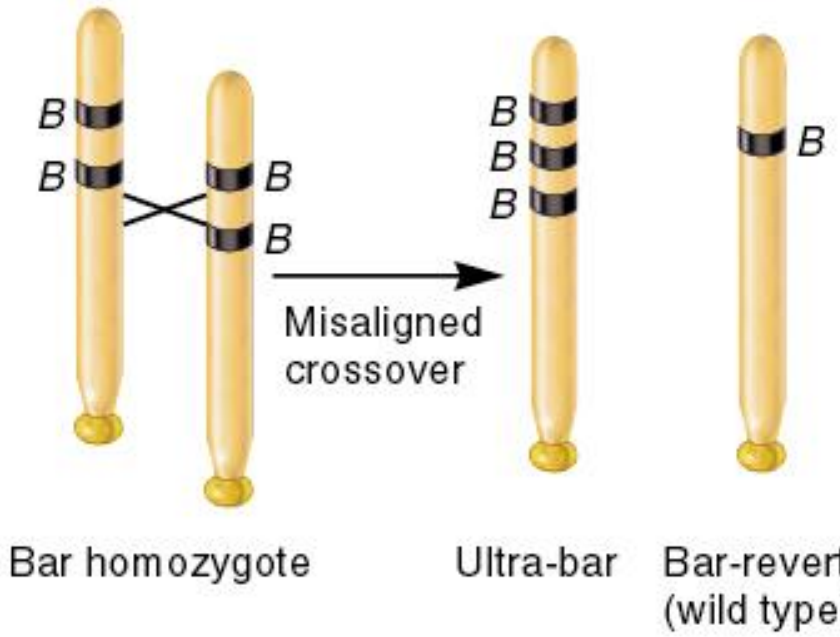
The 16A region duplication returned to the wild-type banding pattern

Bar phenotype is caused by a duplication in region 16A of the X chromosome

Ultra-bar phenotype is caused by three copies in the 16A region

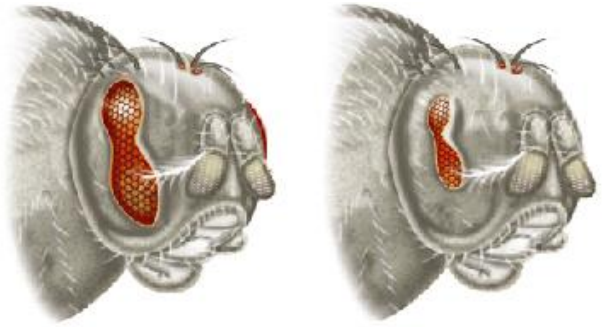
Interpreting the Data

- The mechanism of formation of the *bar* allele can be explained by a misaligned crossover
- Likewise for the formation of *ultra-bar* and *bar-revertant* alleles



Interpreting the Data

- The bar and ultra-bar alleles are also associated with the phenomenon of position effect
  - A female that is homozygous for the *bar* allele has four copies of region 16A
    - And 70 facets
  - A female that is heterozygous for the *ultra-bar* and normal alleles also has four copies of region 16A
    - But only 45 facets



	Bar homozygote	Ultra-bar heterozygote
Approx. number of facets	70	45

- The positioning of three copies next to each other on the X chromosome increases the severity of the defect