**Notes**

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**Unit VI: Alteration in chromosome number and structure.**

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# Position effect

Any phenotypic alteration as a result of chromosome rearrangement involving a change only in the order of the genes in the genetic material and involving no change in the amount of genetic material is referred to as position effect. No gene mutation (no change in the nature of gene) is involved. Due to shift of loci from one position to another, or due to shift of another segment in vicinity of a locus, expression changes.

Position effect are either stable, (Bar eye *Drosophila)* or variegated (*Drosophila* eye colour)

- ✓ **Stable position effects** - uniform phenotypic effects that result from changes of specific segments of chromosomes.
- ✓ **Variegated position effects** result in a mosaic pattern of a trait usually evidenced in a particular structure or area of the body.

Position effect - illustrated by following examples.

**Bar eye in Drosophila –** Sturtevant (1925)

**Duplications of the *Bar* locus of *Drosophila*** - Bar is a sex-linked dominant gene mutation that reduces the size of the eye. While normal wild type flies have round eyes, the X-linked mutation *Bar* (*B*) caused the eyes to be small and slit-like in males and homozygous females; female heterozygotes had kidney or bean-shaped eyes. Flies with the *double-Bar* mutation have even smaller eyes.

Normal (Wild type) ---- $B^+/ B^+$ ---------- 779 facets

Heterozygous bar eye ----- B/ $B^+$ ---------- 358 facets

Homozygous bar eye ------ B/B ---------- 68 facets

The *Bar* mutation was somewhat unstable: it tended to revert to wild-type spontaneously. Females (B/B) homozygous for *Bar* produced progeny with round eyes (wild type) at a frequency of ∼1/1000. A more extreme type was also produced called ultra-Bar (with only 24 facets) although with a lower frequency. Pure ultra-Bar also produced wild type eye with the same rate with which ultra - bar was obtained from them. They also may give rise to Bar. It was observed by Sturtevent that the frequency of these events was much higher than that was expected due to spontaneous mutations. He formulated a hypothesis and confirmed that new types arose due to unequal crossing over in the region of the Bar locus. Alfred Sturtevant found that in every *Double bar* fly, crossover had occurred between loci on either side of *Bar* locus. He suggested that the change to *Double bar* was due to unequal crossing over rather than to a simple mutation of one allele to another.

If the homologous chromosomes do not line up exactly during synapsis, a crossover produces an unequal distribution of chromosomal material. This mechanism is **unequal crossing-over**. This occurs when pairing occurs between homologous chromosomes in regions that are out of register. This pairing can be mediated by sequences that are repeated on the same chromosome. Following recombination (or crossing over) one chromosome will lose DNA sequence whereas the homologous chromosome will gain the sequences lost by the first.

Later, an analysis of the banding pattern of the salivary gland chromosomes by Bridges (1936) confirmed Sturtevant's hypothesis. It was found that Bar is a duplication of several bands in the 16A region of the X chromosome. Double bar is the triplication of the segment.

About 10 years after Sturtevant's paper, cytological studies by Bridges (1936), in the context of interpretations by Muller (1936), confirmed that the *Bar* mutation was indeed a tandem duplication.

A Bar homozygote (B/B) and a double bar/ wild type heterozygote (BB/ $B^+$), both have 4 copies of the 16 A region. It would therefore be reasonable to expect that both genotypes would produce the same phenotype. However, the *Bar* homozygote has about 70 facets in each eye, whereas the heterozygote has about 45 facets. Thus, not only the amount of genetic material but also its configuration, determines the extent of phenotype.

**Variegated position effects**

- ✓ Eg. Specks of different colours may occur in the eyes of *Drosophila* due to rearrangements of the w (white eye) locus.
- ✓ Inversions or translocations that place w+ in heterochromatin may cause white variegation or mosaic pattern for eye colour.
- ✓ Action of gene is depressed when transferred to a heterochromatin region (suppression of gene transcription).
- ✓ Involves two chromosome breaks, one in the heterochromatin region and the other near a euchromatic gene.
- ✓ X-ray treatment caused the X chromosome in some of the flies to break in two places, the middle piece inverted and the chromosome ends were re-joined. One of the breaks occurred in the heterochromatin surrounding the centromere and the other was near to the *white*$^+$ ($w^+$) gene. The $w^+$ gene makes a product that is necessary for the deposition of the eye pigments. When this gene functions normally it results in the normal dark red eye of fruit flies, but if it is inactive then the fly's eye is white. As a result of the inverted piece of the chromosome, the $w^+$ was moved from its normal position, toward the end of the X chromosome, to a position very close to (about 25000 bp from) the new heterochromatic breakpoint. In this position, the gene is expressed in some cells and silenced in others, and thus gives rise to the red and white mosaic pattern.