# DELETION

**Q.What do you mean by structural aberration of chromosome?**

Structural aberrations refer to the cases where normal chromosome numbers of karyotype usually remains unchanged but their genetic composition is altered through loss, gain & rearrangement of particular chromosomal segments involving breaks. Possible causes for chromosomal aberrations include, i)loss of the acentric fragment, ii)restitution of fragments, iii)Exchange or non-restitutional union between fragments of nonhomologues.

**Q.What are the major types of structural aberration found for chromosome?**

A)Deletion, B)Duplication, C)Inversion, D)Translocation.

**A)Deletion:**

**Q.What is deletion? Distinguish between interstitial and terminal deletion. Give suitable example for each.**

Deletion refers to loss of genetic material in the form of chromosomal piece (s) from the genome. The chromosome with missing piece is called deleted/ deficient elements and the affected individual is called deleted heterozygote. This kind of radiation, of which X rays and γ rays are examples, is highly energetic and causes chromosome breaks. The way in which the breaks re-join determines the kind of rearrangement produced.

a)**Terminal deletion:**

i)Deletion at any end of the chromosome involving a single break.

ii)CRI-DU-CHAT syndrome in man develops due to loss of terminal one-half of the short arm of chromosome-5 (J.Lejeune, 1963). (**Figure1**)

**b)Intercalary or interstitial deletion:**

Deletion from the interior of a chromosome involving at least two breaks. E.g. Notch-wing phenotype in *Drosophila*. (**Figure 1**)


**Q.What do you mean by multigenic and intragenic deletion?**

(a)The effects of deletions depend on their size. A small deletion within a gene, called an **intragenic deletion,** inactivates the gene and has the same effect as other null mutations of that gene. If the homozygous null phenotype is viable (as, for example, in **human albinism**), then the *homozygous deletion* also will be viable. Intragenic deletions can be distinguished from single nucleotide changes because they are *nonrevertible*.

(b)For most of this section, we shall be dealing with multigenic deletions, those that remove from two to several thousand genes. **Multigenic deletions** have severe consequences. If by inbreeding such a deletion is made *homozygous* (that is, if both homologs have the same deletion), then the combination is *almost always lethal*. This outcome suggests that most regions of the chromosomes are essential for normal viability and that complete elimination of any segment from the genome is deleterious.

**Q.Mention possible consequences of deletion on phenotypic variation, lethality, dominance.**

1.The consequences of a deletion depend on the genes or parts of genes that have been removed. In diploid organisms, an individual heterozygous for a deletion may be normal. However, if the homolog contains recessive alleles that have deleterious effects, the consequences can be severe.

2.Phynotypic variation—

Alteration of phenotypic characters due to absence of concerned genes (present in deleted parts).

3.Lethality—

Occurs when deletion is large and involves many loci & homozygous, for autosomal loci and hemizygous for sex-linked locus.

4. If the deletion involves the loss of a centromere, the result is an acentric chromosome, which is usually lost

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