Genetics

Biology Dept.3rd stage

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Lec-9- STRUCTURAL CHANGES IN CHROMOSOMES

2. Duplication

Duplication. The presence of a part of a chromosome in excess of the normal complement is known as duplication. A broken part of chromosome attaches itself to a normal homologus chromosome or non-homologus chromosome. Thus, due to duplication some genes are present in a cell in more than two doses. The effects of duplications are generally less harmful than those of deletions. Depending upon the mode of joining of the duplicated region to a chromosome, duplication can be of following types:

- **Tandem.** Here the duplicated region is situated just side by side of the normal corresponding section of the normal chromosome and the sequence of genes are the same in the normal and duplicated regions. e.g.: if the normal chromosome has gene sequence ABCD•EFGH (the point shows the position of centromere), the gene sequence in tandem duplication will be ABCDBCD•EFGH.
- **Reverse.** In this case, the sequence of genes in the duplicated section of a chromosome is just the reverse of the normal sequence. e.g.: if the normal chromosome has gene sequence ABCD•EFGH, the gene sequence in reverse duplication will be ABCDDCB•EFGH.
- **Displaced.** Here, the duplicated section is situated adjacent to the normal, but elsewhere in the chromosome.
- The additional chromosome segment is located in a non-homologous chromosome is **translocation duplication.**

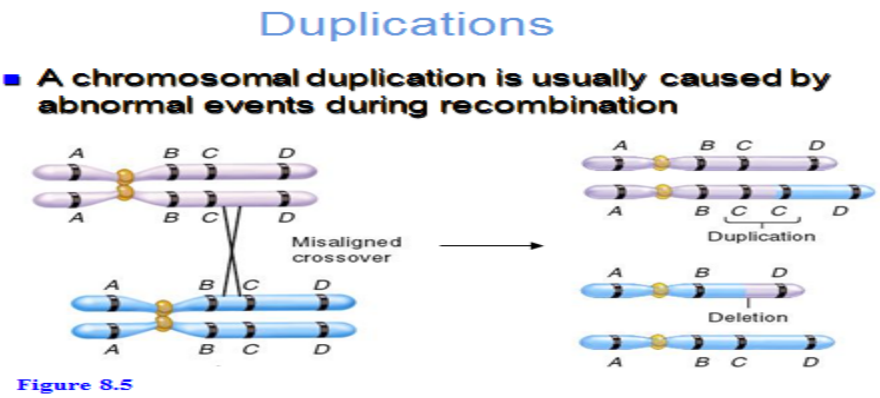

Figure 8.5

3- Translocation.

It involves the transfer of a chromosome section or a set of genes to a non-homologus chromosome. It results in the change in sequence and position of genes (rearrangement) but not in their number (no addition or deletion). e.g.: if the original chromosomes were ABCD•EFGH and PQRS•TUVW, the new ones may be ABCD•EUVW and PQRS•TFGH. Translocation are of following types:

(a) Simple. This involves a single break in a chromosome and the broken part gets attached to one end (terminal) of a non-homologus chromosome. e.g.:

ABC•DEF ⟶ C•DEF
MN•OPQ ABMN•OPQ

(b) Reciprocal. In this case, there is an exchange of chromosome part between two non-homologus chromosomes, so that two translocation chromosomes are formed simultaneously. This is the most frequent type of translocation, and is of two types- **homozygotic**, in which both the homologus chromosomes are involved and **heterozygotic**, in which only one chromosome of a pair of homologus chromosomes is involved. In humans, **Philadelphia chromosome** is an example of translocation, occurring between 22q and 9q (long arm) characterized by ***chronic myelocytic leukemia***, a kind of cancer. e.g.:

ABC•DEF ⟶ MNC•DEF
MNO•PQR ABO•PQR

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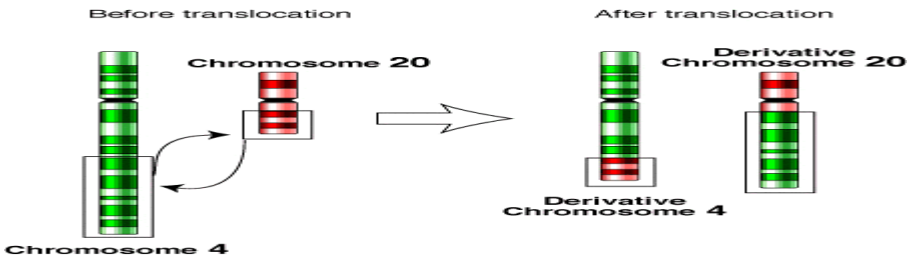
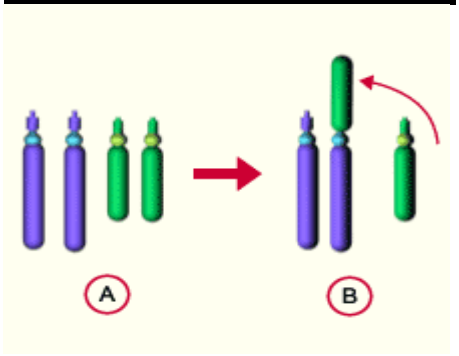


(c) Robertsonian. It also involves the exchange of chromosome segments between two non-homologus chromosomes, but in this case, whole arm of a chromosome is transferred to the other chromosome. In humans, they are the most common structurally abnormal chromosomes. e.g.:

ABC•DEF ⟶ MNO•DEF
MNO•PQR ⟶ ABC•PQR

Robertsonian translocations are phenotypically inconspicuous. Also here, though, problems arise when it comes to **gamete formation** because, normally, the diploid chromosome set is halved thereby. Since, however, in this translocation a chromosome has fused to another one, no ordered segregation can take place. The carrier has a larger probability of having offspring with trisomy/monosomy and this is independent of his age. Often a translocation (e.g., t[14q21q]) is found in families with inherited **trisomy 21.**

- Robertsonian translocation of acrocentric chromosomes



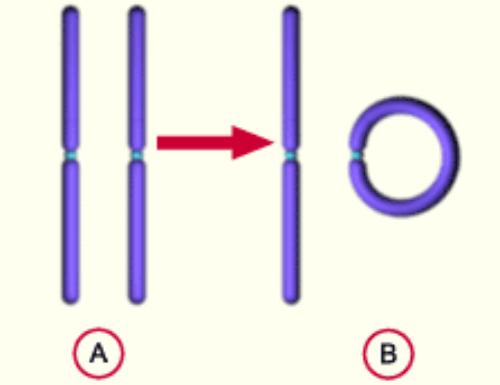
A normales
B Chromosomenpaar centric fusion of two non-homologous chromosomes

Fig.
In a reciprocal translocation two acrocentric chromosomes lose their short arms. Afterwards, the remaining partial pieces (q- arms) fuse to one another.

4- Ring Chromosome

- Ring formation

Ends (telomeres) of chromosomes sometimes break off and are lost. In this case the **formation of chromosome rings** can take place in that the two ends bind to one another



5- Inversion.

It involves rotation of a part of chromosome or a set of genes by 180˚ on its own axis. *Breakage* and *reunion* are essential for inversion. The net effect is neither a gain nor a loss in the genetic material but simply a rearrangement of the gene sequence. e.g.: a chromosome having a gene sequence ABCDEF will have a gene sequence of AB<u>EDC</u>F after inversion. Inversions are of two types on the basis of centromere in relation to the inverted segment.

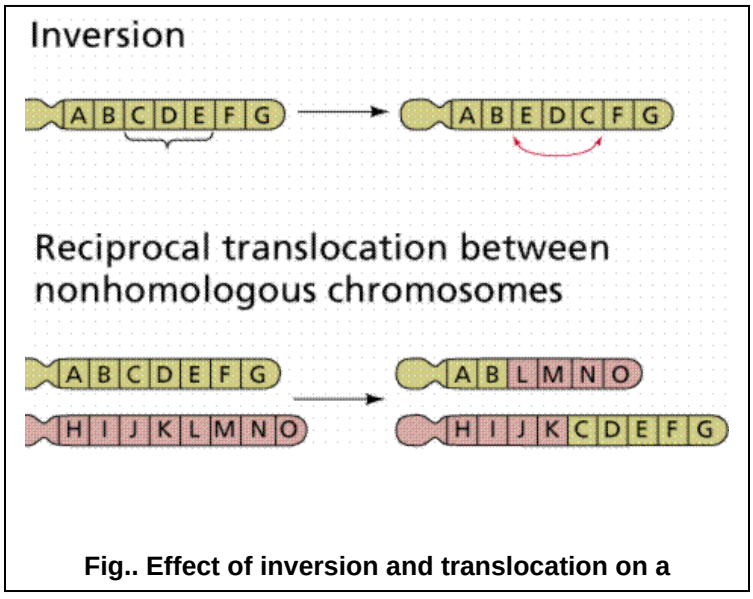


Fig.. Effect of inversion and translocation on a

(a) Paracentric. In this type the inverted segment does not include the centromere, i.e., the centromere is located outside the inversion loop. e.g.: if the normal gene

sequence is ABCD•EFGH, the sequence in paracentric inversion will be ABCD•EGFH. Paracentric inversions are *very rare* because it is less probable for two breaks to be in the same arm. Further they are difficult to detect, since they do not change the length of chromosome.

(b) Pericentric. Here, centromere is involved or located in the inversion loop. e.g.: if the normal gene sequence is ABCD•EFGH, the sequence in pericentric inversion will be ABFE• DCGH.

Differences between chromosomal mutation and gene/point mutation

Chromosomal mutation	Gene/point mutation
A process that cause changes in chromosome structure or number	A process that causes changes in the base sequence of the DNA
Types of chromosomal mutation are chromosomal aberration and alteration in chromosome number.	Types of gene mutation are base substitution, base inversion, base deletion and base insertion.
No frameshift mutation	Base deletion and base insertion cause frameshift mutation
No production of non-functional protein	Leads to production of non-functional protein