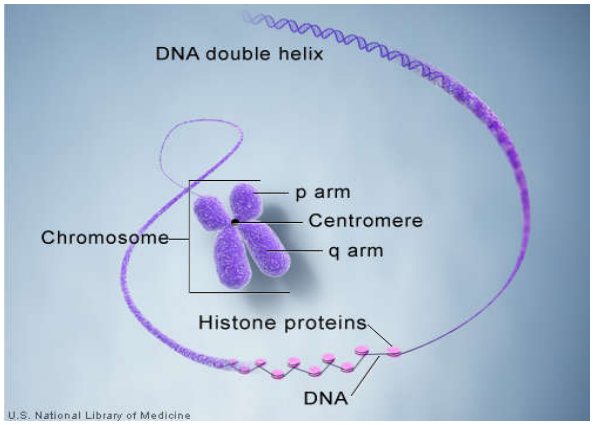
Variation in Chromosome Number and Structure

1. **What is chromosome?**

A long DNA molecule with all of the genetic material of living organisms which tightly coiled many times around the histone proteins within the nucleus called Chromosomes.

Each chromosome has a constriction point which is known as centromere. The location of the centromere on each gives a characteristics shape and can be used to describe the location of genes. Centromere divides chromosome into two arms. The long arm labeled as "q" arm and the short arm labeled as "p" arm.



2. **What are chromosome mutations?**

Each organism has a characteristics number of chromosome with a distinct size and structure. However there can be found chromosomal disorder or aberration regarding the chromosome number and structure. These abnormalities arise from beginning of the error in the normal cell which is called chromosomal mutation.

3. **What do you mean by position effect?**

Position effect is the gene expression when its location is changed in a chromosome by translocation and inversion. These gene expressions enhance the expression of neighboring genes. Fore example, Position-Effect Variegation is caused by gene silencing in some cells due to abnormal juxtaposition with heterochromatin via rearrangement and transposition and also associated with changes in chromatin conformation.

4. **What are the types of chromosome mutations?**

Chromosomal mutations on the basis of chromosome rearrangement:

a. Duplication –

Duplication is the process of mutation that involves production of one or more copies of gene or part of chromosome through which new genetic material generated during molecular evolution.

Fore example, *Drosophila melanogaster*, fruit flies having **bar mutation** due to reduce number of facets in the eye. It makes eye smaller and bar shaped instead of oval.

The human genome consists of several duplication sequence which is known as segmental duplication that are greater than 1000bp in length. When segmental duplication found on the same chromosome is called **intrachromosomal** duplication but the segmental duplication when found in different chromosomes is called **interchromosomal** duplication.

b. Deletion –

Deletion is a second type of mistake in DNA replication process which removes single nucleotide or entire sequence by base flipping in the template DNA. It can be caused by crossing over during meiosis. Deletion is responsible fore numerous genetic diseases such as **cri du chat syndrome (**chromosome 5 is responsible**), cystic fibrosis, male infertility** etc. Three types of deletions are **terminal deletion (**occurs towards the end of chromosome**), intercalary deletion (**occurs from the interior of chromosome**), microdeletion (**a small amount of deletion up to 5mb**)**.

c. Inversion –

Inversion is a third type of mutation in which chromosome segment is inverted or turns 180 degrees. If inversions do not include centromere are termed as paracentric inversion whereas inversions include centromere are termed as pericentric inversion. Inversion is the process where neither loss nor gain any genetic material, just the DNA sequence change their position. Human chromosome 4 differs from chimpanzees in a pericentric inversion.

d. Translocation –

Translocation is the movement of genetic material within same chromosome or between nonhomologous chromosomes. Translocation includes reciprocal, nonreciprocal and robertsonian tranlocation. **Reciprocal translocation** involve in a two way exchange of segments between the chromosomes. In case of reciprocal translocation, chromosome **AB•CDEFG** and **MN•OPQRS** might give rise to chromosomes **AB•CDQRG** and **MN•OPEFS**. In case of **non reciprocal translocation** involve in a movement of genetic material from one chromosome to another without any reciprocal exchange. If nonreciprocal translocation has taken place between two nonhomologous chromosomes **AB•CDEFG** and **MN•OPQRS**, then it produces chromosomes **AB•CDG** and **MN•OPEFQRS**. **Robertsonian translocation** is caused by breaks at or near the centromeres of two acrocentric chromosomes, generating a metacentric chromosome with two long arms and another chromosome with two very short arms.

Chromosomal mutations on the basis of chromosome number:

Euploidy –

Euploidy is the basic set of chromosome number. If an organism have basic set of chromosome number 7, may have euploids with chromosome number 7,14,21,28. Euploids are consists of three types – monoploid (single set of genome), diploid (two sets of genome) and polyploidy (more than two sets of genome). For example, Barley have 2n= X=7 (haploid of diploid species).

Aneuploidy –

Organisms have one or a few chromosomes above or below the normal set of chromosome number. A cell with incomplete set of chromosome is called aneuploid cell. Aneuplody causes several genetic disorder. Types of aneuplody are as follows –

Monosomy refers to lack of one chromosome (45,X) shows **turner syndrome** in human.

Disomy is the presence of two copies of each chromosome. It is the normal condition.

Trisomy refers to the presence of three copies instead of normal set of chromosome. For example down syndrome is called trisomy possess chromosome 21.

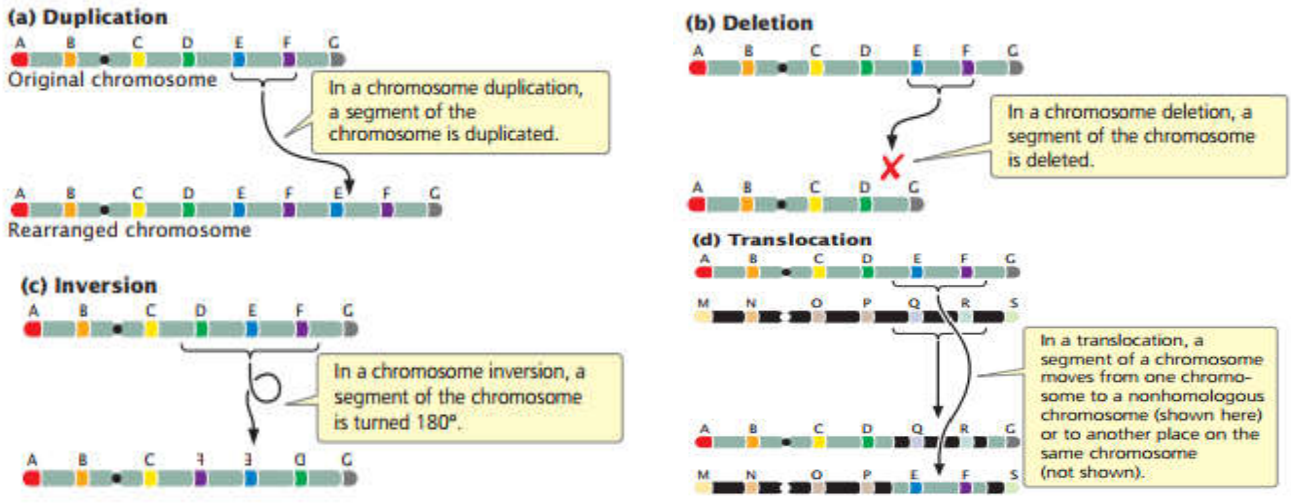


Fig1: Images of Duplication, Deletion, Inversion and Translocation.

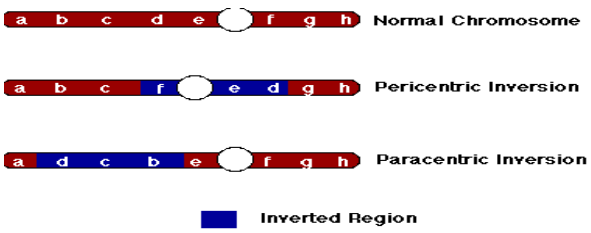


Fig2: Pericentric and Paracentric Inversion.

References:

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(All the informations are collected from above references and will be used only for teaching and learning purposes)