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Faculty of Pharmacy
Second year

Chromosomal Abnormalities

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Introduction

Chromosomal abnormalities result from mutations which change the number of chromosomes (numerical abnormalities) or change the structure of the chromosome (structural abnormalities). They may alter the ability of the cell to survive and function. Chromosomal abnormalities are common causes of birth defects that can affect the brain and other parts of the body. The normal fertilized egg cell contains 23 chromosomes from the mother and 23 from the father. Thus, there are normally 23 pairs of chromosomes in the fertilized egg. These include two sex chromosomes: XX for girls and XY for boys. Some chromosomal abnormalities occur when there is an extra chromosome.

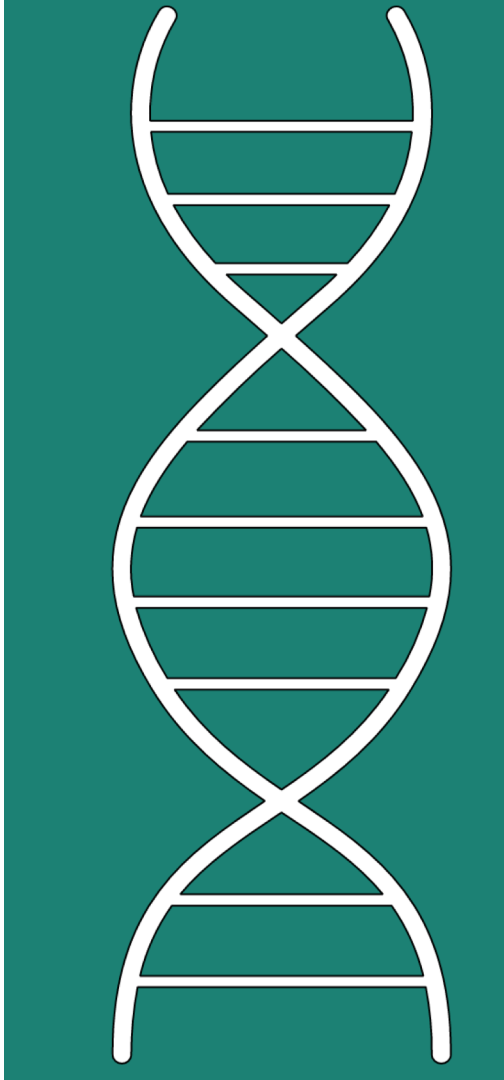
Causes of chromosome abnormalities

Chromosome abnormalities usually occur when there is an error in cell division. There are two kinds of cell division, mitosis and meiosis.

Mitosis results in two cells that are duplicates of the original cell. One cell with 46 chromosomes divides and becomes two cells with 46 chromosomes each.

Meiosis results in cells with half the number of chromosomes, 23, instead of the normal 46.

In both processes, the correct number of chromosomes is supposed to end up in the resulting cells. However, errors in cell division can result in cells with too few or too many copies of a chromosome. Errors can also occur when the chromosomes are being duplicated.



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Other factors that can increase the risk of chromosome abnormalities are:

Maternal Age: Women are born with all the eggs they will ever have. Some researchers believe that errors can crop up in the eggs' genetic material as they age. Older women are at higher risk of giving birth to babies with chromosome abnormalities than younger women. Because men produce new sperm throughout their lives, paternal age does not increase risk of chromosome abnormalities.

Environment: Although there is no conclusive evidence that specific environmental factors cause chromosome abnormalities, it is still possible that the environment may play a role in the occurrence of genetic errors.

Types of abnormalities

Numerical abnormality

A numerical abnormality mean an individual is either missing one of the chromosomes from a pair or has more than two chromosomes instead of a pair.

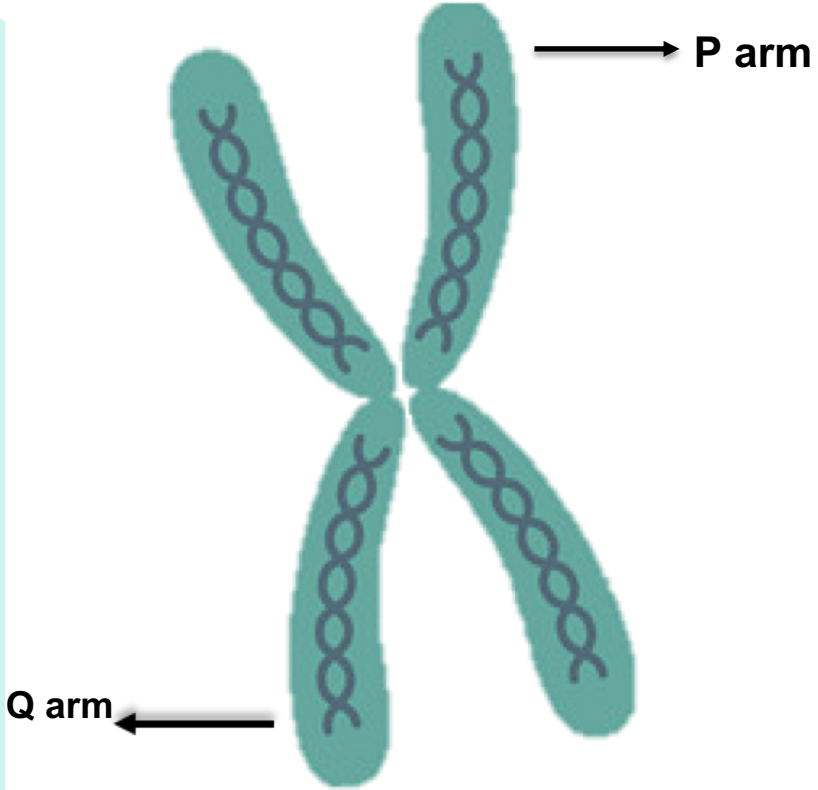
Structural abnormality

Structural chromosome abnormalities occur when there is a change in the structure or parts of a chromosome.

Structural abnormalities

Structural chromosome abnormalities result from a break or breaks that disrupt the continuity of a chromosome, followed by reconstitution in an abnormal combination.

Structural abnormalities occur when the chromosomal morphology is altered due to an unusual location of the centromere and therefore abnormal lengths of the chromosome's short (p) and long arm (q).

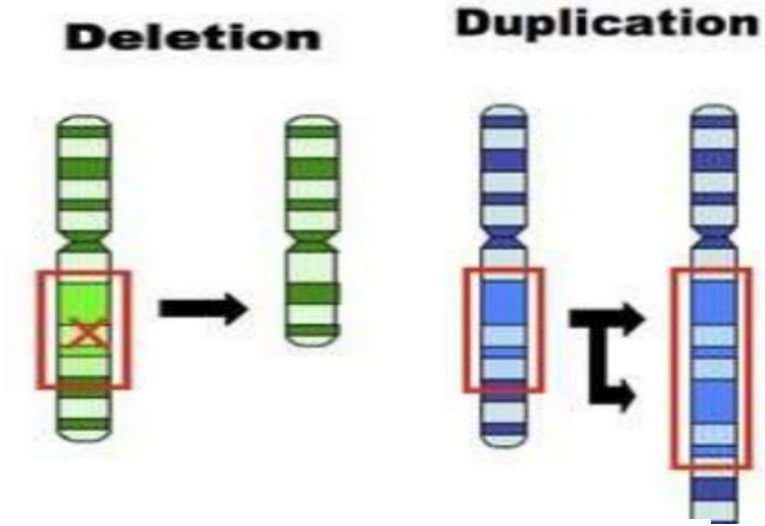


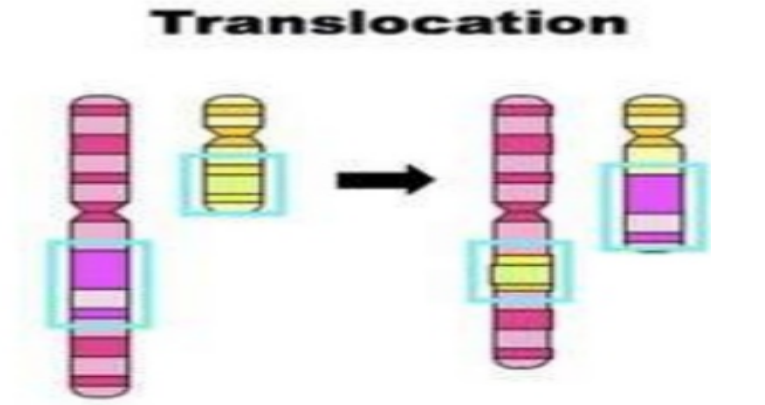
Structural abnormalities

Deletions: A portion of the chromosome is missing or deleted.

Duplications: A portion of the chromosome is duplicated, resulting in extra genetic material.

Translocations: A portion of one chromosome is transferred to another chromosome



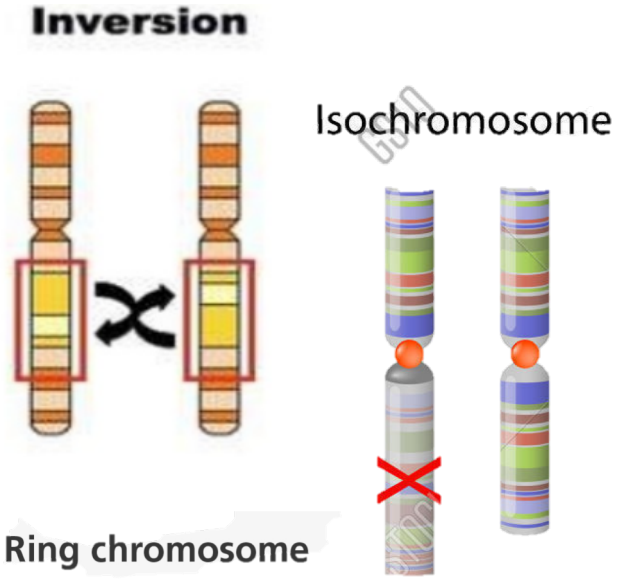


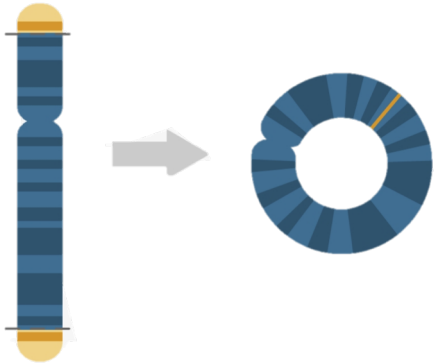
Structural abnormalities

Inversions: A portion of the chromosome has broken off, turned upside down, and reattached. As a result, the genetic material is inverted.

Rings: A portion of a chromosome has broken off and formed a circle or ring. This can happen with or without loss of genetic material.

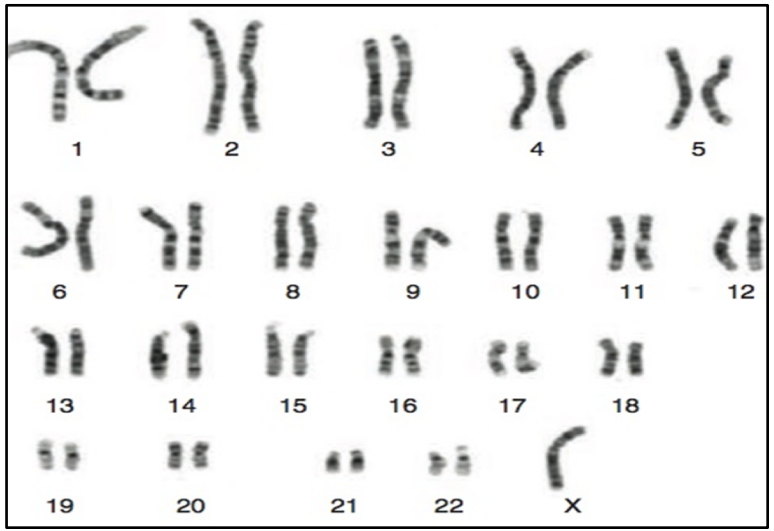
Isochromosome: is an inverted duplication of one arm of a chromosome with loss of the other arm.





Numerical abnormalities

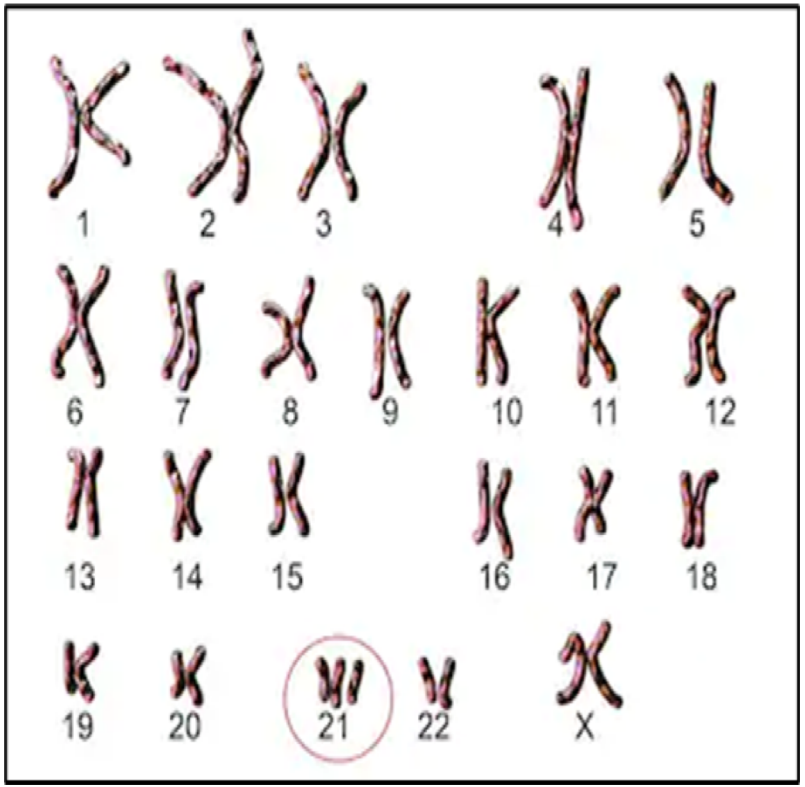
Monosomy: Monosomy is the state of having a single copy of a chromosome pair instead of the usual two copies found in diploid cells. Monosomy can be partial if a portion of the second chromosome copy is present. Monosomy, or partial monosomy, is the cause of some human diseases such as Turner syndrome.



Numerical abnormalities

Trisomy: An individual has three chromosomes instead of a pair For example, if a baby has Down syndrome they, in most cases, have three copies of chromosome 21, rather than the usual pair, and the condition is thus called "trisomy 21."

Other common trisomy's are trisomy 18 and trisomy 13, which means that there are three copies of chromosome 18 or 13, respectively, in each cell of the body, rather than the usual pair.



Summary

- In conclusion chromosome abnormality is a missing, extra, or irregular portion of chromosomal DNA.
- These can occur in the form of numerical abnormalities, where there is an atypical number of chromosomes, or as structural abnormalities, where one or more individual chromosomes are altered.
- Chromosome anomalies usually occur when there is an error in cell division following meiosis or mitosis.
- Chromosome abnormalities may be detected or confirmed by comparing an individual's karyotype, or full set of chromosomes, to a typical karyotype for the species via genetic testing.

Reference

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