

# Chromosomal abnormalities

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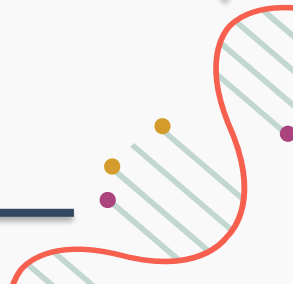
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***At the end of this presentation you will be able to:***

- 1. Discuss Chromosome abnormalities and their types.**
- 2. Explain Causes of Chromosome abnormalities**
- 3. Mention Two Examples Caused by teratogens**

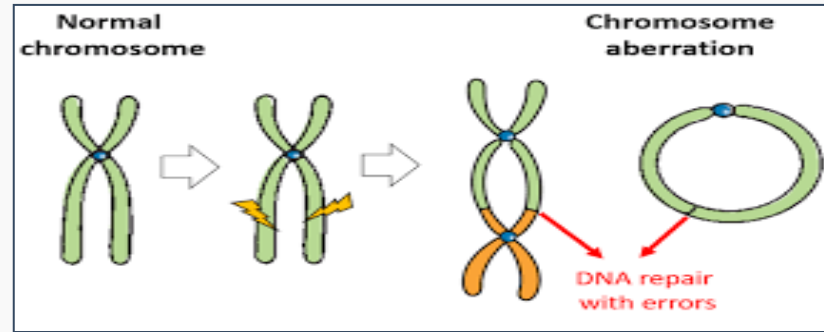


# Genetic disorders traditionally fall into three main categories:

single-gene defects, chromosomal abnormalities, and multifactorial conditions.

## chromosomal abnormality :

is a disorder characterized by a morphological or numerical alteration in single or multiple chromosomes effect autosomes, sex chromosomes or both.



## Types



**Numerical Abnormalities:** When an individual is missing one of the chromosomes from a pair, the condition is called monosomy. When an individual has more than two chromosomes instead of a pair, the condition is called trisomy.



Numerical disorders are considerably more common than structural ones

**structural abnormalities :** a chromosome structure can be altered in several ways 1- deletion , duplication , translocation , inversion , ring  
Most structural abnormality can occur as an accident in the egg or sperm





# • Structure abnormalities

# Unbalanced structure

## Deletions:

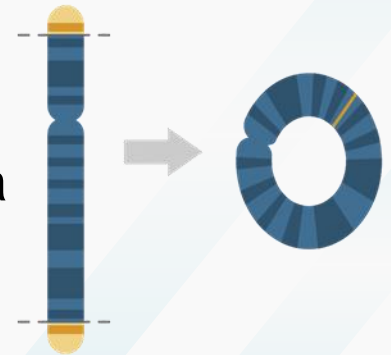
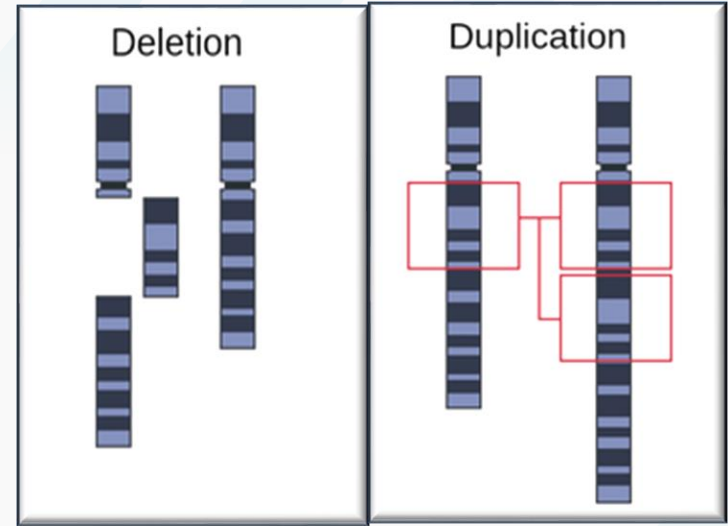
A portion of the chromosome is missing or deleted.

## Duplications:

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

## 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.



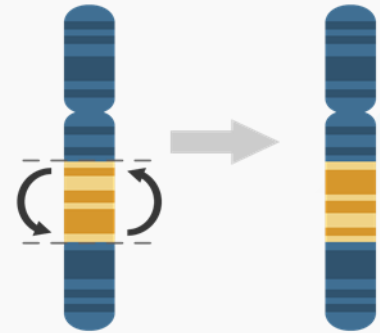
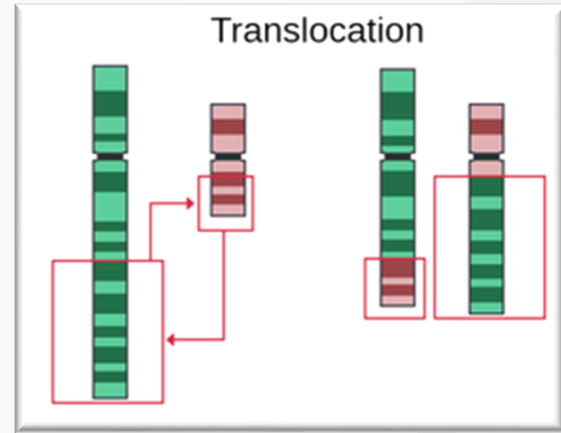
## ↙ **Balanced structure**

### **Translocations:**

A portion of one chromosome is transferred to another chromosome.

### **Inversions:**

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



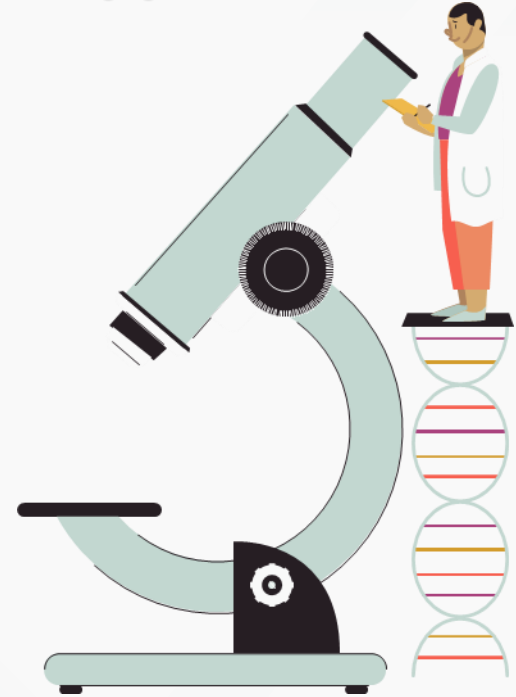


- **Causes abnormalities**



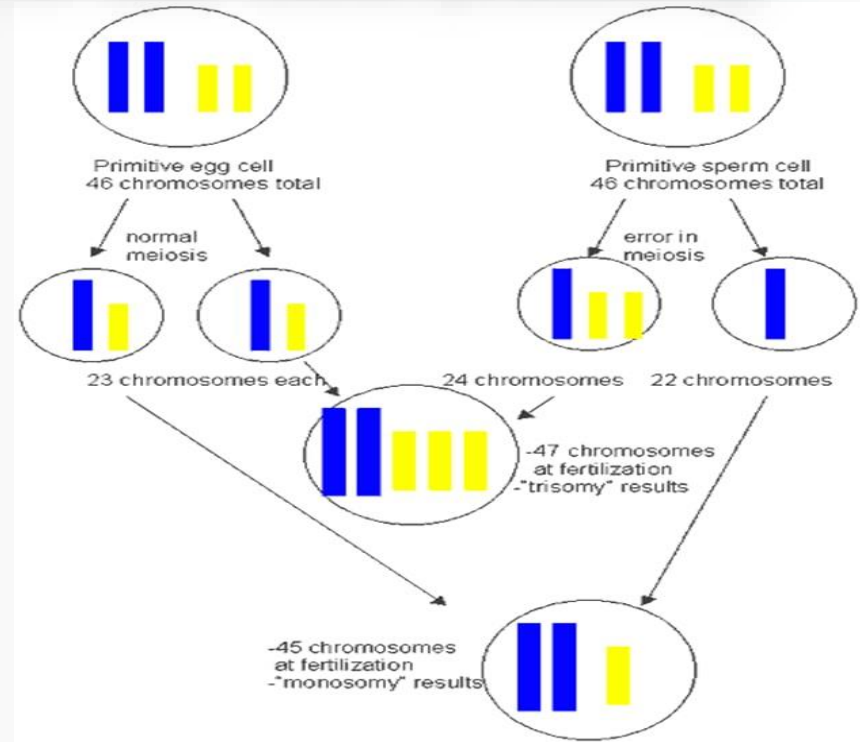
# Chromosome abnormalities often happen due to 1 or more of these:

- Errors during dividing of sex cells (meiosis)
- Errors during dividing of other cells (mitosis)
- Other causes of birth defects (teratogens)



# Errors during dividing of sex cells (meiosis)

pregnancies with a trisomy or a monosomy may go to full-term and result in the birth of a child with health problems, it is also possible that the pregnancy may miscarry, or that the baby is stillborn (not born alive), because of the chromosome abnormality



# Errors during dividing of other cells (mitosis)

- Mitosis is the dividing of all other cells in the body. This process repeats itself, until the entire baby is formed. Mitosis continues throughout our lifetime, to regenerate new skin cells, new blood cells, and other types of cells that are damaged or that simply die off.
- During pregnancy, an error in mitosis can occur, just like the error previously described in meiosis. If the chromosomes do not split into equal halves, the new cells can have an extra chromosome (47 total) or have a missing chromosome (45 total). This is another way a baby can be born with a chromosome abnormality.

# Substances that cause birth defects (teratogens)

- Some medicines
- Street drugs
- Alcohol
- Toxic chemicals
- Some viruses and bacteria
- Some kinds of radiation
- Certain health conditions, such as uncontrolled diabetes

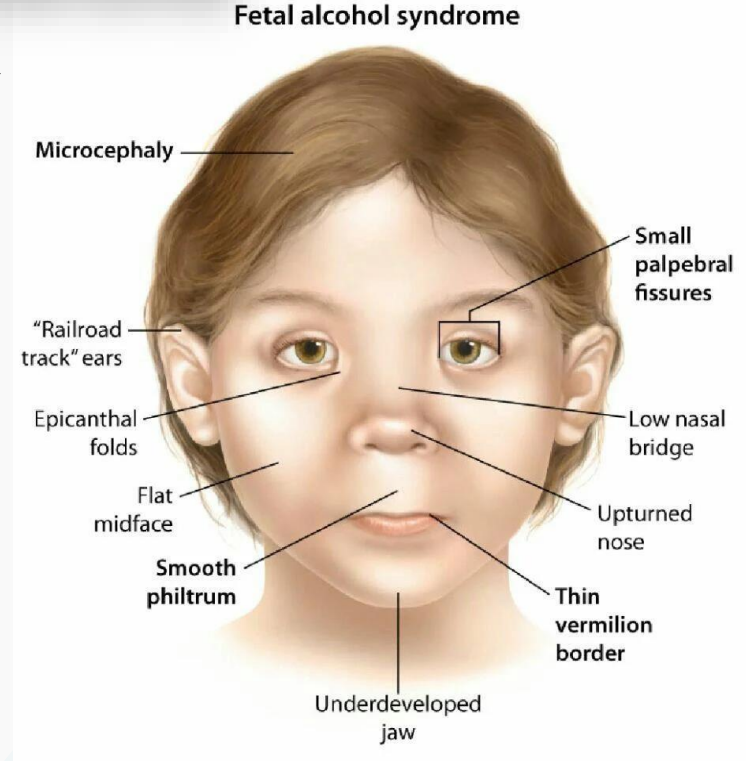


# Examples Teratogens



# Fetal alcohol syndrome

Fetal alcohol syndrome is a condition in a child that results from alcohol exposure during the mother's pregnancy. Fetal alcohol syndrome causes brain damage and growth problems. The problems caused by fetal alcohol syndrome vary from child to child, but defects caused by fetal alcohol syndrome are not reversible.



## Thalidomide syndrome

Thalidomide, a sedative used in treatment of a range of conditions, including morning sickness. Thalidomide embryopathy is characterized by phocomelia (Without limbs)

Thalidomide led to the death of approximately 2,000 children and serious birth defects in more than 10,000 children, with over half of them in Germany. In 1961, thalidomide was taken off the market.





# summary



- Discussed chromosomal abnormalities DNA repair with errors It is a disorder characterized by morphological or numerical change .
- Types:Numerical abnormality When an individual is missing one of the chromosomes from a pair, a chromosome structure can be altered in several ways 1- deletion , duplication , translocation , inversion , ring
- Different Causes of chromosomes abnormality
- Also we mentioned Fetal alcohol syndrome is a condition in a child that results from alcohol exposure during the mother's pregnancy, and thalidomide syndrome.

# Resources for research and reference

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