



Chromosomal Abnormality

Present by:

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ILOs

01. Describe chromosomal abnormality.

02. List causes of chromosomal abnormality.

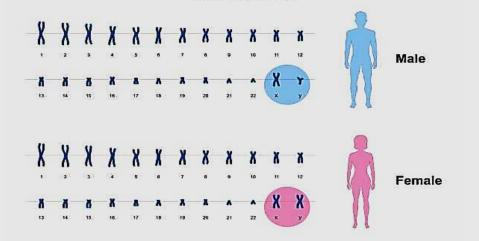
03. Describe types of chromosomal abnormality with example.

Introduction

Almost every cell in our body contains 23 pairs of chromosomes, for a total of 46 chromosomes.

Each chromosome contains thousands of genes, which make proteins

that direct the body's development, growth, and chemical reactions.



CHROMOSOME

01. Describe chromosomal abnormality

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What is chromosomal abnormality :

- A chromosome anomaly, abnormality of aberration is a missing, extra, or irregular pursion of chromosomal DNA
- It can be from an atypical number of chromosomes or a structural abnormality in one or more chromosome
- When a chromosome is abnormal, it can cause health problems in the body

02. List causes of chromosomal abnormality

Causes of chromosomal abnormality:

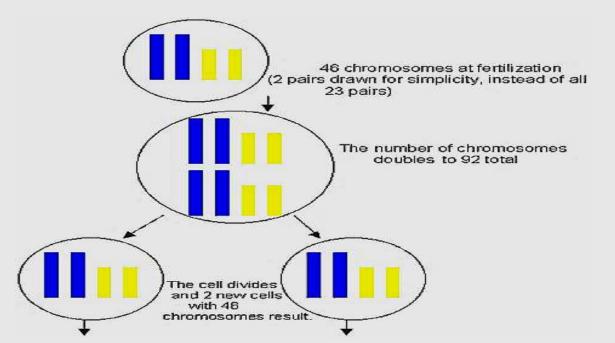
Abnormal chromosomes most often happen as a result of an error during cell division due to one or more of these:

- Errors during dividing of sex cells (meiosis)
- Errors during dividing of other cells (mitosis)
- Exposure to substances that cause birth defects (teratogens)



Mitosis is a process where a single cell divides into two

identical daughter cells (cell division)



Mitosis :

Abnormal mitosis exerts indirect effects on the future stability of the genome.

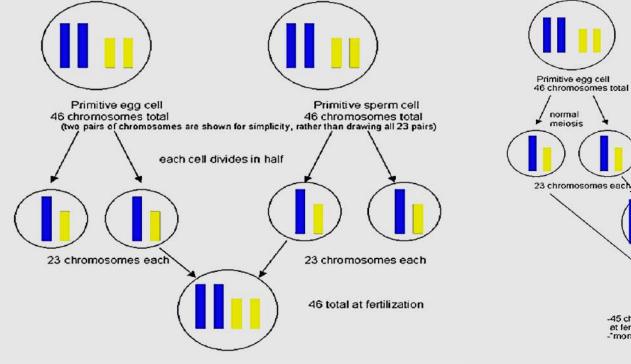
an error in mitosis can cause the chromosomes do not split into equal halves, the new cells can have an extra chromosome (47 total) or have a missing chromosome (45 total).

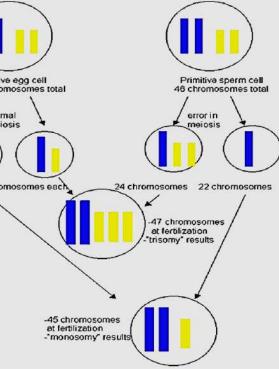


Meiosis:

Meiosis is the process in which sex cells divide and create new sex cells. Meiosis is the start of the process of how a baby grows. It causes each parent to give 23 chromosomes to a pregnancy.

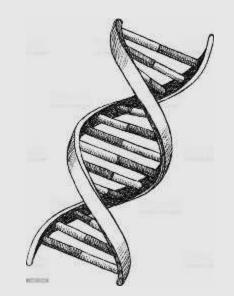
Causes of chromosomal abnormality:





Teratogens:

- They are things that a mother may be exposed to during her pregnancy Teratogens include:
- 1. Some medicines
- 2. Street drugs
- 3. Alcohol
- 4. Tobacco
- 5. Toxic chemicals
- 6. Some viruses and bacteria



03. Describe types of chromosomal abnormality

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Types of chromosomal abnormality:

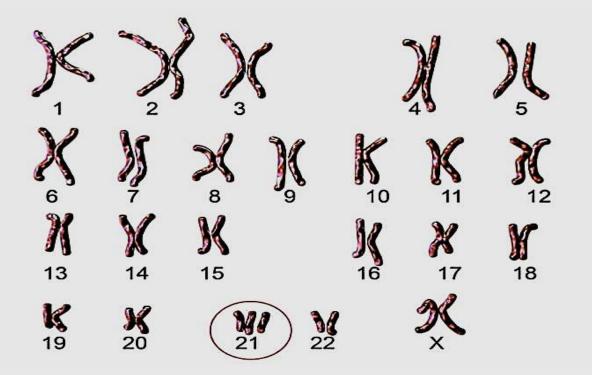
There are many types of chromosome abnormalities, they can be organized into two basic groups:

• Numerical disorder

This is called aneuploidy (an abnormal number of chromosomes), and occurs when an individual is missing either a chromosome from a pair (monosomy) or polyploidy has more than two chromosomes of a pair (trisomy, tetrasomy etc.)

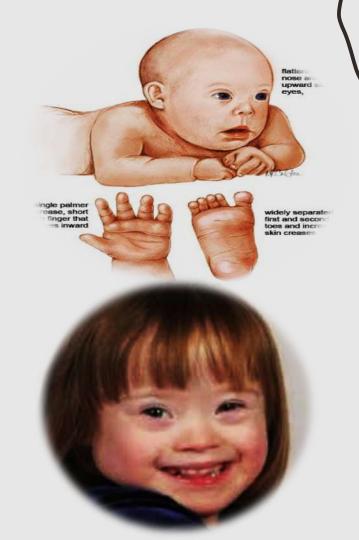
Example about numerical abnormality:

Down syndrome (21 Trisomy)



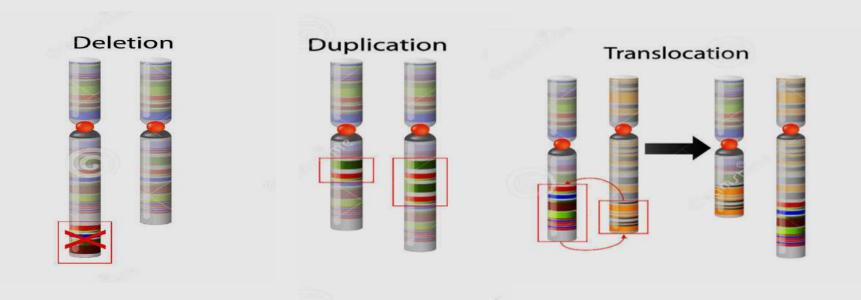
Feature of down syndrome:

- Upward slant to eyes.
- Small ears that fold over at the top.
- Small, flattened nose.
- Small mouth, making tongue appear large.
- Short neck.
- Small hands with short fingers



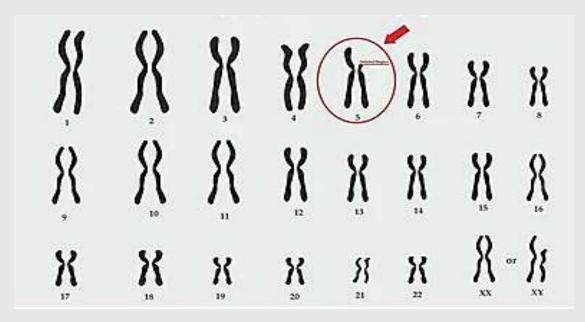
• Structural abnormality:

A chromosome's structure can be altered in several forms:



Example:

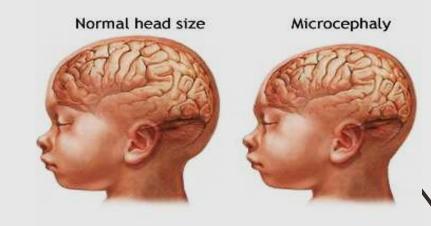
 Cri-du-chat (cat's cry) syndrome, also known as 5p- (5p minus) syndrome



Feature of Cri-du-chat

syndrome:

- Cat-like cry
- Small head size
- Characteristic facial features
- Intellectual disability
- Hypotonia
- Growth delay





Summary

- Chromosomal abnormalities is the numerical or structural error in the natural forms of chromosomes.
- Importantly, this abnormalities usually happens due to error in cell division i.e. mitosis and meiosis.
- The abnormality in the Chromosomes can result different human condition such as downs syndrome and cri-du chat syndrome

References

- <u>https://www.wikilectures.eu/w/Chromosomal_Abnormalities</u>
- <u>https://www.genome.gov/about-genomics/fact-sheets/Chromosome-Abnormalities-Fact-Sheet</u>
- <u>https://childrenswi.org/medical-care/genetics-and-genomics-program/medical-genetics/chromosome-abnormalities/how-chromosome-abnormalities-happen</u>
- <u>https://www.ncbi.nlm.nih.gov/books/NBK115545/</u>
- <u>https://www.cdc.gov/ncbddd/birthdefects/downsyndrome.html</u>
- <u>https://www.urmc.rochester.edu/encyclopedia/content.aspx?ContentTypeID=90&ContentID</u>
 <u>=P02126</u>
- <u>https://image.shutterstock.com/image-vector/cri-du-chat-syndrome-known-260nw-</u> <u>1454593979.jpg</u>

Thank you

Do you have any question?

