MEDICAL ACADEMY NAMED AFTER S.I.GEORGIEVSKY OF VERNADSKY

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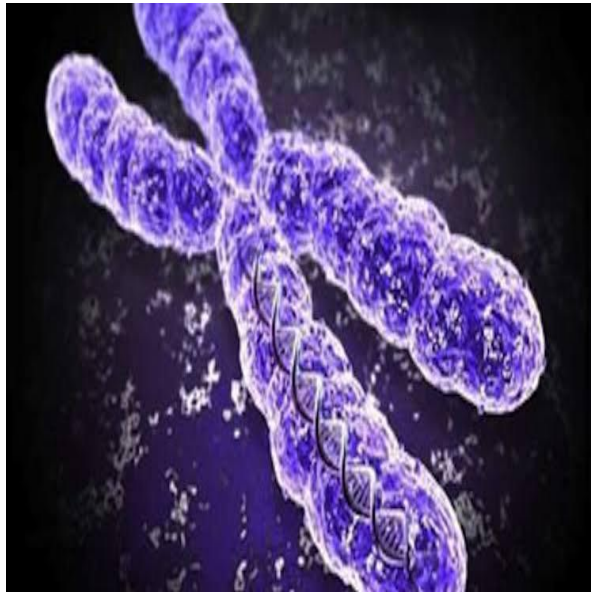
TOPIC: CHROMOSOMAL DISEASES ASSOCIATED WITH CHANGES IN THE NUMBER OF CHROMOSOMES IN HUMANS

TEACHER: PROFESSOR SVETLANA SMIRNOVA

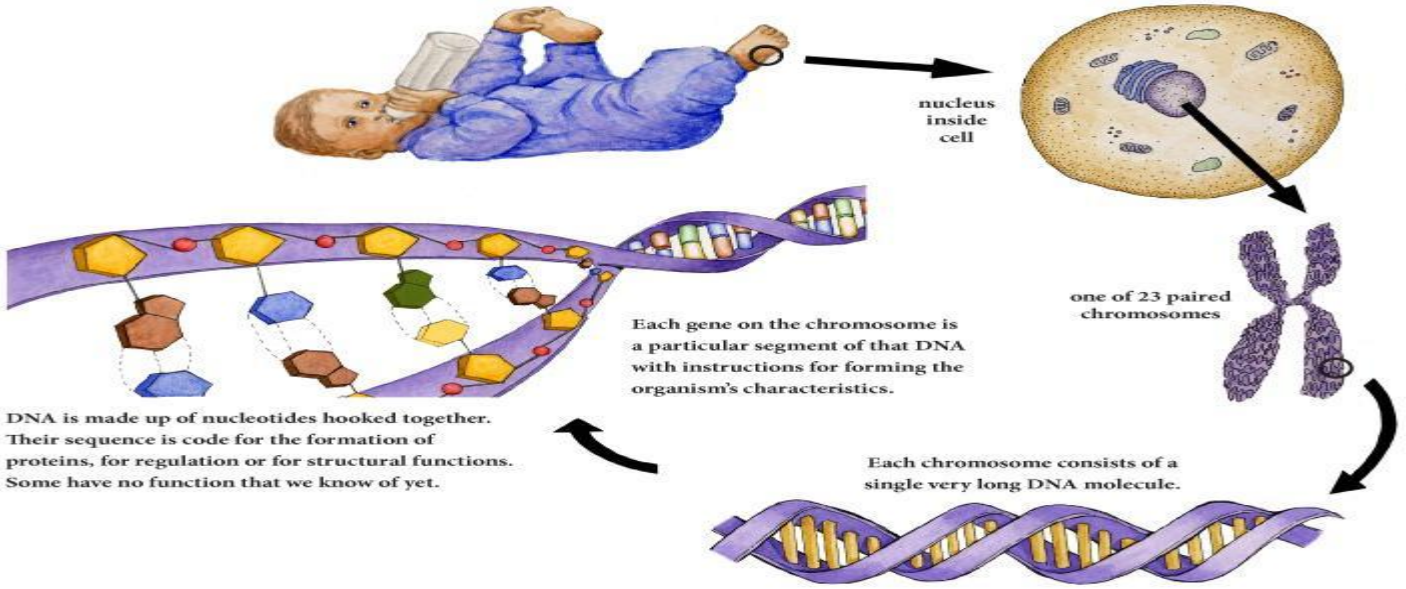
What are chromosomes?

- A chromosome is a long DNA molecule with part or all of genetic material of an organism.
- They are organized packages of DNA found inside your body cells.
- Humans have 23 pairs of chromosomes (total is 46).
- Chromosomes vary in size.
- Each chromosome has a centromere, which divides the chromosome into two uneven sections. The shorter section is called the p arm, and the longer section is called the q arm.



Where are chromosomes found in the body?

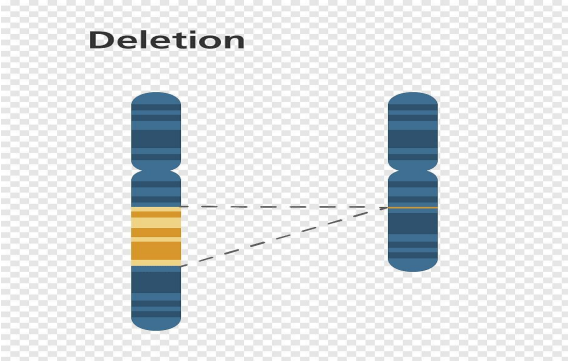
- The body is made up of individual units called cells. Your body has many different kinds of cells, such as skin cells, liver cells and blood cells. In the center of most cells is a structure called the nucleus. This is where chromosomes are located.



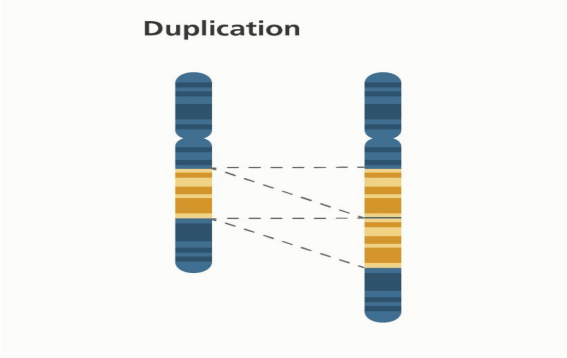
What are the different types of chromosome disorders?

- **Chromosome disorders can be classified into two main types**:
- numerical and structural**. Numerical disorders** occur when there is a change in the number of chromosomes more or fewer than 46. Examples of numerical disorders include trisomy, monosomy and triploidy . Probably one of the most well-known numerical disorders is **Down syndrome** (trisomy 21).Other common types of numerical disorders include trisomy 13, trisomy 18, Klinefelter syndrome and Turner syndrome.
- **Structural chromosome disorders** result from breakages within a chromosome. In these types of disorders there may be more or less than two copies of any gene. This difference in number of copies of genes may lead to clinical differences in affected individuals.

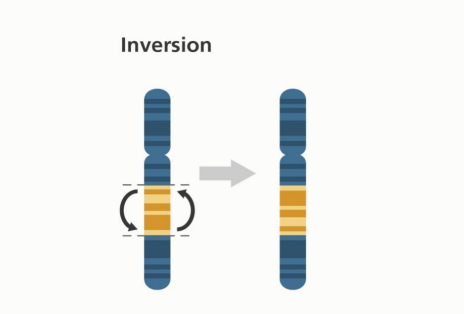
- **Deletions**: A portion of the chromosome is missing or delete.



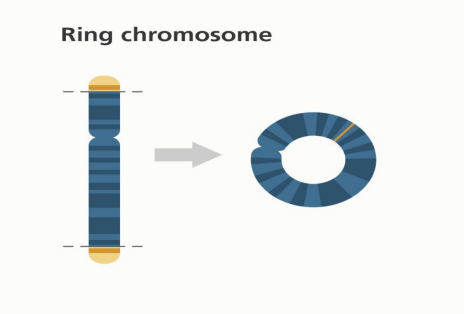
- **Duplications:** A portion of the chromosome is duplicated, resulting in extra genetic material.



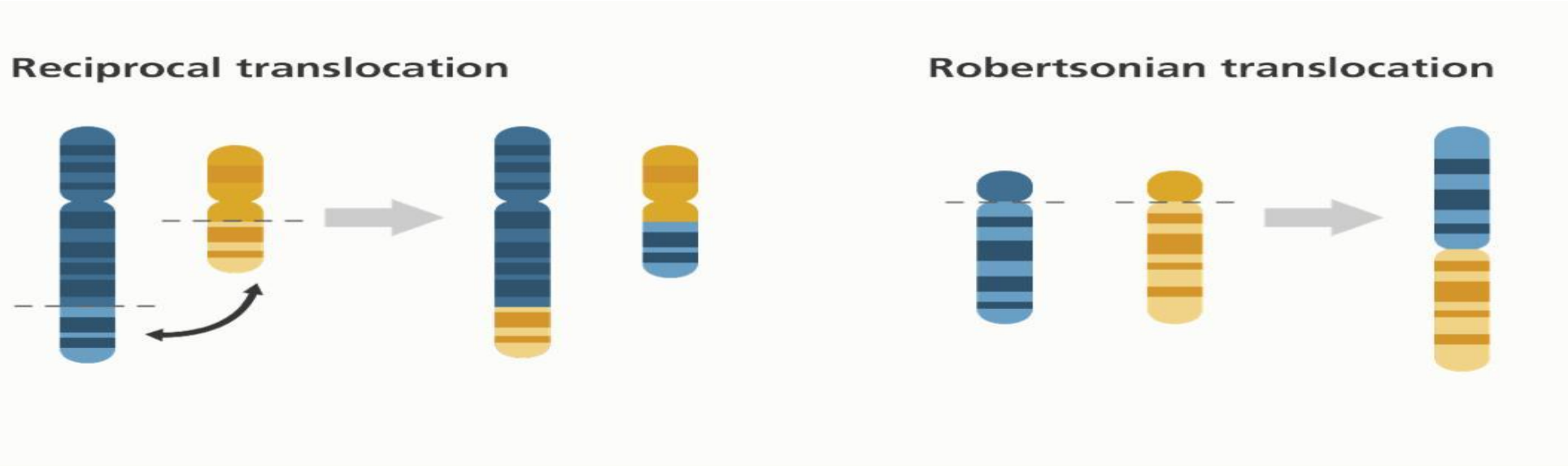
- **Inversions:** A portion of the chromosome has broken off, turned upside down, and reattached. As a result, the genetic material is inverted.



- **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.



- **Translocations:** A portion of one chromosome is transferred to another chromosome. There are two main types of translocation. In a reciprocal translocation, segments from two different chromosomes have been exchanged. In a Robertsonian translocation, an entire chromosome has attached to another at the centromere.

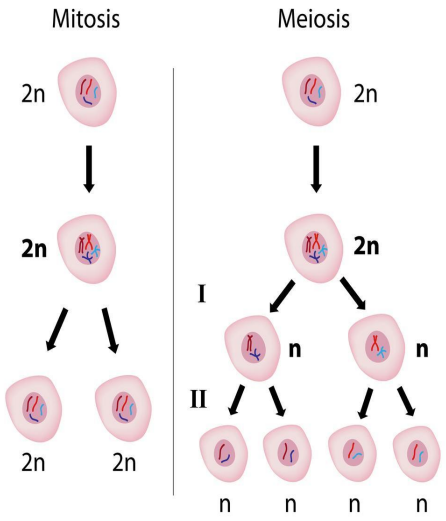


Chromosomal abnormalities:

- **Chromosomal abnormalities** can occur as an accident when the egg or the sperm is formed or during the early developmental stages of the fetus. The age of the mother and certain environmental factors may play a role in the occurrence of genetic errors. Prenatal screening and testing can be performed to examine the chromosomes of the fetus and detect some, but not all types of chromosomal abnormalities.
- Chromosomal abnormalities can have many different effects, depending on the specific abnormality. For example an extra copy of chromosome 21 causes Down syndrome trisomy 21. Chromosomal abnormalities can also cause miscarriage, disease, or problems in growth or development.

How do chromosome abnormalities happen?

- 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. This kind of cell division occurs throughout the body, except in the reproductive organs. This is the way most of the cells that make up our body are made and replaced.
- **Meiosis** results in cells with half the number of chromosomes, 23, instead of the normal 46. This is the type of cell division that occurs in the reproductive organs, resulting in the eggs and sperm.



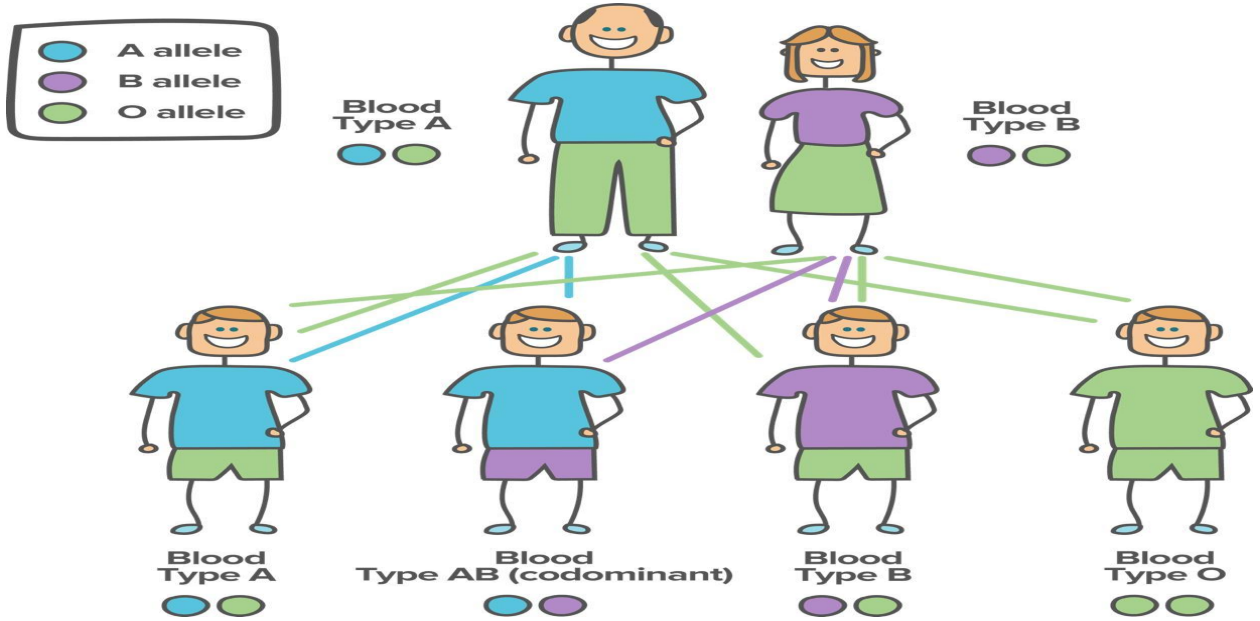
- **Maternal Age**: Women are born with all the eggs they will ever have. Some researchers believe that errors can crop up in the eggs' genetic material as they age. Older women are at higher risk of giving birth to babies with chromosome abnormalities than younger women. Because men produce new sperm 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.

Examples of genetic disease resulting from chromosomal aberrations:

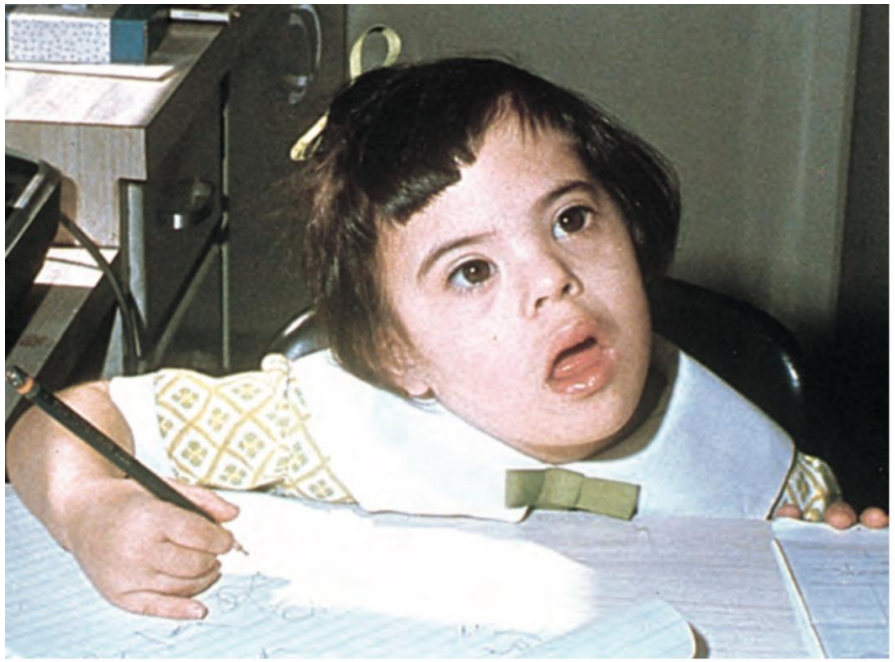
- Down syndrome (trisomy of 21).
- Kleinfelters syndrome (XXY).
- Turner syndrome (single X).
- Triple XXX syndrome.
- Philadelphia chromosome translocation-chronic myelogenous leukemia (CML)

What are genetic factors affecting child development?

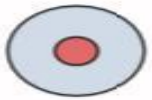
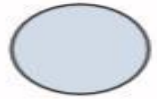
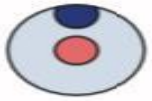
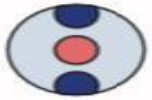
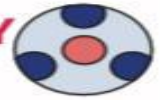
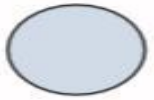
- Genetic Interactions: Genes can sometimes contain conflicting information, and in most cases, one gene will win the battle for dominance. Some genes act in an additive way. For example, if a child has one tall parent and one short parent, the child may end up splitting the difference by being of average height.

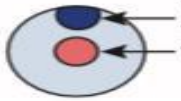


the facial features of Down syndrome.



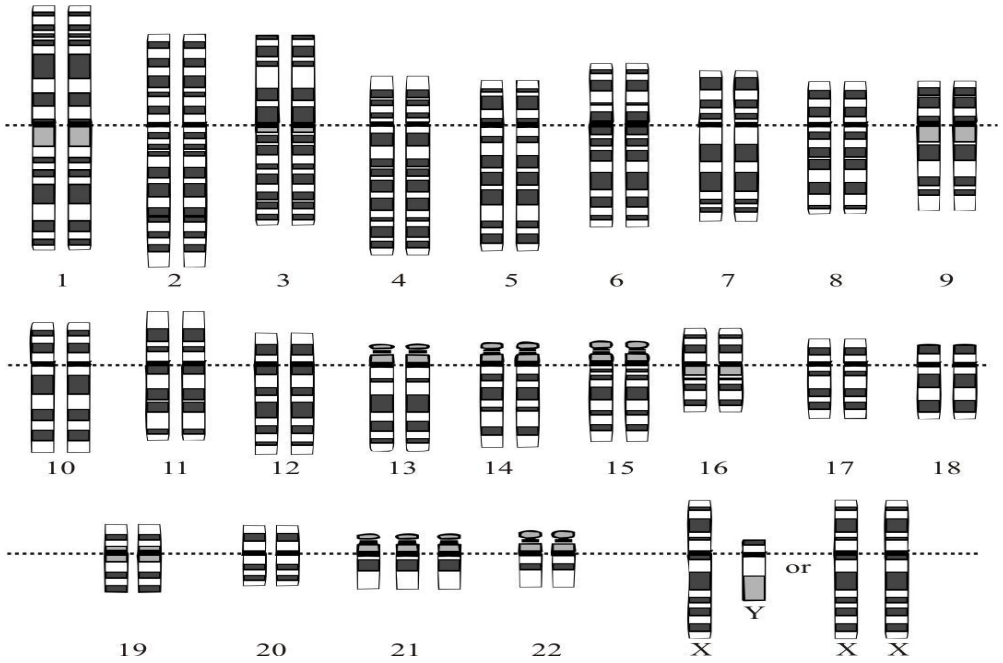
Numerical aberrations of sex chromosomes.

Gametes: Ovum \ Sperm	X	Y	XY	O
X	46,XX Normal ♀ 	46,XY Normal ♂ 	47,XXY Klinefelter ♂ 	45,X Turner ♀ 
XX	47,XXX ♀ 	47,XXY Klinefelter ♂ 	48,XXXY Klinefelter ♂ 	46,XX Normal ♀ 
XXX	48,XXXX ♀ 	48,XXXY Klinefelter ♂ 	49,XXXXY Klinefelter ♂ 	47,XXX Triple X ♀ 
O	45,X Turner ♀ 	45,Y LETHAL	46,XY LETHAL	44 LETHAL



Aneuploidy :

Aneuploidy is the presence of an abnormal number of chromosomes in a cell, for example a human cell having 45 or 47 chromosomes instead of the usual 46. It does not include a difference of one or more complete sets of chromosomes. A cell with any number of complete chromosome sets is called a euploid cell.



Mechanisms:

- Aneuploidy arises from errors in chromosome segregation, which can go wrong in several ways.
- **Nondisjunction** usually occurs as the result of a weakened mitotic checkpoint, as these checkpoints tend to arrest or delay cell division until all components of the cell are ready to enter the next phase.
- For example, if a checkpoint is weakened, the cell may fail to notice that a chromosome pair is not lined with the spindle apparatus. In such a case, most chromosomes would separate normally with one chromatid ending up in each cell, while others could fail to separate at all. This would generate a daughter cell lacking a copy and a daughter cell with an extra copy.
- Completely inactive mitotic checkpoints may cause nondisjunction at multiple chromosomes, possibly all. Such a scenario could result in each daughter cell possessing a disjoint set of genetic material.

Merotelic attachment occurs when one kinetochore is attached to both mitotic spindle poles. One daughter cell would have a normal complement of chromosomes; the second would lack one. A third daughter cell may end up with the missing chromosome .

Multipolar spindles: more than two spindle poles form. Such a mitotic division would result in one daughter cell for each spindle pole; each cell may possess an unpredictable complement of chromosomes.

Monopolar spindle: only a single spindle pole forms. This produces a single daughter cell with its copy number doubled.

A **tetraploid** intermediate may be produced as the end-result of the monopolar spindle mechanism. In such a case, the cell has double the copy number of a normal cell, and produces double the number of spindle poles as well. This results in four daughter cells with an unpredictable complement of chromosomes, but in the normal copy number.


Genetics
sequence
organisms
genes
DNA
strand
heredity
organism
change
inherit
proteins
nucleotides
physical
function
gene
science
traits
amino
acids
protein
cells
Mendel
genetic
role
creating
inheritance
experience
living
order
shape
chain
large

THANK YOU