Chromosomal abnormalities

- A **chromosome anomaly**, **abnormality** or **aberration** reflects on a typical number of chromosomes or a structural abnormality in one or more chromosomes.

- A karyotype refers to a full set of chromosomes from an individual which can be compared to a "normal" karyotype for the species via genetic testing.

- Chromosome anomalies usually occur when there is an error in cell division following meiosis or mitosis.
- There are many types of chromosome anomalies.
- They can be organized into two basic groups, numerical and structural anomalies.

Numerical

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

- In humans an example of a condition caused by a numerical anomaly is Down Syndrome, also known as Trisomy 21 (an individual with Down Syndrome has three copies of chromosome 21, rather than two).
- Trisomy has been determined to be a function of maternal age.

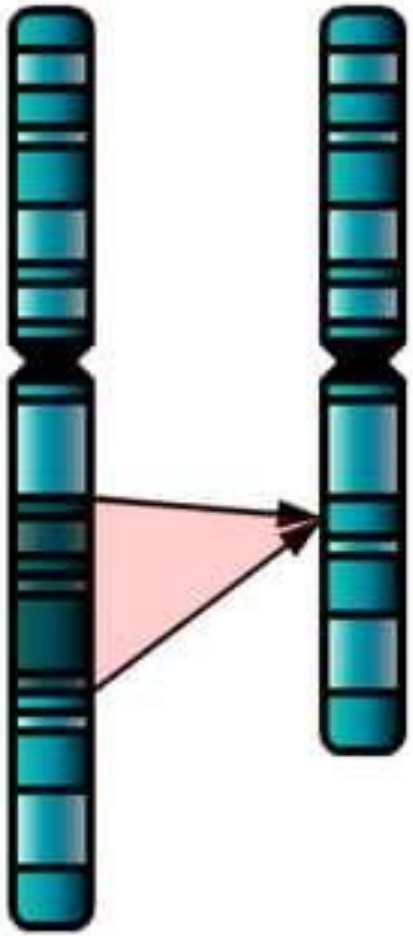
- An example of monosomy is Turner Syndrome, where the individual is born with only one sex chromosome, an X.

Structural

- When the chromosome's structure is altered, this can take several forms:

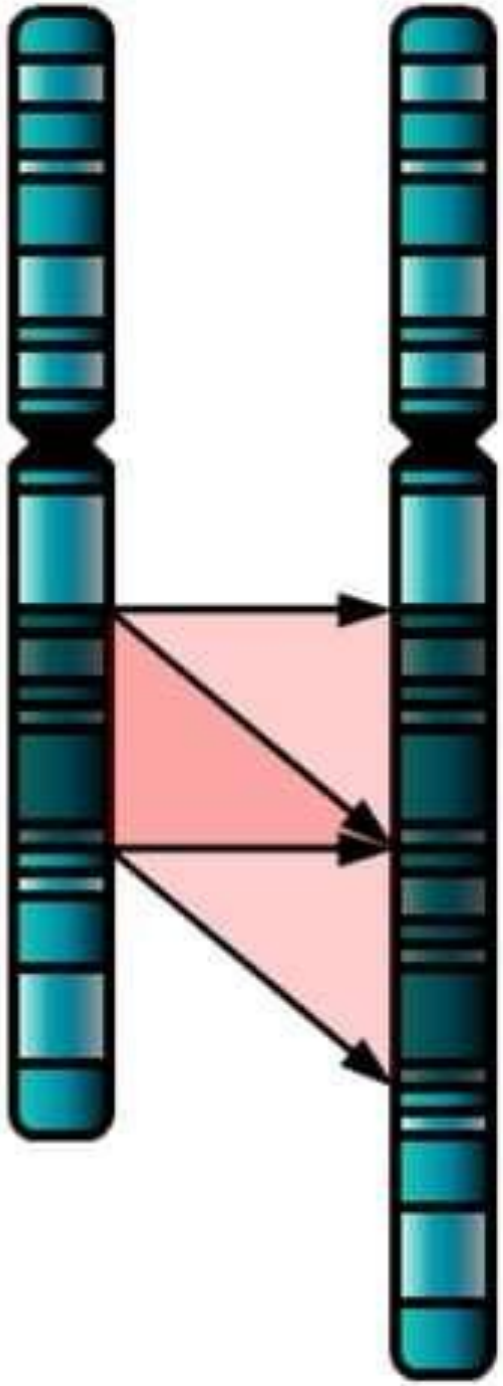
- Deletions:

A portion of the chromosome is missing or deleted.

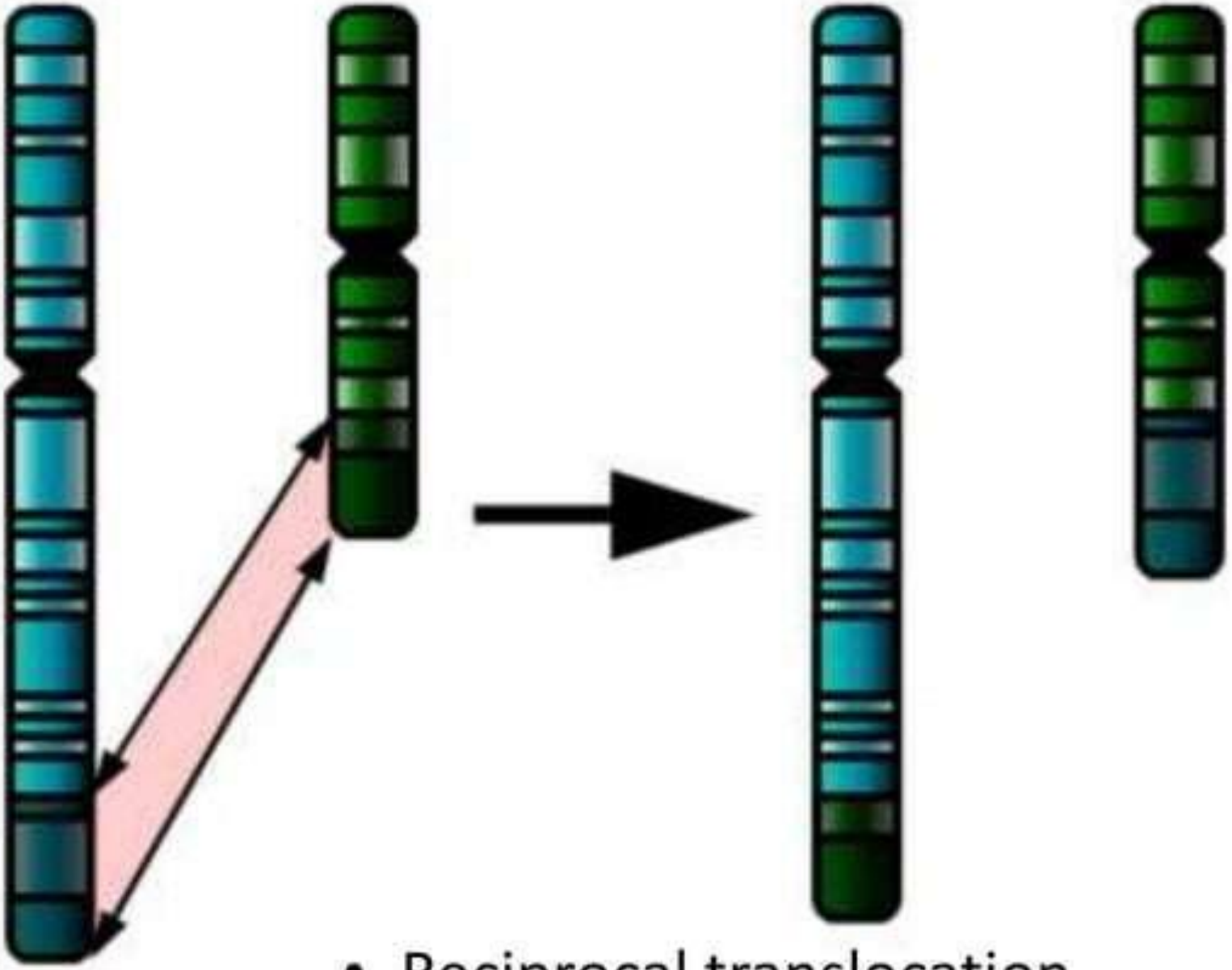


- Known disorders in humans include
- Wolf-Hirschhorn syndrome, which is caused by partial deletion of the short arm of chromosome 4; and
- Jacobsen syndrome, also called the terminal 11q deletion disorder.

- Duplications: A portion of the chromosome is duplicated, resulting in extra genetic material. Known human disorders include Charcot-Marie-Tooth disease type 1A which may be caused by duplication of the gene encoding peripheral myelin protein 22 (PMP22) on chromosome 17.



- Translocations: A portion of one chromosome is transferred to another chromosome. There are two main types of translocations:
- Reciprocal translocation: Segments from two different chromosomes have been exchanged.
- Robertsonian translocation: An entire chromosome has attached to another at the centromere - in humans these only occur with chromosomes 13, 14, 15, 21 and 22.



- Reciprocal translocation

- **Robertsonian translocation** (ROB) is a common form of chromosomal rearrangement that in humans occurs in the five acrocentric chromosome pairs, namely 13, 14, 15, 21, and 22.

- They are named after the American biologist William Rees Brebner Robertson Ph.D. (1881–1941), who first described a Robertsonian translocation in grasshoppers in 1916.
- They are also called

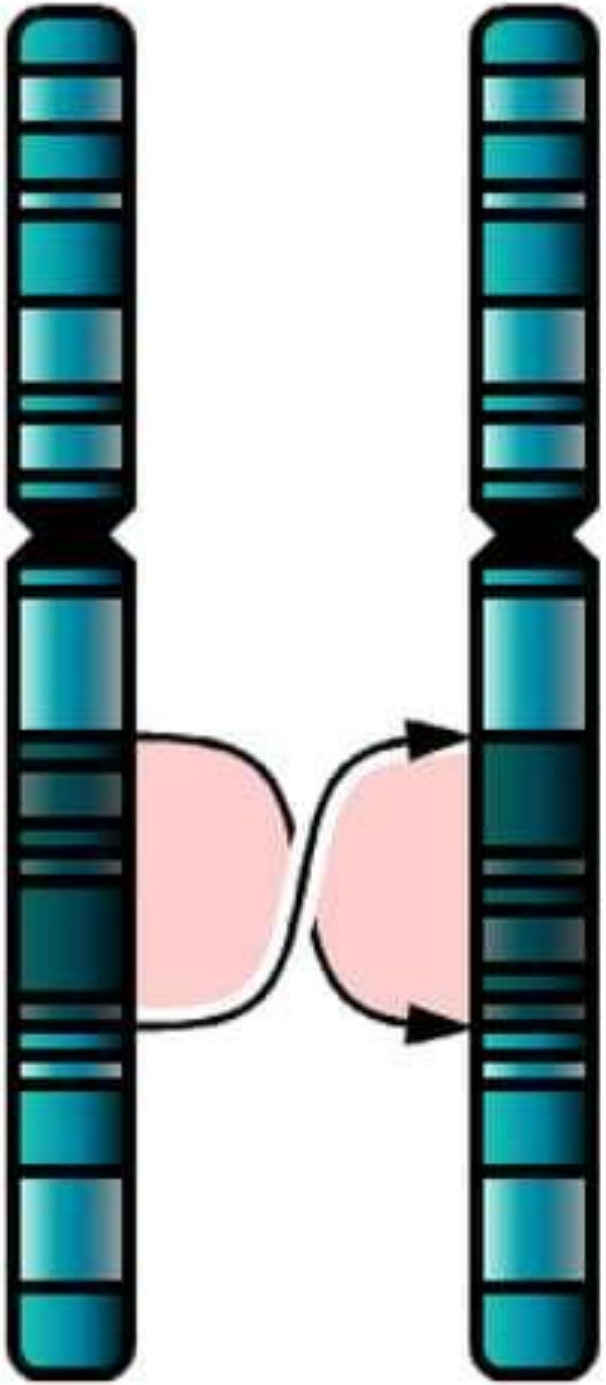
➢ **whole-arm translocations** or

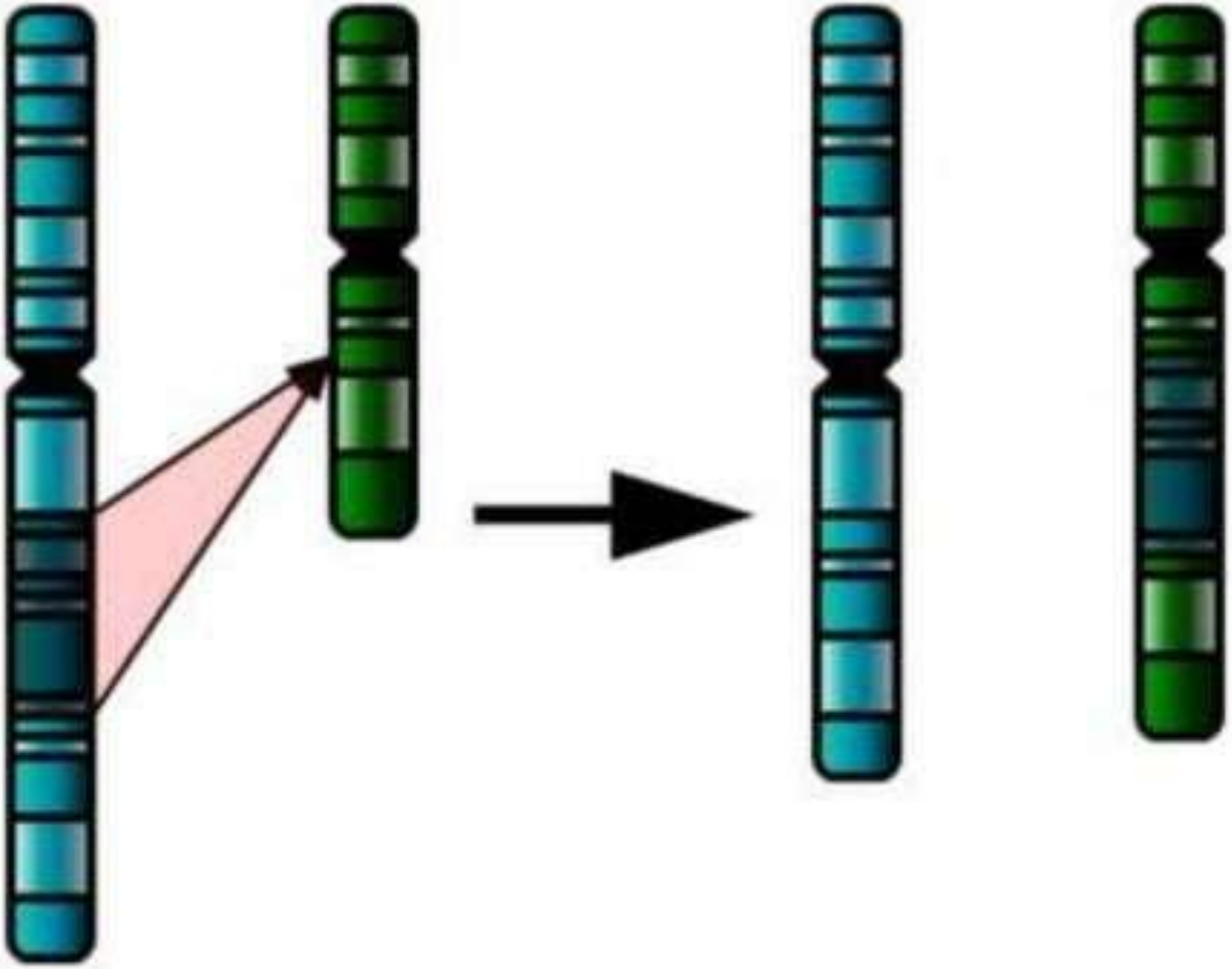
➢ **centric-fusion translocations**.

- A Robertsonian translocation is a type of nonreciprocal translocation involving two homologous chromosomes or non-homologous chromosomes (i.e. two different chromosomes, not belonging to a homologous pair).
- A feature of those chromosomes that possess an acrocentric centromere, partitioning the chromosome into a large arm containing the vast majority of genes, and a short arm with a much smaller proportion of genetic content.

- During a Robertsonian translocation, the participating chromosomes break at their centromeres and the long arms fuse to form a single chromosome with a single centromere.
- The short arms also join to form a reciprocal product, which typically contains nonessential genes and is usually lost within a few cell divisions.

- Inversions: A portion of the chromosome has broken off, turned upside down and reattached, therefore the genetic material is inverted and sequence is disturbed.





- Insertions: A portion of one chromosome has been deleted from its normal place and inserted into another chromosome.

- 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: Formed by the mirror image copy of a chromosome segment including the centromere.

- Chromosome instability syndromes are a group of disorders characterized by chromosomal instability and breakage.
- They often lead to an increased tendency to develop certain types of malignancies.

- Most chromosome abnormalities occur as an accident in the egg or sperm, and therefore the anomaly is present in every cell of the body.
- Some anomalies, however, can happen after conception, resulting in Mosaicism (where some cells have the anomaly and some do not).

- In genetics, a **mosaic** or **mosaicism** denotes the presence of two or more populations of cells with different genotypes in one individual who has developed from a single fertilized egg.

- Chromosome anomalies can be inherited from a parent or be "de novo".
- This is why chromosome studies are often performed on parents when a child is found to have an anomaly.
- If the parents do not possess the abnormality it was not initially inherited; however it may be transmitted to subsequent generations.

- *De novo* mutation, a genetic mutation that neither parent possessed nor transmitted