Chromosomal Aberration

A chromosome is a long DNA molecule with part or all of the genetic material of an organism. Chromosomal abnormalities occur when there is a defect in a chromosome, or in the arrangement of the genetic material on the chromosome. Very often, chromosome abnormalities give rise to specific physical symptoms, however, the severity of these can vary from individual to individual.

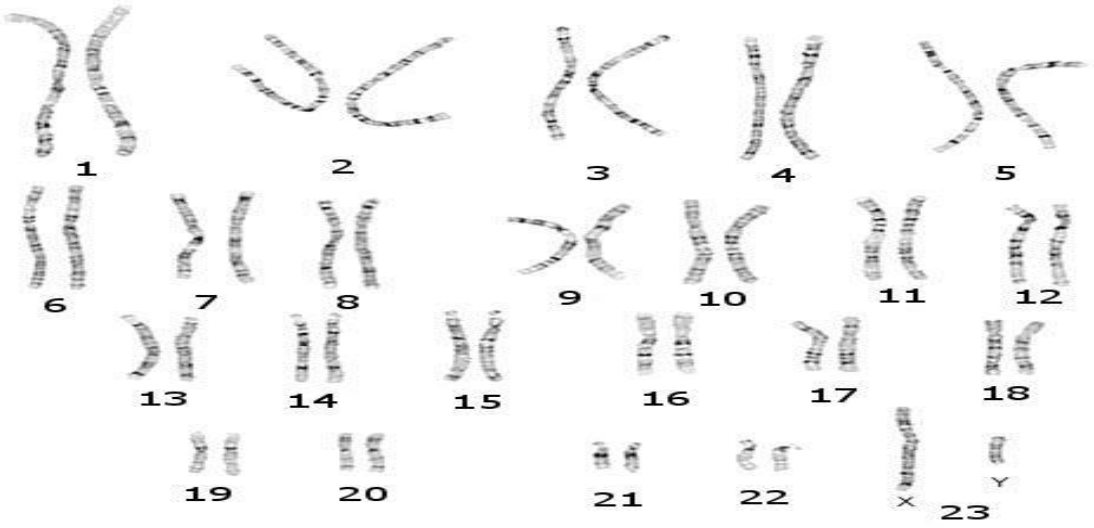
Abnormalities can be in the form of additional material which may be attached to a chromosome, or where part or a whole chromosome is missing, or even in defective formation of a chromosome. Any increases or decreases in chromosomal material interfere with normal development and function.

Karyotyping : refers to a full set of chromosome from an individual, or a photographic arrangement of a set of chromosomes of an individual. ′

Number of chromosomes: 22 pairs of autosomes& a pair of sex chromosomes.

Diploid: 46. haploid : 23

There are two main types of chromosomal abnormality which can occur during meiosis and fertilization: numerical aberrations and structural aberrations.



Numerical Aberrations

These are usually caused by a failure of chromosome division, which results in cells with an extra chromosome or a deficiency in chromosomes.

Gametes with these anomalies can result in conditions such as Down syndrome (who have 47 chromosomes instead of 46), or Turner syndrome (45 chromosomes).

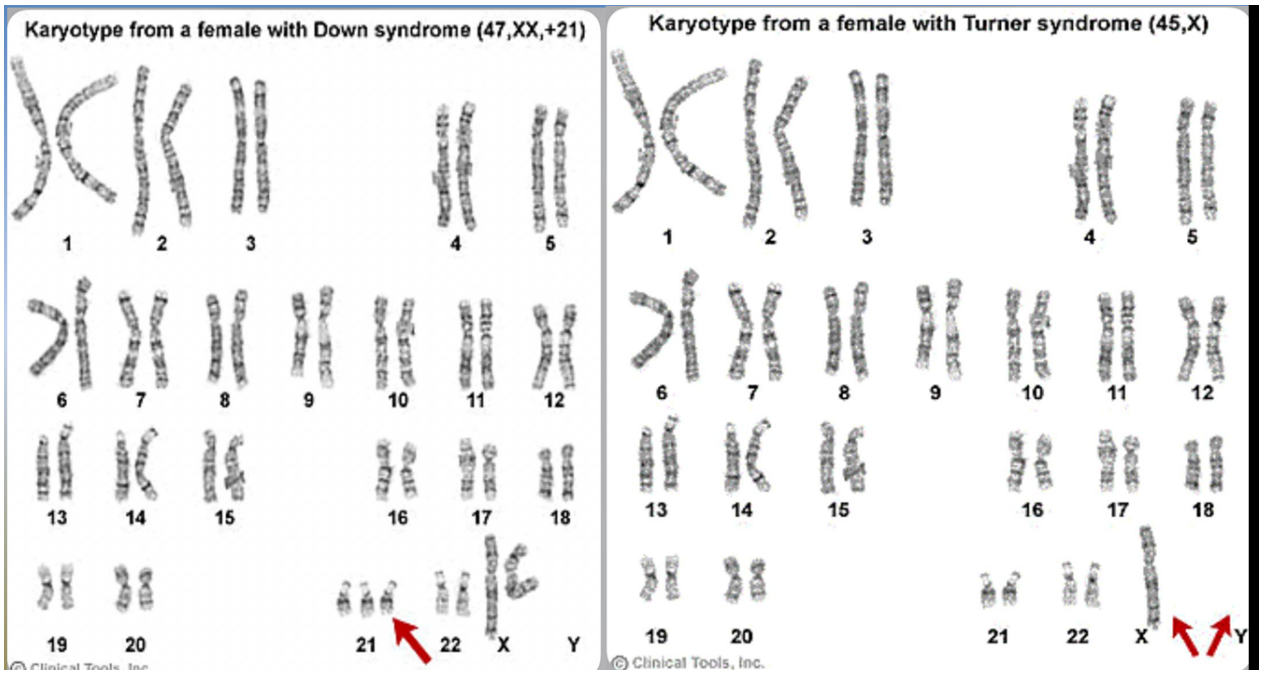
Aneuploidy : having too many or too few chromosomes.

1. Trisomy: 3 copies of particular chromosome. Common trisomy is down syndrome, trisomy 21. Reason : fails to go to opposite poles of the dividing cells.

2. Monosomy: missing of a copy of chromosome. Ex: 45x, and monosomy are always lethal, the only compatible condition is turner syndrome(only one x chromosome)

3. Polyploidy: more than normal diploid number of chromosome. Ex: triploidy – 6pxxx , Tertroploidy – 92XXXX , Reason : Polysperm

4. Mosaicism : half of the cell have normal and the remaining have abnormal in term of structure or number It commonly seen in down syndrome , turner syndrome etc. .



Structural Aberrations

These occur due to a loss or genetic material, or a rearrangement in the location of the genetic material. They include: deletions, duplications, inversions, ring formations, and translocations.

Deletions: A portion of the chromosome is missing or deleted. Known disorders include Wolf-Hirschhorn syndrome, which is caused by partial deletion of the short arm of chromosome 4; and Jacobsen syndrome,

Duplications: A portion of the chromosome is duplicated, resulting in extra genetic material. Known 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: When a portion of one chromosome is transferred to another chromosome. There are two main types of translocations. 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; these only occur with chromosomes 13, 14, 15, 21 and 22.

Inversions: A portion of the chromosome has broken off, turned upside down and reattached, therefore 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: Formed by the mirror image copy of a chromosome segment including the centromere.

Structural aberrations also include some disorders which are characterized by chromosomal instability and breakage. One example, is the creation of a fragile site on the X Chromosome - Fragile X syndrome. Boys are worse affected by this because they only have one X-Chromosome but even in girls, Fragile X syndrome can cause learning difficulties.