Chromosomal Aberrations

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Chromosome Abnormalities

What is Chromosomal aberration?

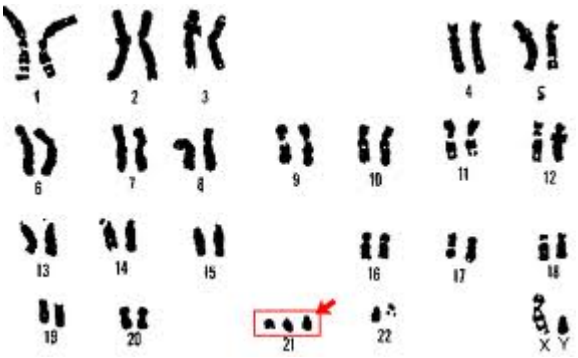
The disruptions in the normal chromosomal content of a cell are known as chromosomal aberrations.

These are the main cause of genetic conditions in humans. For example, ‘Down syndrome’. It occurs because of the aberration of 21st Chromosome. Translocations and chromosomal inversions are also the forms of chromosomal aberrations. But these won’t create any diseases like Down syndrome. But these chromosomal changes may lead to a higher chance of a child with a chromosome disorder during Birth. Abnormal numbers of chromosomes are named as aneuploidy. This may lead to death create many genetic disorders. If any of the people are affected with this abnormalities, the Genetic counseling is offered to them to adapt to the body changes.

Genetic Disorders

The gain or loss of DNA from chromosomes can lead to a variety of genetic disorders. Human examples include:
Cri du chat

- Caused by the deletion of part of the short arm of chromosome 5.
- Symptoms are wide-set eyes, a small head and jaw, moderate to severe mental health issues, and are very short.



Down syndrome

- Caused by an extra copy of chromosome 21 normally termed as trisomy 21.
- Symptoms are decreased muscle tone, stockier build, asymmetrical skull, slanting eyes and mild to moderate developmental disability.

Edwards syndrome

- The second-most-common Trisomy;
- It is a Trisomy of chromosome 18.

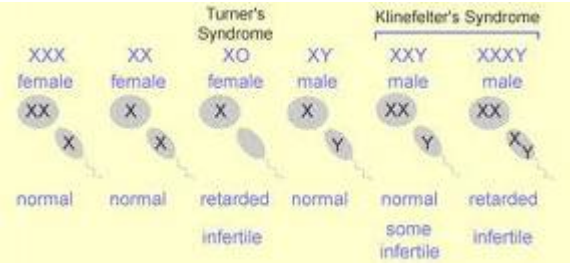
- Symptoms are motor retardation, developmental disability and numerous congenital anomalies causing serious health problems.

Idic15:

- Abbreviation for Isodicentric 15 on chromosome 15
- Also called as IDIC(15), Inverted duplication 15, extra Marker, Inv dup 15, partial tetrasomy 15.

Jacobsen syndrome:

- Rare disorder
- Also known as terminal 11q deletion disorder.
- Symptoms are mild developmental disability, with poor expressive language skills.
- Most have a bleeding disorder called Paris-Trousseau syndrome.



Klinefelter's syndrome (XXY):

- Affected Men are usually sterile, and tend to have longer arms and legs and to be taller than their peers.
- Boys with the syndrome are often shy and quiet, and have a higher incidence of speech delay and dyslexia.
- During puberty, without testosterone treatment, some of them may develop gynecomastia.

Patau Syndrome:

- Known as D-Syndrome or trisomy-13.
- Symptoms are somewhat similar to those of trisomy-18, but they do not have the characteristic hand shape.

Small supernumerary marker chromosome:

- An extra, abnormal chromosome.

Triple-X syndrome (XXX):

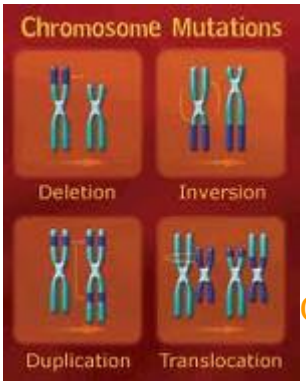
- XXX girls tend to be tall and thin.
- They have a higher incidence of dyslexia.

Turner syndrome (X instead of XX or XY):

- Female sexual characteristics are present but underdeveloped.
- Symptoms are short stature, low hairline, abnormal eye features.

XYY syndrome. (S H 4):

- XYY boys are usually taller than their siblings.
- May have learning difficulties.



Chromosomal Mutations

Chromosomal mutations produce changes in whole chromosomes (more than one gene) or in the number of chromosomes present.

- Deletion (S H 4)– loss of part of a chromosome
- Duplication (S H 4)– extra copies of a part of a chromosome
- Inversion(S H 4) – reverse the direction of a part of a chromosome
- Translocation (S H 4)– part of a chromosome breaks off and attaches to another chromosome.

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