Chromosome abnormalities

A chromosome anomaly, abnormality or aberration reflects an atypical number of chromosomes or a structural abnormality in one or more chromosomes. Chromosome anomalies usually occur when there is an error in cell division. There are many types of chromosome anomalies. They can be organized into two basic groups, numerical and structural anomalies. A chromosome anomaly may be detected or confirmed in A Karyotype.

A- Abnormalities of Chromosome Structure: can be either unbalanced or balanced rearrangements. If the rearrangement is unbalanced, there is additional or missing genetic material, On the other hand, if the rearrangement is balanced, there is no extra or missing chromosome material.

- Deletions: A small section of a chromosome is missing.
- Translocations: A section of a chromosome is attached to another chromosome.
- Inversions: A section of chromosome is snipped out and reinserted upside down.
- Duplications: A section of a chromosome is duplicated, so there is extra genetic material.
- Ring chromosome: Material is deleted at both ends of a chromosome, and the new ends join together to form a ring.
- Isochromosomes: when one arm of a chromosome is missing, and the other arm is duplicated

B- Abnormalities of Chromosome Number:

Polyploidy: the presence of complete set of extra chromosome in a cell and observed in a form:

- Triploidy: 69 chromosome in each cell nucleus, karyotyping is 69, xxx.
- Tetraploidy : 92 chromosome in each cell nucleus ,karyotyping is 92,xxxx.

Aneuploidy: the cell contain missing or additional chromosome .usually only one chromosome are affected.

- Trisomy: (three copy of chromosome) is a type of anueploidy in which there are three copies of a particular chromosome instead of two:
  - ✓ Trisomy21(47,xy,+21) recognized as Down syndrome.
  - ✓ Trisome 18 (47,xy,+18) recognized as Edward syndrome ,is the second most common autosomal trisomy.
  - ✓ Trisomy 13(47,xy,+13) recognized as patau syndrome.
- Monsomy: is another type of aneuploidy where there is one copy of a certain chromosome instead of two copies. When this occurs it is almost always lethal. The only complete monosomy that is Turner syndrome, where there is only one X chromosome instead of the normal pair.

Genetic diseases

- Klinefelter syndrome, (47, XXY) syndrome is a condition in which human males have an extra X chromosome, the condition exists in roughly 1 out of every 500-650, Cause: The extra X chromosome is retained because of a nondisjunction event during meiosis.

Symptoms

- Infertility (azoospermia).
- Psychological or behavioral problems.
- Delays in motor development or dysfunction.
- Tall, a rounded body type.
- Gynecomastia (increased breast tissues).
- Hypogonadism , individuals will often have a low serum testosterone.
- Osteoporosis.

Diagnosis: A karyotype is used to confirm the diagnosis & Diagnosis can also be made prenatally via chorionic villus sampling or amniocentesis, tests.

- Turner syndrome or Ullrich-Turner syndrome , is a conditions in human females, of which monosomy X (absence of an entire sex chromosome), which one of those sex chromosomes is missing, when the chromosome is missing in some cells but not others or Ring chromosome, a condition referred to as mosaicism or 'Turner mosaicism, The condition exists 1 out of every 2000 females. These females do not have Barr bodies, which are those X chromosomes inactivated by the cell.

Symptoms

- Short stature
- Lymphedema (swelling) of the hands and feet
- Low hairline
- underdeveloped breasts and widely spaced nipples
- Non-functioning ovaries
- hearing loss in some cases
- narrowing of the aorta
- Amenorrhoea

many nevi in body