STRUCTURAL ABNORMALITIES OF CHROMOSOMES

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Structural Abnormalities

- Changes that affect the structure of a chromosome
- These changes can affect many genes along the chromosome and hence disrupt the proteins made from those genes.
- They can occur during the formation of an egg or sperm cells, in early fetal development or in any cell after birth.
- The effects of structural changes depend upon their size and location and whether any genetic material is gained or lost.
- Parents may have a "balanced chromosomal rearrangements", Translocation, ring chromosomes, inversions can present as balanced arrangements where the person is phenotypically normal.
- Balanced rearrangements have an increased risk of history of infertility, multiple miscarriages or children affected with unbalanced structural or numerical chromosomal abnormalities and with presentation of clinical features due to formation of unbalanced gametes.
- Microdeletions may present as behavioural problems.
- Unbalanced structural abnormalities cause both physical and mental problems.
Deletions

• Occurs when a chromosome breaks and some genetic material is lost. Deletions can be large or small and can occur anywhere along a chromosome.

46, XY, del (5)
Deletion of short arm of chr 5
CRI DU CHAT SYNDROME: deletion of short arm of chromosome 5

Di George syndrome: deletion of a small piece of long arm of chromosome 22

Velocardiofacial syndrome
Translocations

• A portion of one chromosome is transferred to another.

• 1. RECIPROCAL TRANSLOCATION, segments of different chr have been exchanged.

• 2. ROBERTSONIAN TRANSLOCATION, an entire chr has attached to another at the centromere. In humans this only occurs in chr 13, 14, 15, 21 and 22.
Duplication

- Occurs when part of a chromosome is copied (duplicated) and present in two copies. This type of chromosomal change results in extra copies of genetic material from the duplicated segment. These extra genes present on the duplicated segment do not function properly.

46, XX, dup(2)
partial duplication of the short arm of chr 2
Duplication of short arm of chromosome 12
PALLISTER KILLIAN SYNDROME
Inversion

Involves the breakage of a chromosome in two places, the resulting piece of DNA is reversed and re-inserted into the chromosome. Inversions can be balanced or unbalanced and can predispose an individual to having a child with a duplication or deletion of the involved region.

46, XY, inv (11) – pericentric inversion of chr 11
Pericentric inversion of chromosome 9
iso-chromosomes

- Is a chromosome with two identical arms. Instead of one long arm (q) and one short arm (p), an isochromosome has two long or two short arms thereby having extra copy and missing copy of other genes.

46, X, I (Xq)
Isochromosome of Xq, Turner female.
Isochromosome 18 p syndrome
Ring chromosome

- The chromosome breaks in two places and the ends join to form a ring structure. In many cases, the genetic material near the ends of the chromosome is lost.

46,XY, r(3)(p26 q26) Ring chromosome 3(p26 q29)
Ring chromosome 20 syndrome
Clinical implications of numerical and structural chromosomal abnormalities:

• If the disorder is clinically demonstrated with structural anomaly, then karyotype of the affected child should be done.

• Most of the cases may require karyotyping of the mother and the father.

• Cases of mental retardation and developmental delay and even behavioural problems (such as autism, slow speech etc) may well be due to numerical and structural abnormalities of the chromosomes: so remember to do karyotyping of the affected child and parents.

• A number of cases of infertility, repeat miscarriages and children born with unbalanced structural and numerical abnormalities may be a result of balanced chromosomal re-arrangements in the parents: so do parental KT.

• All proved numerical and structural abnormalities in either the affected child and the parents will require pre-natal invasive karyotyping in future pregnancies.
Minds are like parachutes. They work best when open! - Lord Thomas Dewar