

chromosomal disorder :-

- * The chromosomal disorders are caused due to absence or excess or abnormal arrangement of one or more chromosome.

Polypoidy :- failure of cytokinesis after telophase stage of cell division results in an increase in a whole set of chromosomes in an organism. This phenomenon is known as polyploidy. This condition is often seen in plants.

"change in whole set of chromosome number"

Aneuploidy :- * failure of segregation of chromatids during cell division cycle results in gain or loss of a chromosome(s), called aneuploidy.

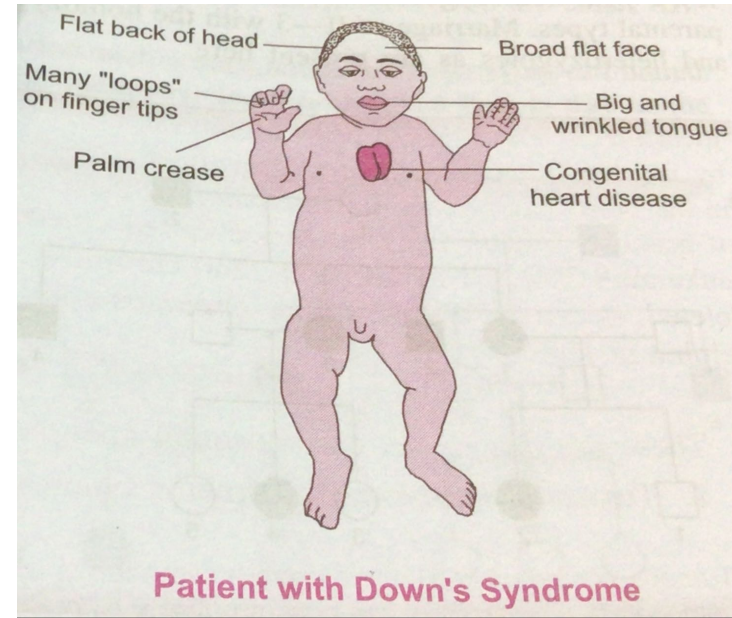
"change in a subset of chromosome number"

Examples :- Down's syndrome
Turner's syndrome
Klinefelter's syndrome

- * The total number of chromosomes in a normal human cell is 46 (23 pairs). Out of these 22 pairs are autosomes and one pair of chromosomes are sex chromosome.
- * Sometimes, though rarely, either an additional copy of a chromosome may be included in an individual or an individual may lack one of any one pair of chromosomes. These situations are known as trisomy or monosomy of a chromosome, respectively. Such a situation leads to very serious consequences in the individual.

(a) Down's Syndrome :- (Mongolism)

- The cause of this genetic disorder is the presence of an additional copy of the chromosome number 21 (trisomy of 21).
- Both the chromosome of the pair 21 pass into a single egg due to non-disjunction during oogenesis.
- This disorder was first described by Langdon Down (1866).
- The affected individual is
 - (i) short statured with small round head
 - (ii) flat back of head & Broad flat face
 - (iii) furrowed tongue and partially open mouth
 - (iv) Palm is broad with palm crease and many loops on fingertips
 - (v) Big and wrinkled tongue.
 - (vi) Congenital heart disease
 - (vii) Physical, psychomotor and mental development is retarded.

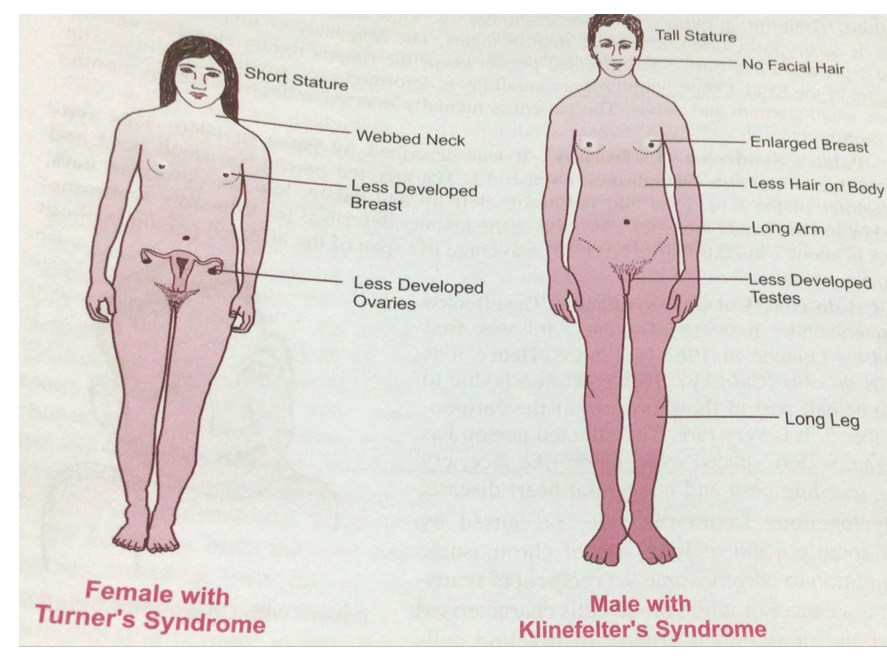


(b) Klinefelter's syndrome :-

- This genetic disorder is also caused due to the presence of an additional copy of X-chromosome resulting into a Karyotype of 47, XXY.
- Such an individual has overall masculine development however, the feminine development (development of breast i.e. Gynaecomastia) is also expressed.
- Such individuals are sterile.

(c) Turner's Syndrome :-

- This disorder is caused due to the absence of one of the X-chromosome, i.e. 45 with XO,
- such females are sterile as ovaries are rudimentary besides other features including lack of other secondary sexual characters.
- Such individuals are sterile. They may not menstruate or ovulate



- * Above chromosomal disorders can be easily studied by analysis of Karyotypes of individual.

** Inheritable mutations can be studied by generating a pedigree of a family.

** chromosomal disorders are non-heritable and pedigree analysis of family does not help in tracing the pattern of inheritance of such chromosomal disorder.