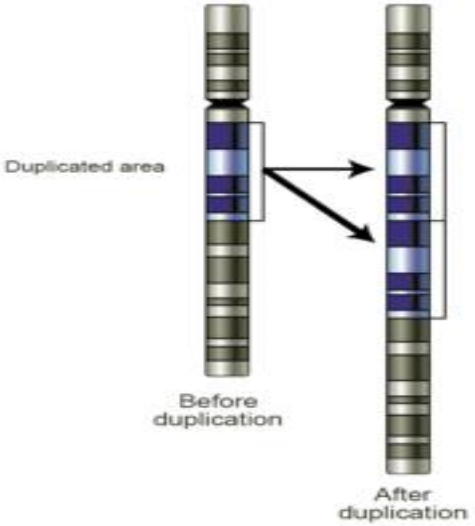
NUMERICAL AND STRUCTURAL VARIATIONS OF CHROMOSOMES

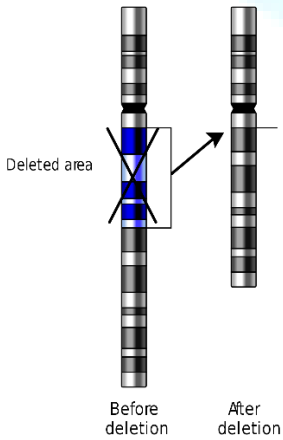
Chromosome mutations

- Chromosome rearrangements alter the structure of chromosomes ;like a piece of a chromosome might be duplicated, deleted or inverted.
- Rearrangements are :-

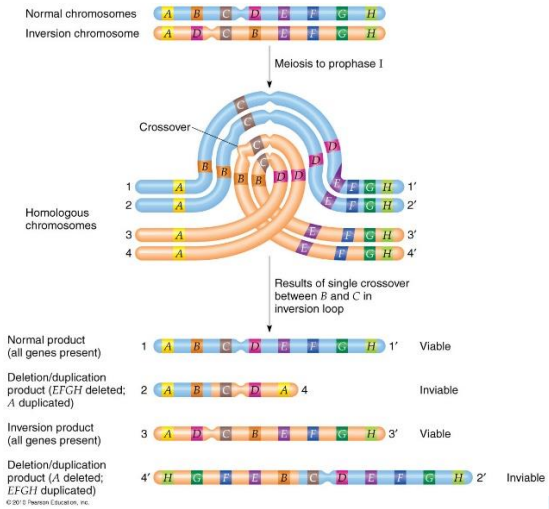
Duplications



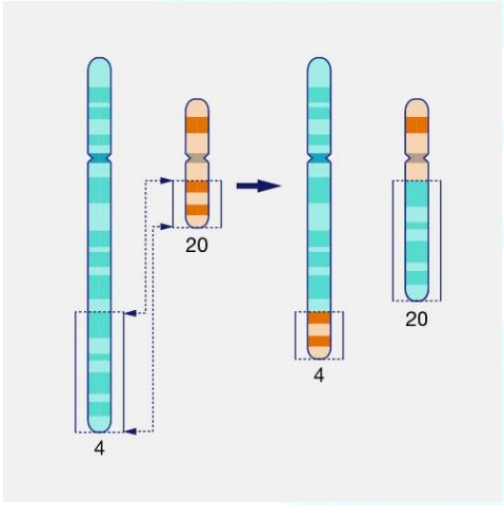
Deletions



Inversions



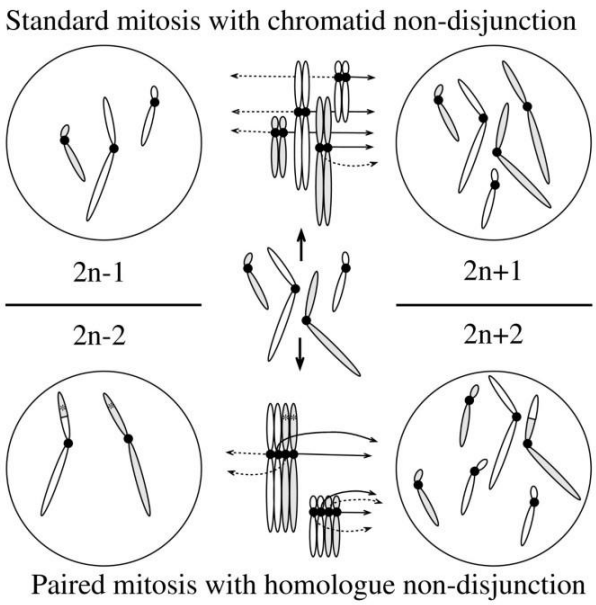
Translocations



- There are basically two Types of mutations
    1. Aneuploidy
    2. Polyploidy

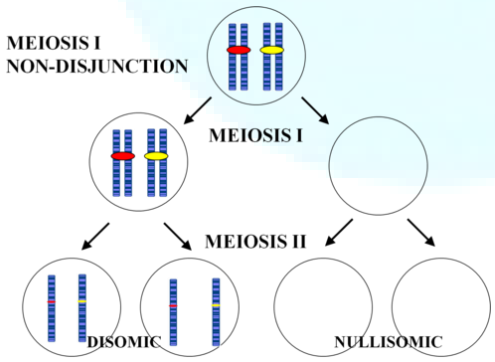
Aneuploidy

- In aneuploidy, the number of chromosome is altered.one or more individual chromosomes are added or deleted.
- There are mainly four types or kinds of aneuploidy:



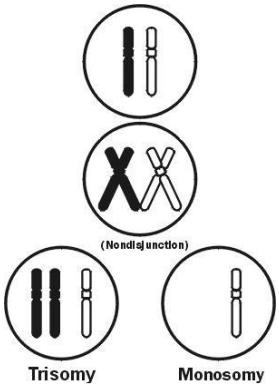
I. Nullisomy
II. Monosomy
III. Trisomy
IV. Tetrasomy

Nullisomy



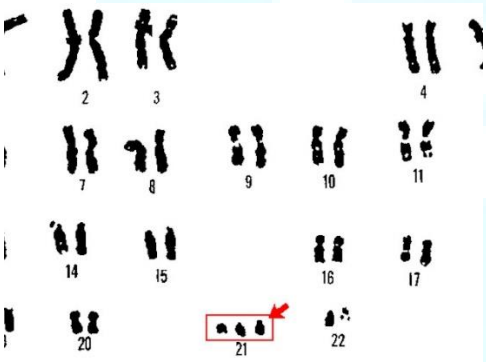
- 2 n - 2
- It is the loss of both members of a homologous pair of chromosome.
- Normally human have 2 n = 46 chromosomes
- But a nullisomic zygote have 2 n – 2 = 44 chromosomes.

Monosomy



- 2 n – 1
- It is the loss of a single crhromosome
- Human having monosomy will have 2 n – 1 = 45 chromosomes.

Trisomy



- 2 n + 1
- It is the condition of gaining a single chromosome
- A human trisomic individual has 2 n + 1 = 46 chromosomes.

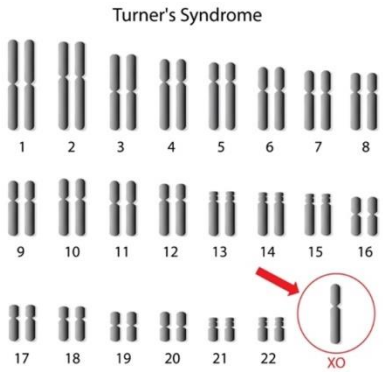
Tetrasomy

- 2 n + 2
- It is the gain of two homologous chromosomes
- Human with this condition will have 2 n + 2 = 48 chromosoms.

Aneuploidy in humans

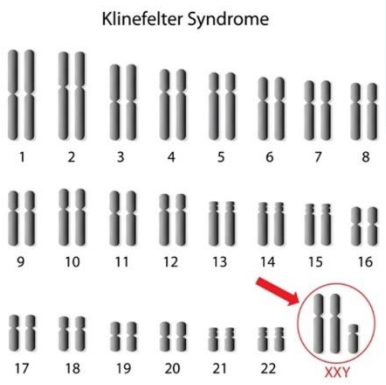
- Sex chromosome aneuploidy:

- **Turner syndrome (XO)**



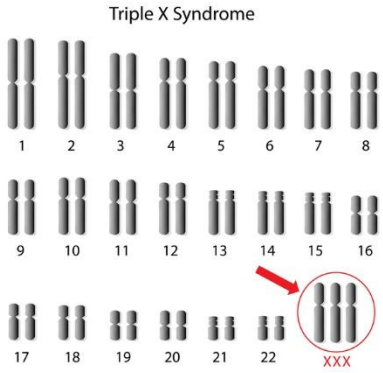
Had only a single X chromosome
At least one X chromosome is necessary for human development.
Usually embryos missing both Xs spontaneously abort in the early stages of developments.
If continued development result in female with under developed secondary sex characters
Thus they are sterile but their intelligence is usually normal.

- **Klinefelter syndrome (XXY)**



One or more Y chromosome and multiple X chromosomes in male .
XXY, XXXY, XXXXY, OR XXYY
Most of them have normal intelligence.

- **Poly X females**

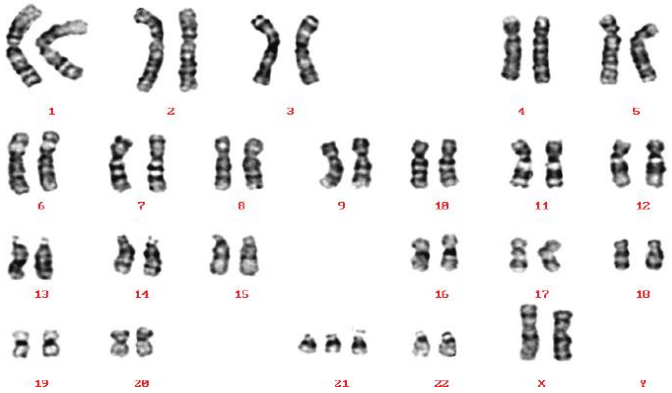


Three X chromosomes.
Also called Triple X syndrome.
No distinct features other than a tendency to be tall and thin.
Few are sterile, many menstruate regularly and are sterile.
Most XXX females have normal intelligence.

- Autosomal aneuploidy

Less common than sex chromosome aneuploids in humans.
Probably because there is no mechanism of dosage compensation for autosomal chromosomes.
Most autosomal aneuploids are spontaneously aborted.
With the exception of aneuploids of some of the small autosomes such as chromosome 21.
Because these chromosomes are small and carry fewer genes, the presence of extra copies is less detrimental than it is for larger chromosomes.

- **Down syndrome**



Trisomy 21 is also Down syndrome
47 chromosomes present
The nondisjunction event can occur in either parent
More in females.
Frequency of njndisjunction increases with maternal age.

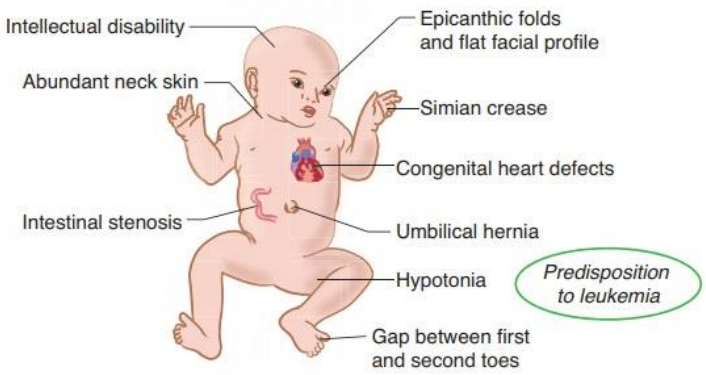


Figure 6-1. Down Syndrome

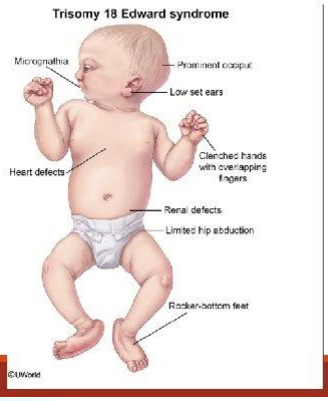
- **Edward syndrome**

Trisomy 18 (Edward Syndrome)

Genetic mechanism is nondisjunction during oogenesis

Features:

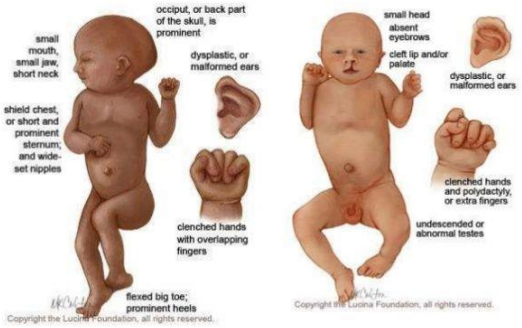
- Clenched fist, overlapping of fingers
- Rocker bottom feet
- Congenital heart defects
- Low-set ears, small lower jaw (micrognathia)
- Microcephaly
- Intellectual disability



Trisomy 18

- **Patau syndrome**

Patau's syndrome. (Trisomy 13)



Trisomy 13

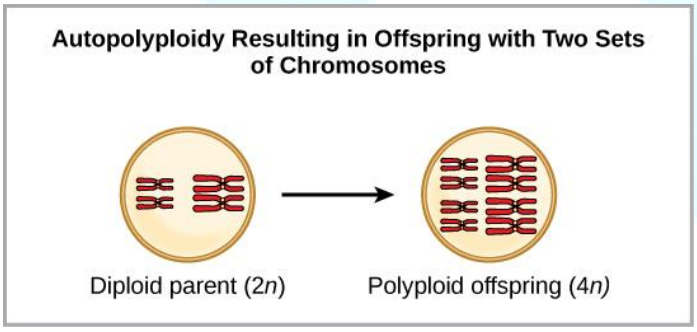
Half of children with trisomy 13 die with in the first month of life, and 95% die by the age of 3.

Polyploidy

- In polyploidy, one or more complete set of chromosomes are added.

- Most eukaryotic organisms are diploid (2n) for most of their life cycle, possessing two sets of chromosomes.
- Occasionally, whole sets of chromosomes fail to separate in meiosis or mitosis, leading to polyploidy, the presence of more than two genomic sets of chromosomes.
- Triploids(3n), tetraploids(4n), pentaploids(5n), and even higher number of chromosome sets.
- Polyploidy is common in plants and is a major mechanism by which new plant species have evolved.
- Approximately 40% of all flowering plant species and from 70% to 80% of grasses are polyploids.
- They include a number of agriculturally important plants, such as wheat, oat, cotton, potatoes and sugar cane.
- Polyploidy is less common in animals but is found in some vertebrates, fishes, salamanders, frogs and lizards.
- There are two major types of polyploids.
  I. Autopolyploidy
  II. Allopolyploidy

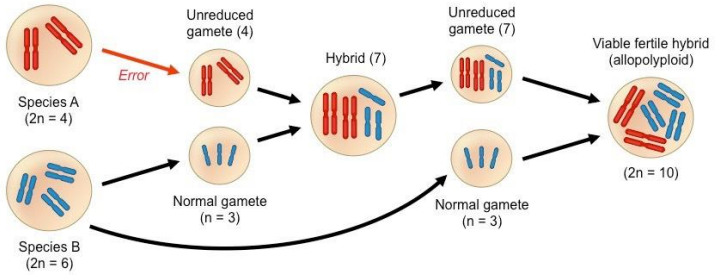
Autopolyploidy



- Here all the chromosome sets are from a single species.
- Accidents of meiosis or mitosis that produce extra sets of chromosomes, njndisjunction of all chromosomes in an early 2n embryo.
- Eg, doubles the chromosome number produces an autotetraploid (4n)
- An autotriploid (3n) is formed by njndisjunction in meiosis produces a diploid gamete and then fuses with a normal haploid gamete to produce a tripoid zydote.

- Alternatively, triploids may arise from a cross between an autotetraploid that produce 2n gametes and a diploid that produce 1n gametes.

Allopolyploids



- Allopolyploidy is the polyploidy in which chromosome sets are from two or more species.
- It means hybridization between two or more species.