Genetics

Biology Dept.3rd stage

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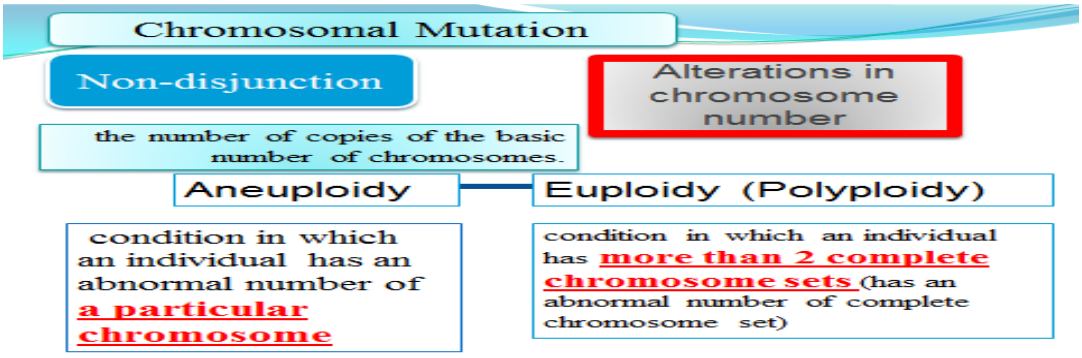
Lec-10-

NUMERICAL CHANGES IN CHROMOSOMES

Are also referred as genomic mutations because they involve variations in chromosome number of a whole genome. The number of chromosome may change in two ways, either the number of sets of chromosomes increases resulting in polyploidy or decreases leading to haploidy (*does not occur in man*), or the number of individual normal chromosomes changes giving rise to aneuploidy.

The only types of polyploidy found in man are triploidy (3n) and tetraploidy (4n) and majority of these cases end up as spontaneous abortions. The only known autosomal monosomy is the extreme rare 21 monosomy, and of the trisomics only 3, those for chromosomes 13, 18, 21.



Variation In Chromosome Number and chromosomal

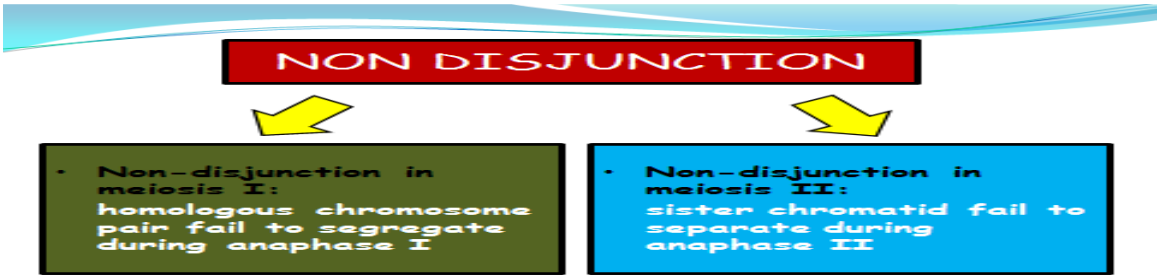
Non-Disjunction

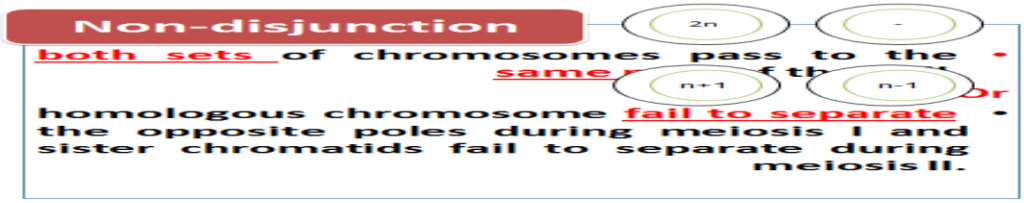
Generally during gametogenesis the homologous chromosomes of each pair separate out (disjunction) and are equally distributed in the daughter cells.

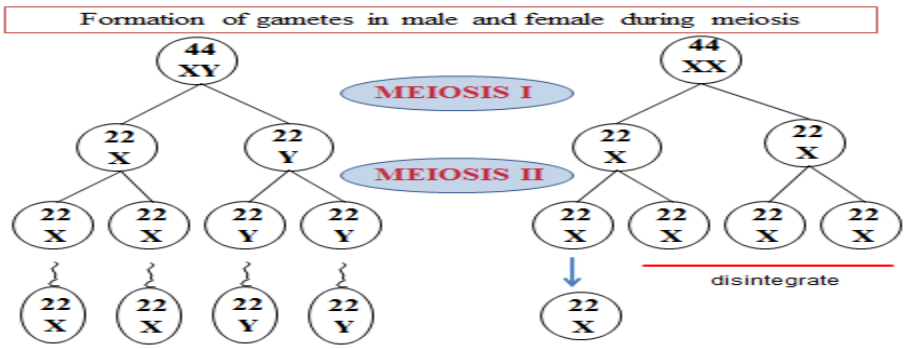
But sometime there is an unequal distribution of chromosomes in the daughter cells.

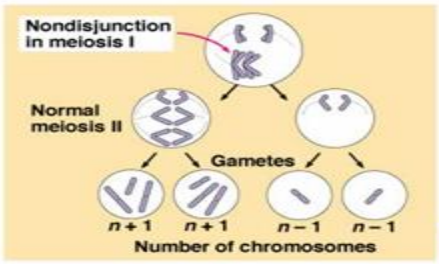
The failure of separation of homologous chromosome is called **non-disjunction**.

This can occur either during **mitosis** or **meiosis** or **embryogenesis**.



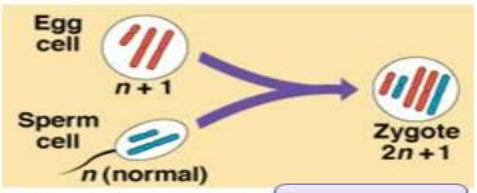




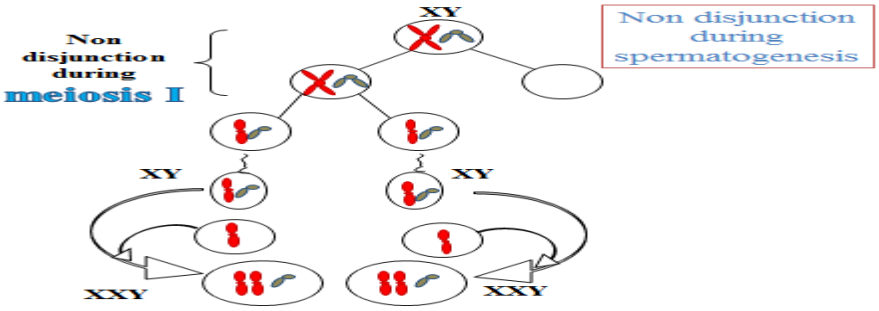


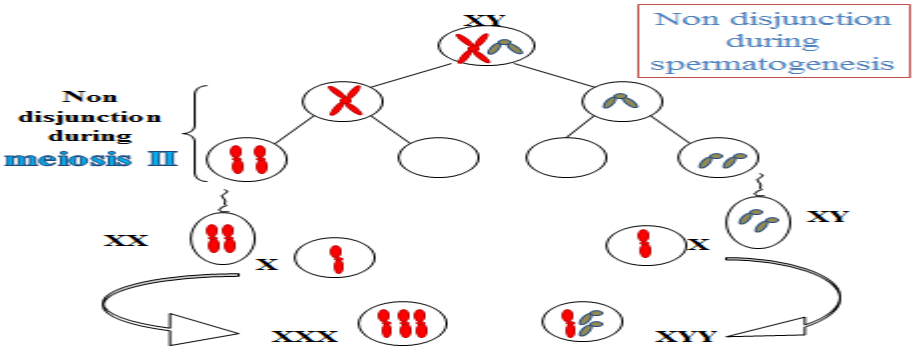
Half the daughter cells produced have an extra chromosomes (n+1) whilst the other half have a chromosome missing (n-1)

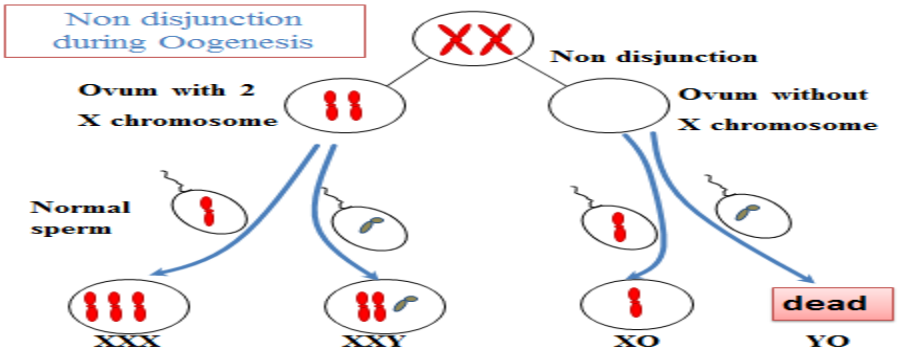
Fusion gametes between chromosome (n+1) and normal gamete (n), produced embryo with chromosome (2n+1) :



Example: Down's syndrome — Trisomy







Human aneuploidies

1. Aneuploidy. Aneuploidy is either due to the loss of one or more chromosomes (hypoploidy) or due to addition of one or more chromosomes (hyperploidy). It is of following types:

(a) Monosomy. It represents the loss of a single chromosome from the diploid set, having the genomic formula 2n-1. Since, there is lack of one complete chromosome, such aberrations create major mortality or reduced fertility.

(b) Nullisomy. It is due to the loss of a single pair of homologus chromosome and have the chromosome complement 2n-2. It should not be confused with double monosomy (2n-1-1) where the missing chromosomes are also two in number, but they are non-homologus as in wheat.

(c) Trisomy. Here, the organisms have an extra chromosome (2n+1). It is commonly found in humans and is responsible for following three syndromes:

- **21 Trisomy (Down's syndrome, Mongolism).** This is a least severe condition of autosomal trisomy, but most frequent.
- **Trisomy 18; Edward Syndrome**

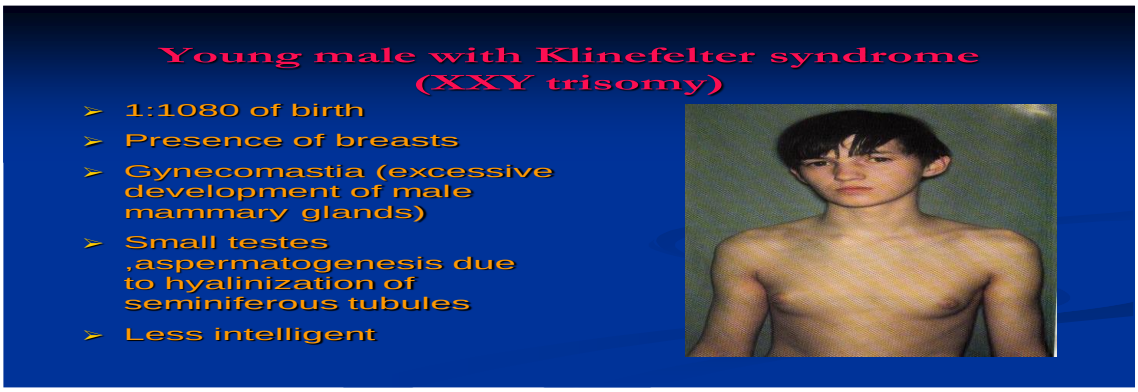
47; +18 ------1/8000 live births; maternal age affect-low birth weight-multiple dysmorphic features-chin, ears, single palmar crease, clenched hands-

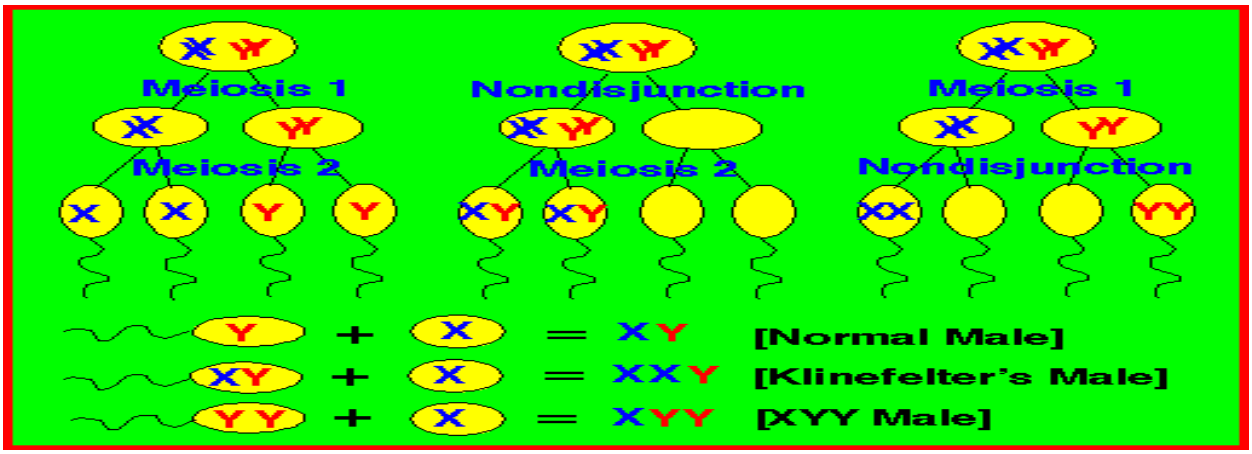
malformations of the brain, heart, kidneys, and other organs-

Trisomy 13; Patau Syndrome

- **Known aneuploidies**

- Turner syndrome (45,X or X0)
- Klinefelter syndrome (47,XXY)
- Jacobs syndrome (47,XYY)
- Down syndrome (47,+21 or Trisomy 21)
- Patau syndrome (47,+13 or Trisomy 13)
- Edwards syndrome (47,+18 or Trisomy 18)





Genome Mutation or Polyploidy

Polyploidy is a change in the quantity of total genomes. This type of mutation affects chromosome content of an organism. Humans are diploid creatures; that is, for every chromosome in our body, there is another one to match it. If an organism possesses multiples of the haploid number of chromosomes, it is called euploid.

Human and other eukaryotes are diploid (2*n*).

It is possible for a species, particularly a plant species, to produce offspring that contains more chromosomes than its parent.. Polyploids have important applications in plant breeding and agriculture and are responsible for the creation of thousands of species in today's planet, and will continue to do so. They are also responsible for increasing genetic diversity and producing species showing an increase in size, vigor, and increased resistance to disease.

Polyploidy

- Organism with multiples of the haploid number of chromosome (3n, 4n, 5n, ...)
- Many plants important for agriculture are the products of polyploidy.
  - For example, oats, cotton, potatoes, tobacco, and wheat are polyploid.

Genome	Polyploidy
3n	Triploid
4n	Tetraploid
5n	Pentaploid
6n	Hexaploid
8n	Octaploid
10n	Decaploid