Lecture 1(a)

Chromosomal Aberrations

Numerical changes in chromosomes

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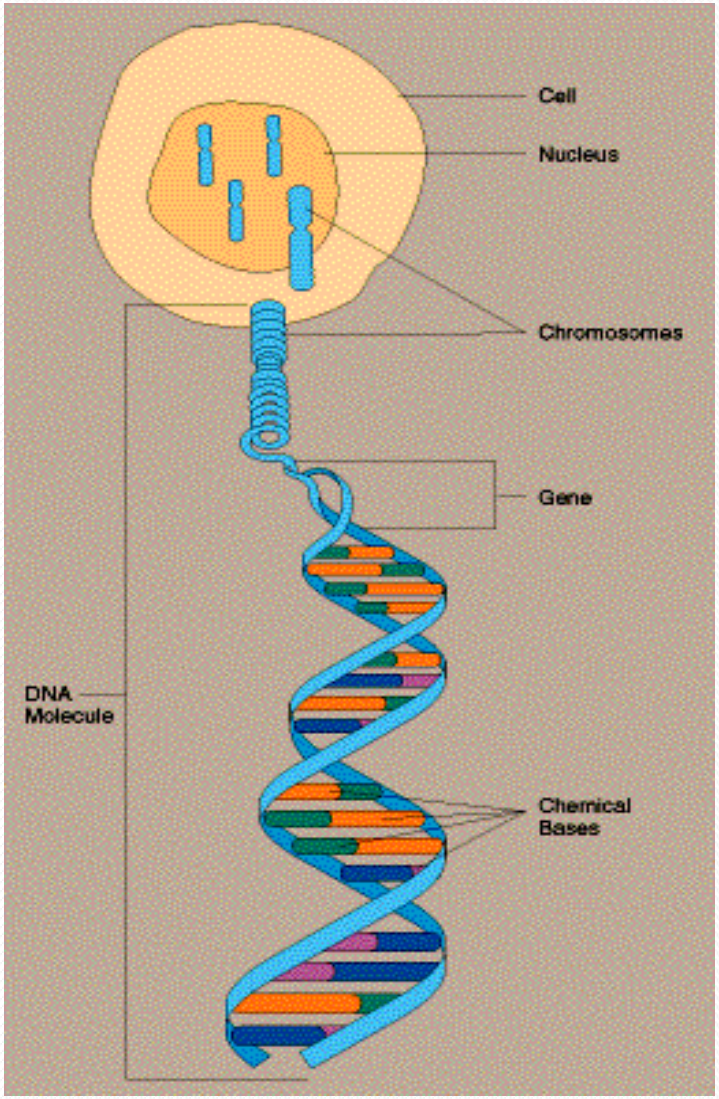
Basic definitions

- Chromosomes, DNA and genes
- Karyotype
- Definition of Chromosomal aberrations
  - Structural and numerical changes
  - Numerical changes
  - Euploidy
  - Aneuploidy

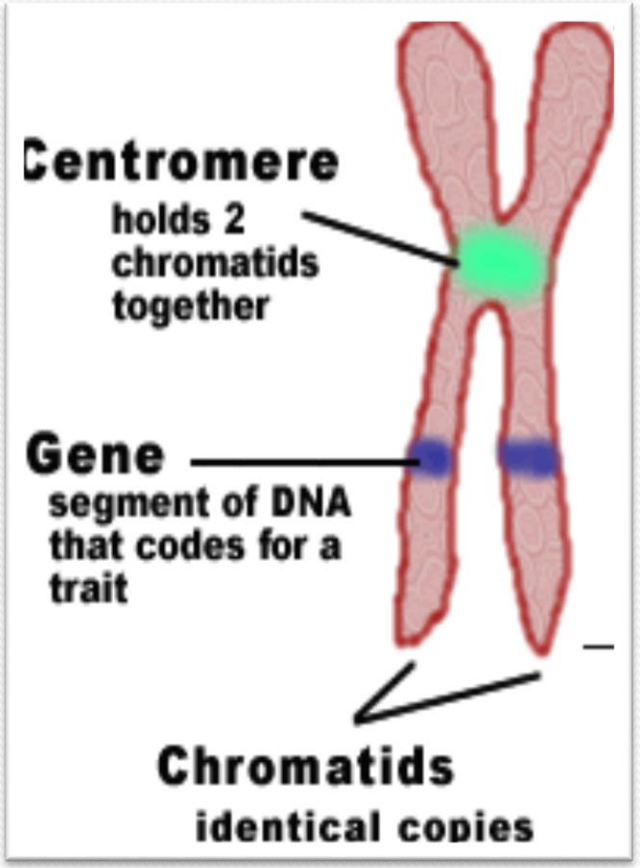
Chromosomes

- The **chromosome** carries the genetic information.
- composed of **deoxyribonucleic acid (DNA)** on framework of protein .
- Segments of DNA molecules comprise the genes; the units of heredity.



Chromosomes

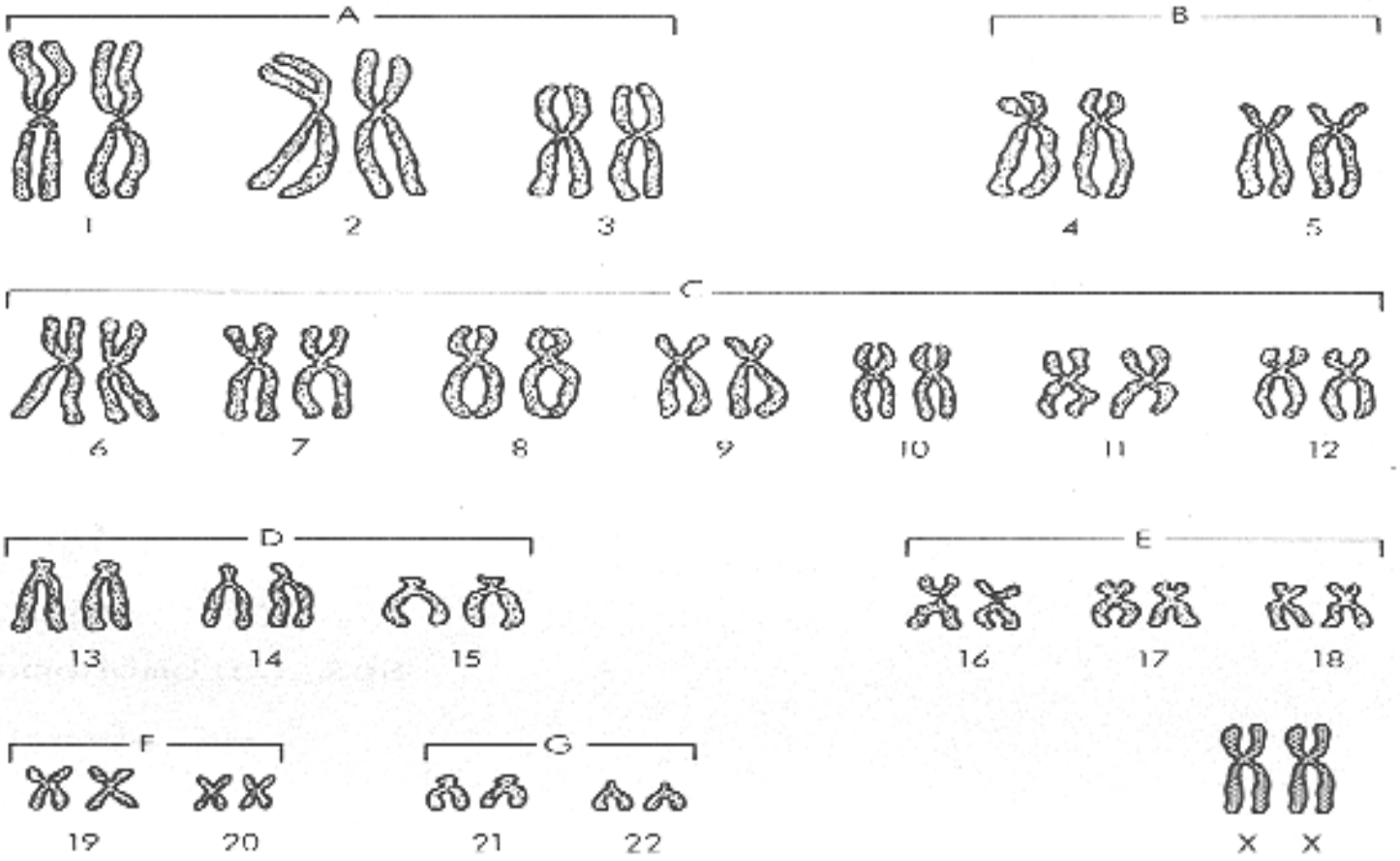
- During cell division, the chromosome can be seen to consist of 2 parallel strands; the **chromatids**, held together at one point, the **centromere.**



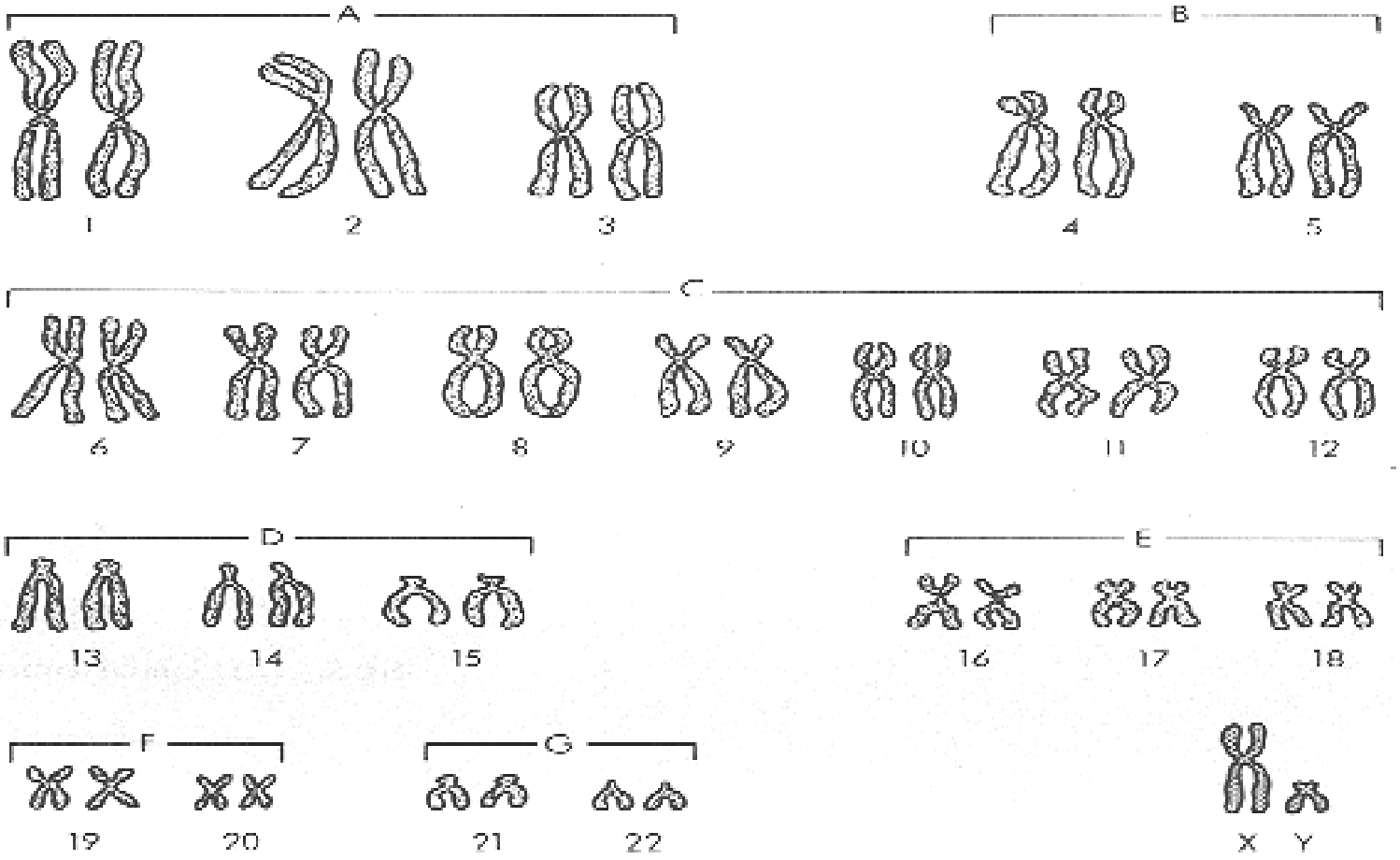
Karyotype

- It is the set of chromosomes of an individual.
- It is the systematized arrangement of the chromosomes of a single cell.
- In the human cell, there are 46 chromosomes or 23 pairs (diploid number); of these 23 pairs, 22 are similar in both sexes and are called the ***autosomes.*** The remaining pair is called ***sex chromosomes*** : XX in the female cells and XY in the male cells .
- Chromosomes are arranged in groups A to G according to their shape & size.

Karyotype of a normal female in human



Karyotype of a normal male in human



Normal Male

CHROMOSOMAL ABERRATIONS

- **A chromosome aberration, disorder, anomaly, abnormality, or mutation is a missing, extra, or irregular portion of chromosomal DNA. OR**
- **Mutations that cause change in the structure or number of chromosomes are called chromosomal aberrations. Generally, the incidence of chromosomal abnormalities is 5-6 persons/1000.**
- **Many children with a chromosomal abnormality have mental or physical birth defects.**

HISTORY OF CHROMOSOMAL ABERRATIONS

- In 1959 two discoveries opened a new era of genetics.
- Jerome Lejeune, Marthe Gautier, and M. Raymond Turpin discovered the presence of an extra chromosome in Down syndrome patients.
- C. E. Ford and his colleagues, P. A. Jacobs and J. A. Strong first observed sex chromosome anomalies in patients with sexual development disorders.

Types of Chromosomal aberrations

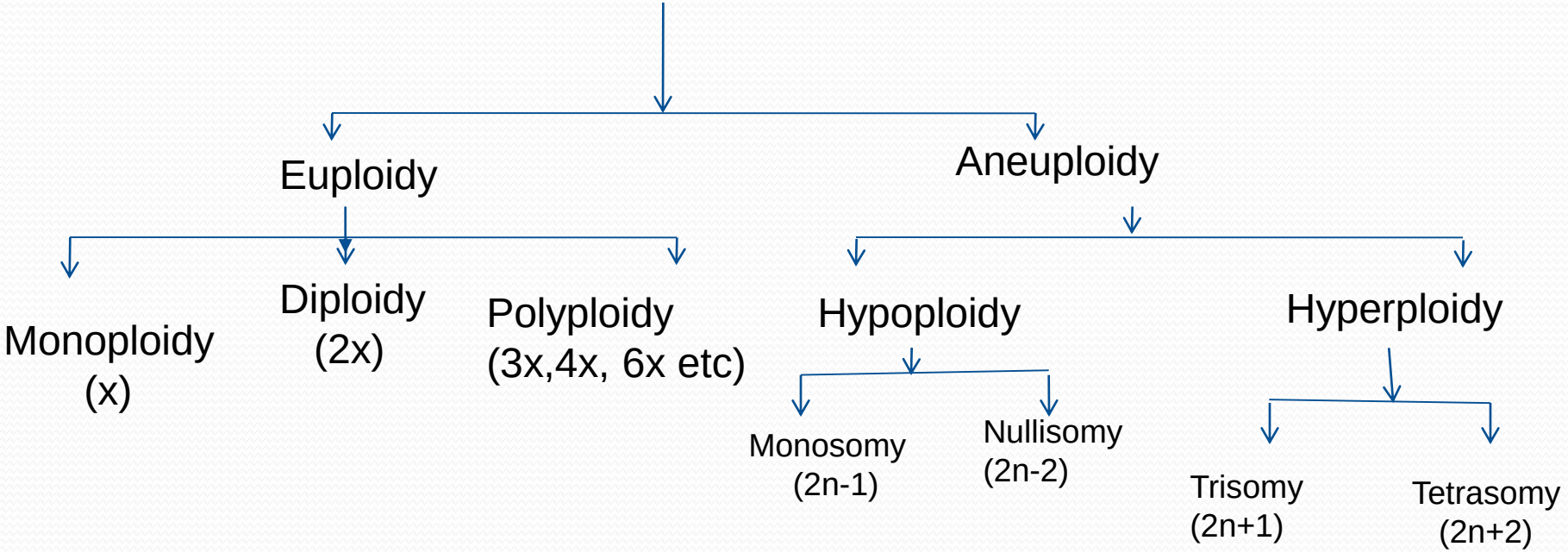
- Chromosomal aberrations are either **numerical or structural.**
- They are a very common cause of early spontaneous miscarriage.
- Usually, but not always, cause multiple congenital anomalies and learning difficulties.

Chromosomal Aberrations (abnormalities)

- **Structural Aberrations**
  - Deletion
  - Duplication
  - Inversion
  - Translocation

- **Numerical Aberrations**
  - Euploidy: the usual number and sets of chromosomes
  - Polyploidy: the presence of three or more complete sets of chromosomes
  - Aneuploidy: the presence of additional chromosomes or missing individual chromosomes

Numerical aberrations in Chromosomes



Euploidy

- Presence of extra chromosome is exact multiple of basic chromosome number or,
- Organism should posses one or, more full sets of chromosomes.
- Euploids can be monoploids, diploids or polyploids

Monoploidy

- An individual that contains one half the normal number of chromosomes is a **monoploid** and exhibits **monoploidy.**
- Some species such as bees, ants and male bees are normally monoploid

Common Wheat posses monoploidy

- The chromosomes of common wheat are believed to be derived from three different ancestral species, each of which had 7 chromosomes in its haploid gametes. The monoploid number is thus 7 and the haploid number is $3 \times 7 = 21$. In general n is a multiple of x. The somatic cells in a wheat plant have six sets of 7 chromosomes: three sets from the egg and three sets from the sperm which fused to form the plant, giving a total of 42 chromosomes. As a formula, for wheat $2n = 6x = 42$, so that the haploid number n is 21 and the monoploid number x is 7.

Polyploidy

- Autopolyploids: polyploids created by chromosome duplication within a species
- Allopolyploids: polyploids created by hybridization between different species (homeologous chromosomes)

1. Autopolyploidy

- Even-numbered multiples of haploid number of chromosomes. e.g.-

(a) Triploidy (23x3 or 63 chromosomes)

(b) Tetraploidy (23x4 or 92 chromosomes)

(c)Hexaploidy (23x6 or 138 chromosomes)

(d)Octaploidy (23x8 or 184 chromosomes)

Polyploidy

conti…

- Triploidy is almost always sterile (bananas are propagated by cuttings, some apple species by grafts,
- Triploids are often formed by fusion of a diploid (2x) gamete from a tetraploid (4x) parent with a normal gamete from a diploid parent.
- Autopolyploidy can also be induced artificially with colchicine.

- **Autotriploids** are known in watermelons, sugarbeet, tomato, grapes and banana, although in several of these cases the polyploids have been artificially produced.
- **Autotetraploids** are known in rye (*Secale cereale*), corn (*Zea mays*), red clover (*Trifolium pratense*), berseem (*Trifolium alexandrium*), marigolds *Tagetes*), snapdragons (*Antirrhinum*), *Phlox*, grapes, apples, etc.

2. Allopolyploidy

- Polyploidy may result from doubling of chromosome number in a F_1 hybrid which is derived from two distinctly different species.
- Let A represent a set of chromosomes (genome) in species X, and let B represent another genome in a species Y. The F_1 will then have one A genome and another B genome. The doubling of chromosomes in this F_1 hybrid (AB) will give rise to a tetraploid with two A and two B genomes . Such a polyploid is called an allopolyploid or **amphidiploid.**

Allopolypoidy

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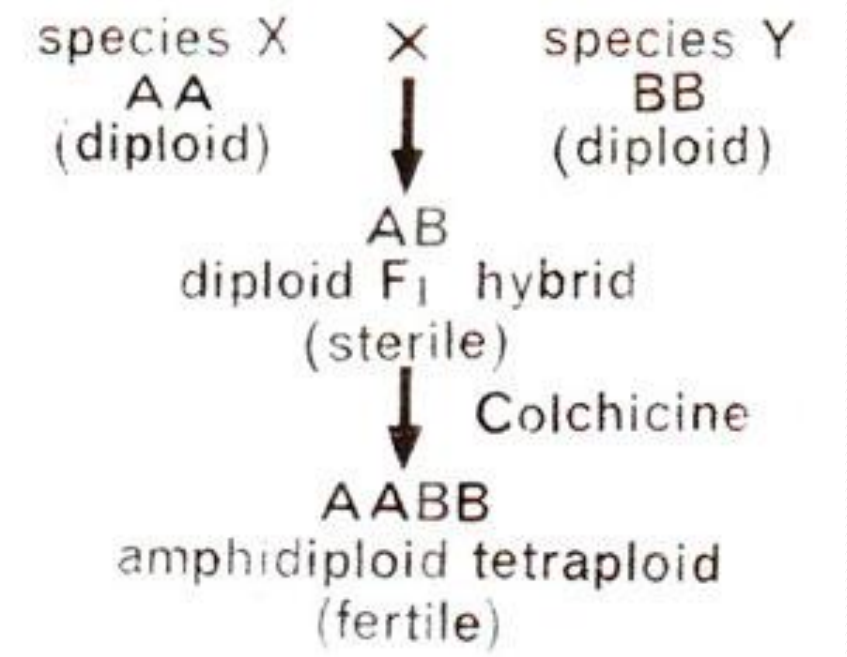
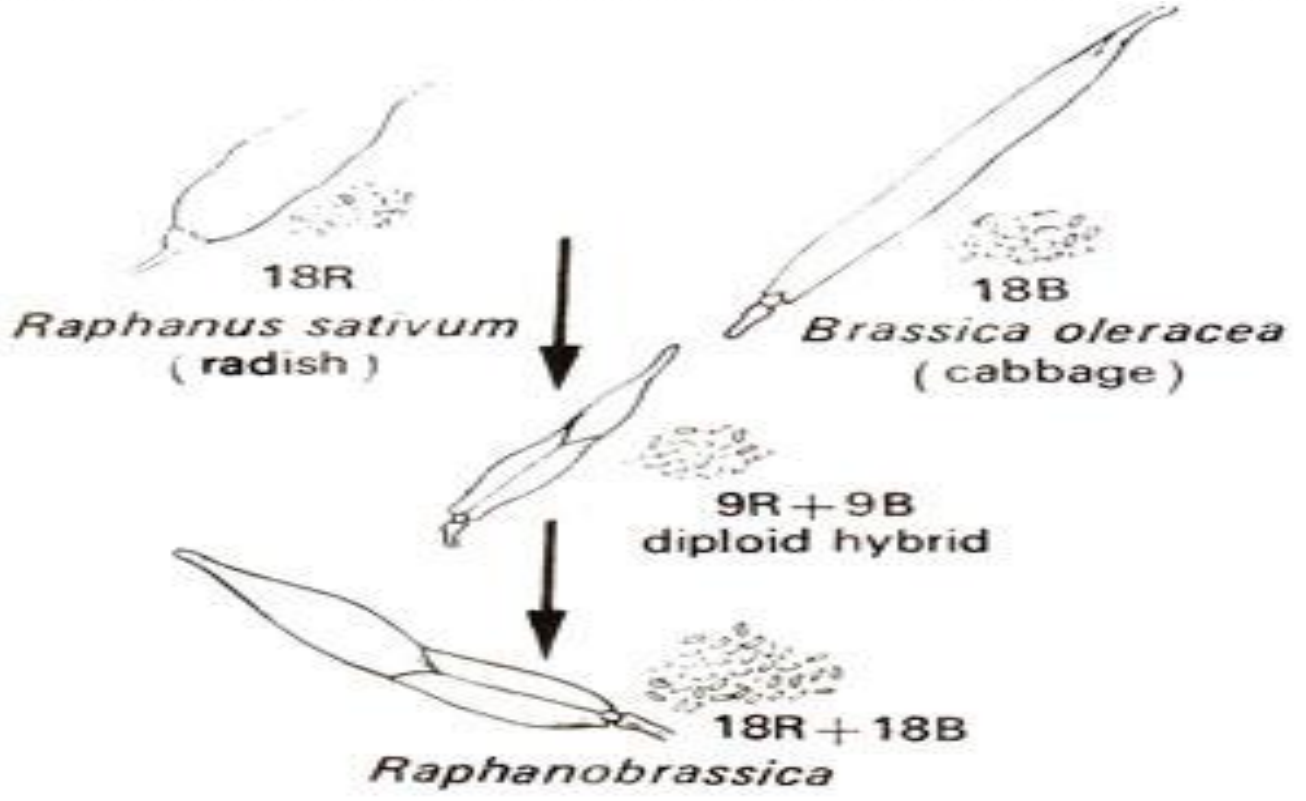


Raphanobrassica is a example of allopolyploidy

- In 1927, G.D. **Karpechenko,** a Russian geneticist, reported a cross between *Raphanus sativus* (2*n* = 18) and *Brassica oleracea* (2*n* = 8) to produce F_1hybrid which was completely sterile.
- On cytological examination these fertile plants were found to have 2*n* = 36 chromosomes, which showed normal pairing into 18 bivalents

Raphanobrassica conti…



Common cultivated wheat is an example of allopolyploidy

- There are three different chromosome numbers in the genus *Triticum*, namely $2n = 14$, $2n = 28$ and $2n$ - 42.
- The common wheat is hexaploid with $2n = 42$, and is derived from three diploid species :
- (i) AA = *Triticum aegilopoides* ($2n = 14$),
- (ii) BB = *Aegilops speltoides* ($2n = 14$)
- (iii) DD = *Aegilops squarrosa* ($2n = 14$).
- The hexaploid wheat is designated as AABBDD,

Allopolyploidy in wheat

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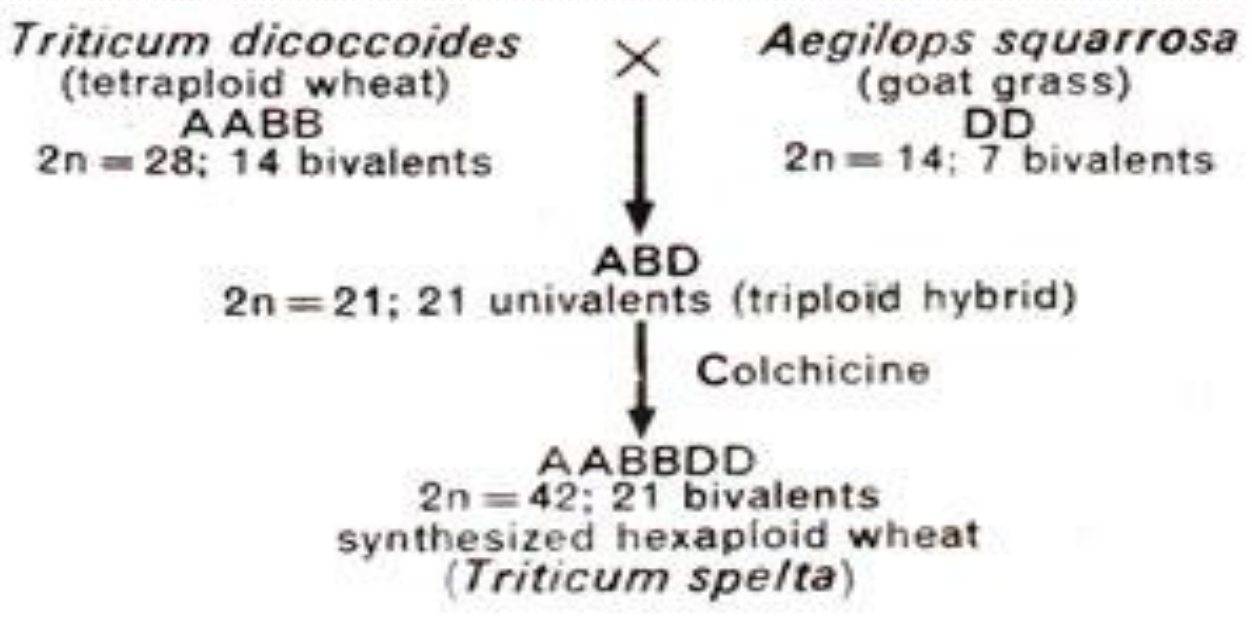


Allopolyploidy examples

(i) **Cotton:** The new world cotton (*Gossypium hirsutum*) J.O. Beasley crossed old world cotton (*Gossypium herbaceum*) with American cotton (*Gossypium raimondii*) and doubled the chromosome number in F1 hybrids. The allopolyploid thus produced resembled the cultivated new world cotton (*Gossypium hirsutum*) and when crossed, with it gave fertile F1 hybrids.

(**ii)Tobacco:** There are two cultivated species of tobacco. i. e. *Nicotiana tabacum* and *Nicotiana rustica*. *Nicotiana tabacum* is an allotetraploid and available evidence suggests that it is derived from a cross between *Nicotiana sylvestris* x *Nicotiana tomentosa*

Aneuploidy

1. **Aneuploidy:** Individuals have a numerical change in part of the genome. The chromosome number of aneuploids is not an exact multiple of the haploid number, n.
2. **Hypoploidy:** an organism in which a chromosome (or part thereof) is under represented.
3. **Hyperploidy:** an organism in which a chromosome (or part thereof) is overrepresented.

Types of Hypoploidy

Monosomy (2n-1):

- The diploid organism which lacks one chromosome of a single homologous pair is called monosomic with genomic formula 2n-1.
- **Turner Syndrome:** (XO) female with retarded sexual development who is usually sterile.

Nullisomy (2n-2):

- Diploid organisms which have lost a pair of homologous chromosomes are called nullisomics with genomic formula 2n-2.
- Humans with this condition will not survive.

Types of Hyperploidy

Trisomy(2*n*+1): A particular chromosome represented three times

- Trisomics were obtained for the first time in *Datura stramonium* (jimson weed) by **A.F. Blakeslee** and his co-workers.

Tetrasomy (2n+2): A particular chromosome represented four times.

- The four homologues tend to form a quadrivalent at meiosis and disjunction often proceeds fairly regularly, two by two. All the 21 possible tetrasomics in wheat are viable.

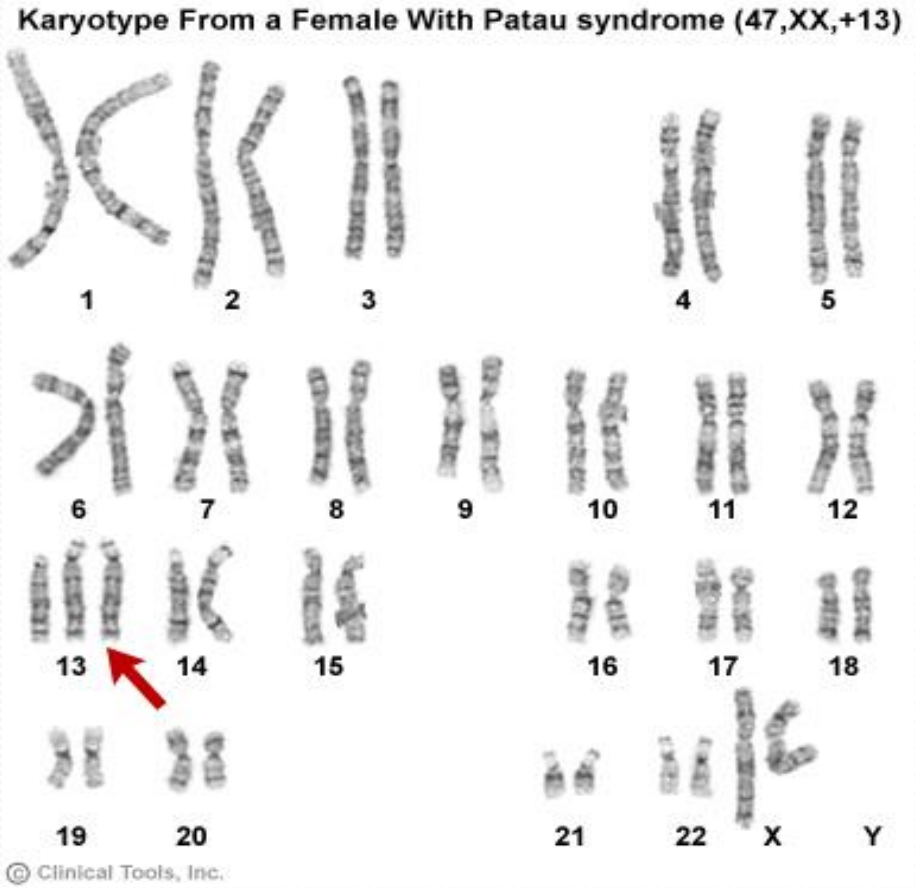
Trisomy of Autosomes in Human beings

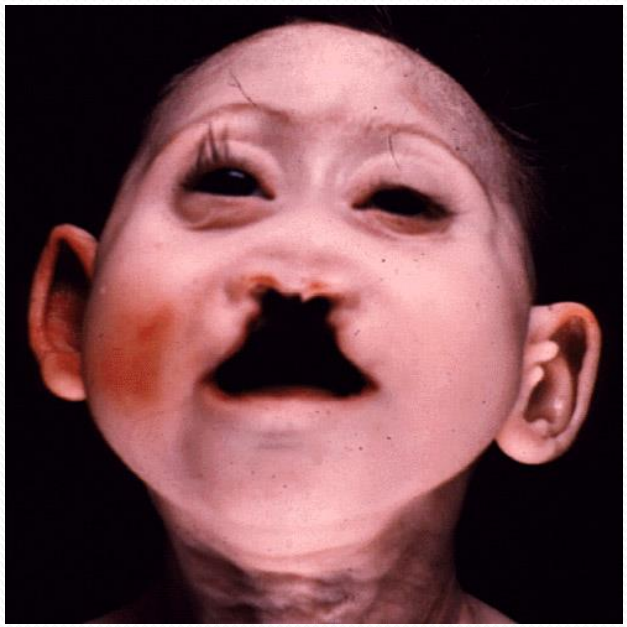
- Trisomy 13 or D-trisomy (Patau syndrome)
- Trisomy 18 or E-trisomy (Edward syndrome)
- Trisomy 21 or G-trisomy (Down syndrome)

Trisomy 13 (Patau Syndrome)

- 1st described by Bartholin (1657) & redefined by Patau (1960).
- Chromosomal complement: 47,XX,+13 (female) or 47,XY,+13 (male)
- Phenotype: Male or female
- Incidence: 1:12,000 (increases with the age of mother)

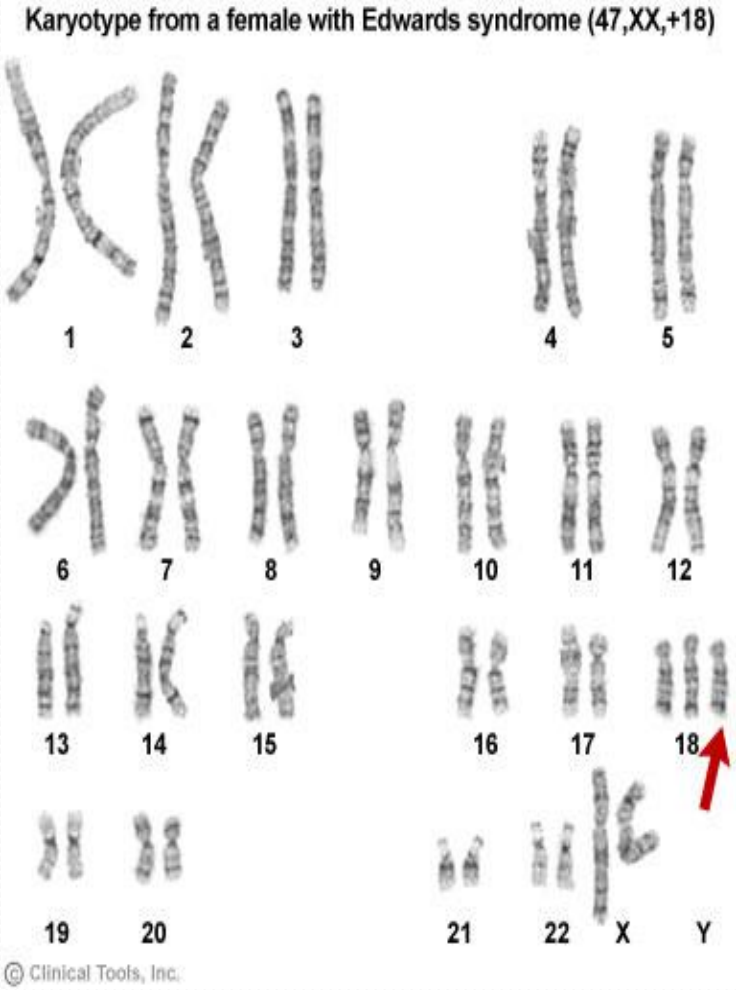
Patau syndrome

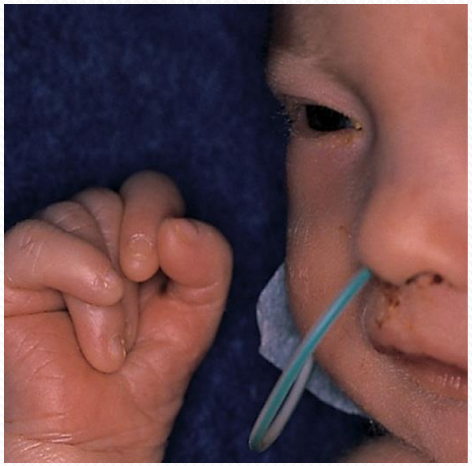




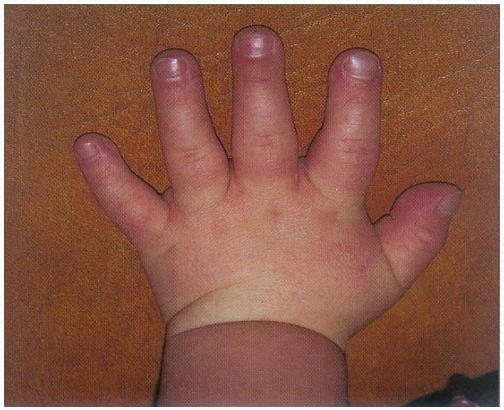
Trisomy 18 (Edward Syndrome)

- Chromosomal complement: 47,XX,+18 (female) or 47,XY,+18 (male)
- Phenotype: Male or female
- Incidence: 1:8000





Overlapping of the fingers in Edwards' syndrome



Short broad hand

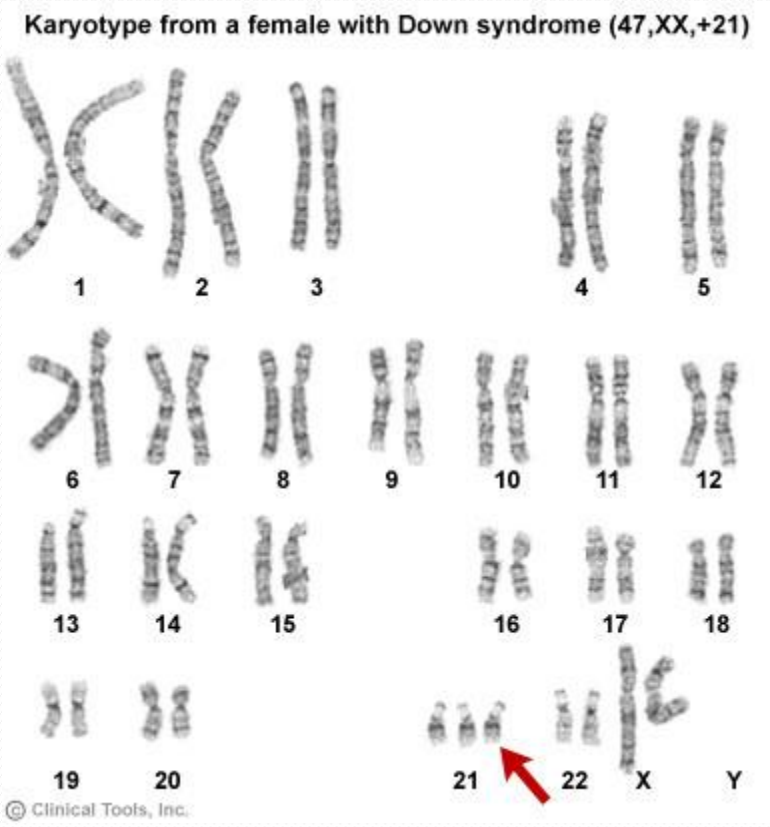
Down Syndrome (Mongolism) Trisomy 21

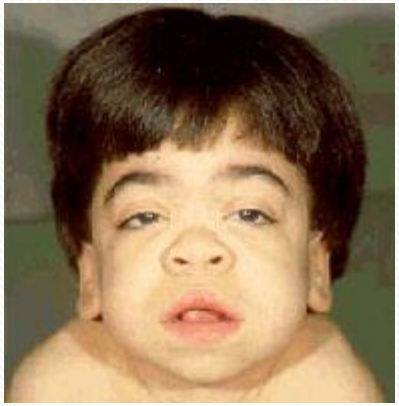
Incidence :

- The most common chromosomal aberration
- Incidence 1/700 live birth & 10 % of M.R.

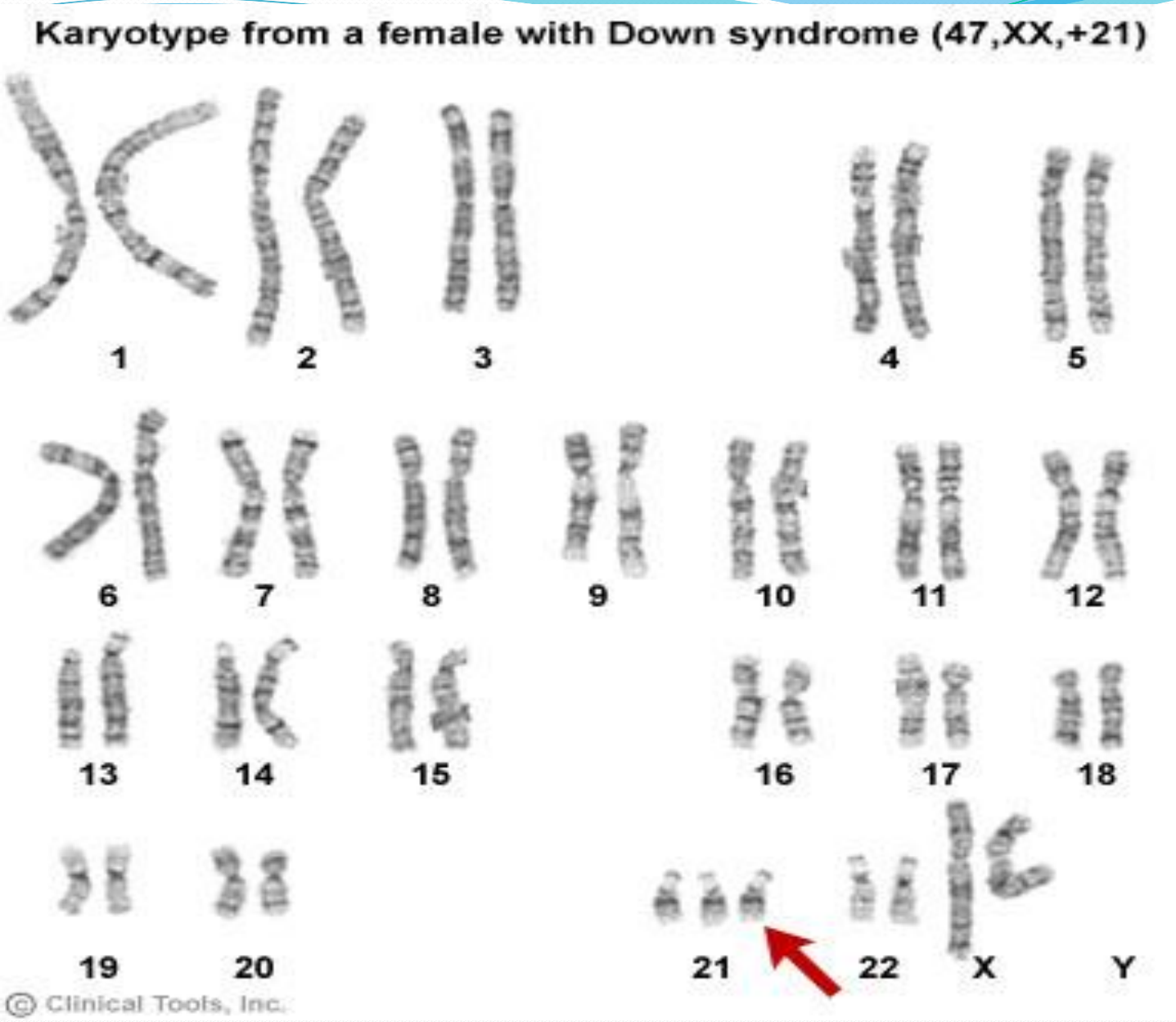
Definition : It is trisomy 21 i.e. the cell contain an extra chromosome, number 21 i.e. the cell contains three 21 chromosomes instead of two .

Down Syndrome





Cardiofaciocutaneous Syndrome
Antimongoloid slant


Karyotype from a female with Down syndrome (47,XX,+21)
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2
3
4
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16
17
18
19
20
21
22
X
Y
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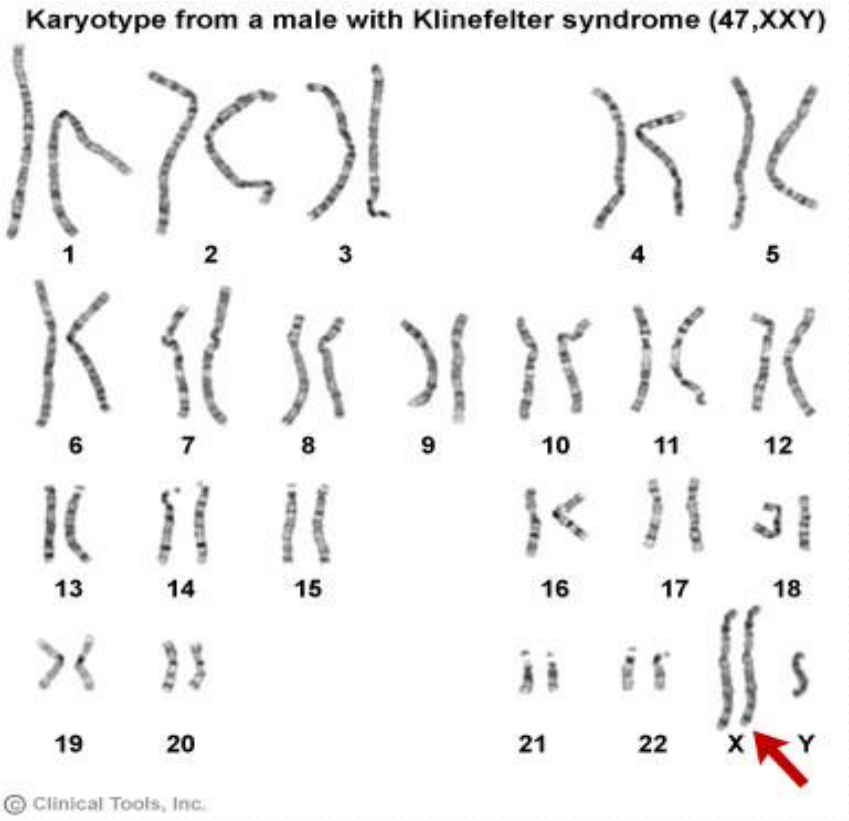
Trisomy of Allosome in Human beings

- **<u>Klinefelter Syndrome</u>**
- Trisomy of sex chromosome - XXY

  (An additional X chromosome in males)
- Occurrence – 1 in 500-1000 males



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Thank you….