ANEUPLOIDY

Subject : Advanced Genetics
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Key Points

- Aneuploidy.
- Non-disjunction.
- Types of Aneuploidy.
- Aneuploidy in Humans.

Aneuploidy

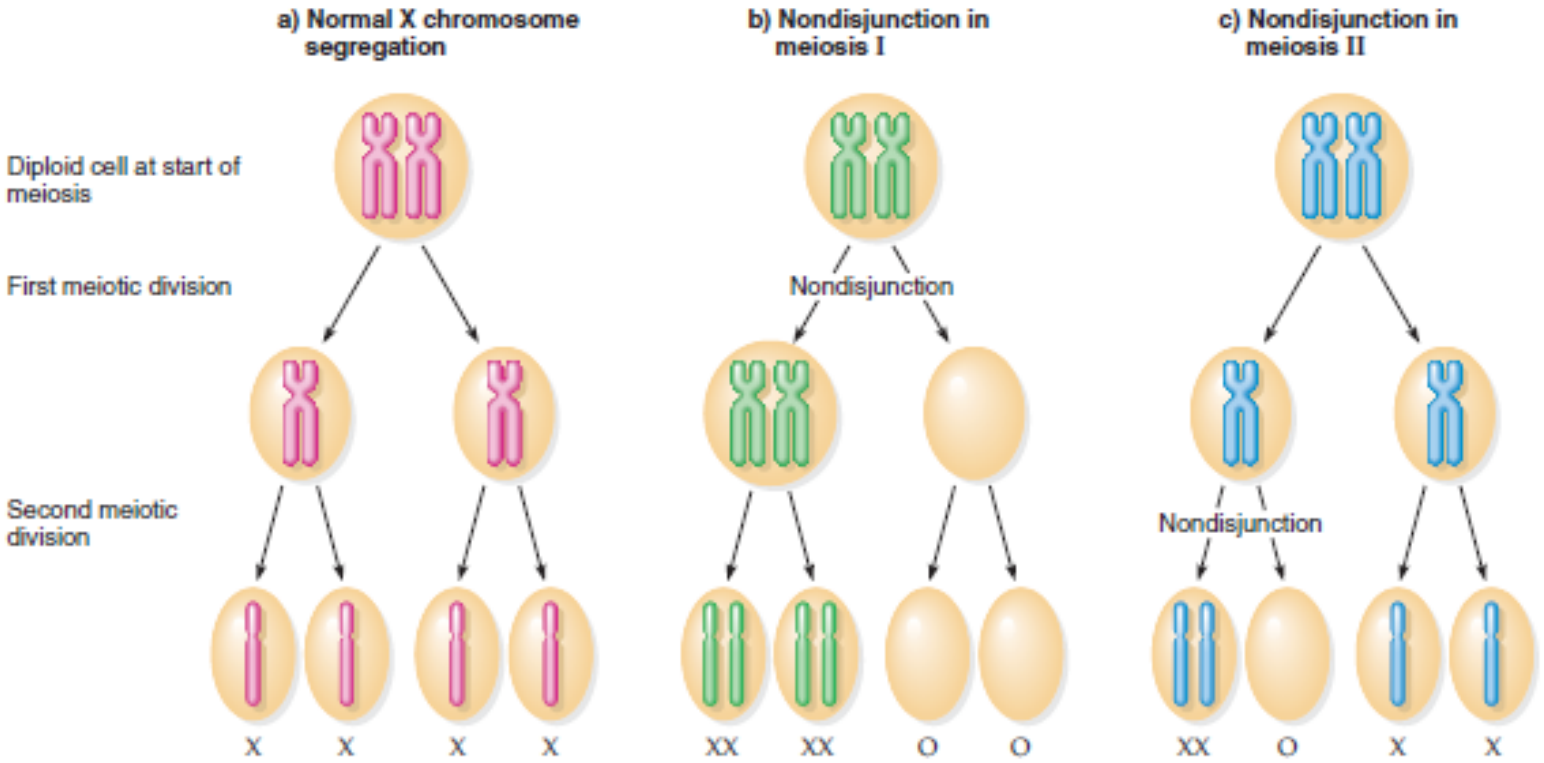
- Aneuploidy is presence of abnormal number of chromosome in a cell.
- Aneuploid cell has one or more chromosome lost or added to a normal set of chromosomes.

Non-disjunction

- Generally during gametogenesis the homologus chromosomes of each pair separate out (disjunction) and are equally distributed in the daughter cells.
- But sometime there is unequal distribution of chromosome in the daughter cells.
- The failure of separation of homologous chromosome is called non-disjunction.
- This can occur in mitosis (in rare) and in meiosis.

Figure 12.18

Nondisjunction in meiosis involving the X chromosome. Nondisjunction of autosomal chromosomes and of all chromosomes in mitosis occurs in the same way.



Types of Aneuploidy

- Nullisomy (2N-2) – Involves a loss of one homologous chromosome pair.
- Monosomy (2N-1) – Involves a loss of single chromosome.
- Trisomy (2N+1) – Involves a single extra chromosome.
- Tetrasomy (2N+2) - Involves an extra chromosome pair.
- Double monosomic (2N-1-1) – It has two separate chromosome present in only one copy each.
- Double tetrasomic (2N+2+2) – It has two chromosomes present in four copies each.

Figure 16.15

Normal (theoretical) set of metaphase chromosomes in a diploid (2N) organism *(top)* and examples of aneuploidy *(bottom)*.

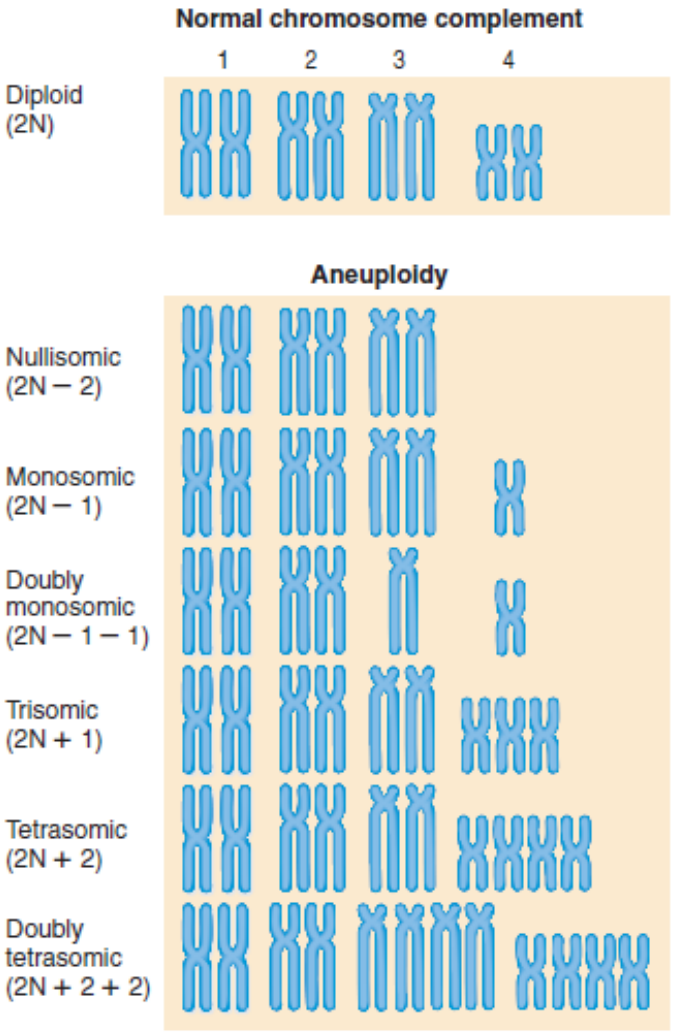
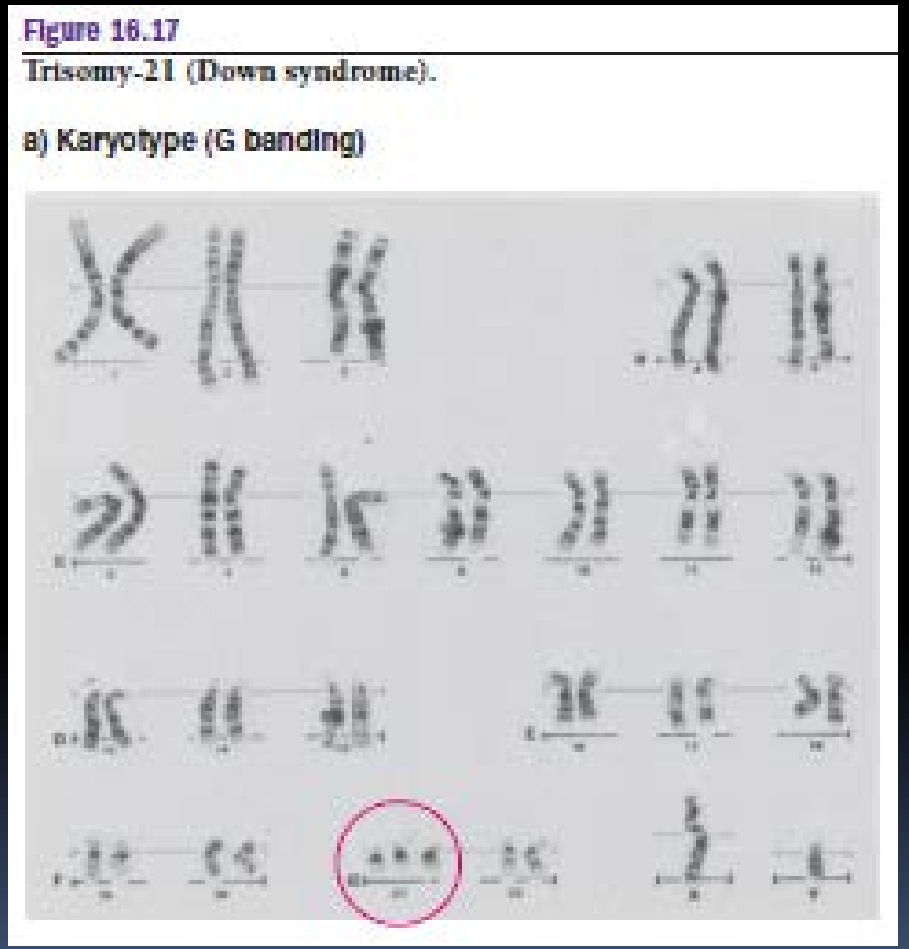


Table 16.1 Aneuploid Abnormalities in the Human Population

Chromosomes	Syndrome	Frequency at Birth
Autosomes		
Trisomic 21	Down	14.3/10,000
Trisomic 13	Patau	2/10,000
Trisomic 18	Edwards	2.5/10,000
Sex chromosomes, females		
XO, monosomic	Turner	4/10,000 females
XXX, trisomic	Viable; most	
XXXX, tetrasomic	are fertile	14.3/10,000 females
XXXXX, pentasomic		
Sex chromosomes, males		
XYY, trisomic	Normal	25/10,000 males
XXY, trisomic		
XXYY tetrasomic	Klinefelter	40/10,000
XXXY, tetrasomic		

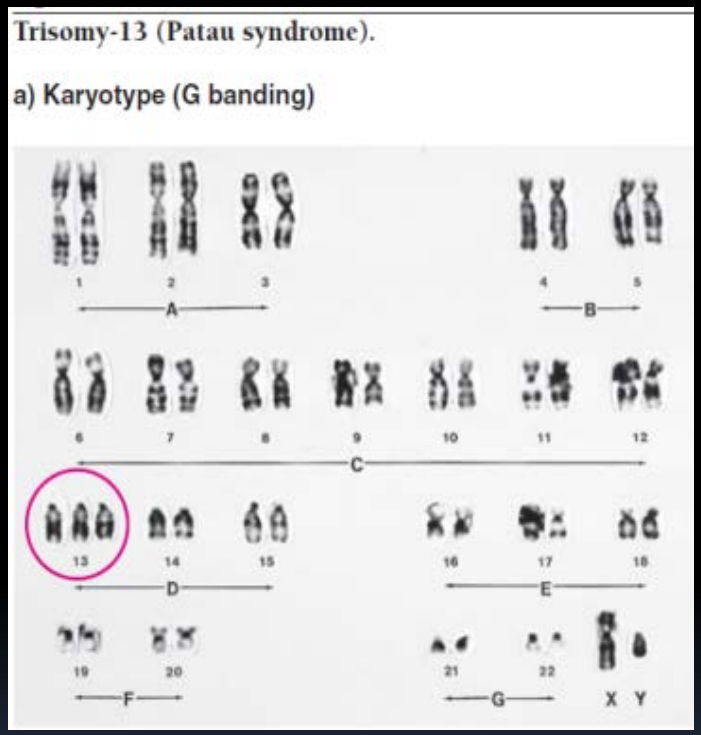
Trisomy-21

- Occurs when there are three copies of chromosome 21.
- Occurs about 14.3 in 1 million live births
- Individuals with trisomy-21 have Down syndrome , characterised by low IQ, epicanthal folds, short and broad hands, and below average height.



Trisomy-13

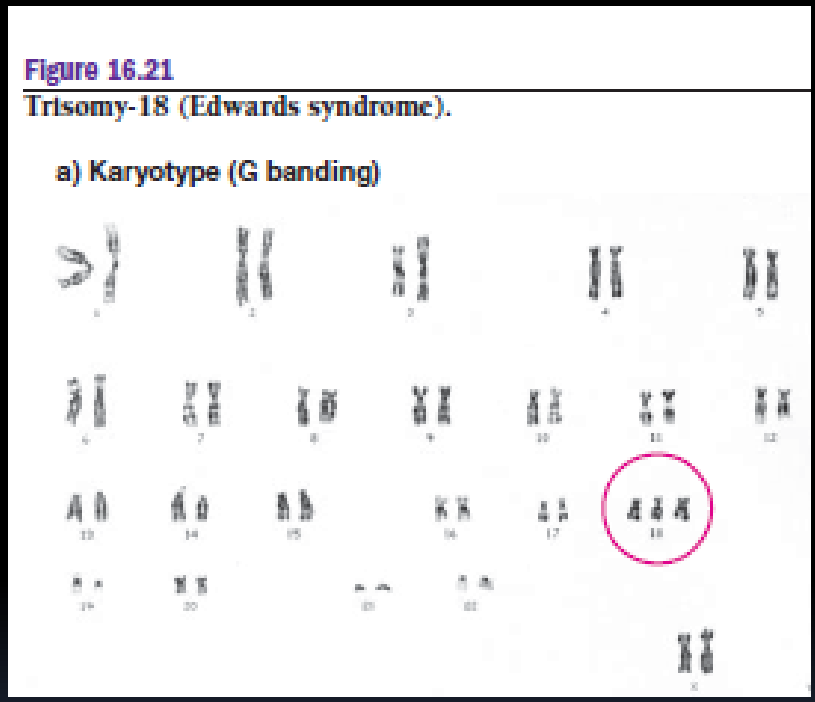
- Trisomy-13 produces Patau Syndrome.
- Occurs in about 2 in 10,1000 live births.
- Characterisitics of individuals with trisomy-13 include cleft lip and palate, small eyes, polydactyly (extra fingers & toes), mental retardation & cardiac abnormalities.
- Most infants die before the age of 3 months.



Trisomy-18

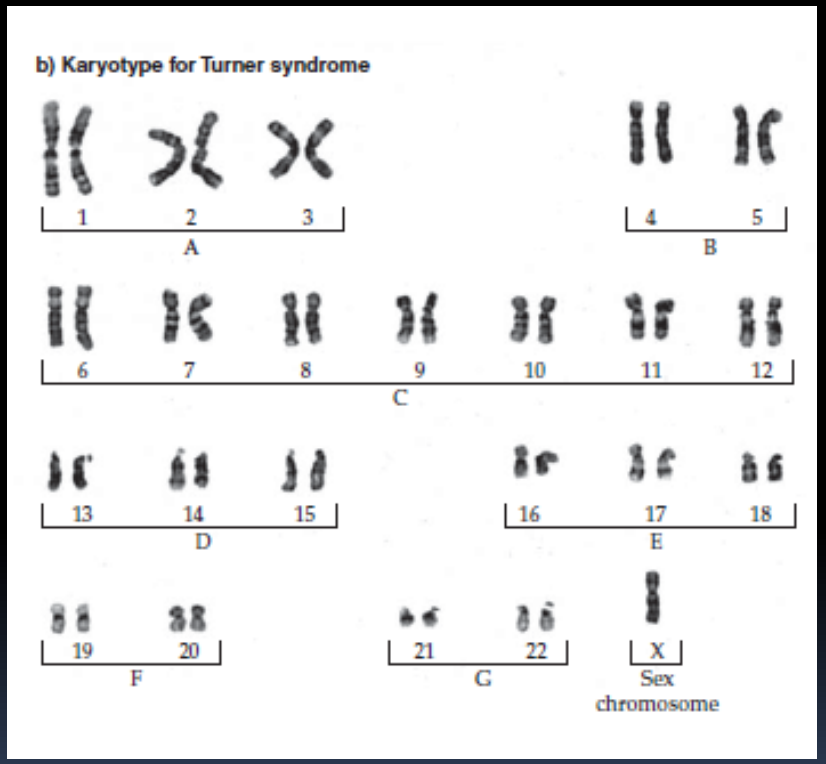
- Trisomy-18 produces Edwards syndrome.
- Occurs in about 2.5 in 10,000 live births.
- 80% of infants with this syndrome are female.
- Characteristics shown by individuals are :

  Clenched fists, an enlongated skull, low-set malformed ears, mental & developmental retardation.
- Most of infants with trisomy-18 die within 6 months.



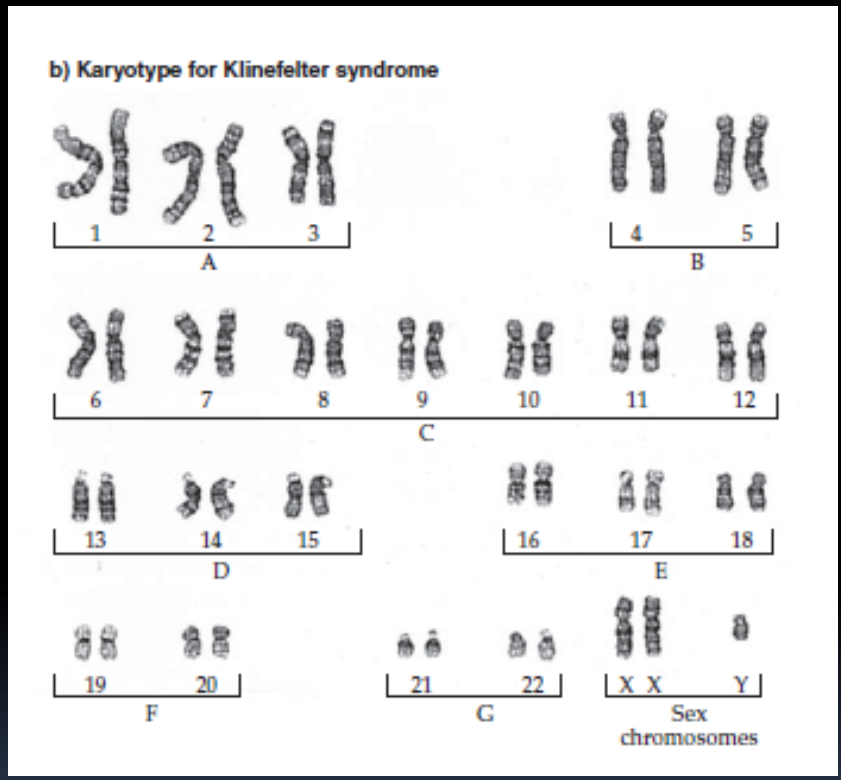
Turner Syndrome (XO)

- Occurs with a frequency of 1 in every 10,000 females born.
- Fail to develop secondary sexual characteristics, tend to be shorter and have weblike necks, poor developed breasts, immature internal sexual organs.



Klinefelter Syndrome (XXY)

- About 1 in 1000 males born have this syndrome.
- Males have underdeveloped testis and are taller than the average male.
- Some breast development is seen in about 50% of affected individuals ; and some show subnormal intellegence.



Reference

- Peter J. Russell igenetics : A molecular approach

Thank you...