Biology Grade 9

CHAPTER 7: CHROMOSOMES, CARRIERS OF GENETIC INFORMATION

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Activity 4: Chromosomes and traits of the individual

- If the chromosomes are the carriers of genetic information, any change in the karyotype is translated into a modification in a certain trait of the individual.
- What are the consequences of a deviation in the number of chromosomes in a human karyotype?

❑ Chromosomal Abnormalities:

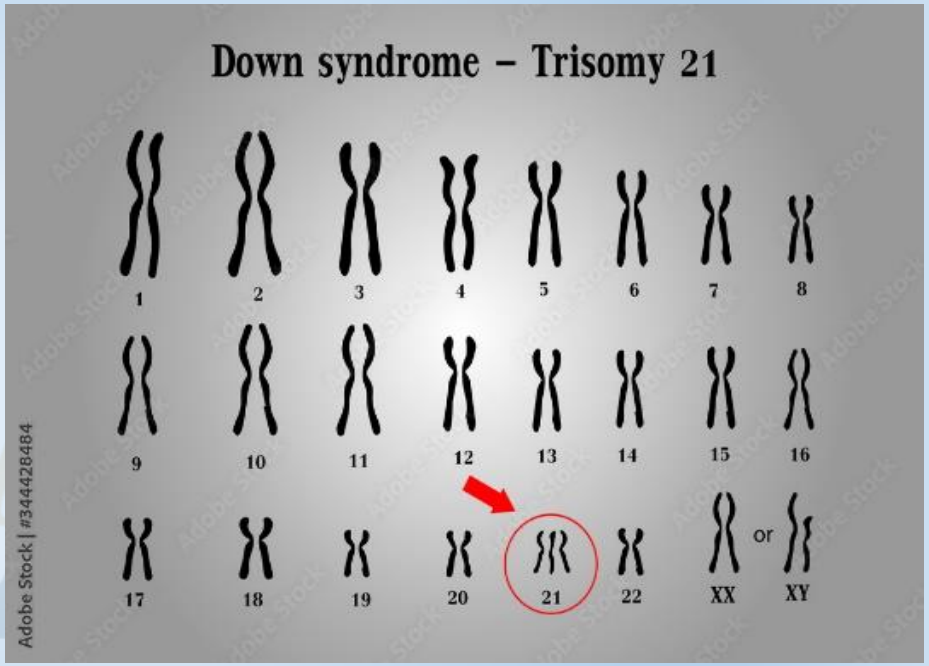
- For several reasons, errors may take place during zygote formation. The problem appears as a missing or extra chromosome.
- So any deficiency or excess in the number of chromosomes of an individual causes disease in that individual.
- It can be autosomal abnormality if the problem is in pair chromosomes 1 to 22 like trisomy 18 or monosomy 8.
- It may be gonosomal if it occurred in pair 23 like: XO, XXX, XXY, or XYY.
- Note that only gonosomal problems lead to a sterile individual.

❖ Autosomal abnormalities:

- Deficiency or excess in the number of autosomes.

❑ Trisomy 21 (Down syndrome):

- The individual suffering from trisomy 21 has an extra chromosome 21.
- In their Karyotype, there are 3 copies of chromosome 21 instead of 2 copies.
- Chromosomal formula: **47, XY, + 21 (or 47, XX, + 21)**.
- This individual shows slim eyes with a round face and inclined back with mental retardation.



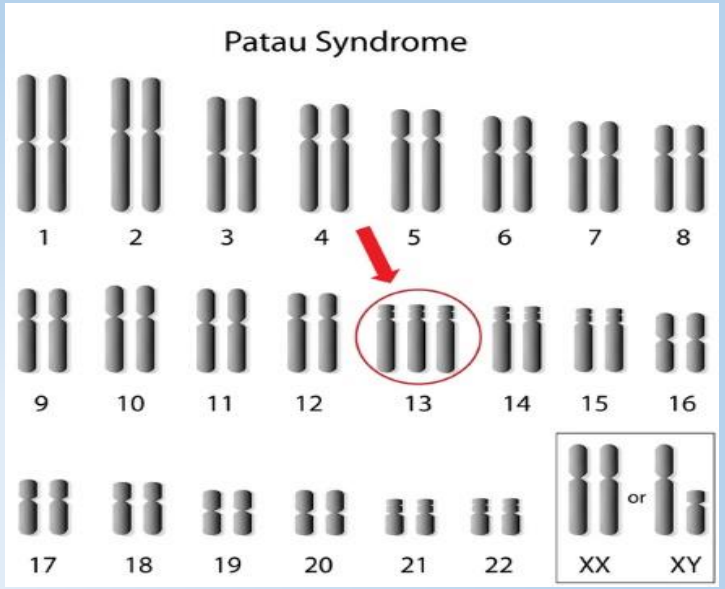
❖ Other abnormalities:

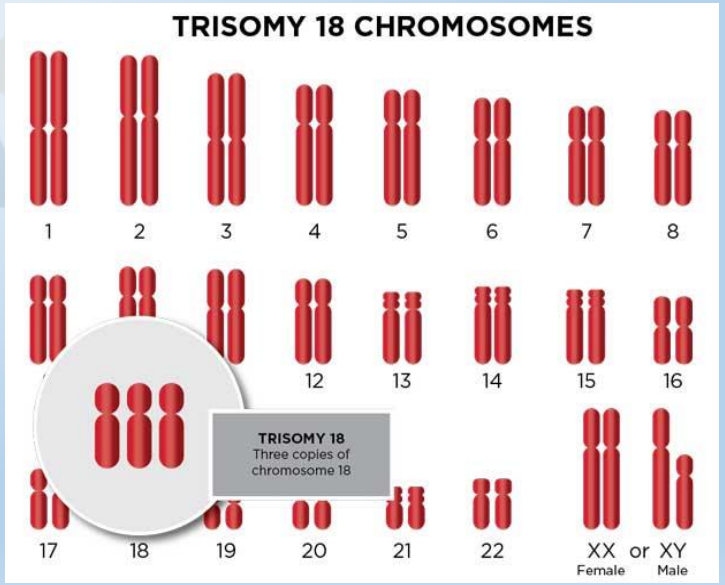
- **Trisomy 13:** 3 copies of chromosome 13 instead of 2 copies.

- **Trisomy 18:** 3 copies of chromosome 18 instead of 2 copies.

- **Monosomy 21:** 1 copy of chromosome 21 instead of 2 copies. **(deficiency)**.

- Chromosomal formula: 45, XY, - 21 (or 45, XX, - 21).



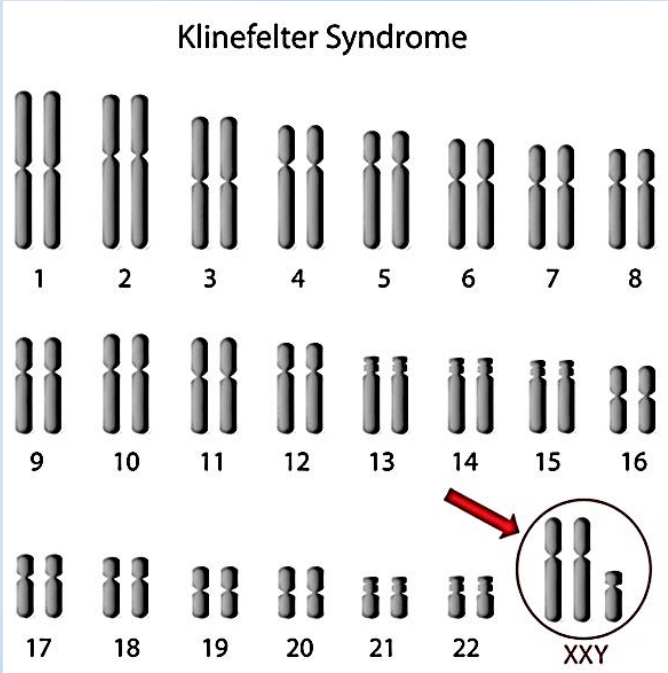


❖ Gonosomal abnormalities:

Excess or deficiency in the number of gonosomes (sex chromosomes).

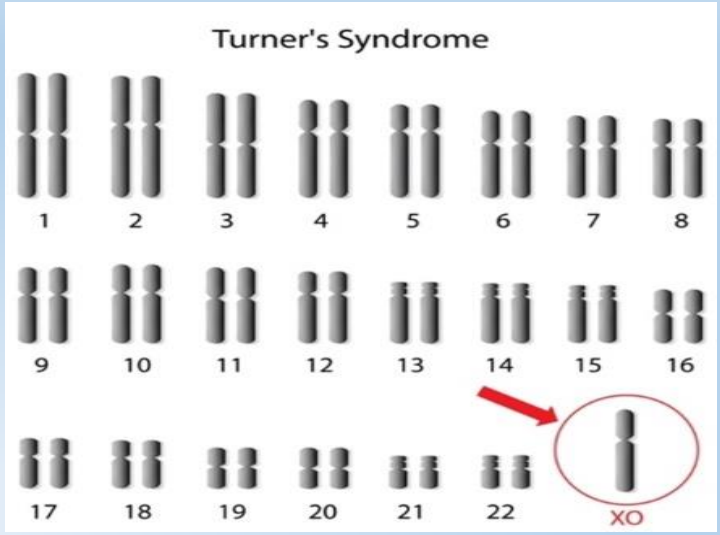
❑ Klinefelter syndrome (trisomy XXY):

- The individual suffering from this abnormality has one extra sex chromosome.
- His karyotype presents 3 sex chromosomes instead of 2.
- (2 X chromosomes and 1 Y chromosome).
- Chromosomal formula: **47, XXY (male)**.
- This individual shows development of breast and hips, besides a feminine voice and absence of facial hair.
- The person is usually sterile (infertile), cannot have children.



❖ Turner syndrome (Monosomy X):

- The individual suffering from this abnormality has one missing sex chromosome.
- She has one sex chromosome (X) instead of 2.
- Chromosomal formula: 45, X0 (female).
- This individual shows an absence of breast development with a weak, abnormal development of the uterus.
- The individual is usually sterile (infertile), cannot have children.



❑ Other abnormalities:

- **Trisomy XXX:** 47, XXX (female)
- **Monosomy Y:** 45, Y (male)

Abnormality	Down syndrome (Trisomy 21)	Klein filter syndrome (Trisomy XXY)	Turner syndrome (Monosomy X)
The cause	3 copies of chromosomes 21 instead of 2 copies	Present of 3 sex chromosomes instead of 2. (2 X chromosomes and 1 Y chromosome)	Present of one sex chromosome (X) instead of 2
Sex	Male / female	Male	Female
Number of chromosomes	47	47	45
Type of this abnormality	Autosomal abnormality	Gonosomal abnormality	Gonosomal abnormality
Chromosomal formula	47,XY,+21 or 47,XX,+21	47,XXY	45,XO
Sterile or fertile	Fertile	Sterile	Sterile
2 symptoms of this disease	• Slim eyes • inclined back	• Development of breasts • Feminine voice	• Absence of breasts. • Weak abnormal development of uterus