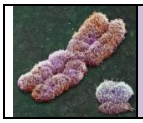


BIOLOGY LABORATORY:
Study of a human karyotype

4º E.S.O.

FOUNDATIONS

The exact number of chromosomes of the human species was unknown until a specific technical point, such as hypotonic shock, was pressed. This gave rise to the fact that in 1956, Tjio and Levan, working with somatic cells, definitively established that humans have up of 22 pairs of autosomes and the pair of sex chromosomes, that is:

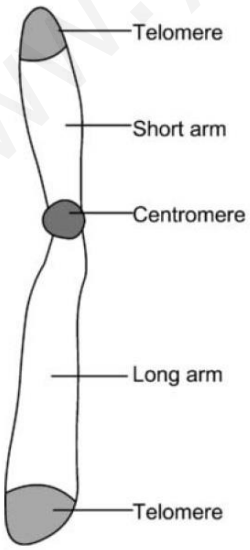
- Women: 2n = 44 + XX
- Men: 2n = 44 + XY

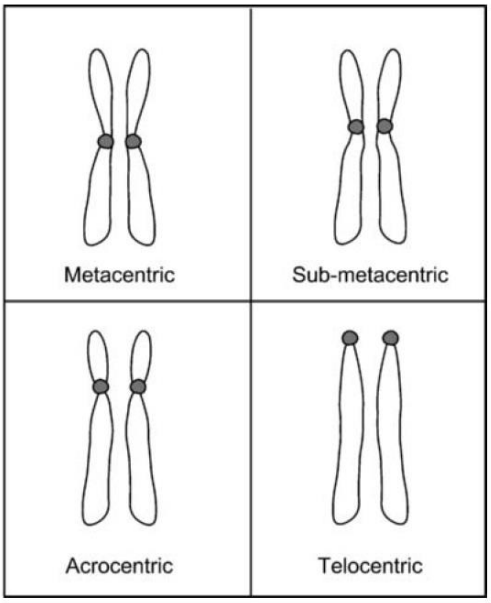
For the unification of criteria in the description of the karyotype, several meetings were held in: Denver (1960), London (1963), Chicago (1966), and Paris (1971), setting up a numerical classification based on the following points:

- The autosomes are numbered from 1 to 22, ordered by decreasing size, and within the same size, by the position of the centromere. Denver (1960).
- Chromosomes of similar length are grouped into groups designated by letters (A through G). Chicago (1966).
- The sex chromosomes X and Y are included in the corresponding groups (C and G respectively) by their size.

Therefore, the karyogram (idiogram) that is the graphic representation of the human karyotype is made up of 7 groups, the characteristics of which are:

- A: Includes chromosomes from 1 to 3. Large, almost metacentric. They are easily distinguished by their size and by the position of the centromere.
- B: Includes chromosomes from 4 and 5. Large and submetacentric. These 2 chromosomes are difficult to distinguish, although 4 is slightly larger.
- C: Includes chromosomes from 6 to 12. Chromosomes of medium size, submetacentric. The X looks like the longest in this group. Especially at 6, so it is difficult to distinguish. This group is the one that represents more identification difficulties.
- D: Includes chromosomes from 13 to 15. Medium size, subtelocentric. Chromosomes 13 has prominent satellites on the short arm. The 14th has them small. At 15 it is difficult to observe them.
- E: Includes chromosomes from 16 to 18. Metacentric small chromosomes (16) or submetacentric chromosomes (17 and 18).
- F: Includes the chromosomes of 19 and 20. Chromosomes smaller than those of group E, and metacentric.
- G: Includes the chromosomes of 21 and 22. Very small, and subtelocentric. Both have satellites. The Y is included in this group.





As in many of the cases it was difficult to differentiate the chromosomes within the same group, banding techniques were introduced that through a differential staining (tinción) along the chromosomes, allowing a better differentiation of them.
The most widely used staining method is Giemsa banding (G Bands), the chromosomes presenting a band pattern:

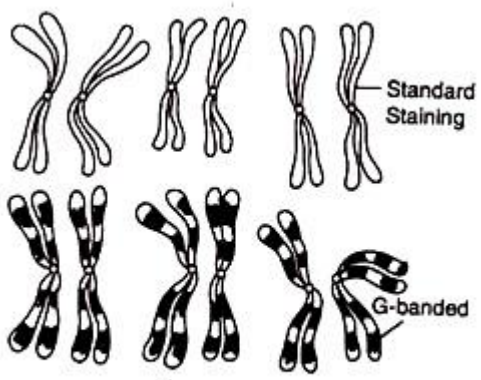


Fig. 36.1 : Chromosomes showing standard staining & G-bands.

ANOMALIES IN THE HUMAN CARIOTIPE.

Abnormalities can be numerical or structural and affect autosomes or sex chromosomes.
Jacobs et al. (1974), out of a total of 43,000 observations in newborns, give a total frequency of 0.56% (1 in 172) of detectable chromosomal abnormalities of any type without the need to apply banding techniques.

I.- ANOMALIES THAT APPEAR IN THE AUTOSOMES

- **Trisomy 21: Down syndrome**. Discovered by Lejeune et al (1959). It appears with a frequency of 1 in 700 live births, increasing the odds with the mother's age. Individuals have mental retardation. Women are fertile, giving 50% of Mongolian descent; instead the males are usually sterile. 4% of cases are due to the so-called translocation mongolism, in which the carriers, even when they have 2n = 46, present a translocation between a group D chromosome and a 21.
- **Trisomy D: Patau syndrome**. It appears in 1 in 10,000 live births. The half-life is about 130 days. They present multiple deformations and very deep mental retardation.
- **Trisomy E: Edwars syndrome**. 1 out of every 4000 live births appears. The half-life is about 8 months, with girls living longer than boys. The syndrome actually corresponds to chromosome 18.

II.- ANOMALIES THAT APPEAR IN SEXUAL CHROMOSOMES;

A) Males:

- **XXY Constitution**: Klinefelter Syndrome. Frequency of 1 in 400 live births. Eunucoids, low hairiness, gynecomastia. 25% with mental retardation. They are sterile, since they lack spermatogenesis.
- **Duplo Y syndrome:** Corresponds to men of high stature (from 1.80 m), somewhat lower than normal intelligence, sexual desinhibition. It has been linked to aggressive and antisocial behavior.

B) Women:

- **Constitution XO:** Turner Syndrome. Frequency of 1/5000, almost always sterile. Wide membranous neck, low ear implantation, cardiovascular deficiencies.

OBJECTIVES:

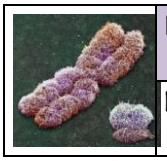
- Build a human karyotype.
- Characterize our chromosomes by their shape and size.
- Determine the genetic sex of an individual.
- Know some genetic alterations as well as their consequences.

MATERIAL

- Photocopies, glue and scissors
- If you don´t have a printer, don´t worry, you can use Paint programme or any photo editor tu cut and order the chromosomes.

ACTIVITIES:

- Observe the attached idiogram and describe in the correct order the steps of the technique used to represent a karyotype.
- Look at the metaphase chromosomes and cut (in a rectangle) each chromosome. Pair it with the same size and shape.
- Place them on a paper using the example of the attached idiogram, from largest to smallest, to obtain your karyotipe
- Analyze your results:
  - Indicate if they have alterations in their shape:
    - If they are all the same size.
    - If their arms are the same.
    - If everyone has a centromere.
    - If everyone has the same dark bands.
- Write the chromosome formula: number of autosomes and number of sexual chromosomes
- Indicate chromosomal sex: XX/XY…
- Indicate if it has any alteration and what are its consequences

	**BIOLOGY LABORATORY** - WORKSHEET Study of a human karyotype	**GROUP:**	**4º E.S.O.**
	NAME:	**DATE:**	**MARK:**

1. Write the correct order of how to build a karyotype

SOLUTION THE ORDER IS:									

ORDER THE CHROMOSOMES HERE:

A						B						
	1		2		3		4		5			
C												
	6		7		8		9		10		11	12
D						E						
	13		14		15		16		17		18	
F						G						
	19		20				21		22			

SEXUAL CHROMOSOMES			ALTERATION AND CONSEQUENCES IF NEEDED:
	X	Y	
CASE NUMBER°: SEX:			