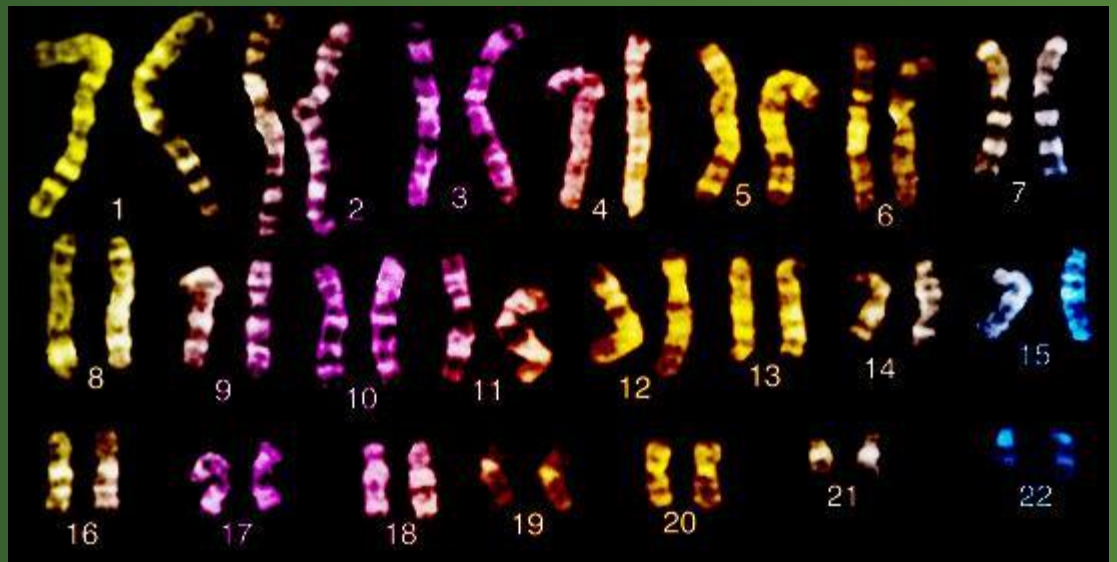
Chromosomes and Karyotypes

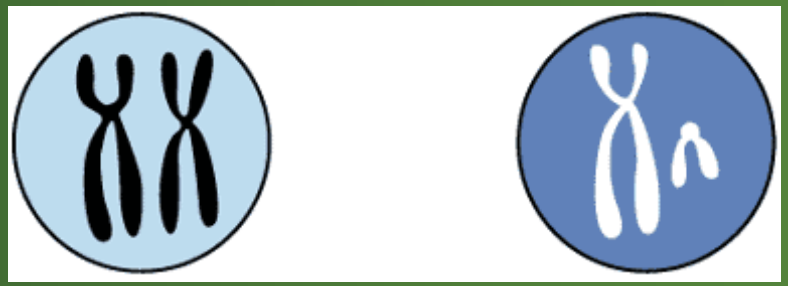
Two Types of Chromosomes:

1. Autosomes
   - ALL chromosomes except the sex chromosomes
   - 22 pairs (Chromosomes #1-22)



Two Types of Chromosomes:

2. Sex Chromosomes:
   - 1 pair (human chromosome #23)
   - Determine the sex of an organism
     - In mammals XX is female, XY is male



Female Male

THE ONLY GENETIC DIFFERENCE BETWEEN (HUMAN) MALES AND FEMALES IS THIS:

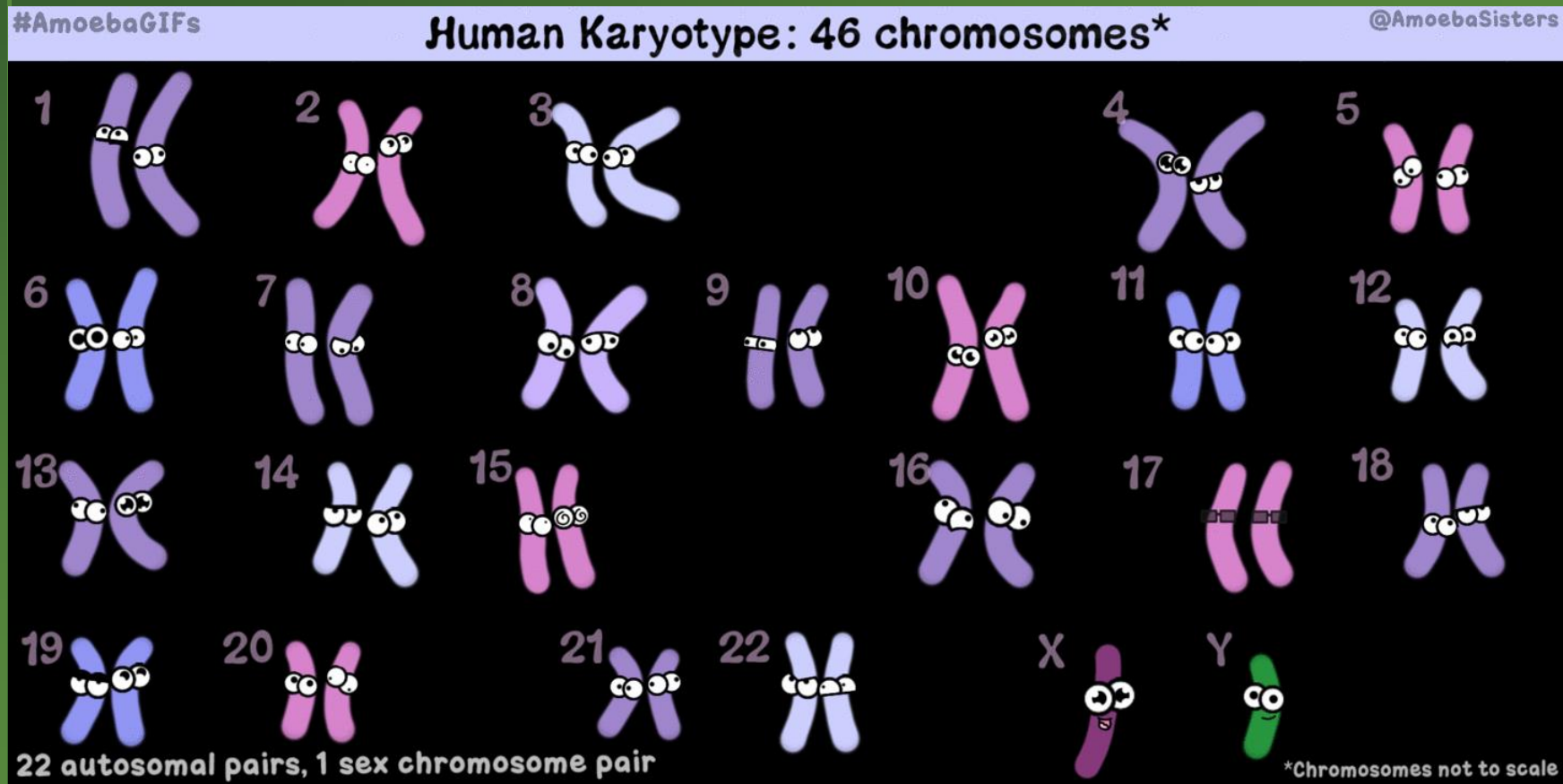
FEMALES HAVE TWO X CHROMOSOMES:

WHILE MALES HAVE ONE X AND ONE Y:

THE OTHER 22 OTHER PAIRS OF CHROMOSOMES ARE THE SAME.

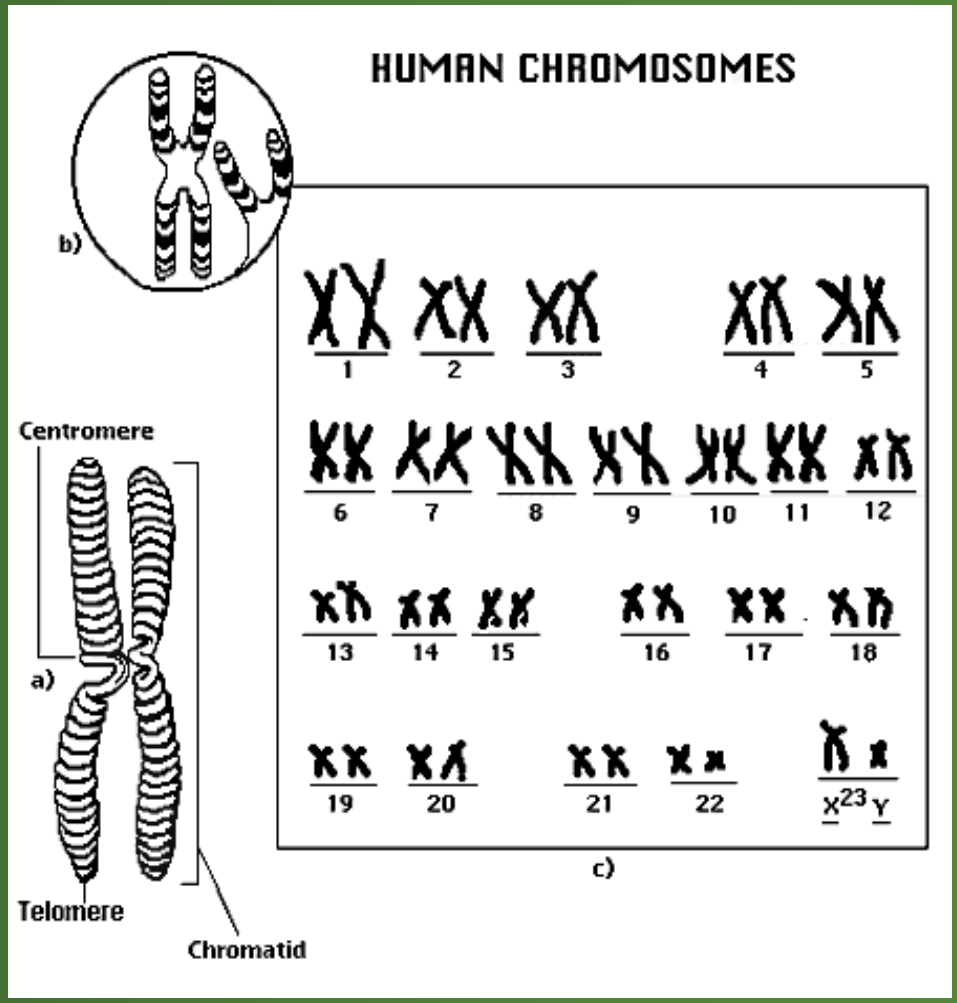
Karyotypes

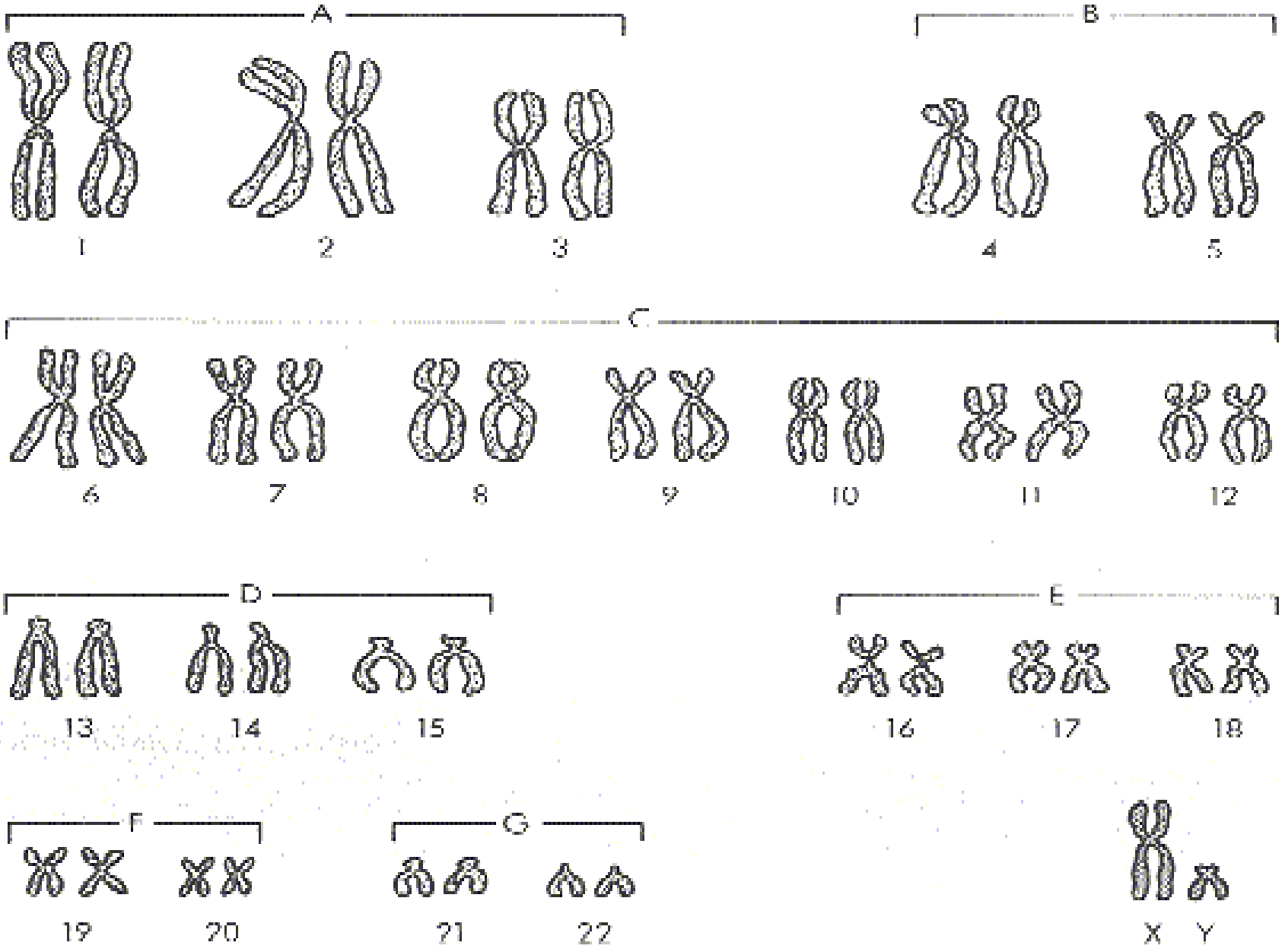
A picture of the chromosomes in which the chromosomes arranged in matching (homologous) pairs



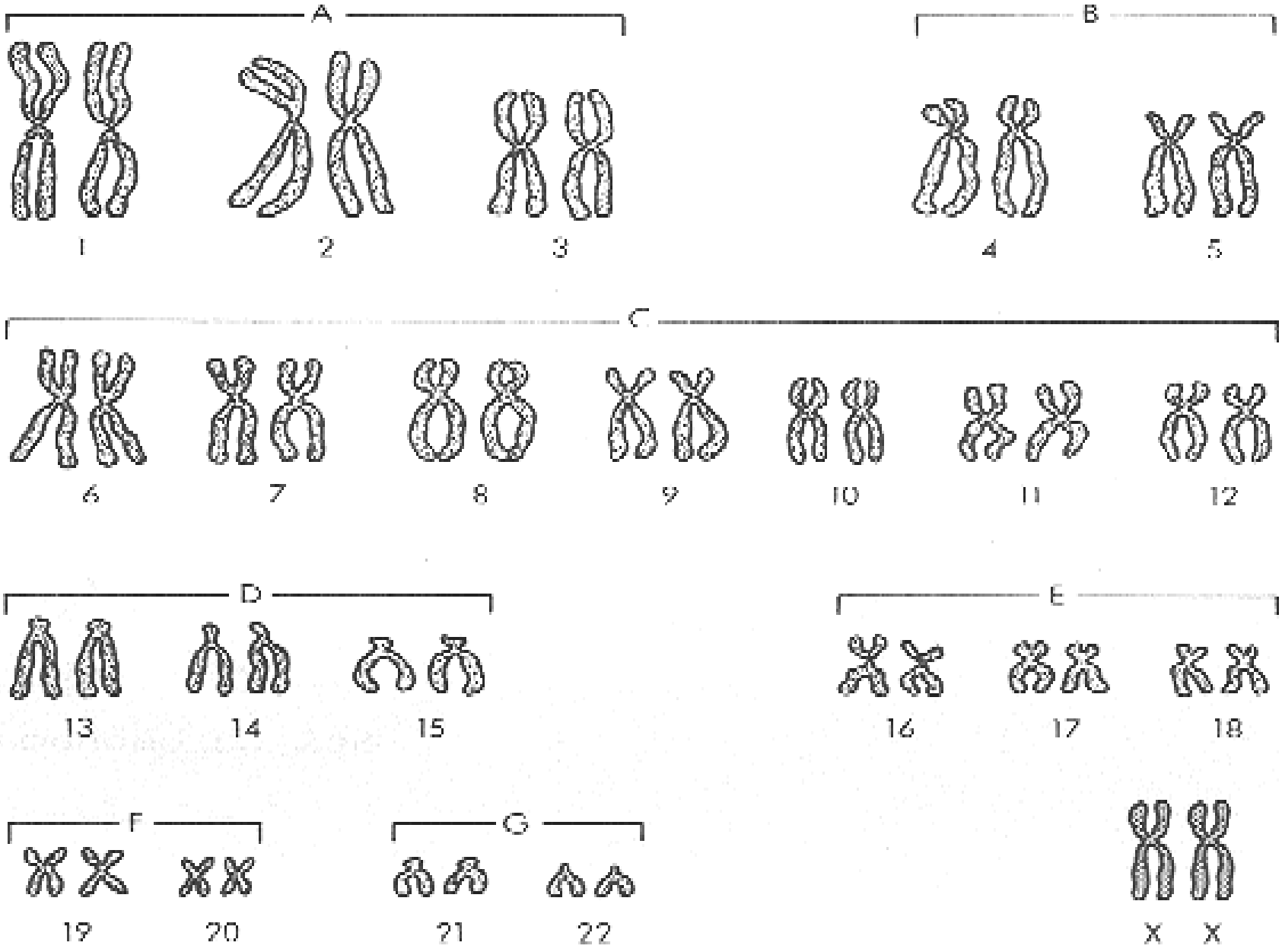
Karyotypes

- Arranged in size order from largest to smallest pair
- The sex chromosomes (X and Y) are usually the last pair, though they are not the smallest.





Normal Male



Normal Female

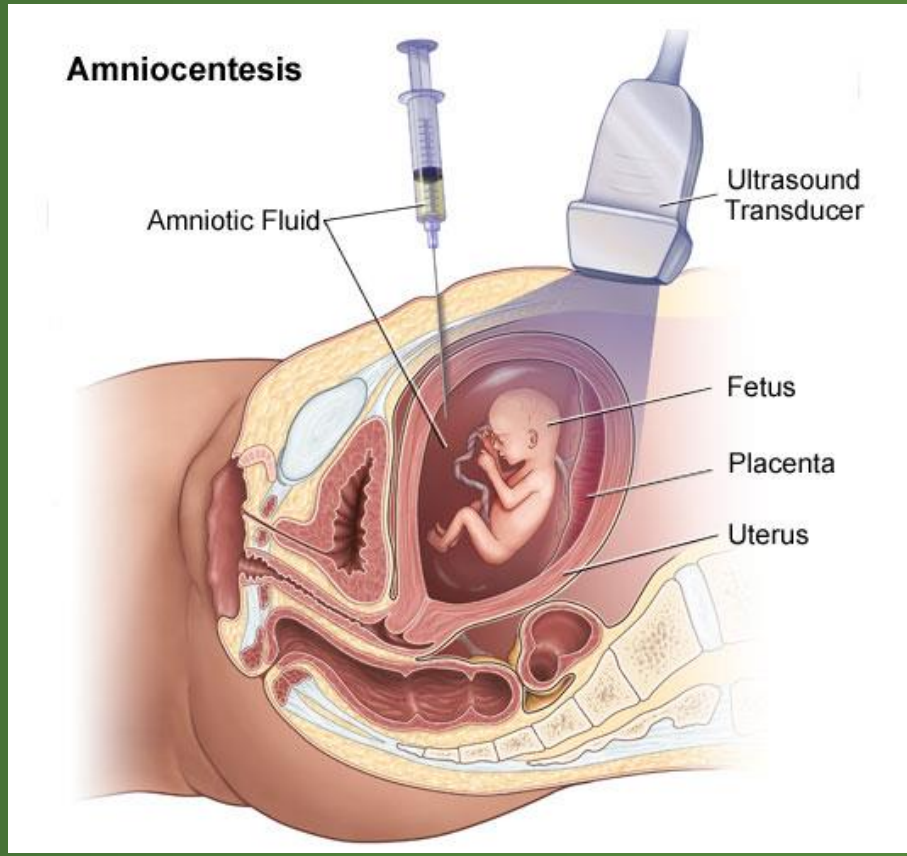
Karyotypes

- How are they used?
  - They are used for diagnosis of genetic abnormality based on the number of chromosomes.
  - They are used to determine the sex of an unborn child.

Karyotypes : How They Are Prepared

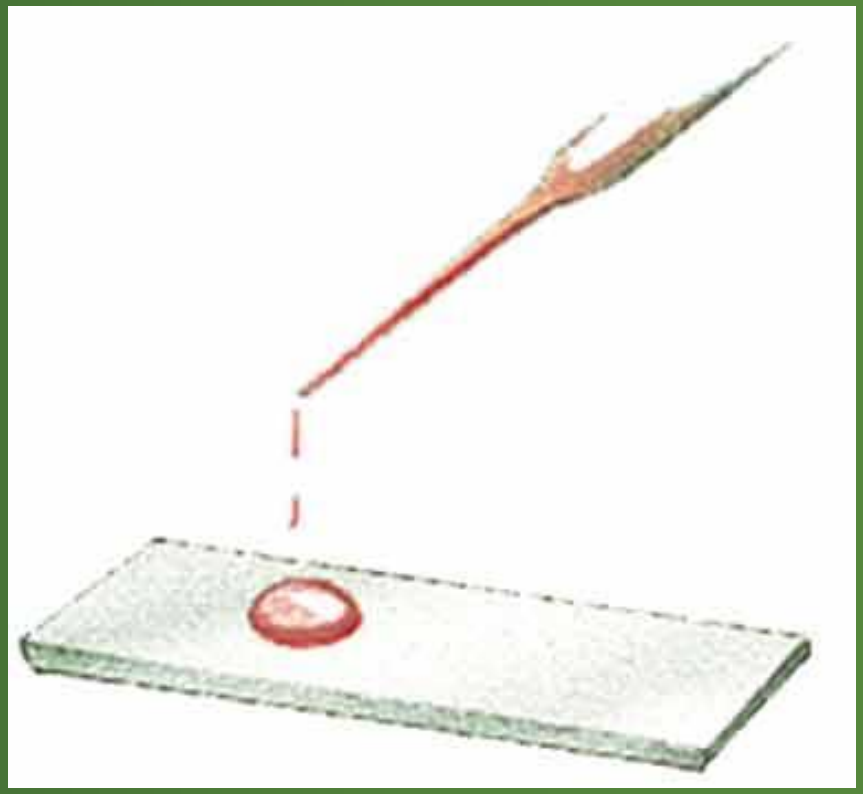
Cells are collected from a variety of sources:

- Amniotic fluid via a prenatal amniocentesis
- Blood Sample



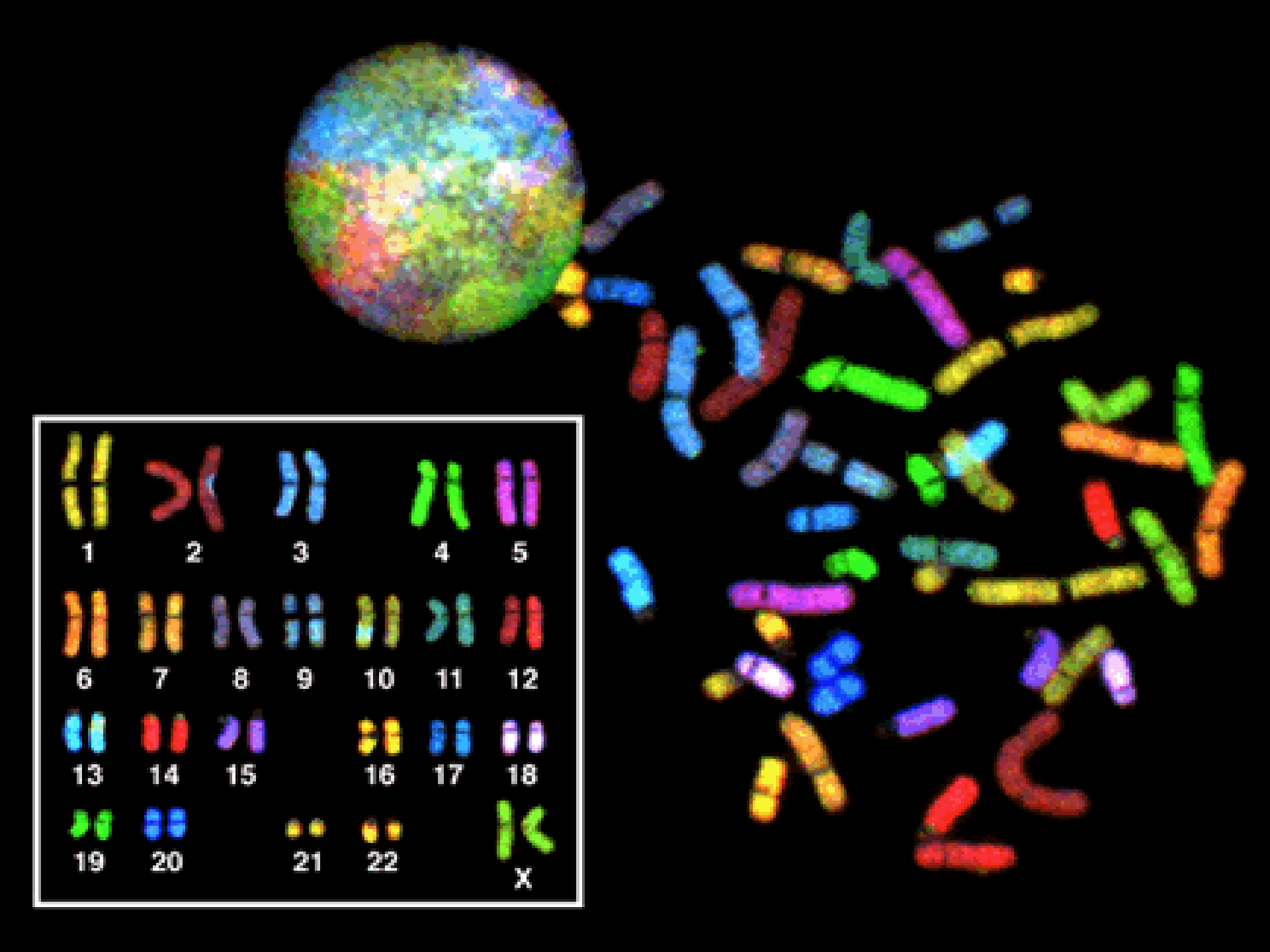
Karyotypes: How They Are Prepared

- Sample of cells are allowed to continue dividing
- Cells are stopped when in METAPHASE of MITOSIS.



Karyotypes: How They Are Prepared

- A photograph of the chromosomes is taken and enlarged.
- A trained technician matches the chromosomes into the homologous pairs based on three characteristics:
  - Size
  - Banding
  - Centromere position


1
2
3
4
5
6
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11
12
13
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15
16
17
18
19
20
21
22
X

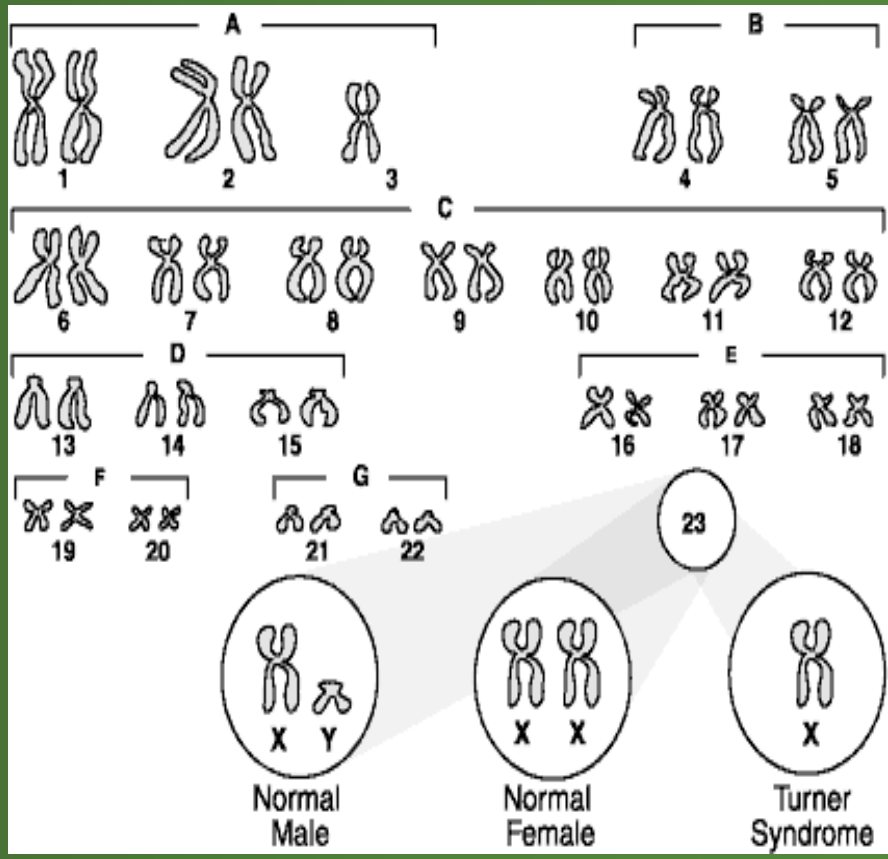
Chromosomal Disorders

- Normal:
  - Have 2 matching chromosomes for each of the 23 pairs
- Aneuploidy:
  - Having one more or one less of one of the chromosomes of the 23 pairs.

Chromosomal Disorders

- Monosomy: Missing one chromosome of one of the pairs

  Example: Turner syndrome; Monosomy 23
  - Missing one of the X chromosomes
  - Female who is X0 instead of XX

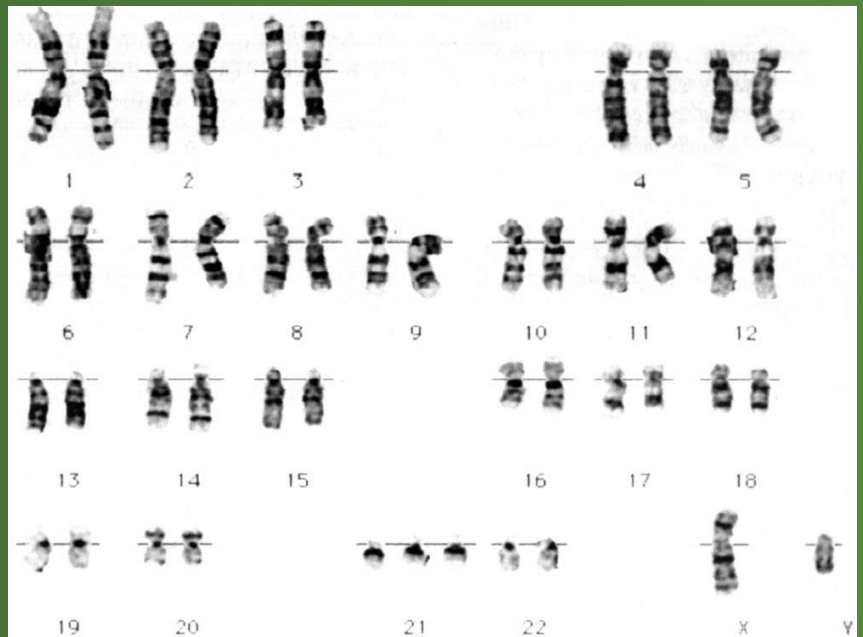


Turner Syndrome

- Symptoms:
  - Short stature
  - Webbed neck
  - Lack of secondary sex characteristics
  - A hollow appearance to the chest
  - Lack of menstruation
  - Low hairline
  - "Droopy" eyelids

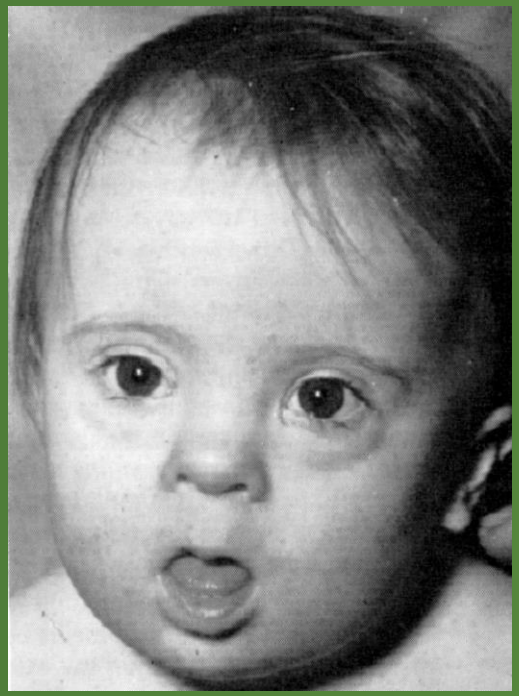
Chromosomal Disorders

- Trisomy: An extra chromosome of one of the pairs
  - Down syndrome; Trisomy 21
    - Extra chromosome #21 (so, there are 3 chromosome #21)



Down Syndrome

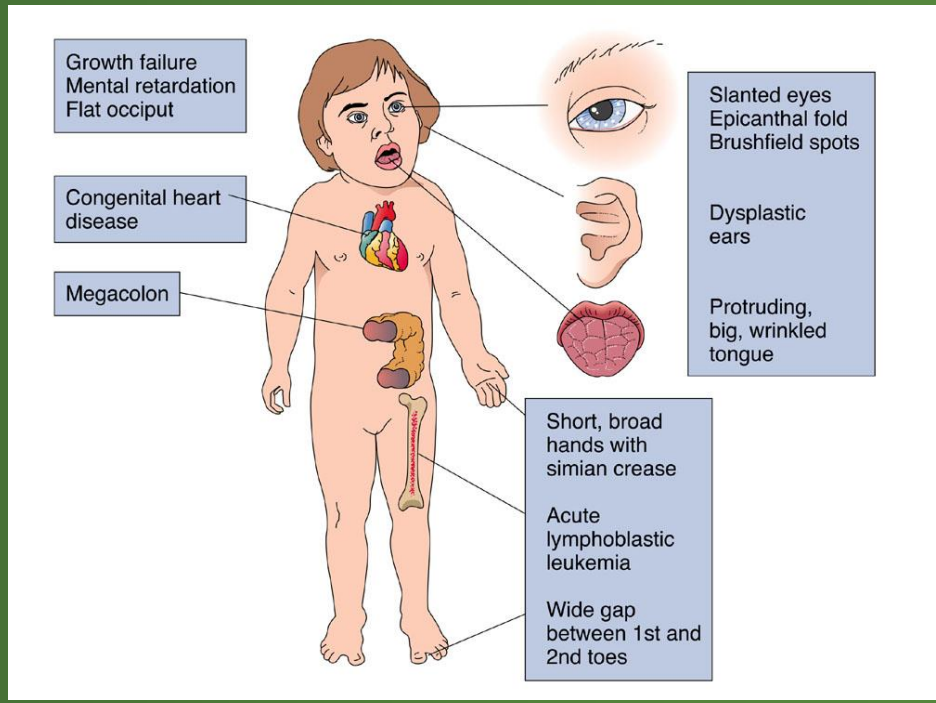
- Incidence
  - One of the most common chromosomal abnormalities
  - Frequency varies a lot according to the age of the mother.
    - The rate is only 1 in 2,000 for women 20 years old
    - In those 40 or older, it is 1 birth in 100.



Down Syndrome

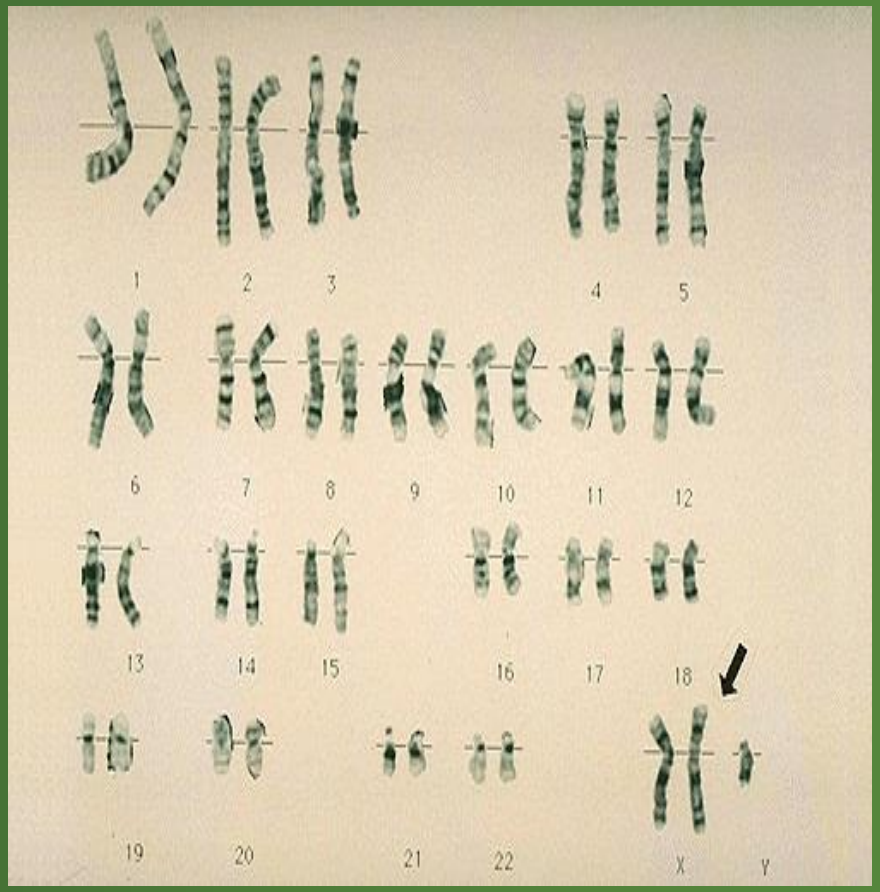
Symptoms:

- Small head, flattened in the back
- Broad, flat face
- Relatively small eyes, turned up at the outer corners
- Oversize tongue in a small mouth
- Single horizontal line across the palm, instead of the usual "head" and "heart" lines
- Short stature, with short limbs and stubby fingers



Genetic Disorders

- Klinefelter's Syndrome; Trisomy 23
  - Extra sex chromosome
  - Male who is XXY instead of XY
  - The most common sex chromosome abnormality in males



Klinefelter's Syndrome

- Symptoms:
  - Arm span exceeds height by more that an inch.
  - No or very little body hair and no facial hair.
  - High voice
  - Minimal muscle growth in arms/legs
  - Small testicles
  - Breast Tissue (not just fat, but actually firm breasts)
  - Low Testosterone Level