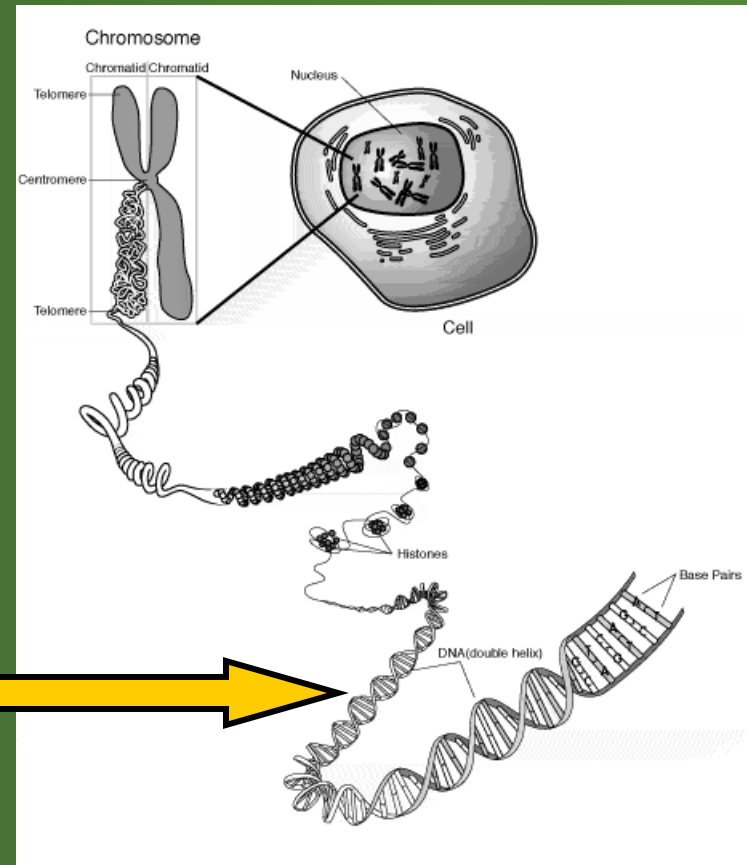
A microscopic view of numerous chromosomes, appearing as bright orange-red, X-shaped structures against a dark background. The chromosomes are scattered across the frame, with some showing distinct centromeres and arms. An orange rectangular box is overlaid on the right side of the image, containing the title text.

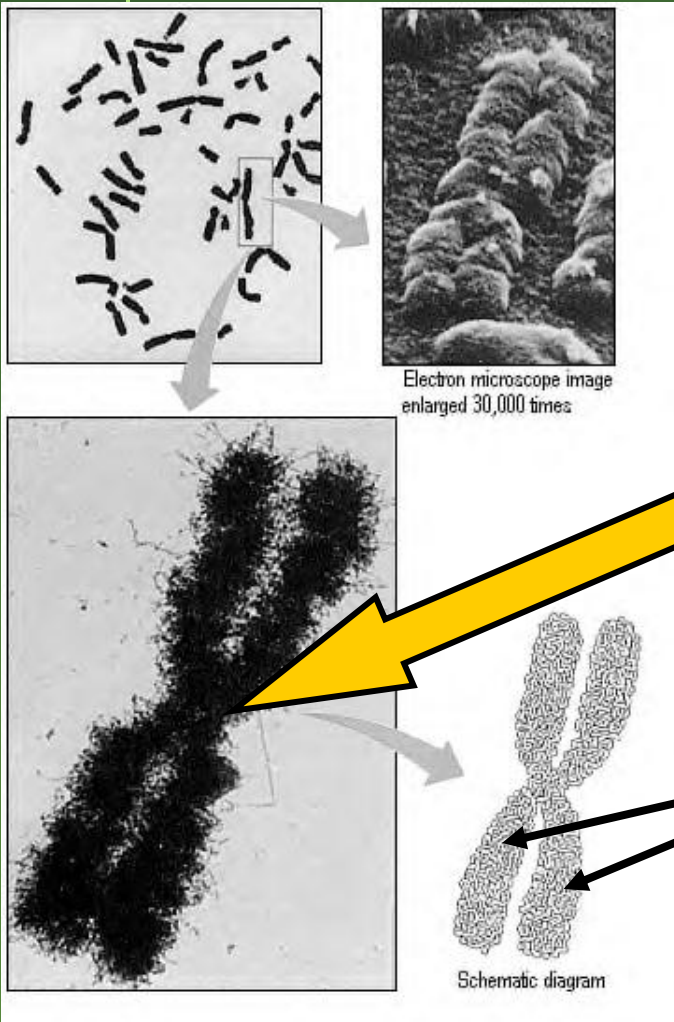
# Chromosomes and Karyotypes

# Review of Chromosomes

- Super **coiled** DNA
- Structure: It may be
  - A **single** coiled DNA molecule



# Chromosomes



- Or after **replication**, it may be **two** coiled DNA molecules held **together** at the center.
  - The area it is held together is called the "**centromere**."
  - **Chromatid**: Each DNA molecule in a **double** stranded chromosome (therefore, each **replicated** chromosome has **2** chromatids).

# Chromosomes

- **Humans** have **46** chromosomes
  - **23** chromosomes from **Mom**
  - **23** chromosomes from **Dad**

## Two Types of Chromosomes:

### 1. **Autosomes**

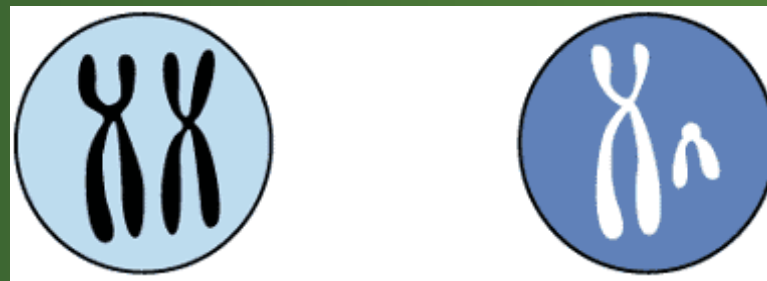
- **ALL** chromosomes **except** the **sex** chromosomes
- **22** pairs (Chromosomes #**1-22**)



# Two Types of Chromosomes:

## 2. Sex Chromosomes:

- 1 pair (human chromosome #23)
- Determine the **sex** of an organism
  - In **mammals** & **fruit flies** XX is **female**, XY is male



Female

Male

- In birds **ZZ** is male, **ZW** female
  - \*FIX IN NOTE TEMPLATE

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.

# Two Types of Cells

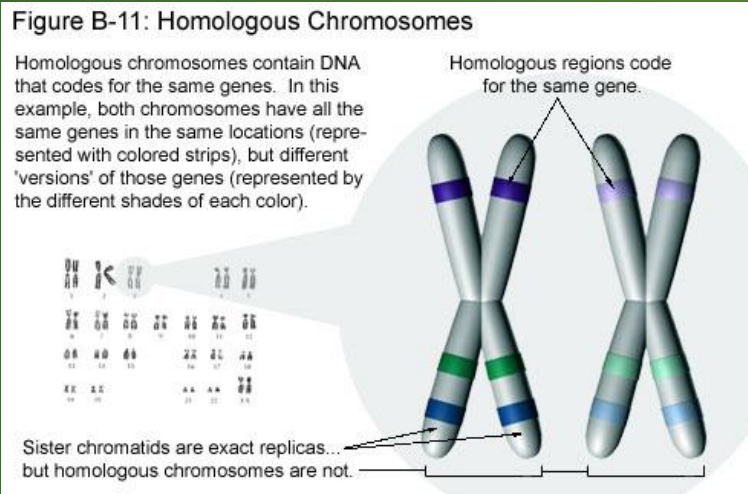
1. **Somatic Cells:**
  - All cells of the **body**
2. **Sex Cells:**
  - **Egg** and **Sperm**

# 1. Somatic Cells:

## Chromosomes are Homologues:

“Homo” means **same**.

- Homologous chromosomes are the same **size** and **shape**, and carry genes for the same **traits**.
- Called a “**homologous pair**”





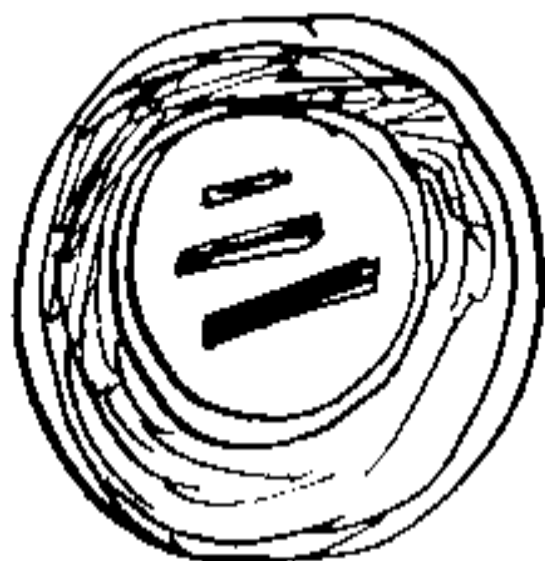
# Homologous Chromosomes

- Humans have **23** homologous pairs in all cells **except** sex cells
  - Cells with 23 homologous pairs are called "**DIPLOID**" or **2N**
    - **N** stands for number of **unique** chromosomes
    - Cells with 23 homologous pairs are called "**somatic cells**"
    - In humans, all cells **except** sperm and egg are somatic cells

## 2. Sex cells (sperm and egg)

- only have **one** of each chromosome
  - **No** homologous **pairs**
  - Called "HAPLOID" or N (think "half")
  - Sex Cells are called "GAMETES."

A cell with a single set of chromosomes is called **HAPLOID**; one with two sets is called **DIPLOID**. Our body cells are diploid, while our germ (sex) cells are haploid.



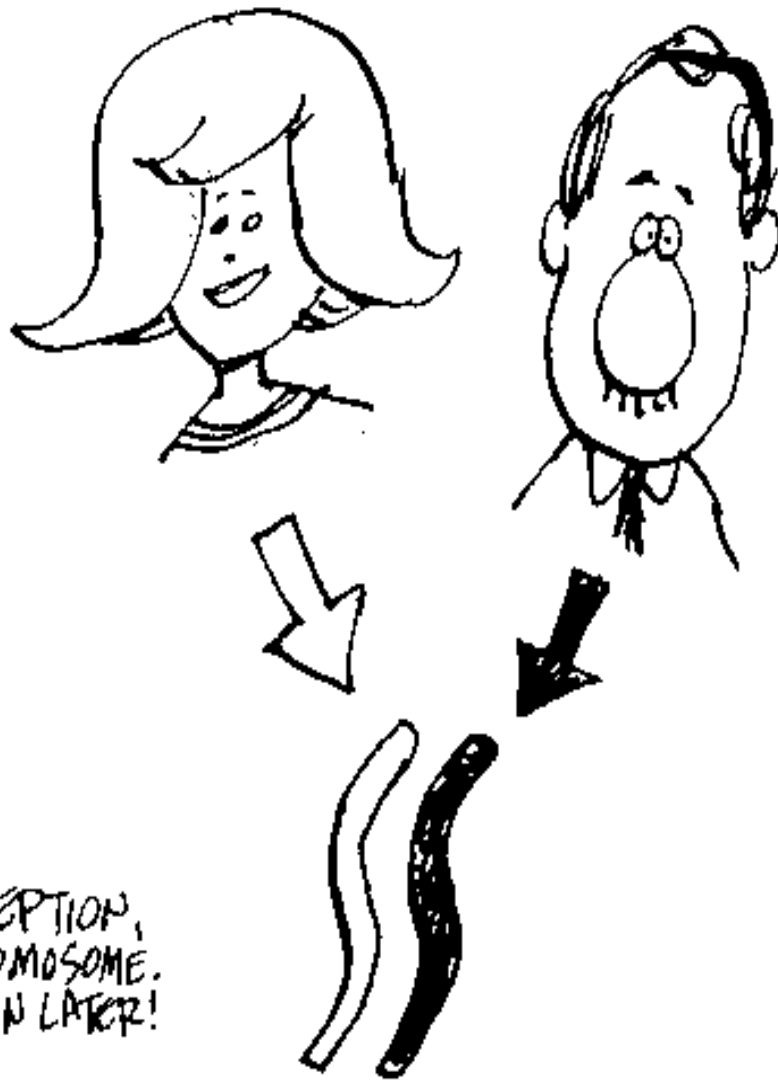
HAPLOID

DIPLOID



DIPLOID ORGANISMS INCLUDE ALL THE FAMILIAR MAMMALS AND BIRDS AND MANY PLANTS. HAPLOIDS INCLUDE MALE HONEY BEES, MANY FUNGI, AND ASEXUAL ONE-CELLED CREATURES.

HUMANS, FOR EXAMPLE, WITH 46 CHROMOSOMES, REALLY HAVE 23\* HOMOLOGOUS PAIRS: ONE FROM EACH PAIR COMES FROM MOM AND ONE FROM DAD.



\*WITH ONE EXCEPTION,  
THE SEX CHROMOSOME.  
WE'LL EXPLAIN LATER!

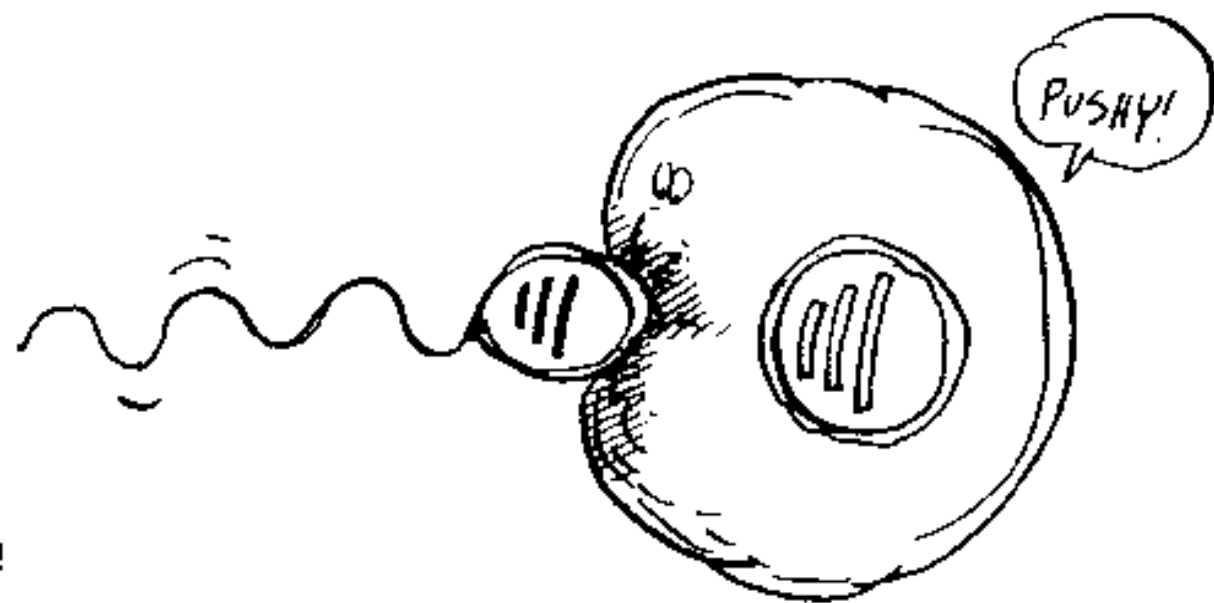
IT WAS THIS

**FACT**

SPERM AND EGG ARE  
SINGLE CELLS WITH  
ONLY HALF THE NORMAL  
NUMBER OF CHROMOSOMES.

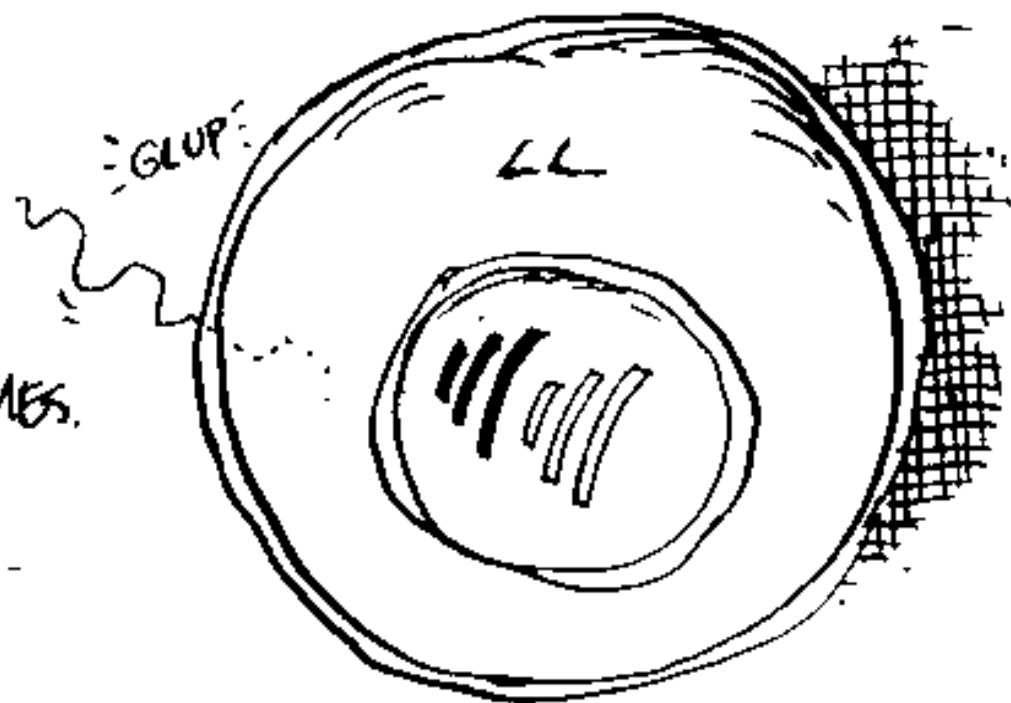
DAZZLING!

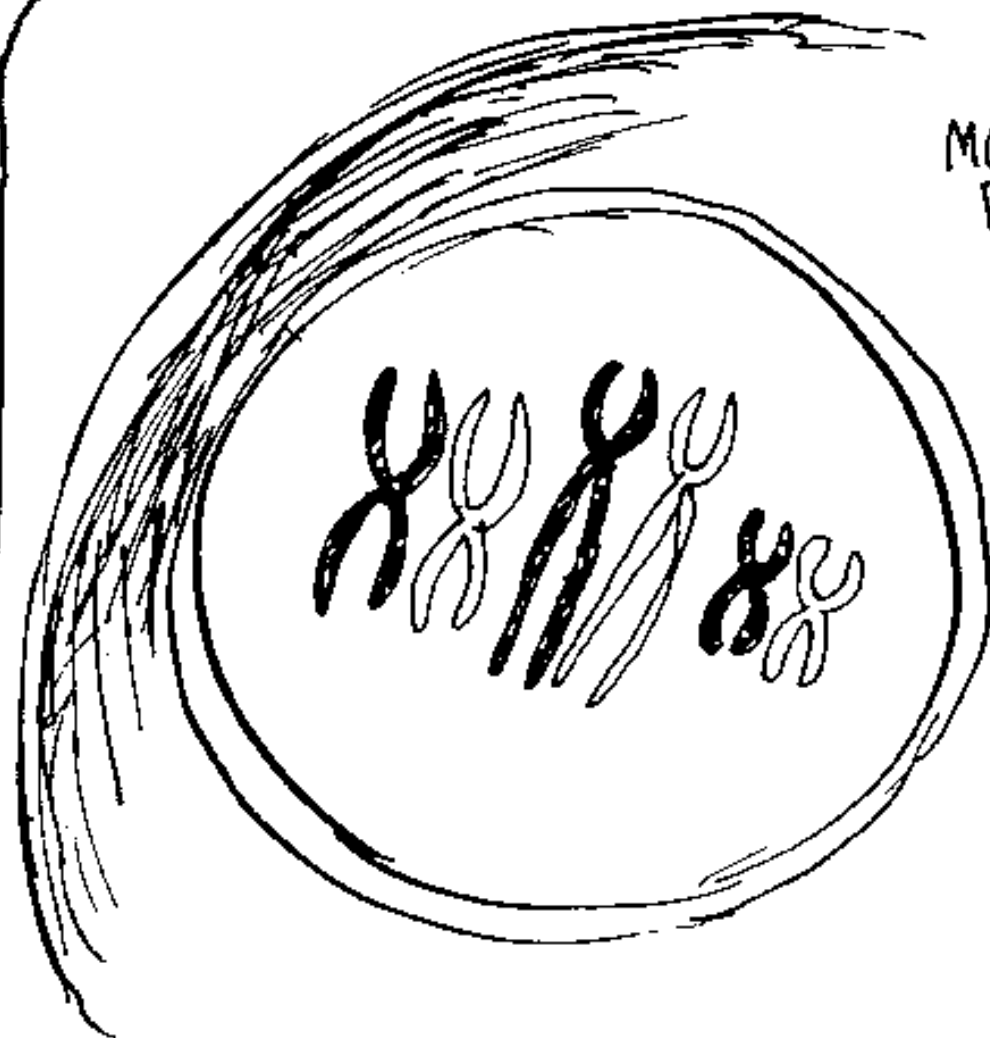




IT WORKS LIKE THIS:  
THE SPERM AND EGG—  
THE GERM CELLS, OR  
**GAMETES**, AS THEY  
ARE KNOWN—EACH  
CARRIES A HALF SET  
OF CHROMOSOMES.

AT FERTILIZATION, THEIR  
NUCLEI UNITE, GIVING  
THE FERTILIZED EGG,  
OR **ZYGOTE**, A FULL  
COMPLEMENT OF CHROMOSOMES.  
FROM THIS CELL ARISE  
ALL OTHERS BY MITOSIS.



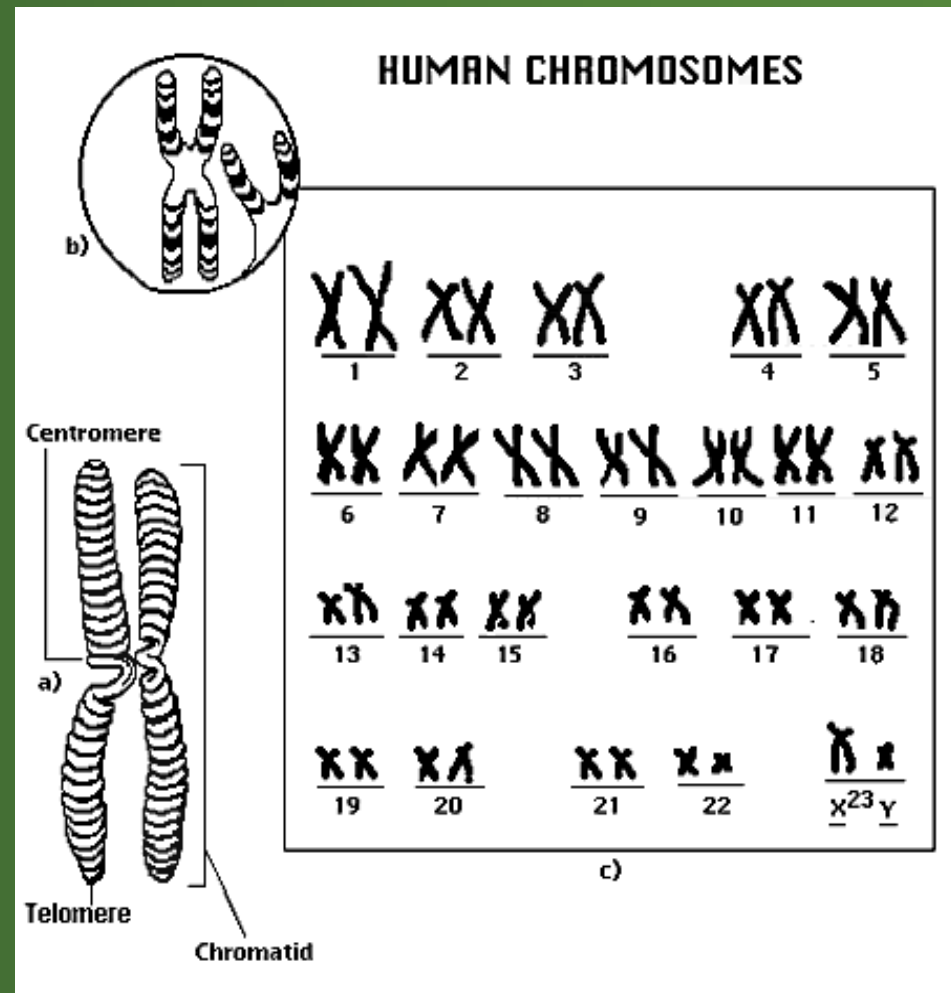


MOREOVER, IT WAS DISCOVERED (BY THE AMERICAN WILLIAM SUTTON IN 1902) THAT EACH CHROMOSOME FROM THE SPERM CAN BE MATCHED WITH A VIRTUALLY IDENTICAL ONE FROM THE EGG. (IT'S EASIER TO SEE WHEN THEY'RE DOUBLED AND CONTRACTED.)

THUS, THERE ARE REALLY ALREADY TWO COPIES OF EVERY CHROMOSOME IN THE CELL. THESE ARE CALLED "HOMOLOGOUS PAIRS"—"HOMOLOGOUS" MEANING "SAME SHAPE."

# Karyotypes

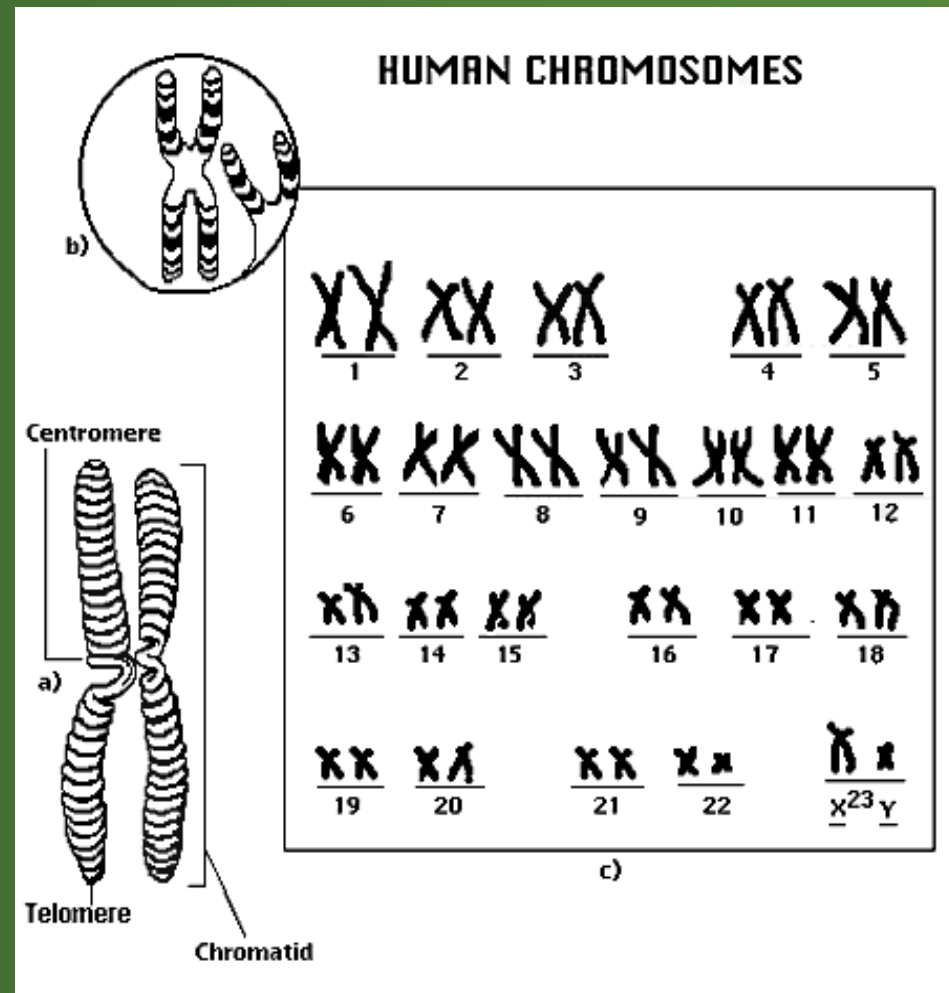
- A **picture** of the chromosomes in which the chromosomes arranged in **matching** (homologous) pairs

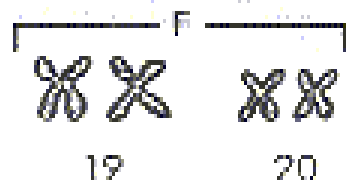
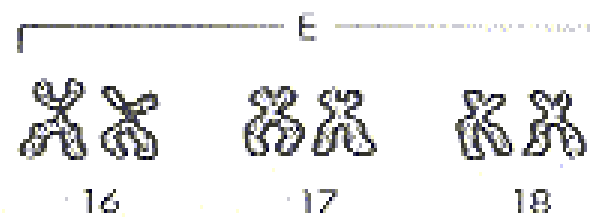
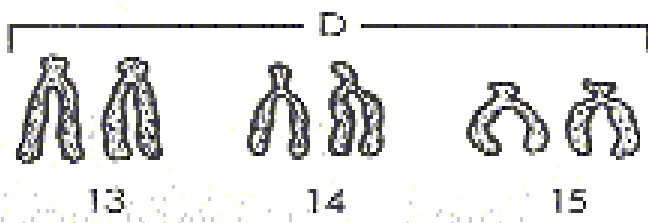
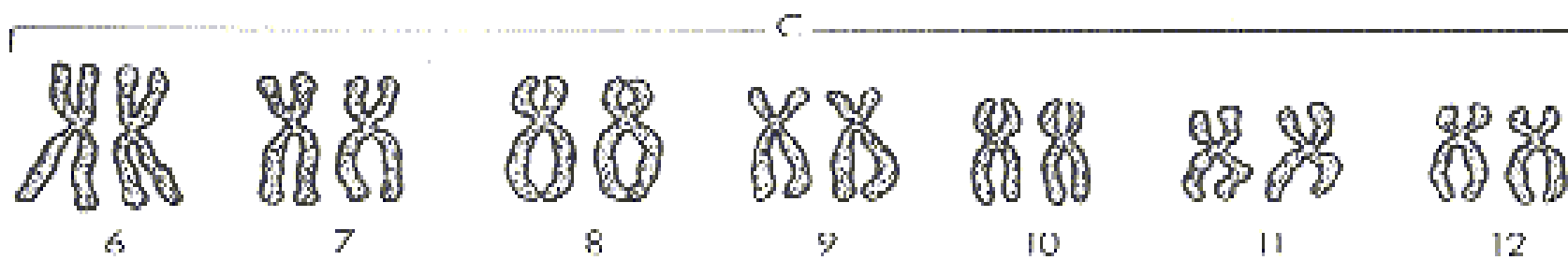
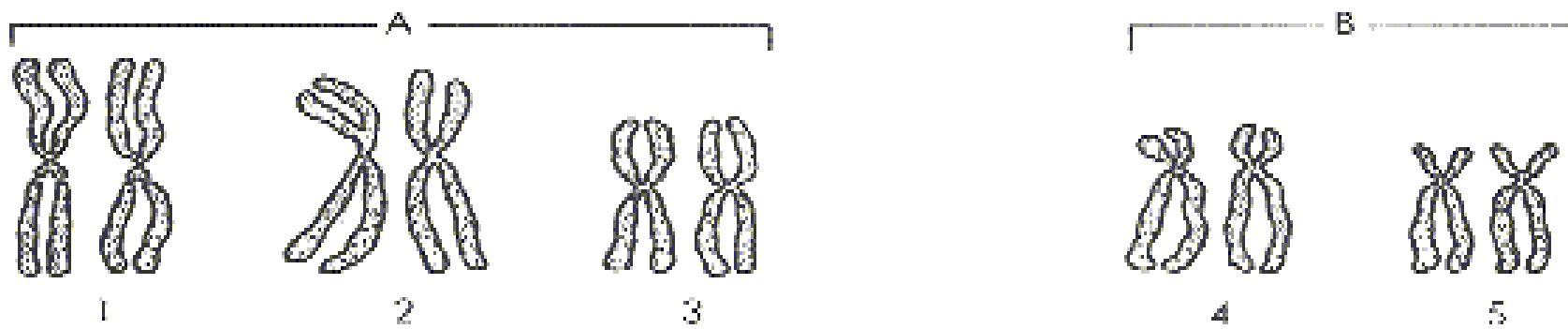




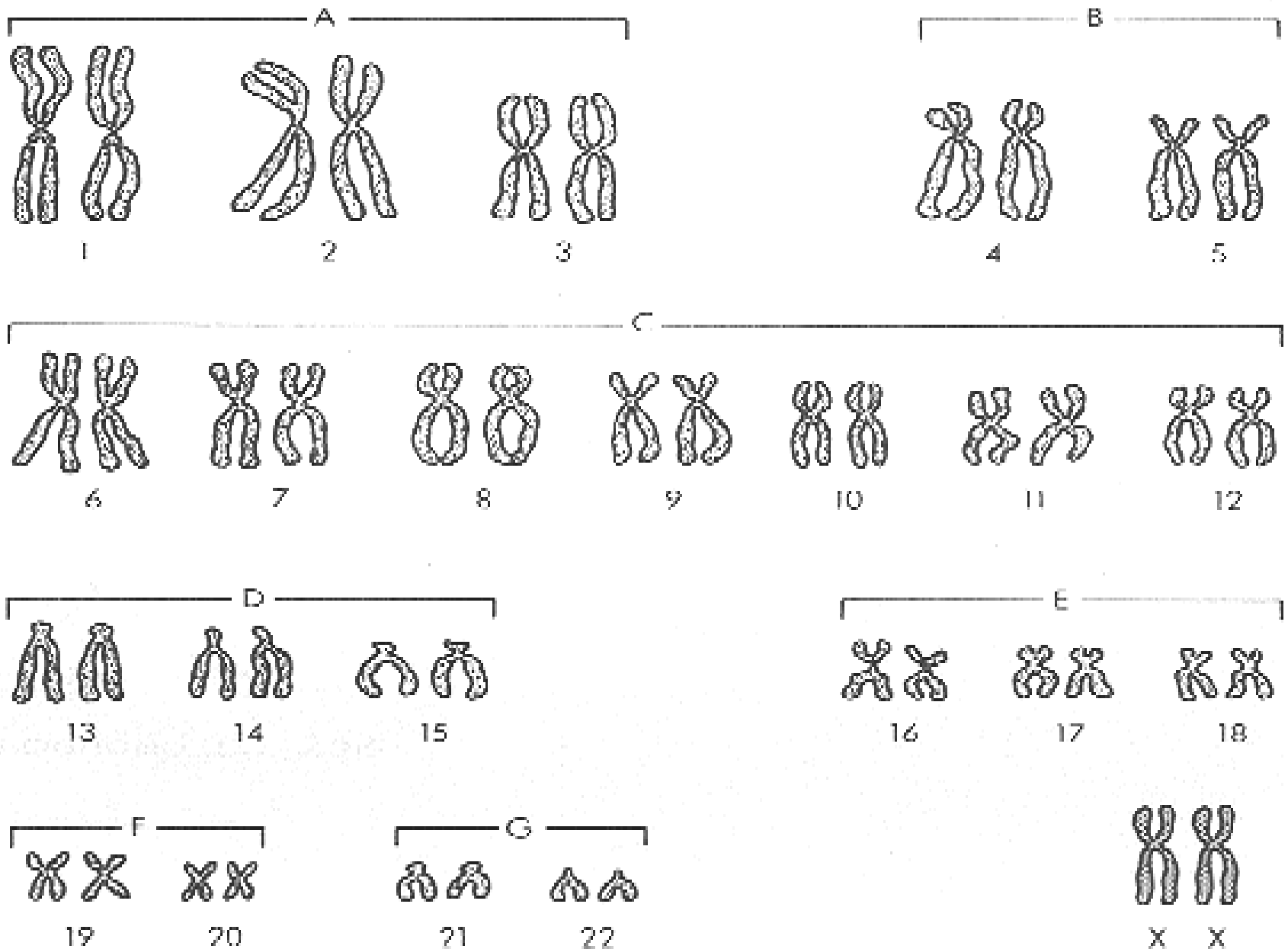
# Karyotypes

- Arranged in **size** order from **largest** pair 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

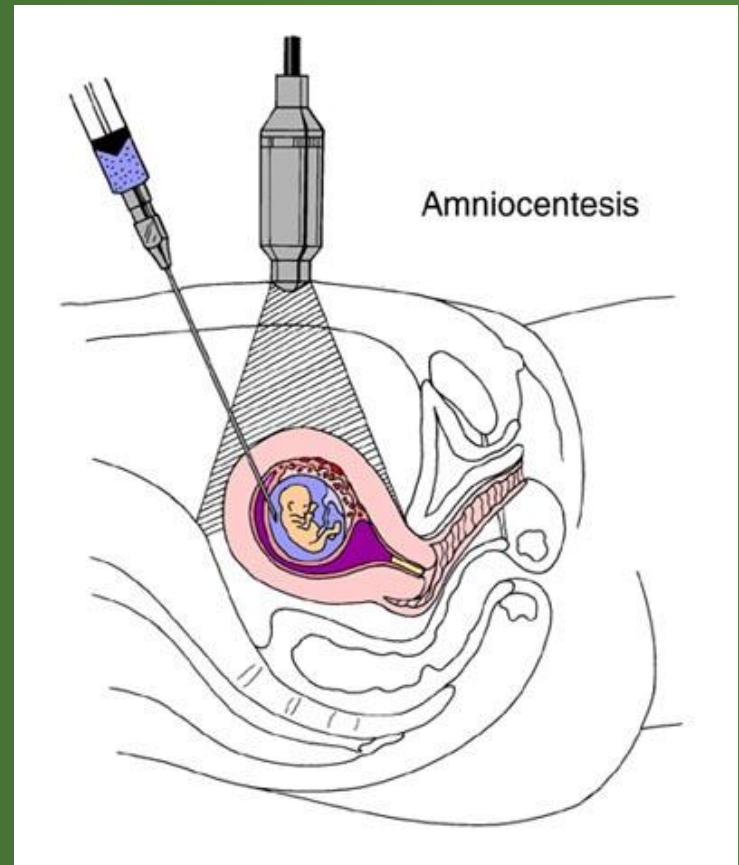
- How are they **used**?

- 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 are they prepared?
  - Cells are collected from a variety of sources:
    - Amniotic fluid via a pre-natal “amniocentesis”
    - Blood Sample



# Karyotypes

- How are they prepared?
  - Sample of cells are allowed to continue **dividing**
  - Cells are stopped when in **METAPHASE** of MITOSIS.



# Karyotypes

- 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



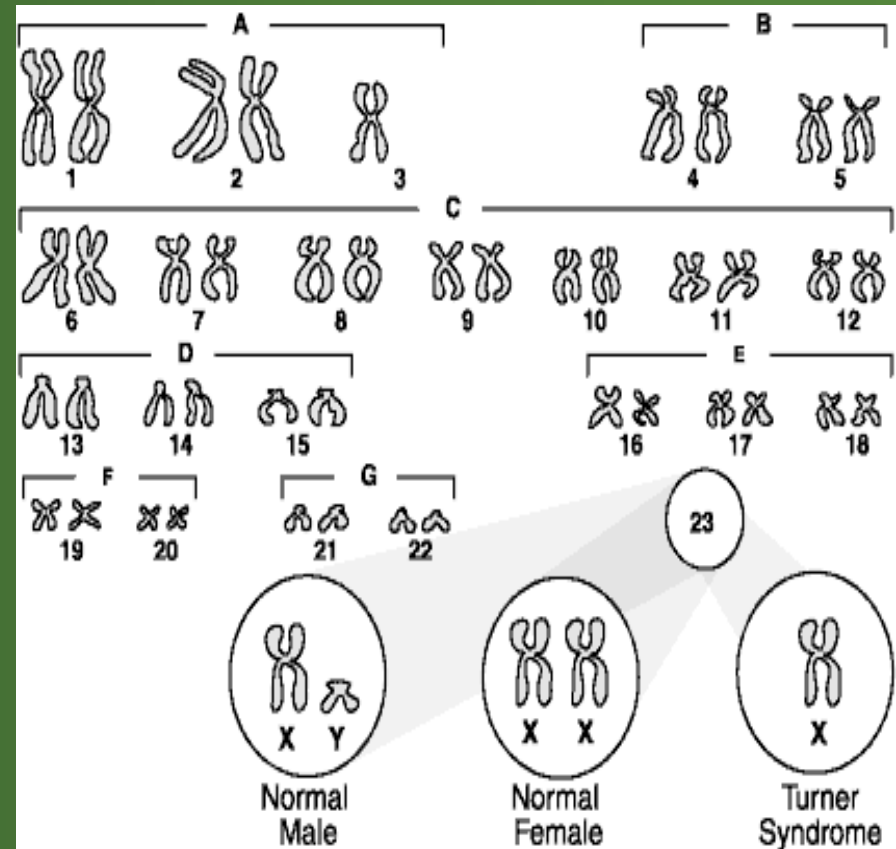
# Chromosomal Disorders

- 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
  - **Turner's syndrome;**  
**Monosomy 23**
    - **Missing** one of the **X** chromosomes
    - **Female** who is **X0** instead of **XX**



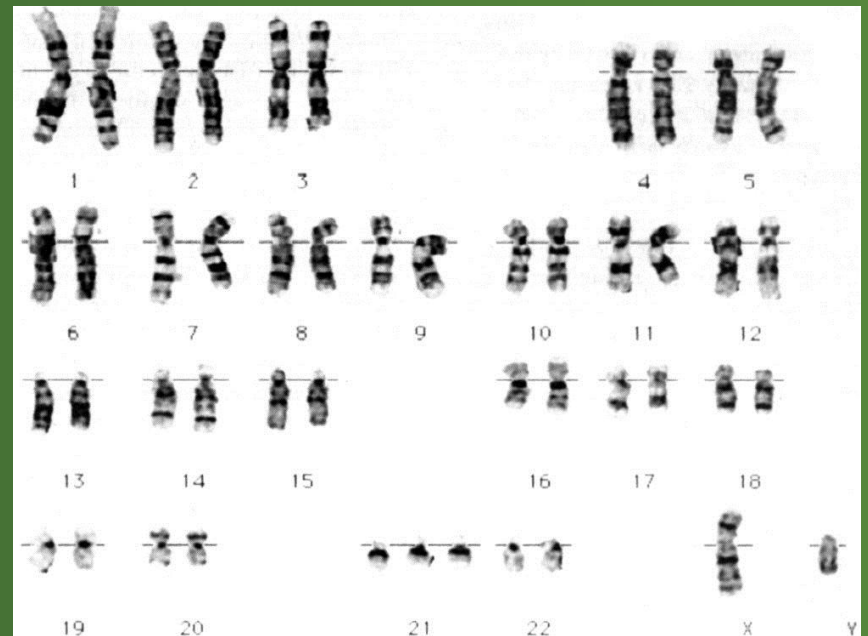
# Chromosomal Disorders

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



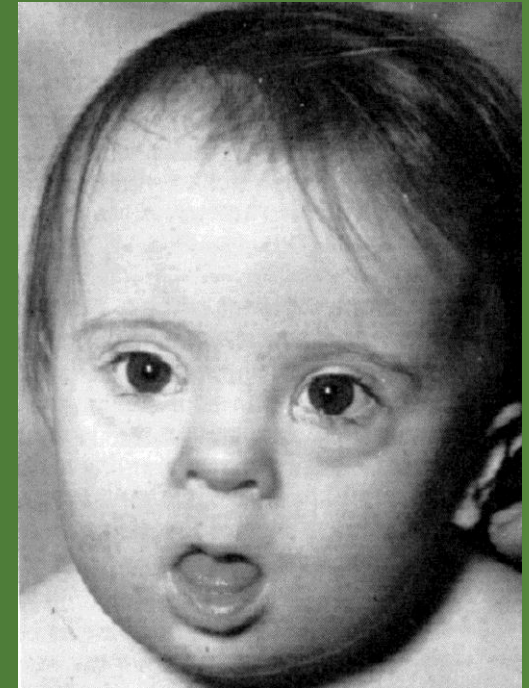
# Chromosomal Disorders

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



# Chromosomal Disorders

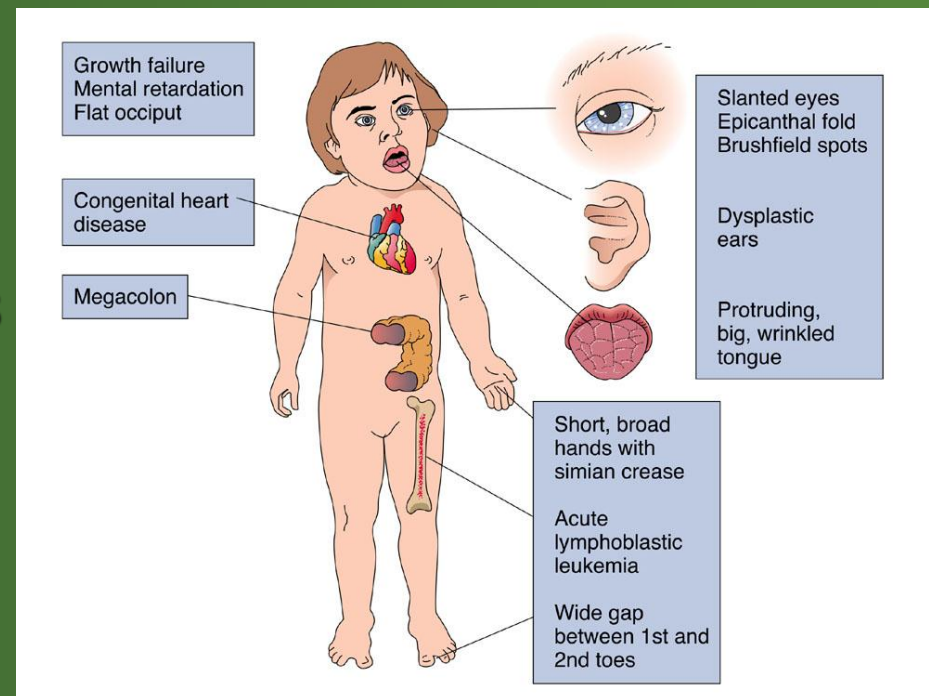
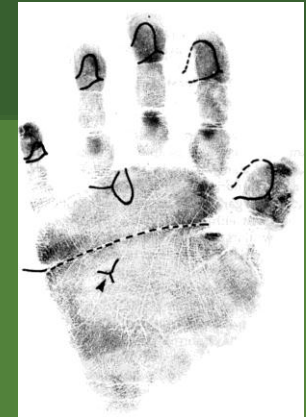
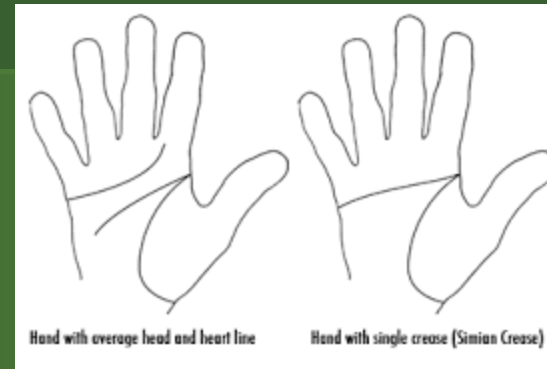
- Incidence
  - One of the most **common** chromosomal abnormalities
  - Frequency varies a lot according to the **age** of the mother.
    - The rate is only 1 in **2,000** for women **20** years old
    - In those **40** or older, it is 1 birth in **100**.



# Genetic Disorders

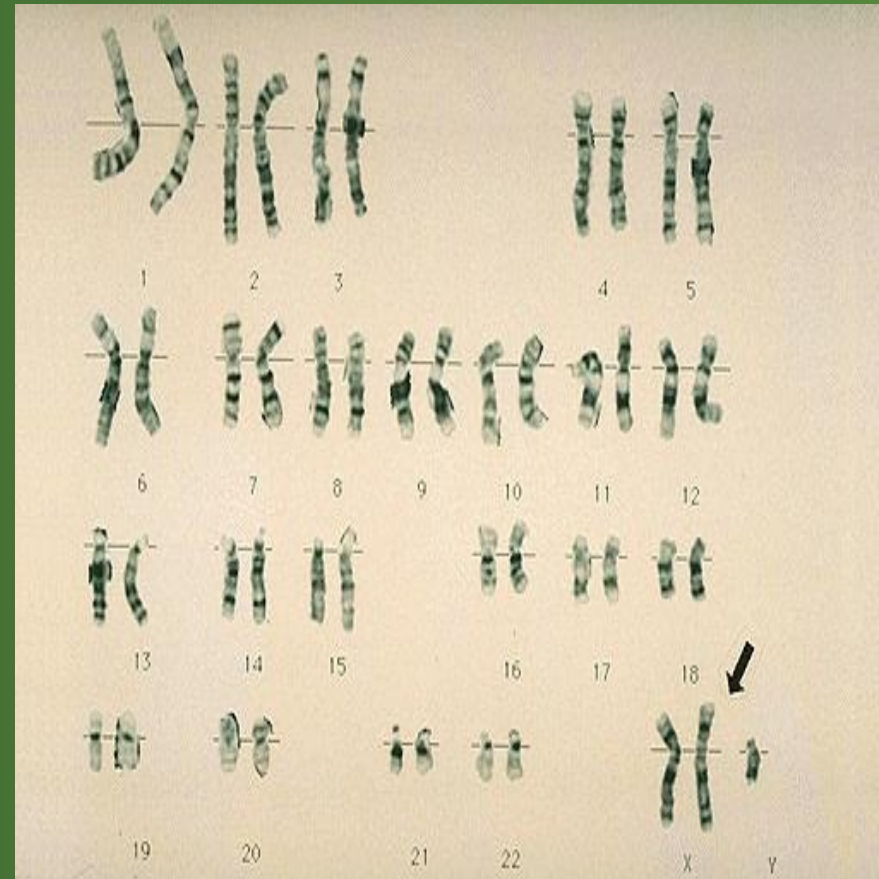
## Symptoms:

- 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**



# Genetic Disorders

- Symptoms:
  - Arm span exceeds height by more than 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