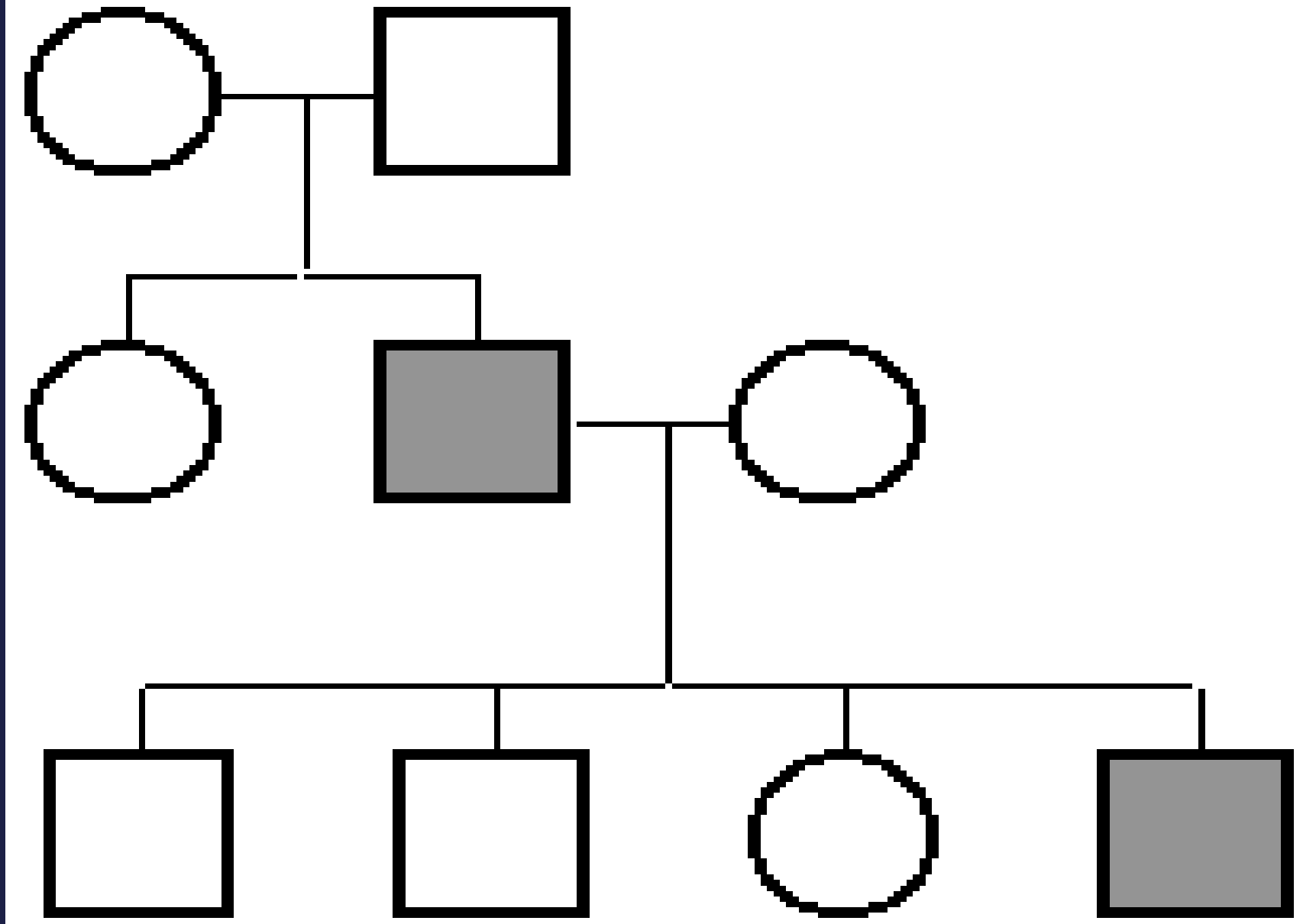
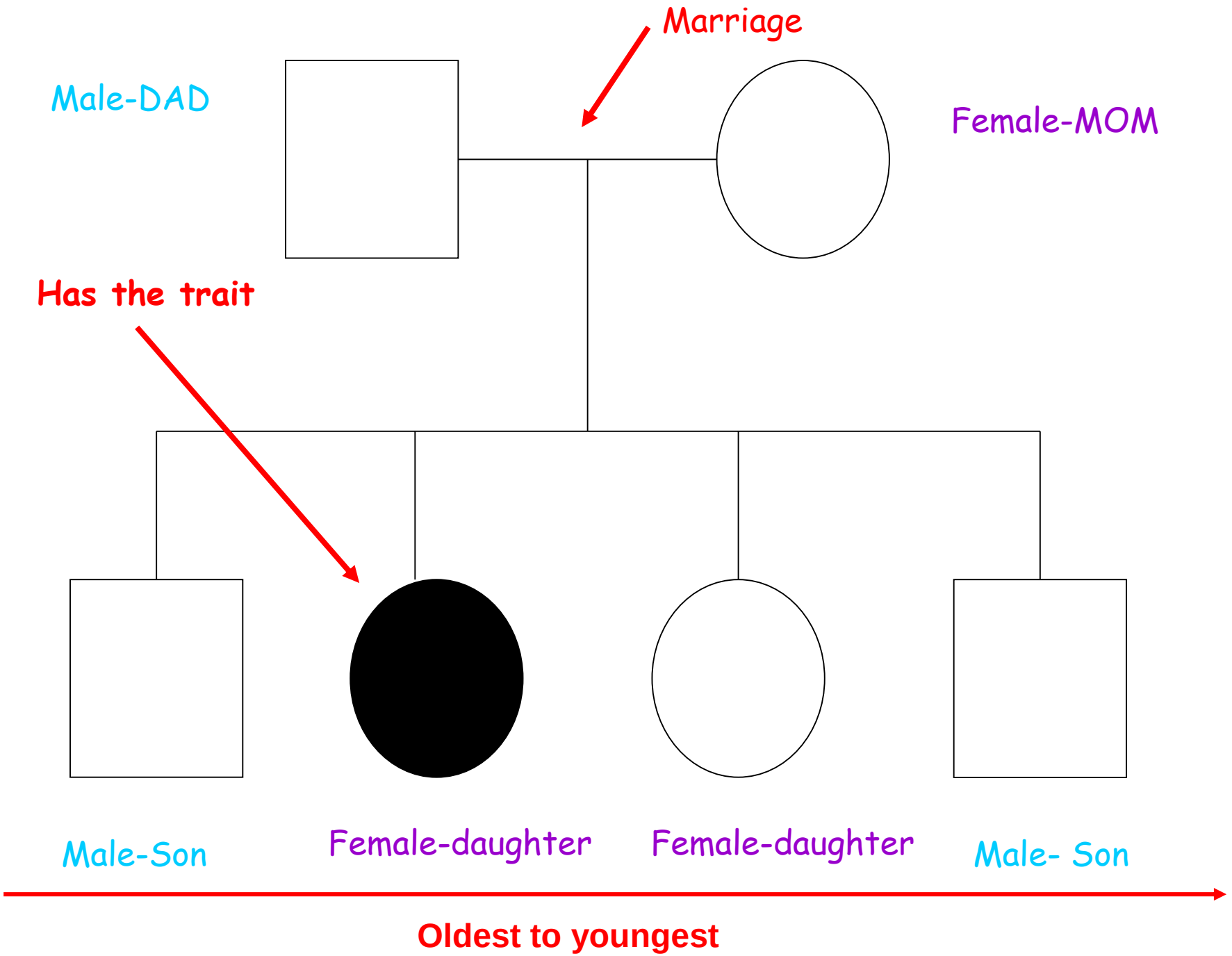
Pedigrees and Karyotypes



- On a pedigree:
  - A circle represents a female
  - A square represents a male
  - A horizontal line connecting a male and female represents a marriage
  - A vertical line and a bracket connect the parents to their children
  - A circle/square that is shaded means the person **HAS** the trait.
  - A circle/square that is not shaded means the person does not have the trait.
  - Children are placed from oldest to youngest.
  - A key is given to explain what the trait is.


Marriage
Male-DAD
Female-MOM
Has the trait
Male-Son
Female-daughter
Female-daughter
Male- Son
Oldest to youngest

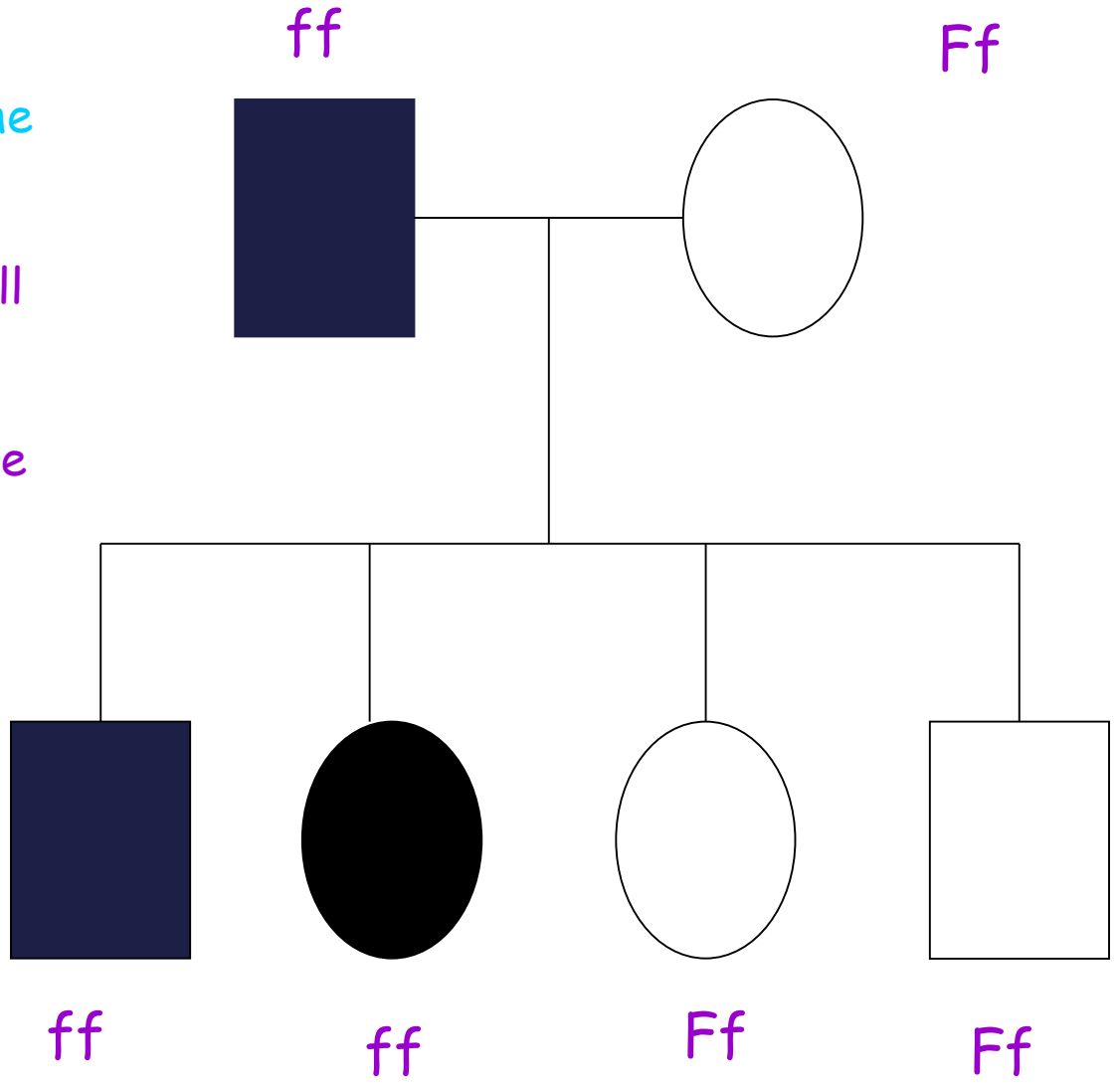
Steps:

- Identify all people who have the trait.

- For the purpose of this class all traits will be given to you. In other instances, you would have to determine whether or not the trait is autosomal dominant, autosomal recessive, or sex-linked.

- In this example, all those who have the trait are homozygous recessive.

- Can you correctly identify all genotypes of this family?
  - F- Normal
  - f- cystic fibrosis



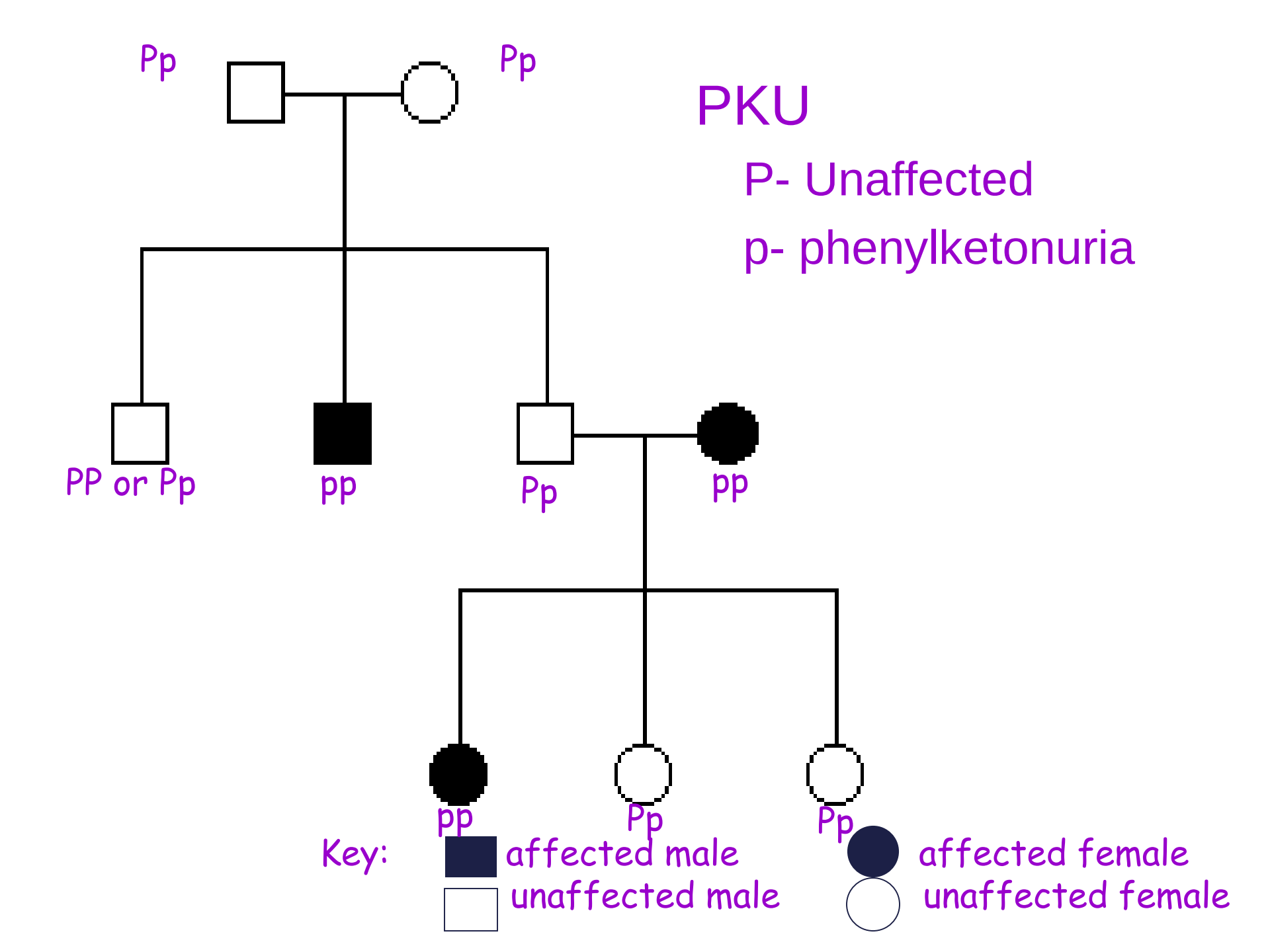
Key:

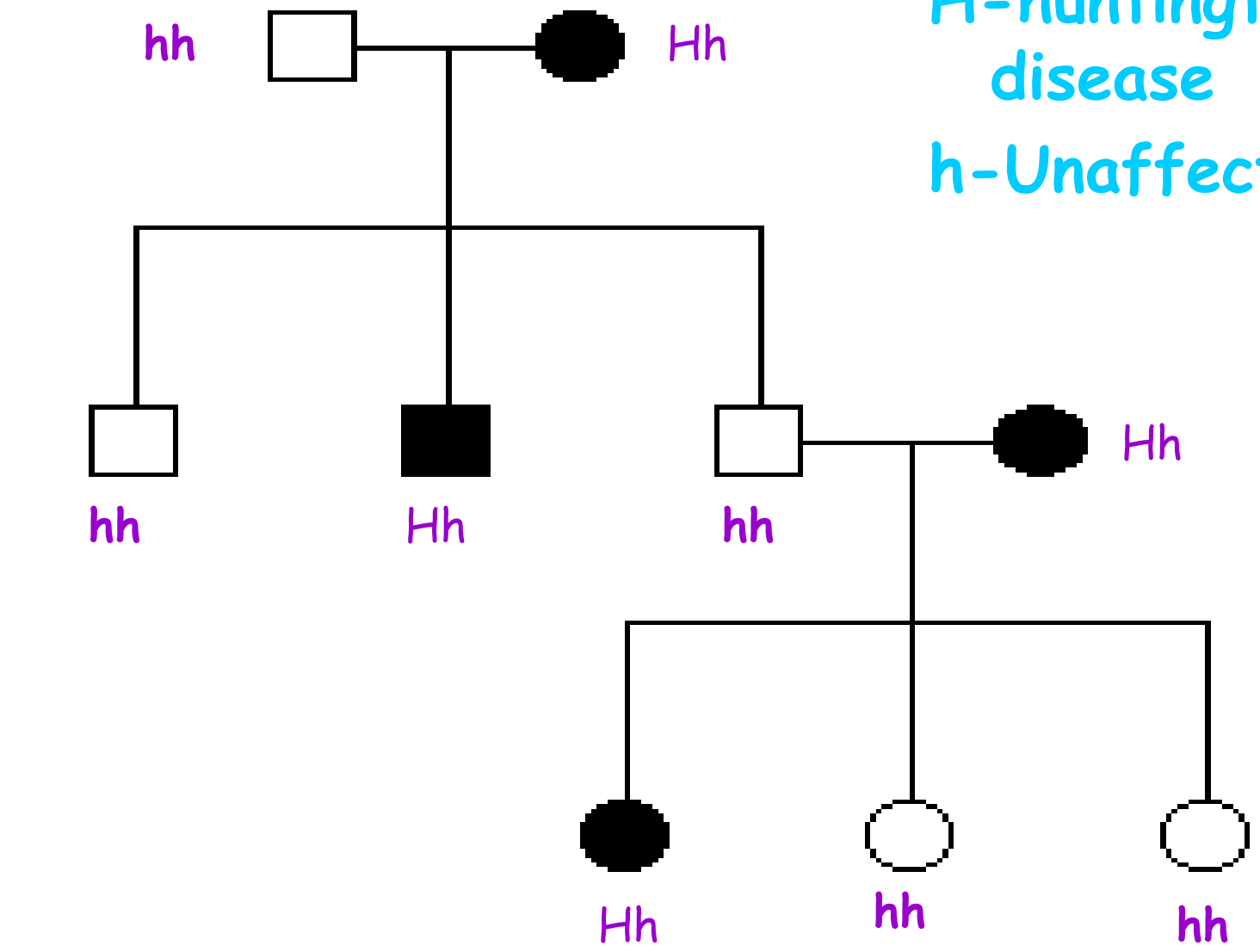
affected male

unaffected male

affected female

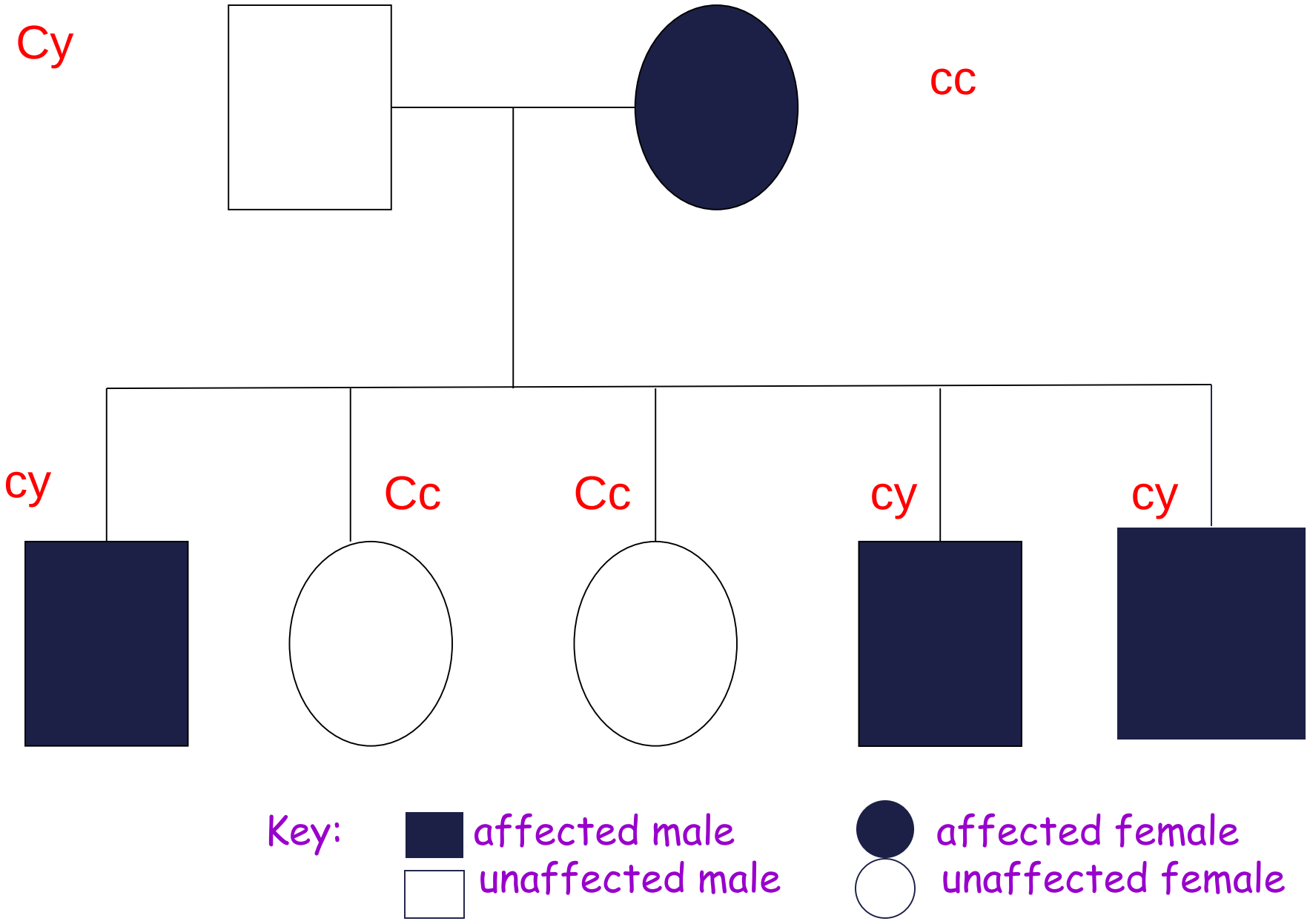
unaffected female


Pp
Pp
PKU
P- Unaffected
p- phenylketonuria
PP or Pp
pp
Pp
pp
pp
Pp
Pp
Key:
affected male
unaffected male
affected female
unaffected female


H-huntington's disease
h-Unaffected
hh
Hh
hh
Hh
hh
Hh
Hh
hh
hh
Key:
affected male
unaffected male
affected female
unaffected female

Sex-Linked Inheritance

Colorblindness



Karyotypes

- To analyze chromosomes, cell biologists photograph cells in **mitosis**, when the chromosomes are fully condensed and easy to see (usually in **metaphase**).

- The chromosomes are then arranged in homologous pairs.

Karyotypes

- The homologous pairs are then placed in order of descending size. The sex chromosomes are placed at the end.

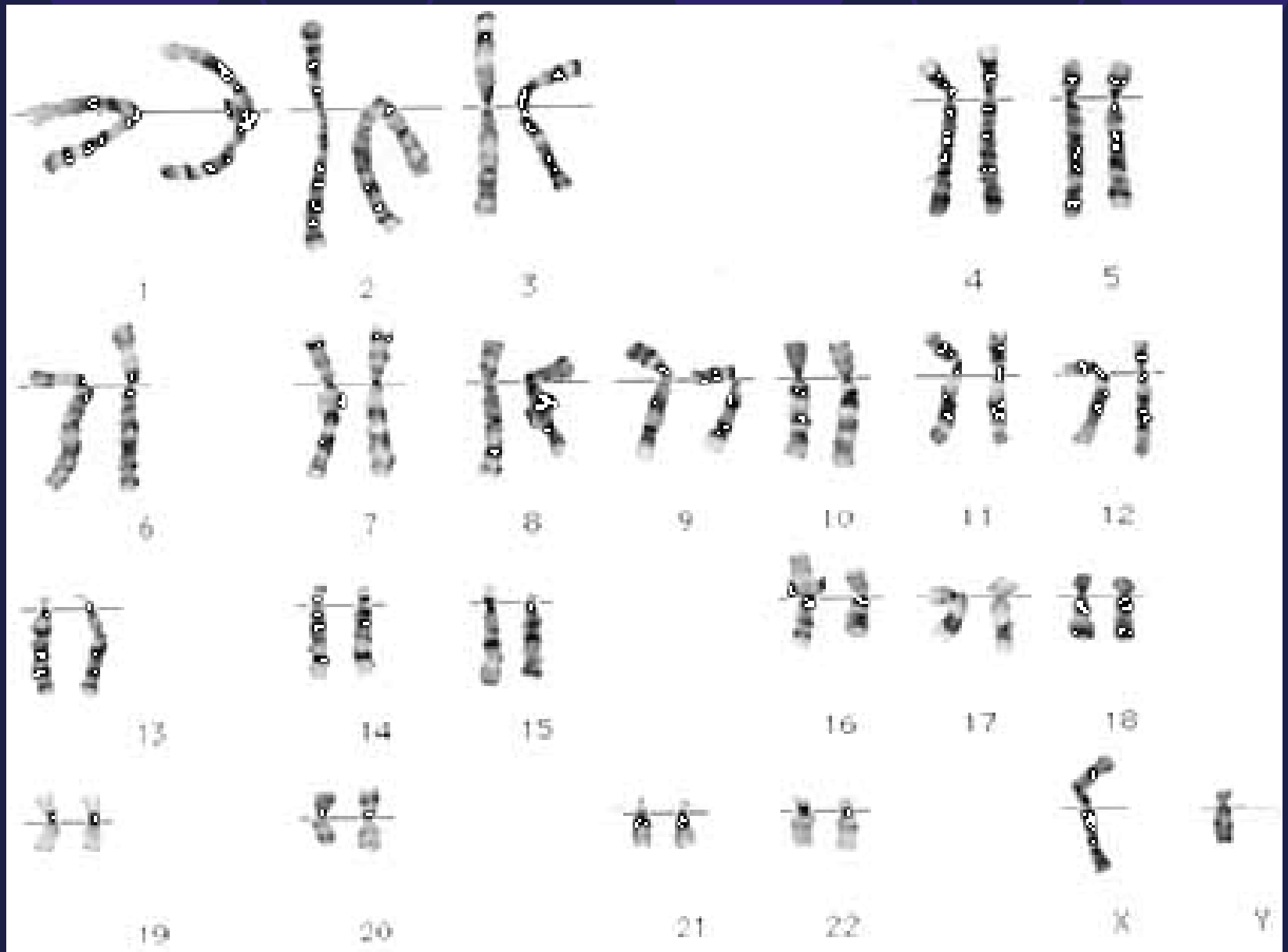
- A picture of chromosomes arranged in this way is known as a **karyotype.**

Karyotypes

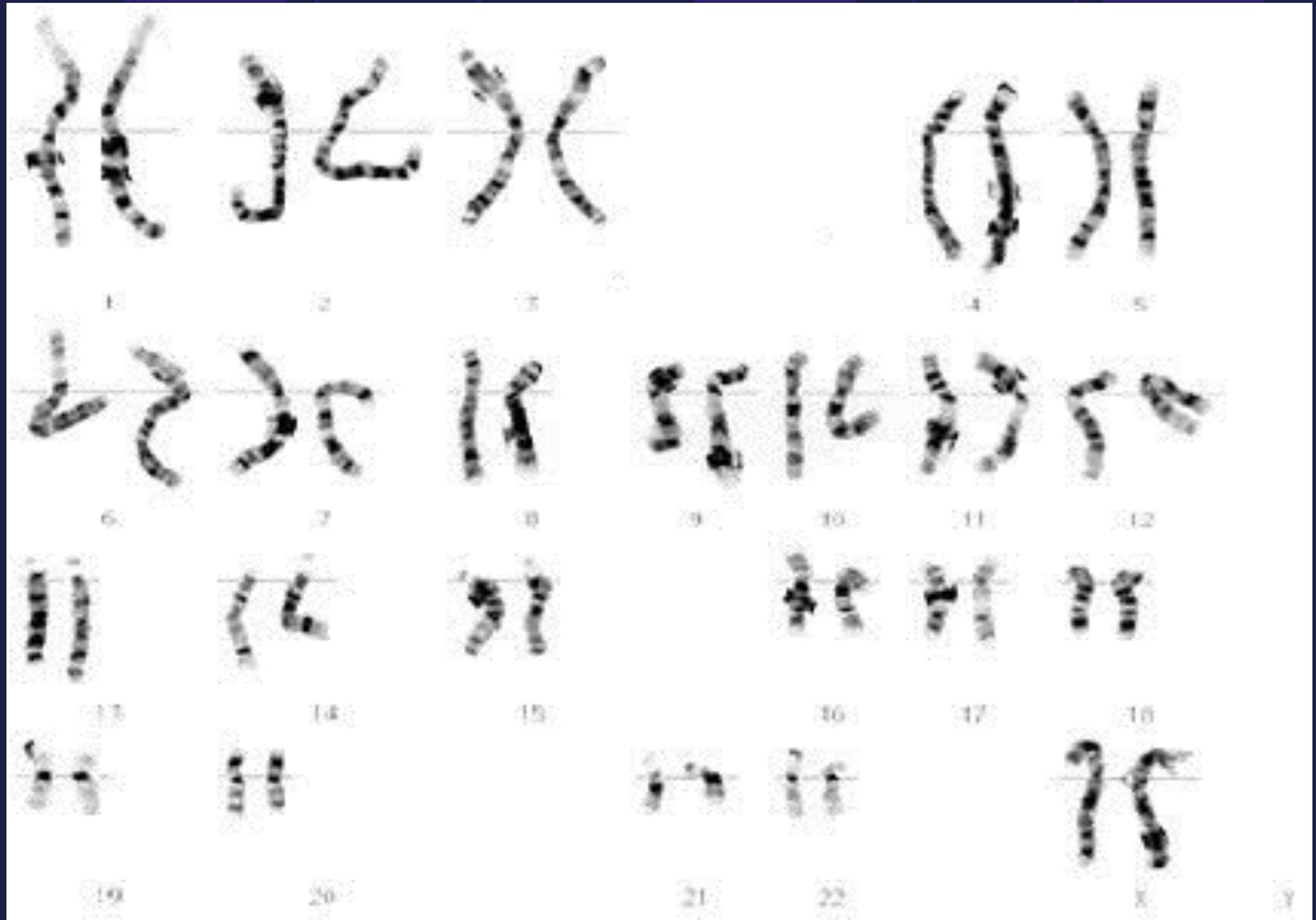
- The **karyotype** is a result of a haploid sperm (23 chromosomes) fertilizing a haploid egg (23 chromosomes).

- The **diploid** zygote (fertilized egg) contains the full 46 chromosomes. (in humans)

Normal Human Male Karyotype: 46,XY



Normal Human Female Karyotype: 46,XX



Labeling a Karyotype

- To label a karyotype correctly, first list the number of chromosomes found in the karyotype. Ex. 46
- Secondly, list the type of sex chromosomes found in the karyotype. Ex. XX
- Lastly, list the any abnormalities at the appropriate chromosome number.

Normal Human Female: 46, XX

Normal Human Male: 46, XY

What are abnormalities?

- Sometimes, during meiosis, things go wrong.
- The most common error is **nondisjunction**, which means "not coming apart".
- If nondisjunction occurs , abnormal numbers of chromosomes may find their way into gametes, and a disorder of chromosome numbers may result.

Autosomal Chromosome Disorders

- Two copies of an autosomal chromosome fail to separate during meiosis, an individual may be born with **THREE** copies of a chromosome.

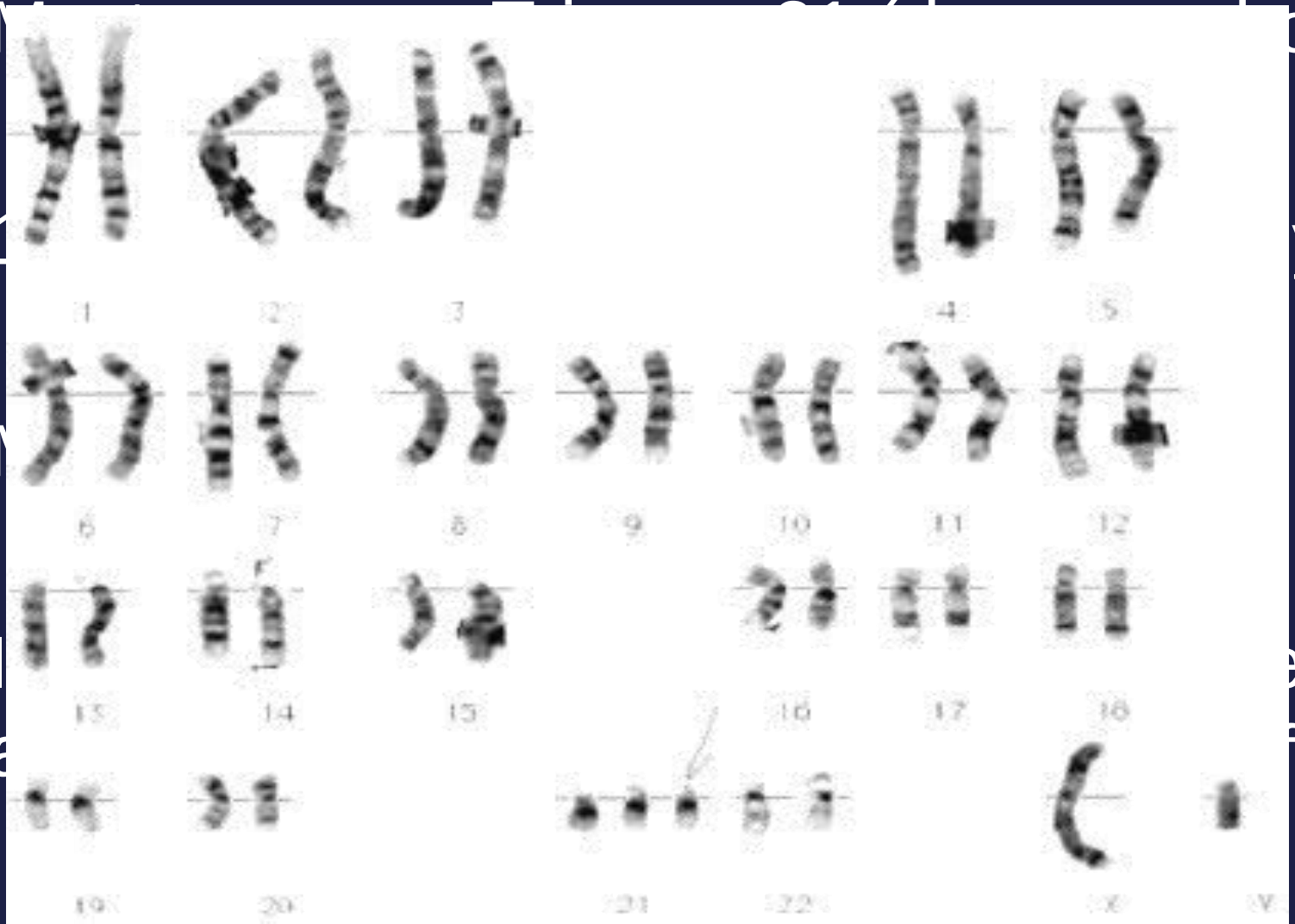
- This is known as a **“Trisomy”**
- Trisomy 13, Trisomy 18, Trisomy 21.

Down Syndrome

Male: 47, XY, +21

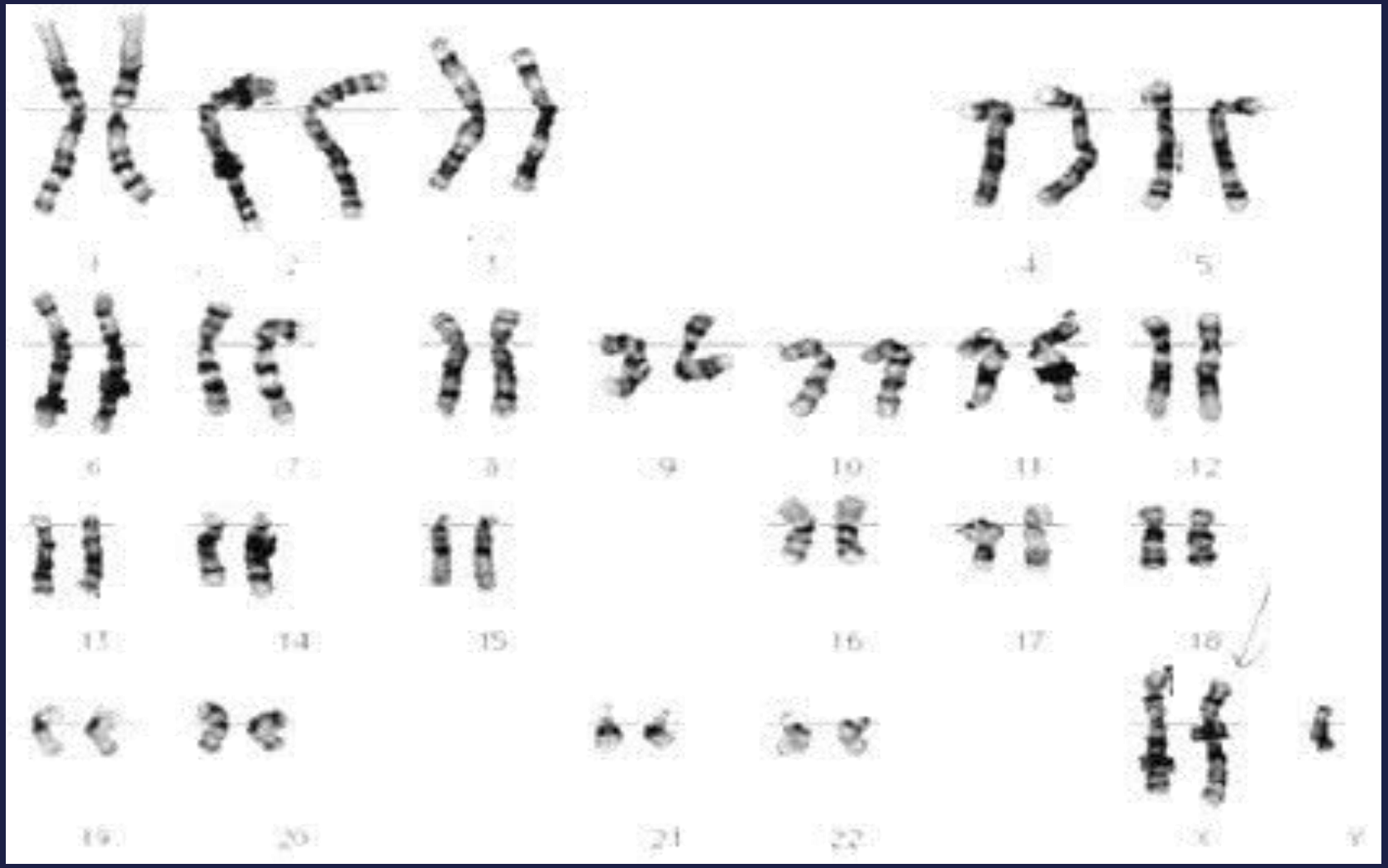
Female: 47, XX, +21

- M ome)
- 1 y 21.
- M
- I es
  a fects.



Sex Chromosome Disorders

Klinefelter's Syndrome, 47 XXY



Other Genetic Disorders

- Sickle Cell Disease
  - Characterized by the bent and twisted shape of the red blood cells.
  - More rigid and get stuck in capillaries. Blood stops flowing and can damage cells, tissues, and organs.
  - Produced physical weakness and damage to the brain, heart, and spleen…could be fatal.
  - Most commonly found in African Americans (can be linked to the incidence of malaria).

Other Genetic Disorders

- **Duchenne Muscular Dystrophy**
  - Sex-linked, defective gene for muscle protein.
  - Progressive weakening and loss of skeletal muscle.
  - In U.S., 1 out of every 3000 males born has condition.