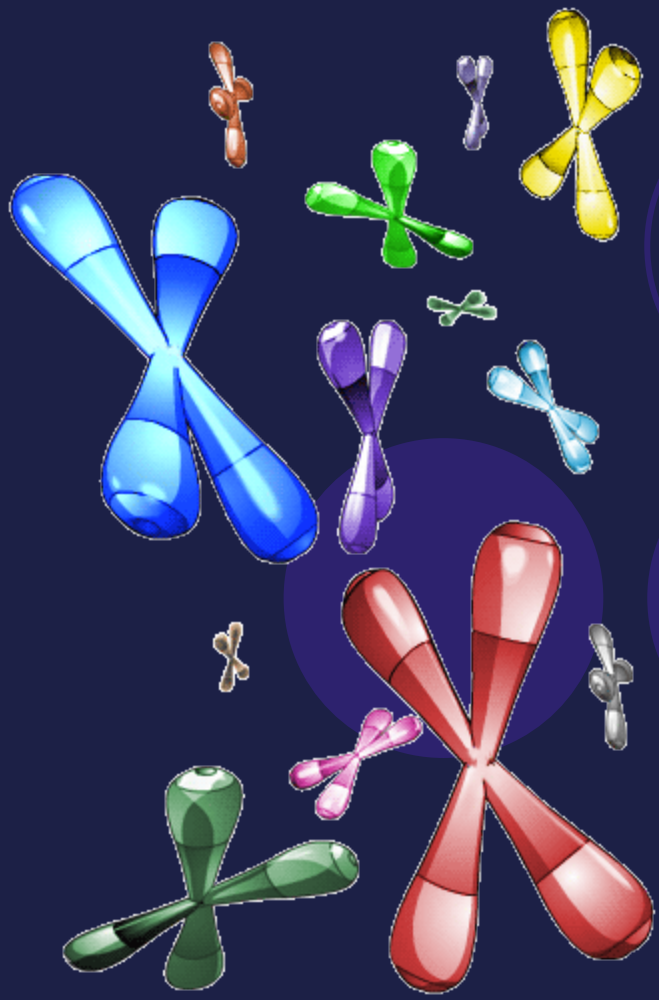
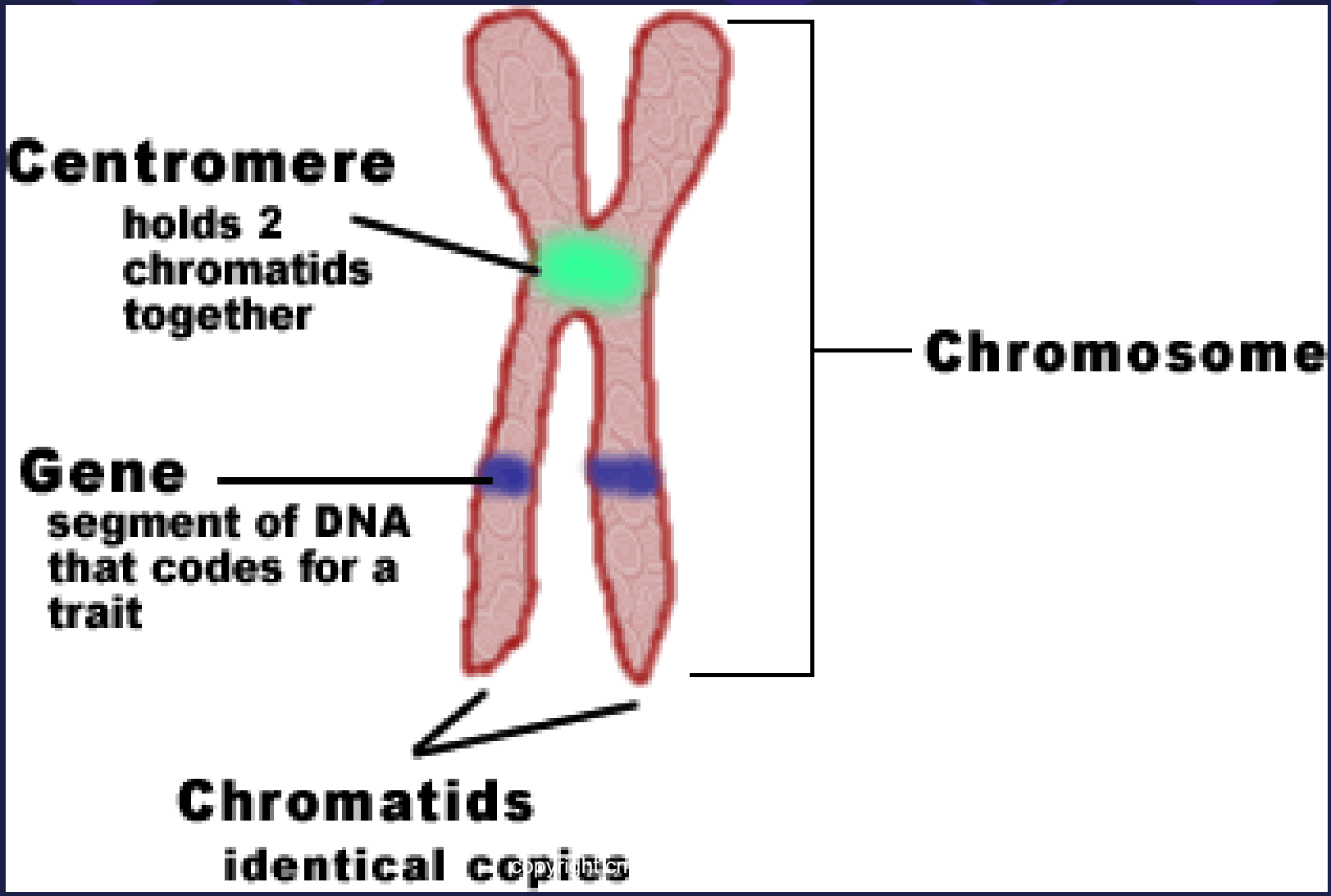


Chromosomes, Karyotypes, and Pedigrees Oh My! 14.1/14.2

Chromosomes



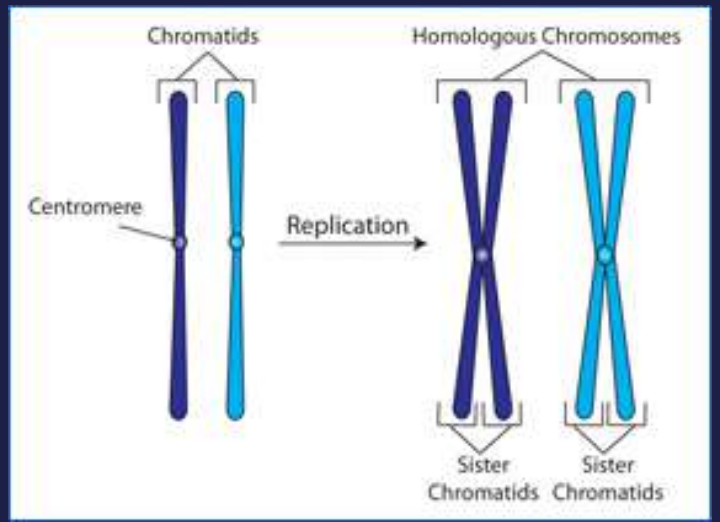
Chromosome Number

- All cells in the human body (SOMATIC CELLS) have 46 or 23 pairs of chromosomes
- Called the DIPLOID or 2n number
- GAMETES (eggs & sperm) have only 23 chromosomes
- Called the HAPLOID or 1n number

Karyotypes

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

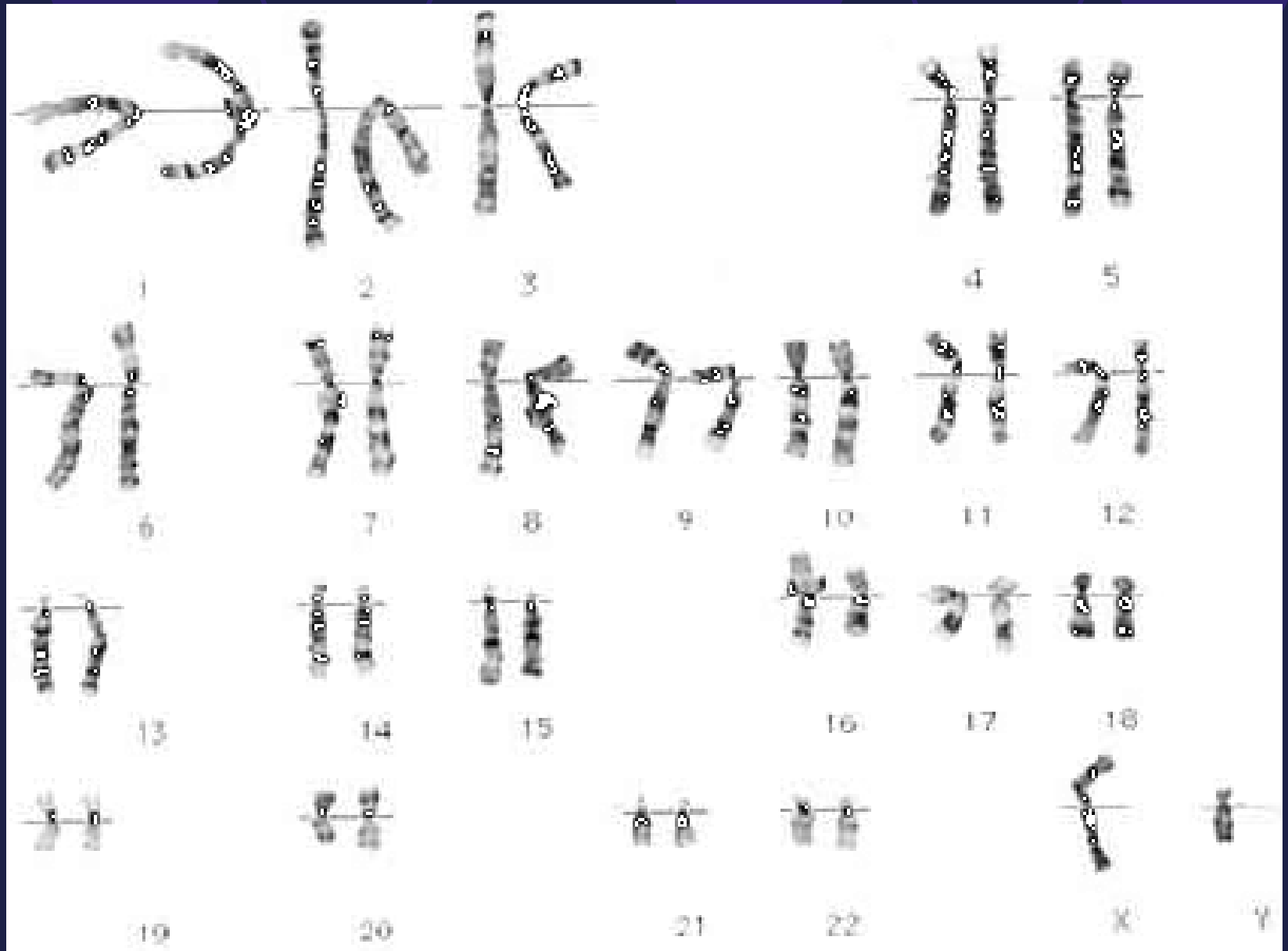
- The chromosomes are then arranged in homologous pairs.



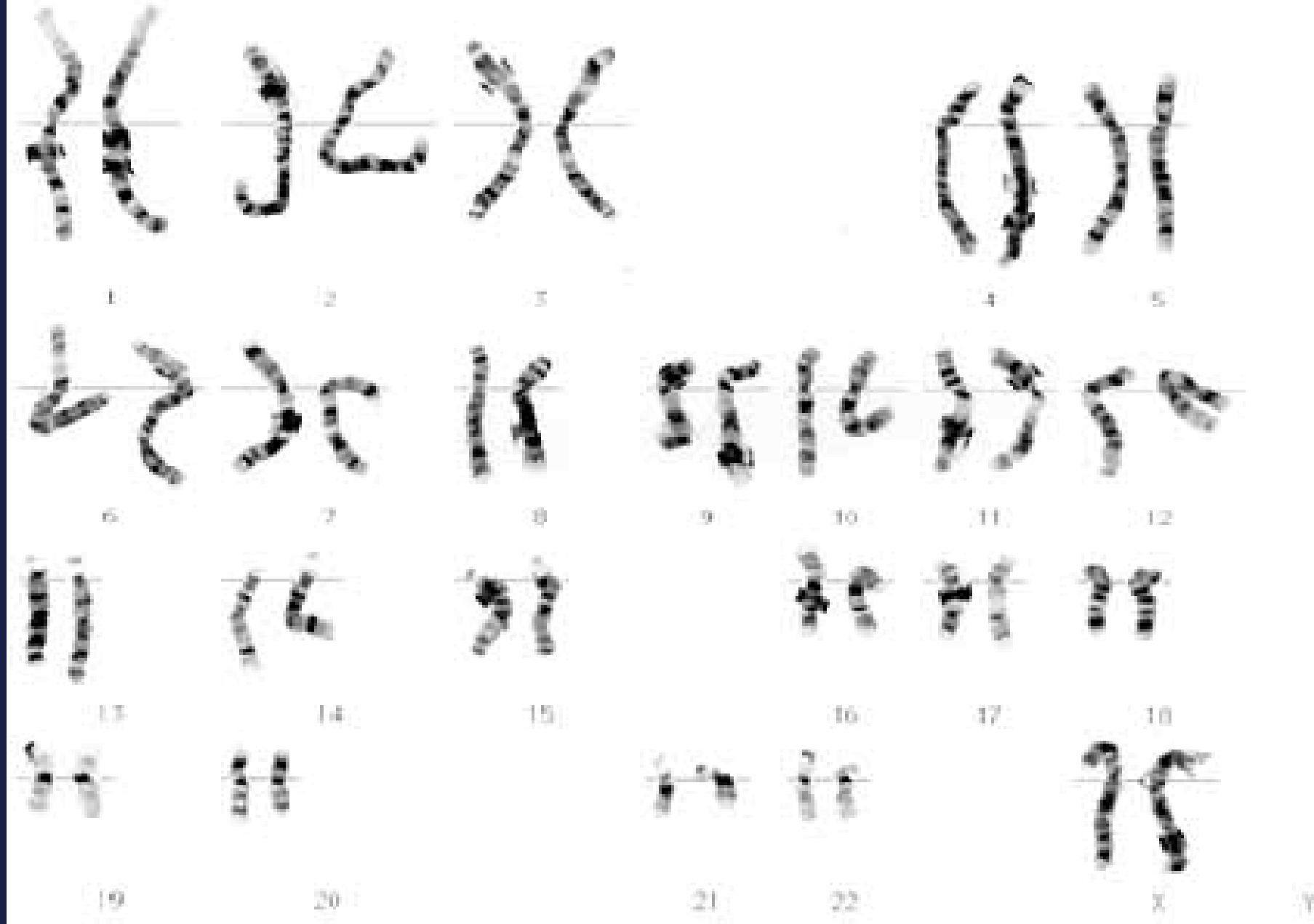
Karyotypes

- @The homologous pairs are then placed in order of descending size@. (autosomal chromosomes)
- The sex chromosomes are placed at the end.
- A picture of chromosomes arranged in this way is known as a **karyotype.**

Normal Human Male Karyotype: 46,XY



Normal Human Female Karyotype: 46,XX



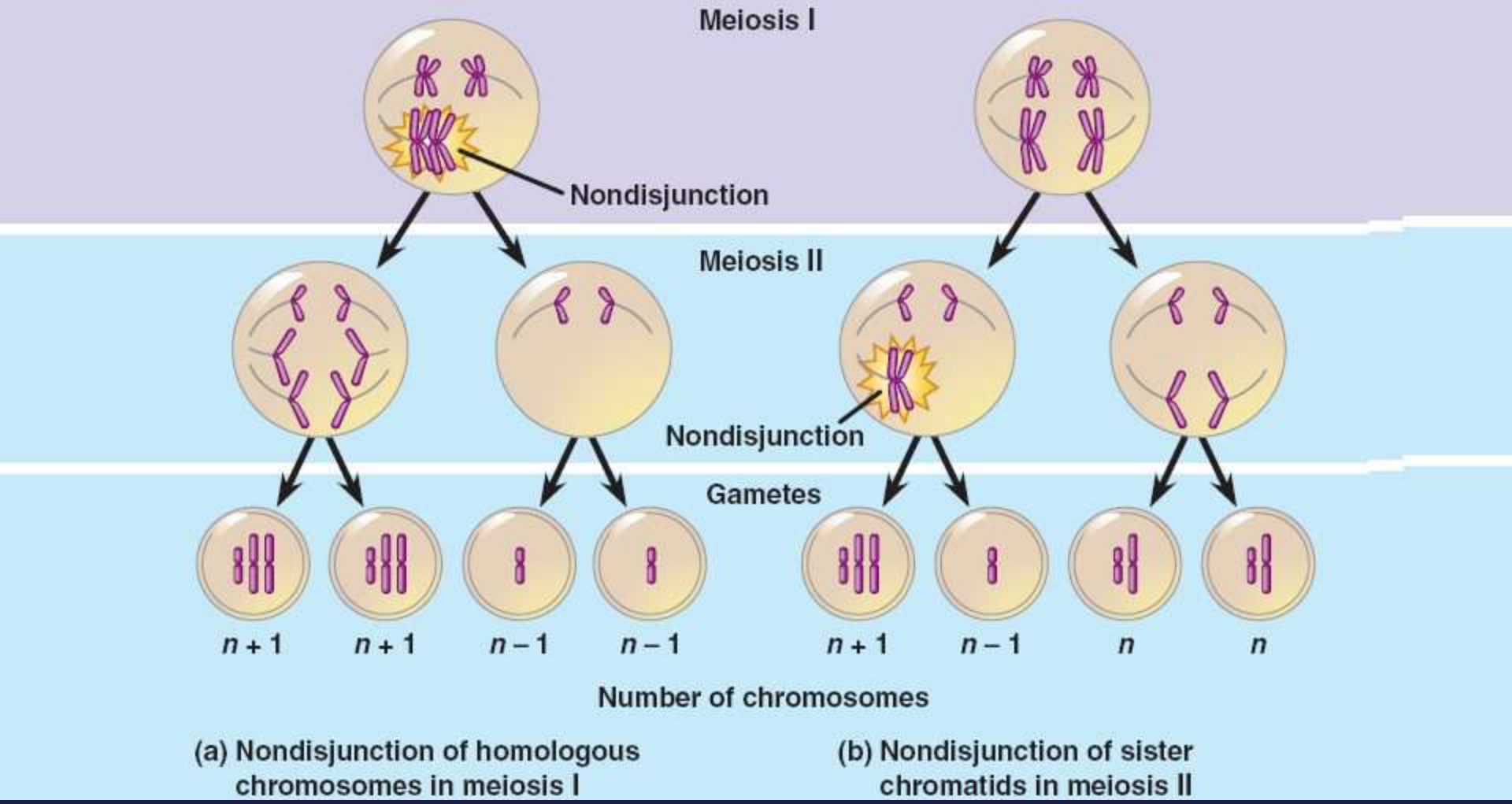
Labeling a Karyotype

- 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.

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.

Nondisjunction



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.

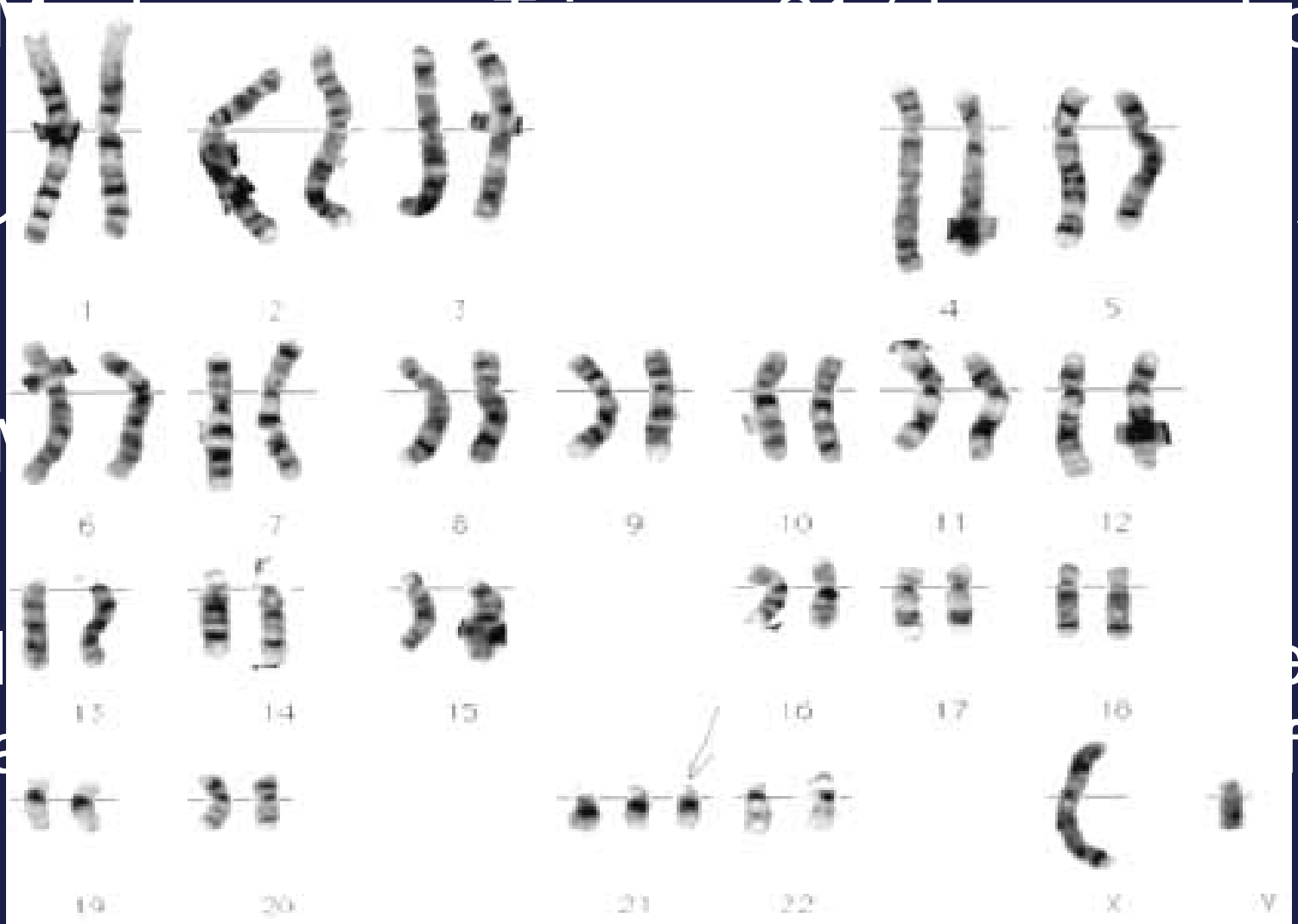
- This is known as a **"Trisomy"**
- Trisomy 13, Trisomy 18, Trisomy 21.

Down Syndrome

Male: 47, XY, +21

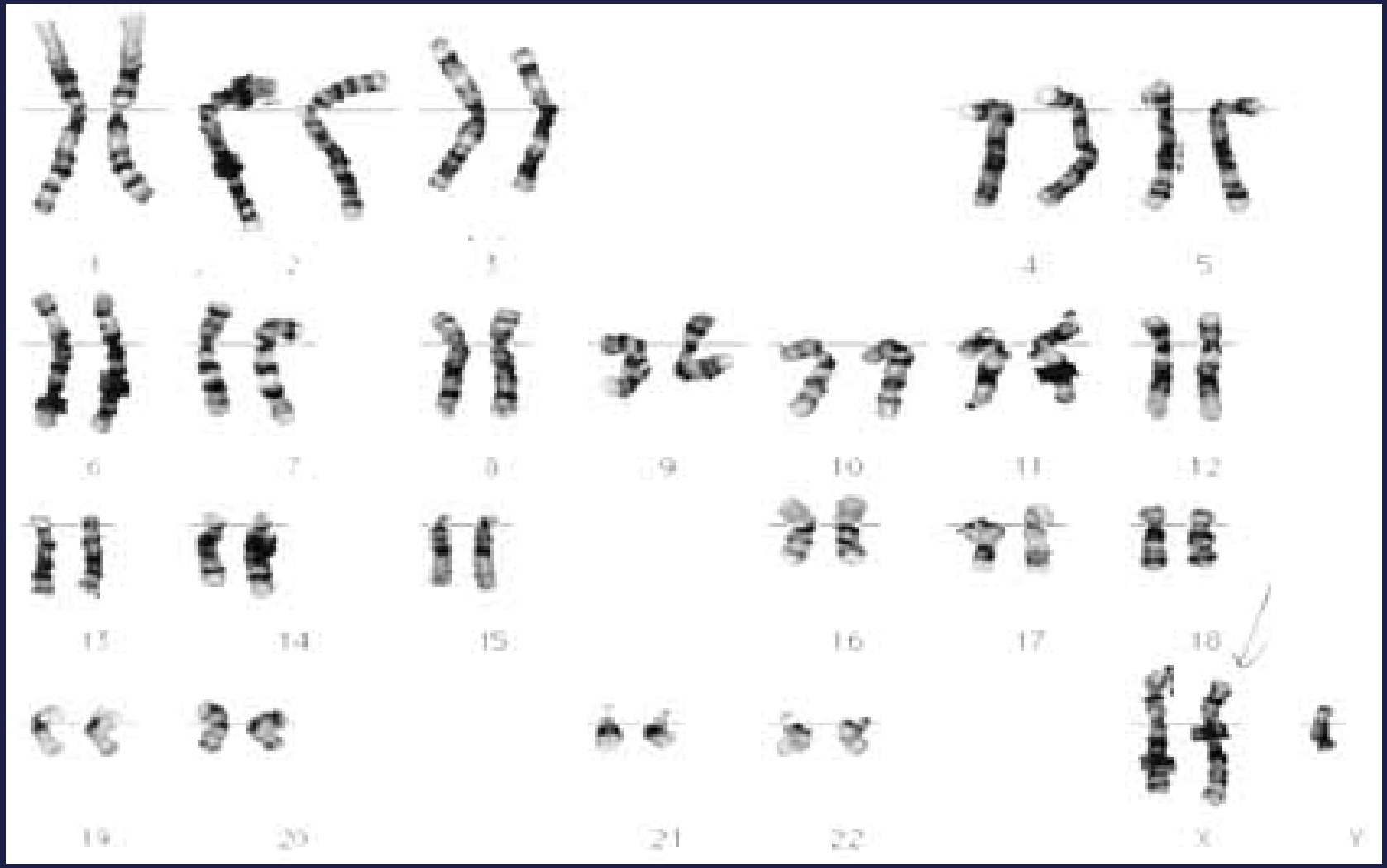
Female: 47, XX, +21

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



Sex Chromosome Disorders

Klinefelter's Syndrome, 47 XXY

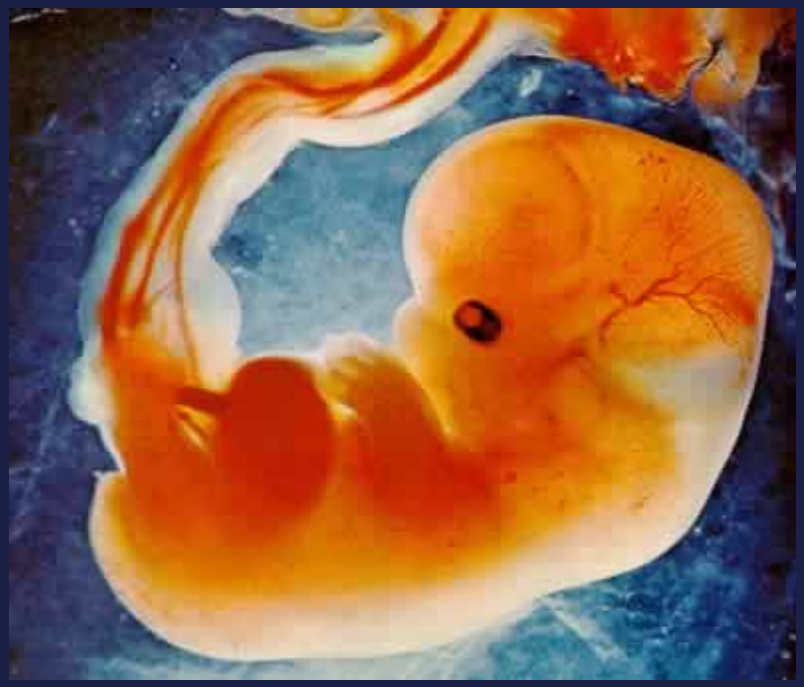


Other Genetic Disorders

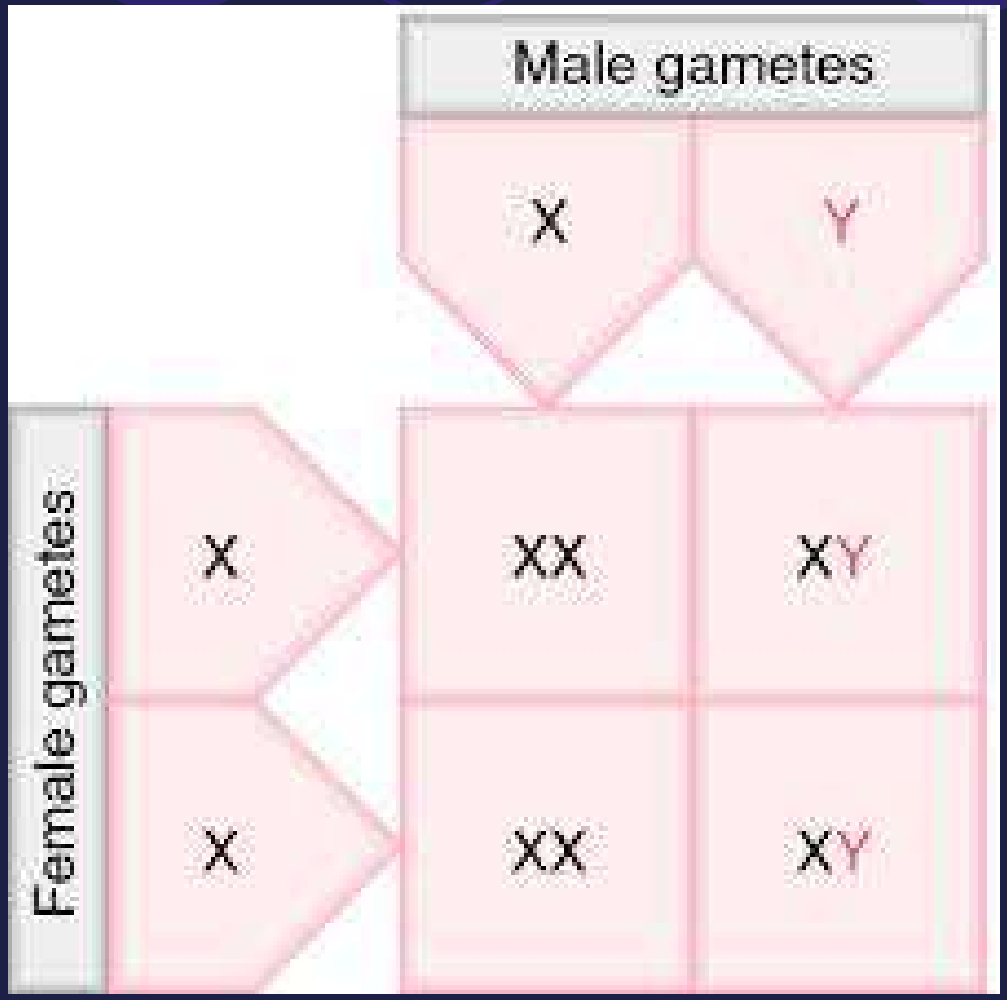
- Sickle Cell Disease
  - Characterized by the bent and twisted shape of the red blood cells.
  - More rigid and get stuck in capillaries. Blood stops flowing and can damage cells, tissues, and organs.
  - Produced physical weakness and damage to the brain, heart, and spleen…could be fatal.
  - Most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

The inheritance of gender

Is it going to be a boy or a girl?



The inheritance of gender

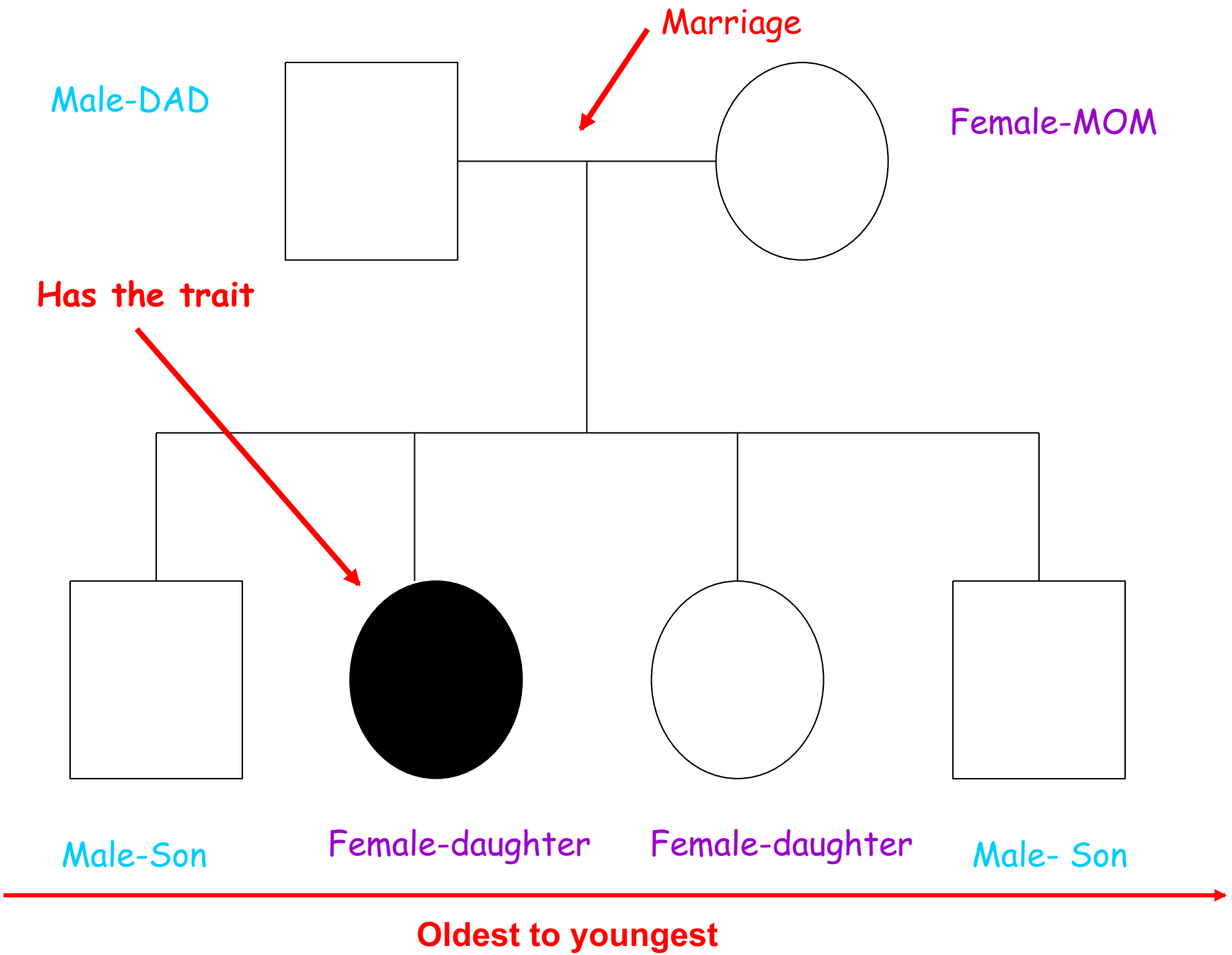


Chance of a girl 50%
Chance of a boy 50%

Pedigree

- A pedigree shows the relationships within a family and it helps to chart how one gene can be passed on from generation to generation.

- @Pedigrees are tools used by genetic researchers or counselors to identify a genetic condition running through a family@, they aid in making a diagnosis, and aid in determining who in the family is at risk for genetic conditions.


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

- 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.

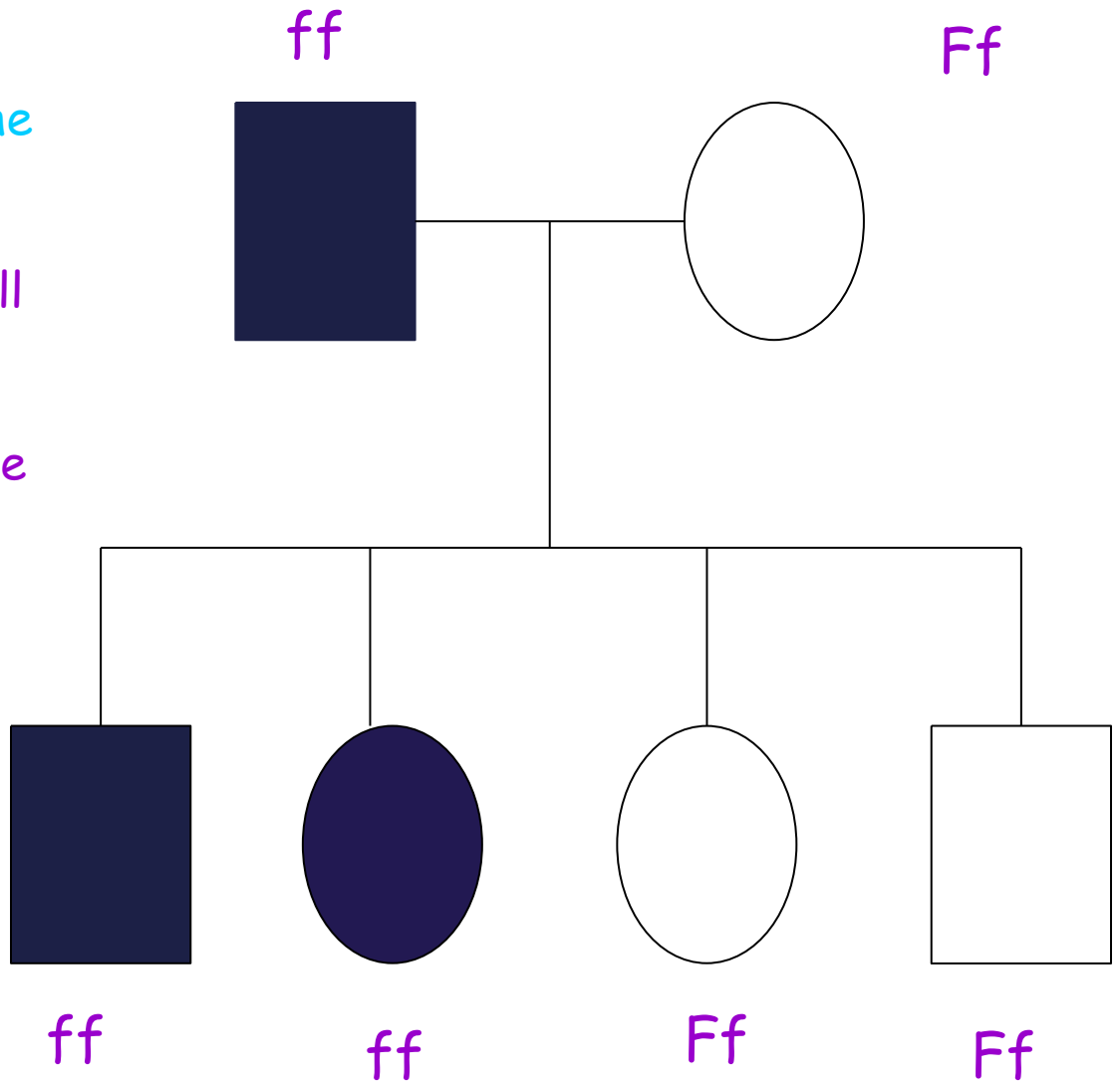
Steps:

- 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

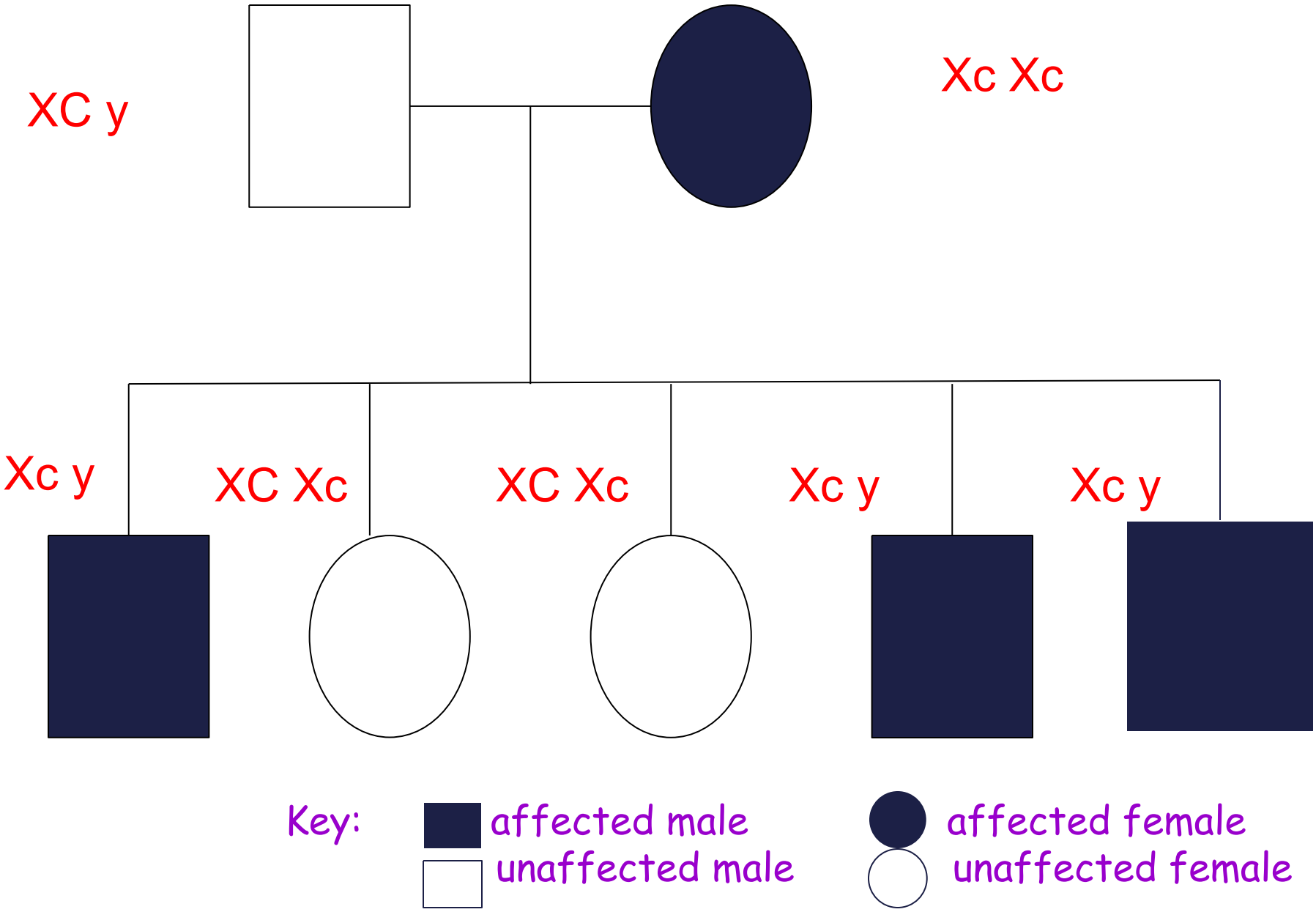
unaffected male

affected female

unaffected female

Sex-Linked Inheritance

- Colorblindness



A male with down's syndrome (extra 21st chromosome) would be labeled as

A. 45 XY

B. 46 XY

C. 47 XX

D. 47 XY

When chromosomes do not separate properly in meiosis, this is called

A. Karyotype
B. Pedigree
C. Kleinfelters
D. Nondisjunction

If a female is a carrier for hemophilia (recessive) $X^{H}X^{h}$ and is married to a man with hemophilia $X^{h}Y$, what is the probability that she will have a daughter with hemophilia?

	Xh	Y
XH	XH Xh	XH Y
Xh	Xh Xh	Xh Y

50% Xh Xh