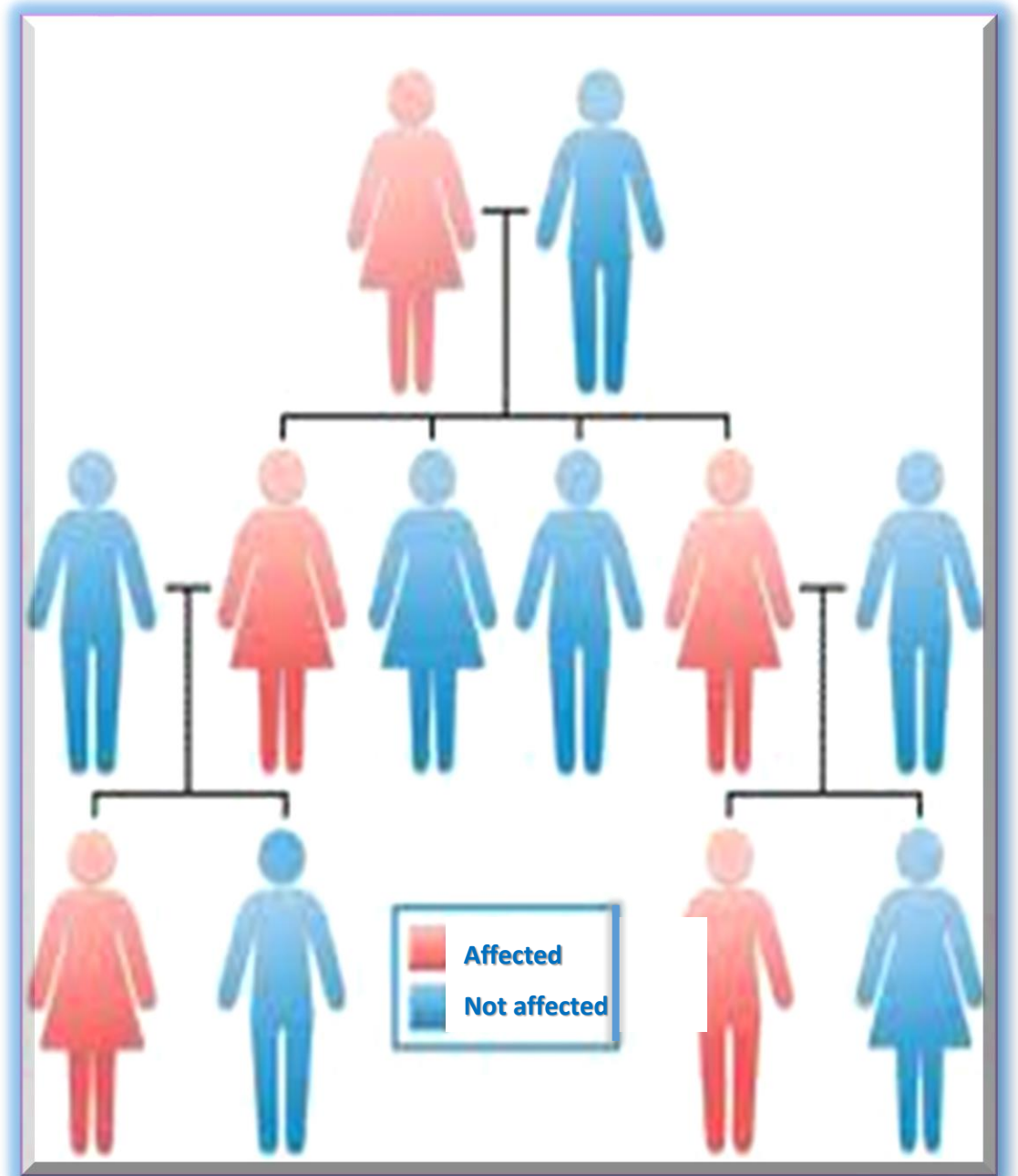


# Laws of inheritance

By:

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Associate Professor of biochemistry &  
Molecular Biology



# Learning outcomes

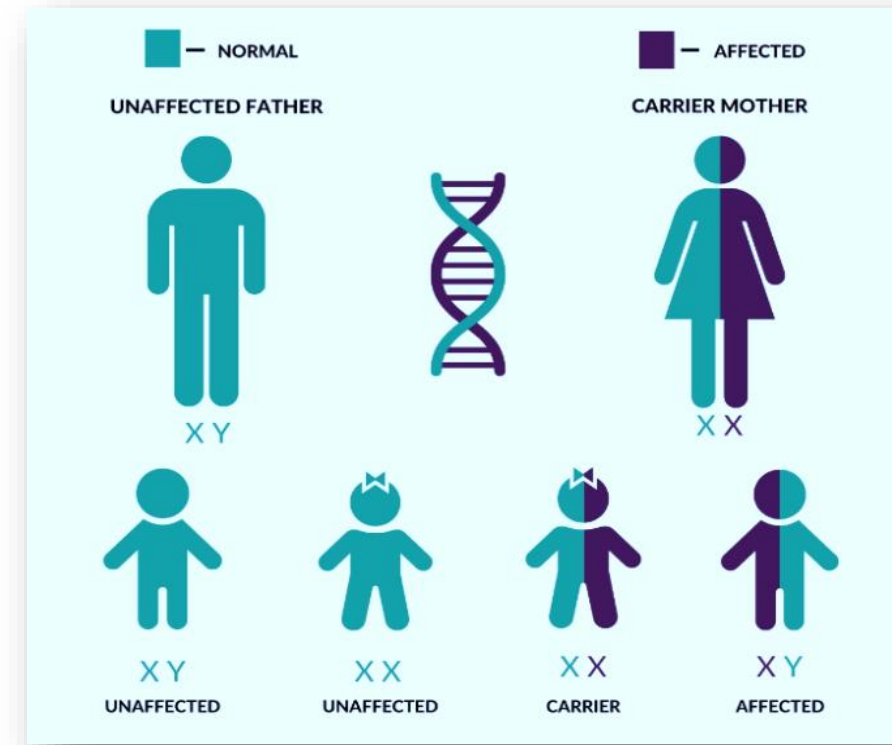


By the end of the lecture, the students should be able to:

- Identify different principles & patterns of heredity (Mendelian and Non-Mendelian Inheritance).
- List & recognize the non-traditional patterns of inheritance.
- Draw a pedigree for a given family and label it.
- Determine different patterns (Mode) of inheritance and correlate different patterns of inheritance with genetic diseases

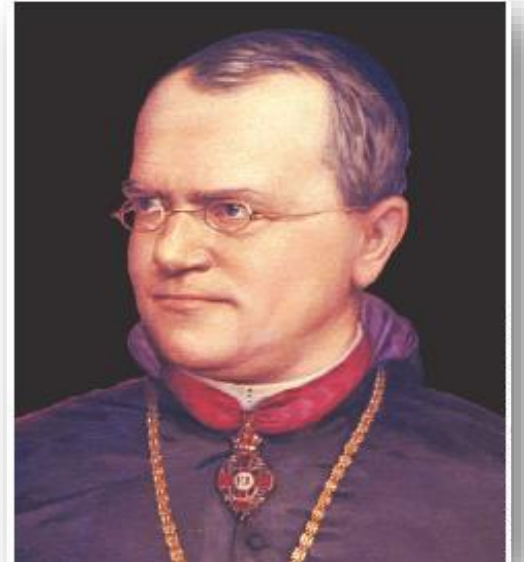
# Inheritance

- Trait is any genetically determined characteristics of the individual
- Inheritance is how traits, or characteristics, are passed on from generation to generation (from parent to offspring).

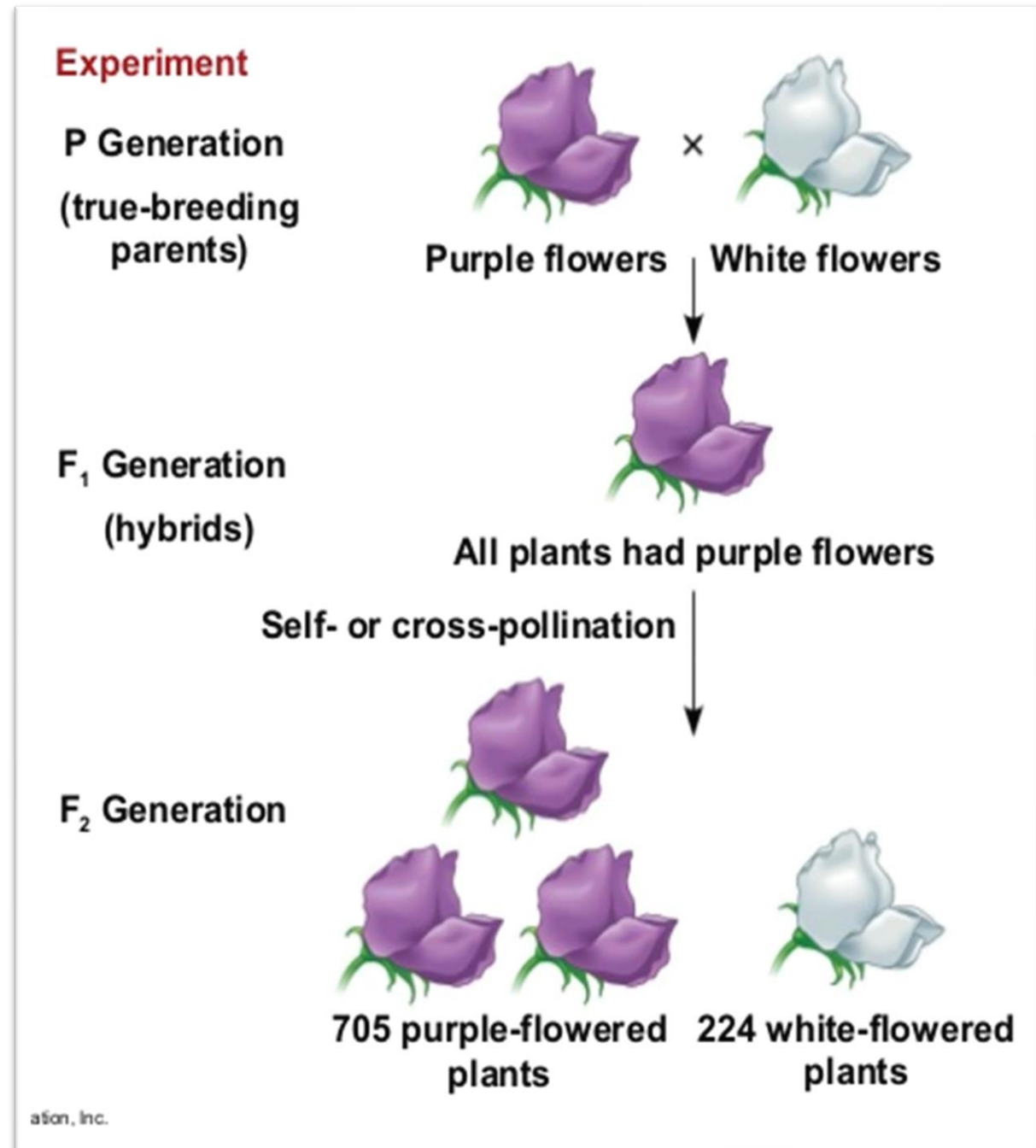


# Genetic Principles

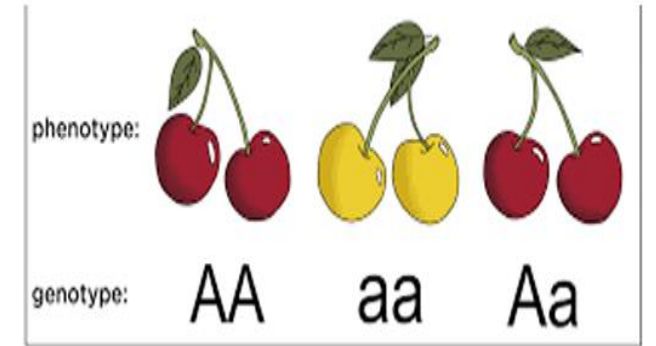
- The study of genetics started with the work of Gregor Mendel (**The Father of Genetics**)
- He laid down **the basic principles of heredity**.
- These principles assisted the modern understanding of how traits may be inherited



- Mendel demonstrated that **traits are passed from parents to offspring through discrete factors, genes, located on chromosomes.**
- His work showed that some traits are **dominant** over others and that **segregation** and **independent assortment** of **alleles** allows for prediction of phenotypic ratios in offspring.



# Mendel's Principles (Laws of inheritance)

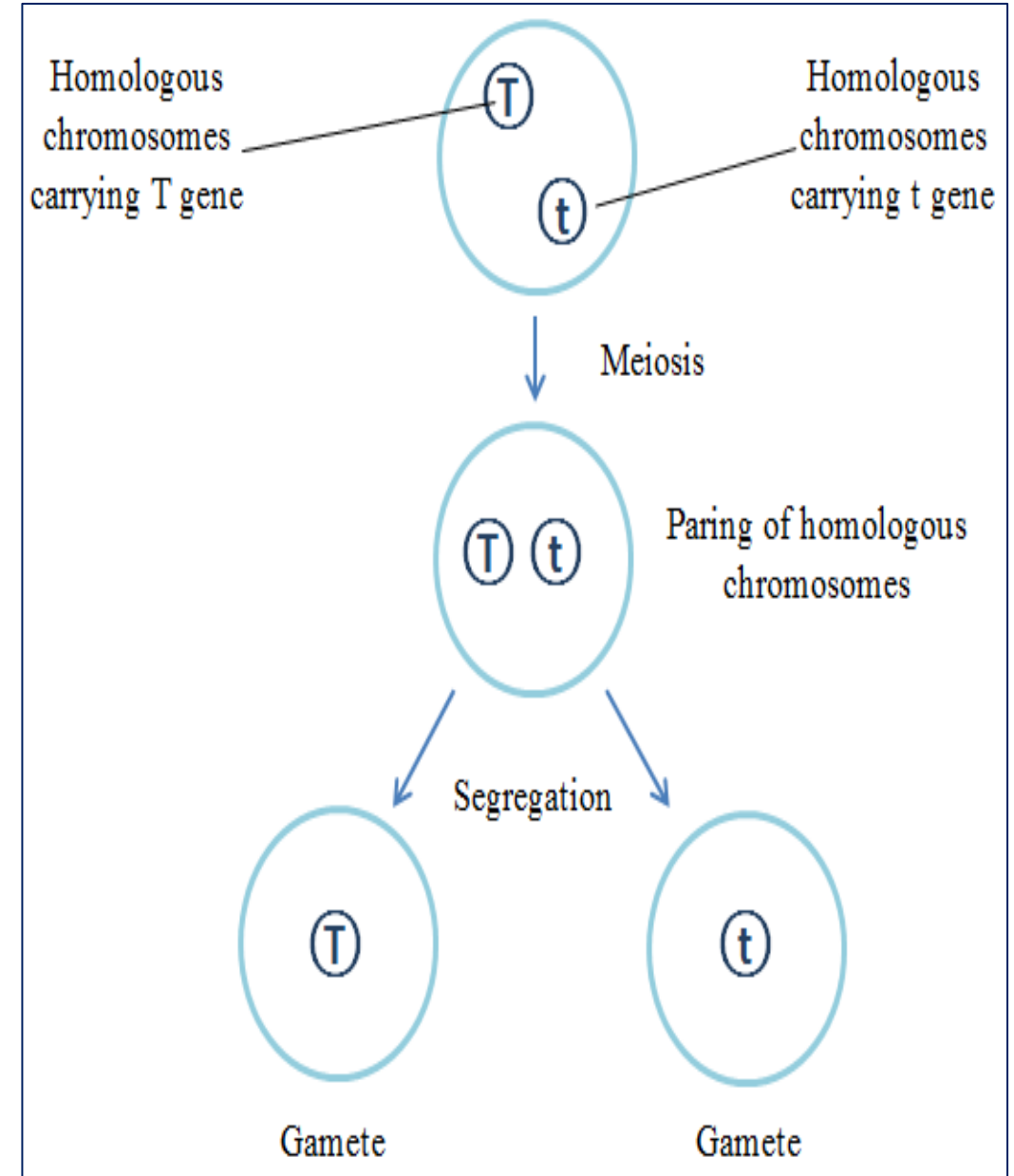


## 1. Principle (Law) of Dominance:

- The inheritance of traits controlled by a **single gene** with **two alleles**, one of which may be completely **dominant** to the other.
- An organism with at least one dominant allele displays the effect irrespective of the presence of recessive one.

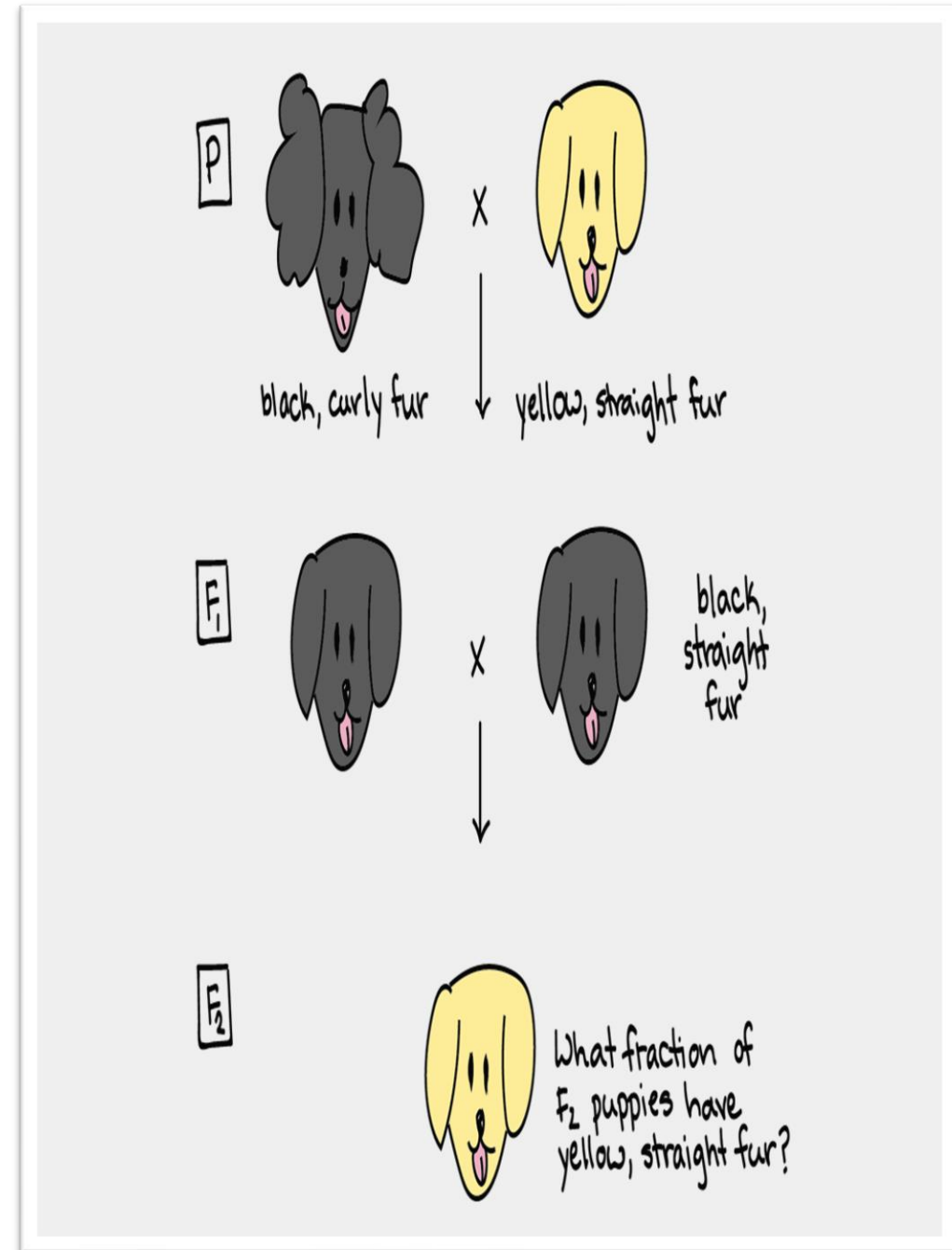
## 2.Principle of Segregation:

When gametes are formed, the 2 alleles for each gene are packed into separate gamete (segregate), so that **each gamete carries only one allele** of the pair for each gene.



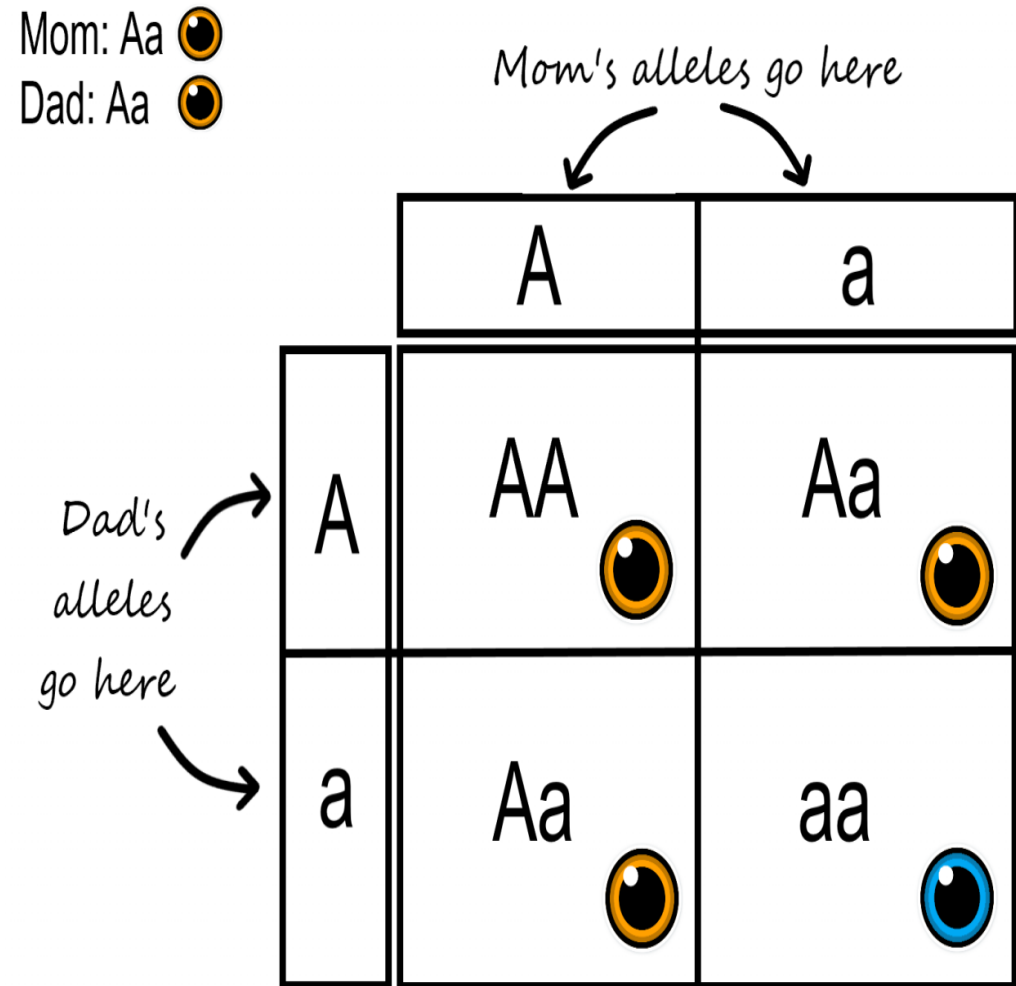
### 3. Principle of Independent Assortment:

- ❑ a pair alleles of one trait separate independently of another pair of another traits during gamete formation =The expression of certain trait (gene) does not affect the expression of another trait.
- ❑ As the individual heredity factors assort independently, different traits get equal opportunity to occur together.
- ❑ Eg –Skin color and hair type  
- Person height and eye color



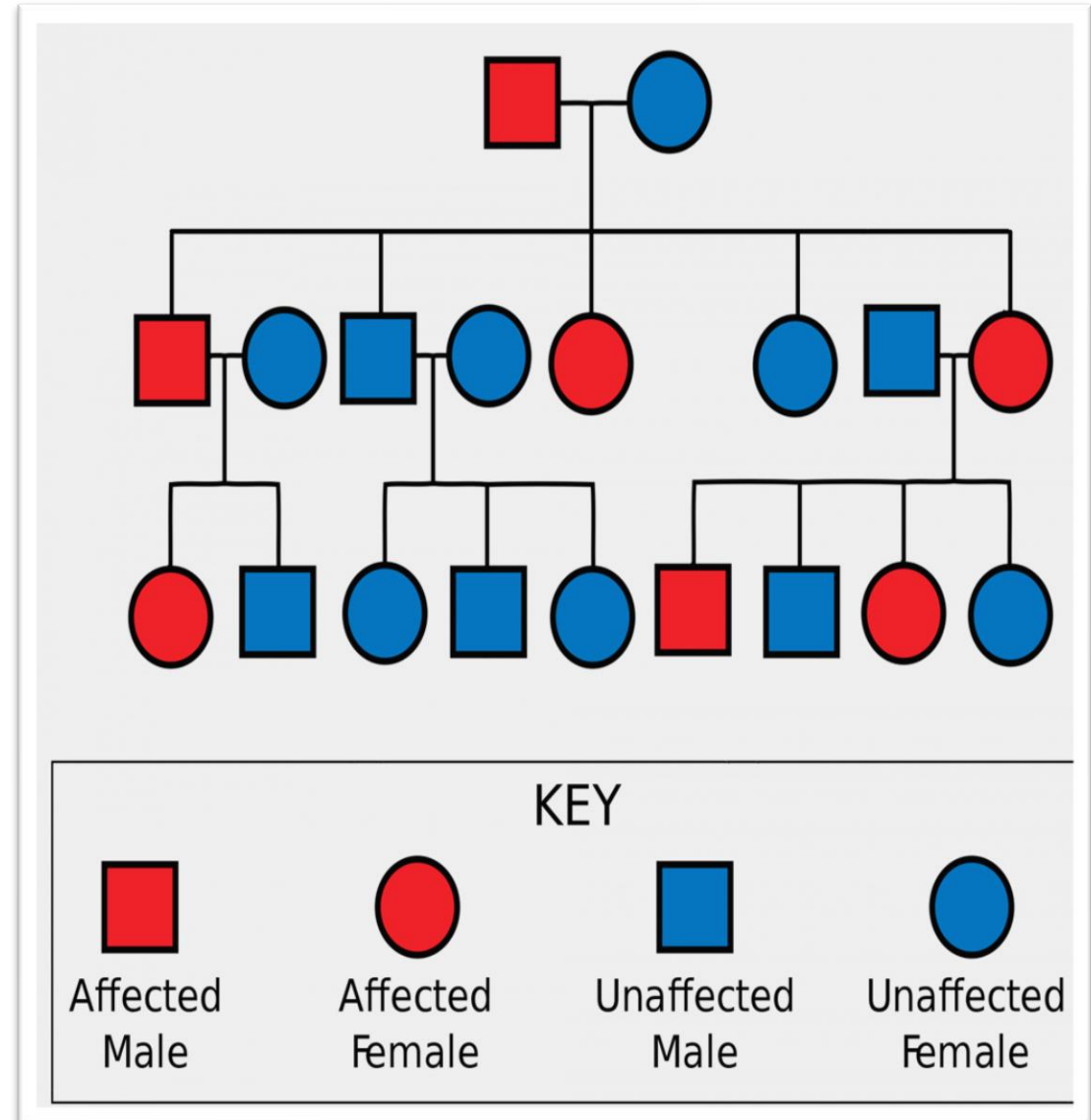
# Punnett square

- It is a chart that allows to easily determine **the expected ratios** “Probability” of **possible genotypes** in the offspring of two parents.
- Considered a method of determining the **probabilities of which traits** the offspring of two organisms **will inherit**



# Family pedigree

- A **family** pedigree shows how a trait is passed from generation to generation within a family.
- It is used to track the appearance of one trait or disease
- A pedigree can show the **hereditary pattern** “whether a Mendelian trait is **autosomal or X-linked trait/ dominant or recessive**”



# Non-Mendelian Inheritance

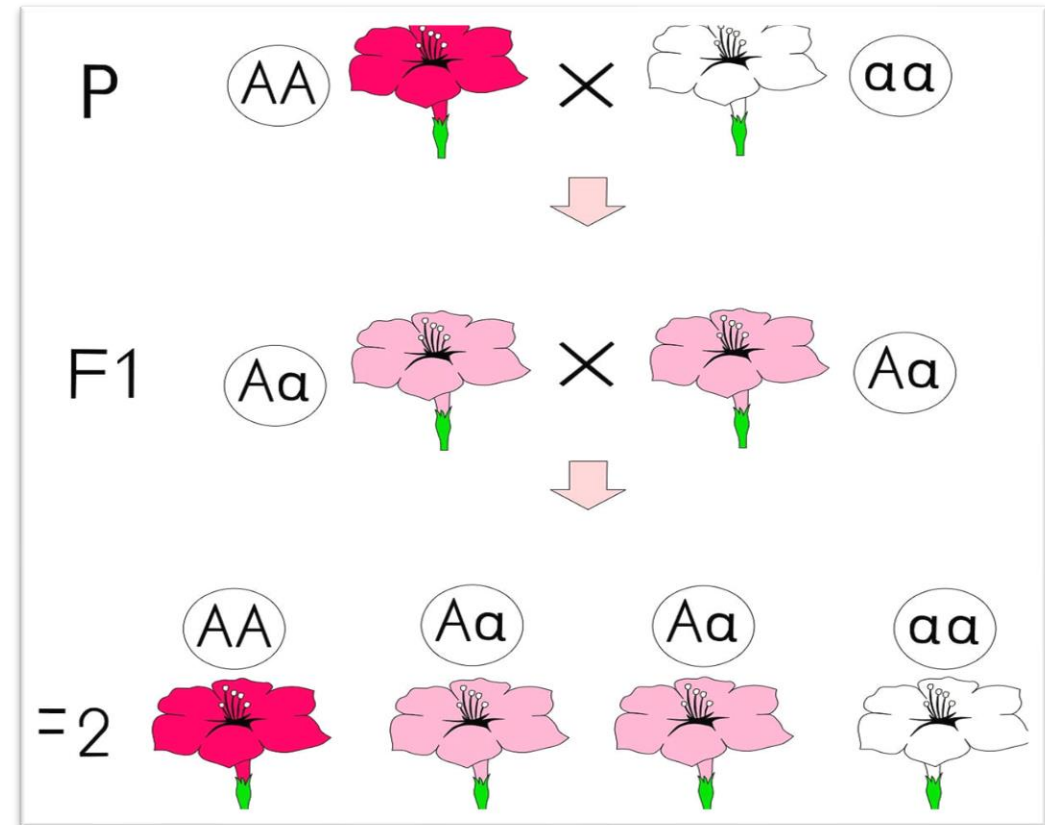
- **Mendelian Genetics (Complete Dominance):**
  - ❑ Genes have only two alleles.
  - ❑ So, there are only 2 possible Phenotypes: either **dominant** or **recessive**.
- **But not all inheritance is based on the rules of Complete Dominance!!**



# Non-Mendelian Genetics

There are other types of inheritance that **Mendel never considered**:

- **Incomplete Dominance**
- **Multiple Alleles**
- **Codominance**
- **Polygenic Traits**



## (Incomplete Dominance)

- The heterozygous offspring displays a **THIRD Phenotype!!**  
**(intermediate phenotype)**
- A good example of incomplete dominance **in humans** is **hair type**.
- There are genes for **straight** and **curly hair**, and if an individual is heterozygous, they will typically have the phenotype of **wavy hair**.



**Homozygous  
Straight**

**Heterozygous**

**Homozygous  
Curly**

## Multiple Alleles







☐ Genes often exist in several different forms and are therefore said to have multiple alleles.

☐ **Example: Human Blood group**  
(A, B, O, or AB)

- **Multiple allele (A, B, and O)**
  - 2 alleles are **dominant** “A and B”
  - one allele is **recessive** “O”

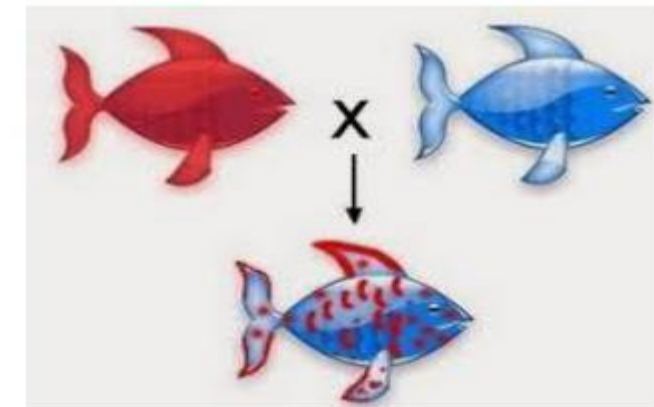
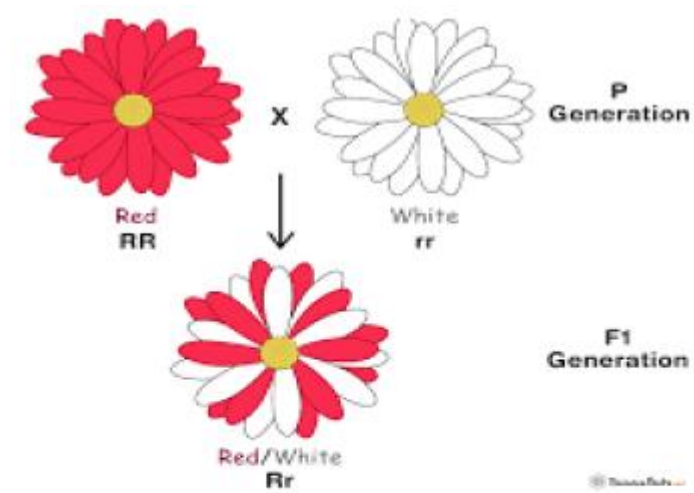
**[3 alleles & 4 phenotypes]**

**NB:** Codominance [A and B alleles / **AB blood type**]

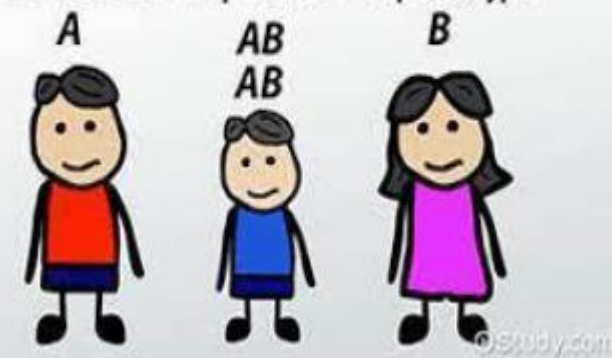
Genotype	Phenotype(blood type)
AA	A 
AO	A 
BB	B 
BO	B 
OO	O 
AB	AB 

# Co-dominance

- ❑ Both alleles of a gene are expressed simultaneously (together) in the phenotype of individuals. [Co means “together”].
- ❑ Both traits are dominant and expressed in phenotype
- ❑ Example in human: AB blood group {A is dominant , B is dominant , When present together in the same person called Codominant}



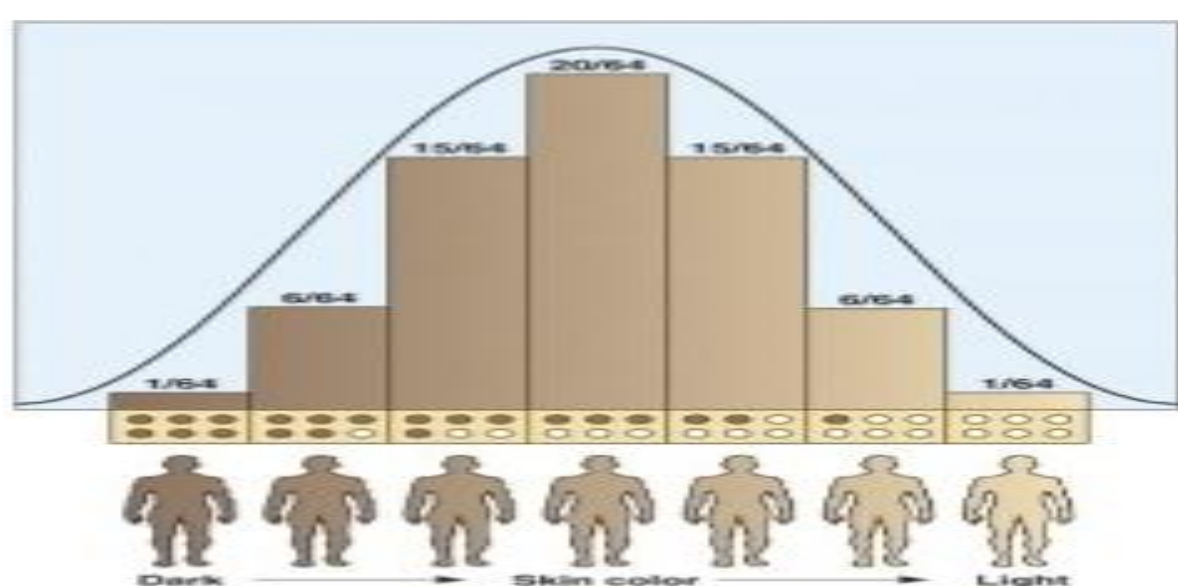
**Codominance**  
both alleles are expressed in the phenotype



# Polygenic Inheritance

## Polygenic Traits

- Traits controlled by interaction of two or more genes (**multiple genes**) are said to be **polygenic traits**.” additive effect:
- **E.g. skin color and height.**
- Skin color is a common example of a polygenic trait it is governed by **6 genes and at least 12 alleles**.



# Environmental Effects on Phenotype (Multifactorial inheritance)

- ❑ Many traits are affected by the environment, as well as by genes.
- ❑ This may be especially true for **polygenic traits**.

❑ E.g.

- **Adult height**, for example, affected by **availability of nutrients**
- **Skin color**
  - There is a **wide range of skin colors** in people worldwide.
  - In addition to differences in **genes**, differences in **exposure to UV light** cause some variation.

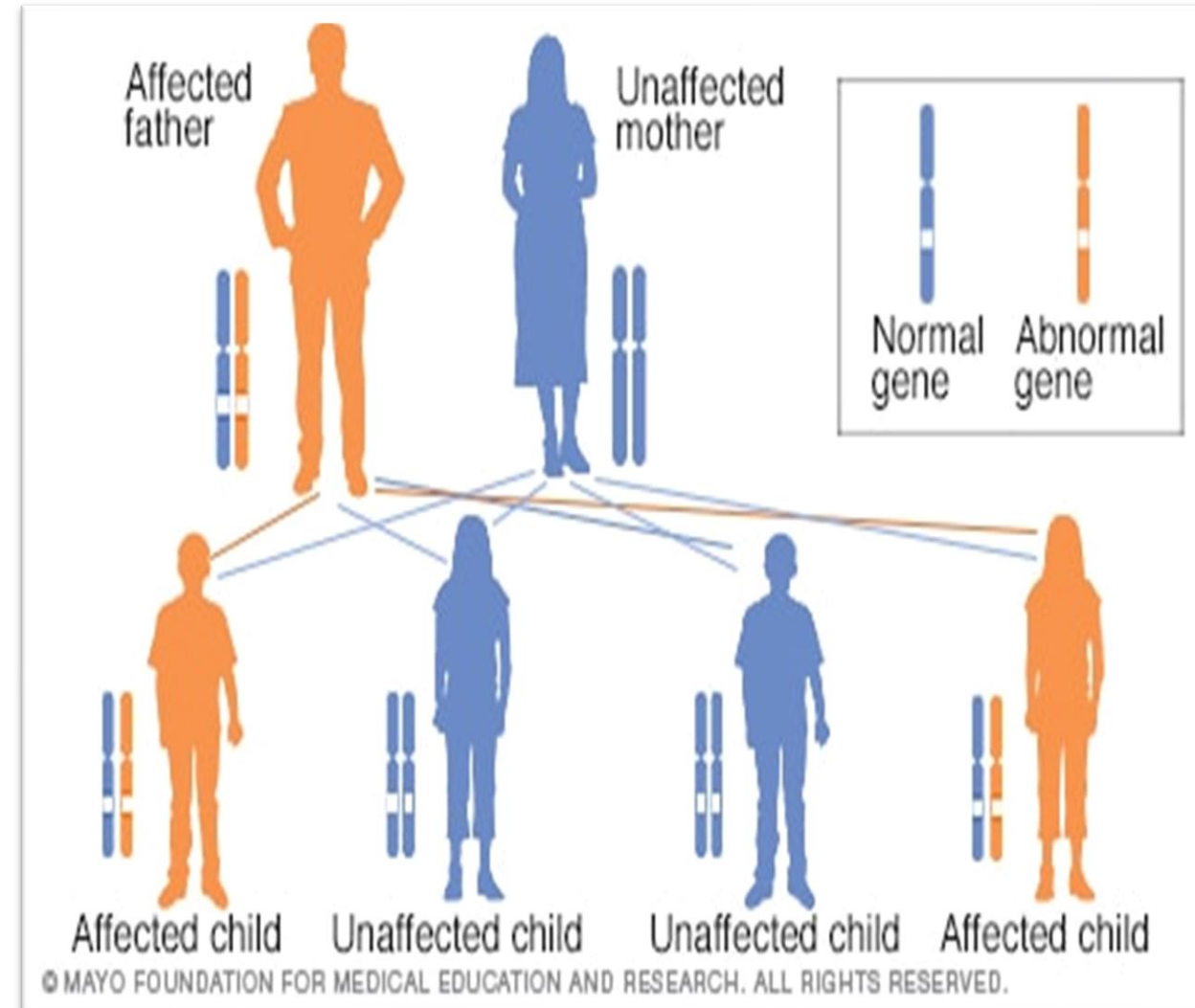


# Patterns (Mode) of inheritance

- The pattern of inheritance describe how a disease is transmitted in families, determining the risk of inheritance for offspring.
- In general, inheritance patterns for **single gene disorders** are classified based on whether the traits are controlled by genes on **autosomes**, or by genes on **sex chromosomes** **Following one of 4 patterns of inheritance**
  - 1) Autosomal dominant
  - 2) Autosomal recessive
  - 3) Sex/ X linked dominant
  - 4) Sex/ X linked recessive

# Autosomal Dominant Inheritance (AD)

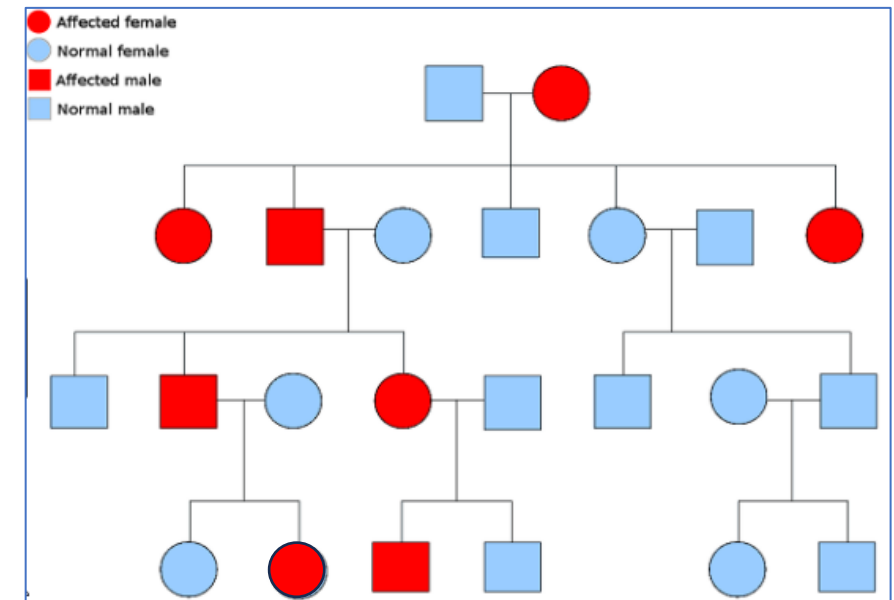
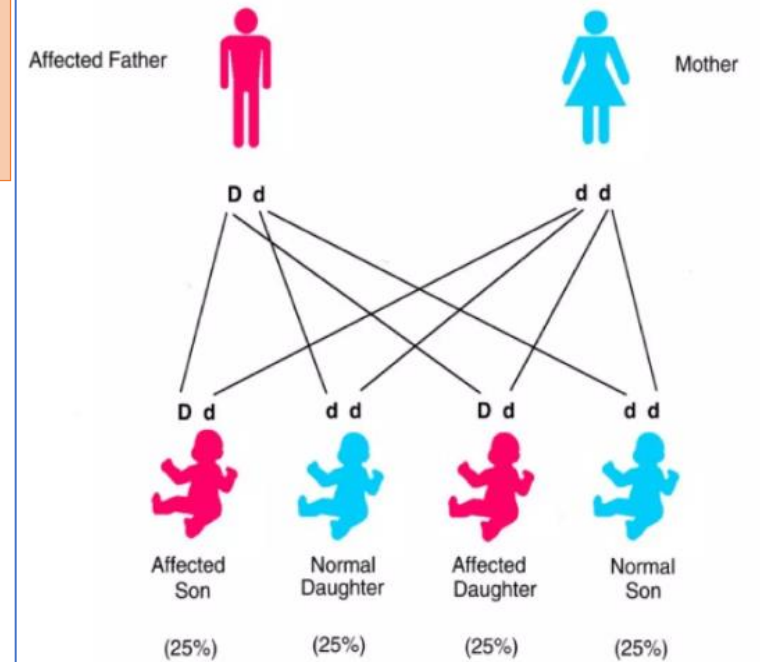
- Only **one copy of a disease allele** is necessary to affect the individual
- =It manifests in heterozygous state (i.e. a person having both normal and mutant alleles)
- With each pregnancy, there is a one in two (**50%**) chance the offspring will inherit the disease allele.



# Autosomal Dominant Inheritance (AD)

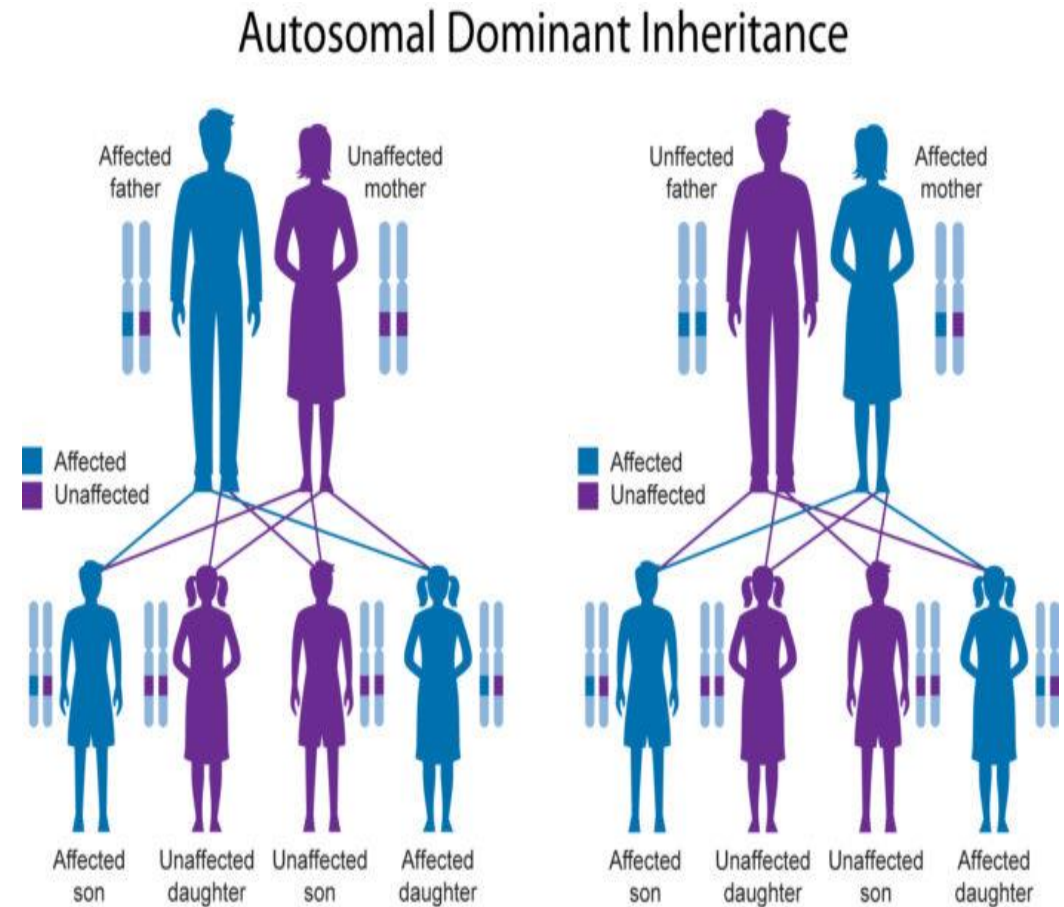
- Male and female are **equally affected**
- The trait appears in **every generation** (Affected people usually have at least one parent carries the diseased allele)
- **Examples:** Huntington's disease, Achondroplasia, Neurofibromatosis

## Autosomal dominant



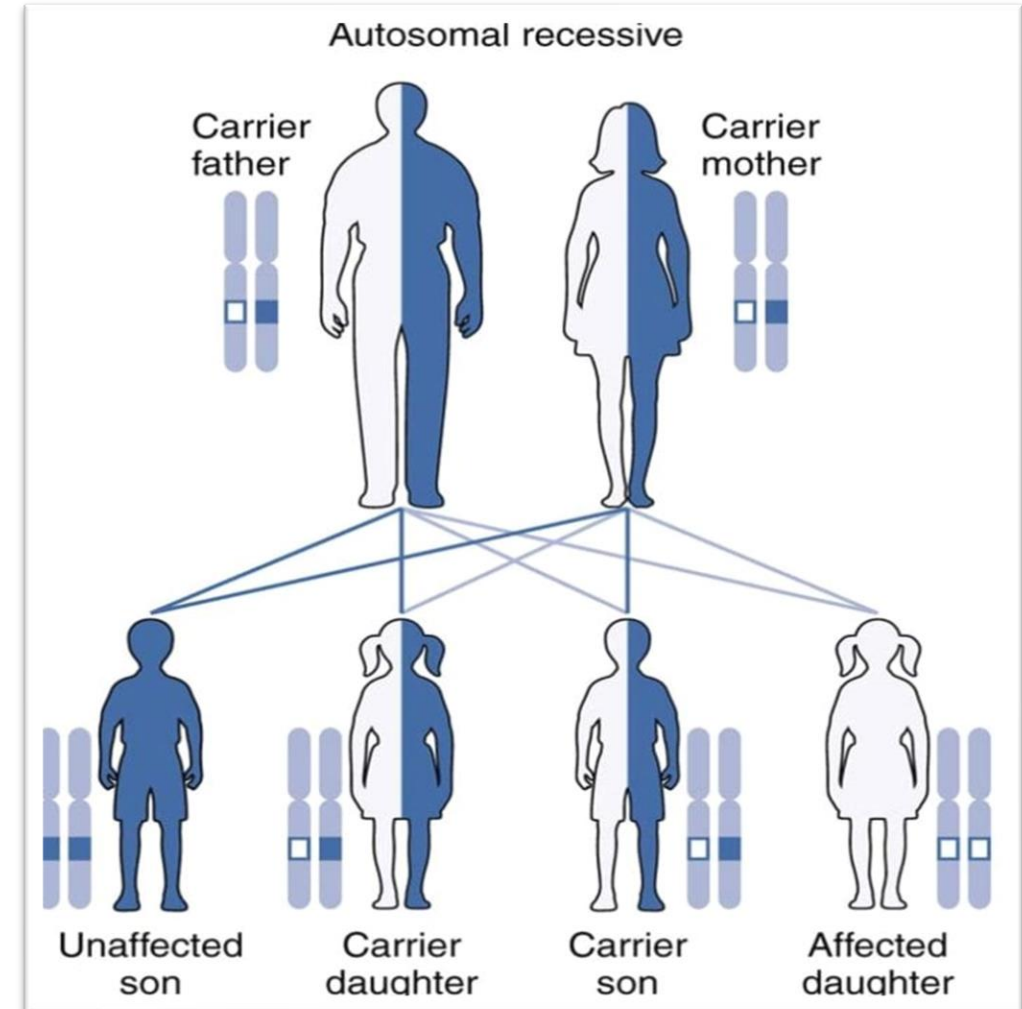
# Autosomal Dominant Inheritance (AD)

- All types of transmission between both sexes could occur i.e. **Male parent can transmit the diseased allele to male or female offspring and the same for female parent (important to differentiate between AD & sex-linked disorders)**



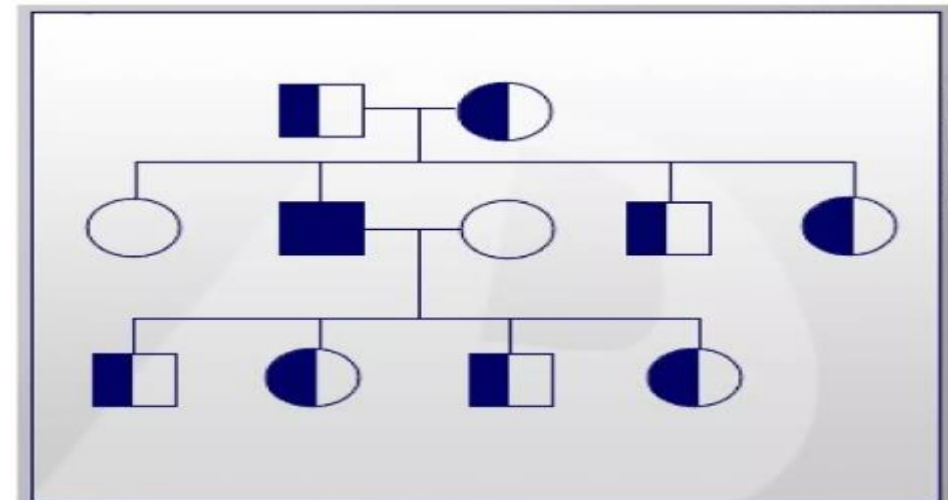
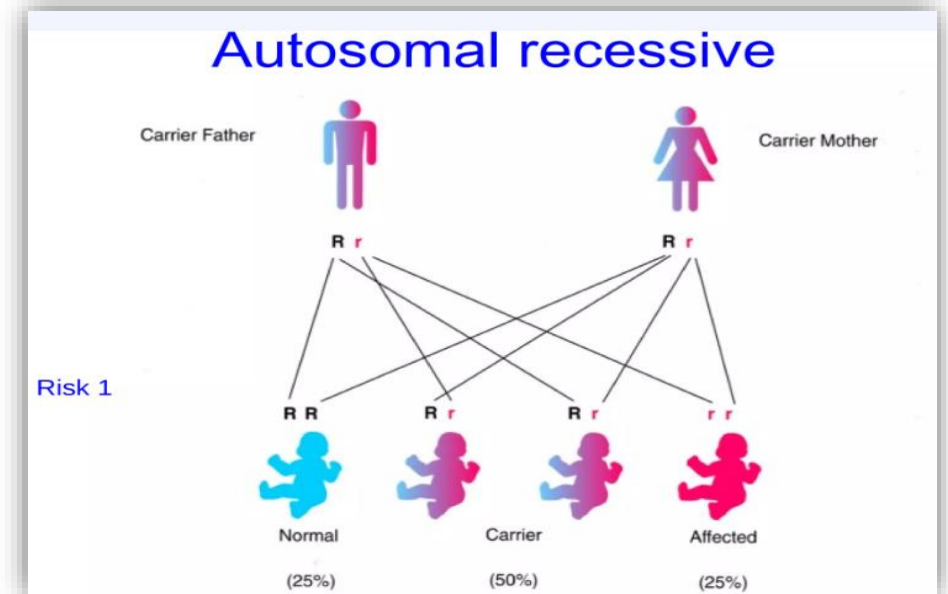
# Autosomal Recessive Inheritance (AR)

- Male and female are **equally affected**
- Manifest only in individuals **homozygous** for the mutant allele (Two copies of the altered gene, one from each parent, are required to cause the disease).
- Individuals heterozygous for the mutant allele are apparently healthy (show no features of the disorder), i.e. carriers



# Autosomal Recessive Inheritance (AR)

- Parents are typically unaffected carriers.
- The **consanguinity increases the risk** (that both parents carry the same recessive allele)
- With each pregnancy of carrier parents:
  - **25%** of the offspring : **affected**
  - **50%** : a carrier.
  - **25%** : Normal
- Example: **Sickle cell anemia**. Albinism, Cystic fibrosis.



# Sex-linked inheritance = X-linked inheritance

- ❑ Sex-linked inheritance are determined by genes located on X chromosome
- ❑ In humans, the X-chromosome contains hundreds of genes that are unrelated to sex determination that required for both males and females & have no counterpart on the Y chromosome
- ❑ A male receives ALL of his X-linked genes from his mother while a female receives her X-linked genes from both parents.

# X-Linked Recessive inheritance (XR)

- Since a **female** has two X chromosomes, **two copies** of a disease allele on the X chromosome are required for a female **to be affected**.
- A **male** with **mutant allele on his single X chromosome**, is said to be **hemizygous** for that allele. They need **one copy** of an X-linked recessive disease allele to be **affected**.
- **Females** only have **one copy** of the disease allele, are unaffected carriers.

# XR-inheritance Genetic risk

## ❑ For a carrier (unaffected) female:

- **Sons: (50%)** chance **affected**.
- **Daughters: (50%)** chance **carriers**.

## ❑ Affected males transmit the disease allele to:

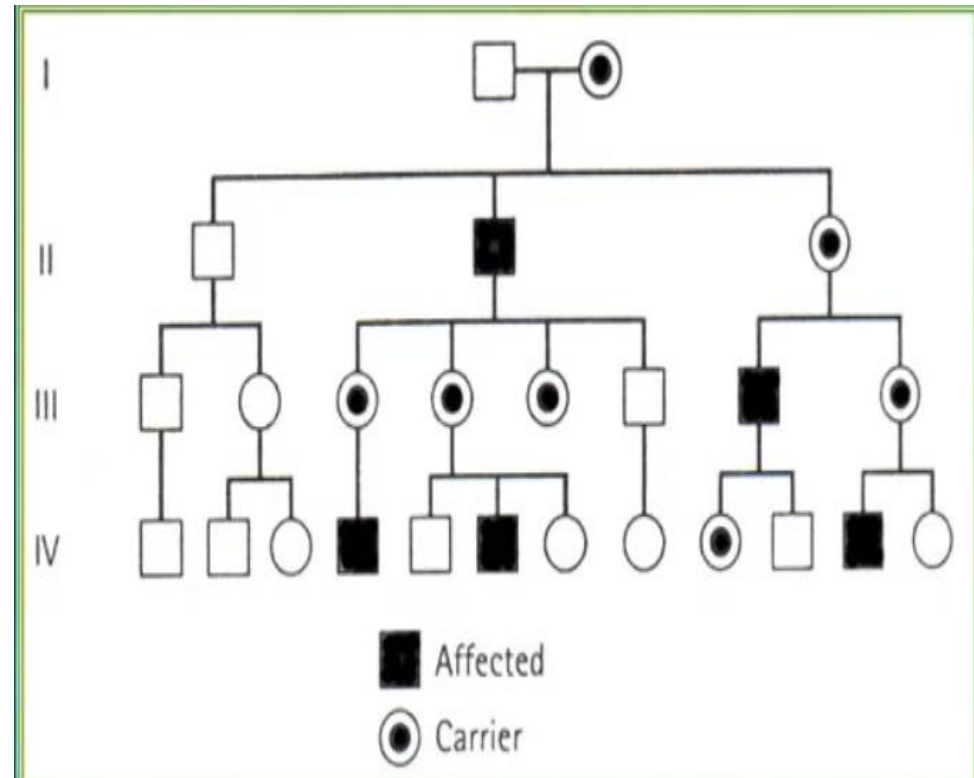
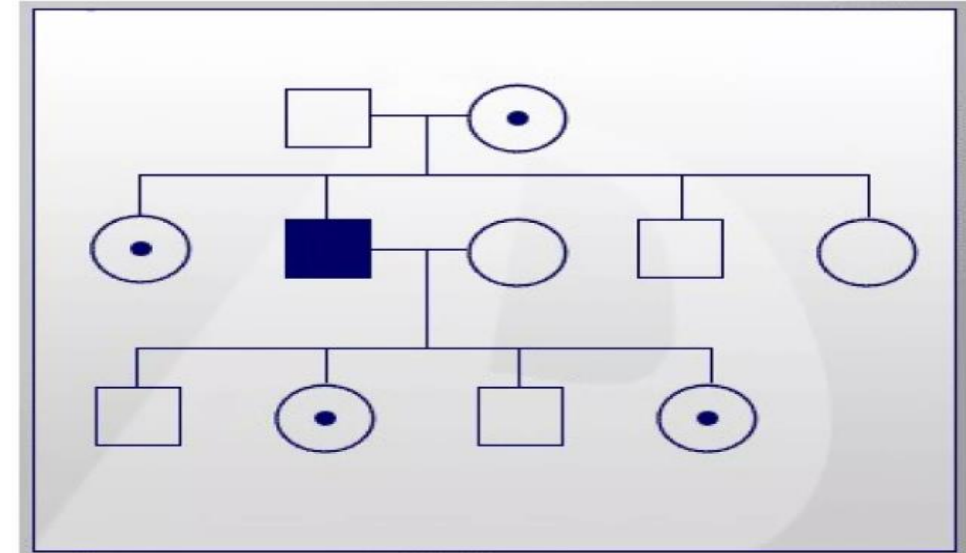
- **All of their daughters**, who are usually **carriers**
- **None of their sons** (**no male-to-male (father-to-son) transmission**, since sons will inherit the Y rather than the X chromosome).

## ❑ Affected female:

- **All of their sons: affected**.
- **All of their daughters: unaffected carriers**.

○ **Examples:** Hemophilia, Colorblindness.

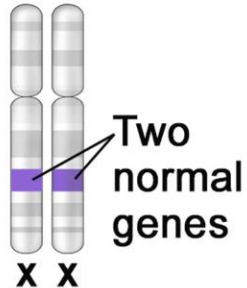
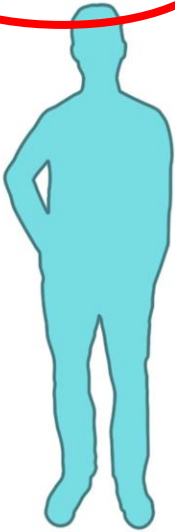
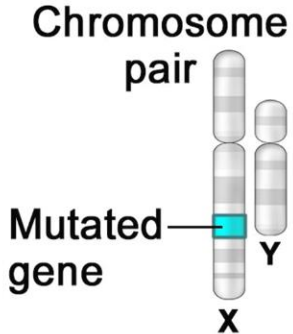
## X-Linked recessive inheritance



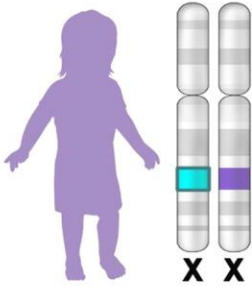
# X-Linked Recessive Inheritance

Affected father

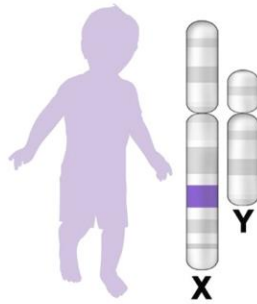
Unaffected mother



Unaffected daughter (Carrier)

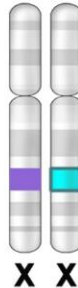
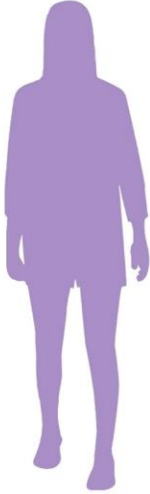


Unaffected son

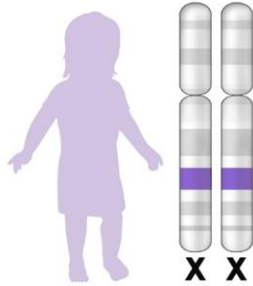


Unaffected father

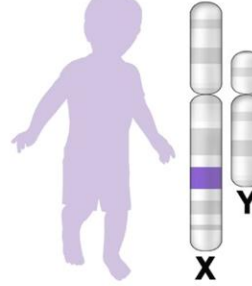
Unaffected mother (Carrier)



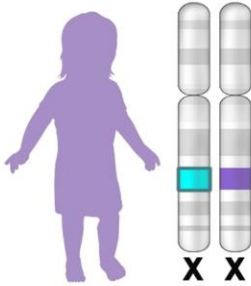
Unaffected daughter



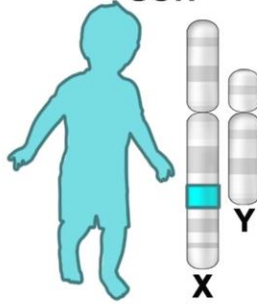
Unaffected son



Unaffected daughter (Carrier)



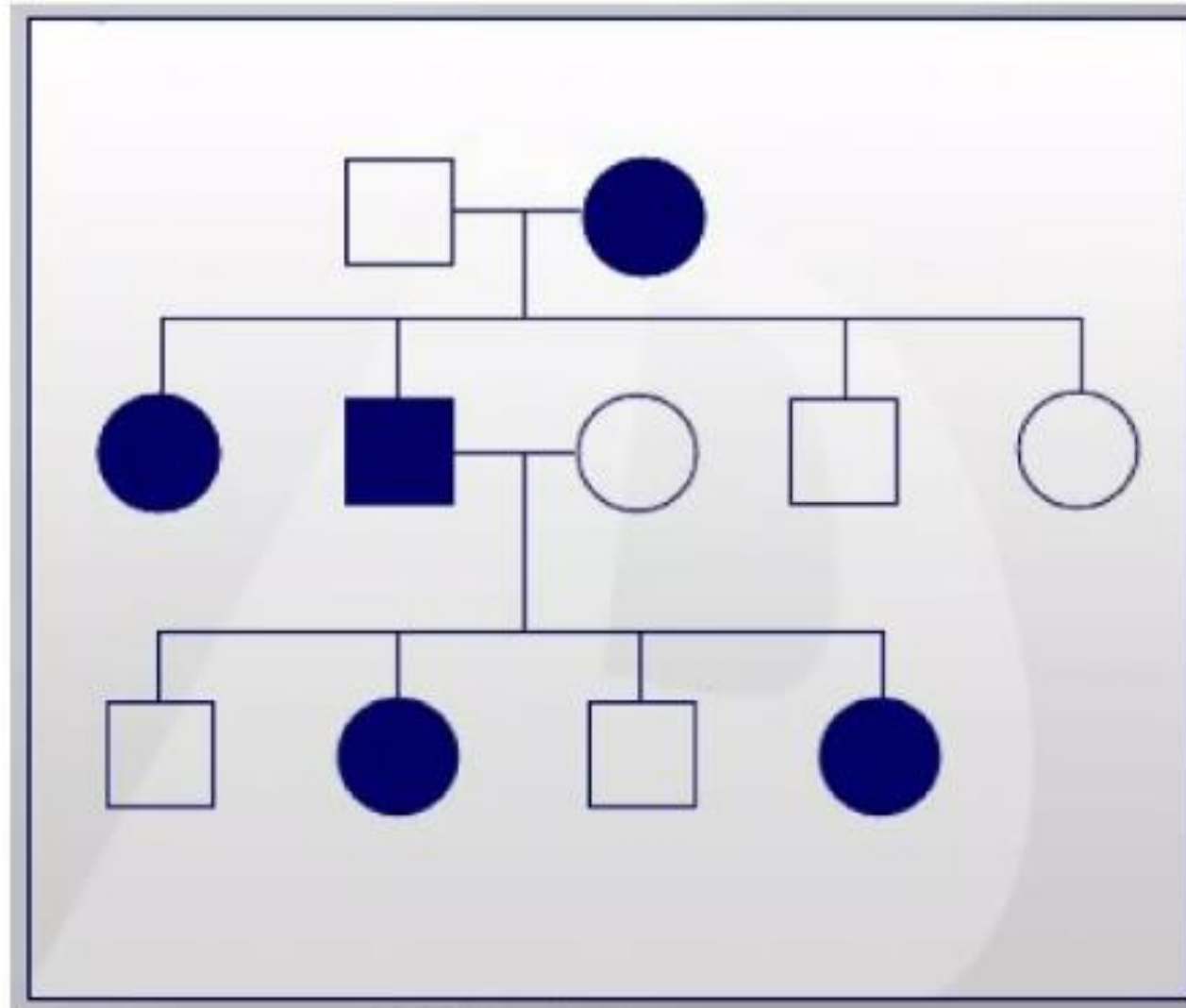
Affected son



# X-Linked Dominant inheritance (XD)

- Only **one copy** of a disease allele on the X chromosome is required for an individual to be **susceptible to the disease**.
- **Both males and females** can be affected, although **males may be more severely affected because they only carry one copy** of genes found on the X chromosome.
- When a **female** is affected, each pregnancy will have a **(50%)** chance for both male and female offspring to inherit the disease allele.
- When a male is affected, all **his daughters will be affected**, but none of his sons will be affected.

# X-Linked Dominant inheritance (XD)



A top-down view of a spiral-bound notebook with a white cover, lying on a light-colored wooden surface. The notebook is open to a page with the words "THANK YOU!" printed in a large, bold, black sans-serif font. The exclamation point in "YOU!" is a vibrant red. To the left of the notebook, a pair of gold-rimmed glasses with black temples is placed. To the right, a sleek black and silver ballpoint pen lies horizontally. In the bottom right corner, a portion of a black circular object, possibly a magnifying glass or a lens, is visible. On the left side of the notebook, a small, light-colored ceramic pot holds a green succulent with thick, pointed leaves. The entire scene is set against a dark blue background that frames the image.