

Pedigrees

The Royal Hemophiliacs

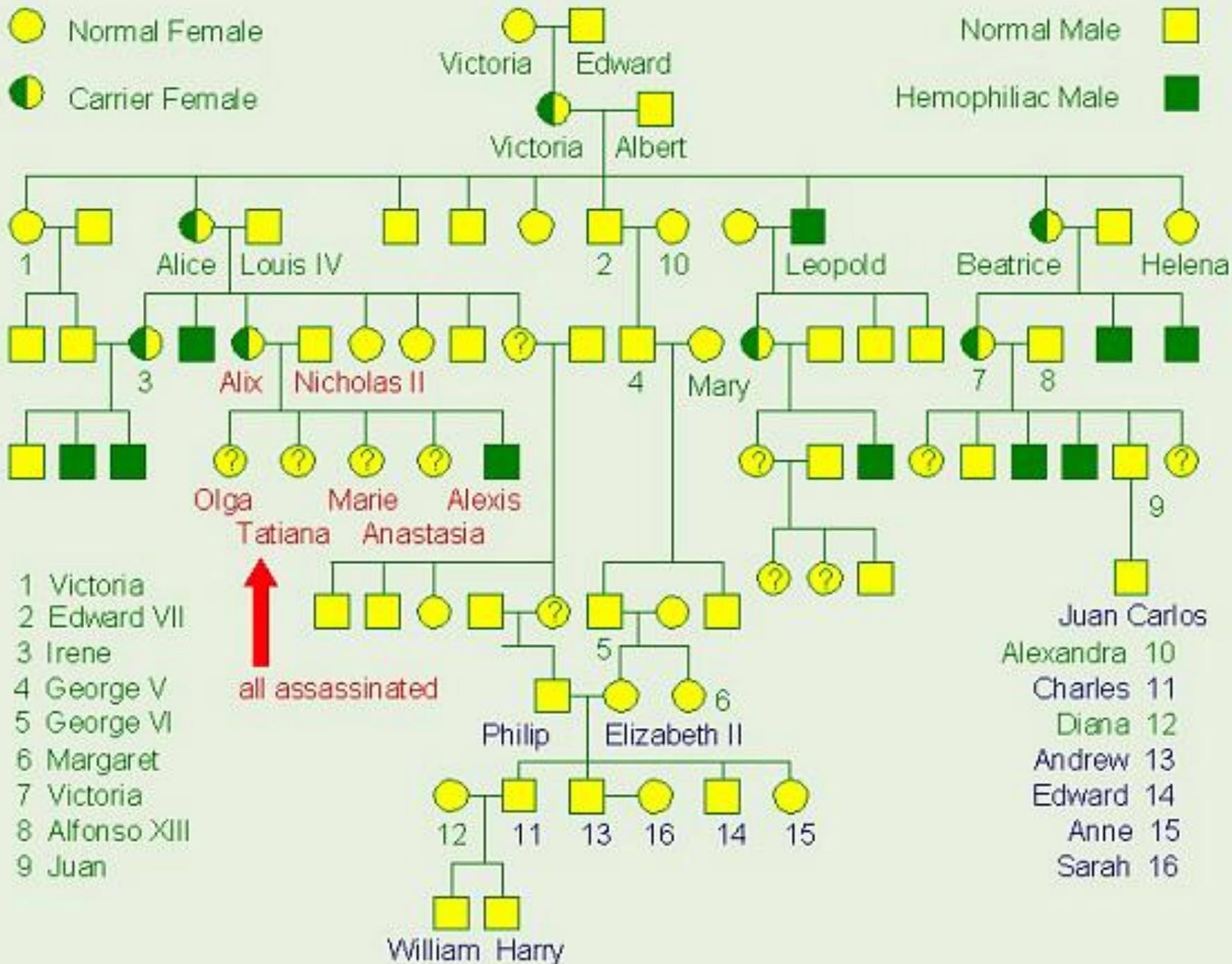


Normal Female

Carrier Female

Normal Male

Hemophiliac Male



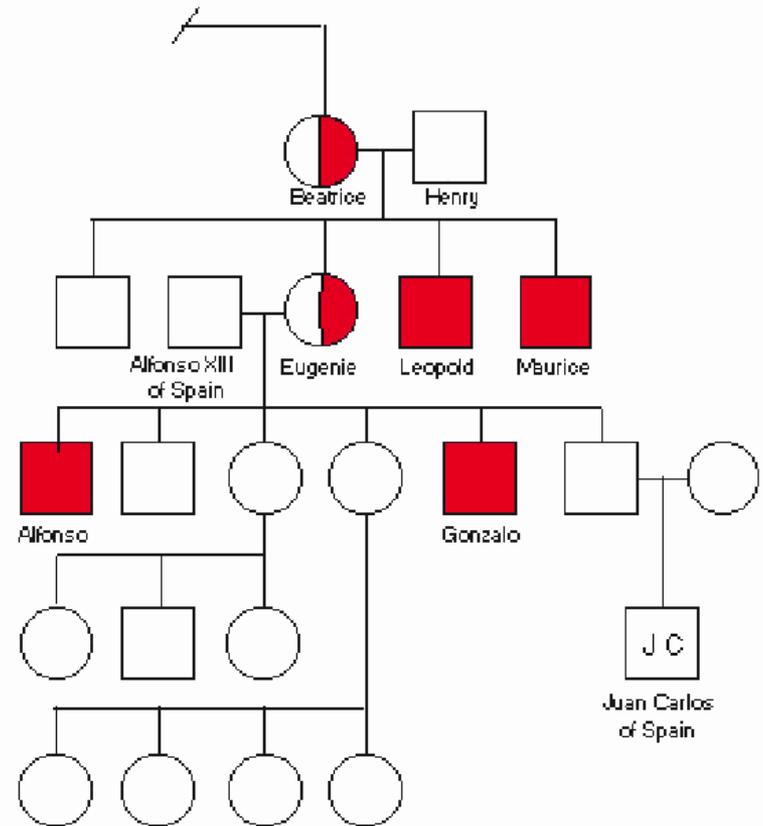
- 1 Victoria
- 2 Edward VII
- 3 Irene
- 4 George V
- 5 George VI
- 6 Margaret
- 7 Victoria
- 8 Alfonso XIII
- 9 Juan

all assassinated

- Juan Carlos
- Alexandra 10
- Charles 11
- Diana 12
- Andrew 13
- Edward 14
- Anne 15
- Sarah 16

William Harry

- Now for the Spanish connection: Victoria's youngest child, Beatrice, gave birth to one daughter, one normal son, and two hemophiliac sons. Looking at the pedigree of the royal family, identify which of Beatrice's children received the hemophiliac gene; why can you make this conclusion?
- Notice that Beatrice's daughter, Eugenie, married King Alfonso XIII of Spain and had six children, one of whom was the father of Juan Carlos, the current King of Spain. Would you predict that Juan Carlos was normal, a carrier, or a hemophiliac?



The Basics of a Pedigree

- Each person is represented by a symbol
- Birth order is from left to right (oldest → youngest)

What symbols are used?



Normal male



Affected male



Normal female

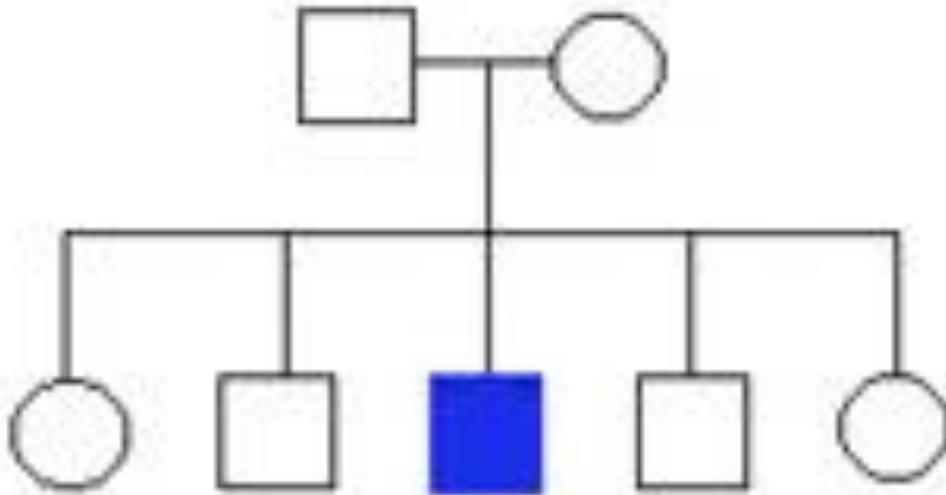


Affected female

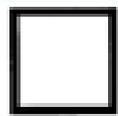


Marriage

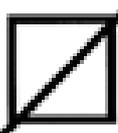
- Here we see a family with 5 children.
- The pedigree chart shows that the second son (third child) is affected by the condition.



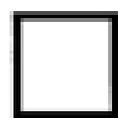
Eldest child ↔ Youngest child

 Unaffected male

 Unaffected female

 Deceased male

 Deceased female

 Propositus

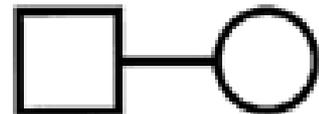
 Affected male

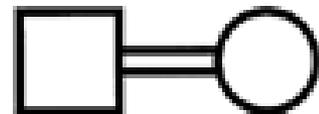
 Affected female

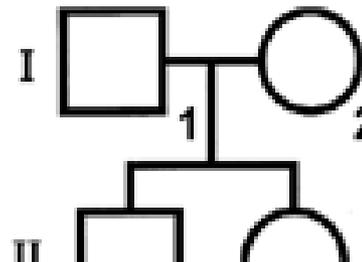
 Carrier for X-linked condition

 Sex unspecified

 Abortion or stillbirth,
sex unspecified

 Mating

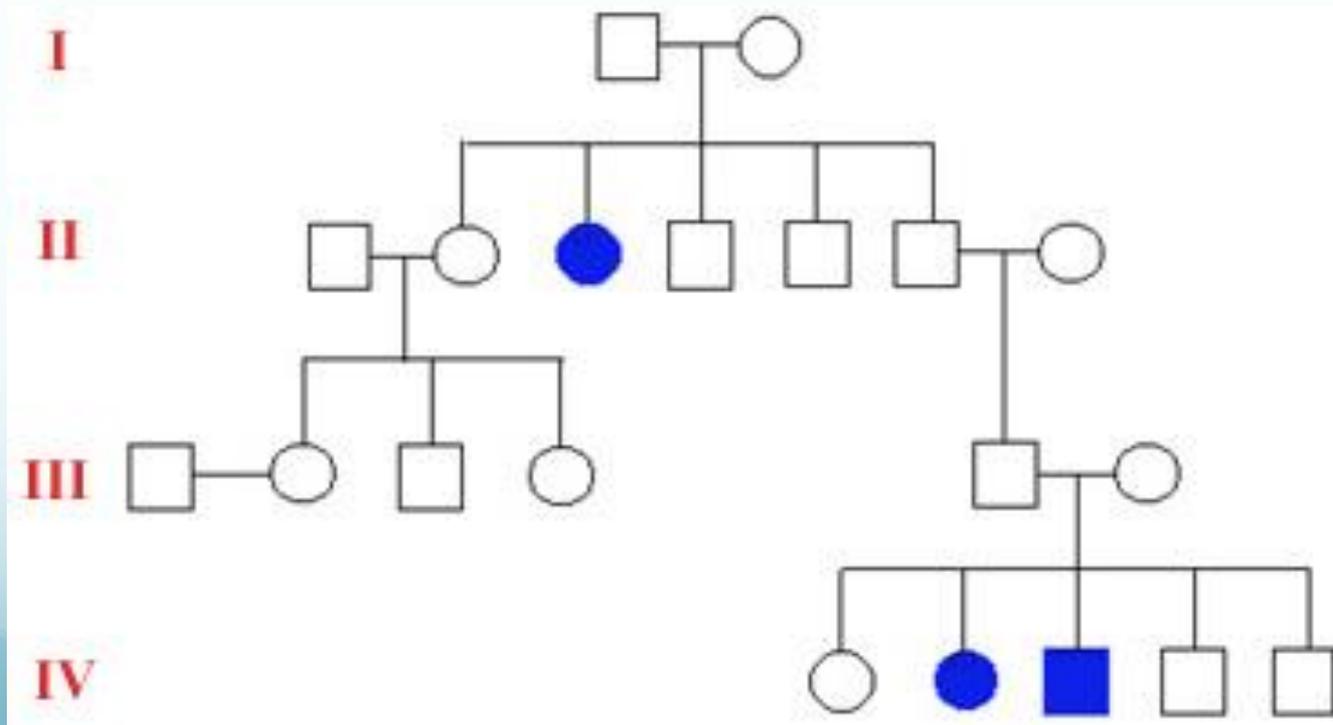
 Consanguineous mating

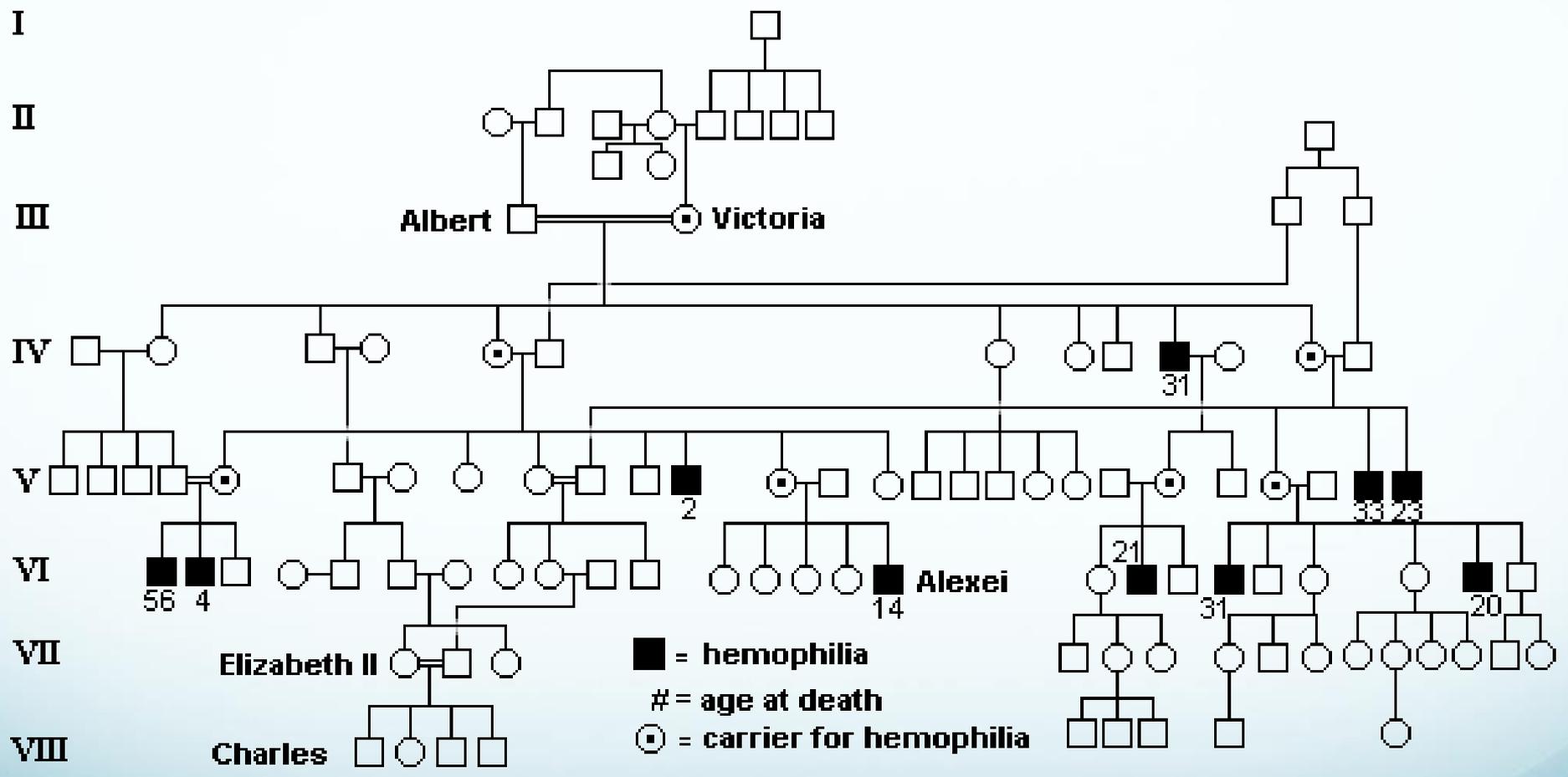
 Parents with offspring:
Roman numerals = generation
Arabic numerals = individual
(propositus is II-1)

 Non-identical twins

 Identical twins

- Below is a pedigree of 4 generations – 20 people.
- Generations are identified by Roman numerals.
- Individuals in each generation are identified by Roman numerals numbered from the left. Therefore the affected individuals are **II3**, **IV2** and **IV3**.



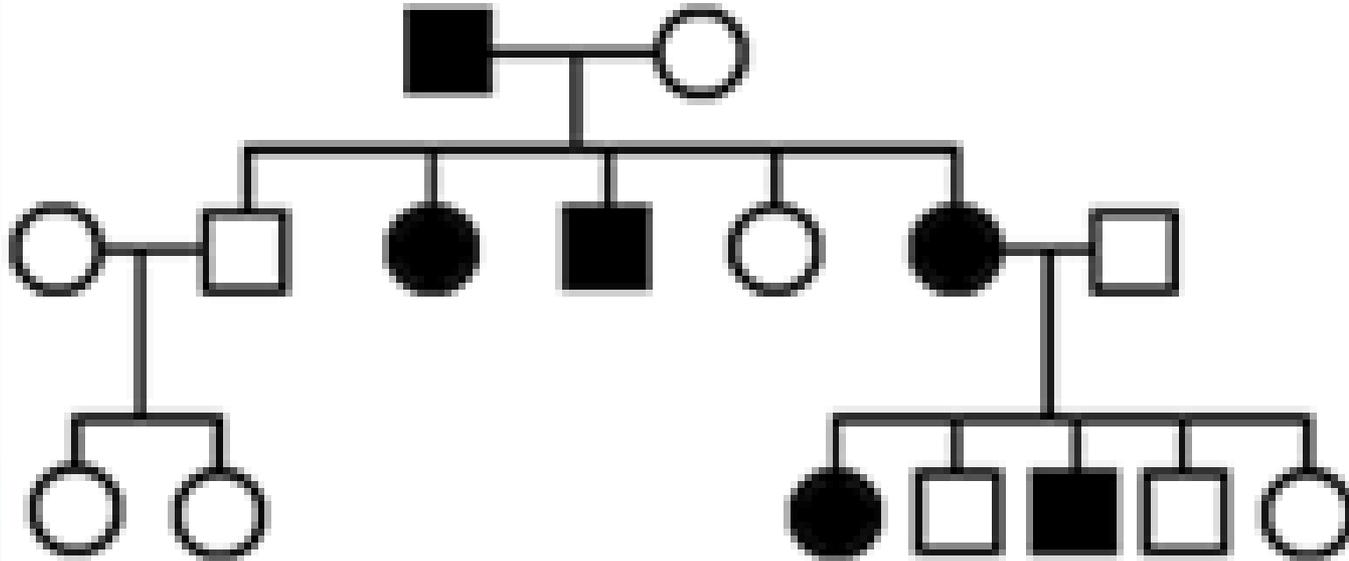


4 Main types of Inheritance

- **Autosomal Dominant**
- **Autosomal Recessive**
- **Sex-Linked Dominant**
- **Sex Linked Recessive**

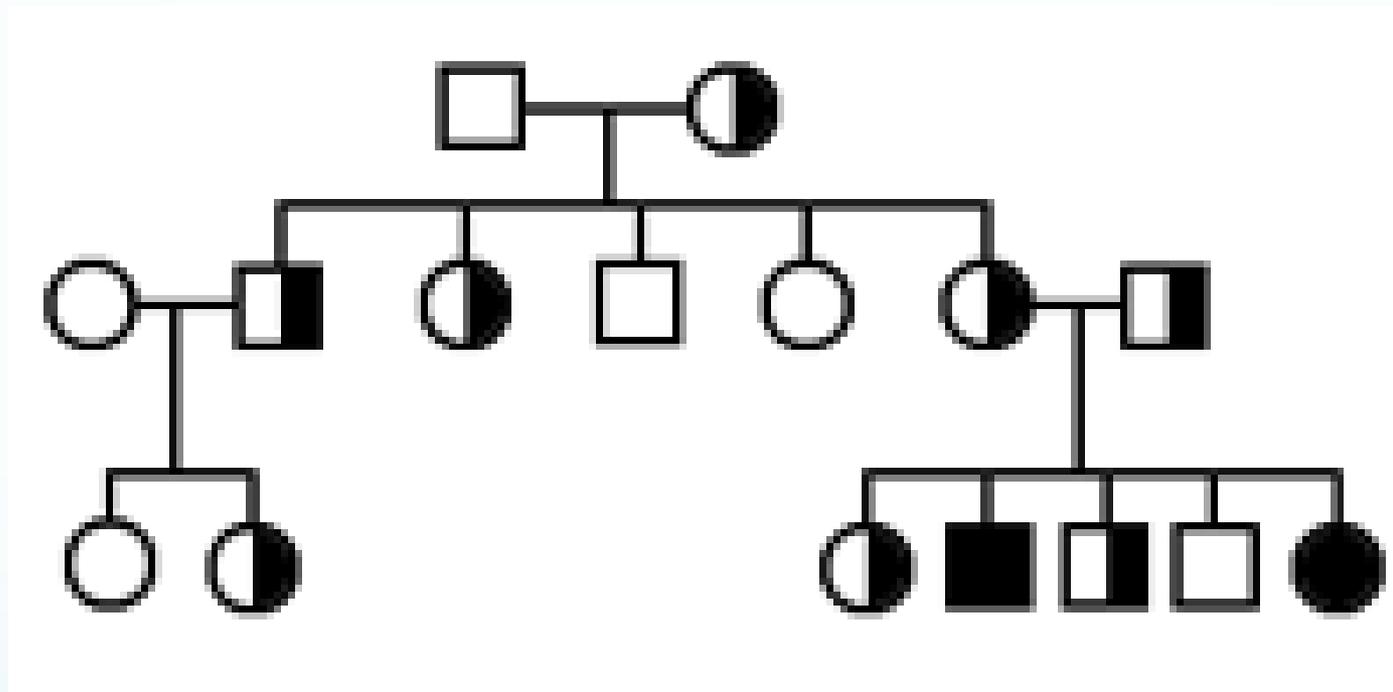
Patterns of Inheritance

Autosomal Dominant



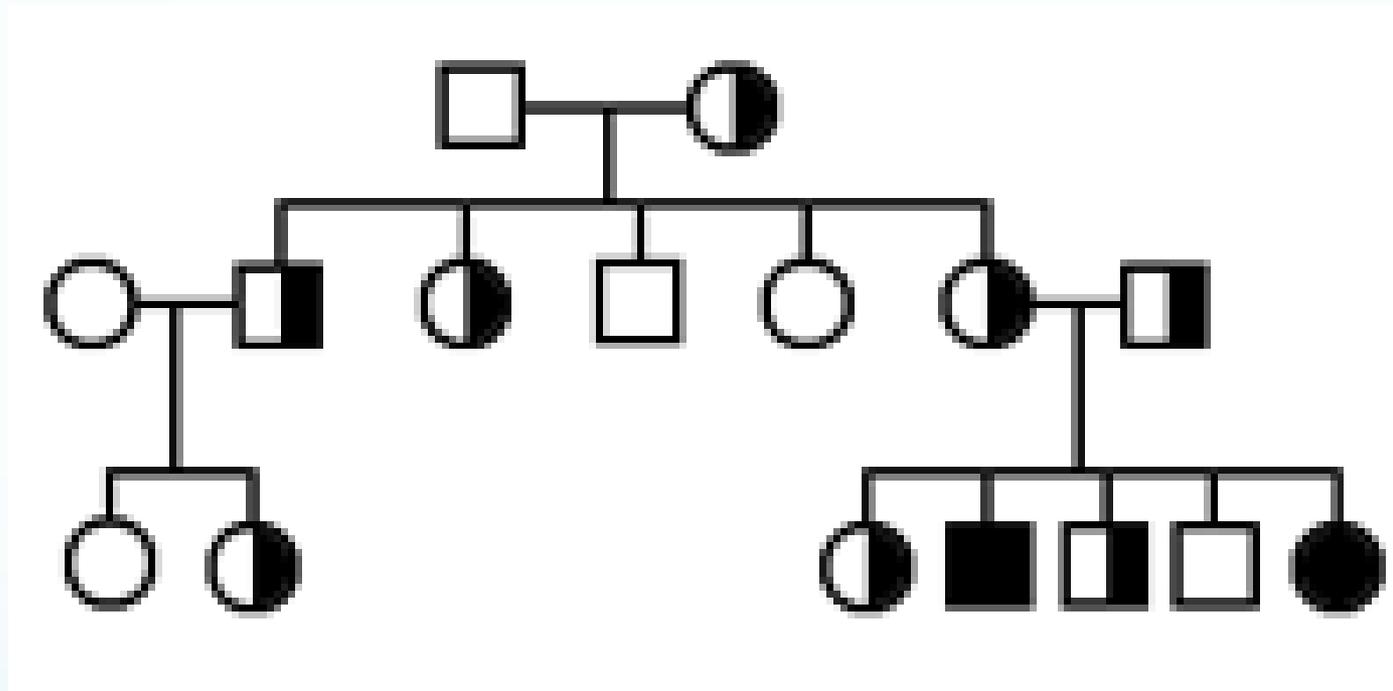
- Affected individuals must have at least one affected allele for the trait to be expressed
- Each offspring has a 50% chance of inheriting the mutant gene if one parent is heterozygous

Autosomal Recessive



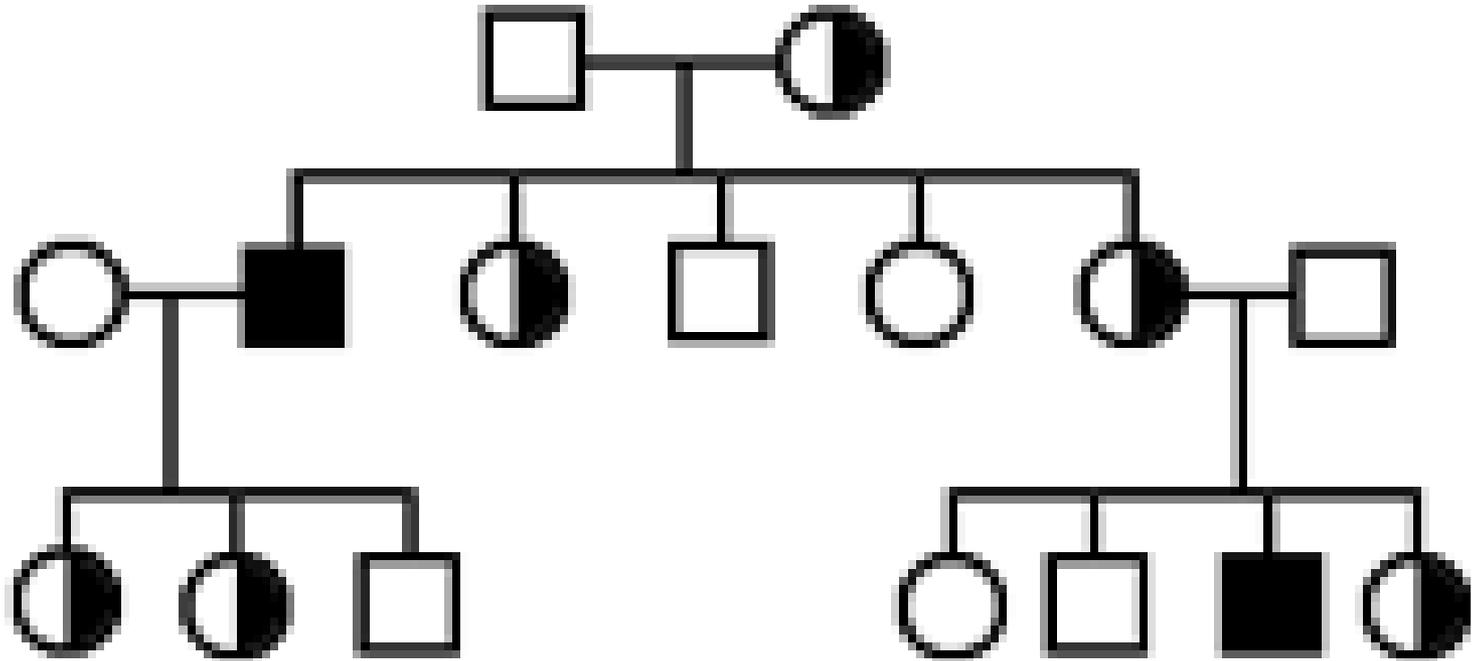
- Affected only when have TWO copies of the mutant gene
- 1 mutant copy/1 normal = carrier
- Females and males are equally affected

Autosomal Recessive



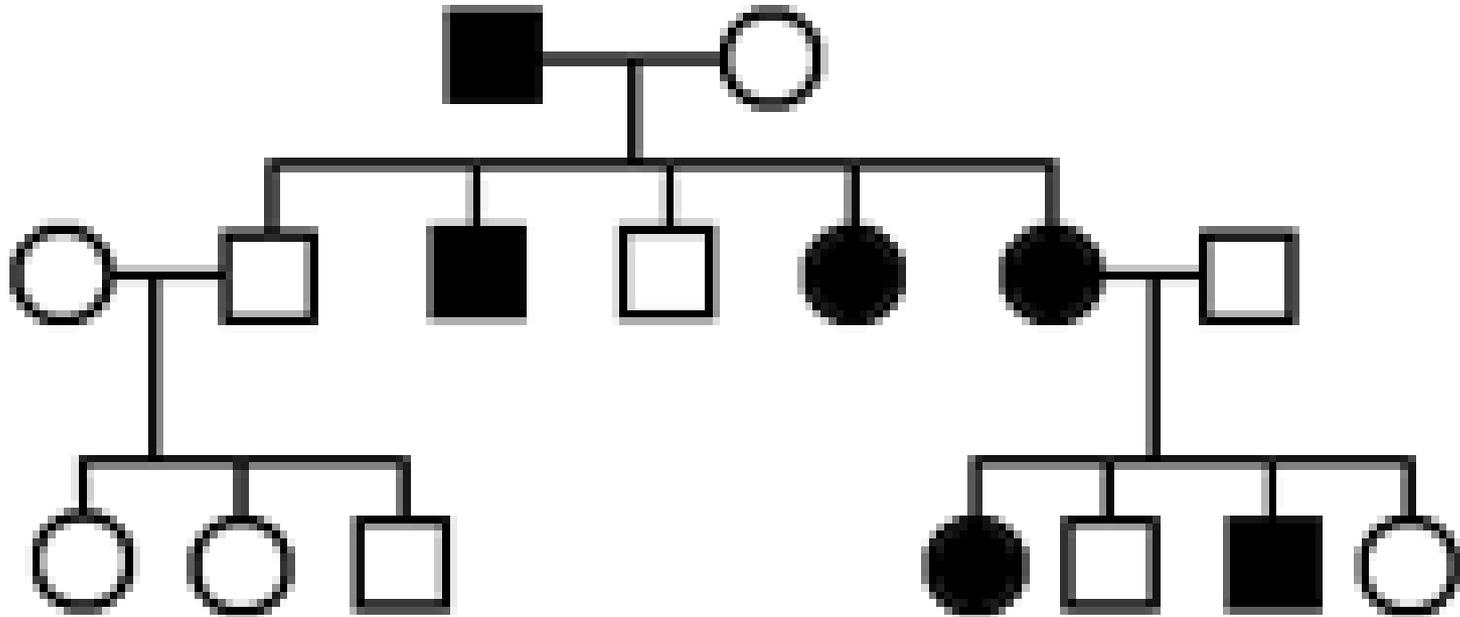
- When 2 carriers mate:
- 25% homozygous unaffected,
- 50% carriers, 25% affected

X-Linked Recessive



- All females who have it are carriers but look normal
- All males who have it are affected
- No father to son transmission but if a father is affected, his daughter will be a carrier

X-linked Dominant



- No father to son transmission
- All of an affected man's daughters will be affected
- 50% of an affected woman's children will be affected

Your Pedigree must have:

- Generation numbers
- Individual numbers
- Affected/Deceased/Carriers marked
- A Key

Individual with multiple diagnoses



Key



High cholesterol



Type II Diabetes