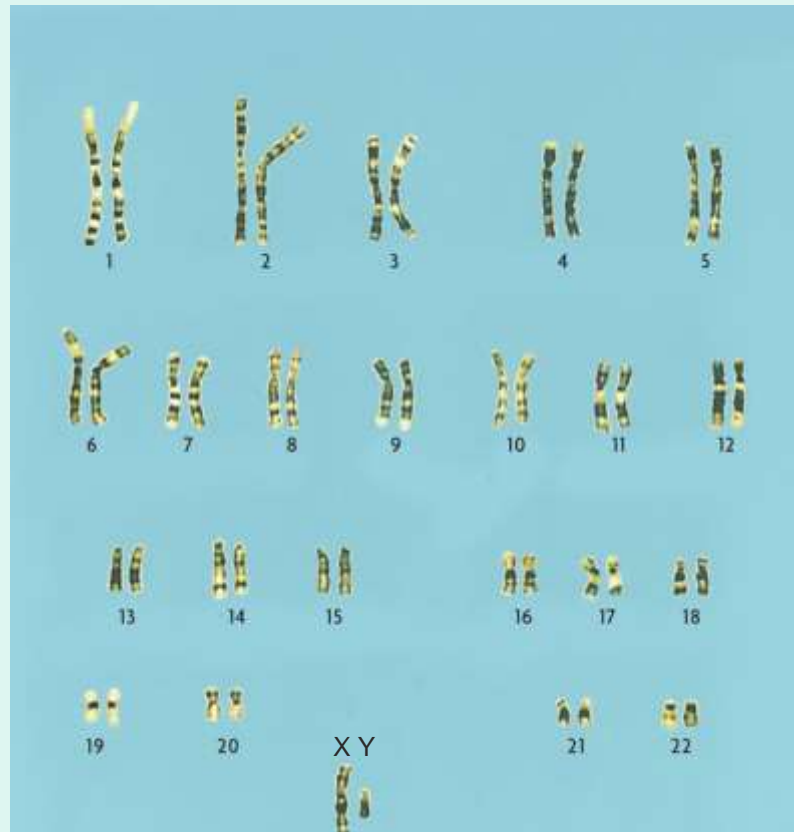


Several methods help map human chromosomes.

- A **karyotype** is a picture of all chromosomes in a cell.

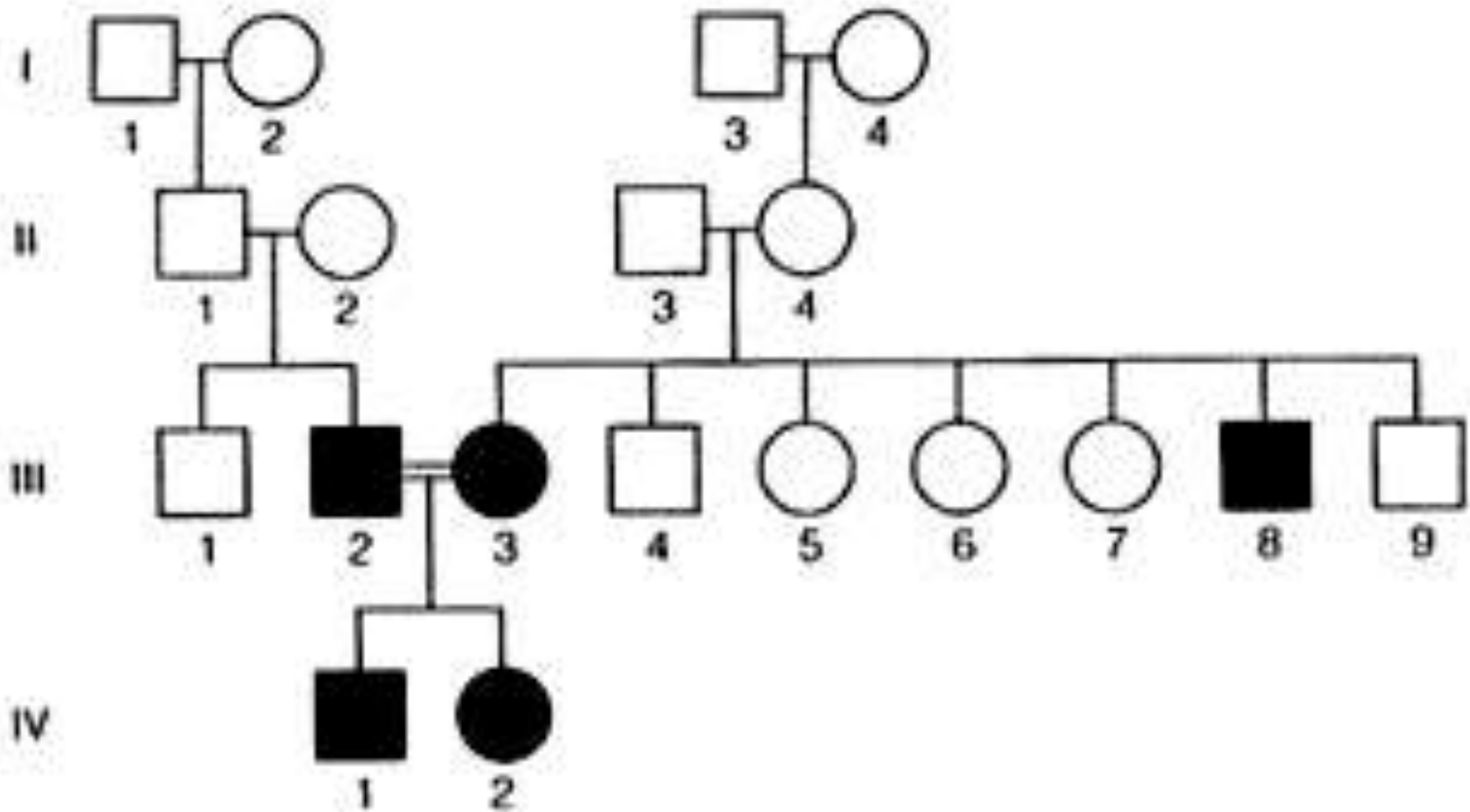


Karyotypes can show changes in chromosomes.

- deletion of part of a chromosome or loss of a chromosome
- large changes in chromosomes
- extra chromosomes or duplication of part of a chromosome



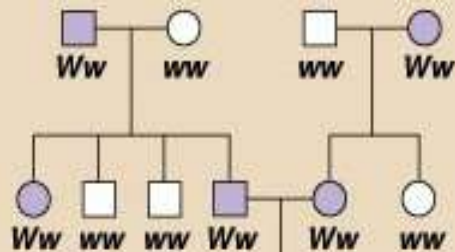
Pedigrees show how a trait is inherited in families



Pedigree analysis

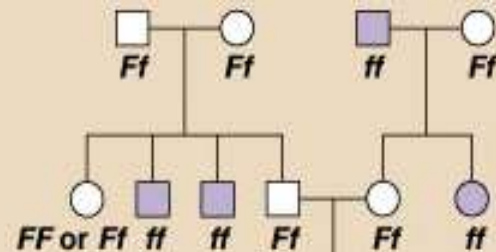
- Pedigree analysis reveals Mendelian patterns in human inheritance
 - data mapped on a family tree

□ = male ○ = female ■ = male w/ trait ● = female w/ trait



First generation
(grandparents)

Second generation
(parents plus aunts
and uncles)



Third generation
(two sisters)



Widow's peak



No widow's peak



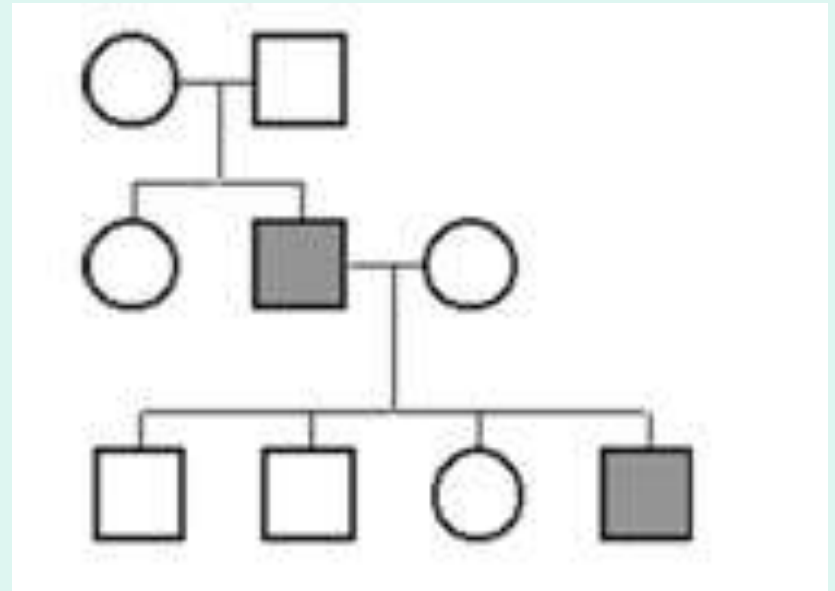
Attached earlobe



Free earlobe

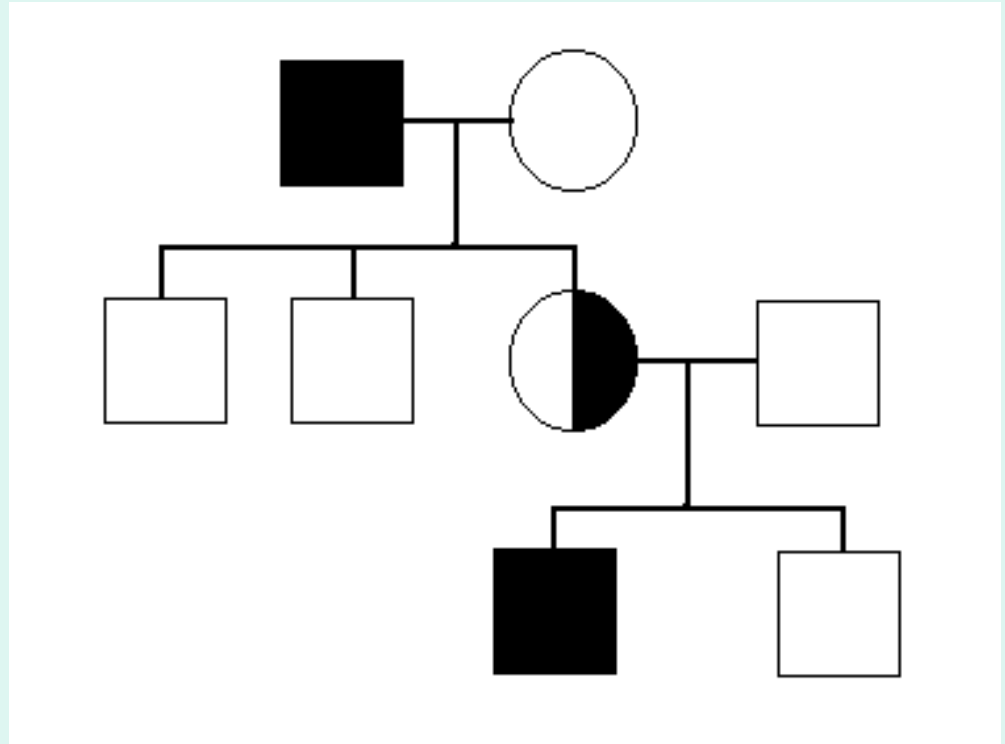
Symbols

- Circle = female
- Square = male
- Normal or without trait = unshaded
- Trait = shaded



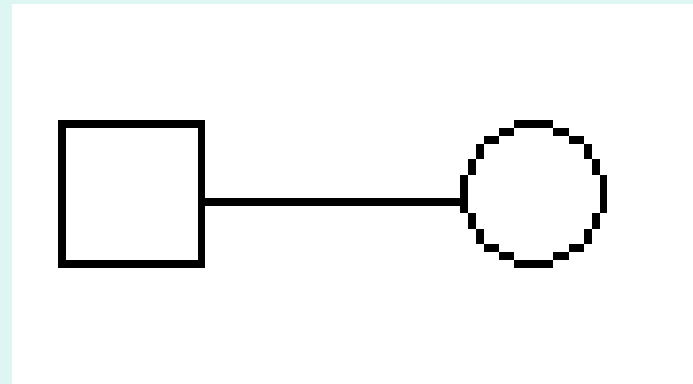
Symbols

- If an individual has one recessive allele for a trait, the person is a **carrier**. This is shown with a half-shaded square or circle.



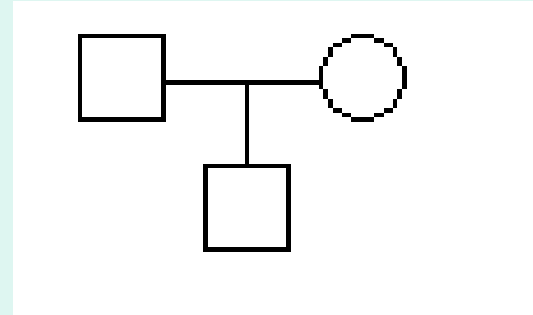
Symbols

- Married couple or couple producing offspring = horizontal line connecting midpoint of circle and square

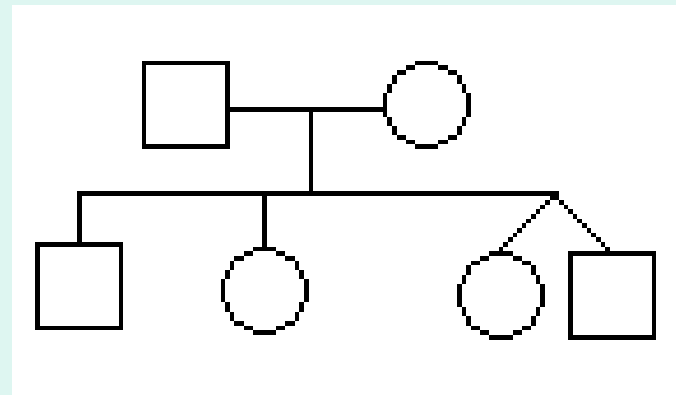


Symbols

- One offspring 



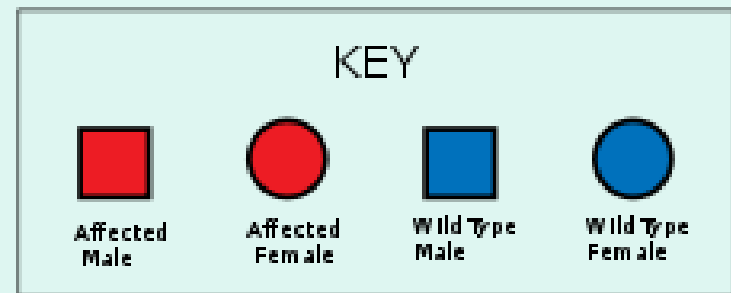
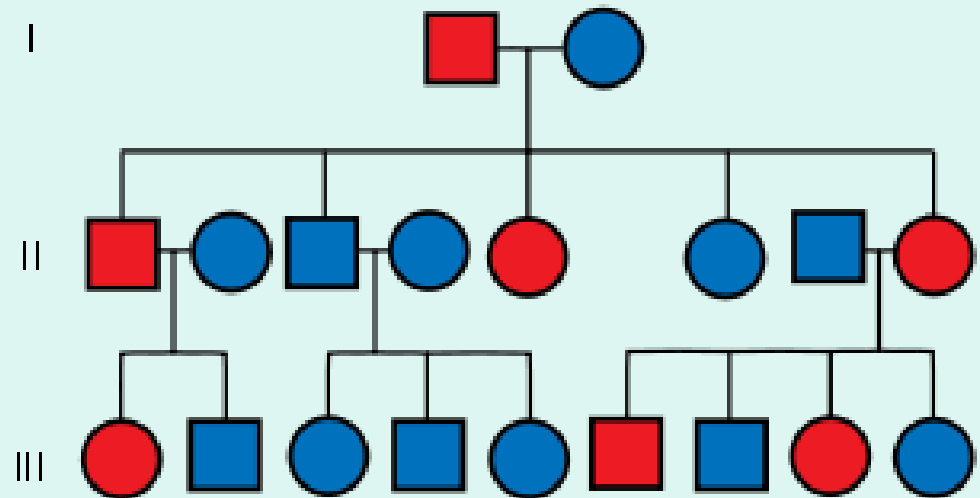
How do you differentiate between a **brother** and **sister** and a **husband** and **wife** on a pedigree?



- Multiple Offspring 

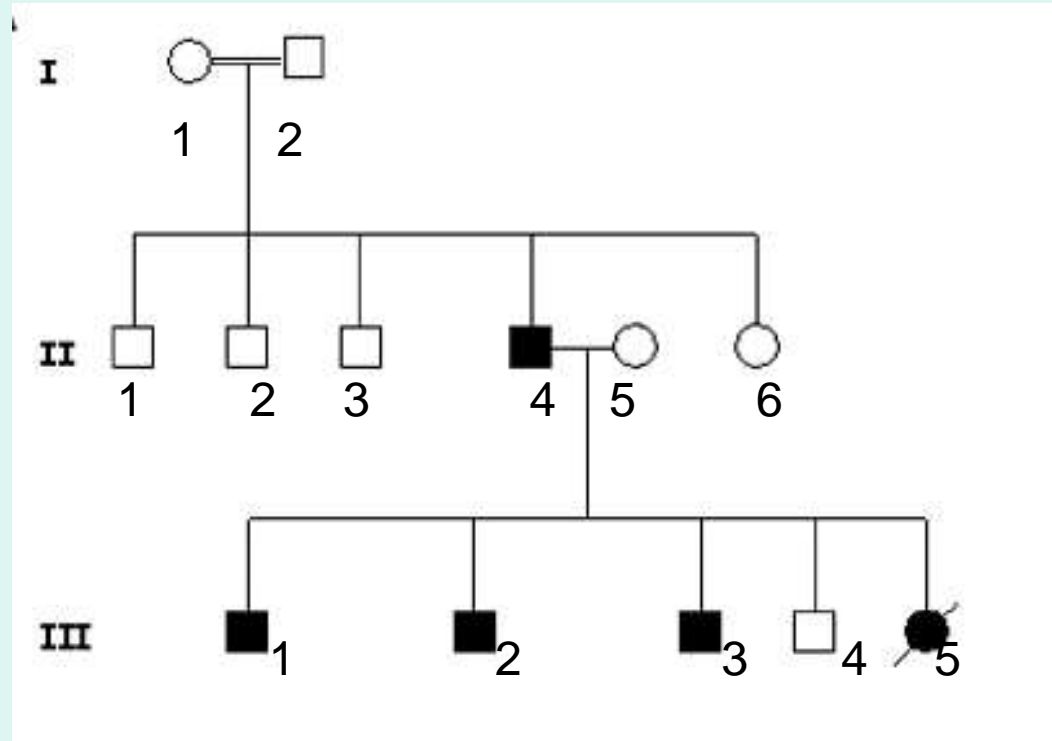
Generations

- Each generation is (often) labeled with a Roman numeral.
- Oldest generation at top of pedigree
- Current generation at bottom



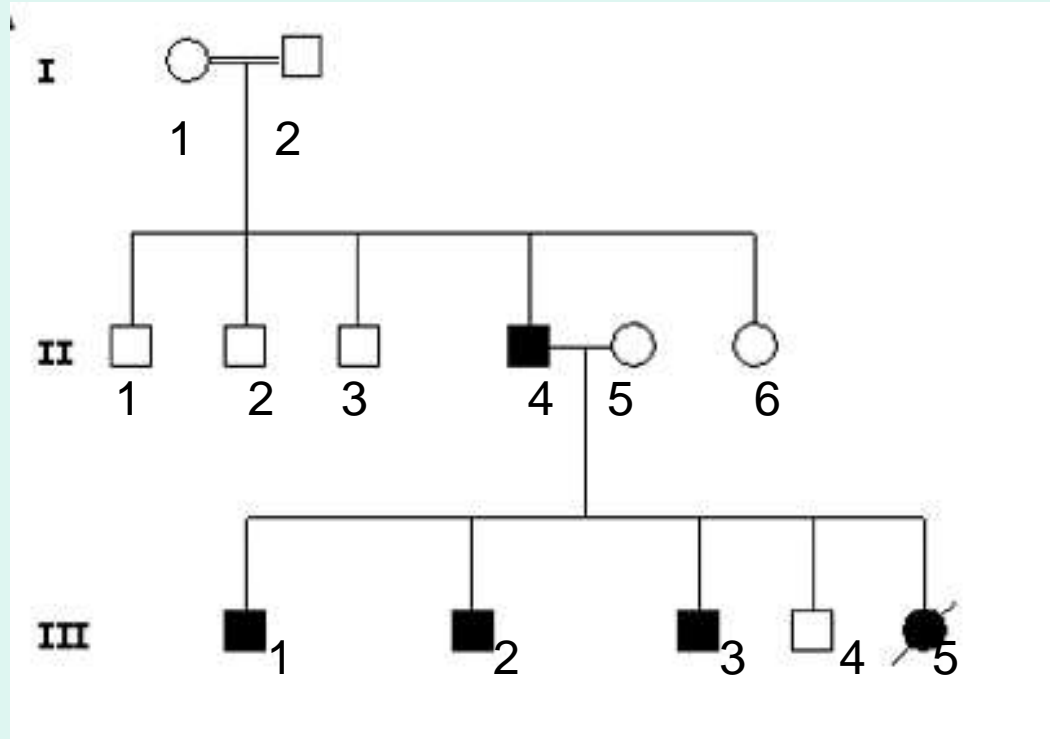
Generations

- Individuals in each generation may be numbered or named.
- Siblings are placed in birth order from left to right.



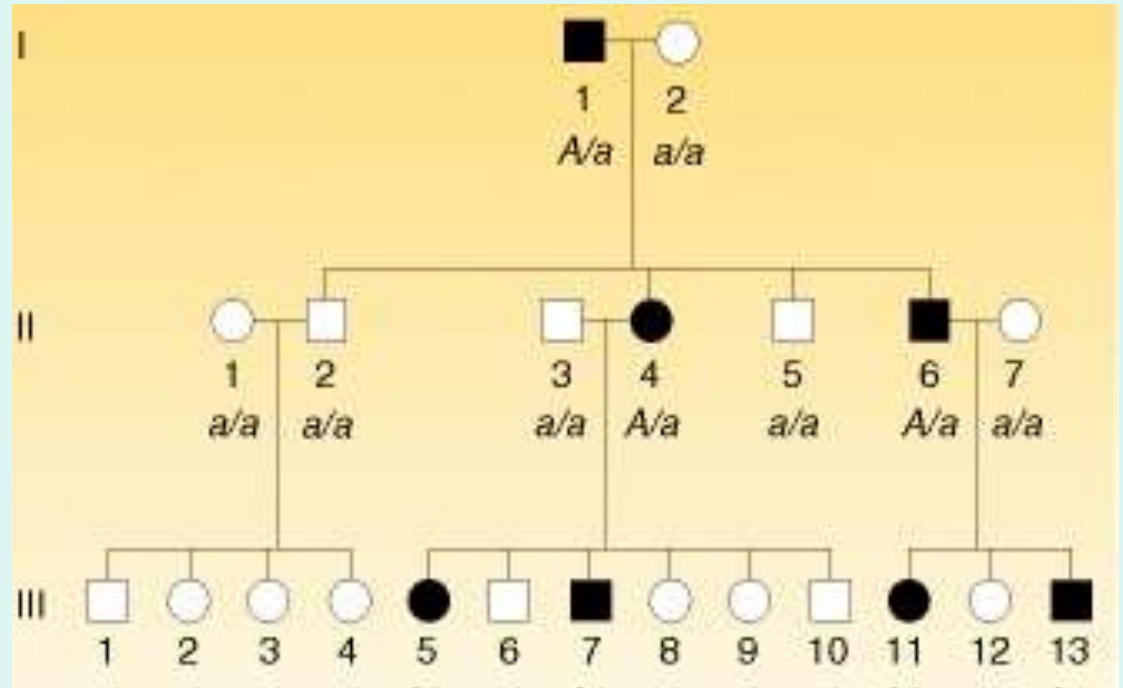
Analysis

- How many offspring were produced by generation 1?
- Number of boys? Girls?
- How many of generation **11** were married with children?
- Deaths are shown with a slash.



Polydactyl Trait

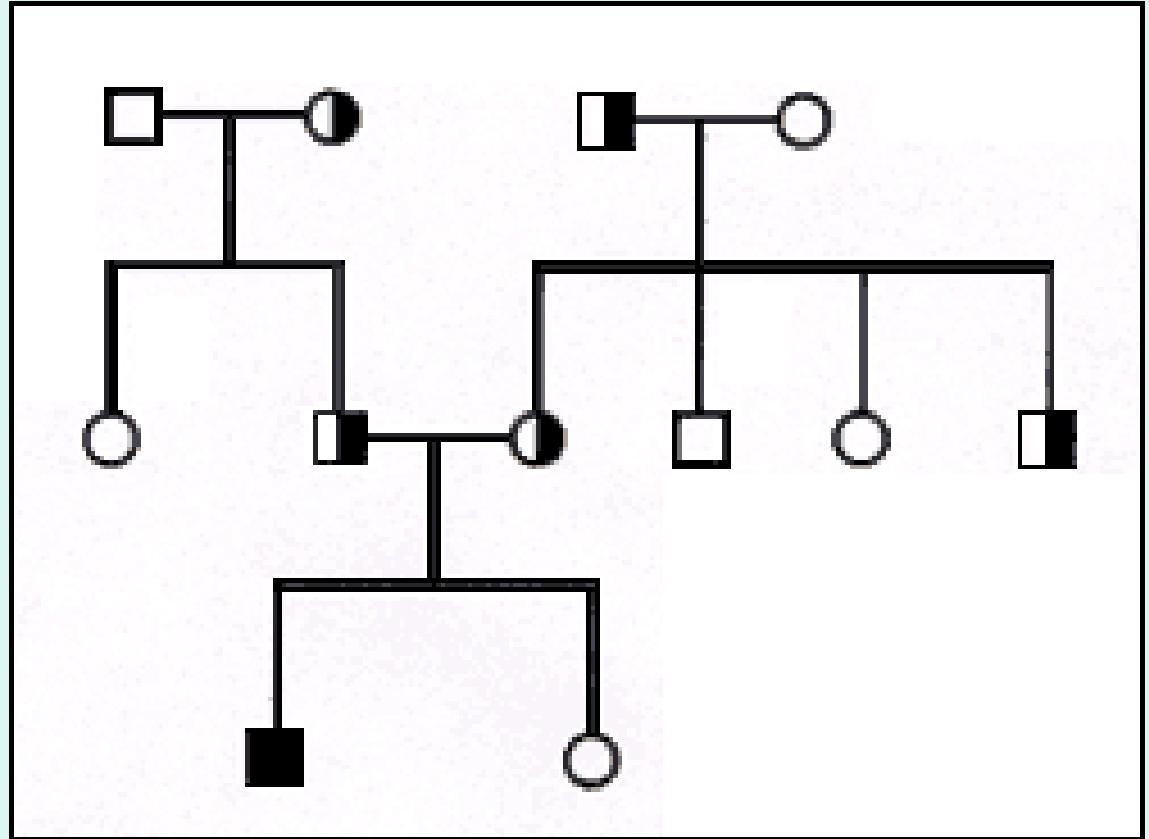
- Who is the first individual with the trait on the pedigree chart?
- Who did individual #7 generation III inherit the trait from?





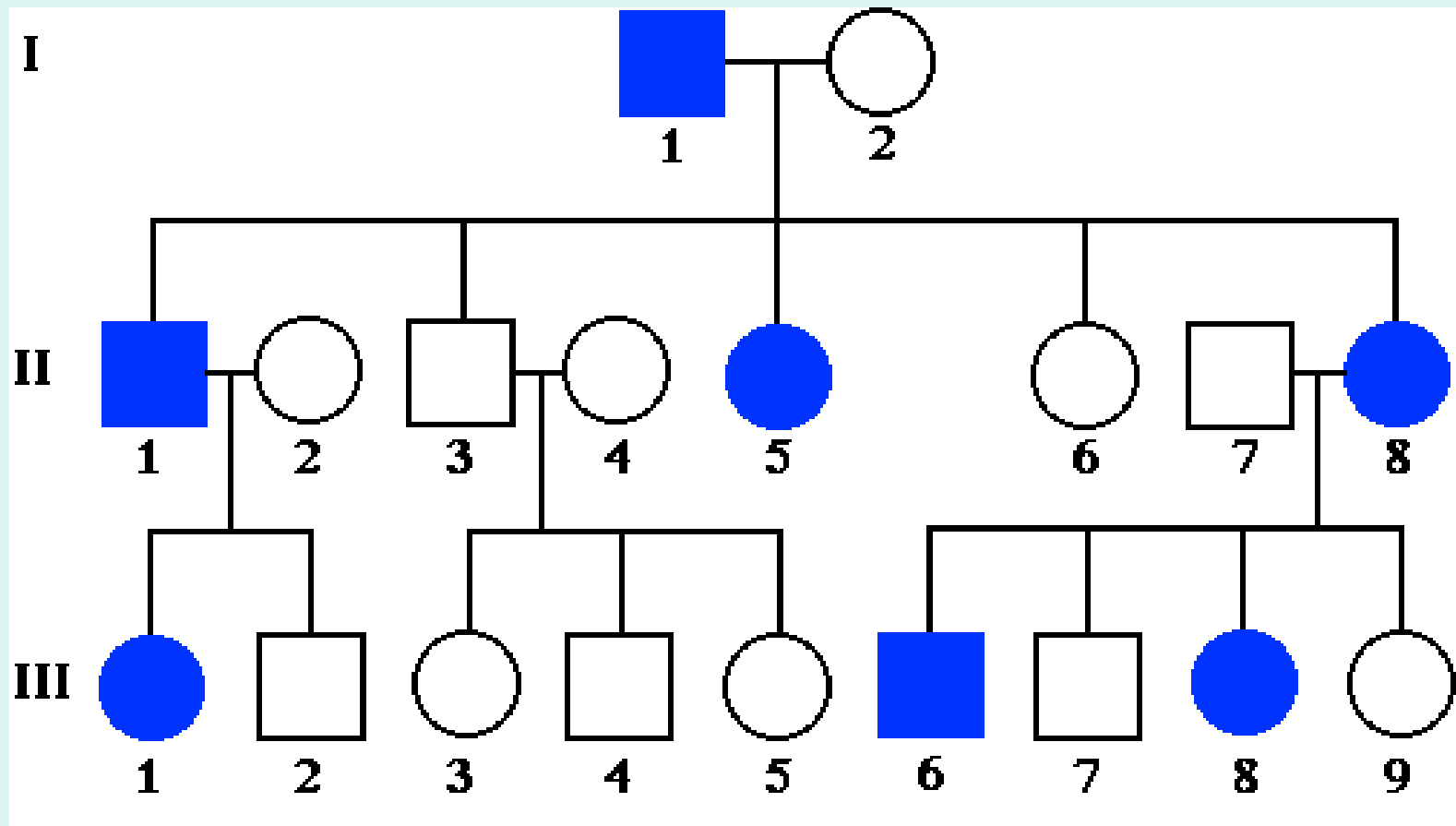
Analysis of a Recessive Trait

- How many individuals on this pedigree **have** the trait?
- How many are carriers?

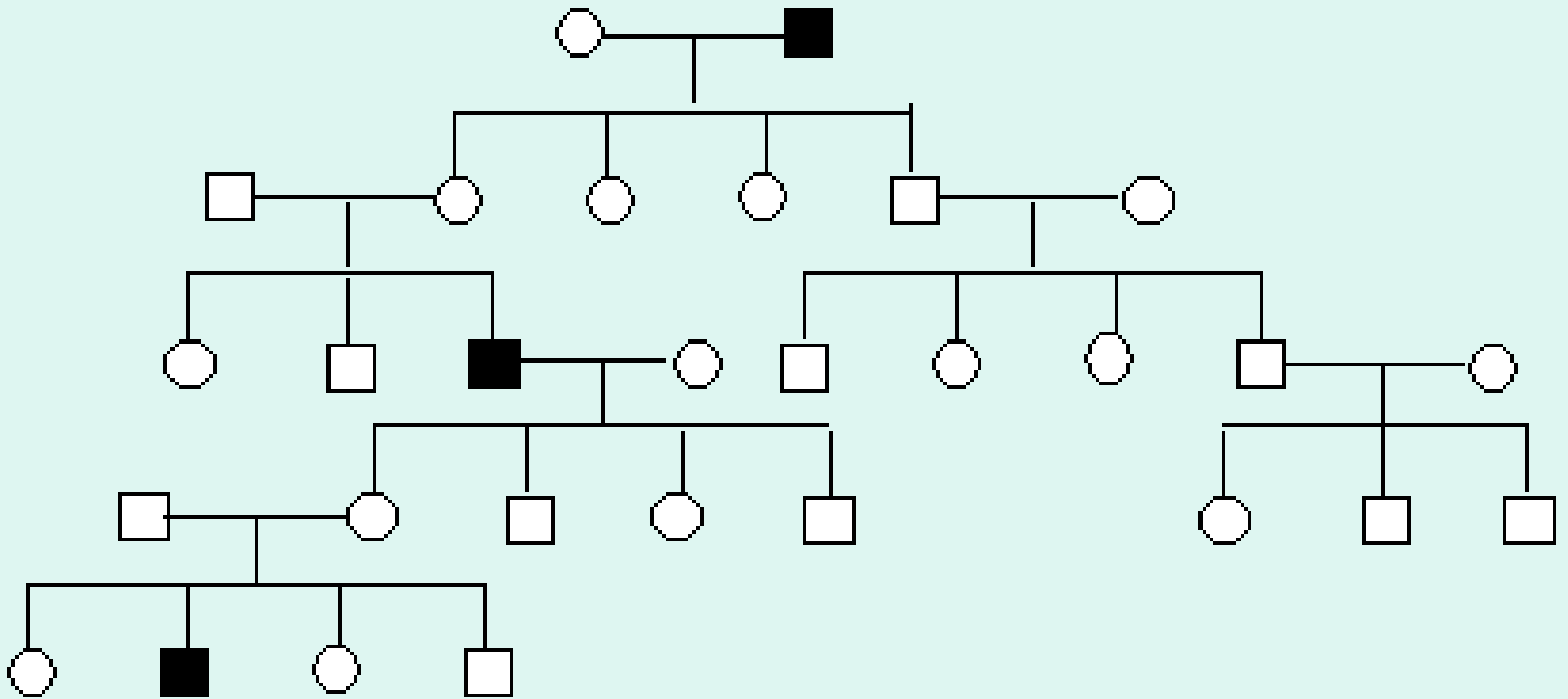


HINT: Dominant
Traits NEVER skip
a generation

Dominant or Recessive Trait?



Dominant or Recessive Trait?



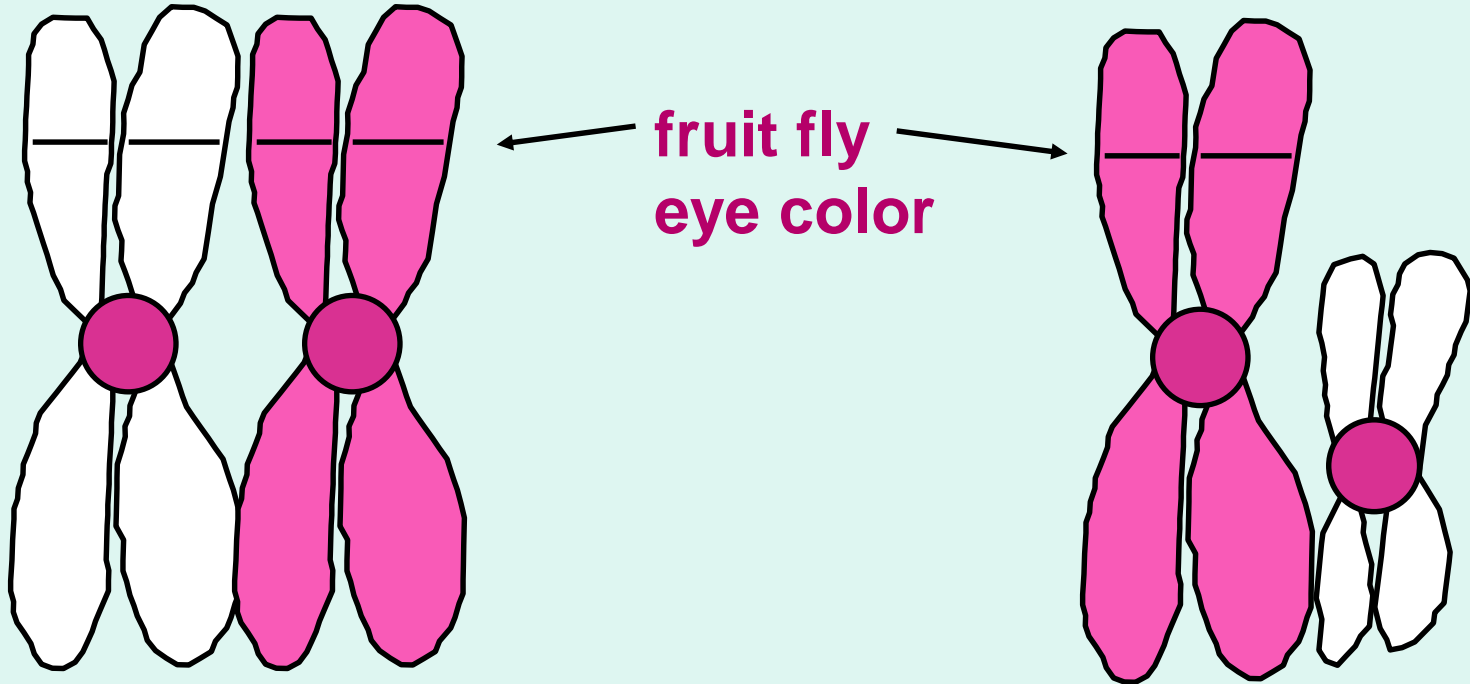
Sex-linked Traits

- Traits (genes) located on the sex chromosomes
- Sex chromosomes are X and Y
- XX genotype for females
- XY genotype for males
- Many sex-linked traits carried on X chromosome

Sex-linked Traits

Example: Eye color in fruit flies

Sex Chromosomes

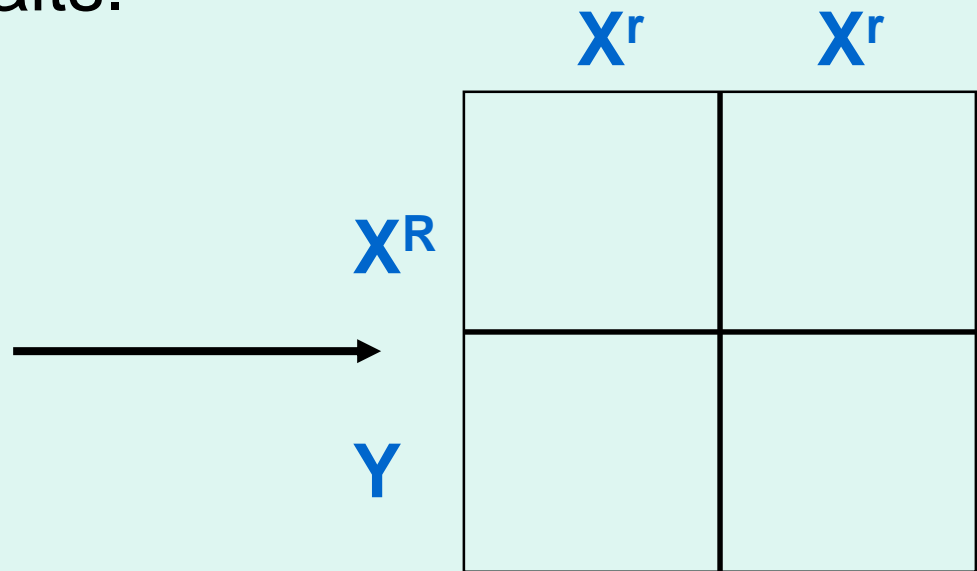


XX chromosome - female

Xy chromosome - male

Sex-linked Trait Problem

- Example: Eye color in fruit flies
- (red-eyed male) x (white-eyed female)
 $X^R Y$ x $X^r X^r$
- Remember: the Y chromosome in males does not carry traits.
- RR = red eyed
- Rr = red eyed
- rr = white eyed
- XY = male
- XX = female



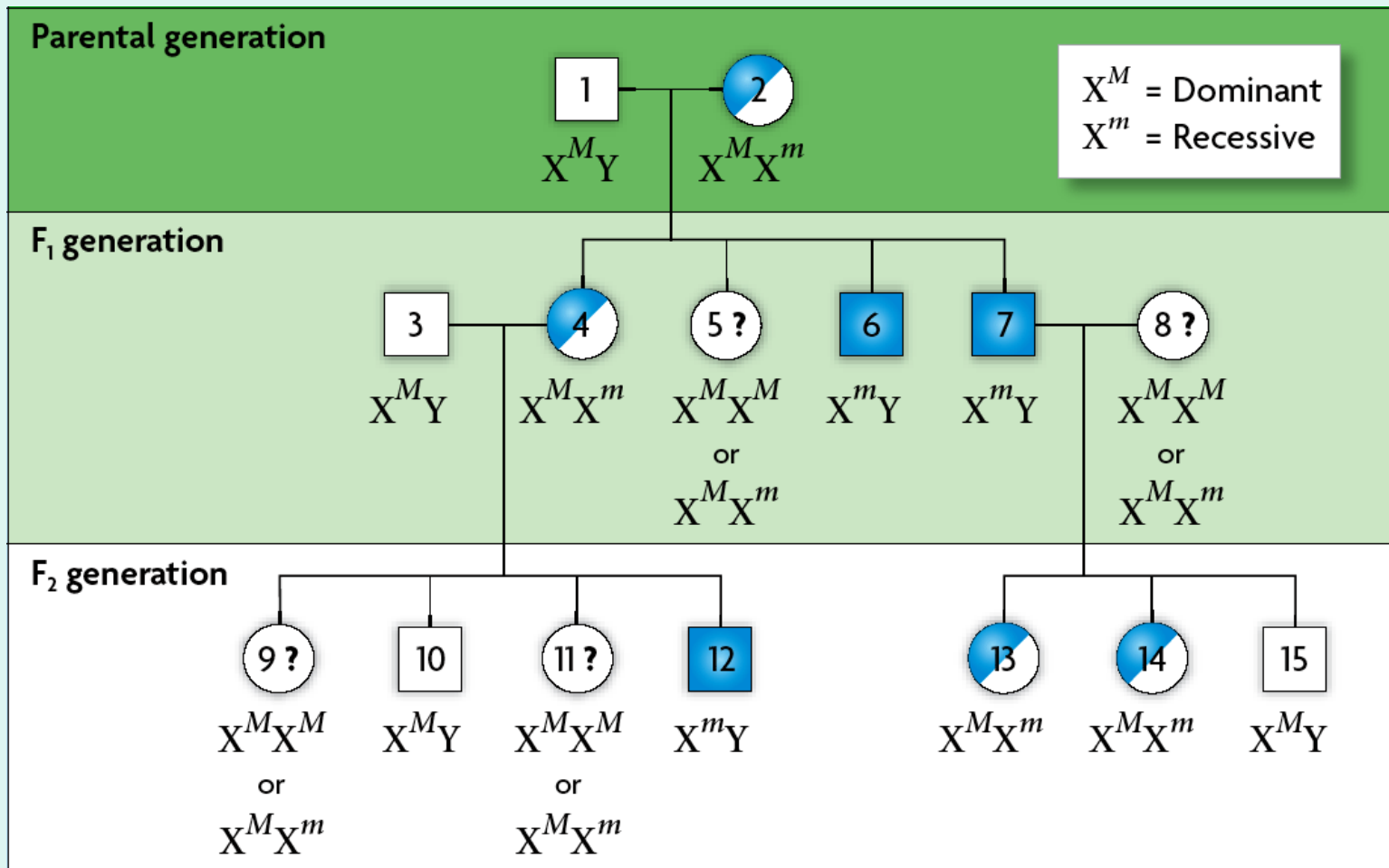
Sex-linked Trait Solution:

	X^r	X^r
X^R	$X^R X^r$	$X^R X^r$
Y	$X^r Y$	$X^r Y$

50% **red eyed**
female

50% **white eyed**
male

- If the phenotype is more common in males, the gene is likely sex-linked.

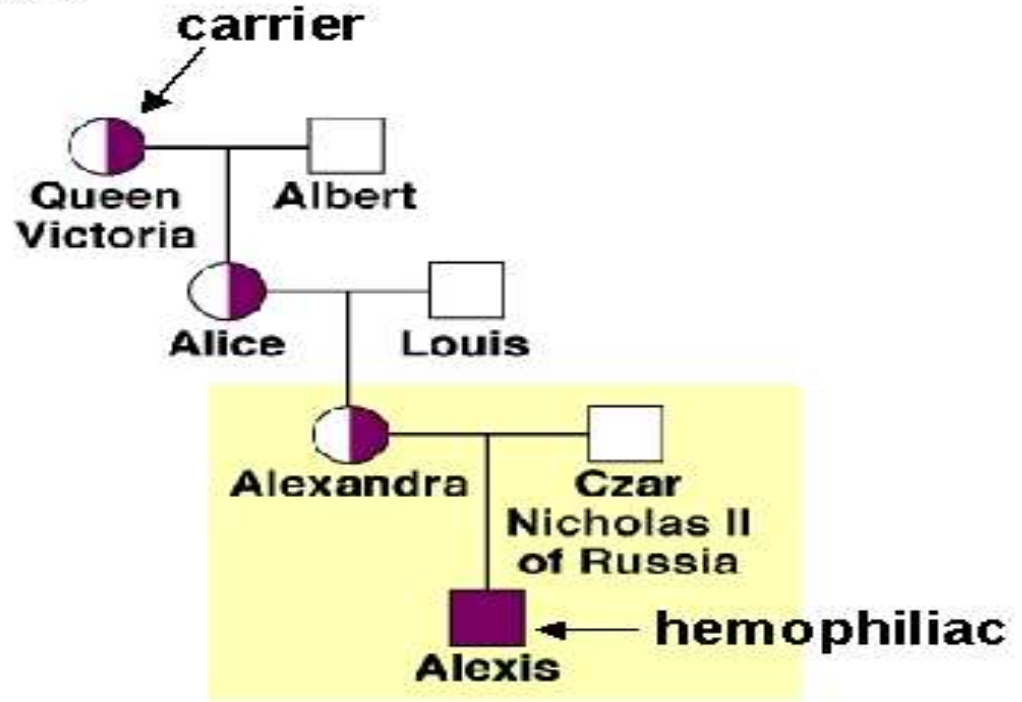


Female Carriers

In a sex-linked trait (like hemophilia), women are carriers, and men have the phenotype more often.



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Create a Pedigree

- A man and woman marry.
- They have five children, 2 girls and 3 boys.
- The mother is a carrier of hemophilia, an X-linked disorder.
- She passes the gene on to two of the boys and one of the daughters is also a carrier.
- Both daughters marry men without hemophilia and have 3 children (2 boys and a girl).
- The carrier daughter has one son with hemophilia.
- One of the non-carrier daughter's sons marries a woman who is a carrier and they have twin daughters.
- What is the percent chance that each daughter will also be a carrier?