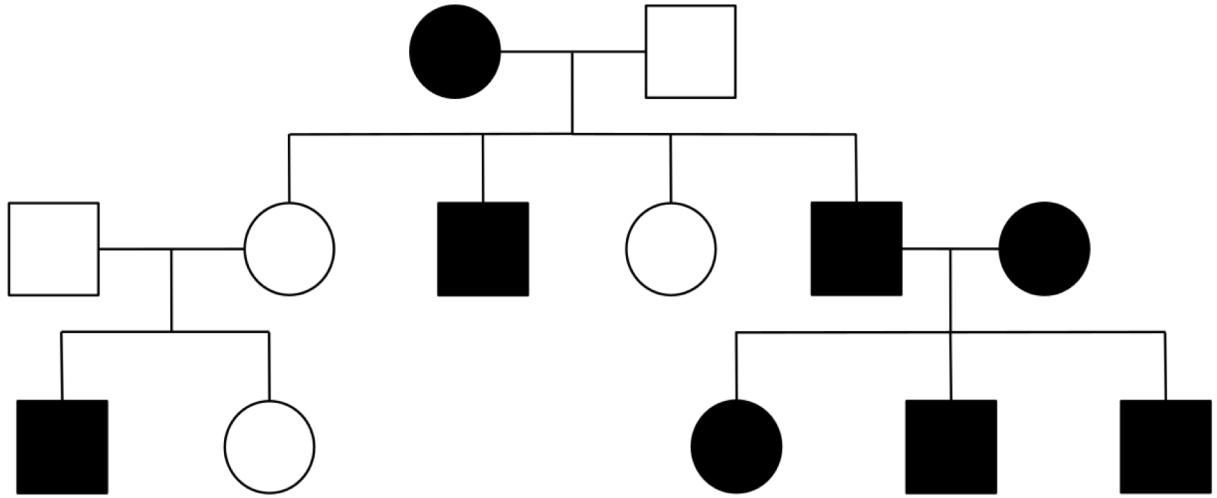
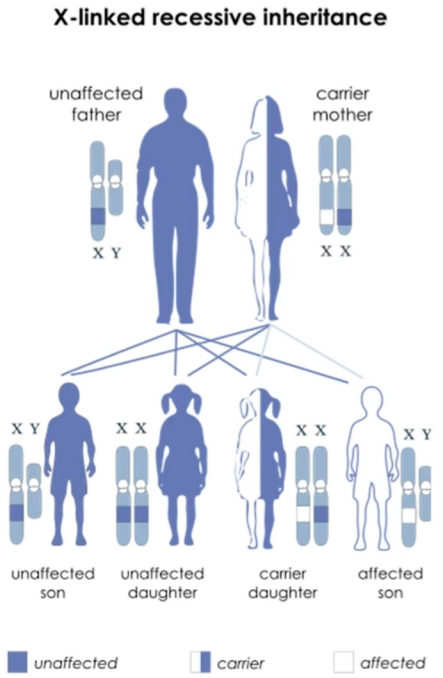
Lesson 16
Pedigrees



Pedigrees

- Sometimes, the genetics of a particular trait are very complicated
  - There might be many genes involved
    - There might be traits that are associated with sex
    - There might be traits that are influenced in ways that we just don't understand
- **Pedigrees** = another set of tools to understand complex traits
  - Especially important to understand diseases



Pedigree analysis uncovers type of inheritance suggests gene(s) characteristics

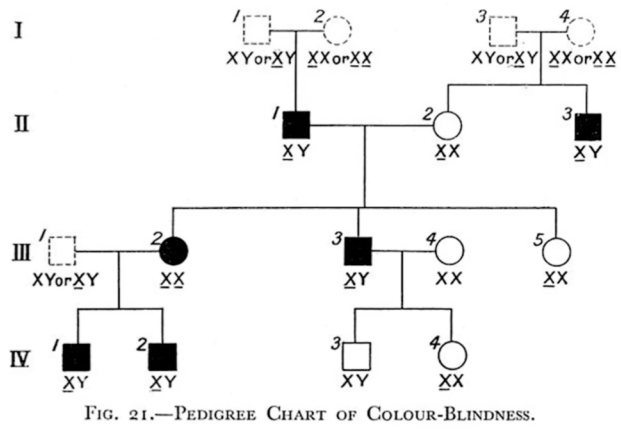


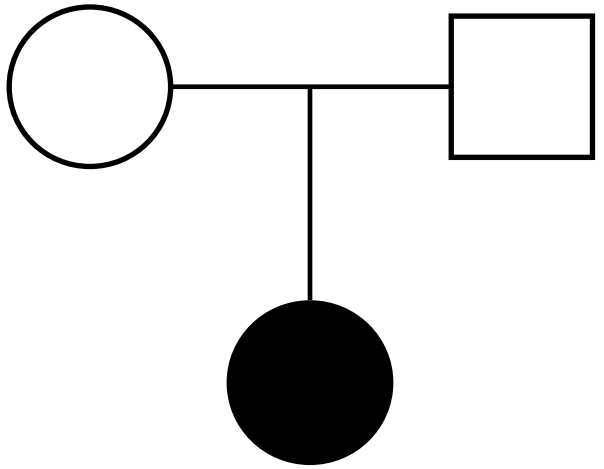
FIG. 21.—PEDIGREE CHART OF COLOUR-BLINDNESS.

Pedigrees

- **Pedigrees** = **genetics from family history**
- Conventions in pedigree writing:
  - ○ female
  - □ male
  - ●■ the trait you are looking at is present (*e.g.*, a disease, brown eyes, etc.)

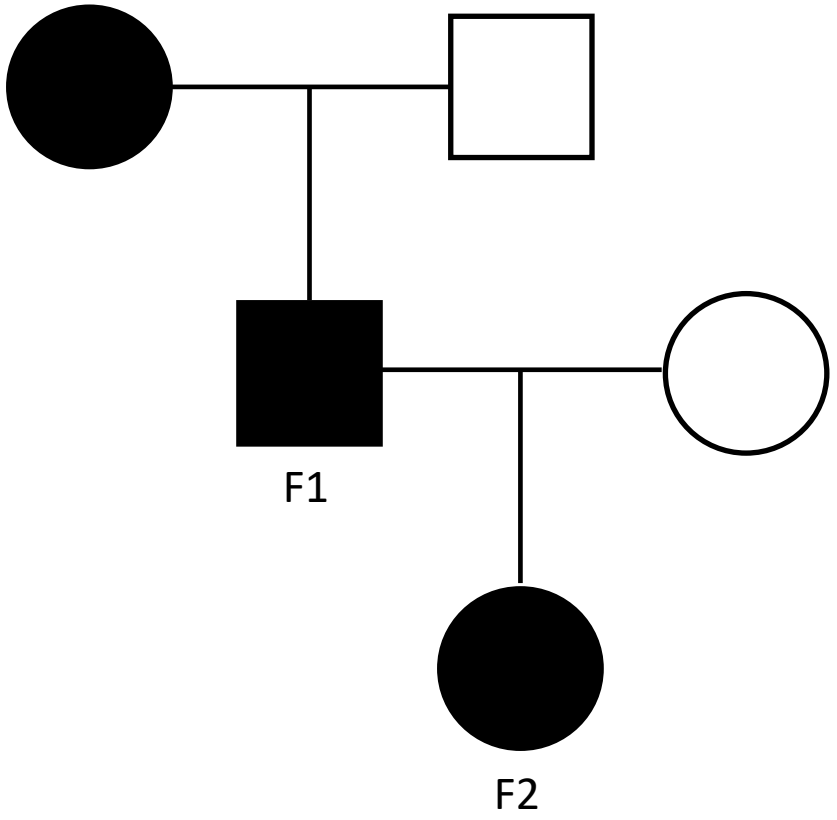
Pedigrees - 1

- This pedigree (*e.g.*, disease):
  - Two healthy parents
    - One affected daughter
- **This is a characteristic of a recessive trait**

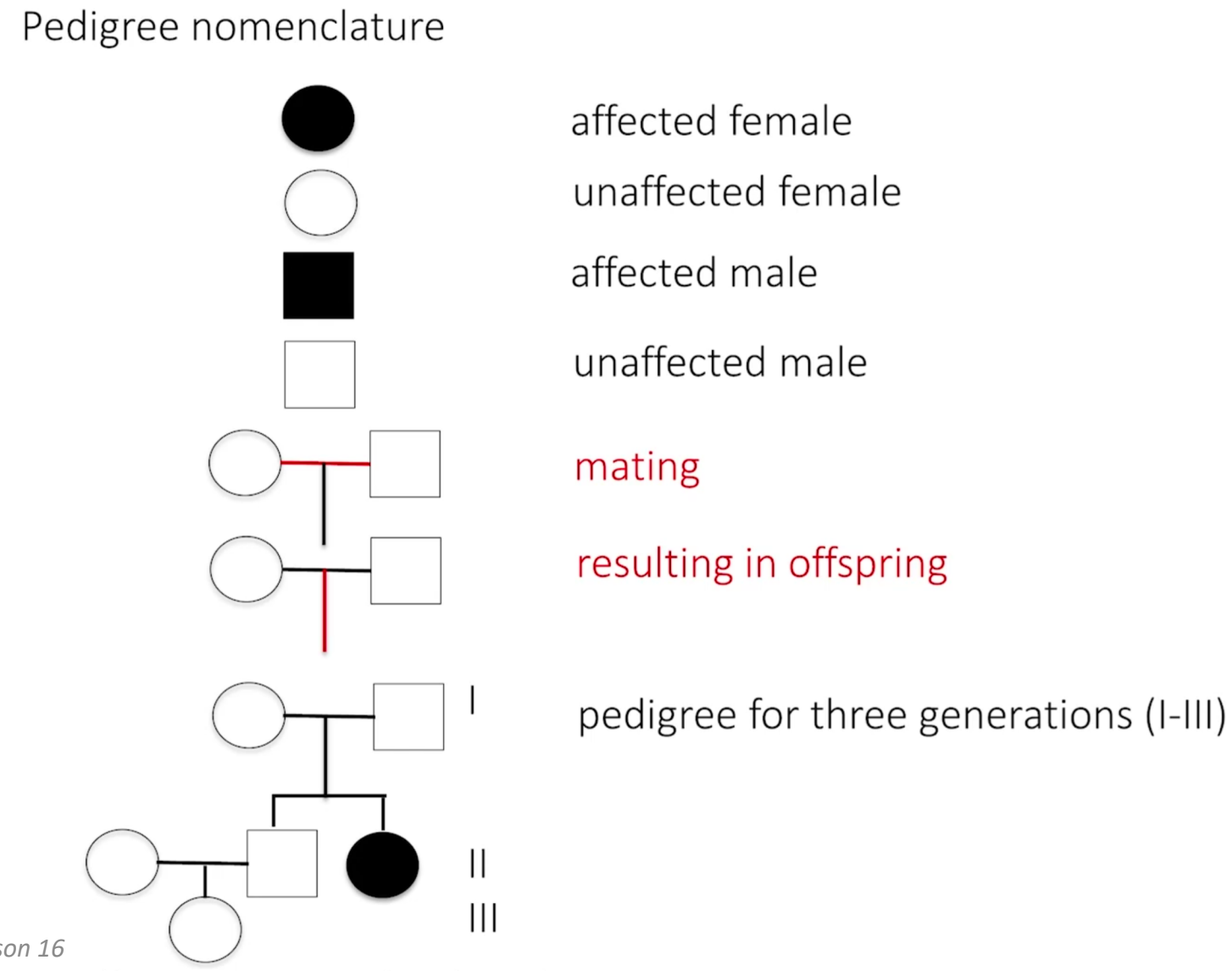


Pedigrees - 2

- This pedigree (*e.g.*, disease):
  - One affected parent and one unaffected parent (*e.g.*, father)
    - One affected son (F1)
  - The affected son mates an unaffected female
    - One affected daughter (F2)
- **The pedigree pattern where every affected offspring has an affected parent is characteristic of a dominant trait**

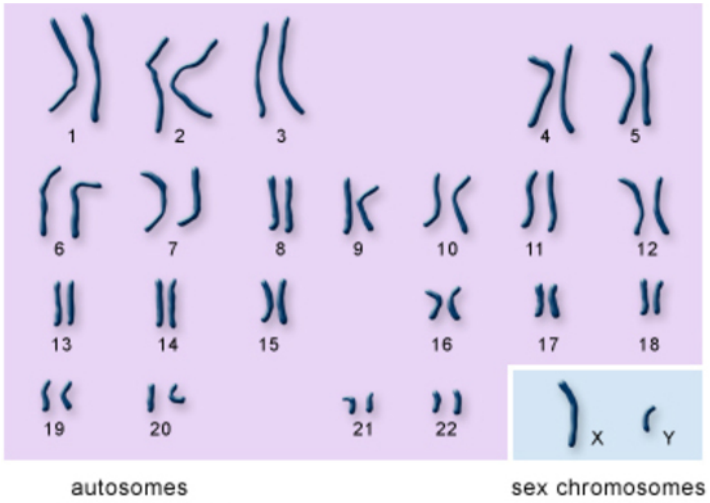


More pedigree nomenclature



Chromosomes and pedigrees

- Eukaryote cells have two types of chrs:
  - Autosomes = 22 paired chrs (2 chrs 1, 2 chrs2, etc….)
  - Sex chrs*:
    - XX (paired) → female * simplified
    - XY (unpaired) → male



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- 3 types of pedigree based on autosomes or sex chromosomes:
- Autosomal recessive
- Autosomal dominant

} Males and females affected equally

Chromosomes and pedigrees

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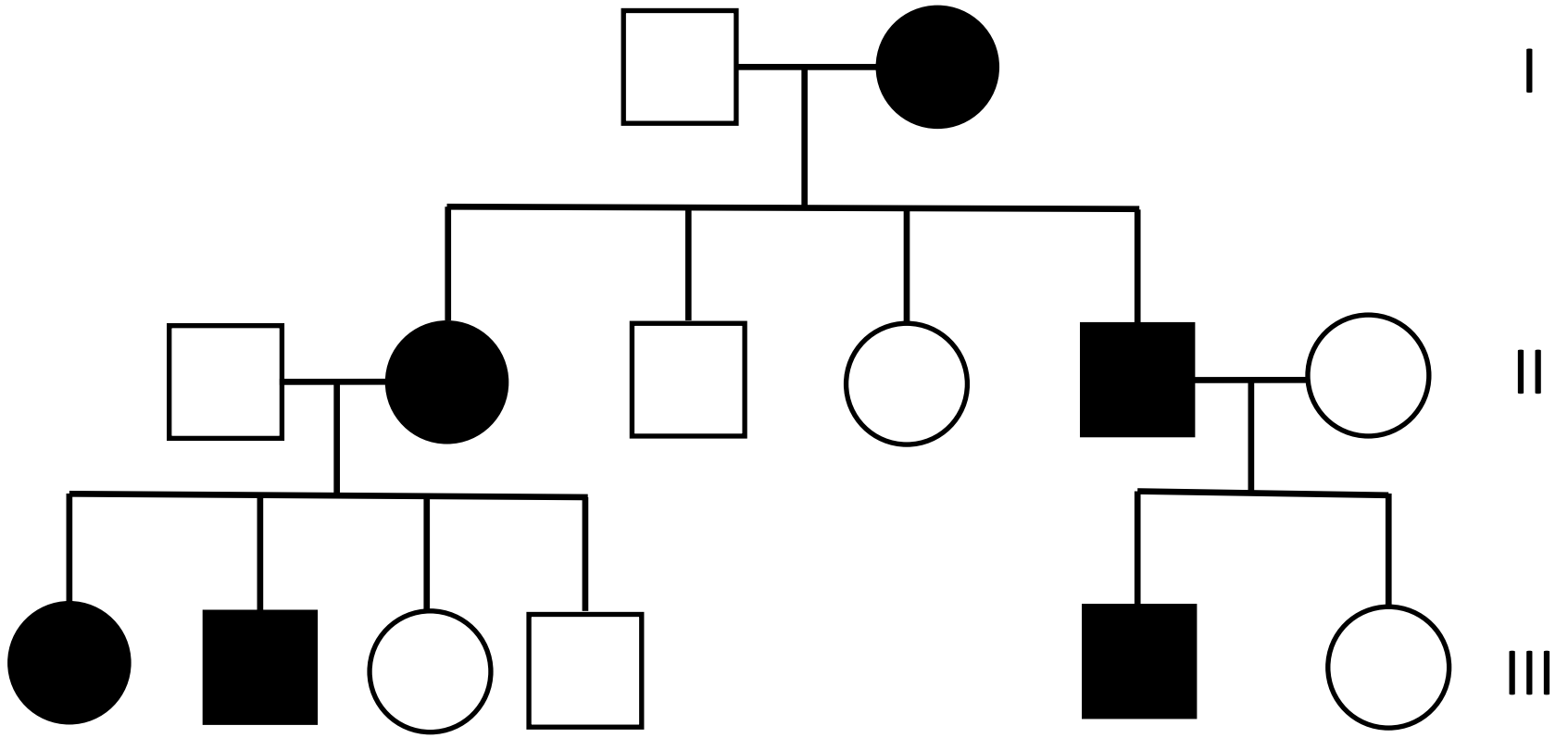
  * simplified
- 3 types of of pedigree based on autosomes or sex chromosomes:
- Autosomal recessive
- Autosomal dominant } Males and females affected equally
- X-linked recessive → Males affected more than females
  - there is only one X (the other allele is Y) and is affected –> the trait will show itself)
  - It is X-linked recessive because this trait on the X allele is always transmitted to the SONS (XY) from the mother

Pedigree for autosomal dominant trait

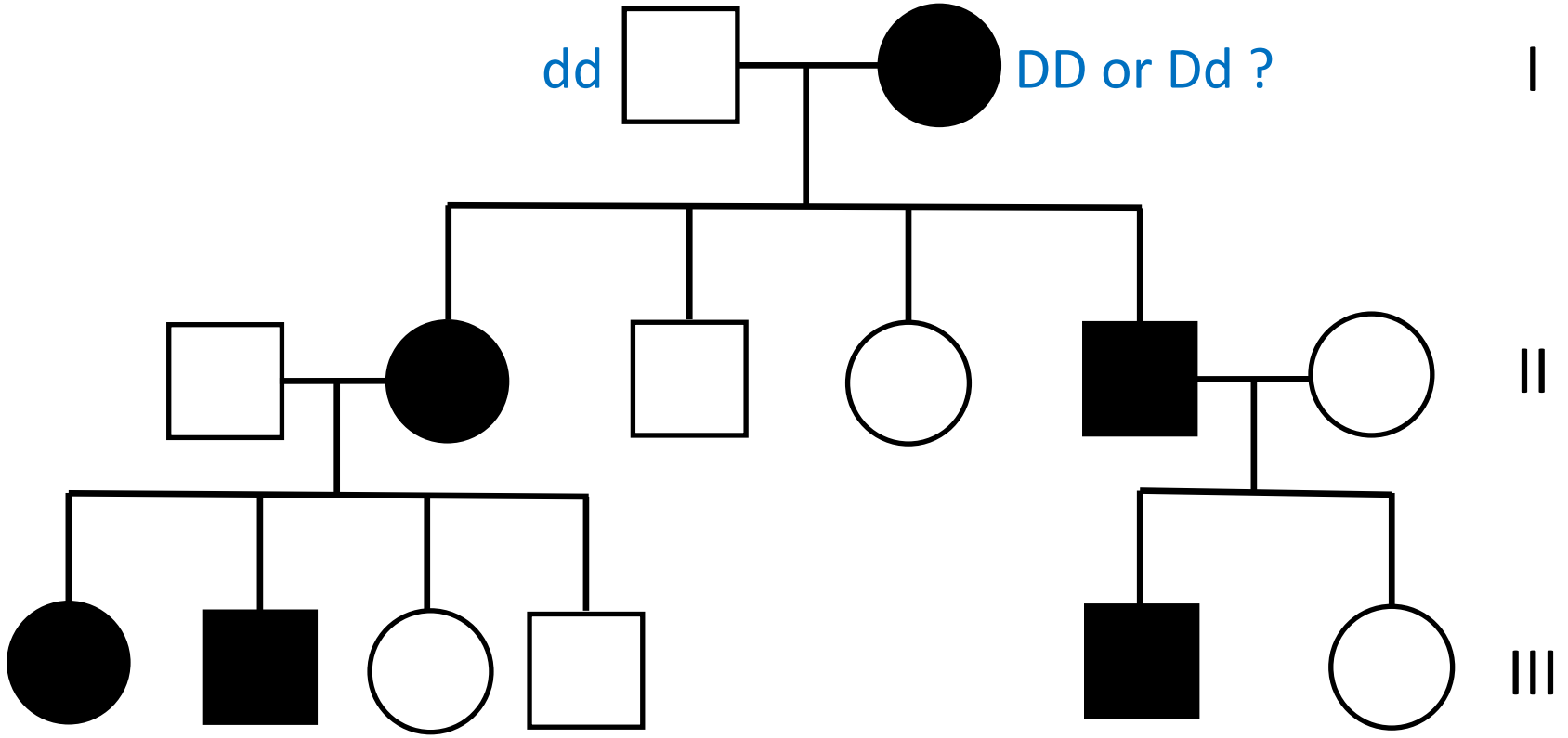
Clues

Autosomal: both males and females are affected

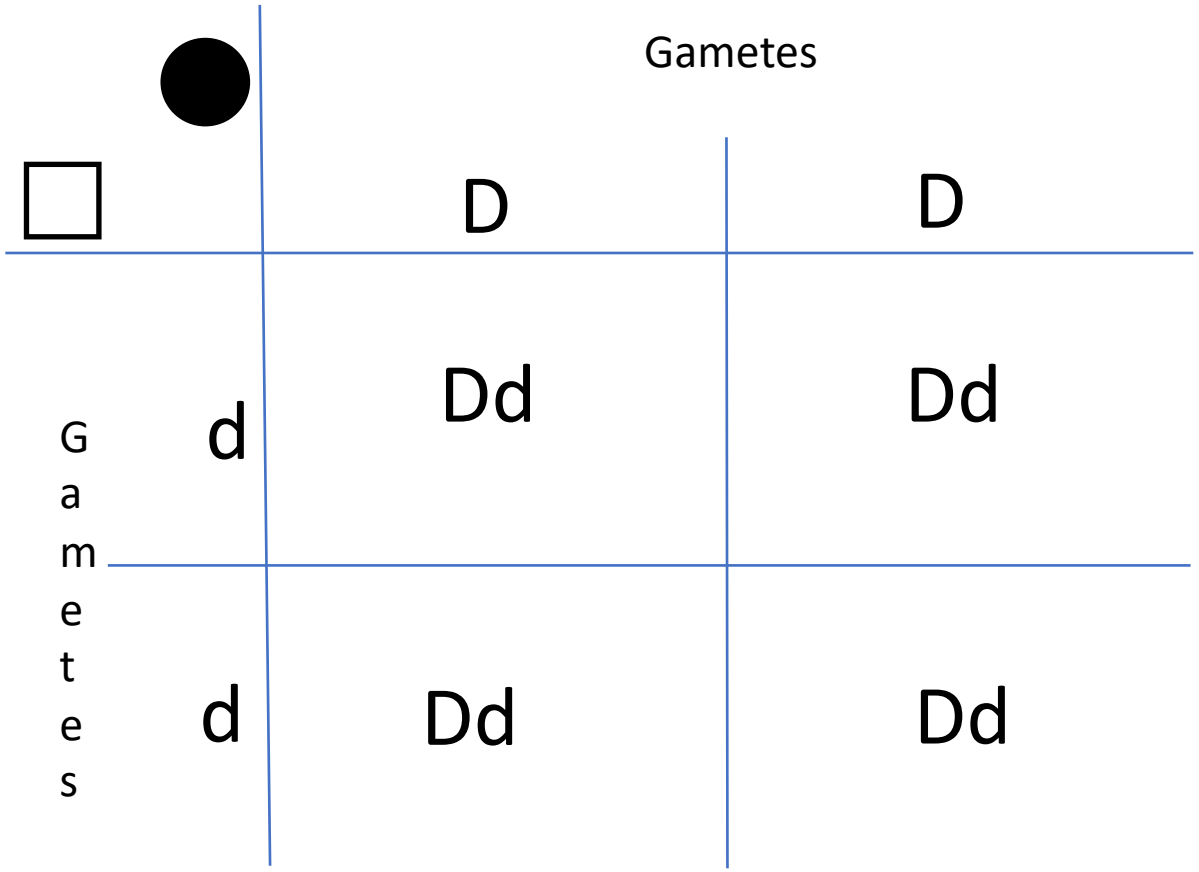
Dominant: <u>every affected</u> child has <u>one affected parent</u>



Pedigree for autosomal dominant trait (with genotypes)

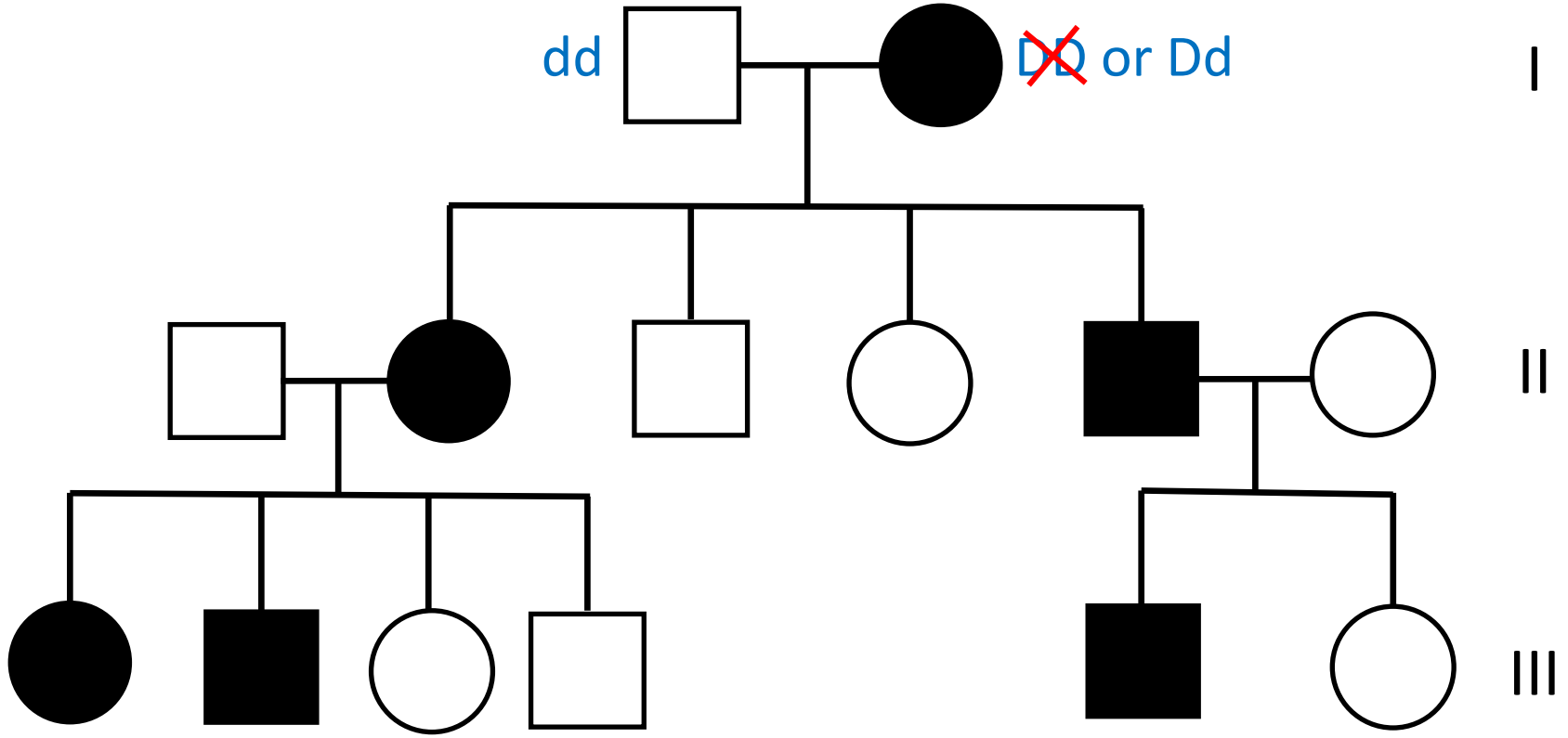


Punnett squares for gen II (gen 1 = parents)

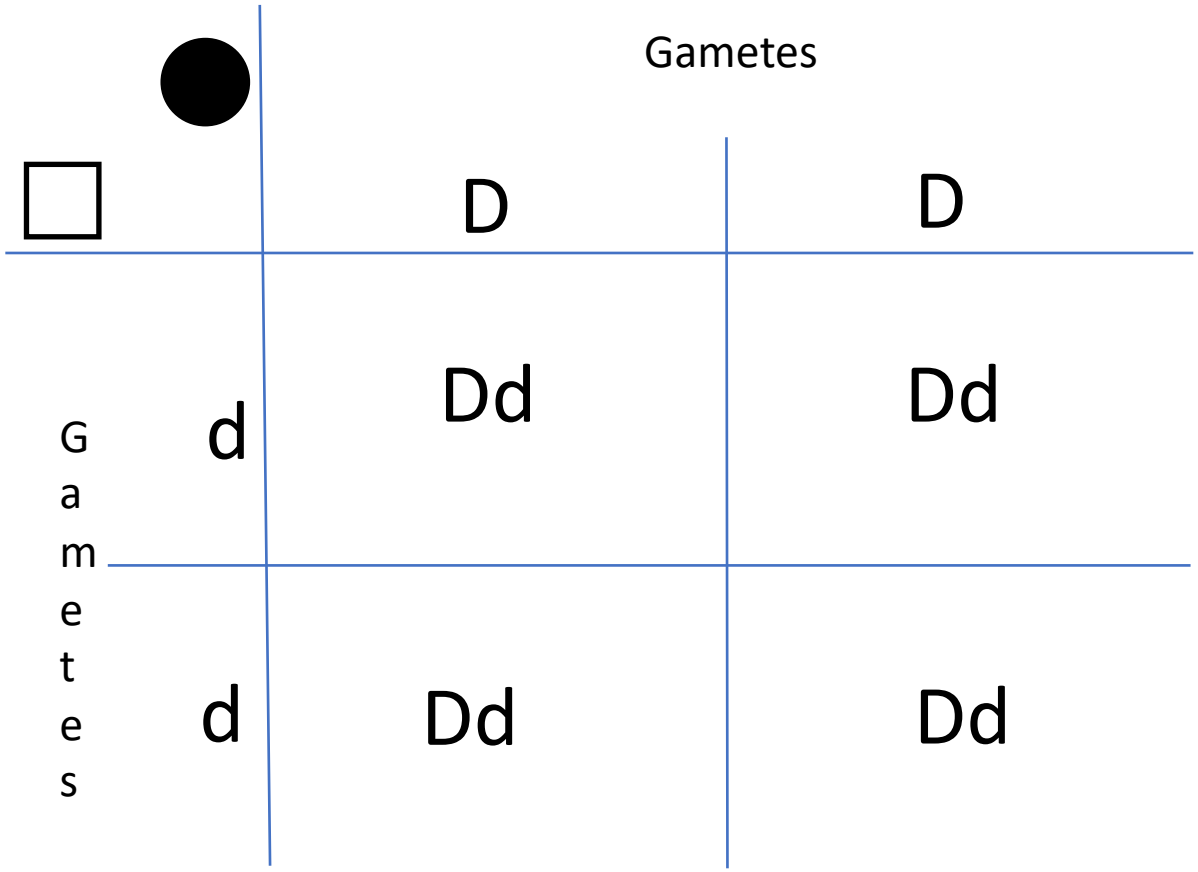


100% of F1 would be affected (all have the dominant allele D)

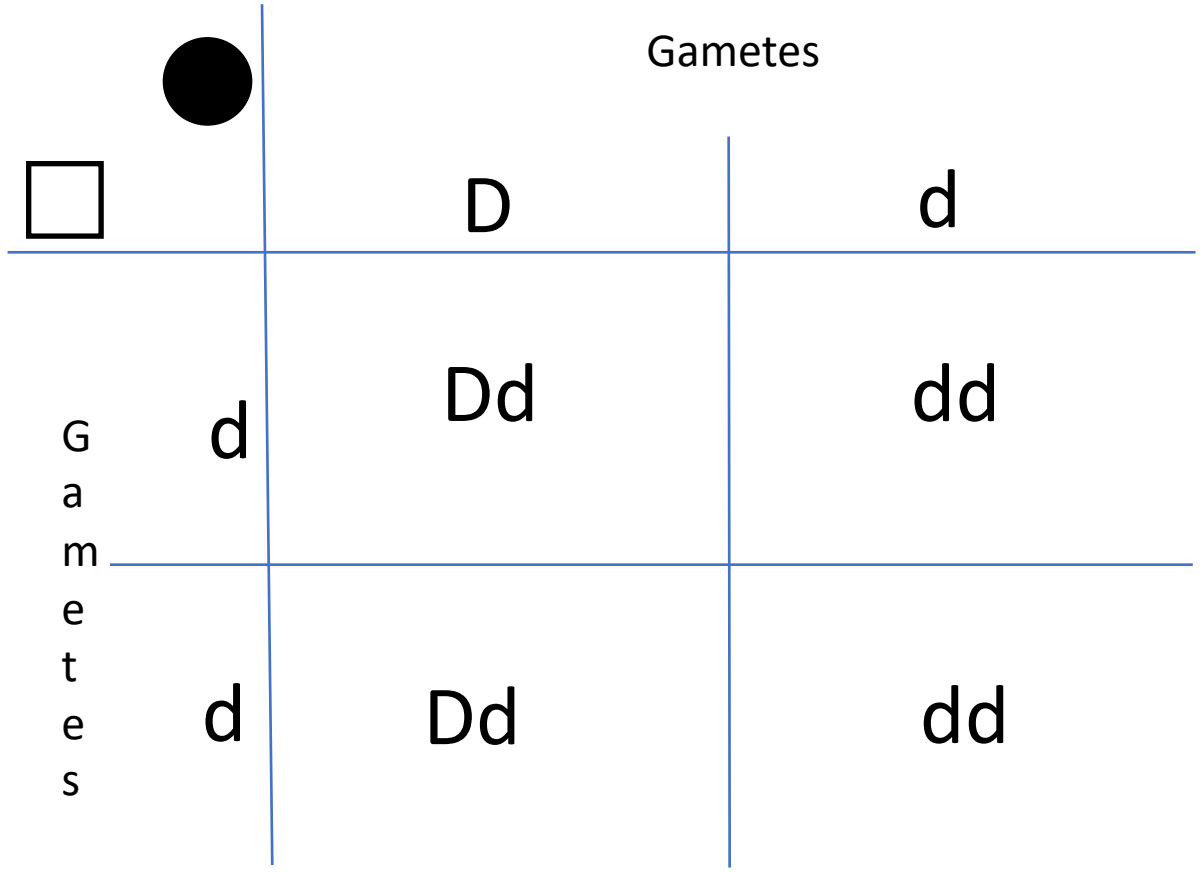
Pedigree for autosomal dominant trait (with genotypes)



Punnett squares for gen II (gen I = parents)

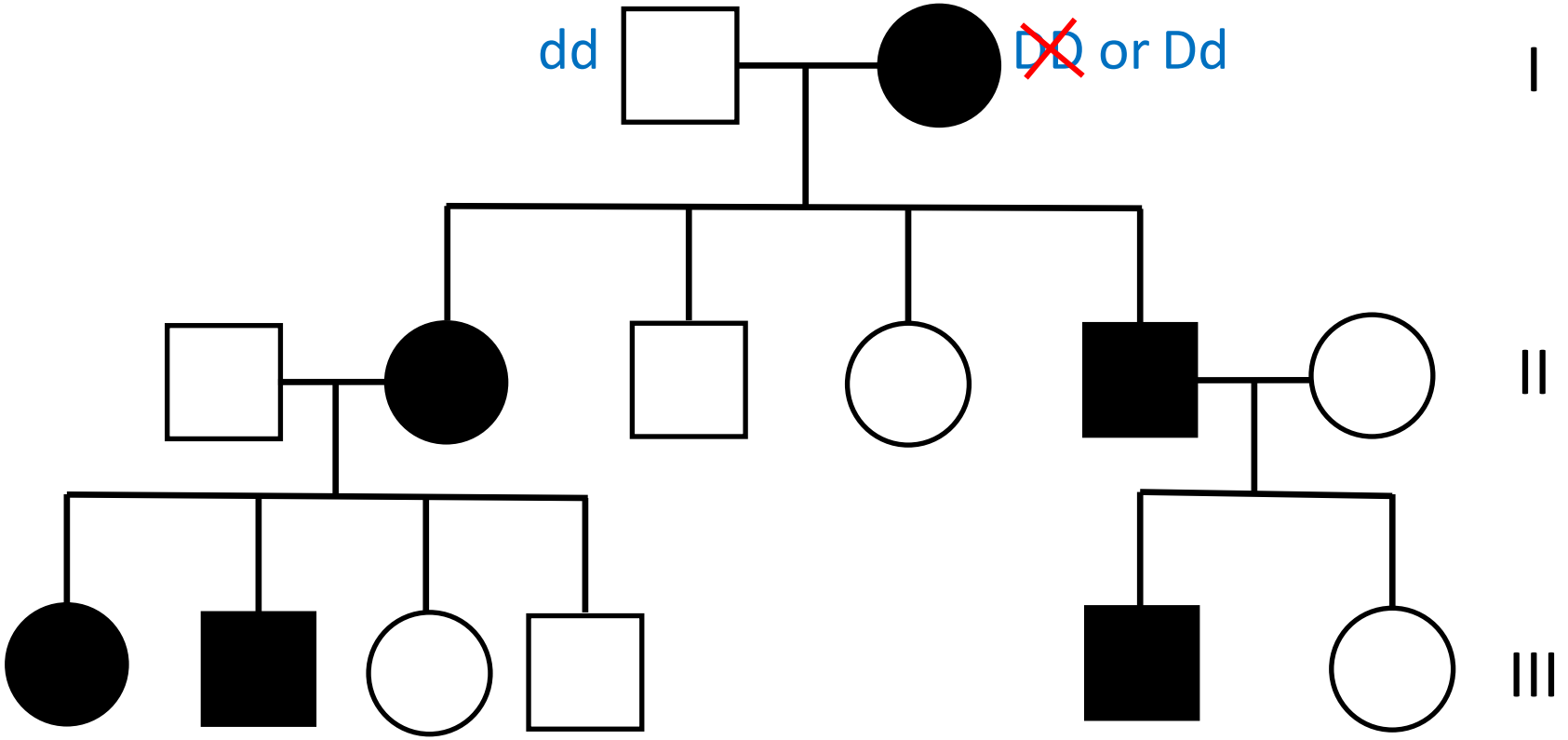


100% of F1 would be affected (all have the dominant allele D)



50% of F1 would be affected (half have the dominant allele D)

Pedigree for autosomal dominant trait (with genotypes)



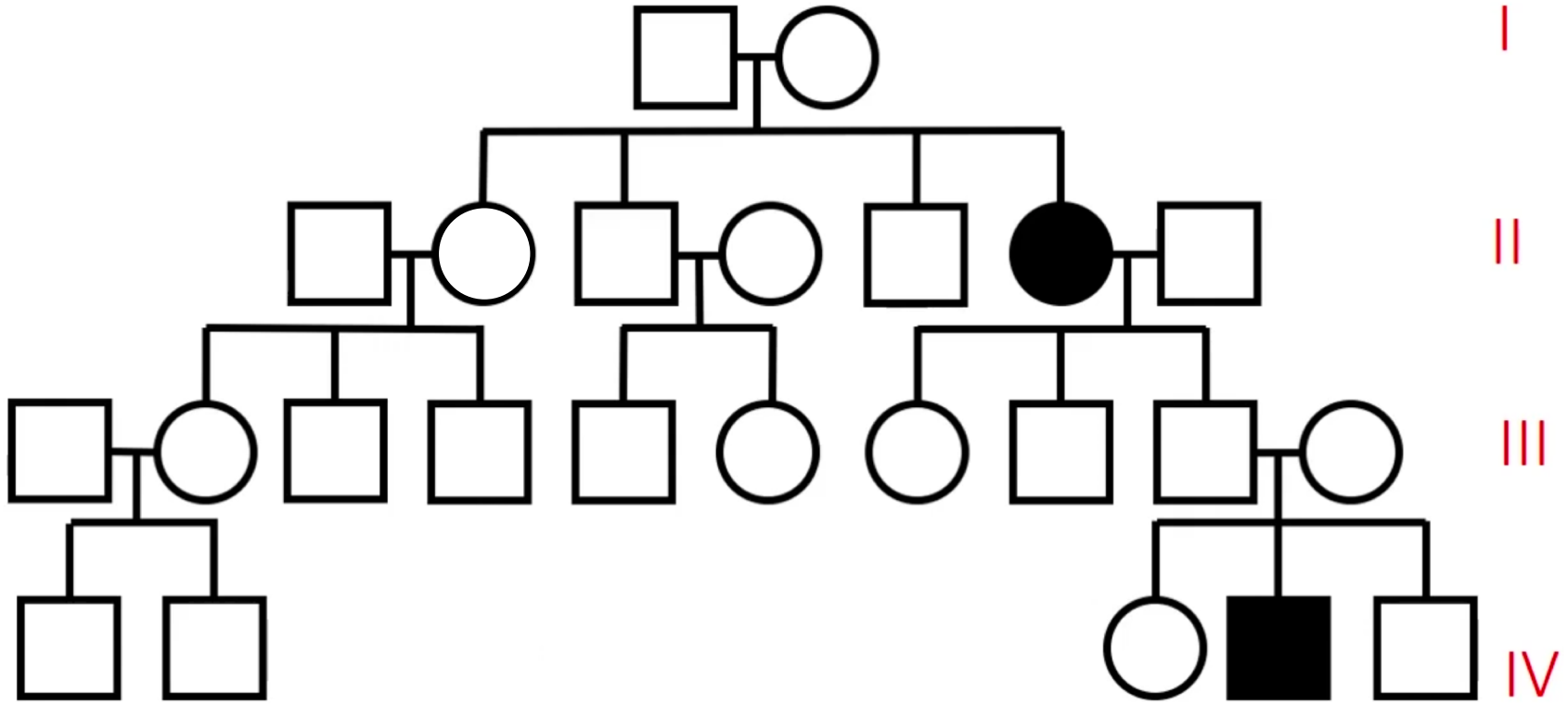
Note: you may not always get ratios identical to predicted due to probability of using specific gametes

Pedigree for autosomal recessive trait

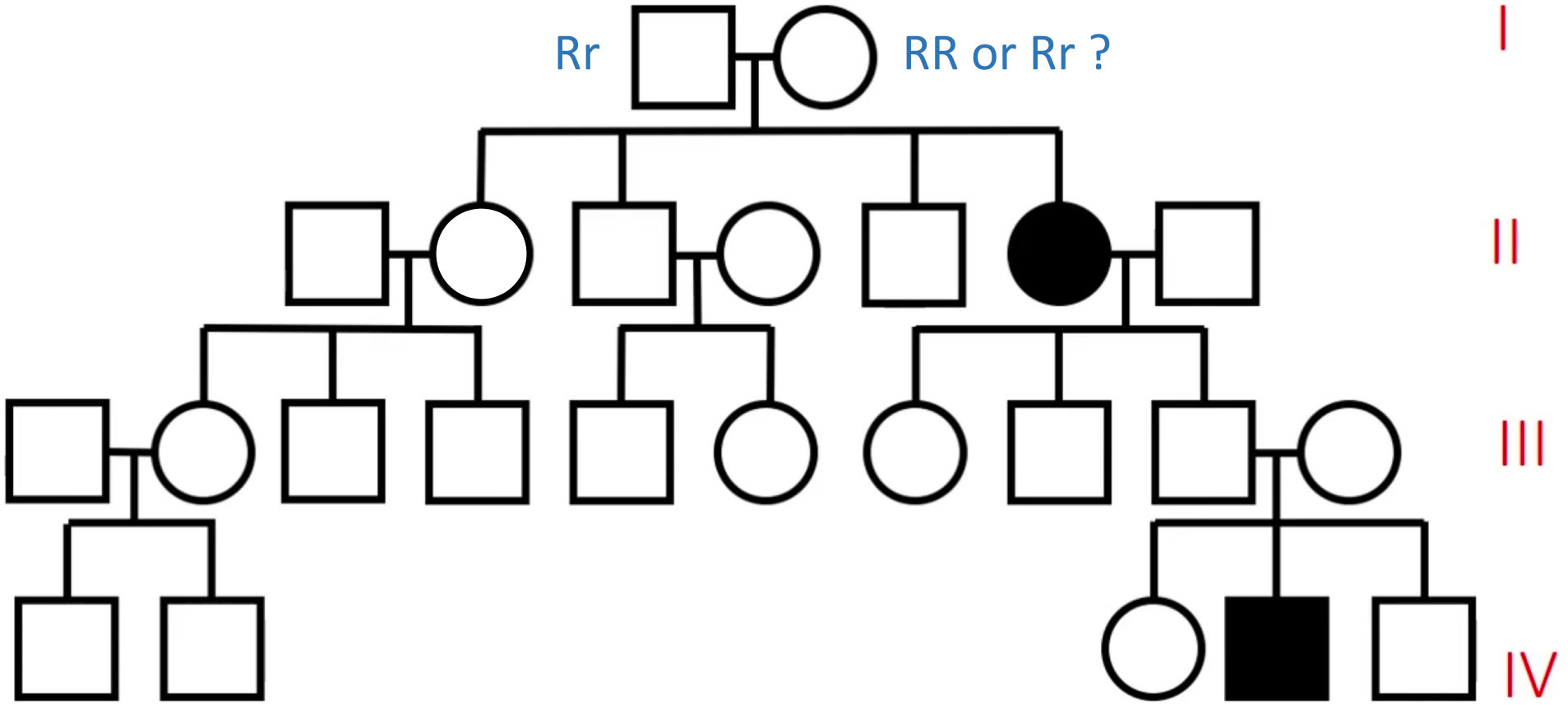
Clues

Autosomal: both males and females are affected

Recessive: affected child(ren) from unaffected parents not many affected offspring

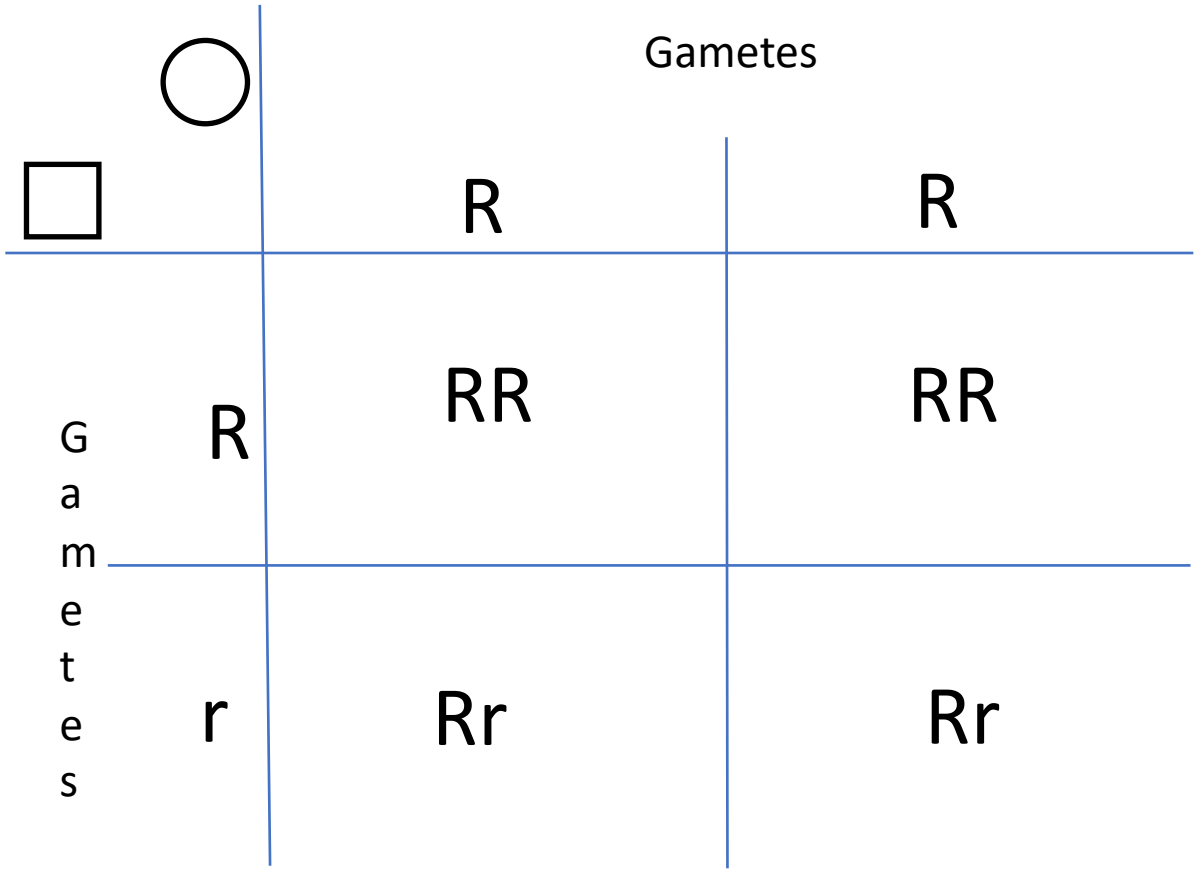


Pedigree for autosomal recessive trait (with genotypes)



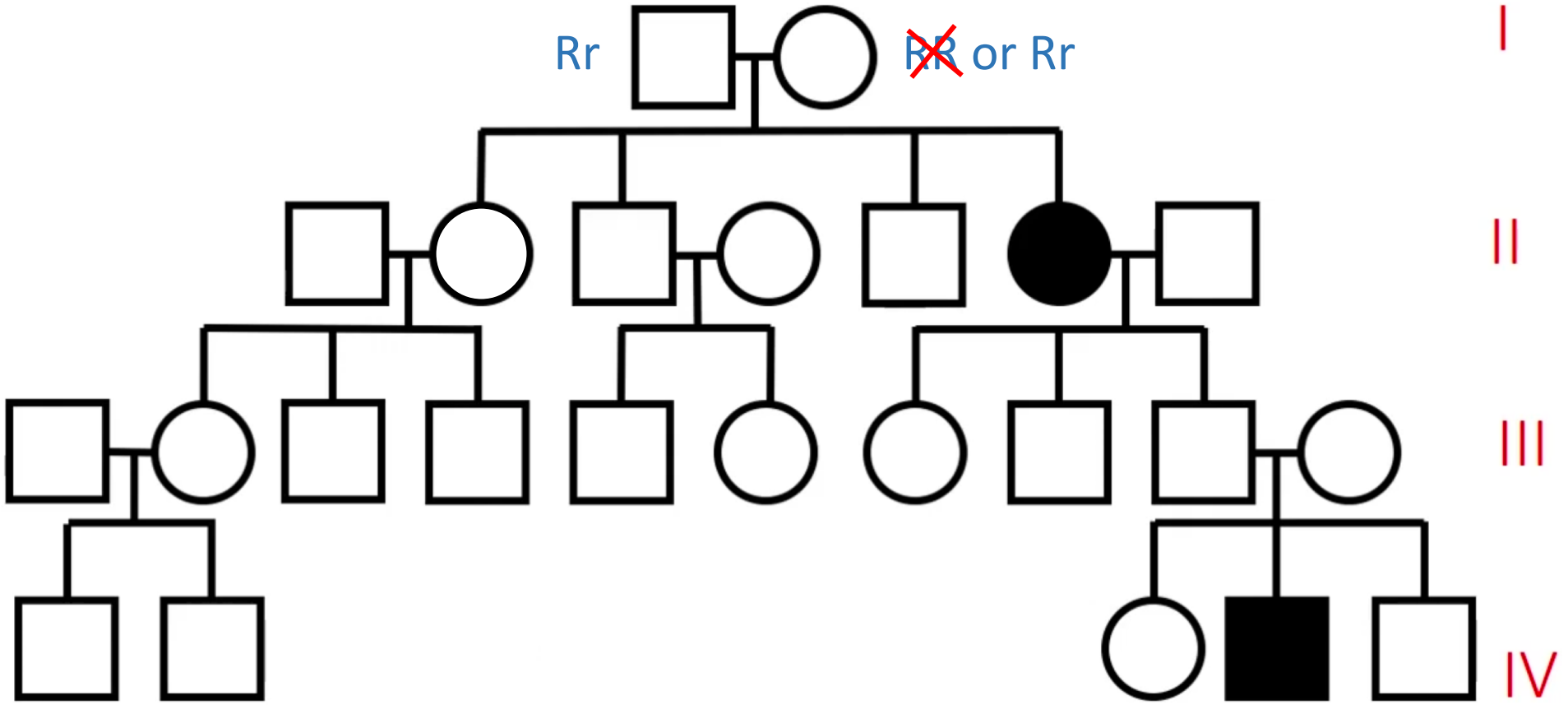
Remember: it is the recessive allele r that expresses the disease

Punnett squares for gen II (gen 1 = parents)

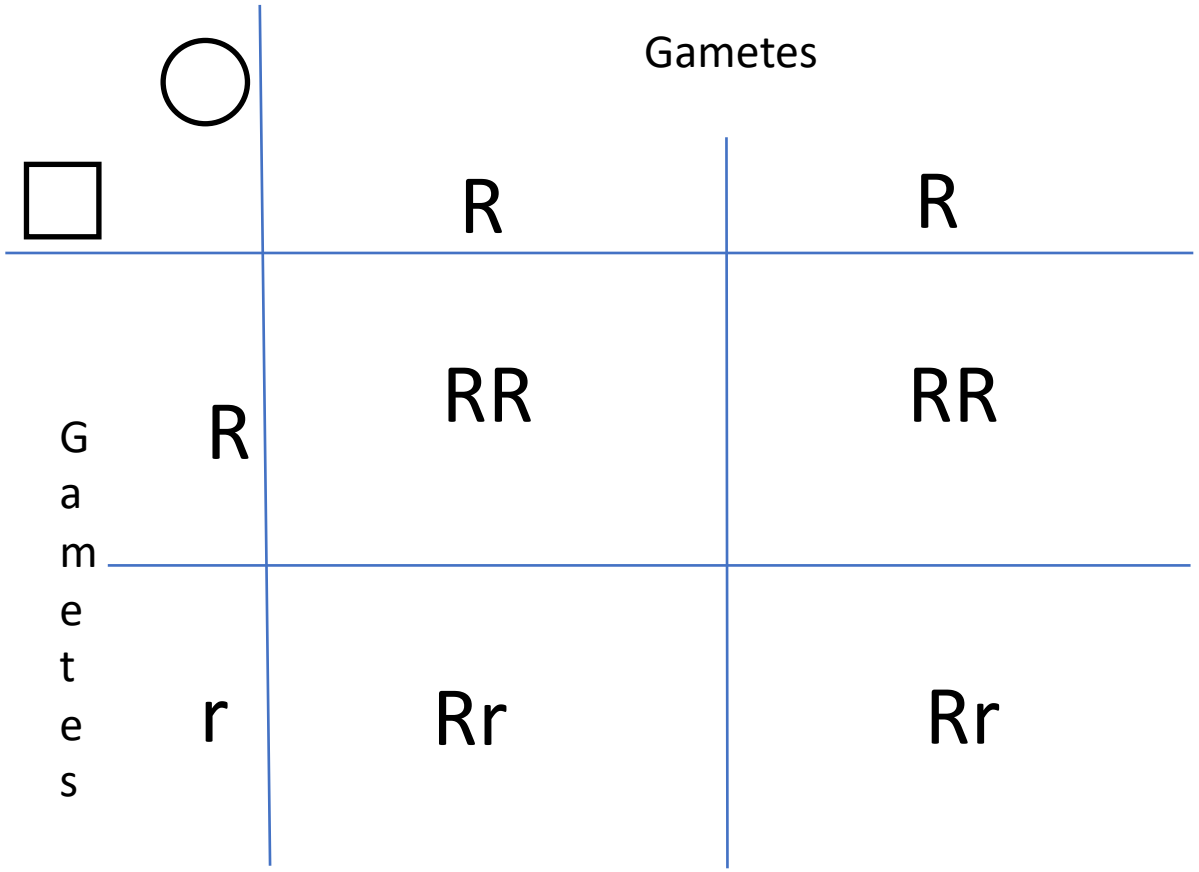


0% of F1 would be affected (none has genotype rr)

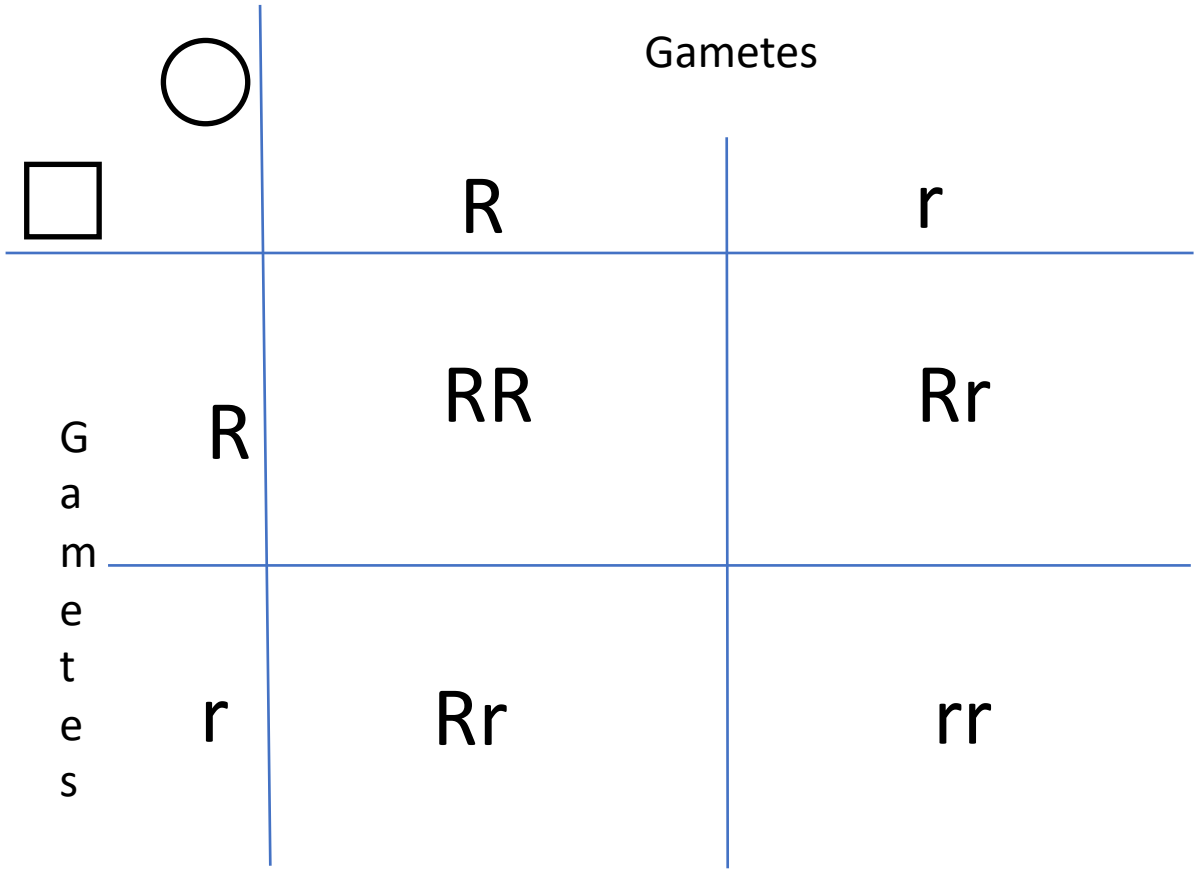
Pedigree for autosomal recessive trait (with genotypes)



Punnett squares for gen II (gen 1 = parents)

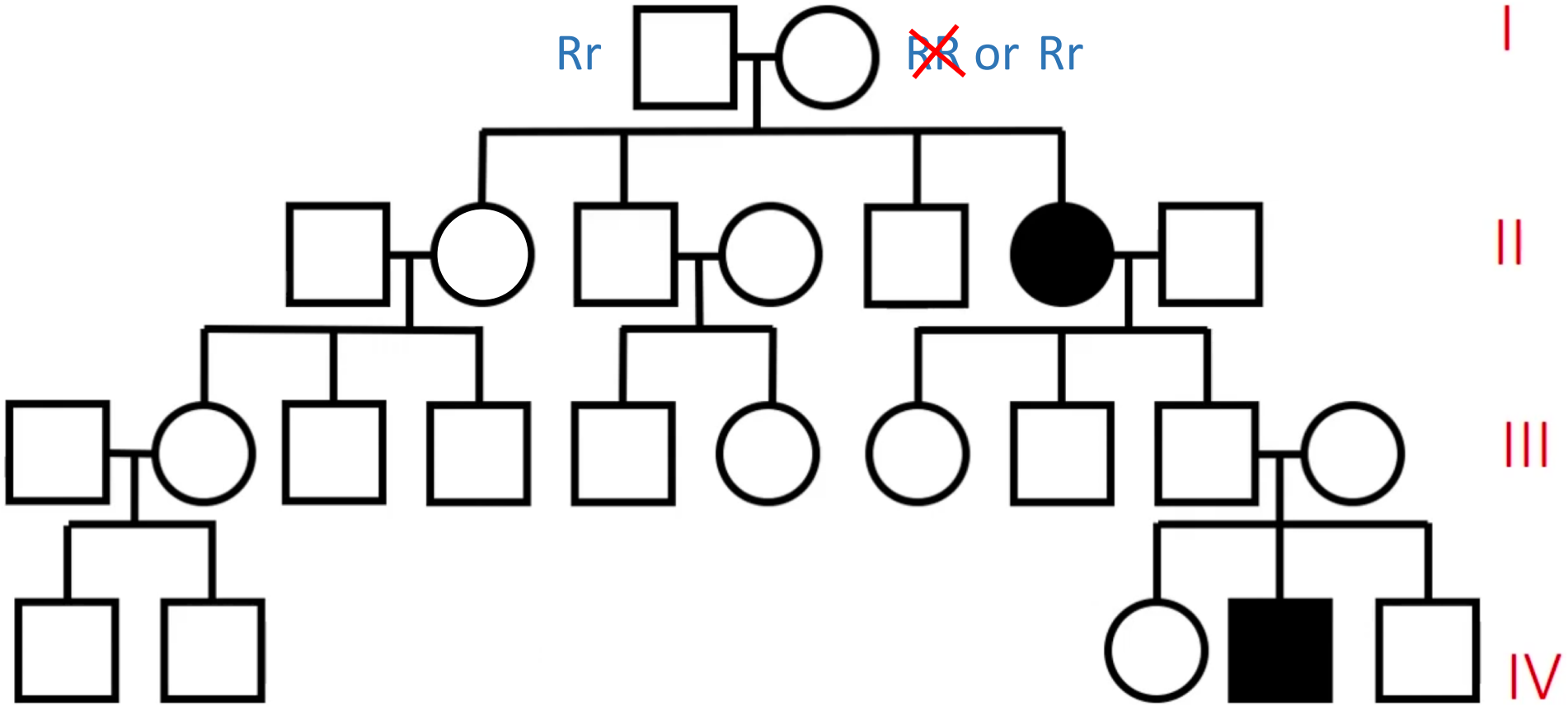


0% of F1 would be affected (none has genotype rr)



25% of F1 (1:3) would be affected (only 1 has rr)

Pedigree for autosomal recessive trait (with genotypes)



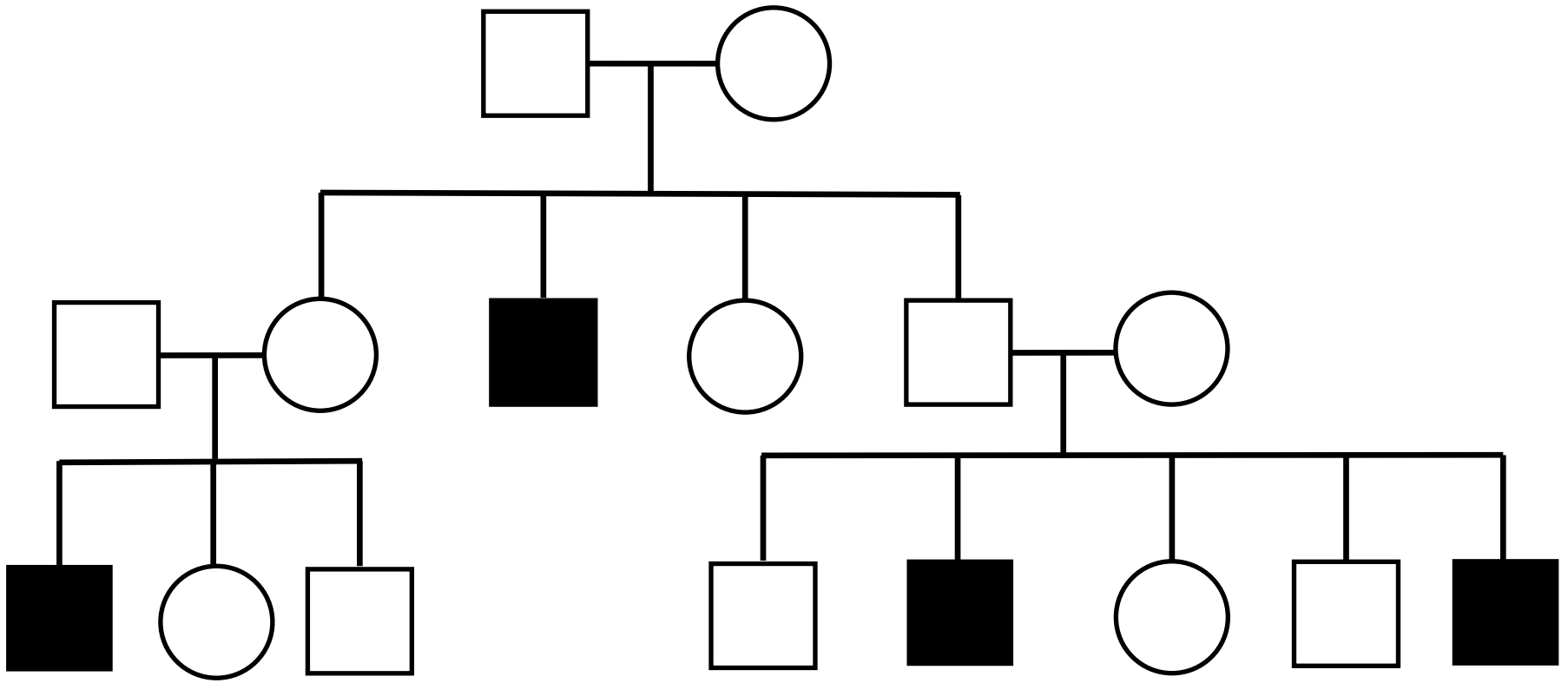
Note: you may not always get ratios identical to predicted due to probability of using specific gametes

Pedigree for X-linked recessive trait

Clues

X-linked: only males affected

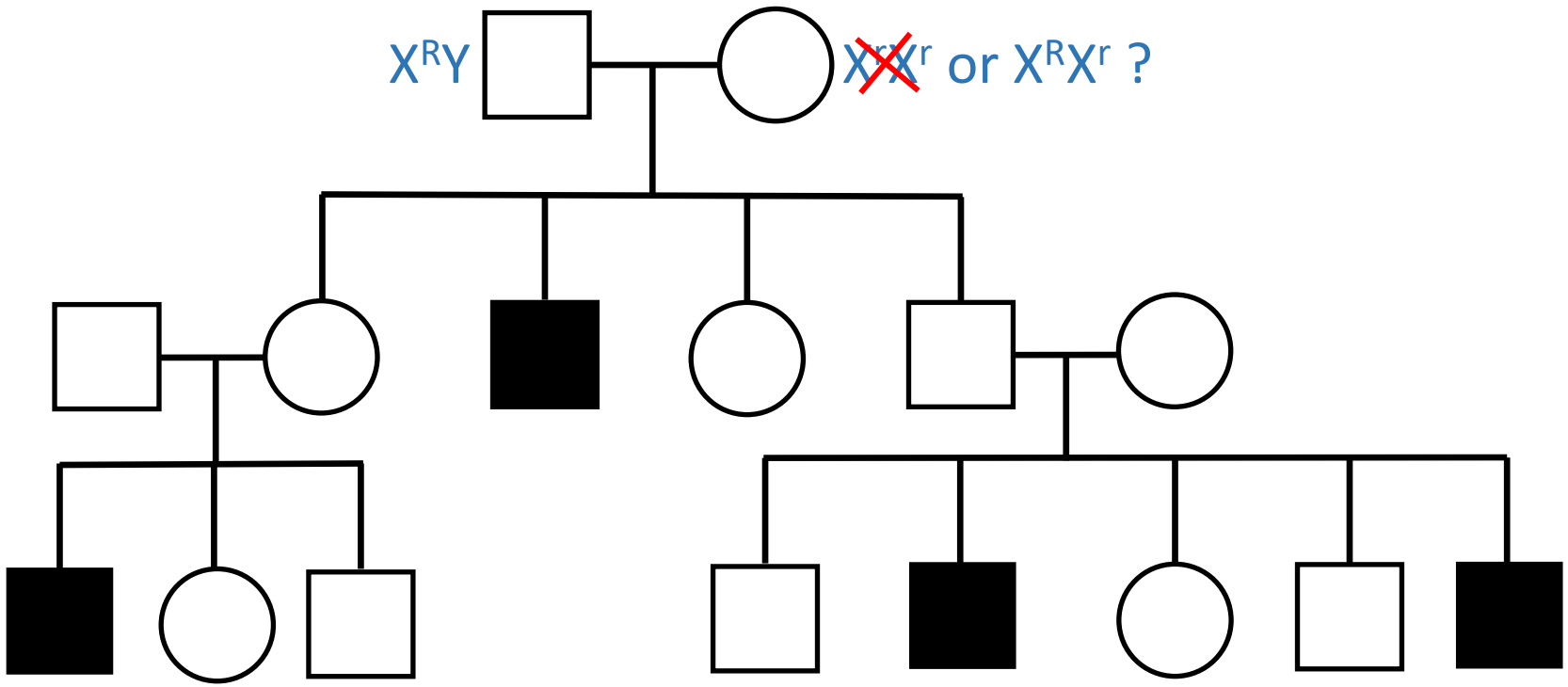
Recessive: affected child(ren) from unaffected parents not many affected offspring



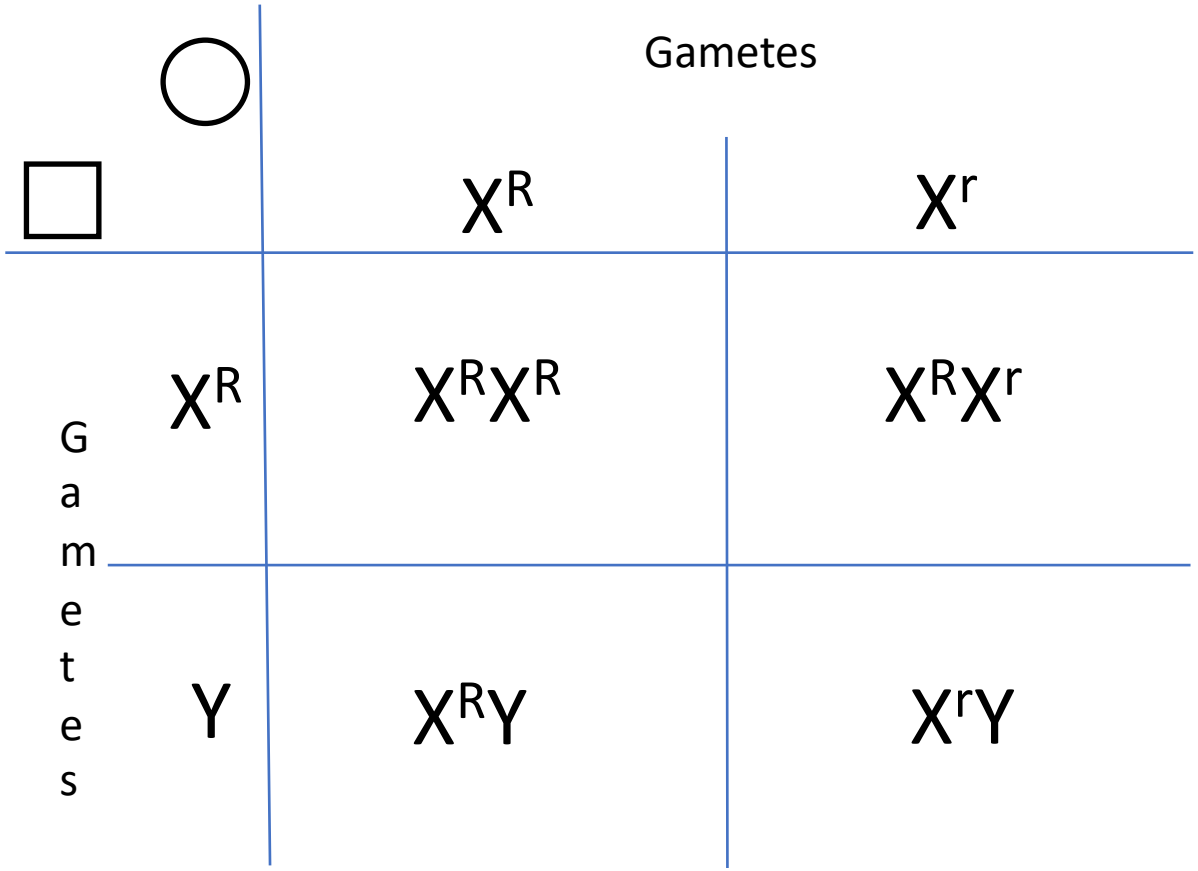
Pedigree for X-linked recessive trait (with genotypes)

Remember: it is the recessive allele r that expresses the disease

X^rX^r can be immediately excluded otherwise the mother would have the trait

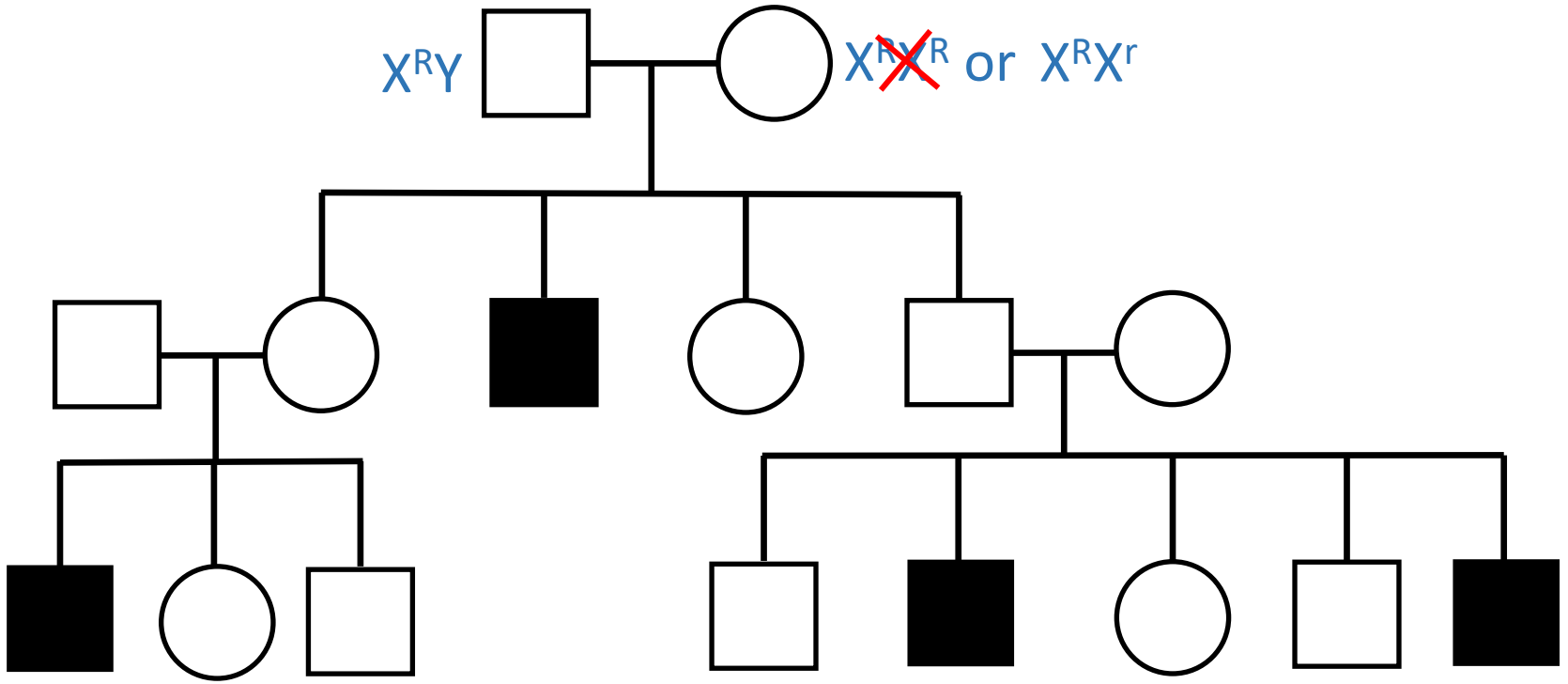


Punnett squares for gen II (gen 1 = parents)



50% of MALE F1 (1:1) would be affected (has genotype X^rY)

Pedigree for X-linked recessive trait (with genotypes)



Note: you may not always get ratios identical to predicted due to probability of using specific gametes

Notes

- The discussed pedigrees are simple and ideal
- You may not always get ratios identical to predicted due to probability of using specific gametes
- You may not have enough offspring at F1 to interpret pedigree
  - You need to analyze F2, F3, ...

Pedigrees

- Take assignment 16: Pedigrees