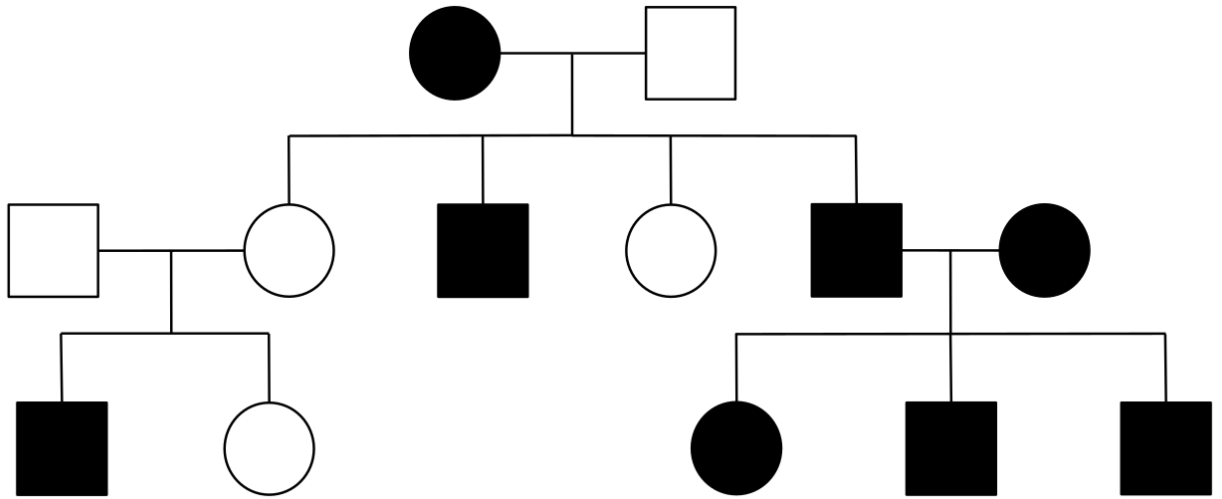
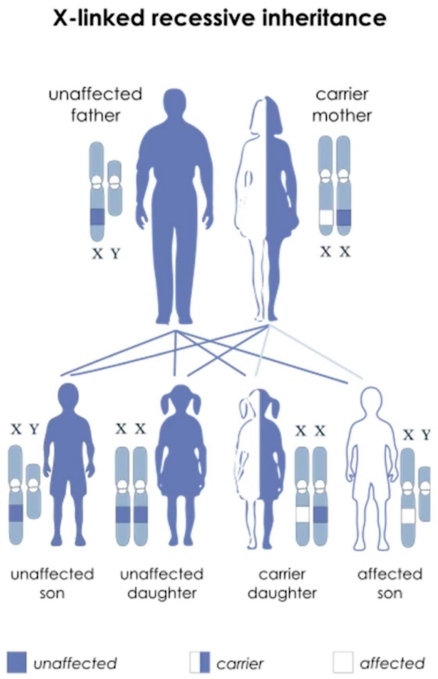
Lesson 16
Pedigrees



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



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

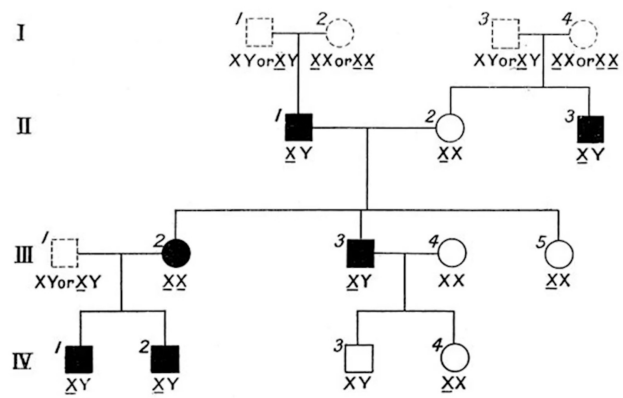


FIG. 21.—PEDIGREE CHART OF COLOUR-BLINDNESS.

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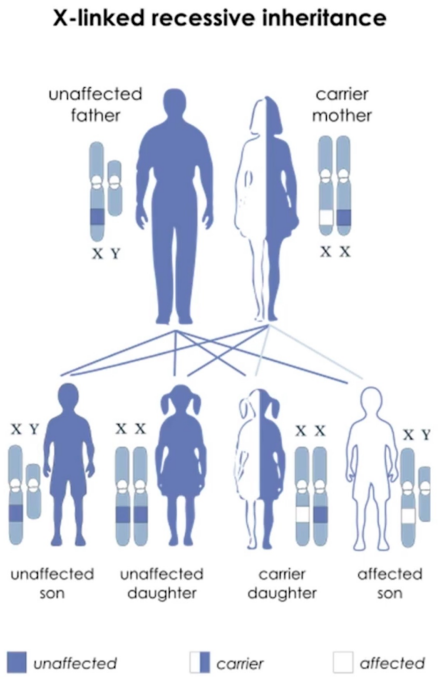
- Pedigrees track traits through generations, helping us understand inheritance patterns
- They are used in genetic counseling, medicine, breeding programs, and forensic science
- Pedigree analysis **helps diagnose genetic disorders** and **predict inheritance risks**



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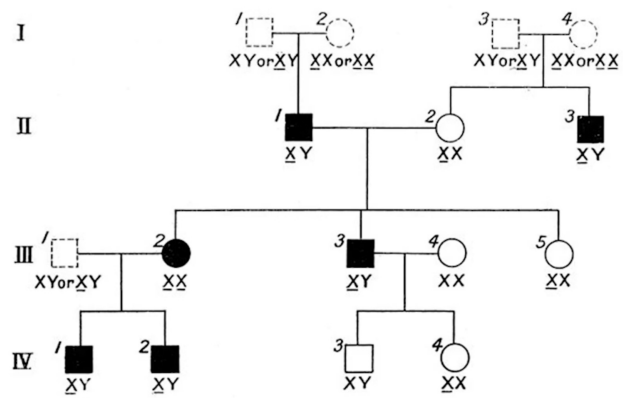

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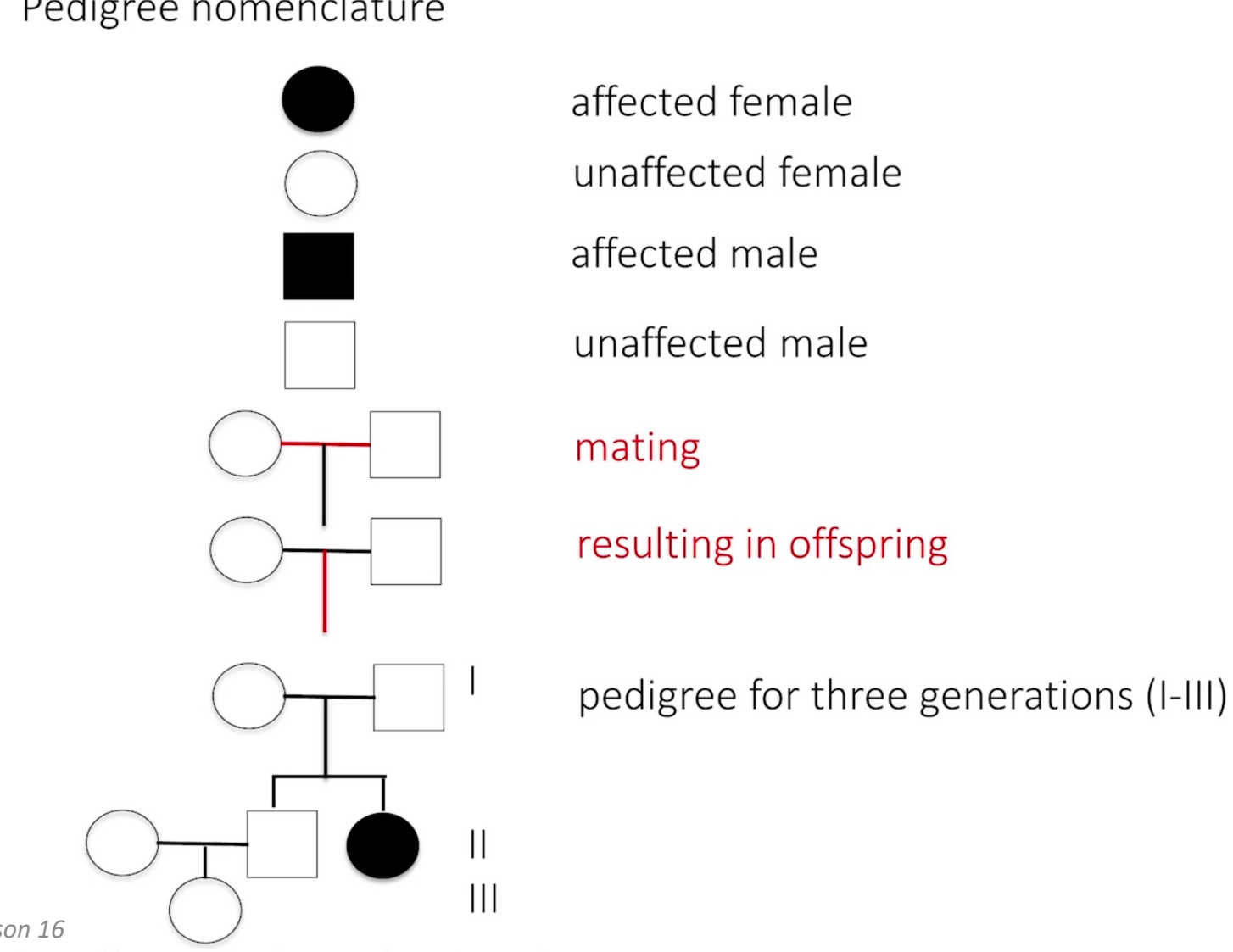
Pedigrees

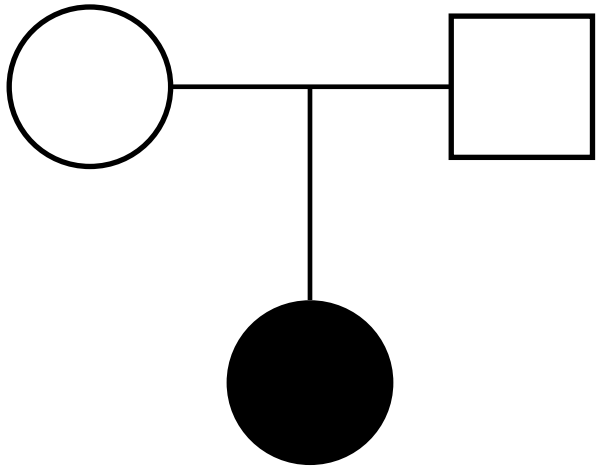
- **Pedigrees** = **genetics from family history**
- Conventions in pedigree writing:
  - ◯ female
  - □ male
  - ●■ the trait you are looking at is present (*e.g.*, a disease, brown eyes, etc.)

More pedigree nomenclature



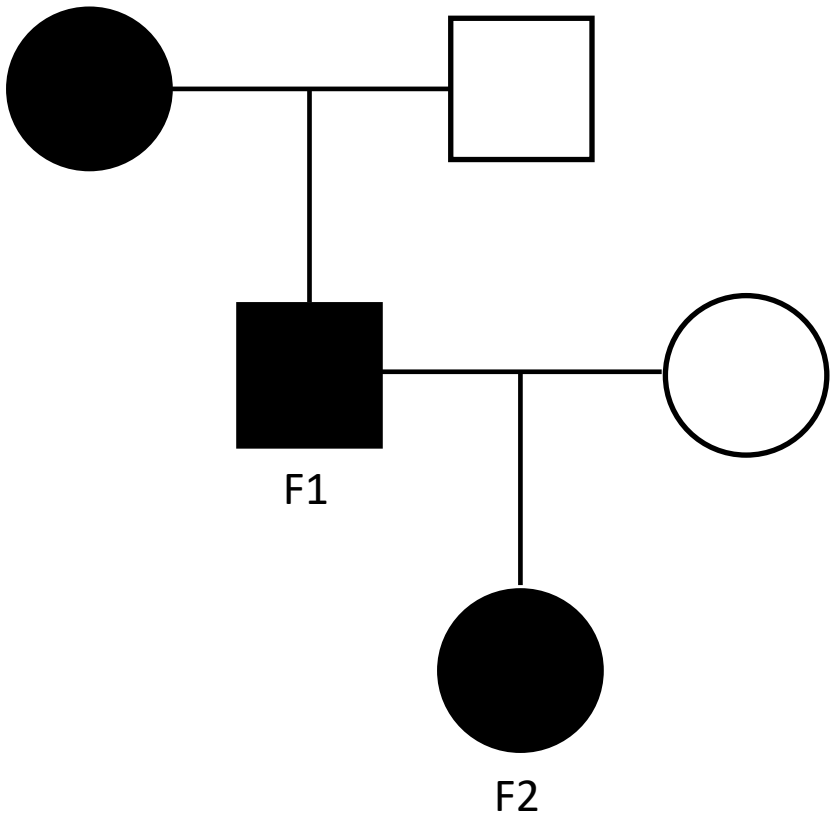
Pedigrees - 1

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



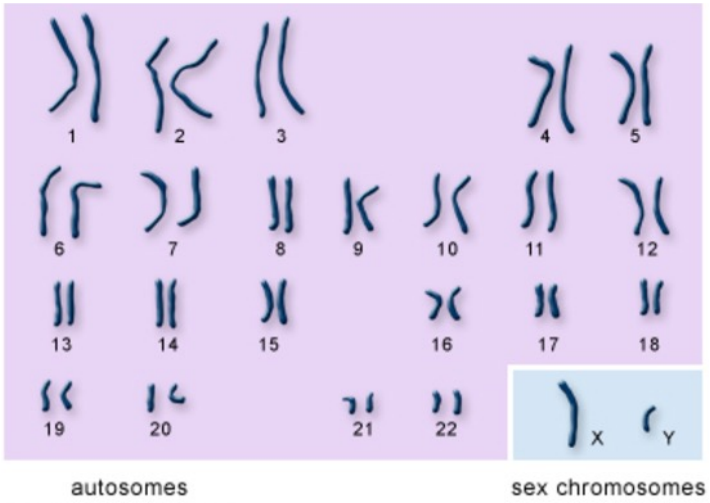
Pedigrees - 2

- 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**



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

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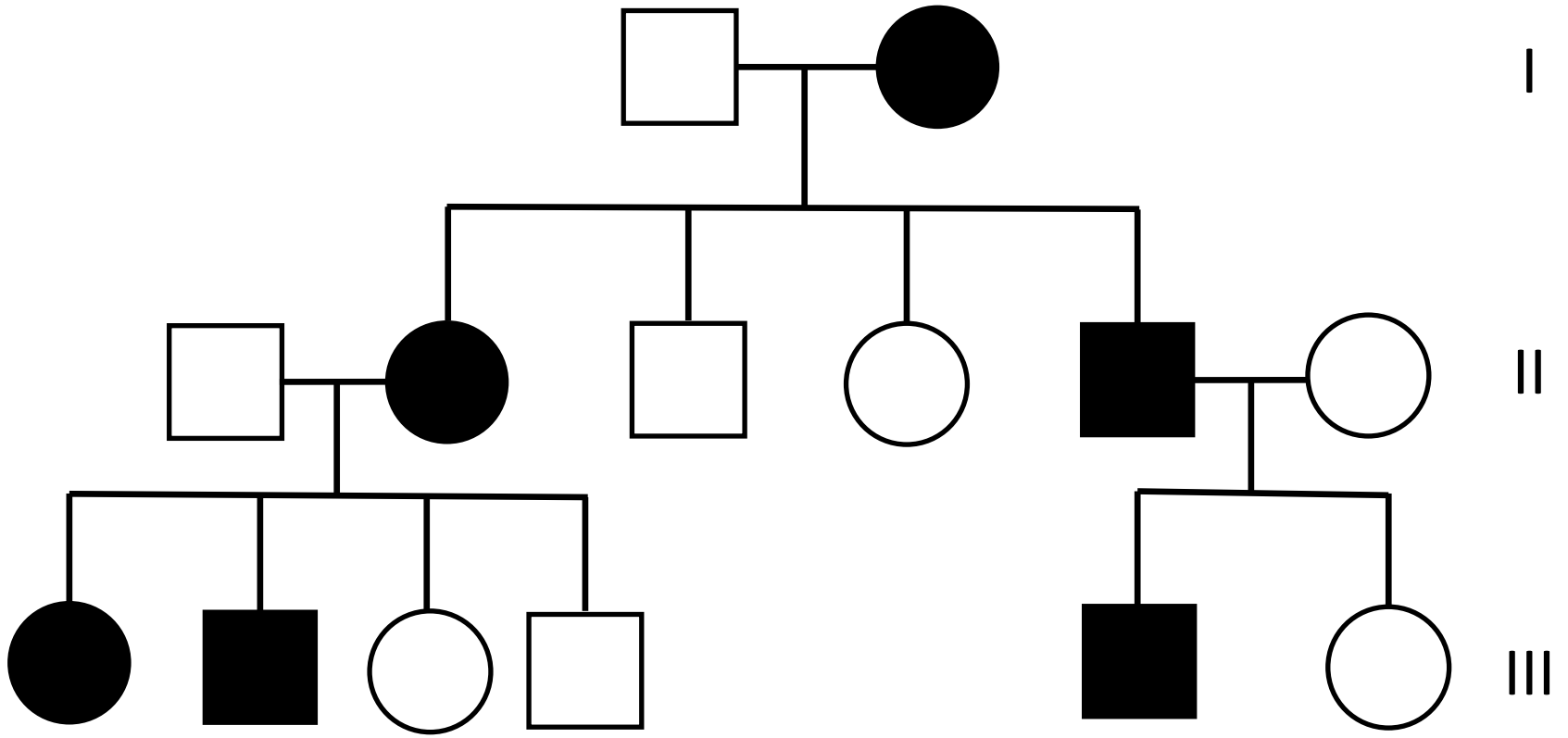
- 3 types of of pedigree based on autosomes or sex chromosomes:
- Autosomal recessive
- Autosomal dominant } Males and females affected equally
- X-linked recessive → Males affected more than females
  - there is only one X (the other allele is Y) and is affected –> the trait will show itself)
  - It 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

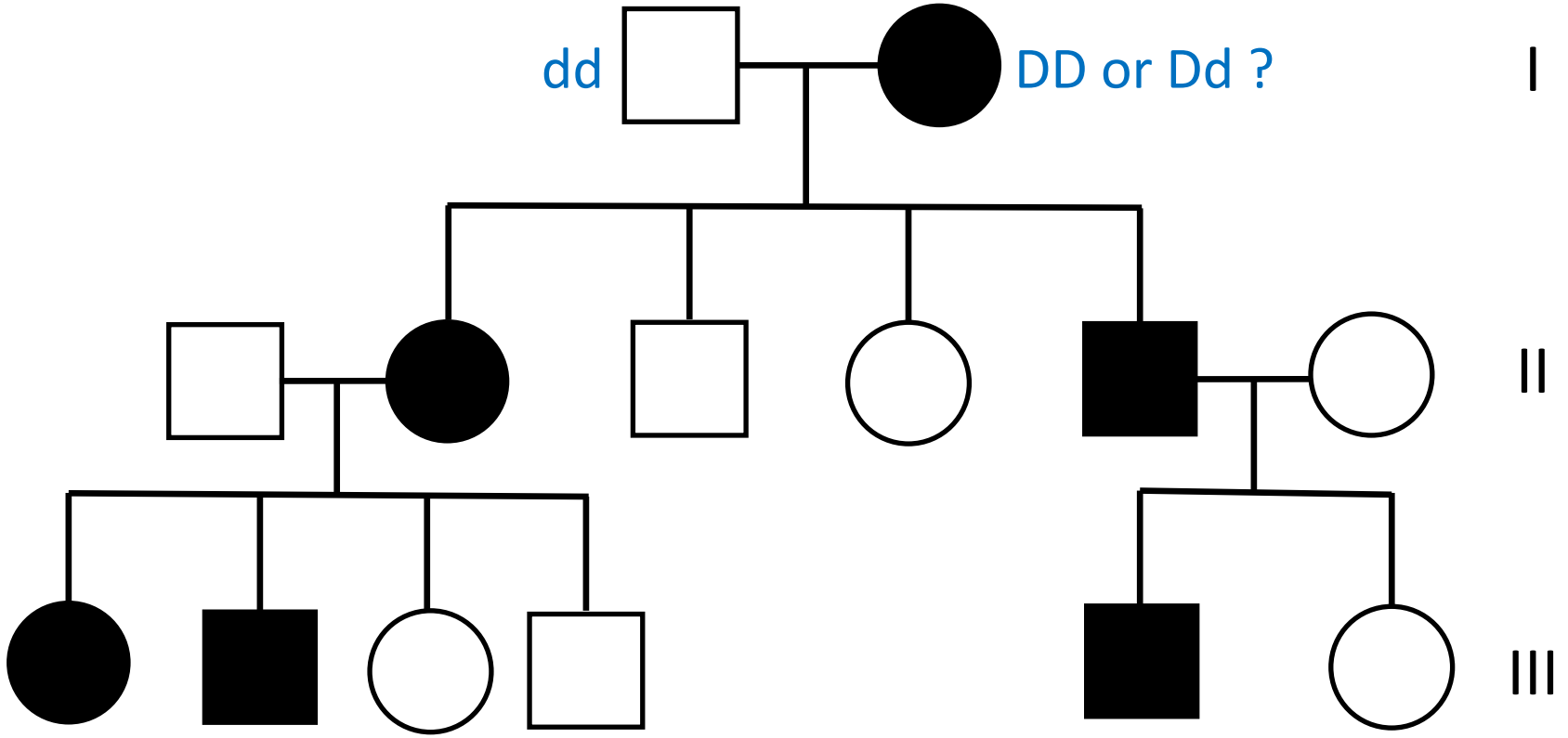
Clues

Autosomal: both males and females are affected

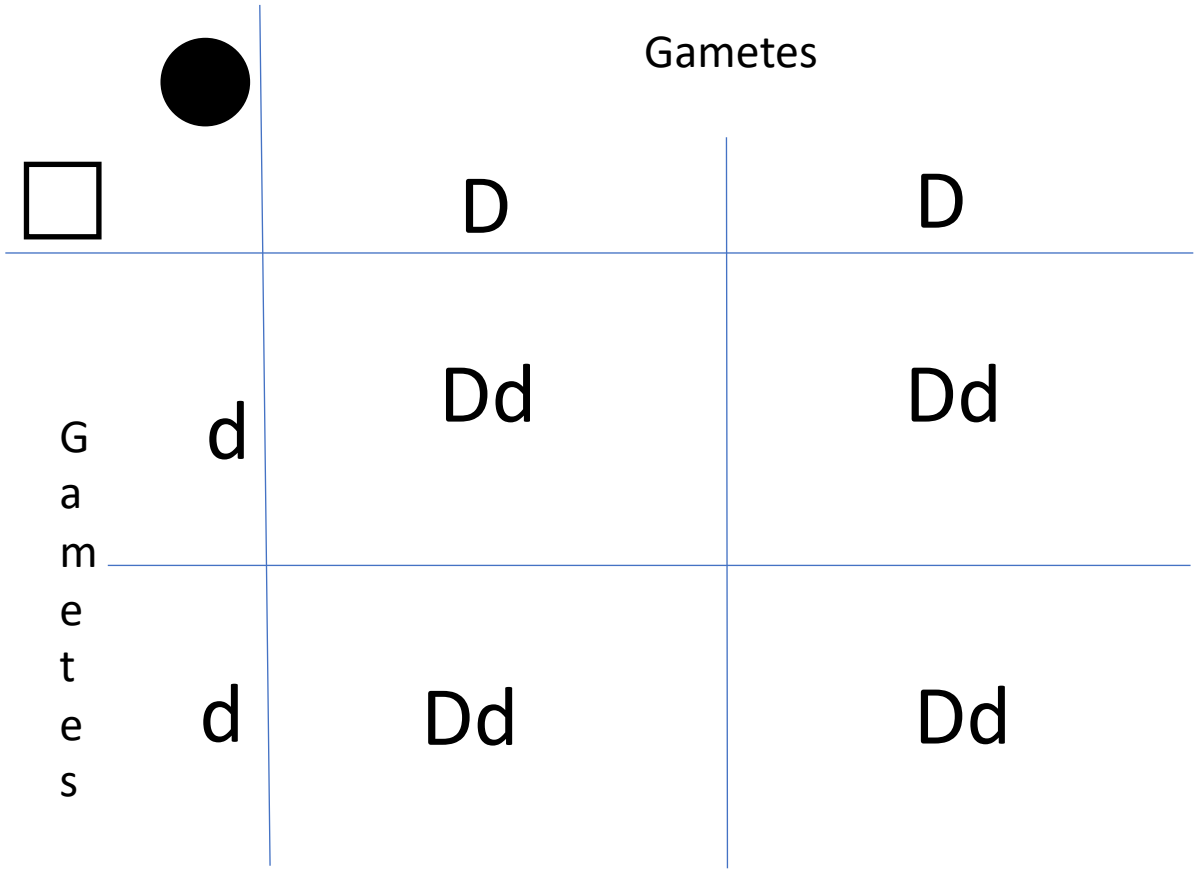
Dominant: <u>every affected</u> child has <u>one affected parent</u>



Pedigree for autosomal dominant trait (with genotypes)



Punnett squares for gen II (gen 1 = parents)



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

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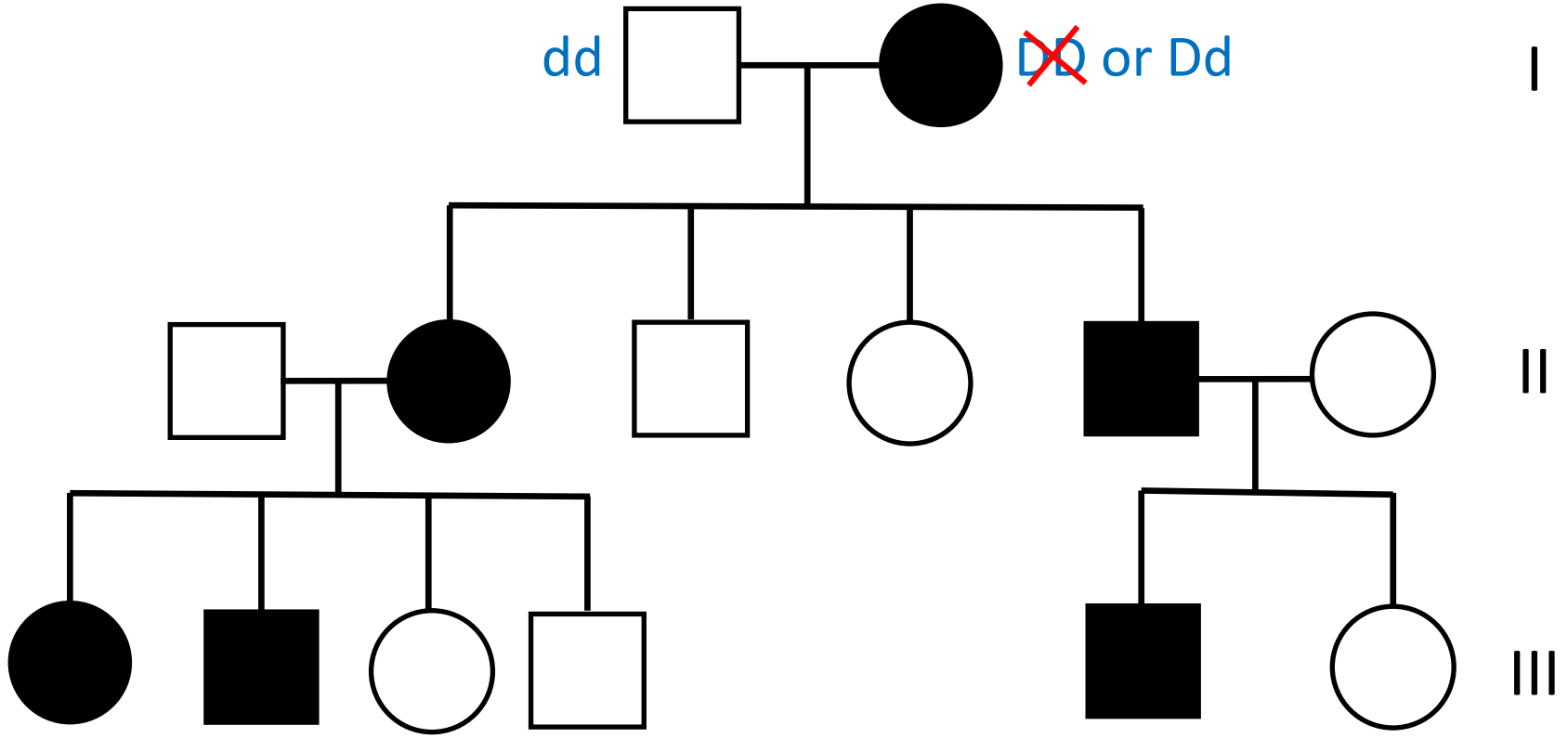
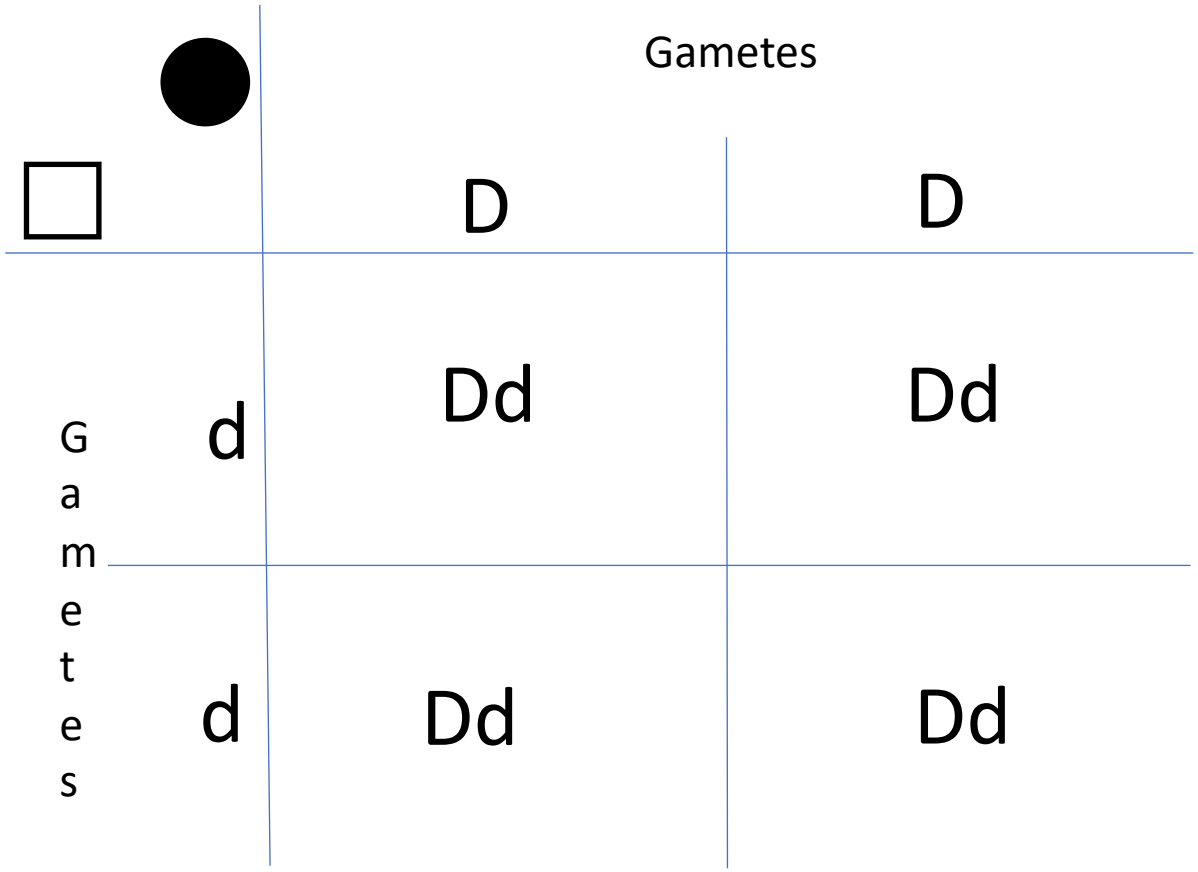
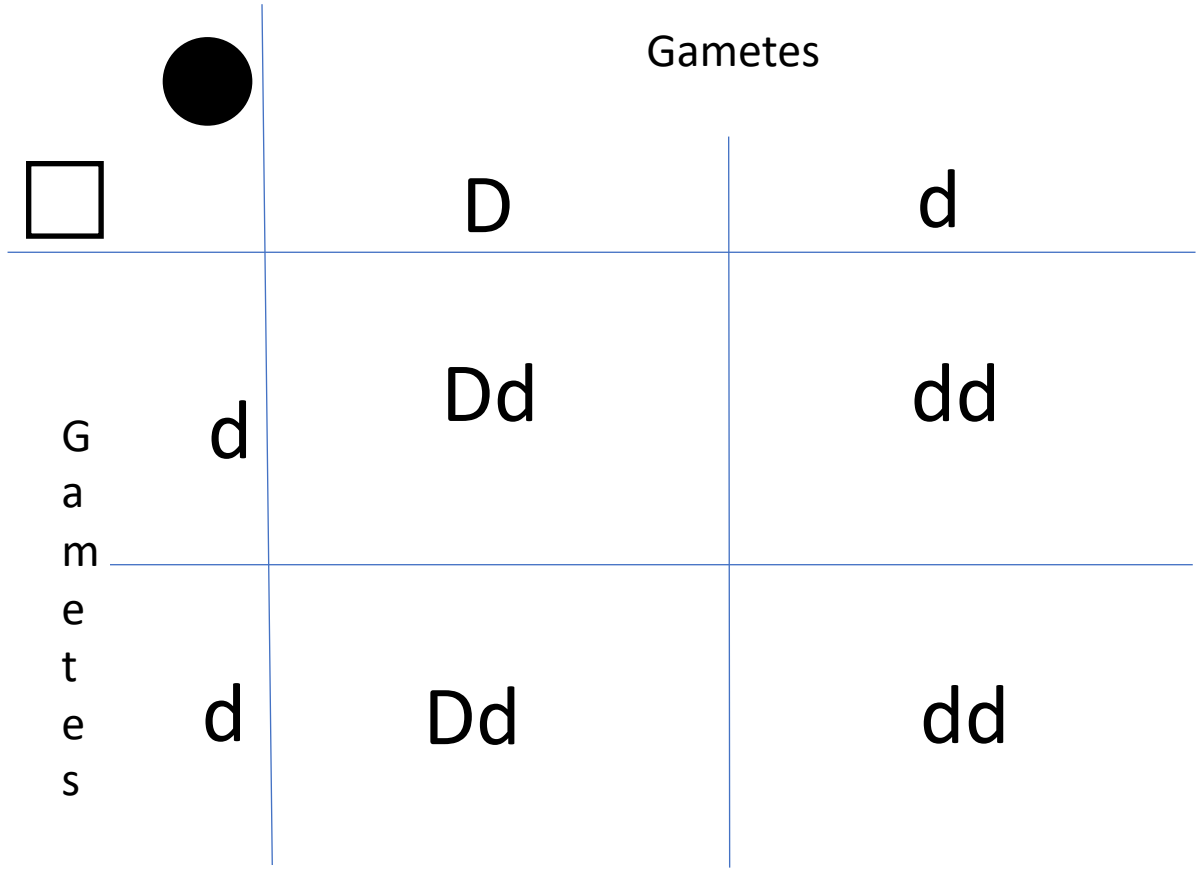


Punnett squares for gen II (gen I = parents)

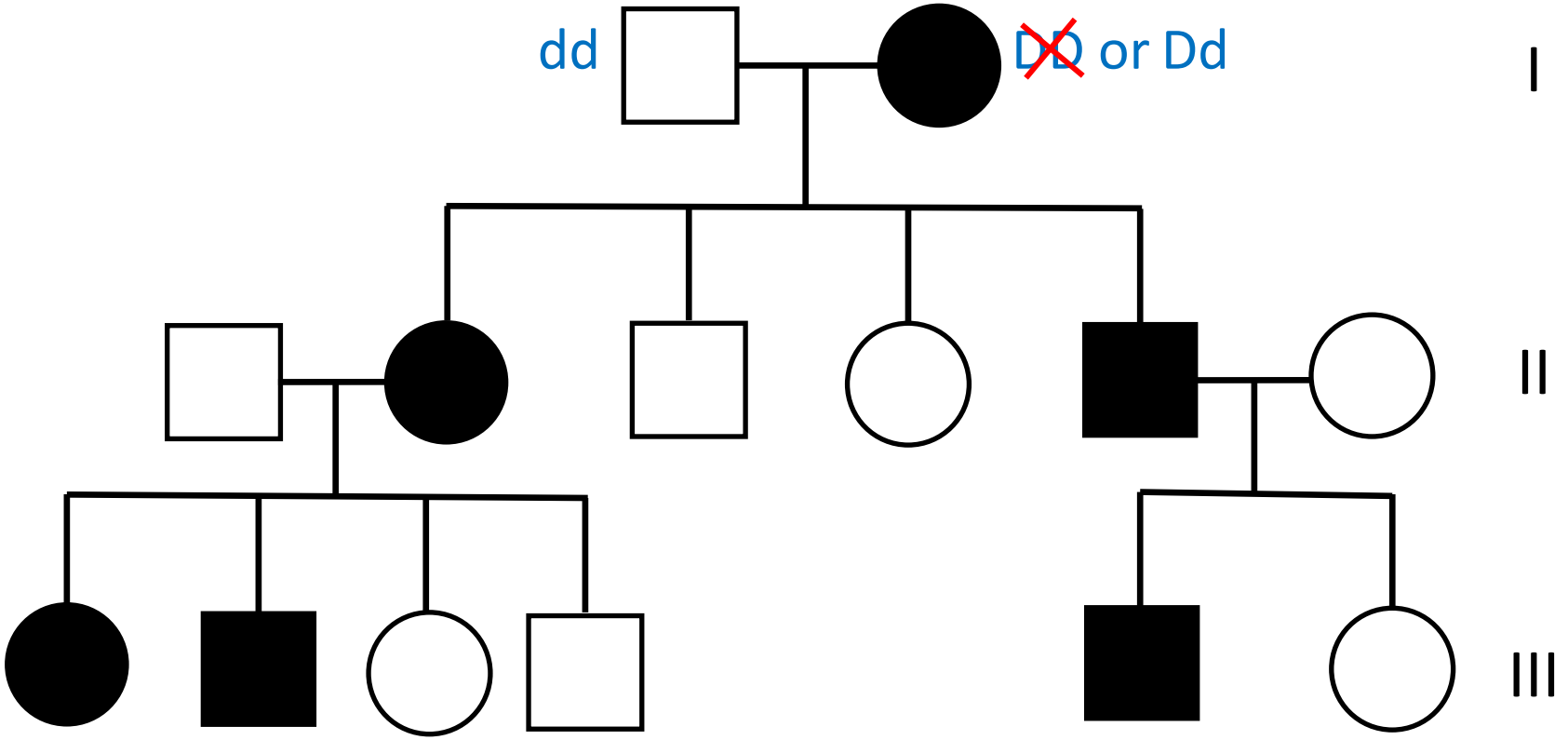


100% of F1 would be affected (all have the dominant allele D)



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

Pedigree for autosomal dominant trait (with genotypes)



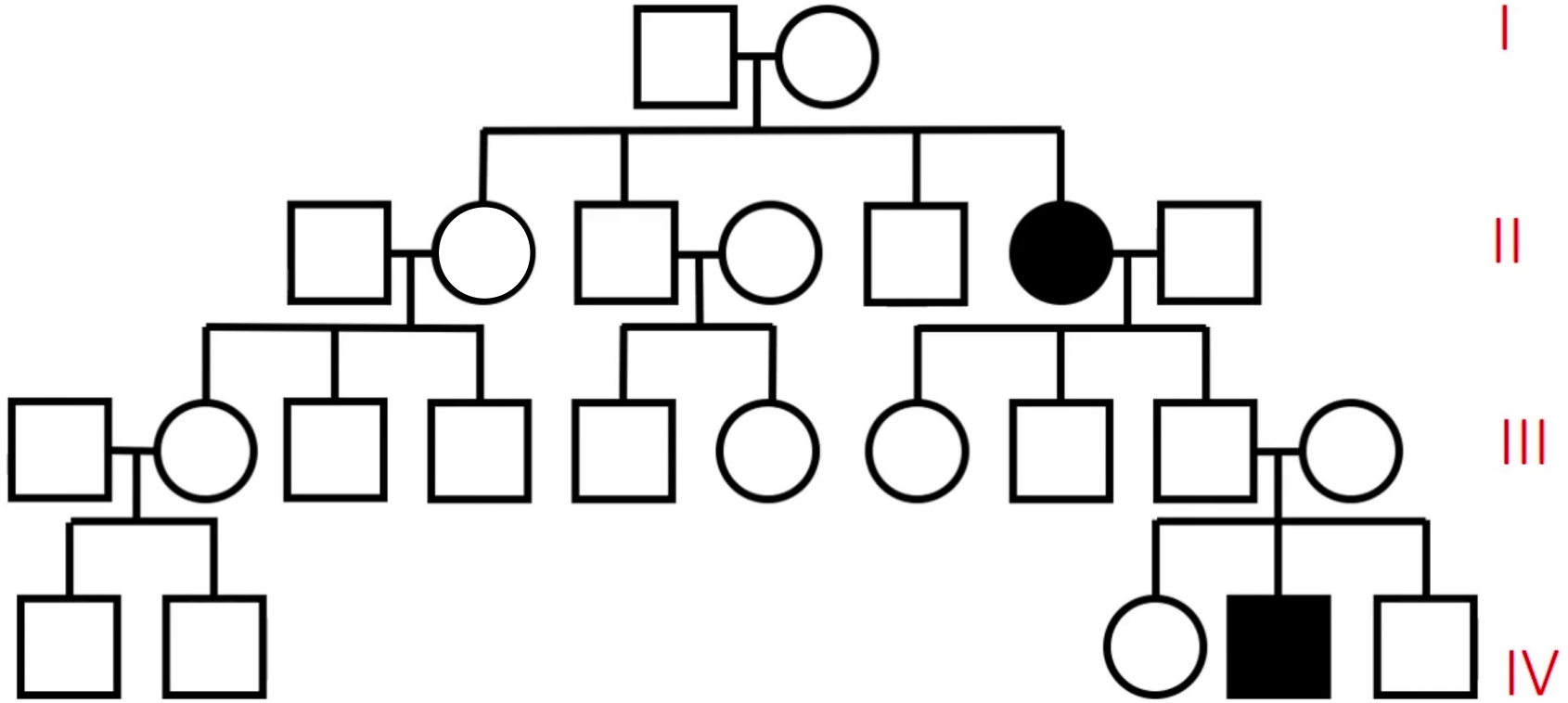
Note: you may not always get ratios identical to predicted*

Pedigree for autosomal recessive trait

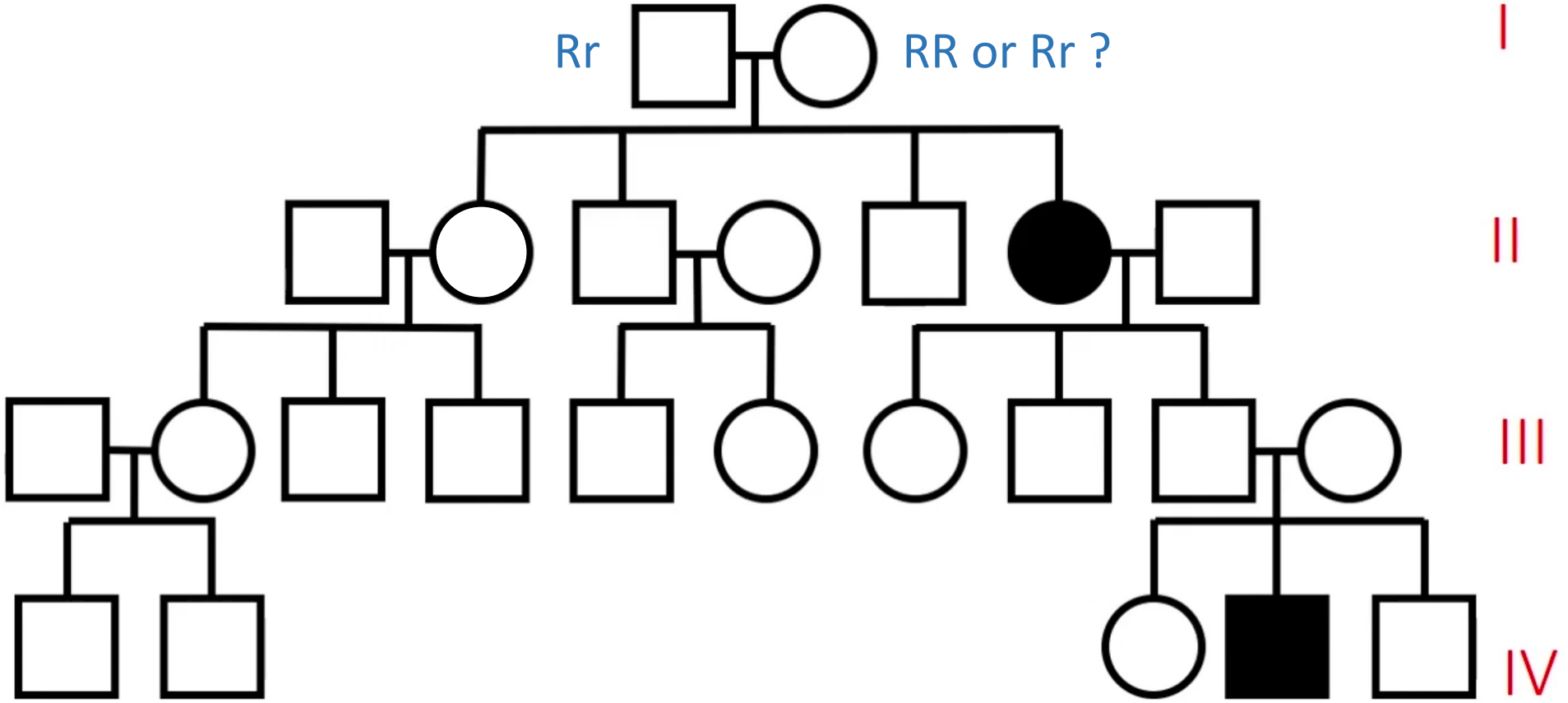
Clues

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)



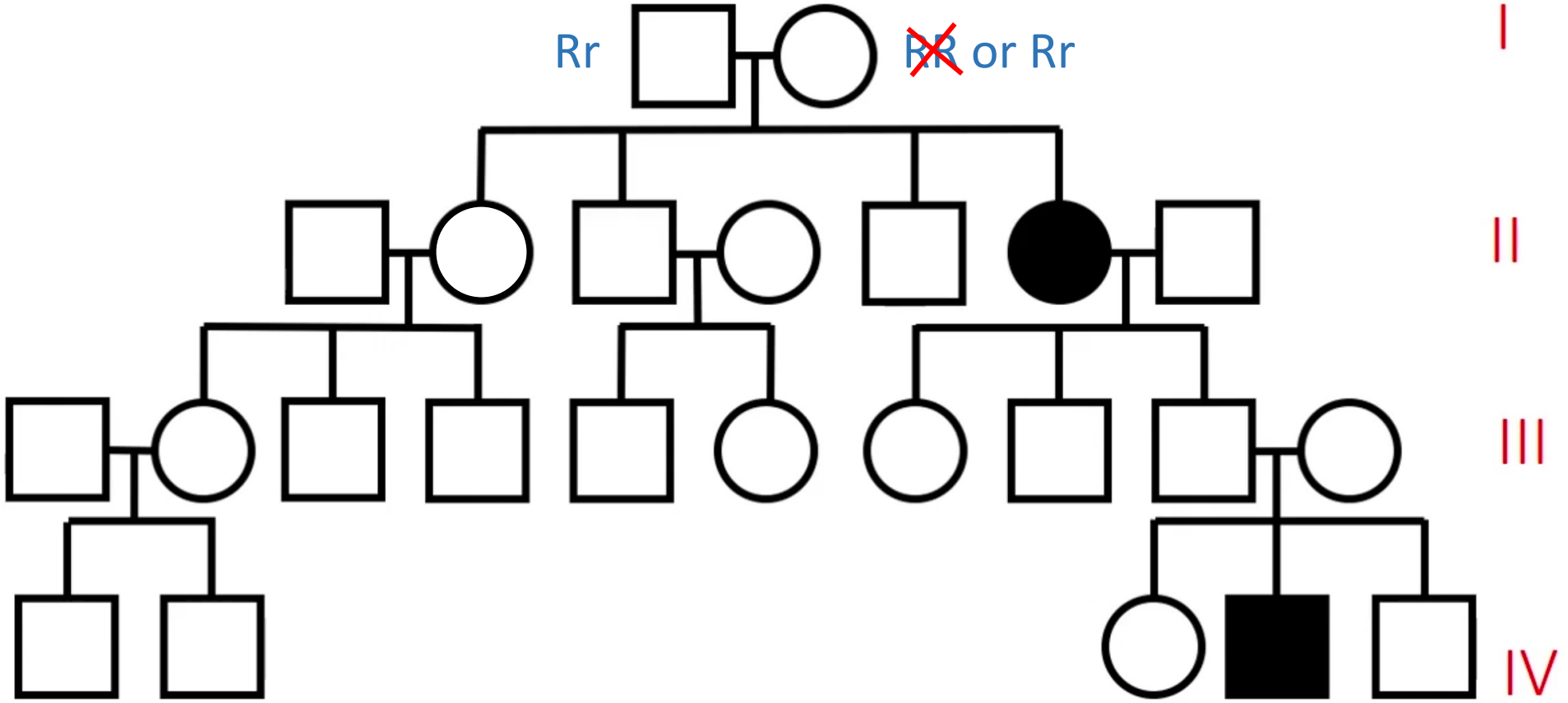
Remember: it is the recessive allele r that expresses the disease

Punnett squares for gen II (gen 1 = parents)

♂ Gametes	♀ Gametes: R	♀ Gametes: R
R	RR	RR
r	Rr	Rr

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

Pedigree for autosomal recessive trait (with genotypes)



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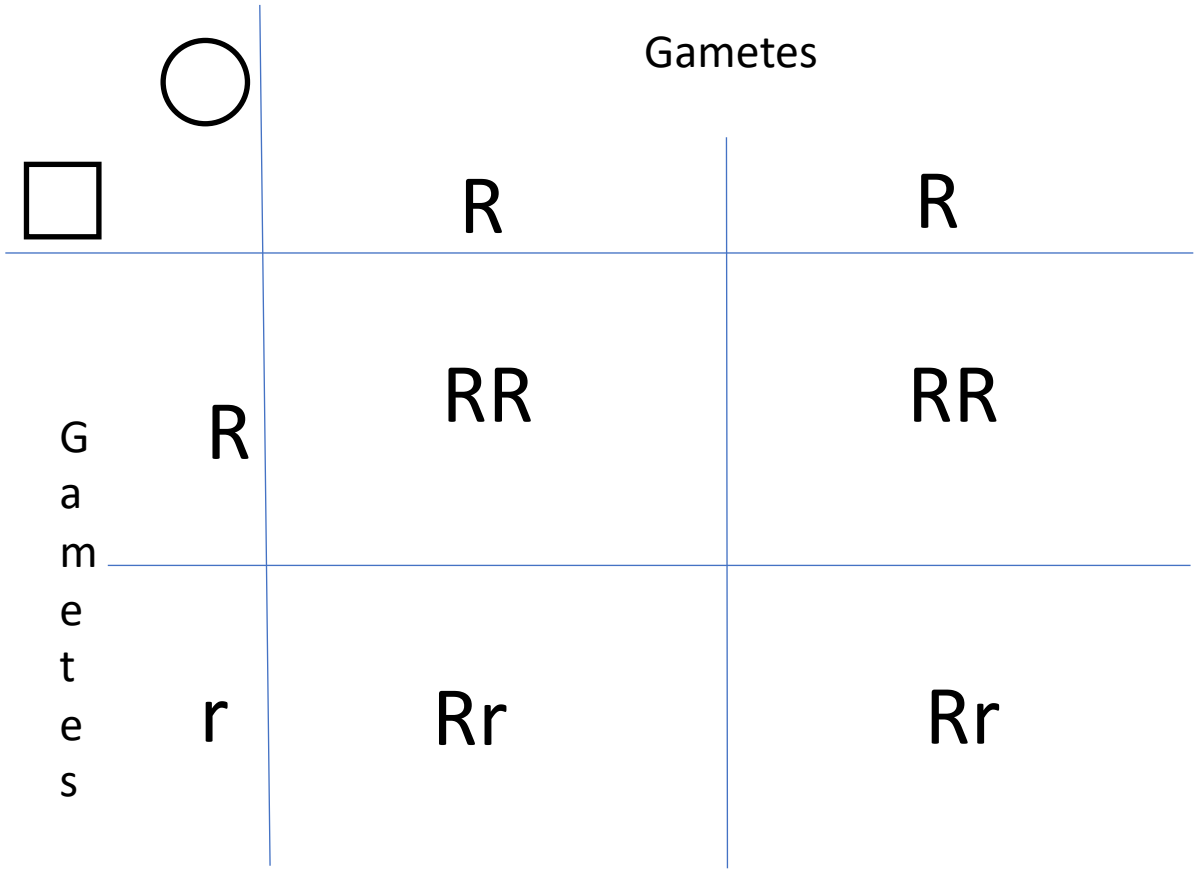

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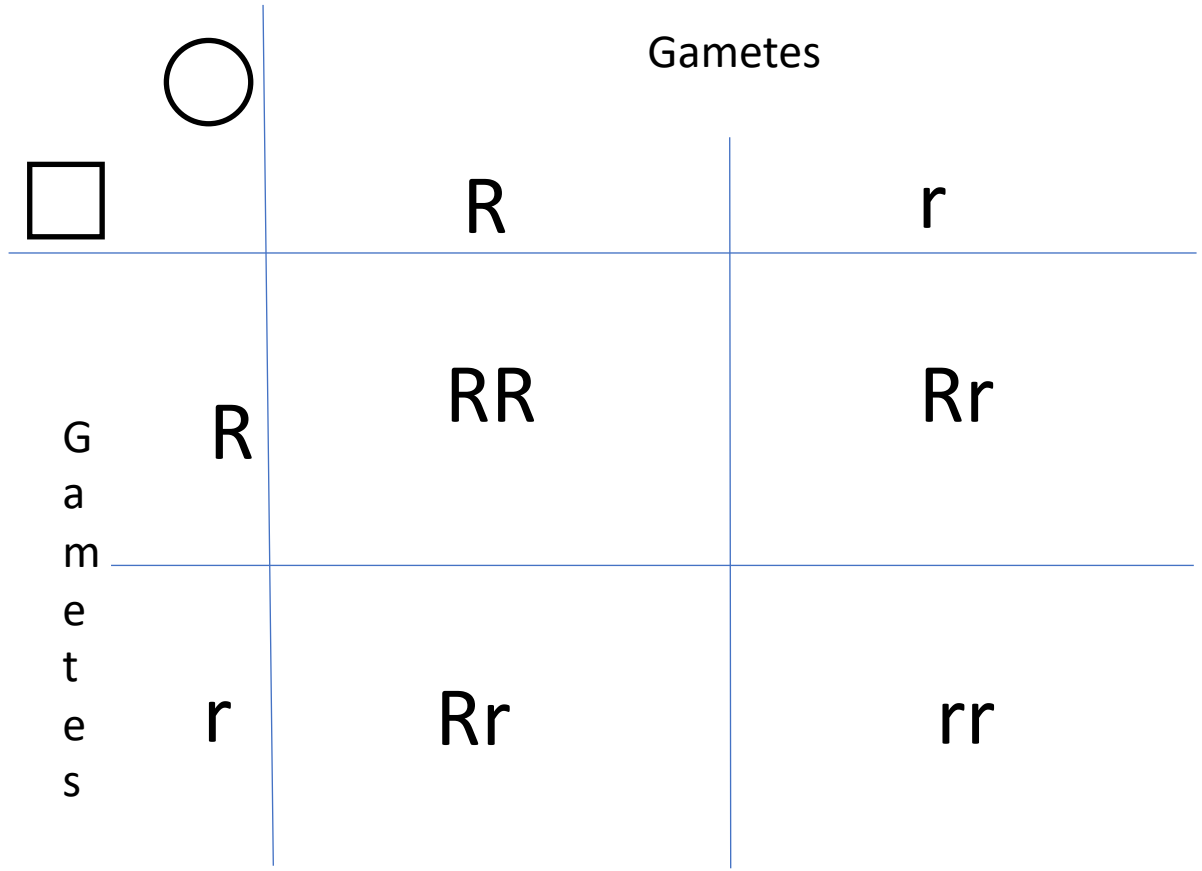
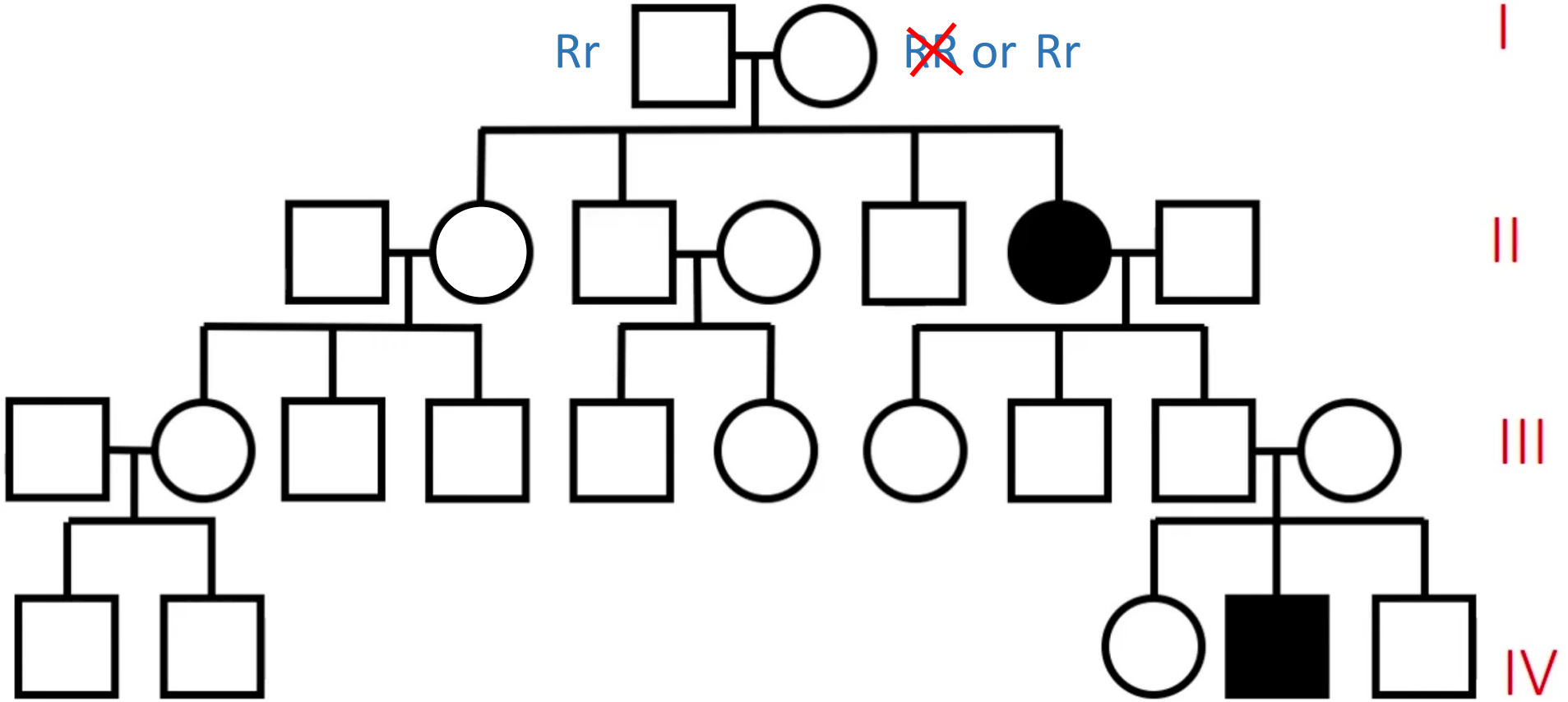


25% of II (1:3) would be affected (only 1 has rr)

Pedigree for autosomal recessive trait (with genotypes)



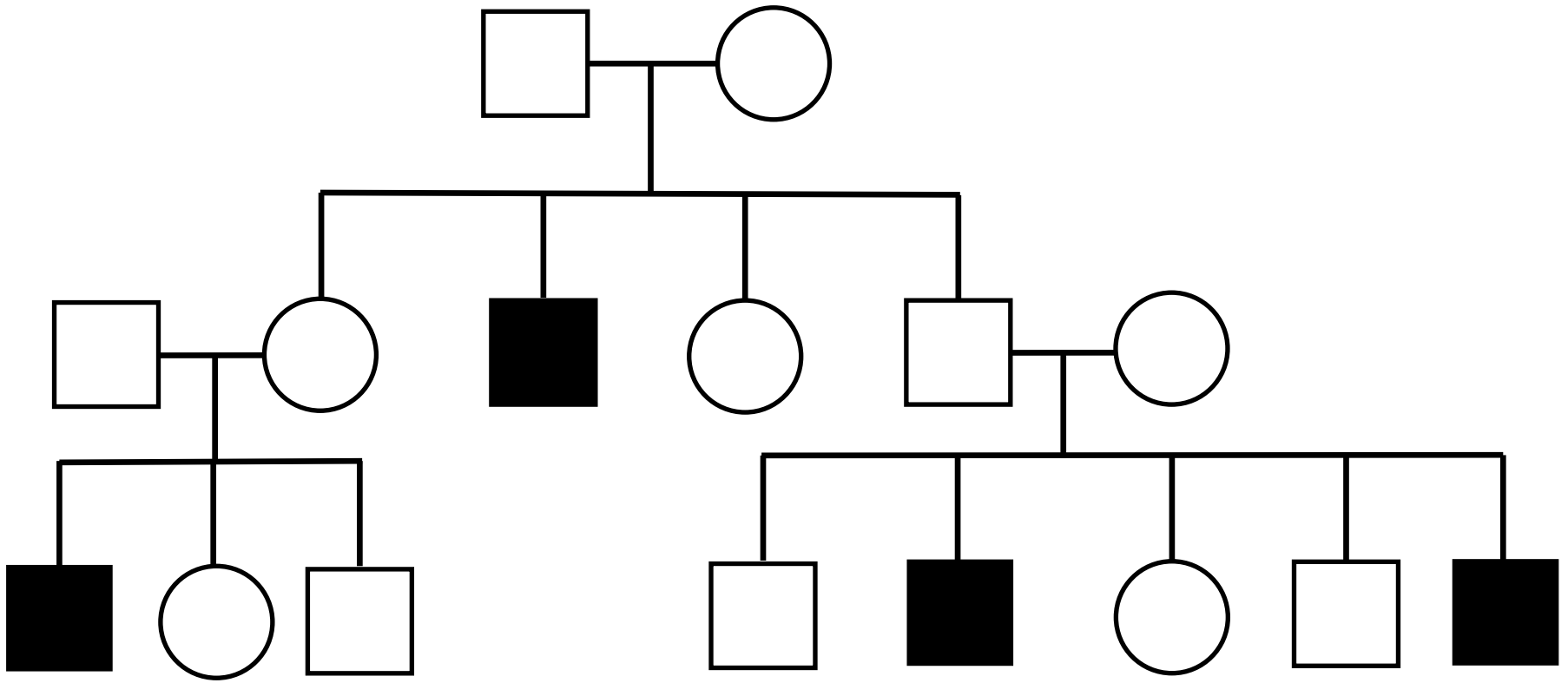
Note: you may not always get ratios identical to predicted*

Pedigree for X-linked recessive trait

Clues

X-linked: only males affected

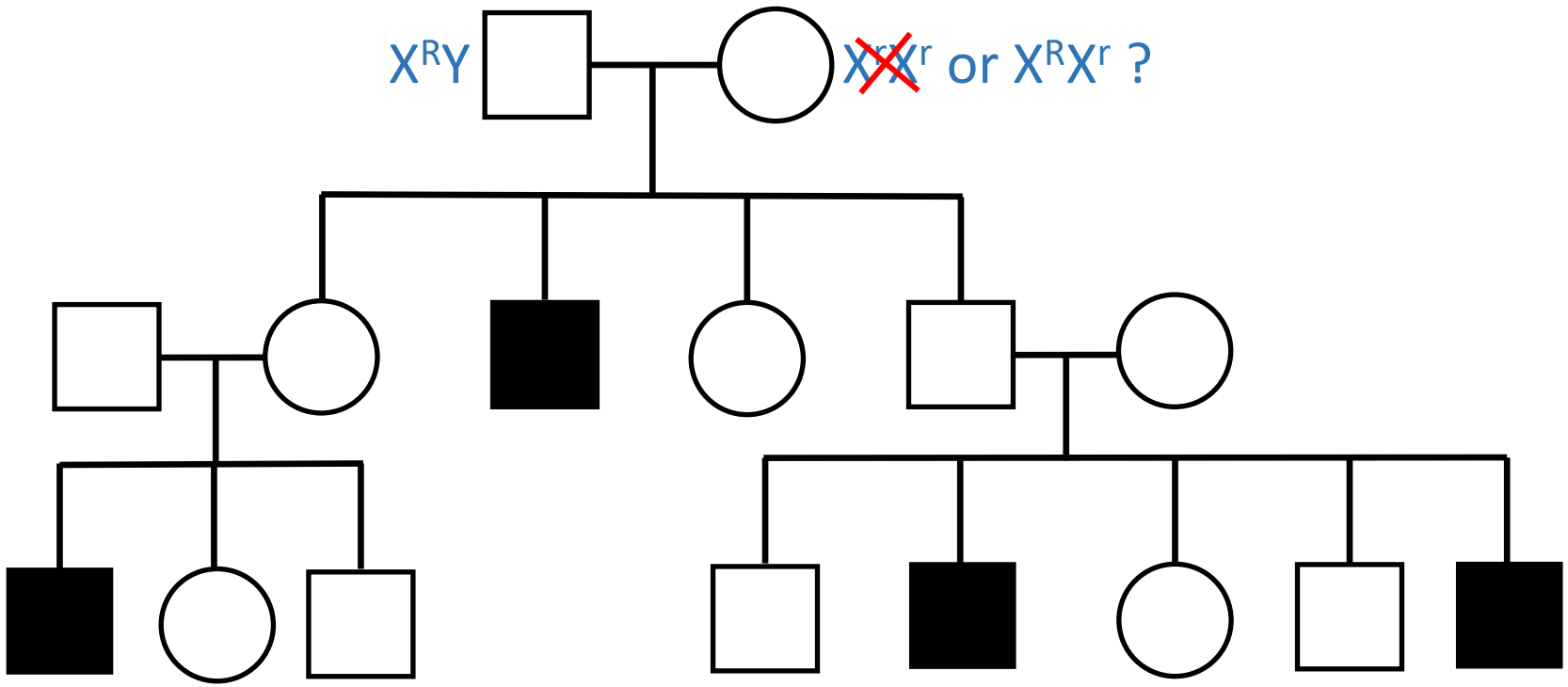
Recessive: affected child(ren) from unaffected parents not many affected offspring



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



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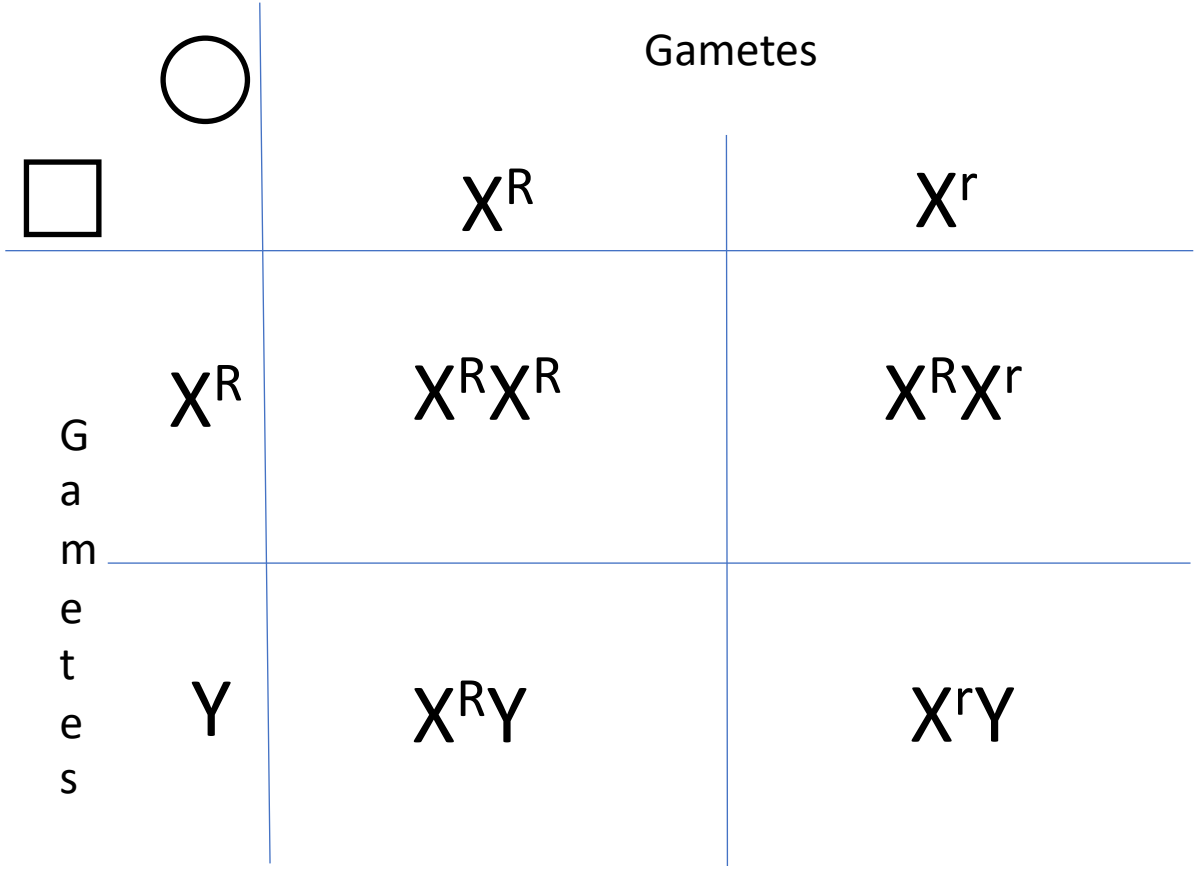
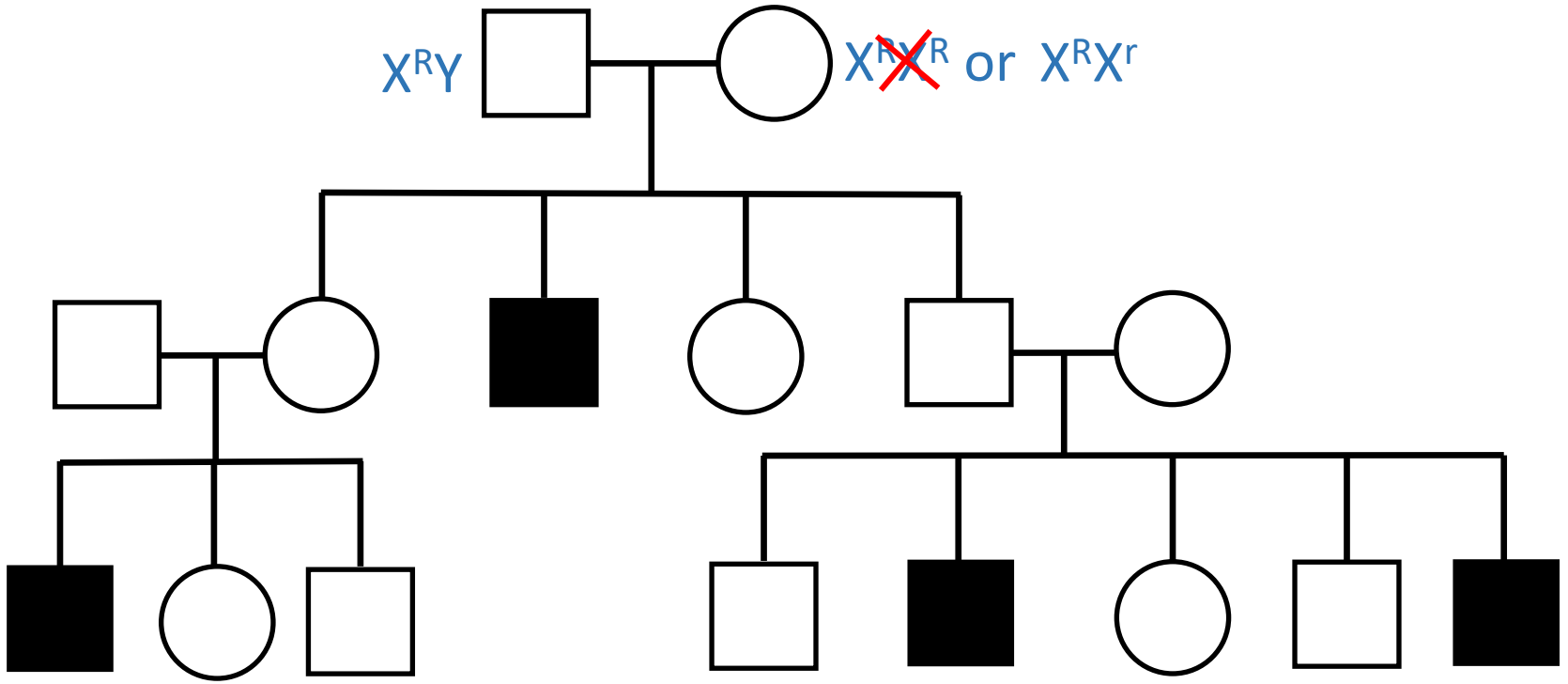


50% of MALE II (1:1) would be affected (has genotype X^rY)

Pedigree for X-linked recessive trait (with genotypes)



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Notes

- The discussed pedigrees are simple and ideal
- You may not always get ratios identical to predicted due to non-Mendelian inheritance patterns, which include (among others):
  - Codominance and incomplete dominance
  - Polygenic traits: some traits are determined by the combined effect of more than one pair of genes → polygenic (or continuous) traits (an example of this is human stature)
  - Multiple-allele series: the ABO blood type system is also an example of a trait that is controlled by more than just a single pair of alleles
  - Modifying and regulator genes: modifying genes alter how certain other genes are expressed in the phenotype (for instance, here is a dominant cataract gene which will produce varying degrees of vision impairment depending on the presence of a specific allele for a companion modifying gene)

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  - Incomplete penetrance: some genes are incompletely penetrant
    - their effect does not normally occur unless certain environmental factors are present. For example:
    - you may inherit the genes that are responsible for type 2 diabetes but never get the disease unless you become greatly overweight, persistently stressed psychologically, or do not get enough sleep on a regular basis;
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  - Pleiotropy: a single gene may be responsible for a variety of traits. The complex of symptoms that are collectively referred to as sickle-cell trait or sickle-cell anemia, is an example.
    - A single gene results in irregularly shaped red blood cells that painfully block blood vessels, cause poor overall physical development, as well as related heart, lung, kidney, and eye problems

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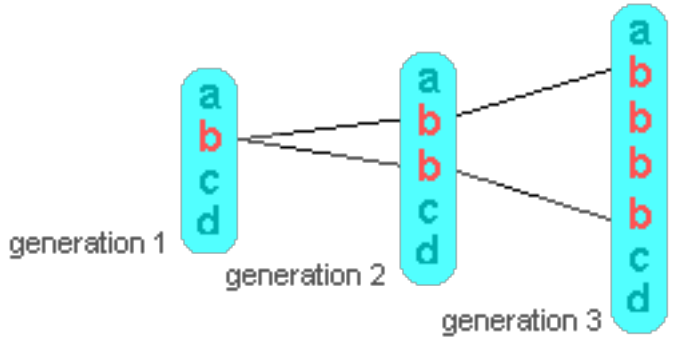

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  - Stuttering alleles: some genetically inherited diseases have more severe symptoms each succeeding generation due to segments of the defective genes being doubled in their transmission to children (typically fatal degenerative nerve disorders

- You may not have enough offspring at F1 to interpret pedigree
  - You need to analyze F2, F3, ...