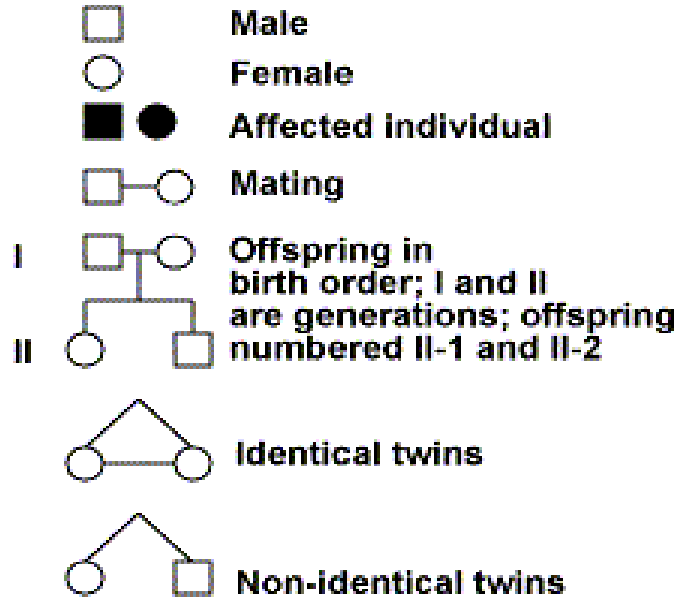
Pedigrees NOTES

OBJECTIVE: Analyze pedigrees to determine the genetic make-up of individuals and modes of inheritance.

All of the conclusions regarding mode of inheritance (dominant versus recessive) we have discussed so far have been obtained from analyzing the results of controlled crosses. In some situations, we do not have the opportunity to perform controlled crosses (for example, with human beings). In such cases, scientists analyze an existing population in an approach called **pedigree analysis**. **For further information see also** https://www.migeneticsconnection.org/genomics/family%20history/family%20history.htm



A series of symbols are used to represent different individuals within a pedigree is shown to the right.

Once phenotypic data is collected from several generations and the pedigree is drawn, careful analysis may allow you to determine the **mode of inheritance** for the trait being studied. Here are some rules to follow.

Determining whether a trait is AUTOSOMAL DOMINANT or AUTOSOMAL RECESSIVE

In the case of **DOMINANT** traits,

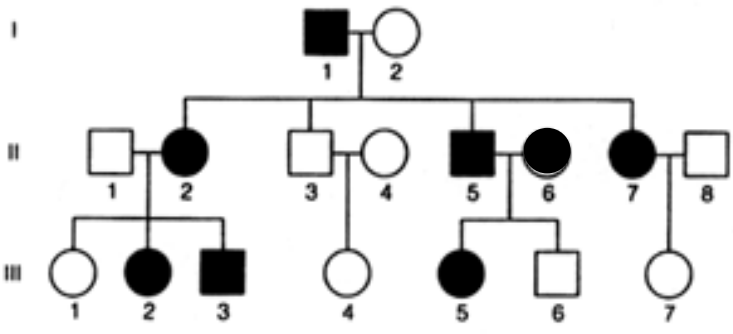
- the phenotype generally appears in very generation
- **An AFFECTED individual MUST have AT LEAST ONE AFFECTED parent**
- **2 UNAFFECTED parents can ONLY have UNAFFECTED offspring**
- **2 AFFECTED parents can have UNAFFECTED offspring**

Therefore, if in a given pedigree two unaffected parents have an affected offspring, the trait cannot be dominant and **must** be recessive.

Similarly, if in a given pedigree two affected parents have unaffected offspring, the trait cannot be recessive and **must** be dominant.

Practice DOMINANT PEDIGREE: In the space below, make THREE OBSERVATIONS from this pedigree to suggest that the trait being studied is **dominant**. In your observations, refer to **specific individuals**.

1. The phenotype appears in every generation, suggesting that the trait is dominant.
2. All affected individuals have at least one affected parent.
3. The wholly unaffected couple in the second generation (individuals II 3 + II 4) has only unaffected children (individual III 4).
4. **THE CLINCHER:** The wholly affected couple in the second generation (individuals II 5 + II 6) has an unaffected child (individual III 6), therefore the trait **CANNOT** be recessive.



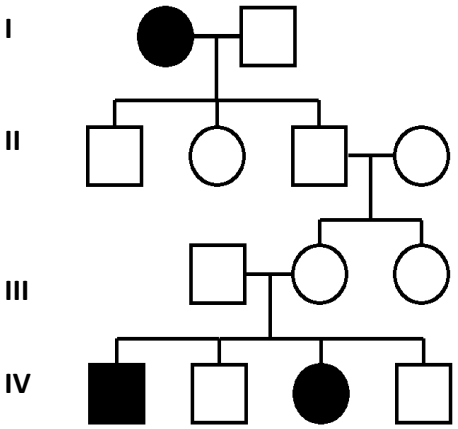
CONCLUSION: This trait MUST be dominant!!!!

In the case of **RECESSIVE** traits,

- The phenotype may skip generations
- **2 UNAFFECTED parents CAN have AFFECTED offspring**
- **2 AFFECTED parents can ONLY have AFFECTED offspring**

Therefore, if in a given pedigree two unaffected parents have affected offspring, the trait cannot be dominant and **must** be recessive.
Similarly, if in a given pedigree two affected parents have unaffected offspring, the trait cannot be recessive and **must** be dominant.

Practice RECESSIVE pedigree: In the space below, make THREE OBSERVATIONS from this pedigree to suggest that the trait being studied is **recessive**. **Explain** how you can tell by providing **specific observations** from the pedigree. Then, identify individuals who **must** be **carriers** by half-shading of his/her square/circle.



1. The phenotype appears to "skip" generations, suggesting that the trait is recessive.
2. **THE CLINCHER:** The unaffected couple in the third generation (individuals III 1 + III 2) have affected children (individuals IV 1 and IV 3), therefore the trait cannot be dominant and **MUST** be recessive.
3. The following individuals all **MUST** be carriers:
   a) II 1, II 2, and II 3 –because their mom had the trait.
   b) Male III 1 and Female III 2 – because two of their children show the trait

CONCLUSION: This trait MUST be RECESSIVE!!!!