Rules Governing Genetic Transmission of Diseases

There are certain rules governing the genetic transmission of diseases.

1. A normal individual cannot have (one or both) alleles of the dominant trait. This is logical, because by definition, even if one allele of a dominant disease trait is present, the individual would be affected.

2. A normal individual, however, can be a carrier of a recessive trait. This means that an individual may have one allele of a recessive trait and still be phenotypically normal. The recessive allele do not express and is masked by the dominant allele. This may show in subsequent generations. So the distinctive feature of recessive trait is that, it skips generations.

3. If trait is X-linked, the male would be affected even by a single recessive allele. This is because male is heterozygous regarding the sex chromosome. So a single allele on X-chromosome (even recessive) will create the expression and male would be affected.

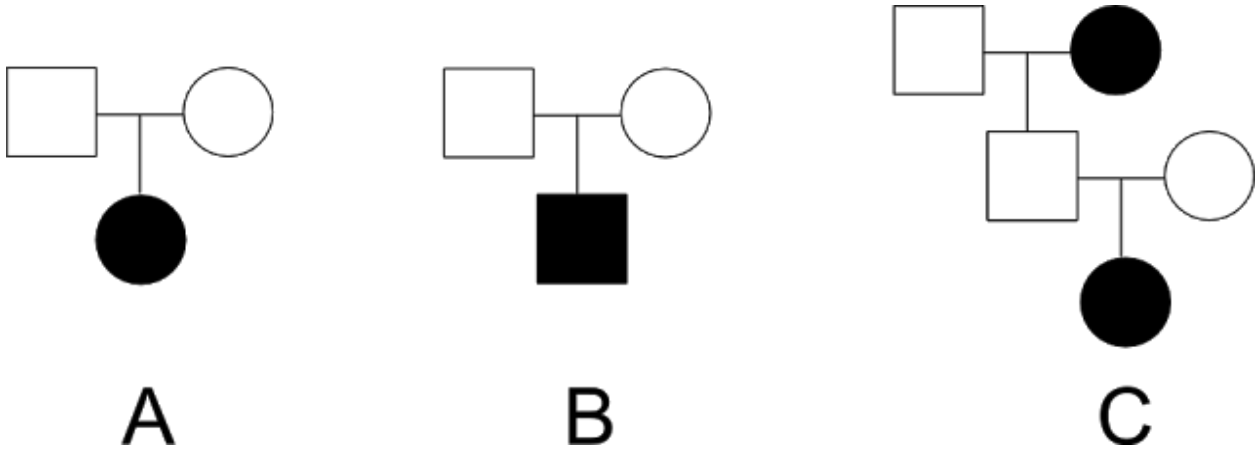
4. If a trait is X-linked, and the father is affected, he transmits the trait to his daughters but not the sons. But mother transmits both to sons and daughters. This is understood by the fact that father has (XY) and X has the affected allele, then that allele is inherited by daughters (XX) and not sons (XY). However, if mother has affected allele, the X chromosome is going to sons as well as daughters equally.

5. **D-D-D Rule for X-Linked Dominant Trait**: If trait is **(D)ominant**, and if **(D)ad** is affected, then all **(D)aughters** will be affected.

6. When father transmits a trait to his son, it is always an Autosomal trait.

Inheritance Patterns in Pedigree Analysis

Based on the above rules, there are a few patterns of genetic inheritance which gives clue to the nature of the trait.

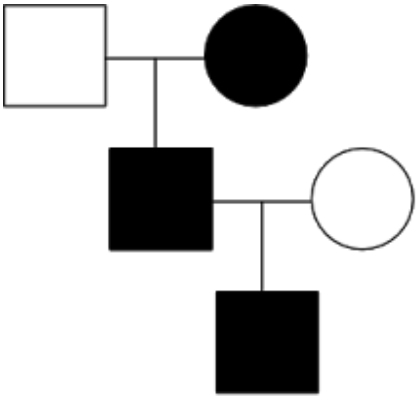


1. If there is skipping of generations, meaning that the trait is observed in generation-I and not in generation-II, but re-appears in subsequent generations, then the trait is "Recessive".

2. If the affected individual has both parents normal, then we can conclude that the trait is "Recessive".

3. If the normal parents have the affected "daughter" ('A' in figure above), then the trait is "Autosomal Recessive". The reason for this is that if daughter is affected and parent are normal, then the trait is "Recessive". But now the question remains whether it is X-Linked or Autosomal. Let's assume that it is X-linked. Since the daughter has (XX) and affected, then both X-chromosomes have the trait allele. This means, she got them from both father and mother. But if father has X-chromosome with the trait allele, then he must also be affected. Since he is not, then the trait has to be "Autosomal".

4. If the normal parents have the affected "son" ('B' in figure above), then the trait is "Recessive". We cannot conclude that its Autosomal or X-Linked. If Autosomal, he got it from both parents. But if X-linked he got it from his mother only.



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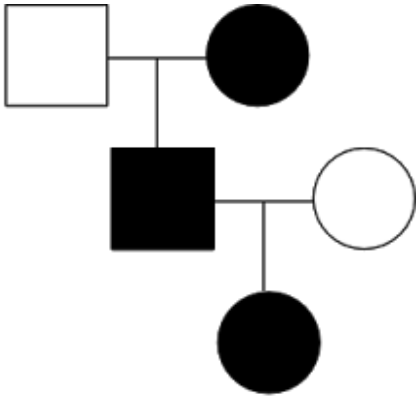
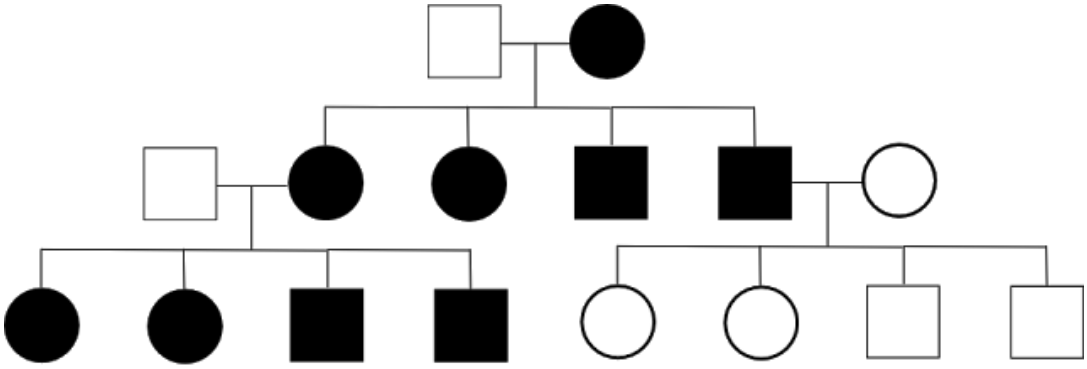


E

5. Trait is dominant, if each generation has a trait. Remember! For a trait to express only one allele is sufficient.

6. When father is affected and son is also affected ('D' in figure above), then the trait is "Autosomal Dominant". This is because if the trait is X-linked, it could not be transmitted to a son. So the trait has to be Autosomal.

7. However, when an affected father transmits the disease to a daughter ('E' in figure above), it may be X-linked or autosomal; we can't say for sure. We need have to have more information to ascertain the case.



F

8. When a mother is affected and "all" the children - sons as well as daughters, are affected ('F' in figure above), then the trait is maternal (cytoplasmic) inheritance.