

Title of the lecture: Pedigree charts





Pedigree charts

Pedigree chart:- are diagrams that show the phenotypes and/or genotypes for a particular organism, its ancestors, and descendants. While commonly used in human families to track genetic diseases, they can be used for any species and any inherited trait. Geneticists use a standardized set of symbols to represent an individual's sex, family relationships and phenotype. These diagrams are used to determine the mode of inheritance of a particular disease or trait, and to predict the probability of its appearance among offspring.

Pedigree analysis is therefore an important tool in basic research, agriculture, and genetic counseling. Each pedigree chart represents all of the available information about the inheritance of a single trait (most often a disease) within a family. In real pedigrees, further complications can arise due to incomplete penetrance including age of onset) and variable expressivity of disease alleles, the phenotype accurately reflects the genotype. A pedigree may be drawn when trying to determine the nature of a newly discovered disease, or when an individual with a family history of a disease wants to know the probability of passing the disease on to their children. In either case, a tree is drawn, as shown in Figure 1 below.

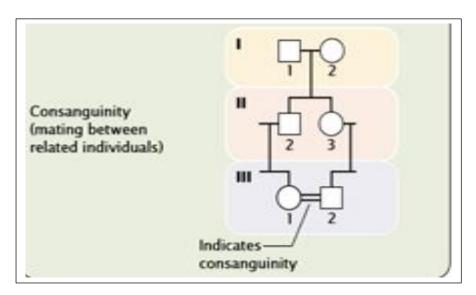


Figure.1 pedigree example

With circles to represent females, and squares to represent males. Mattings are drawn as a line joining a male and female, while a consanguineous mating (closely related, such as siblings or first cousins) is two lines. The symbols commonly used in pedigrees are summarized in figure 2.



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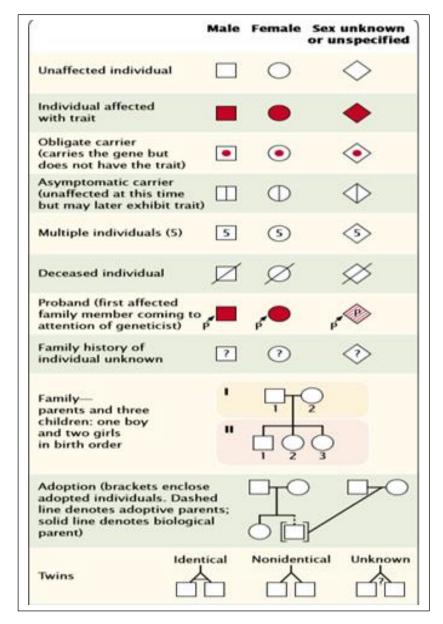


Figure.2 The symbols of pedigree

Before starting the analysis of any family pedigree, it must be taken into a considers whether the trait is autosomal dominant or recessive, X-linked dominant trait or Xlinked recessive trait or Y-linked trait. Therefore, attention must be paid to the notes in the figure 3.



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Autosomal recessive trait

- Appears in both sexes with equal frequency.
- Trait tends to skip generations.
- Affected offspring are usually born to unaffected parents.
- When both parents are heterozygous, approximately 1/4 of the offspring will be affected.
- Appears more frequently among the children of consanguine marriages.

Autosomal dominant trait

- Appears in both sexes with equal frequency.
- Both sexes transmit the trait to their offspring.
- 3. Does not skip generations.
- Affected offspring must have an affected parent, unless they possess a new mutation.

- When one parent is affected (heterozygous) and the other parent is unaffected, approximately 1/2 of the offspring will be affected.
- Unaffected parents do not transmit the trait.

X-linked recessive trait

- More males than females are affected.
- Affected sons are usually born to unaffected mothers; thus, the trait skips generations.
- A carrier (heterozygous) mother produces approximately 1/2 affected sons.
- 4. Is never passed from father to
- All daughters of affected fathers are carriers.

X-linked dominant trait

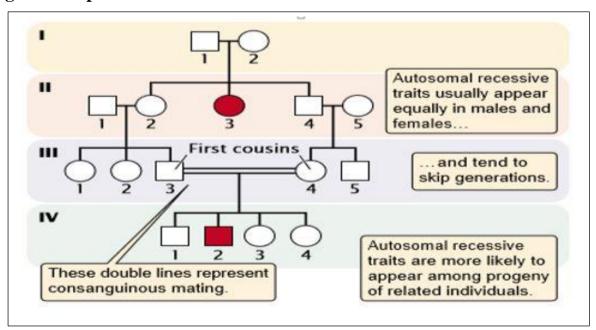
- Both males and females are affected; often more females than males are affected.
- Does not skip generations. Affected sons must have an affected mother; affected daughters must have either an affected mother or an affected father.
- Affected fathers will pass the trait on to all their daughters.
- Affected mothers (if heterozygous) will pass the trait on to 1/2 of their sons and 1/2 of their daughters.

Y-linked trait

- 1. Only males are affected.
- Is passed from father to all sons.
- 3. Does not skip generations.

Figure.4 pedigree characteristics

Pedigree examples:-



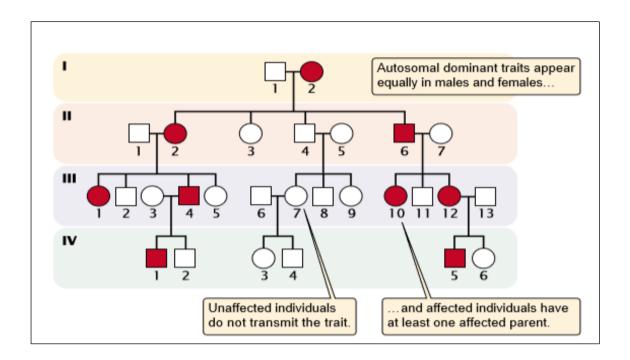
Autosomal recessive traits



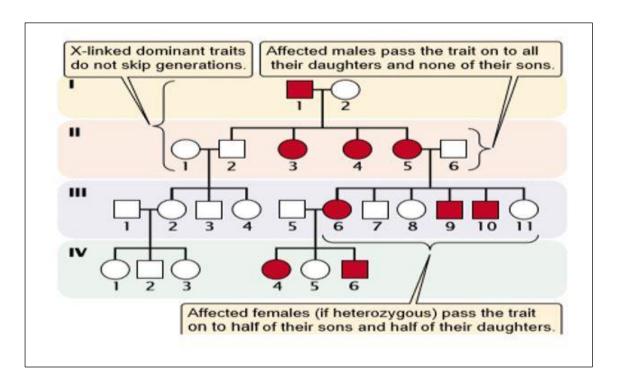
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Autosomal dominant traits



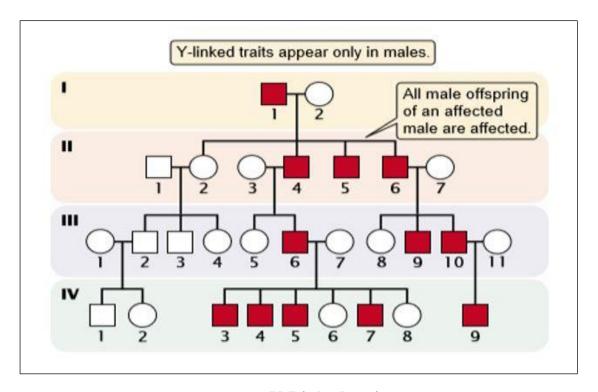
X-Linked dominant traits



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Y-Linked traits