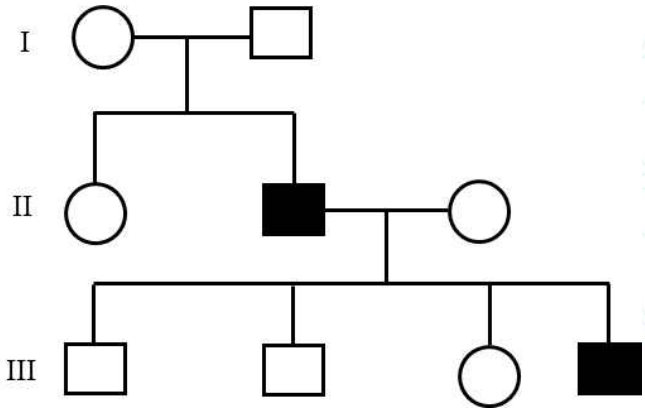
BIOLOGY

Live eBook

01. Pedigree analysis

A pedigree is a family tree or chart made of symbols and lines that represent a person's genetic family history. In pedigree, symbols represent people and lines represent genetic relationships. The pedigree is a visual tool for documenting the biological relationship in families and determine the mode of inheritance (dominant, recessive etc.) of genetic diseases. Pedigrees are most often constructed by medical geneticists or genetic counselors. A sample pedigree is given below:



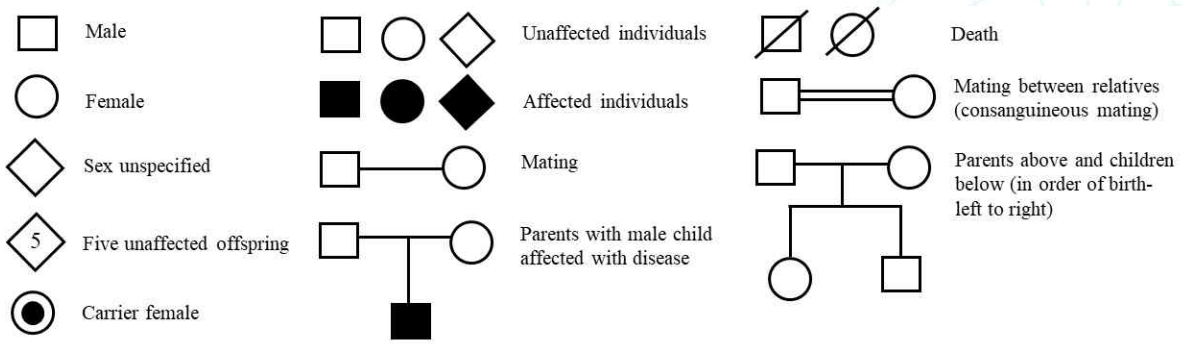
In this pedigree, squares represent males and circles females. Horizontal lines connecting a male and female stand for mating. Vertical lines extending downward from a couple represent their children. Subsequent generations are therefore written underneath the parental generations and the oldest individuals are found at the top of the pedigree. Each generation should be given a Roman numeral designation and individuals within the same generation should be numbered from left to right.

On the basis of the information in a pedigree, geneticist tries to determine the mode of inheritance of a trait. The pedigree might be used to answer two types of questions;

First, and there patterns within the pedigree that are consistent with a particular mode of inheritance?

Second, are there patterns within the pedigree that are inconsistent with a particular mode of inheritance?

Symbols used in the human pedigree analysis



The symbols used in the human pedigree analysis.

Expected patterns for various types of inheritance in pedigrees

Autosomal recessive inheritance

(i) Trait often skips generations.
(ii) An almost equal number of affected males and females.
(iii) If both parents are affected, all children should be affected.
(iv) In most cases of unaffected people mating with affected individuals, all children produced are unaffected. When at least one child is affected (indicating that the unaffected parent is heterozygous), then approximately half the children should be affected.
(v) Most affected individuals have unaffected parents.

Autosomal dominant inheritance

(i) Trait should not skip generations.
(ii) An affected person mating with an unaffected person should produce approximately 50% affected offspring (indicating also that the affected individual is heterozygous).
(iii) The distribution of the trait among sexes should be almost equal.
(iv) Transmitted by either sex.

Sex-linked (X-linked) recessive inheritance

(i) Most affected individuals are male.
(ii) Affected males result from mothers who are affected or who are known to be carriers (heterozygotes).
(iii) Affected females come from affected father and affected or carrier mothers.
(iv) The sons of affected females should be affected.
(v) Approximately half the sons of carrier should be affected.

Sex-linked (X-linked) dominant inheritance

(i) The trait does not skip generations.
(ii) Affected males must come from affected mothers.
(iii) Approximately half the children of an affected heterozygous females are affected.
(iv) Affected females come from affected mothers or fathers.
(v) All the daughters, but none of the sons, of an affected father are affected.

Y-linked inheritance

(i) Affects only males.
(ii) Affected males always have an affected father.
(iii) All sons of an affected man are affected.