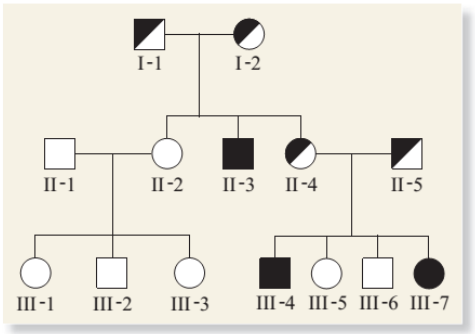
pedigree drawing

- A pedigree shows generations and relationships among biological parents and offspring.
- It also tracks which of those individuals have a specific trait.
- is aimed at determining the type of inheritance pattern that a gene will follow.
- Determine the probability of an affected offspring for a given cross.
- In this section, we will examine a few large pedigrees that involve diseases inherited in different ways



(a) Human pedigree showing cystic fibrosis

Symbols used in pedigree analysis

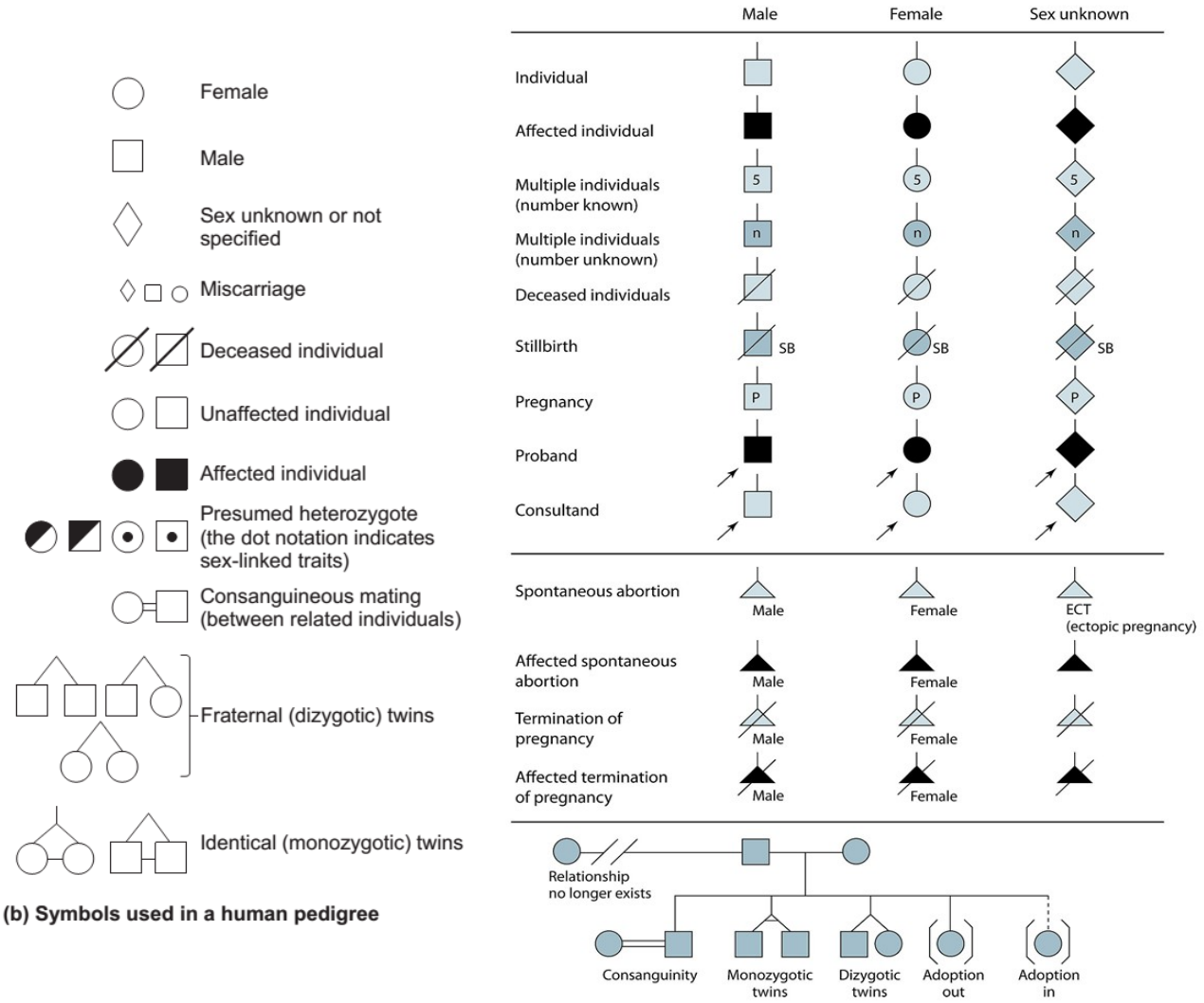
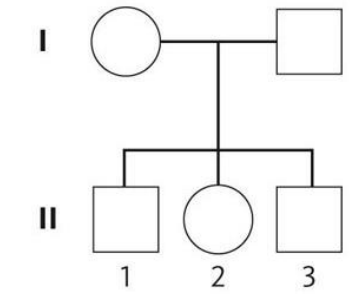


Figure 3.2 Main symbols used in pedigrees. The dot symbol for a carrier, and the double marriage lines for consanguineous mattings, are used to draw attention to those features, but their absence does not necessarily mean that a person is not a carrier, or that a union is not consanguineous.

Designation of generations and individuals

1. Each horizontal line is a generation
2. Place the oldest generation at the top
3. Use Roman numerals to identify generations
4. Use Arabic numbers to identify individuals within a generation
5. List siblings from oldest to youngest, from left to right
6. Male partner is usually placed to the left of the female partner
7. Record full name, current age and date of birth, or age at death for each individual
8. Record race and ethnic origin of each individual
9. Note health problems and/or cause of death for each individual

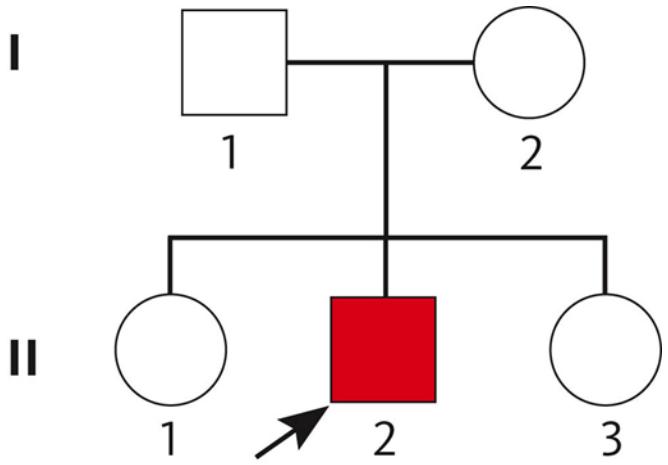


Parents and children. Roman numerals symbolize generations. Arabic numbers symbolize birth order within generation (boy, girl, boy)

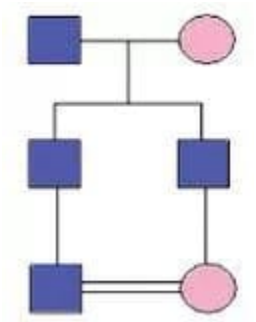
KEY

I, II, III, etc. = each generation
1, 2, 3, etc. = individuals within a generation

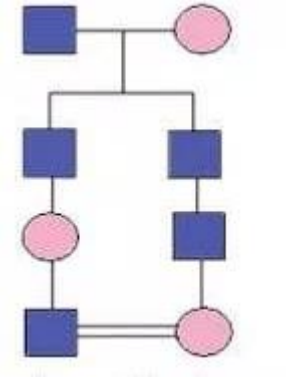
- The proband is an affected individual coming to medical attention independently of other family members.
- The proband is designated with an arrow in the pedigree, and there may be more than one proband per family.



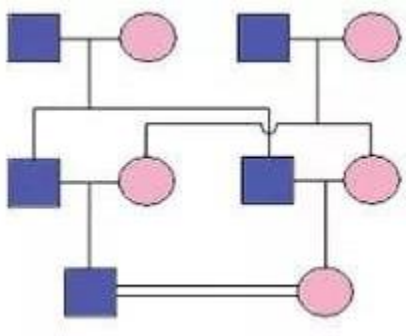
Consanguinity



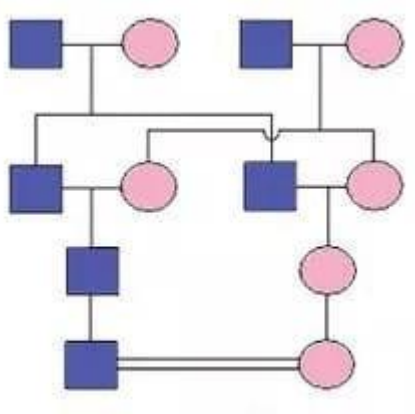
Ahmad Amal



First Cousins

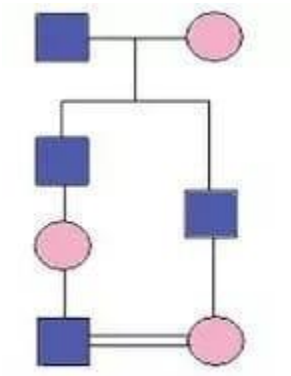


Second Cousins



Double First Cousins

Double Second Cousins



First Cousins
one's removed

Cousins:

- Ahmad and Amal are _____ **cousins** if one of Ahmad's parents is a sib of one of Amal's parents.
- If both of Ahmad's parents are sibs of both of Amal's parents, Ahmad and Amal are _____**cousins**.
- They are _____**cousins** if one of Ahmad's parents is first cousin of one of Amal's parents.

Dominant and Recessive Inheritance

- Nomenclature: For dominant traits the capital letter (e.g. A) represents the mutant allele and the small letter (e.g. a) represents the normal allele. For recessive traits, the small letter (e.g. a) represents the mutant allele and the capital letter (e.g. A) represents the normal allele.

- **Autosomal dominant traits** are those traits in which the phenotype of the heterozygote and the homozygote for the dominant allele are the same, i.e., Aa and AA have the same phenotype where A=dominant allele. These traits are expressed when only one copy of the dominant allele is present. In practice, if the heterozygote expresses the trait, then the trait is classified as dominant, even if the phenotype of the homozygote (AA) and heterozygote (Aa) are different.

- **Autosomal recessive traits** are those traits in which the phenotype is expressed only if homozygous for the recessive allele, i.e., aa where a=recessive allele. Two copies of the recessive allele are necessary for expression.

Dominant and Recessive Inheritance

- If the heterozygote (AB) has a different phenotype than either of the homozygotes (AA or BB), then the alleles are said to be **codominant**.

- **X-linked dominant traits** are those expressed when either males or females have one copy of the dominant allele, i.e., X^AY or X^AX^a where A=dominant allele.

- **X-linked recessive** traits are those expressed in males who carry one copy of the recessive allele (i.e., are hemizygous, XaY where a=recessive allele). Two copies of the recessive allele are generally required for females to express the trait, i.e., XaXa.

pedigree drawing

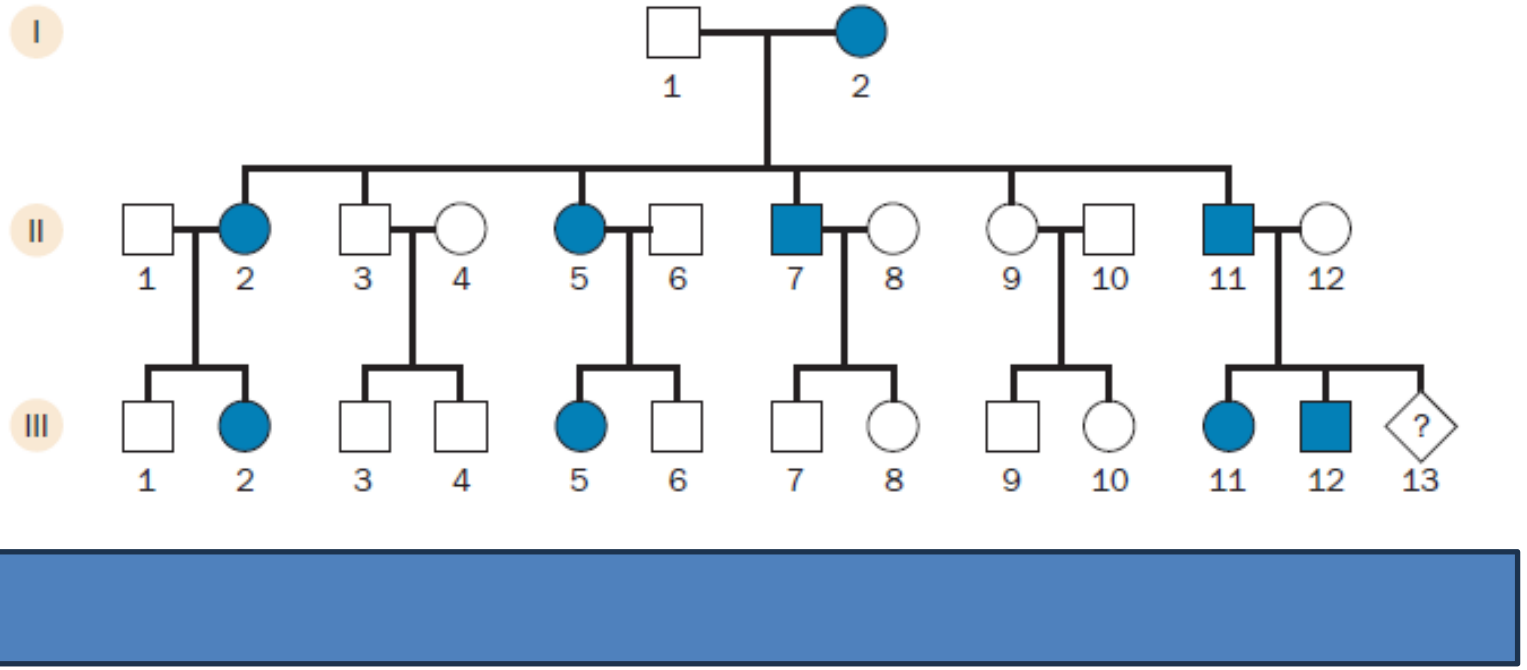


Figure 3.2 shows the symbols commonly used for pedigree drawing. **Generations are usually labeled in Roman numbers, and individuals** within each **generation in Arabic numerals** .

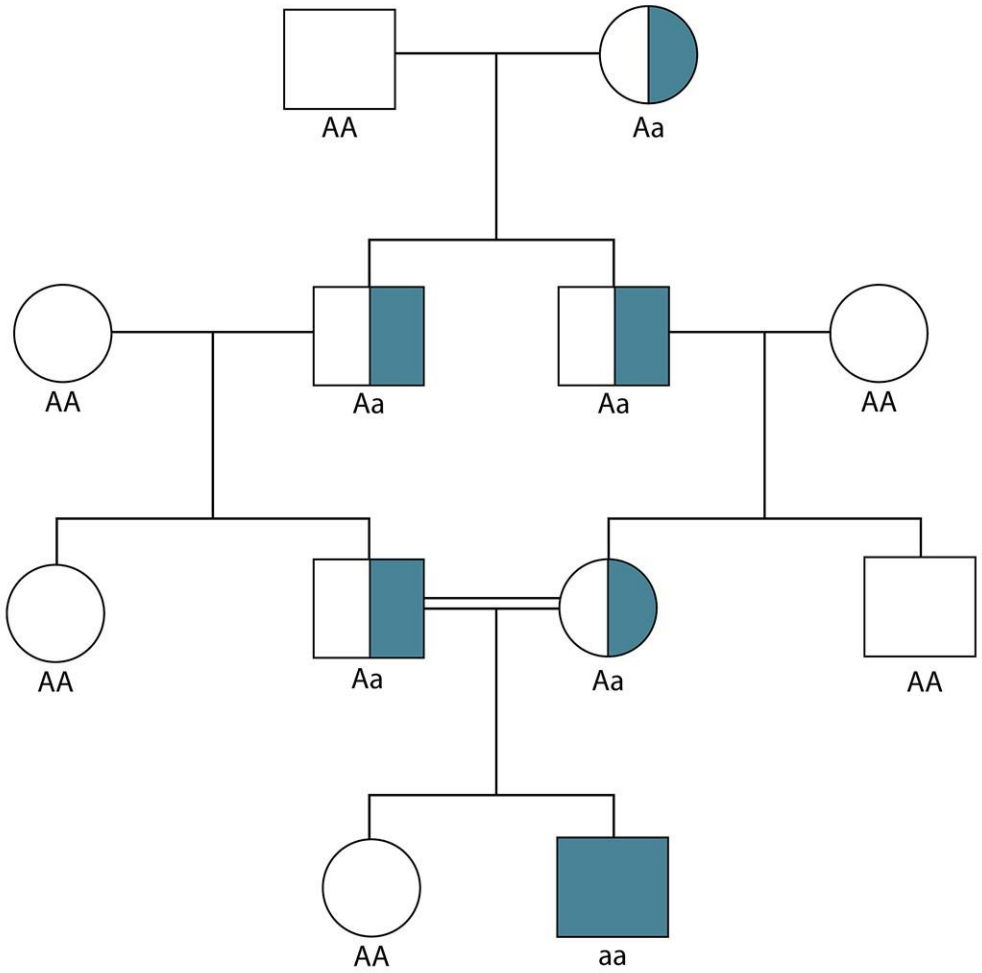
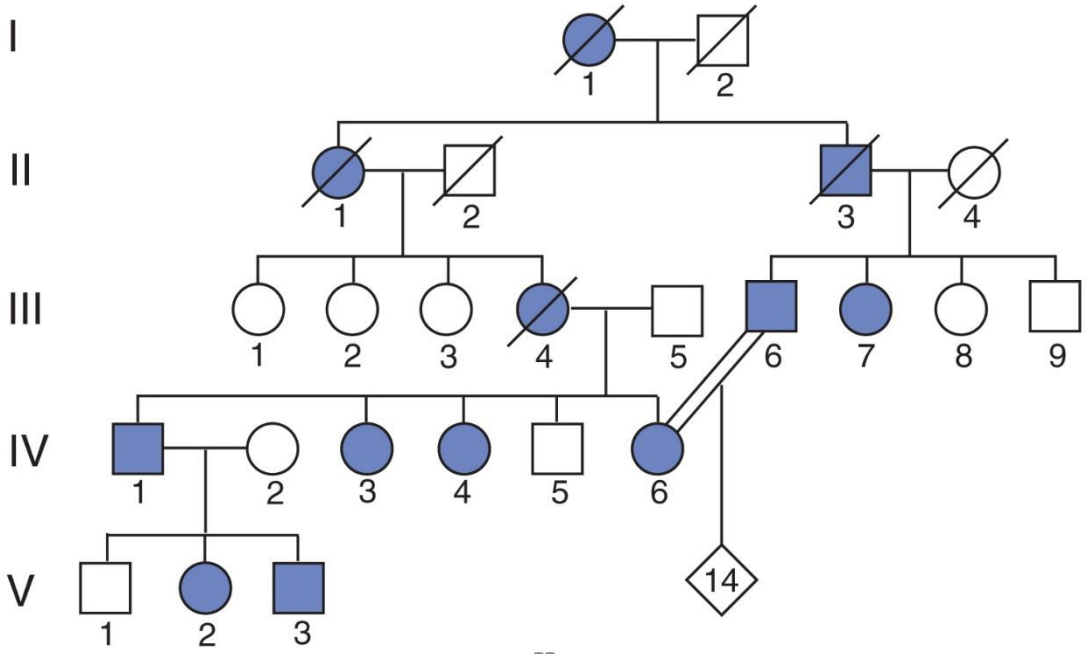


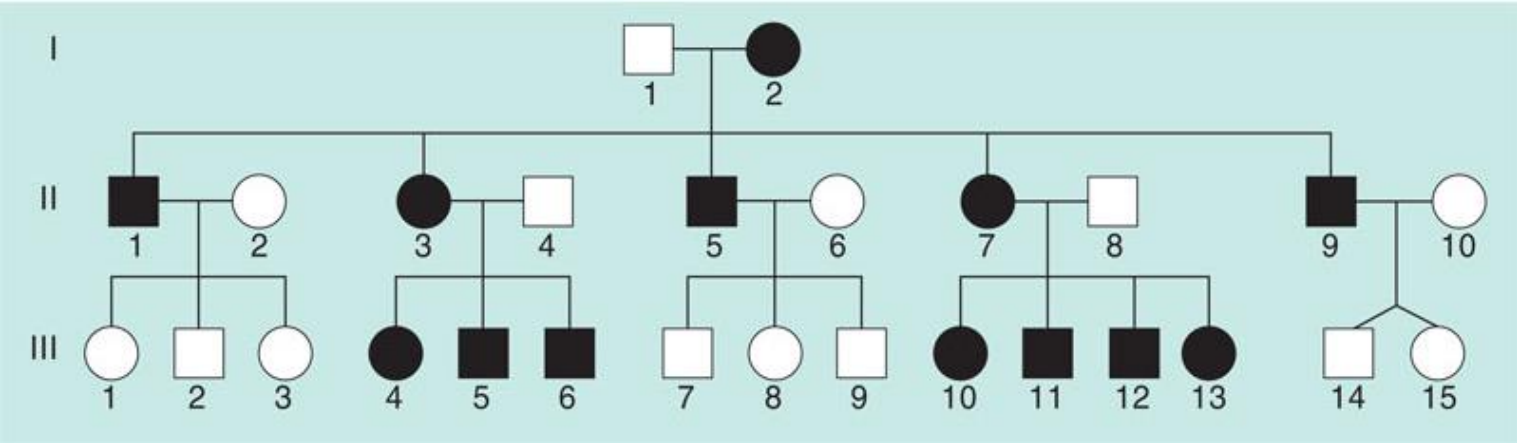
Figure 3.4 The individual who is homozygous for aa has inherited the a allele from his great-grandmother, transmitted through both parents.

A pattern of inheritance indicates a rare dominant trait; e.g Huntington disease

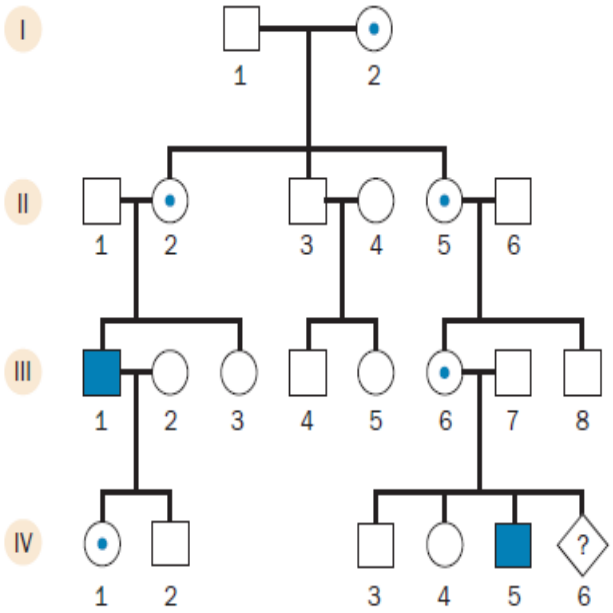
Every affected person has at least one affected parent

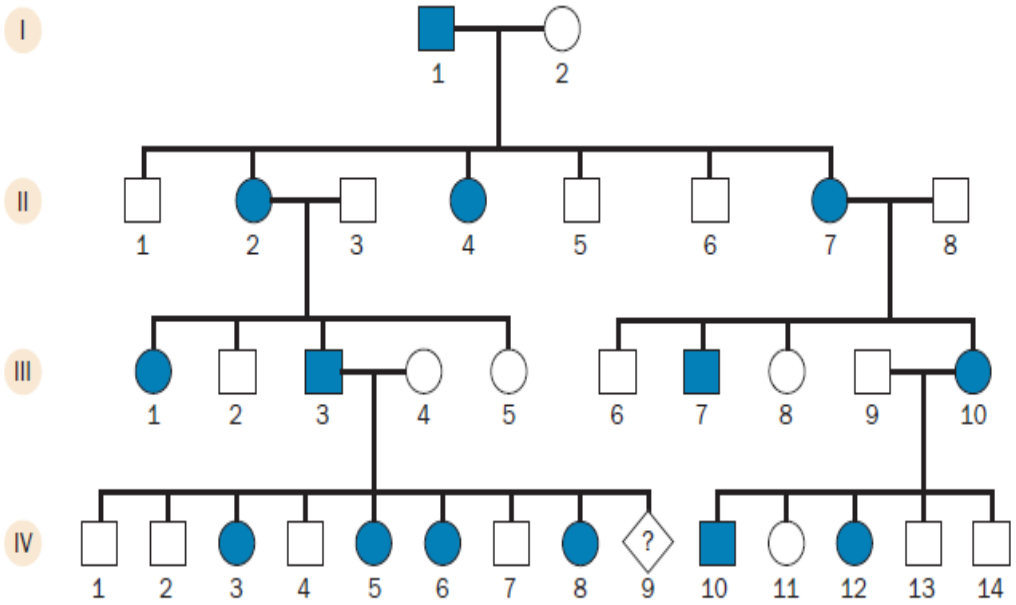


What features characterize this pattern of inheritance?



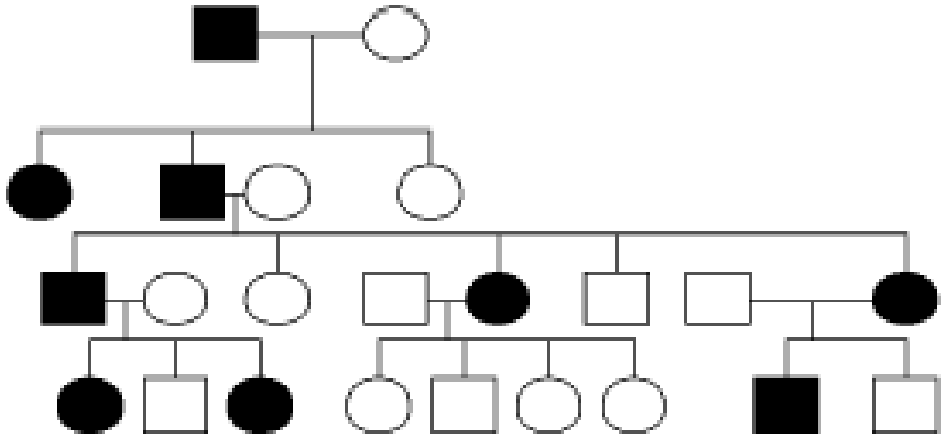
Pedigree


I
1 2
II
1 2 3 4 5 6
III
1 2 3 4 5 6 7 8
IV
1 2 3 4 5 6
?


I
1 2
II
1 2 3 4 5 6 7 8
III
1 2 3 4 5 6 7 8 9 10
IV
1 2 3 4 5 6 7 8 9 10 11 12 13 14
?

Which of the modes of inheritance is the most likely based on the pattern of people with the condition in this family?

a. Chromosomal translocation
b. Autosomal dominant
c. X-linked dominant
d. Mitochondrial
e. Autosomal recessive



Which of the modes of inheritance is the most likely based on the pattern of people with the condition in this family?

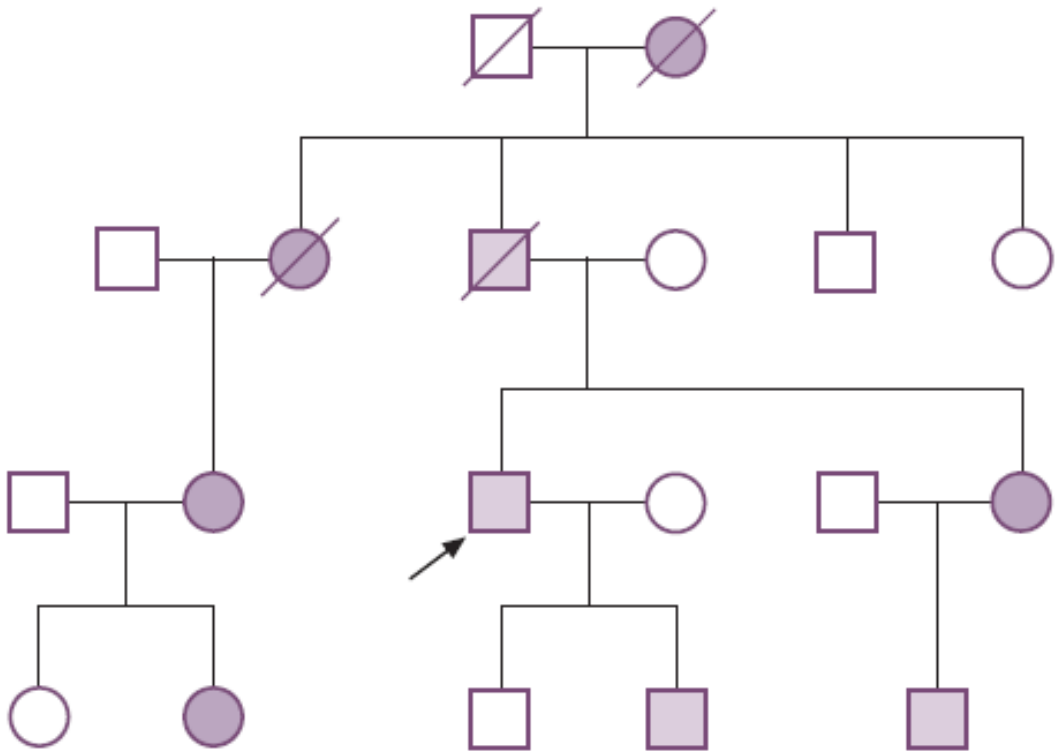
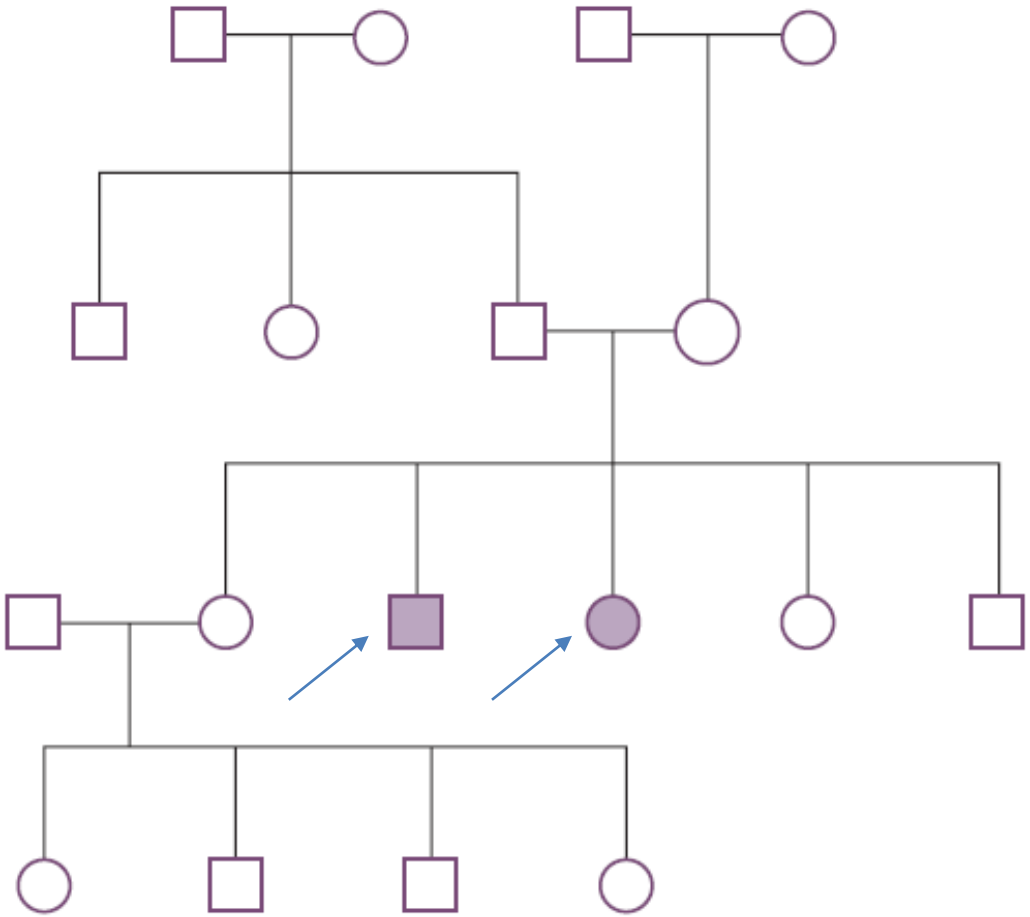
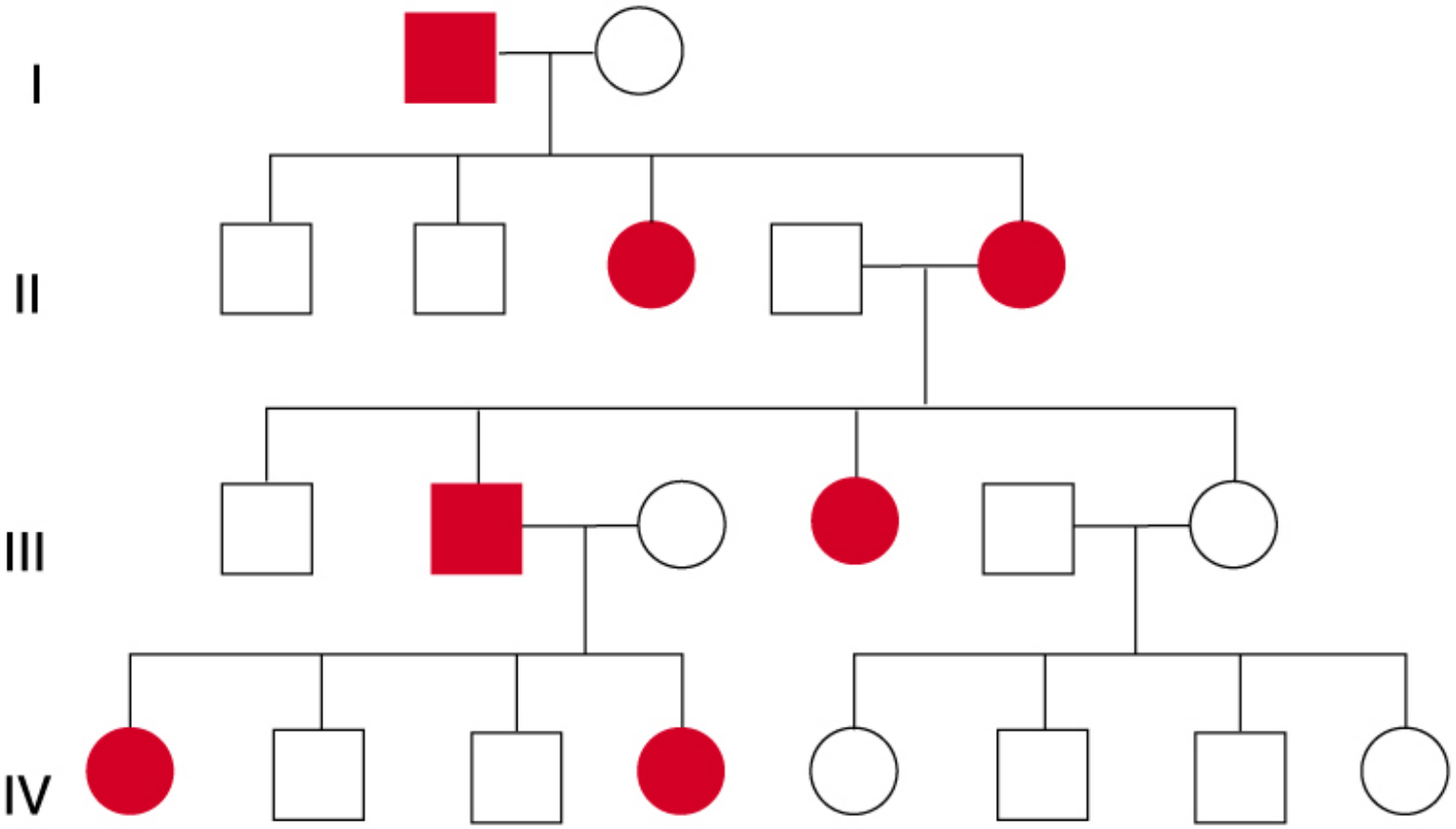


Fig. 8.3 Pedigree of a family with familial hypercholesterolaemia. The patient in Fig. 8.2 is indicated by an arrow.

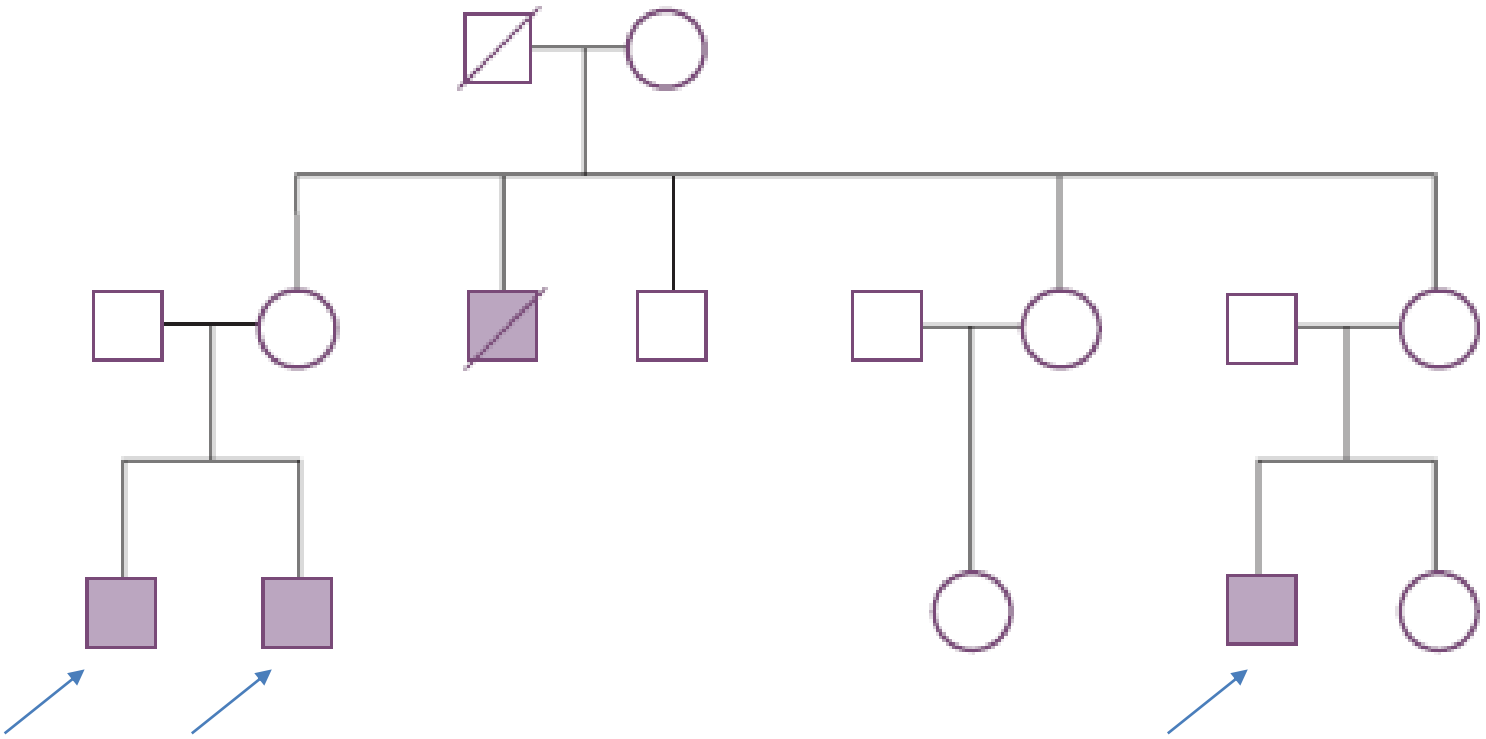
Which of the modes of inheritance is the most likely based on the pattern of people with the condition in this family?



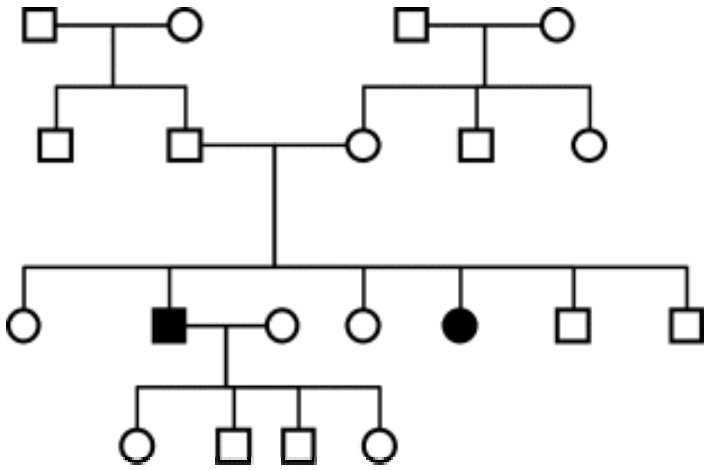
Which of the modes of inheritance is the most likely based on the pattern of people with the condition in this family?



Which of the modes of inheritance is the most likely based on the pattern of people with the condition in this family?



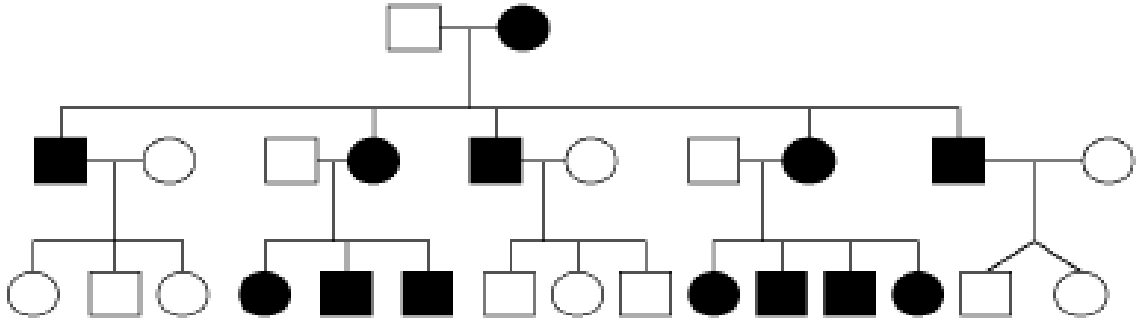
Which of the modes of inheritance is the most likely based on the pattern of people with the condition in this family?



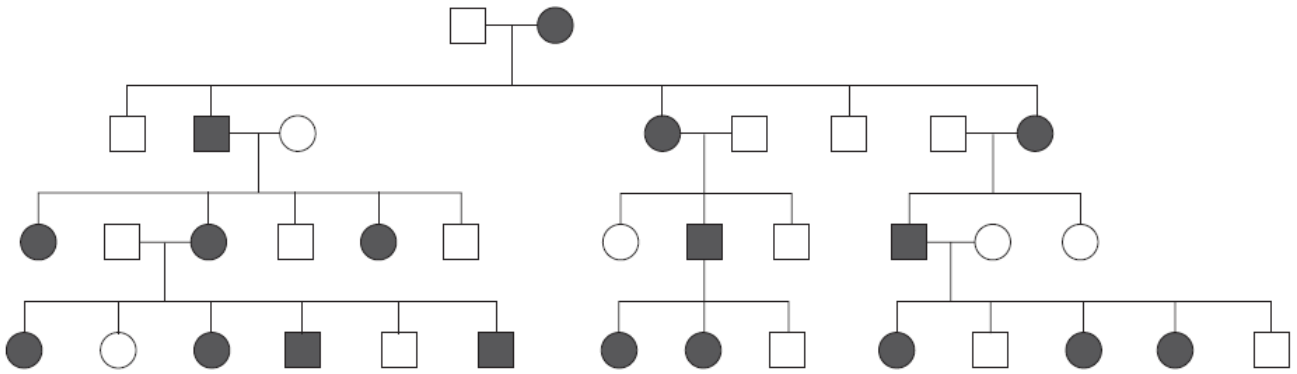
a. Y linked
b. Autosomal recessive
c. Autosomal dominant
d. X-linked recessive
e. X-linked dominant

Which of the modes of inheritance is the most likely based on the pattern of people with the condition in this family?

a. Reciprocal chromosomal translocation
b. Autosomal dominant
c. X-linked dominant
d. Mitochondrial
e. Autosomal recessive



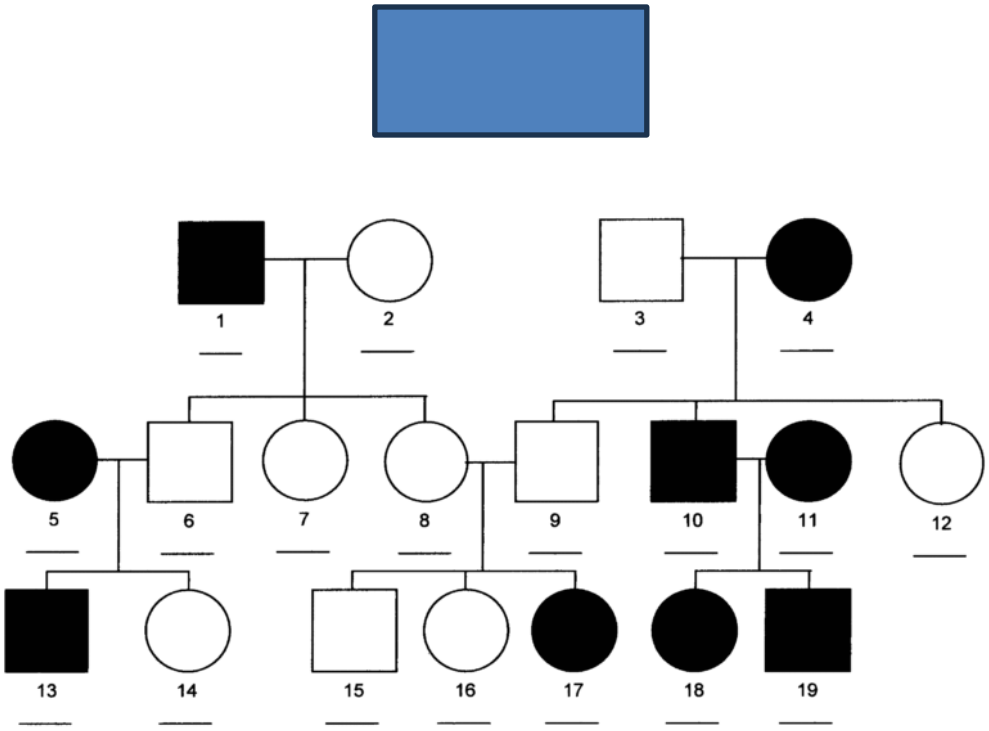
Which of the modes of inheritance is the most likely based on the pattern of people with the condition in this family?



Which of the modes of inheritance is the most likely based on the pattern of people with the condition in this family? Genotype?

A: wild-type allele

a: Mutant allele



Which of the modes of inheritance is the most likely based on the pattern of people with the condition in this family? Genotype?

A: Dominant allele

a: Recessive allele