Pedigree Analysis

(cf. chapters 4.4, 5.2, 6.2 of textbook)

- **"crosses" and "matings" in human(s) (families) are shown and analyzed in pedigrees**
  - **pedigrees are formalized ways using standard sets of symbols to depict family trees and lineages**
  - **pedigrees provide concise and accurate records of families**
  - **pedigrees are helpful in following and diagnosing heritable traits (e.g. diseases and medical conditions), i.e. describing patterns (or modes) of inheritance**
  - **pedigrees are useful in mapping (locating and isolating) genes "responsible" for certain traits (will be covered later)**

Pedigree Construction

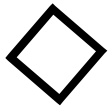
- use standard set of symbols (cf. figure 4.13 of textbook)
- one generation per row (oldest at the top)
- siblings are shown in order of birth (from left to right)
- generations are given roman numerals (I, II, III, IV, etc.)
- individuals within a generation (row) are given arabic numerals (1, 2, 3, 4, etc.)
- (show overhead)


normal female


normal male


female with trait


male with trait


sex unspecified


male, carrying allele for trait but does not express it (“carrier”)

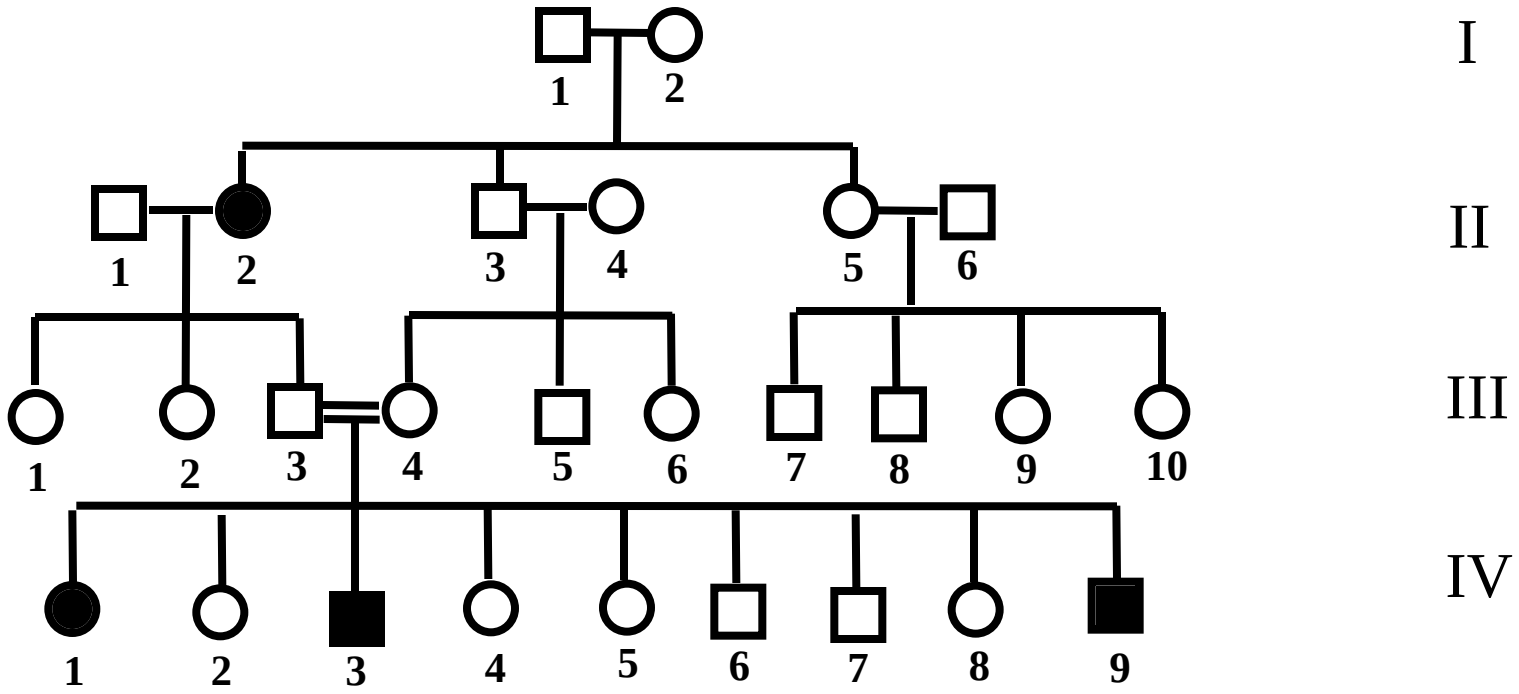
etc. etc. etc.

Analyzing Pedigrees

- trial and error: consider one mode of inheritance at a time for each mating in pedigree and try to find evidence against it; repeat for each mode of inheritance; e.g. autosomal recessive or dominant, sex chromosome recessive or dominant, etc. etc.
- assumption: e.g. for rare traits unaffected people entering into a family pedigree (e.g. by marriage) are considered homozygous normal
- result: pedigrees can frequently rule out, but not necessarily prove, a certain mode of inheritance

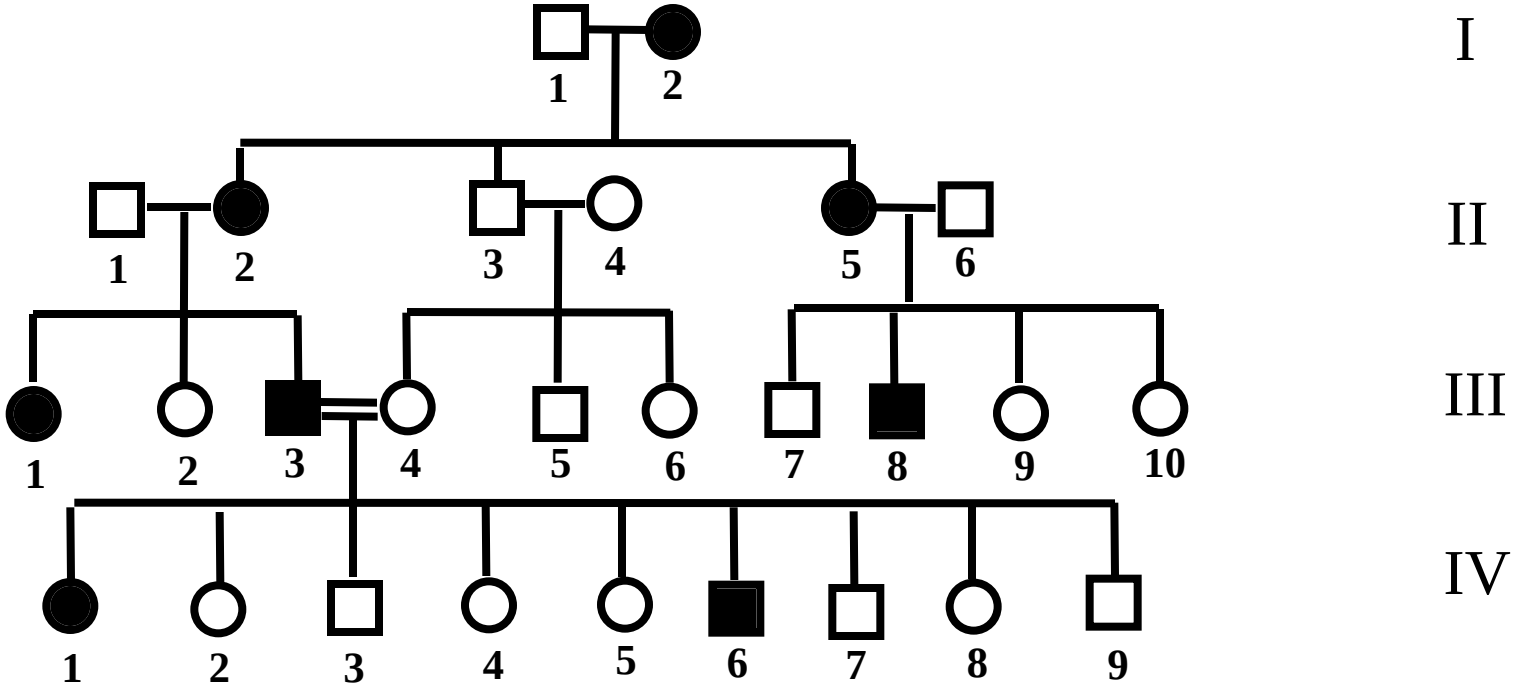
- **NB: Pedigrees and Punnett-Squares are tools that apply Mendel's first rule ("segregation of alleles") to predict recurrence risks of inherited traits, diseases or medical conditions.**

autosomal recessive



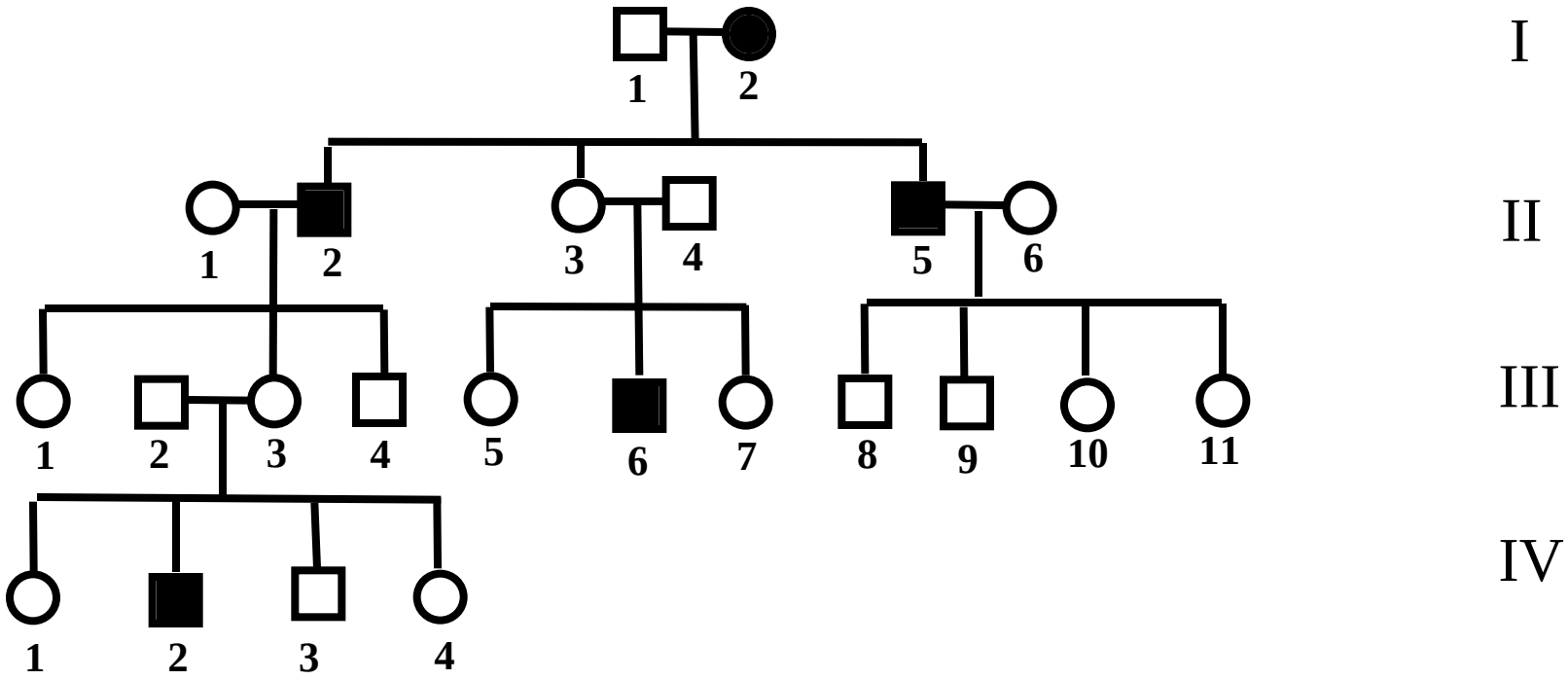
- both sexes affected (males and females)
- trait can skip generations
- e.g. cystic fibrosis, albinism (cf. figure 4.15 of textbook)

autosomal dominant



- both sexes affected (males and females)
- trait does NOT skip generations
- e.g. myotonic dystrophy, hypotrichosis (hair loss that begins in childhood; cf. figure 4.16 of textbook)

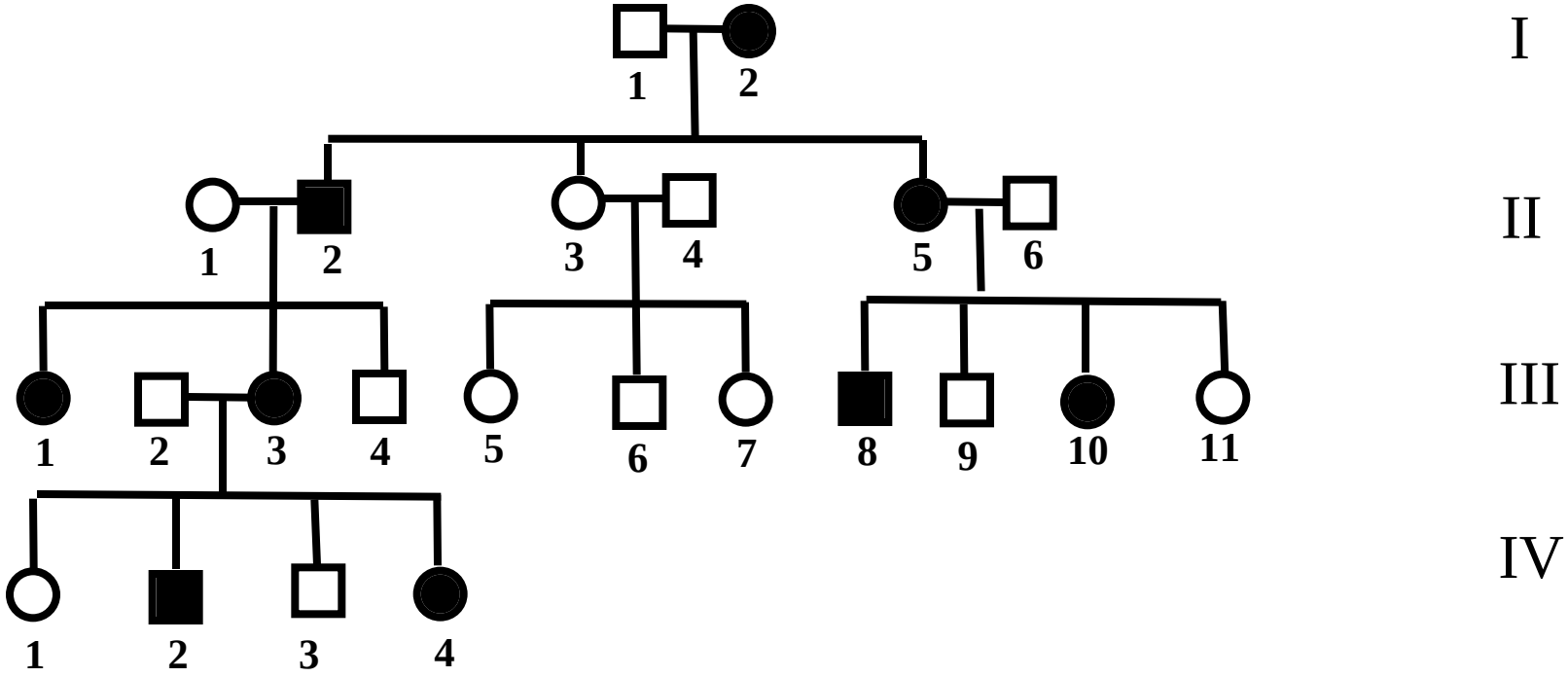
X chromosome linked recessive



- **more affected males than females**
- **males never transmit to sons**
- **daughters of affected males always inherit (recessive!) mutation, thus are "carriers"**

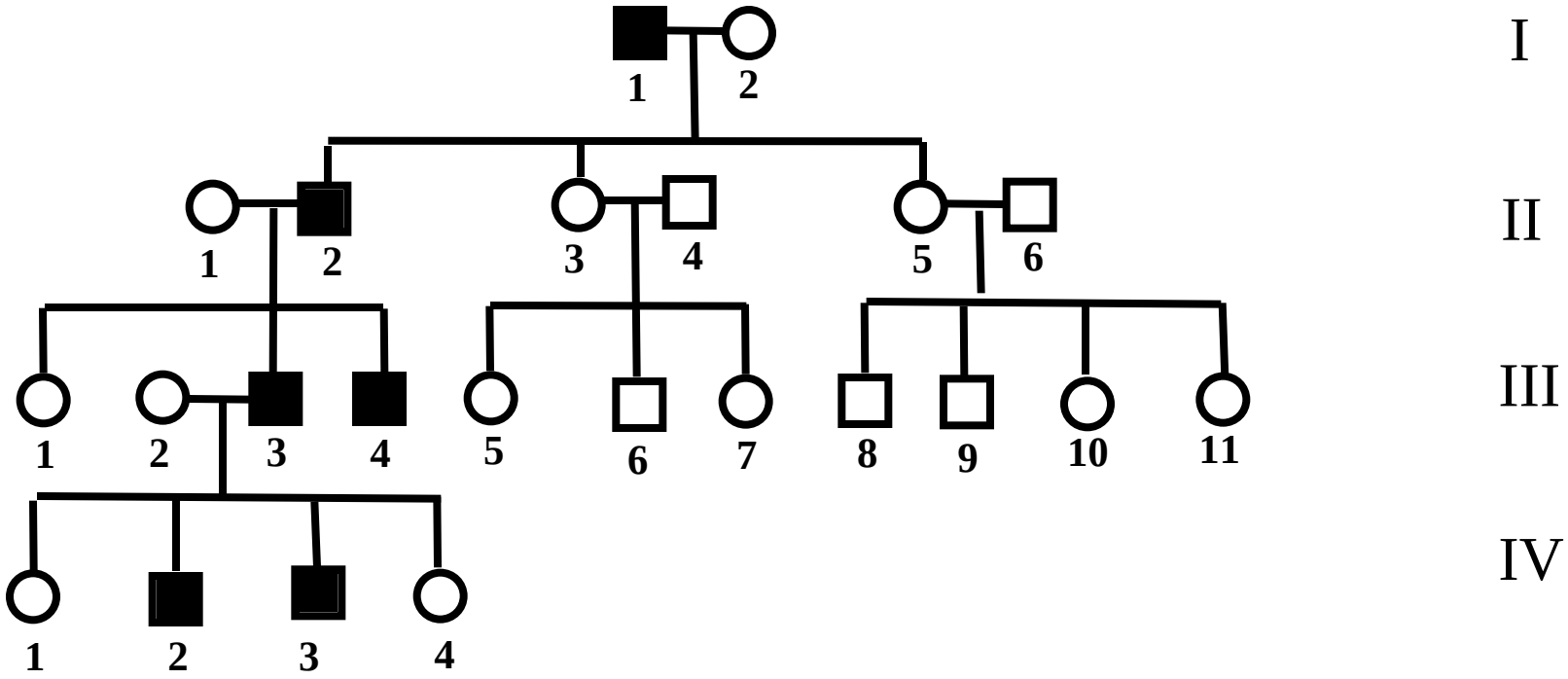
- **trait can skip generations, e.g. when females are heterozygotes ("carriers")**
- **e.g. hemophilia and muscular dystophy (Duchenne form)**
- **cf. chapter 6.2 of textbook**

X chromosome linked dominant



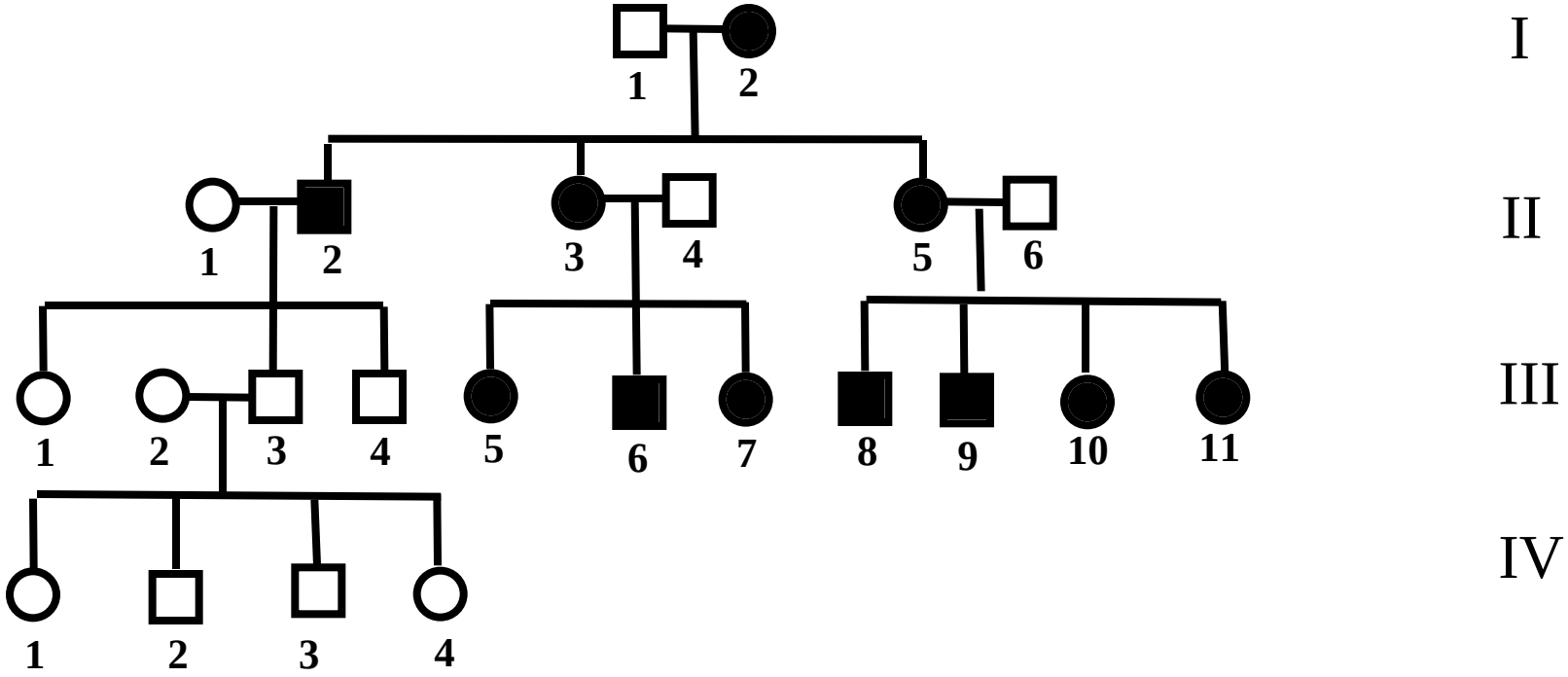
- **both sexes affected (males and females)**
- **females transmit to daughters and sons**
- **males ALWAYS transmit to daughters, but NOT AT ALL to sons**
- **trait does NOT skip generations**
- **e.g. Rett Syndrome (mental retardation, neural degeneration)**
- **cf. chapter 6.2 of textbook**

Y chromosome linked



- **only males are affected**
- **males ALWAYS transmit to sons**
- **cf. chapter 6.2 of textbook**

mitochondrial inheritance



- **both sexes are affected**
- **females transmit to ALL of their progeny**
- **males do NOT transmit to any of their progeny**
- **cf. chapter 5.2 of textbook**

gender effects on phenotype

(cf. chapter 6.4 of textbook)

- **sex-limited inheritance (e.g. traits affected by hormones)**
  - **only one gender is capable of showing trait, the other gender is NOT, regardless of underlying genotype**
  - **the genes involved are typically autosomal, but the expression of these genes is dependent on the gender of the individual**
  - **e.g. beard growth, breast size**
- **sex-influenced inheritance (e.g. traits affected by hormones)**
  - **the gender of the individual determines whether a particular phenotype assumes dominant or recessive state**
    - **e.g. a phenotype that is dominant in one gender is recessive in the other gender**
  - **e.g. pattern baldness (dominant in males and recessive in females)**

problems in constructing a pedigree

- **poor medical records**
- **scattering of family members**
- **inaccurate and anecdotal information**
- **miscarriages and still births**
- **infidelity / concealed adoptions**
- **variable expressivity of genotype (as phenotype)**
- **incomplete penetrance of genotype (showing phenotype)**

- **AGAIN: pedigrees can frequently rule out, but not necessarily prove, a certain mode of inheritance**