How to Solve Problems Involving Pedigrees

1. Since the pedigrees presented are **simple** pedigrees, the following assumptions can be made:
    a. No new mutations occur in the families. This means that a dominant trait must occur in a parent if it appears in a child. This also means that if a recessive trait appears in a child but does not occur in either parent, both parents must be heterozygous carriers for the trait.
    b. All traits are 100% penetrant. This means that if the genotype for the trait occurs in an individual, the trait will appear.
2. If neither parent is affected,
    a. the trait cannot be dominant.
    b. the trait could be recessive and either parent or both could be heterozygous carriers.
3. If one parent is affected,
    a. the trait could be dominant and the affected parent could be heterozygous while the unaffected parent is not a carrier
    b. the trait could be recessive and the affected parent is homozygous while the unaffected parent could be a heterozygous carrier
4. If both parents are affected
    a. the trait could be dominant & both parents could be heterozygous carriers which means that some of the children could be unaffected
    b. the trait could be recessive meaning that both parents would have to be homozygous and all the children would have to be affected
5. Consideration of X-linked or Y-linked traits:
    a. X-linked recessive
        i. affected male children must have, at least, an heterozygous mother
        ii. an affected female will transmit the trait to **all** male children
    b. X-linked dominant
        i. affected males transmit the trait to **all** daughters
        ii. affected sons must have an affected mother
    c. Y-linkage
        i. male to male transmission only
        ii. no affected females
6. Mitochondria associated traits
    a. affected females transmit the trait to all children
    b. affected males do not transmit the trait to any children

Pedigree Analysis Chart - A how to guide!

- Pedigree comes from the Greek word for Foot of a crane
- Pedigree's are used to chart the genealogy of a family
- Pedigree is also the name for a dog food → WHY?
- Pedigree's show how a particular trait can be passed through generations!

PEDIGREE RULES!

- **Females** → Circles **Males** → Squares **Unknown sex** → Diamonds
- **I, II, III** → generations **1,2,3** → individuals within a generation
- **Deceased** → slash through circle/square
- **Known Carriers** → Circle or Square is half shaded
- **Twins**: **Identical** → joined **Fraternal** → un-joined
- **Shaded Symbols**: Affected Individuals for the trait in question

WHAT PEDIGREES TELL YOU!

1) What is the chance one or both of the parents is a carrier for a trait?
2) What is the chance the F1, F2 or so on will express the trait?
3) Enable you to predict the mode of inheritance for the trait.
4) Enable you to determine individual genotypes and chance of inheriting a certain trait.

Pedigree analysis diagram

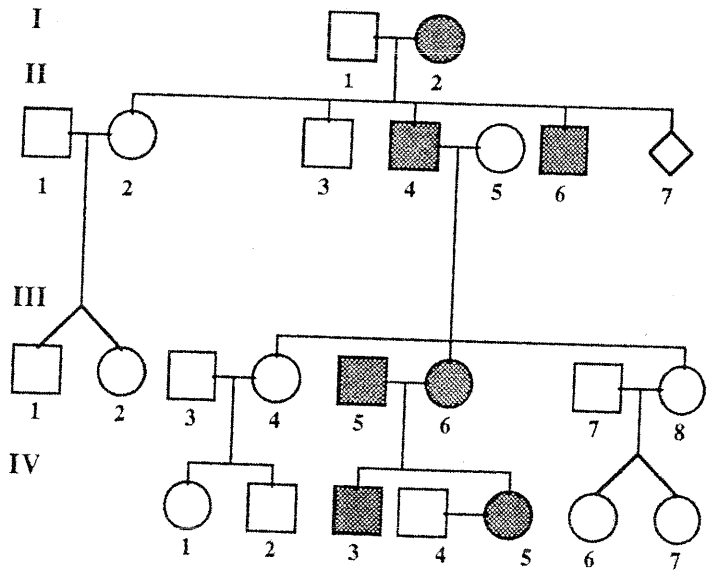
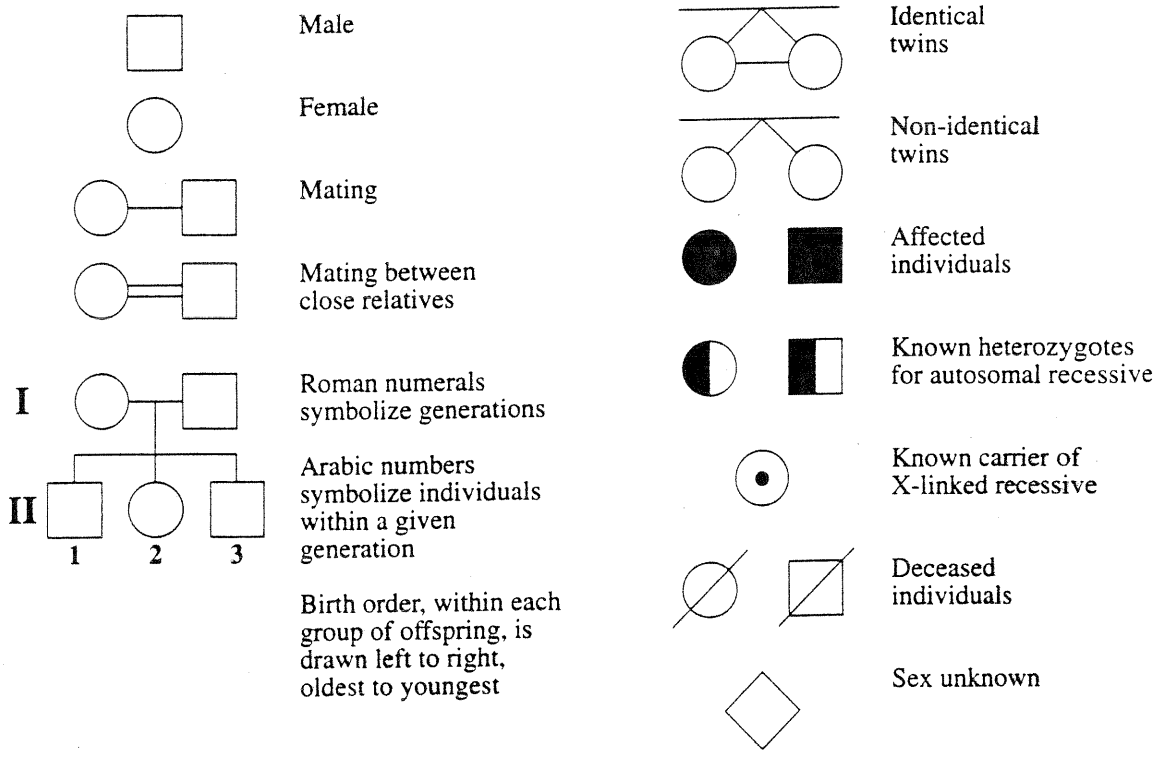


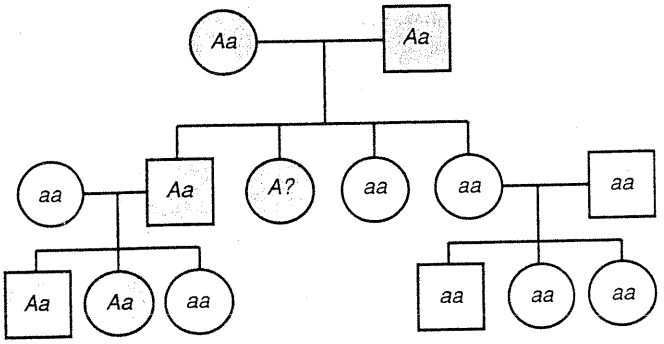
Figure 2-2 Four patterns of inheritance

Autosomal dominant	Autosomal recessive
Males and females are equally likely to have the trait. Traits do not skip generations (generally). The trait is present whenever the corresponding gene is present (generally). There is male-to-male transmission.	Males and females are equally likely to have the trait. Traits often skip generations. Often, both parents of offspring who have the trait are heterozygotes (they carry at least one copy of the allele). Only homozygous individuals have the trait. Traits may appear in siblings without appearing in their parents. If a parent has the trait, those offspring who do not have it are heterozygous carriers of the trait.
X-linked dominant	**X-linked recessive**
All daughters of a male who has the trait will also have the trait. There is no male-to-male transmission. A female who has the trait may or may not pass the gene for that trait to her son or daughter.	The trait is far more common in males than in females. All daughters of a male who has the trait are heterozygous carriers. The son of a female carrier has a 50 percent chance of having the trait. There is no male-to-male transmission. Mothers of males who have the trait are either heterozygous carriers or homozygous and express the trait. Daughters of female carriers have a 50 percent chance of being carriers.

Pedigree Symbols

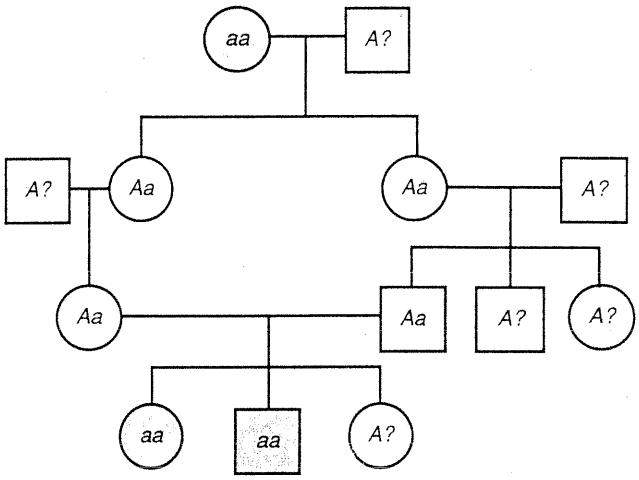


Autosomal Dominant Pedigree Chart
Figure 22.12



Autosomal Dominant Genetic Disorders
- Affected children usually have an affected parent.
- Heterozygotes are affected.
- Two affected parents can produce an unaffected child.
- Two unaffected parents do not have affected children.
- Both males and females are affected with equal frequency.

Key:
AA = Affected
Aa = Affected
aa = Normal



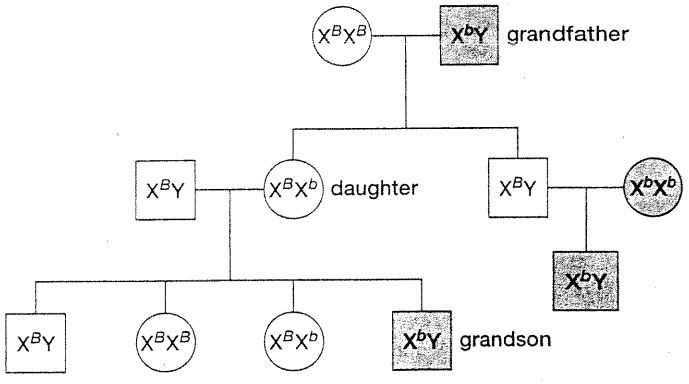
Autosomal Recessive Genetic Disorders
- Most affected children have normal parents.
- Heterozygotes have a normal phenotype.
- Two affected parents will always have affected children.
- Affected individuals who have noncarrier spouses will have normal children.
- Close relatives who marry are more likely to have affected children.
- Both males and females are affected with equal frequency.

Key:
aa = Affected
Aa = Carrier (appears normal)
AA = Normal

Autosomal Recessive Pedigree Chart
Figure 22.9

X-linked Recessive Genetic Disorders

- More males than females are affected.
- An affected son can have parents who have the normal phenotype.
- In order for a female to have the characteristic, her father must also have it. Her mother must have it or be a carrier.
- The characteristic often skips a generation from the grandfather to the grandson.
- If a woman has the characteristic, all of her sons will have it.



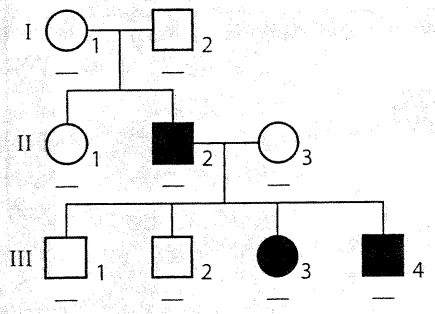
Key:
X^BX^B normal female
X^BX^b carrier female
X^bX^b color-blind female
X^BY normal male
X^bY color-blind male

X-Linked Recessive Pedigree Chart
Figure 23.10

Sample Problem

Problem

The following pedigree shows the inheritance of cystic fibrosis, an autosomal recessive trait, in a family. Identify the genotypes of each family member represented in the pedigree.



What Is Required?

The genotype of each individual in the pedigree.

What Is Given?

The pedigree is given.

Plan Your Strategy

Step 1 Look for an individual with a phenotype that differs from the corresponding phenotype in both parents. This phenotype must result from a homozygous recessive phenotype.

Step 2 Write the symbol for the dominant allele below every individual who does not show the trait.

Step 3 Both parents of the individuals showing the trait must have at least one recessive allele. All the children of a person showing the trait had to receive one recessive allele from this parent.

Act on Your Strategy

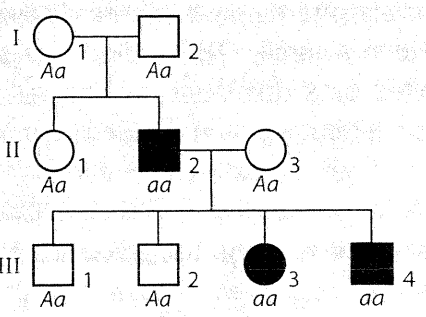
Step 1 Individual II 2 has a different phenotype. Write a homozygous recessive genotype (*aa*) below the symbol for all the individuals who show the trait (II 2, III 3, and III 4).

Step 2 Write the symbol for the one dominant allele (*A*) below all open symbols.

Step 3 Write "*a*" beside "*A*" for I 1, I 2, and II 3.

Step 4 Write "*a*" beside "*A*" for III 1 and III 2.

Step 5 You cannot determine whether II 1 is heterozygous or homozygous dominant.

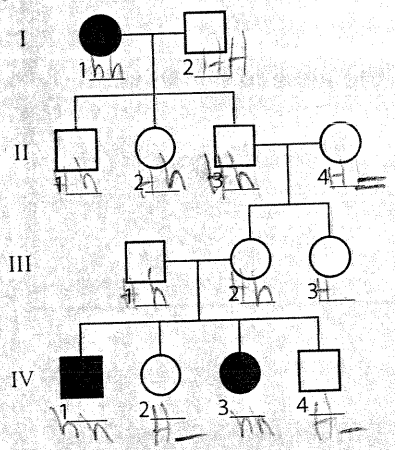


Check Your Solution

Upon checking the pedigree, all genotypes are correct.

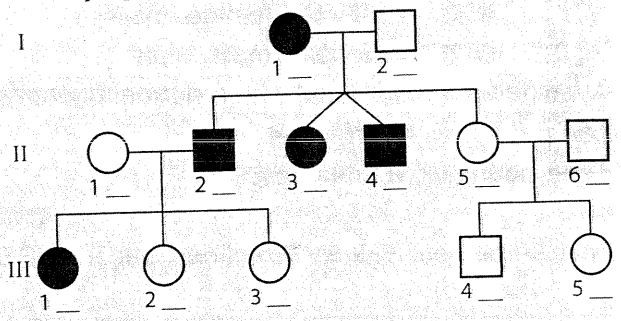
Practice Problems

18. A curved "hitchhiker's thumb" is recessive to a straight thumb. The following pedigree traces the presence of hitchhiker's thumb in a family. Identify the phenotypes and genotypes of all the people shown in the pedigree. Whose genotypes can you not be certain of?

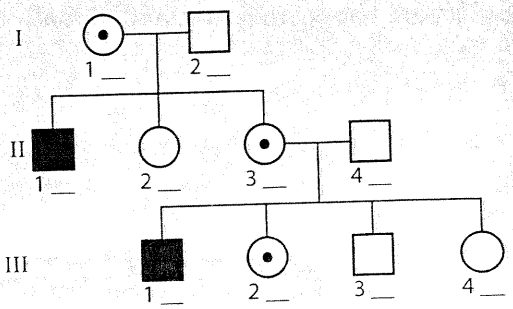


19. In certain families in Norway, woolly hair (hair that looks like sheep's wool) is passed down through the generations. In order for children to have this trait, at least one of their parents must have woolly hair. How is this trait most likely inherited? Draw a pedigree for a family where one of three children and both parents have woolly hair. Identify the genotypes and phenotypes for each individual in the family. Whose genotype can you not be certain of?

20. This pedigree traces tongue rolling in a family. The ability to roll your tongue is controlled by a dominant allele; people with the recessive allele cannot roll their tongue. Identify the phenotypes and genotypes of all the people shown in this pedigree.



21. Duchenne muscular dystrophy is an X-linked recessive trait. The following pedigree shows the occurrence of this disorder in an extended family. Provide the phenotypes and genotypes of all the individuals in the pedigree.



ACTIVITY 7-6

HUMAN PEDIGREES

BACKGROUND

Geneticists often draw *pedigree diagrams* to show the inheritance of a particular genetic trait within a family. These diagrams can help them determine whether a phenotype is controlled by a dominant, recessive, or sex-linked allele. For example, if both parents show a trait but some of their children do not, then the trait is controlled by an *autosomal* (not on a sex chromosome) dominant allele. If neither parent shows the trait, but it appears in one or more of their children, then the trait is controlled by an autosomal recessive allele. If the trait appears primarily in males and it "skips a generation", it is probably a *sex-linked* recessive trait. Many of the traits examined using pedigree diagrams are hereditary disorders.

In a pedigree diagram, each generation is numbered using Roman numerals, with the oldest generation always number I. Each individual within each generation is numbered with an arabic numeral, so that each individual is known by the combination of the generation and the individual numbers, e.g., III-4. The gender and genotype of the individuals are indicated by the following symbols:

○ = normal female
□ = normal male
●, ■ = afflicted female, male
⊘, ▨ = carrier female, male

A *carrier* is an individual with a normal phenotype, but who has the gene in question and can pass it on to offspring.

Time estimate: 60 minutes.

Text Reference: *Biology Directions*, pages 503-507, 533.

PROBLEM

Can human pedigrees be used to determine the probability of a trait appearing in an individual?

HYPOTHESIS

Form your own hypothesis about the effectiveness of pedigrees in determining the probability of a trait appearing in a particular individual.

PROCEDURE

Examine each pedigree and answer the questions that follow.

Part 1. Tay-Sachs Disease

Tay-Sachs disease is caused by a lethal autosomal recessive gene (t). Individuals with Tay-Sachs disease first show the condition at the age of 6 months and die at about 4 years old.

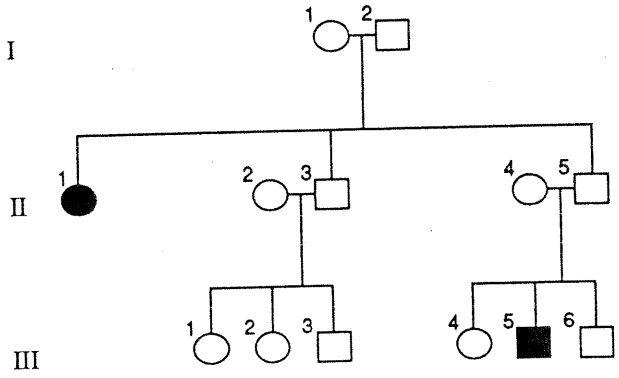


Figure 7.9: Pedigree diagram for a family with a history of Tay-Sachs disease

1. Draw the pedigree in your notebook, and write the genotype for each individual below the circle or square.

Part 2. Sickle-cell Anemia

Sickle-cell anemia is a disease that occurs in about 1 in 500 black children born in North America. Individuals with this disease can suffer from "sickle cell crises" when they are deprived of oxygen by exertion or respiratory ailment. When this happens, red blood cells collapse into a "sickle" shape which can block capillaries (see *Biology Directions* page 533). This causes severe pain, and the lack of blood flow can result in oxygen deprivation which can make the condition even worse.

1. How is this trait inherited?
2. Draw the pedigree in your notebook, and write the genotype for each individual below the circle or square.
3. About 1 in 10 North American blacks are heterozygous for sickle-cell anemia. This number is very high for a trait that can be so detrimental for homozygous individuals. However, research has shown that heterozygous individuals have enhanced resistance to malaria. What effect might this factor have had on the frequency of the sickle-cell gene in populations of North American blacks?

Part 3. Hemophilia

Hemophilia is a recessive sex-linked (found on the X chromosome) condition that used to be called the "bleeder's disease" because the blood

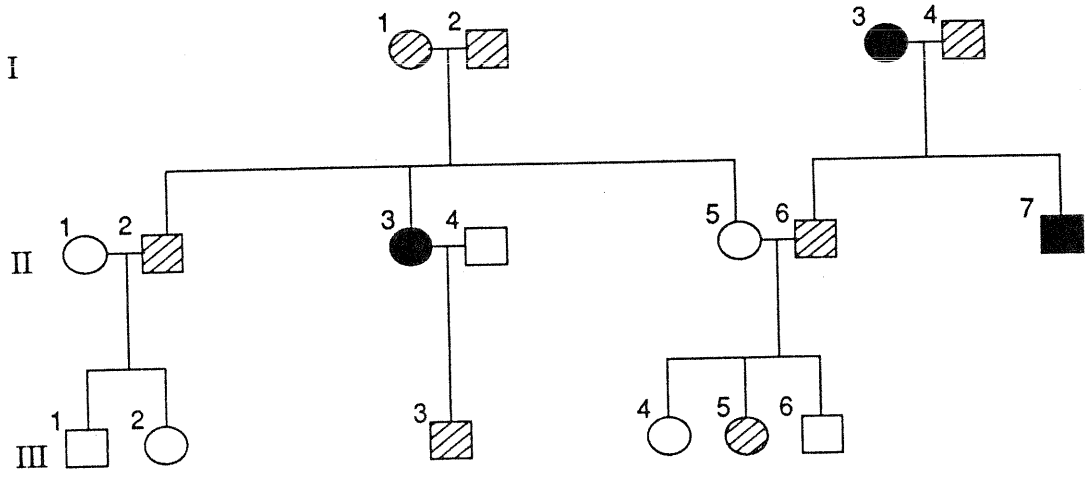


Figure 7.10: Pedigree of a family with some members afflicted with sickle-cell anemia

of affected individuals takes a long time to clot. Normal blood usually clots within five minutes after being placed in a test tube, whereas hemophilic blood may require several hours to clot. Without treatment, even common bruises often lead to serious internal bleeding in the hemophilic individual.

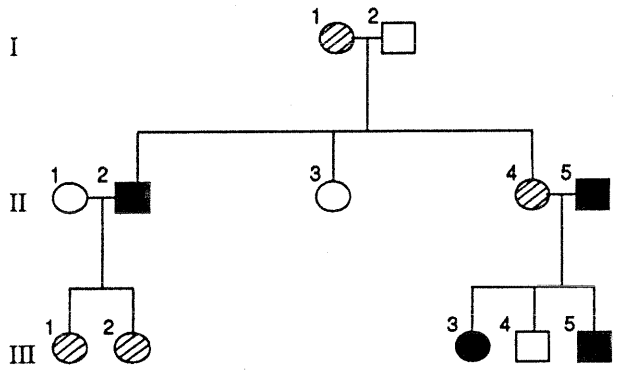


Figure 7.11: Pedigree of a family with a history of hemophilia

1. Draw the pedigree in your notebook. Write in the genotype for each individual, using:
   X^HX^H for a normal female
   X^HX^h for a carrier female
   X^hX^h for a hemophiliac female
   X^HY for a normal male
   X^hY for a hemophiliac male
2. Why do more males have hemophilia than females?
3. Explain how the female (III-3) can have hemophilia.

Part 4. Problems

1. Answer the following questions for both Figures 7.12 and 7.13.
   a. How is this trait inherited?
   b. Draw the pedigree in your notebook, and write in the genotype for each individual.

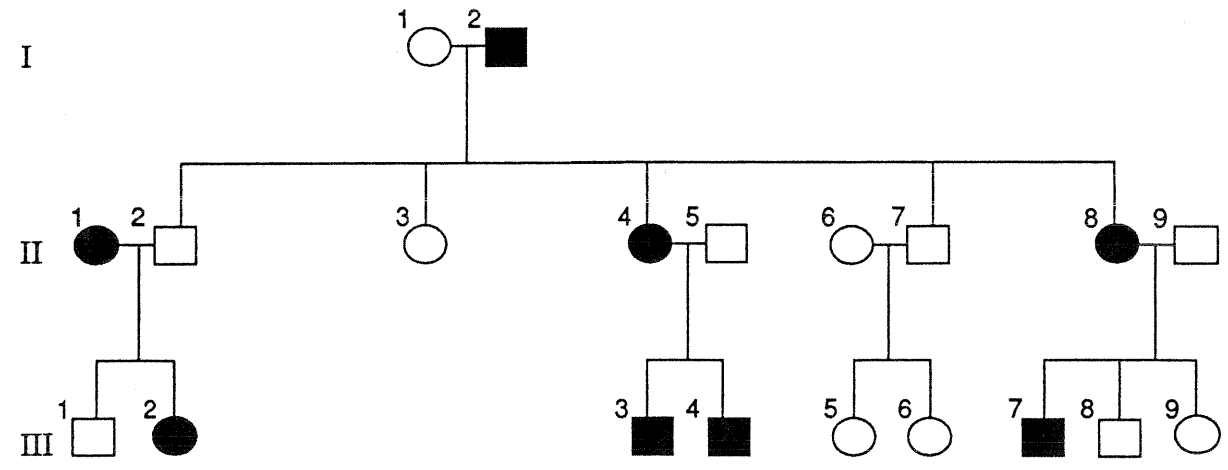


Figure 7.12: Pedigree problem 1

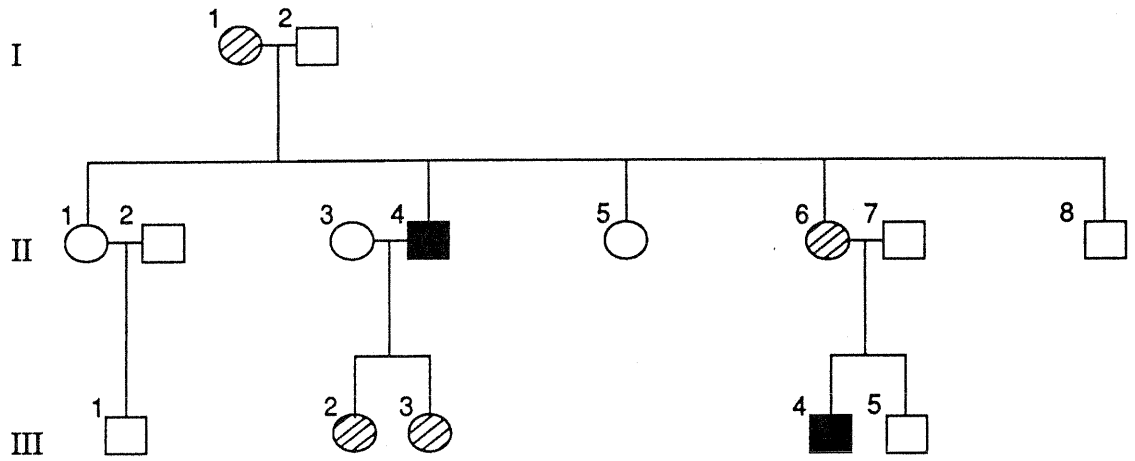


Figure 7.13: Pedigree problem 2

ANALYSIS AND INTERPRETATION

1. Could you determine the genotype of each individual in this Activity?
2. Give an example of an individual where you could work out only the probability of a particular genotype, and calculate that probability.
3. Suppose that you discovered that you and your spouse are both heterozygous for a lethal recessive gene. Would you have children? Explain. Would your decision change if the trait involved was debilitating but not lethal? Explain.
4. Many genetic diseases extract a high cost in terms of health care dollars. Since medicare is funded by government funds, should the government be allowed to legislate who may and who may not have children? Defend your position.