

Progress in human genetics has proceeded slowly. A geneticist uses experimental results to verify the genotypes and phenotypes of living organisms. In humans, the generation time is about 15 to 40 years. Also, the time involved for a new generation to show a certain trait can be several decades. For example, diabetes mellitus in some people may not show up for 30 or 40 years. Other organisms have shorter generation times. For example, fruit flies are easy to study because new generations develop in only ten to twelve days. Traits in these organisms show up much sooner.

Genotypic and/or phenotypic data of human genetic patterns are usually presented as a diagrammed chart called a pedigree. Some pedigree symbols are shown in Table 19-1. A pedigree shows several generations of a family and the people who married into the family (Figure 19-1). Each person in a pedigree can be identified by the Roman numeral for the generation and by an individual number. One difficulty in making a pedigree chart is that sometimes the information is obtained from only a few members of the family. Therefore, it could be inaccurate or unreliable.

Today, geneticists are able to provide a service called "genetic counseling." Families affected with unfavorable traits such as sickle cell anemia or PKU can be given advice about the chances of their future children having these traits.

In this investigation you are to analyze three family pedigrees. From these pedigrees, you are to determine whether the gene for the trait listed is dominant or recessive and determine the genotype and phenotype of each individual in the family.

Procedure

Study each of the following pedigrees of families with certain hereditary traits. Determine whether each of the traits is dominant or recessive. Determine the genotype and phenotype of each individual in each pedigree. Record the genotypes and phenotypes.

SYMBOL	MEANING
□	male
○	female
■ or ●	affected individual
☐ or ☉	stillbirth
○—○	fraternal twins

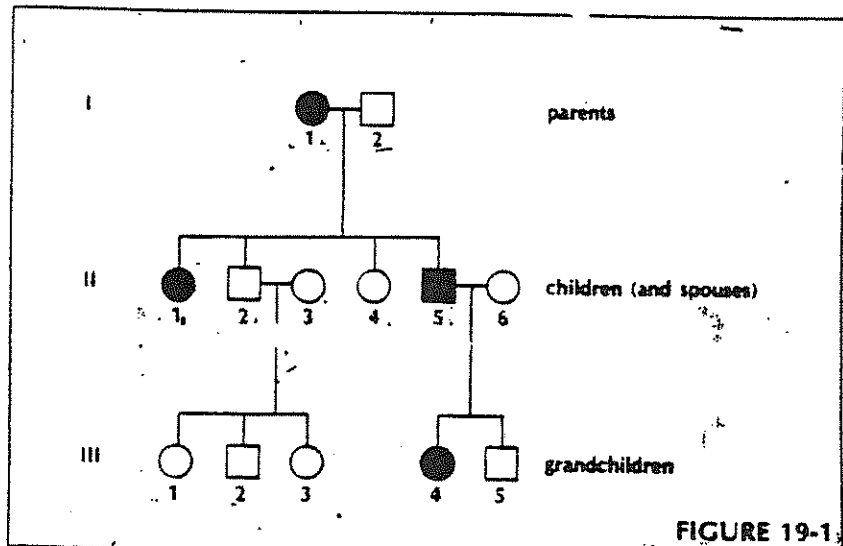


FIGURE 19-1

ENRICHMENT ACTIVITY

Genetics Problem Solving

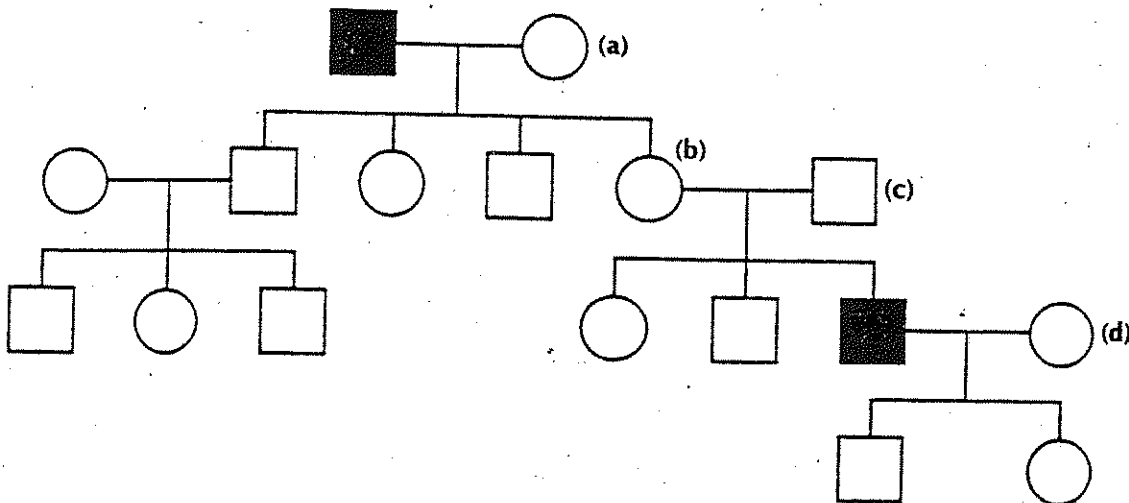
Human Pedigrees

A pedigree is a type of diagram that shows the phenotypes of the members of several generations in a family line with respect to a particular trait. The trait recorded in a pedigree could be a normal trait, such as eye color, or a defective trait, such as a genetic disease. From the patterns of inheritance revealed in a pedigree, it is often possible to determine if a trait is dominant or recessive, and sex-linked or not sex-linked. Pedigrees help genetic counselors assess the probability of a couple having a genetically defective child.

Pedigrees are constructed according to certain rules. Females are represented by circles and males by squares. Shading of a circle or of a square indicates that the individual exhibits the trait. Marriages are represented by horizontal lines connecting a male and a female. Vertical lines connect offspring to marriage lines.

SAMPLE PROBLEM 1

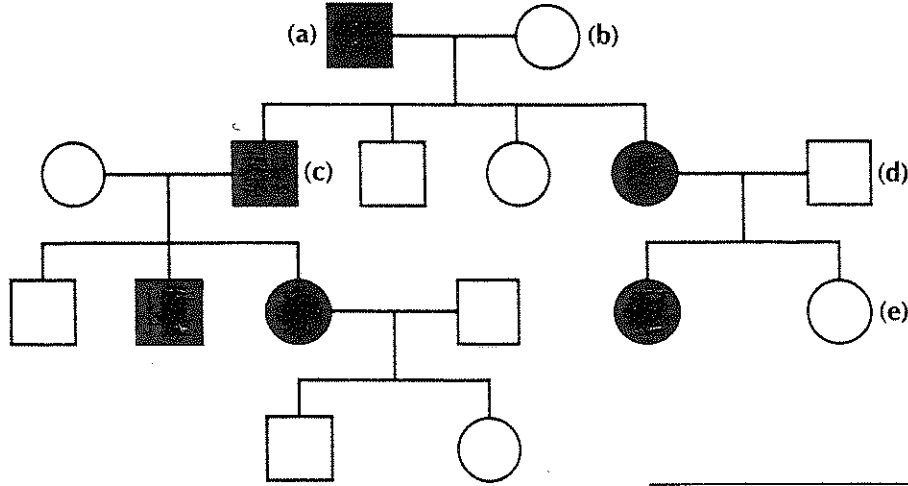
Below is a pedigree for four generations of a family, some of whose members exhibit a particular trait. From the information in the pedigree, determine if the trait is dominant or recessive, and sex-linked or not sex-linked. Which of the lettered individuals is a carrier—that is, heterozygous—for the trait?



Answer One feature of this pedigree is that the trait of interest occurs only in males and appears to be transmitted directly by carrier females to affected offspring. Thus the trait is sex-linked. Since none of the parents of any of the affected individuals exhibits the trait, it must also be a recessive trait. Therefore, the trait is due to a sex-linked recessive allele. Individual (b) must be a carrier in order to have a male offspring with the trait.

SAMPLE PROBLEM 2

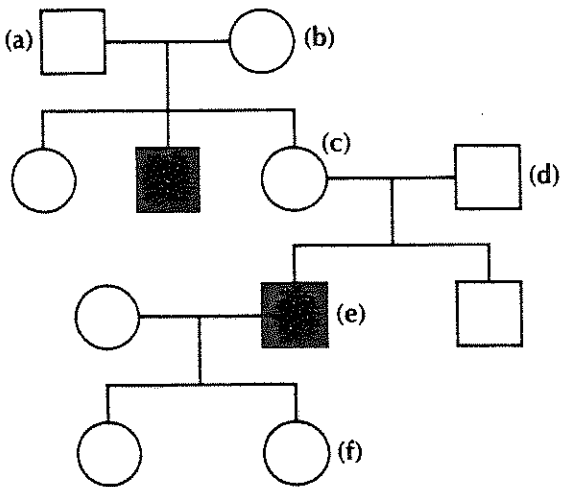
Determine if the trait recorded in the pedigree below is dominant or recessive. Is it sex-linked or not?



Answer The trait appears in at least some offspring of all marriages where one parent has the trait, indicating the trait is dominant. Since the trait occurs in males and females, it is probably not sex-linked. In this pedigree, affected males pass on the trait to some of their sons, a condition that cannot occur when a trait is sex-linked. Thus the trait is a non sex-linked dominant trait.

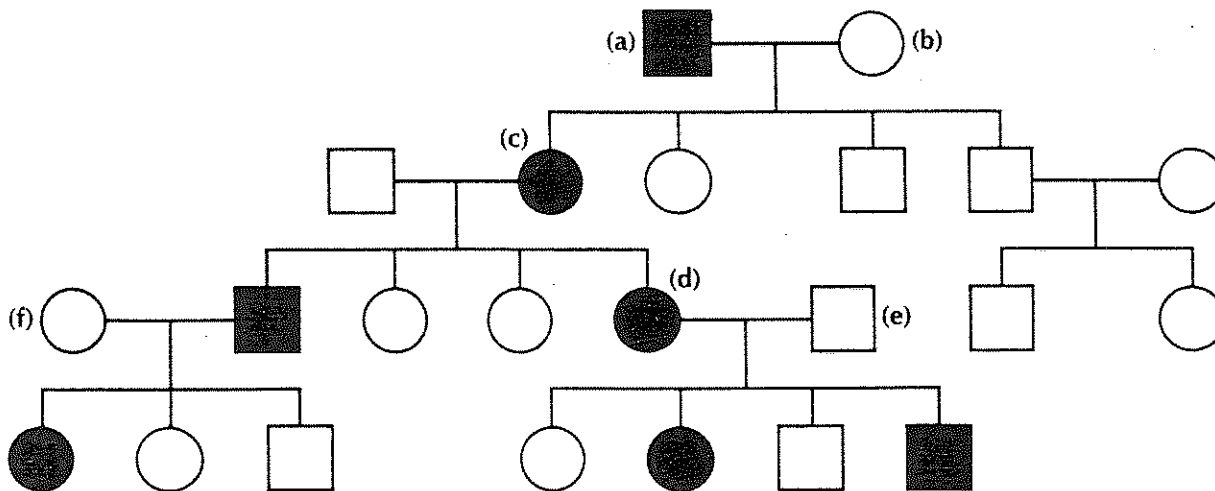
EXERCISES

1. Determine if the trait recorded in the pedigree below is dominant or recessive and sex-linked or not. Which of the lettered individuals carry an allele for the trait?



ENRICHMENT ACTIVITY (continued)
Genetics Problem Solving Human Pedigrees

2. Determine if the trait recorded in the pedigree below is dominant or recessive and sex-linked or not. Which of the lettered individuals are heterozygous for the trait?



3. The pedigree below shows the blood types of some individuals. List all the possible genotypes of each lettered individual.

