

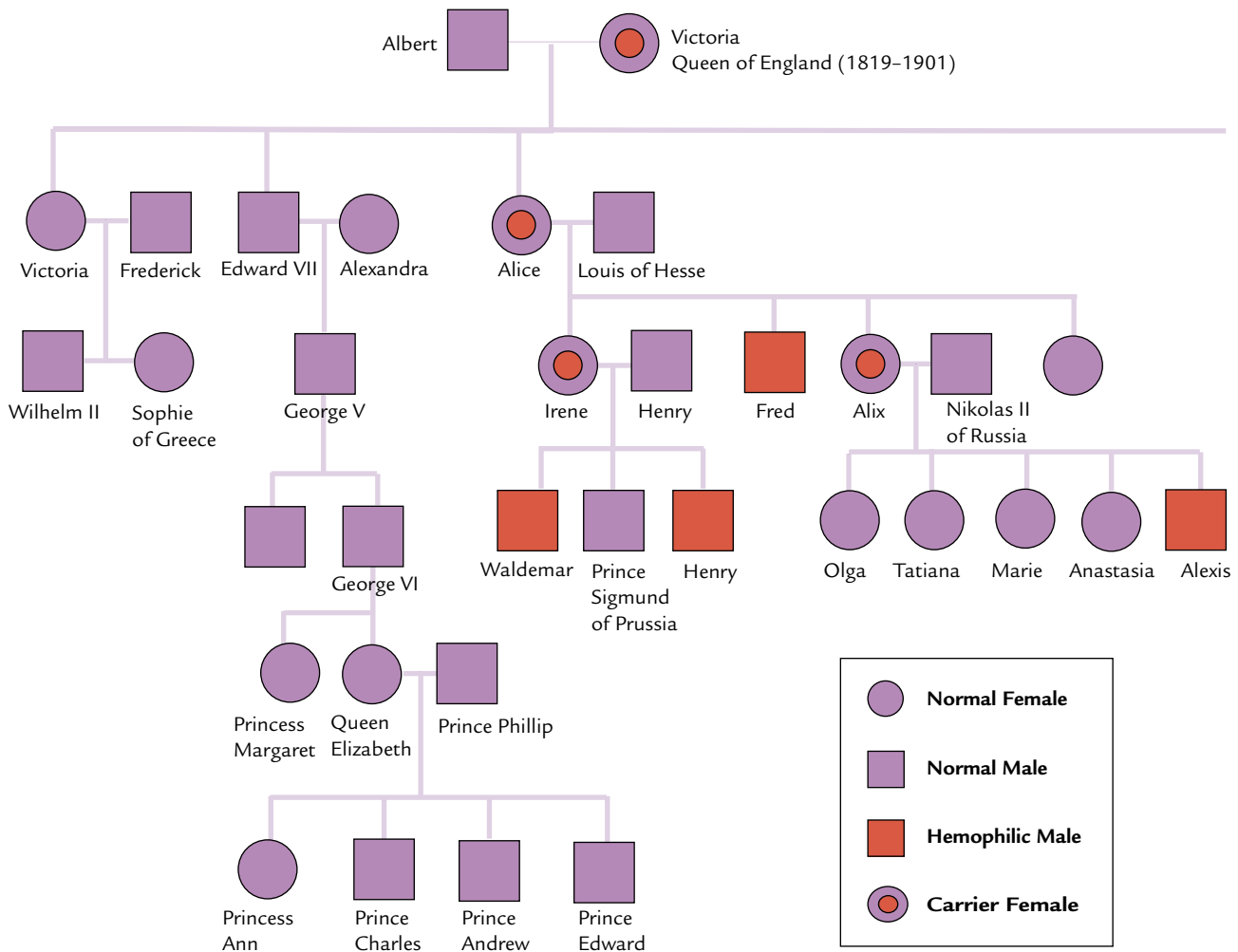
66 Patterns in Pedigrees



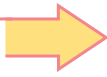
As you now know, genes are inherited and affect the characteristics of an organism. By growing *Nicotiana* seedlings, you've seen how a trait is inherited. You have also seen how Punnett squares can help make predictions about inherited traits in large numbers of offspring.

Studying human inheritance is more difficult. Scientists cannot perform breeding experiments on people. They must use other approaches when studying human genetics. Family histories, such as the one shown below, provide one way to gather evidence about inherited traits in humans.

A PARTIAL PEDIGREE OF HEMOPHILIA IN THE ROYAL FAMILIES OF EUROPE



CHALLENGE



How can you use pedigrees to study human traits?

MATERIALS



For each student

- 1 Student Sheet 66.1, “Pedigree Puzzles”

PROCEDURE

Work with your group to read and discuss pedigrees.

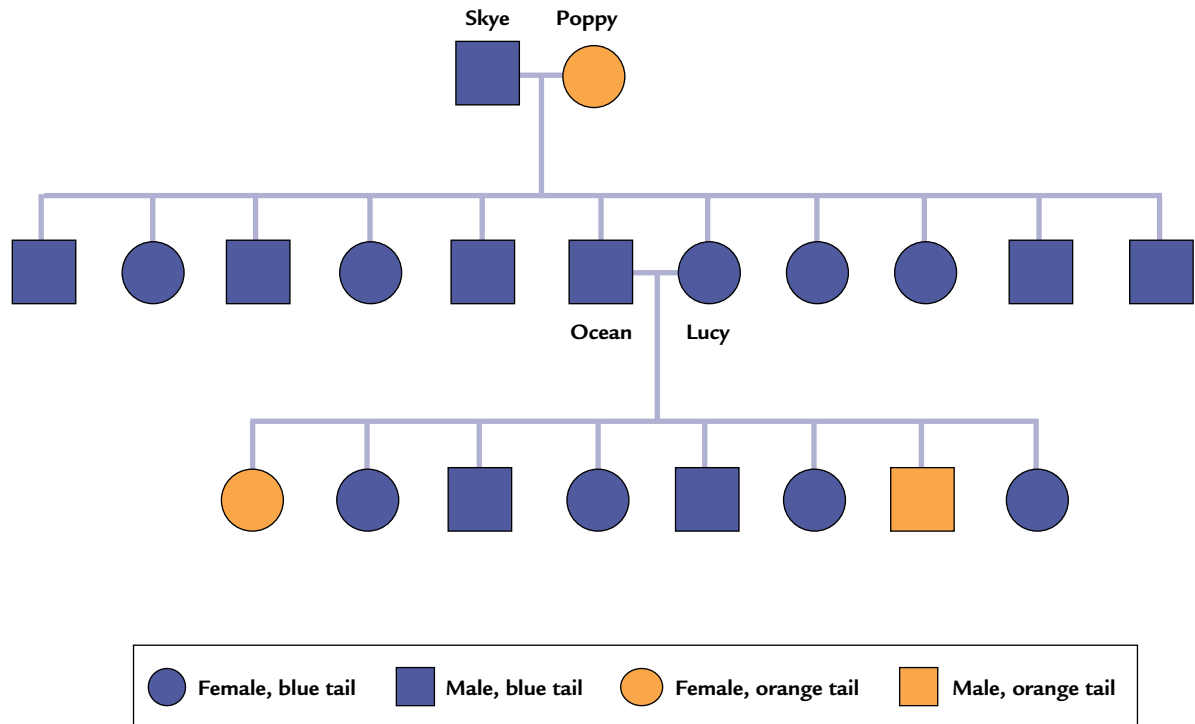
What is a Pedigree?

One approach to studying genes in humans and other organisms is to collect data on a single trait within a family. These data can be used to construct a family tree used for genetic analysis. Such a tree is called a **pedigree**. Researchers use a pedigree to look for patterns of inheritance from one generation to the next. These patterns can provide clues to the way the trait is passed from parents to their offspring (or children).

The figure on the next page shows the pedigree for tail color in the breeding family of Skye and Poppy. Squares are used to represent males and circles are used to represent females. The zoo breeders were concerned that the orange tail color trait might be lost when it did not appear in the second generation, but it reappeared in the third generation.

Activity 66 • Patterns in Pedigrees

CRITTER TAIL-COLOR PEDIGREE



You have used Punnett squares and your knowledge of which trait is dominant to predict what fraction of the offspring of a particular pair of parents are likely to have each trait. But how do scientists find out which trait is dominant when they can't do breeding experiments? They analyze data provided by pedigrees.

STOPPING TO THINK 1

- Look carefully at the figure above. Explain how the information in the pedigree tells you whether orange tail color is dominant or recessive.
- You have used the symbols \mathbf{T} and \mathbf{t} for the alleles of the critter tail-color gene. On Student Sheet 66.1, "Pedigree Puzzles," label each critter in the diagram with the gene combination you think it must have.
- Why don't you know whether each blue-tailed critter in Generation Three is homozygous or heterozygous?

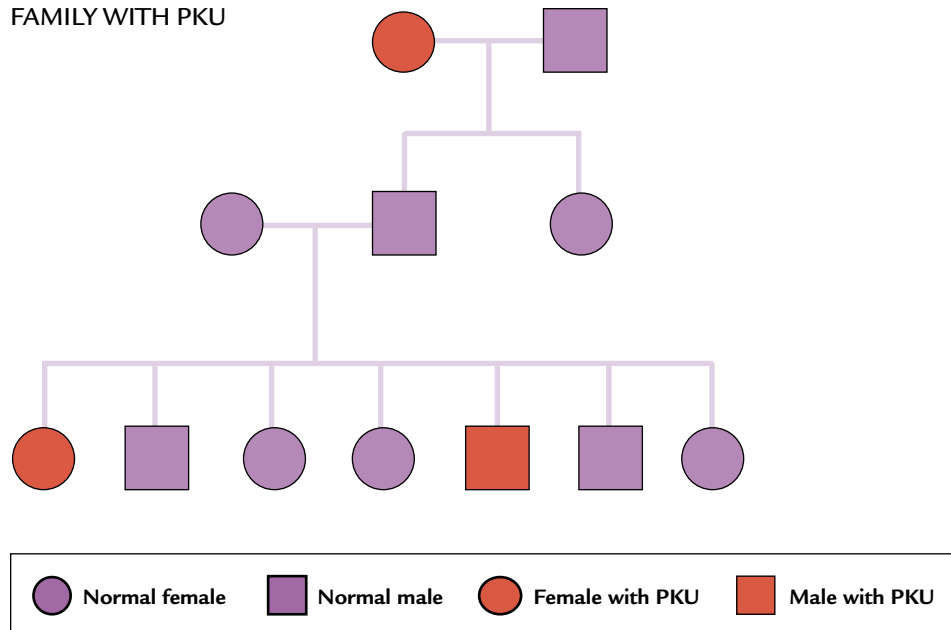
Learning from Data on Human Conditions

Most human traits, such as height, for example, are the results of interactions between many genes and environmental factors. But some hereditary diseases in humans, such as the Marfan syndrome (see Activity 56, “Joe’s Dilemma”), are caused by a single gene. Pedigrees from several generations of a family enable scientists to figure out whether such a condition is dominant or recessive. Once doctors know this, they can predict how likely it is that a child of particular parents will have the condition.

For example, individuals with a condition called PKU, or phenylketonuria (feh-null-key-tun-YUR-ee-uh), cannot break down protein normally. This leads to the build-up of a chemical that causes mental retardation. If PKU is diagnosed shortly after birth, the child can be given a special diet. Children given this special diet for at least the first 10 years of life do not develop the symptoms of the condition. In most of the United States, newborns are routinely tested for PKU within a few days after birth.

Analysis of pedigrees indicates that PKU almost always appears in children of people who do *not* have the condition. The figure below shows a family in which two grandchildren inherited PKU and five did not. These numbers vary from family to family.

FAMILY WITH PKU



STOPPING TO THINK 2

Is PKU likely to be a dominant or a recessive trait? How was it inherited by the individuals as shown in the pedigree? On Student Sheet 66.1, label each individual with the genotype(s) he or she might have.

Hint: Remember that if the condition is dominant, an affected individual could be homozygous or heterozygous. But if it's recessive, an affected individual must be homozygous for the trait.

In the study of genetic diseases, a person who is heterozygous for a recessive genetic condition is called a **carrier**. Such a person does not have the condition, but can pass on an allele for it to his or her children. The recessive allele is hidden, or **masked**—until it shows up in a homozygous individual who has the condition. A person who has a recessive condition is not called a carrier.

Note: In the Cell Biology and Disease unit of *Issues and Life Science*, you learned a very different use of the term *carrier*. When describing infectious diseases, a carrier is someone who does not show symptoms of an infectious disease but can infect other people with the microbe responsible for that disease.

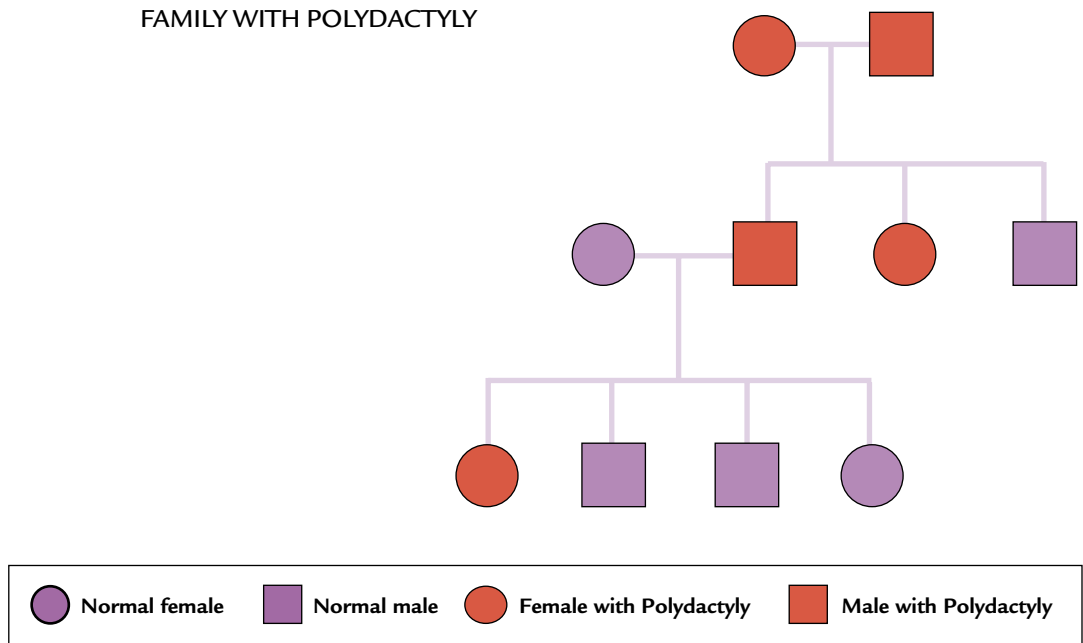
STOPPING TO THINK 3

Why is it impossible for an individual to inherit a recessive condition if only one parent is a carrier for that condition?

Are All Hereditary Conditions Recessive?

PKU is a recessive trait; both parents must be carriers for a person to inherit it. Is this true for all other hereditary diseases, such as the Marfan syndrome? Consider polydactyly (paw-lee-DAK-tul-ee), which causes individuals to have an extra finger or toe on each hand or foot. This is not a dangerous condition, but it does run in families. The figure on the next page shows a typical pedigree of a family with polydactyly. One grandchild in this family has polydactyly, but her three siblings do not. These numbers vary from family to family.

FAMILY WITH POLYDACTYLY



STOPPING TO THINK 4

Is polydactyly likely to be a dominant or a recessive trait? How did the individuals in the pedigree above inherit it? On Student Sheet 66.1, label each individual with the allele combination(s) he or she might have.

Hint: Remember that if the condition is dominant, an affected individual could be homozygous or heterozygous. But if it's recessive, an affected individual must be homozygous for the trait.

Based on such pedigrees, scientists have concluded that polydactyly is a dominant trait. The condition does not skip a generation the way recessive traits can (see the pedigree for the family with PKU).

The Genes for ABO Blood Groups

In certain plants, when purebred red flowers are crossed with purebred white flowers, the offspring are not red or white, but pink. These pink heterozygous flowers show both alleles they possess—the overall effect is an intermediate appearance. Red and white flowers are not dominant or recessive traits, but show **incomplete dominance**.

Activity 66 • Patterns in Pedigrees

Recall the chemical PTC, which you tasted (or did not taste!) in Activity 54, “Investigating Human Traits.” The ability to taste PTC is a dominant trait. However, there can be some incomplete dominance as well: heterozygous individuals may taste PTC less strongly than people with two copies of the tasting allele.

For some other characteristics, two traits both appear at the same time. Since both traits can be observed distinctly in a heterozygous individual, this is known as **co-dominance** (equal dominance).

STOPPING TO THINK 5

Look back at Activity 65, “Breeding Critters—More Traits.” Which characteristic modeled incomplete dominance? Which characteristic modeled co-dominance?

If you completed Activity 46, “Disease Fighters,” in Unit C, “Cell Biology and Disease,” you learned about the ABO blood groups. You saw that people with some blood types have an immune reaction to blood of certain other types; they cannot be given transfusions of these incompatible blood types. In Activity 68, “Searching for the Lost Children,” you will see how blood typing can help solve real-life problems. There are *four* different blood types, A, B, O, and AB. How are blood groups inherited?

After investigating pedigrees from many families, scientists obtained the results listed in the table below.

| ABO Blood Types | | |
|----------------------|---|---------------------------------|
| Parents' Blood Types | | Children's Possible Blood Types |
| O | O | O |
| A | O | A or O |
| B | O | B or O |
| A | A | A or O |
| B | B | B or O |
| A | B | AB, A, B, or O |

Based on these results, figure out the possible genotypes for each of the four blood types. Use Student Sheet 66.1 (page 2) to record your work.

| Blood Type | Possible Allele Pairs |
|------------|-----------------------|
| O | _____ |
| A | _____ or _____ |
| B | _____ or _____ |
| AB | _____ |

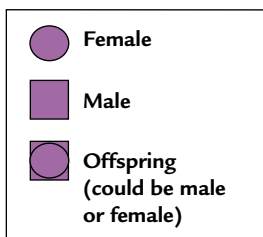
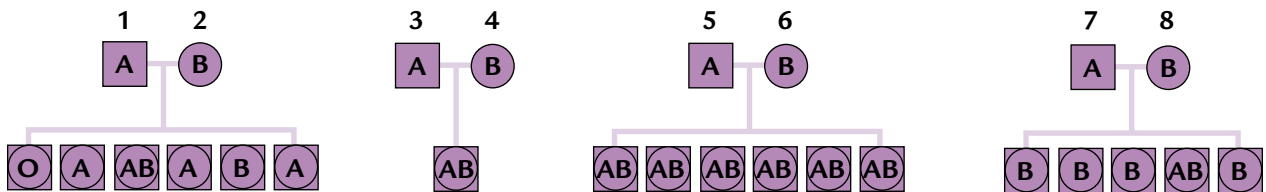
STOPPING TO THINK 6

Which two blood types are co-dominant? Which blood type is recessive?

ANALYSIS

- The following pedigrees represent the blood types in four unrelated families. In each case, the parents have Type A and Type B blood.

FOUR BLOOD TYPE PEDIGREES



- Which of the eight parents are definitely heterozygous for the Type O allele? Explain.
- Which of the eight parents are probably not heterozygous for the Type O allele? Explain.
- Can you be certain that the parents you named in response to Question 1b do not have a Type O allele? Explain.

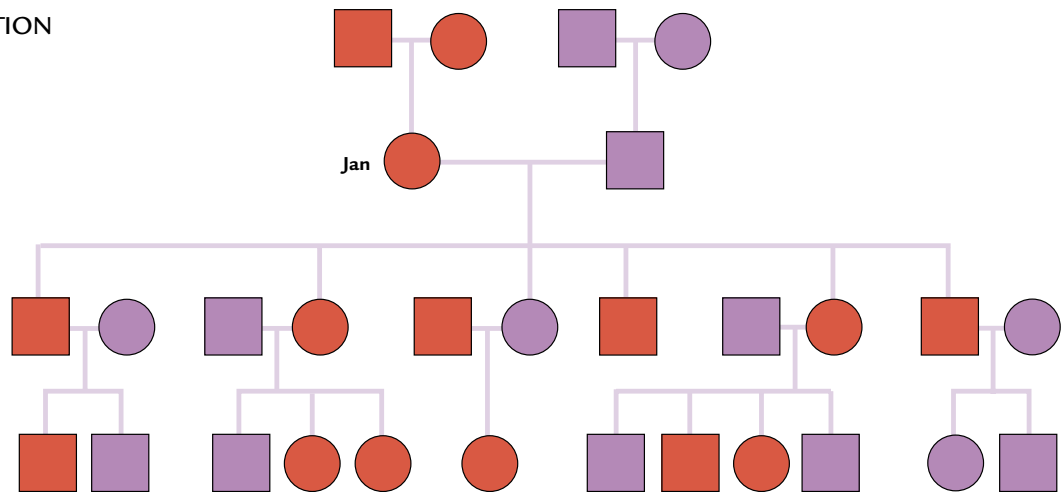
Activity 66 • Patterns in Pedigrees


RED represents the presence of a genetic condition in all the pedigrees for Analysis Questions 2–4.

2. The pedigree shown below represents Genetic Condition One. Use your understanding of pedigrees and this pedigree to answer the following questions. Use Student Sheet 66.1 (page 3) to try out allele combinations for related individuals.

- Is Genetic Condition One most likely a dominant or a recessive trait? Explain your reasoning.
- Is Jan most likely to be homozygous dominant, heterozygous, or homozygous recessive?

GENETIC CONDITION ONE

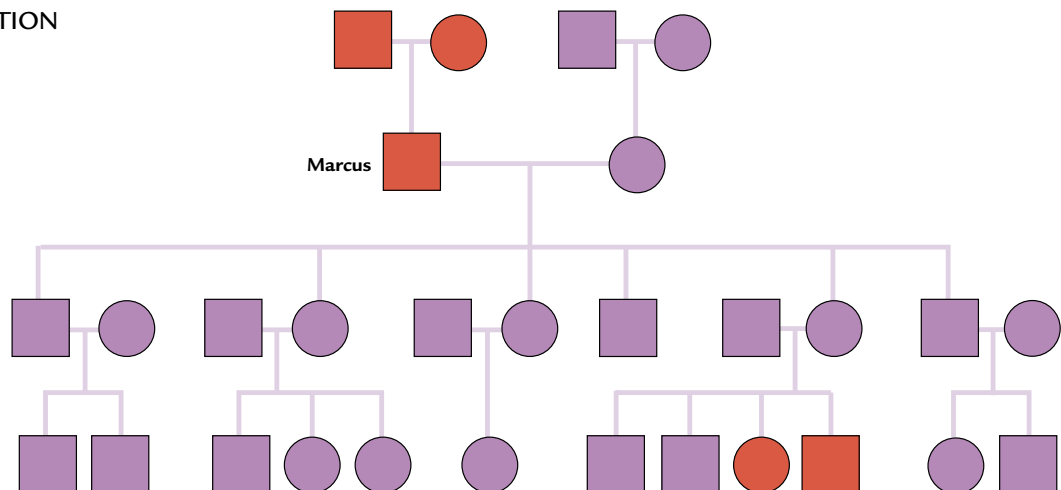


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3. The pedigree shown below represents Genetic Condition Two.

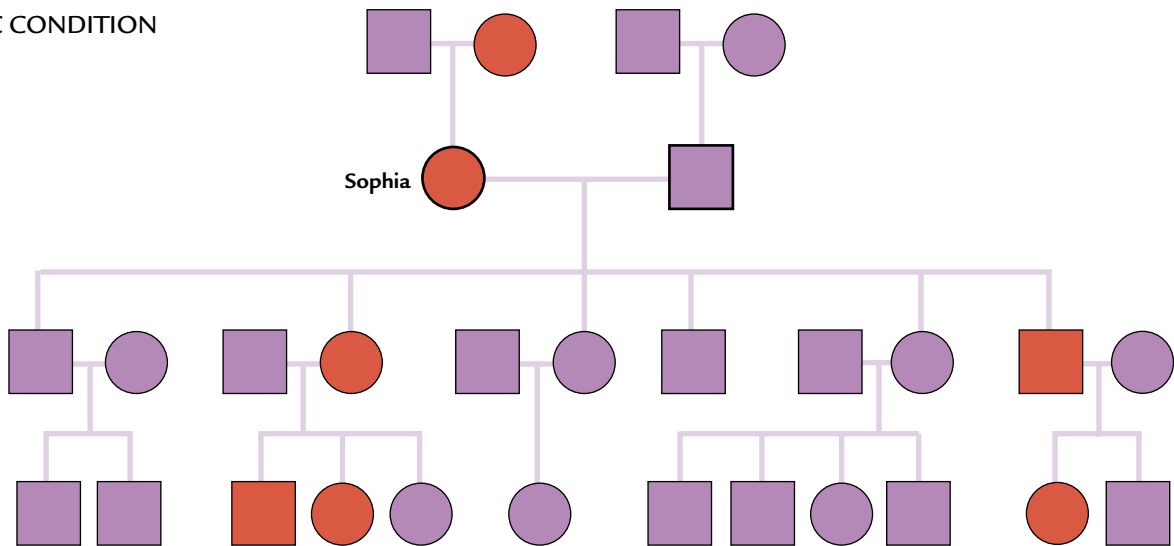
 - Is Genetic Condition Two most likely a dominant or a recessive trait? Explain your reasoning.
 - Is Marcus most likely to be homozygous dominant, heterozygous, or homozygous recessive?

GENETIC CONDITION TWO



4. The pedigree shown below represents Genetic Condition Three.
- Is Genetic Condition Three most likely a dominant or a recessive trait? Explain your reasoning.
 - Is Sophia most likely to be homozygous dominant, heterozygous, or homozygous recessive?

GENETIC CONDITION THREE



5. The term *carrier* is used very differently in genetics than in the study of diseases.
- What is being “carried” by a genetic carrier? What is being “carried” by a disease carrier?
 - How does transmission occur for genetic conditions? How does transmission occur for infectious diseases?