7.2

Mendel and Heredity

These snapdragons show the wide variety of colors possible within this species. This is just one example of the endless variation found in nature.

CAN YOU EXPLAIN IT?

FIGURE 1: Certain types of vegetables, such as broccoli, make some people cringe. Others cannot get enough of it. What causes this difference in taste preferences?

If the idea of a big plate of broccoli makes you want to push your chair away from the table, you are actually not alone. Some people do not like the flavor of broccoli. However, plenty of others do enjoy the taste of this vegetable. Humans have variations in taste preferences, just as we have variations in hair or eye color. What accounts for these differences? Do you like all the same foods as your parents or siblings? Are taste preferences determined by your genes, or do they depend on other influences, such as your environment?

Predict Do you think food preferences are passed down from parents to their children, or does the environment play a role? Explain your answer.

Gather Evidence
As you explore the lesson, gather evidence for how traits are inherited through genes passed down from parents to offspring.

Mendel's Groundwork for Genetics

One of the most important outcomes of sexual reproduction is the variety in traits that results from a shuffling of genes. These traits are distinguishing characteristics that are inherited. Scientists have known for a long time that traits in organisms vary. Scientists also saw that offspring often looked similar to their parents, but not always. What remained a mystery was *why* traits vary.

FIGURE 2: These cats show a variety of inherited traits.

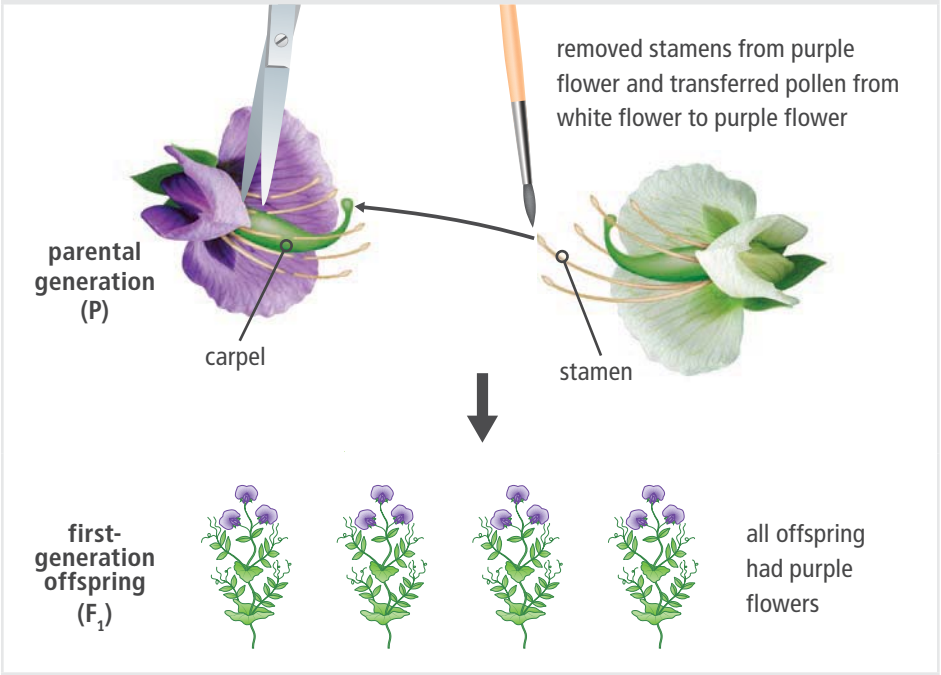
Collaborate With a partner, identify at least three traits that vary among the cats shown in Figure 2.

Mendel's Experimental Design

Our current understanding of heredity comes from a foundation laid in the mid-1800s by an Austrian monk named Gregor Mendel. Mendel's detailed experiments using pea plants led to some important changes in the way scientists viewed the transmission of traits. Scientists of the time commonly thought that parents' traits were blended in offspring, like mixing two colors of paint. However, this idea failed to explain how specific traits on one end of the trait spectrum are observed throughout many successive generations, without all being blended or "diluted."

Mendel chose to work with pea plants based on their fast rate of reproduction and the fact that he could easily control their pollination. He began with purebred plants as the parent generation. Purebred means, for example, that a purple flowering pea plant only produces offspring that have purple flowers when allowed to self-fertilize. During his experiments, Mendel prevented self-fertilization by controlling which plants were able to reproduce. He crossed plants with specific traits by interrupting the self-fertilization process. He then observed the results of each cross. Mendel also used mathematics to analyze the experimental data gathered from hundreds of pea plant crosses.

FIGURE 3: Mendel removed the male parts of flowers and then fertilized the female parts with pollen from a different plant.



Explore Online

Hands-On Lab

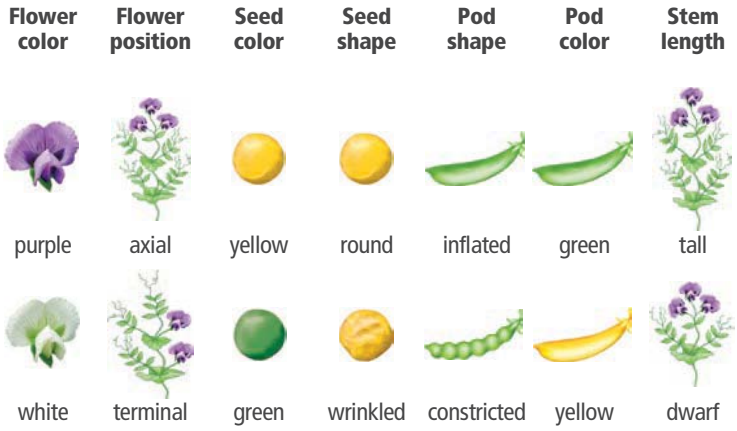
Investigating Traits and Heredity Plan and conduct an investigation to determine how albinism is inherited in tobacco plants.

Analyze Why did Gregor Mendel pollinate the plants himself rather than let the plants self-fertilize?

Mendel's Observations

During his experiments, Mendel observed seven traits in the pea plants. We now know that these specific traits are associated with genes on different chromosomes or are far enough apart on the same chromosome to allow for crossing over. However, Mendel did not know this. The traits Mendel studied are shown in Figure 4. Each trait shows a simple "either-or" characteristic; they do not show an intermediate form. For example, the plant is either tall or short, but not medium in height. The selection of these traits that occur in the "either-or" fashion played a crucial role in helping Mendel identify the patterns he observed. Had he chosen different traits or a different species for his experiments, he may not have come to the same conclusions.

FIGURE 4: Mendel worked with seven traits in pea plants for his experiments.

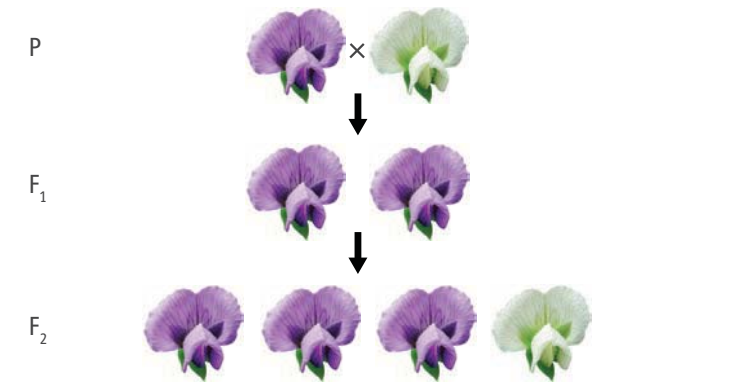


Explain Figure 4 shows the characteristics that Mendel noticed before he set up his experiments. What is one question you would ask about how these traits are passed down from one plant generation to the next?

A genetic cross is the mating of two organisms. When Mendel pollinated a specific female flower of a plant with the pollen from another plant, he carried out a cross. Through his experiments, Mendel was able to observe the results of specific crosses.

Two of Mendel's experimental crosses are shown in Figure 5. In the first experiment, he crossed a purebred white-flowered pea plant with a purebred purple-flowered pea plant. These original plants are the parents—or P—generation. The offspring that result from such a cross are called the first filial—or F_1—generation. In the second experiment, Mendel let the F_1 generation self-fertilize, meaning he did not control their pollination himself. Recall that both F_1 plants had purple flowers. The offspring from these crosses, referred to as the F_2 generation, showed a different set of traits.

FIGURE 5: Purebred white and purple plants were crossed to make the F_1 generation. F_1 plants then self-fertilized, making F_2 plants.



Collaborate
Discuss these questions with a partner.

1. What pattern occurred when the P generation was crossed?
2. What patterns occurred when the F_1 generation was crossed?
3. What questions do you think Mendel would have asked after seeing these results?

Mendel performed similar crosses with F_1 generation plants, which are monohybrids. A monohybrid results from crossing two parents with different variations of a trait. He observed the original traits in the F_2 plants. In all cases, the offspring of these crosses showed many plants with one version of a trait and some plants with the alternate version. The results of his crosses are shown in Figure 6.

Data Analysis

Mendel's Data

FIGURE 6: Mendel allowed the F_1 hybrid plants to self-fertilize, resulting in the reappearance of some previously hidden traits.

Mendel's Monohybrid Cross Results			
F_1 Traits	Dominant	Recessive	Ratio
Pea shape	5474 round	1850 wrinkled	2.96:1
Pea color	6020 yellow	2001 green	3.01:1
Flower color	705 purple	224 white	3.15:1
Pod shape	882 inflated	299 constricted	2.95:1
Pod color	428 green	152 yellow	2.82:1
Flower position	651 axial	207 terminal	3.14:1
Plant height	787 tall	277 short	2.84:1

Analyze Answer the following questions about Mendel's data.

1. What patterns do you notice in the data?
2. What questions might Mendel have asked after seeing this data?

Mendel's Conclusions

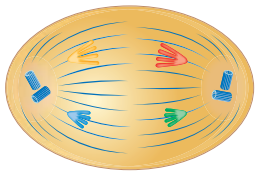
After making careful observations of his experiments and reviewing the data, Mendel realized that certain traits, such as white flowers, had not disappeared; they were just temporarily masked. They also had not been altered by other traits or blended to form a new trait. Mendel concluded that traits are inherited as discrete "factors" that pass from the parental generation to the offspring.

Recall that each gamete of a diploid organism has only one version of a gene, because gametes are haploid, or have half the number of chromosomes as body cells. During meiosis, homologous chromosomes separate and are deposited into gametes. Two gametes fuse during fertilization, so the resulting organism has two copies of each gene, one from each parent. This knowledge, unknown to Mendel, parallels his experimental results and his conclusions about inheritance. The separation of alleles during gamete formation became known as the Law of Segregation.

Gather Evidence What forms of evidence offer support for Mendel's conclusion that traits are inherited as discrete units from the parental generation?

Explain During anaphase I of meiosis, copies of the same gene are separated as homologous chromosomes move to opposite sides of the cell. These chromosomes may or may not contain the same genetic information. Use evidence from meiosis to explain how gene separation occurs and why gametes only have one copy of each gene. How does the process of meiosis support the Law of Segregation?

FIGURE 7: Anaphase I



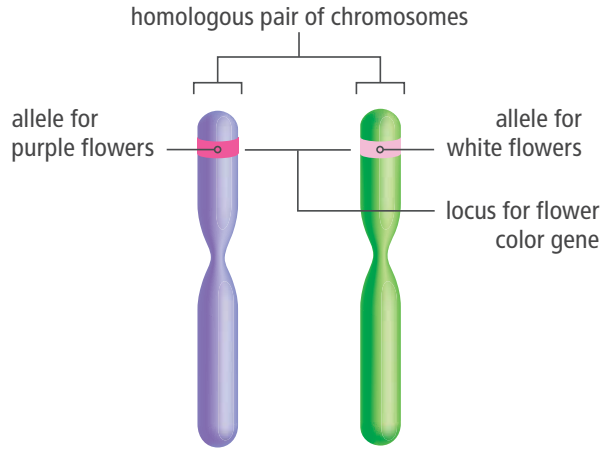
Traits, Genes, and Alleles

We know a lot about DNA and genes today, but this information was discovered long after Mendel's time. However, Mendel did correctly hypothesize that there was a hereditary factor that carried genetic information. We now call those factors genes.

Genes and Alleles

A gene is a piece of DNA that provides a set of instructions to a cell to make a certain protein. Each gene has a locus, which is a specific location on a pair of homologous chromosomes. You can think of the locus as the "address" that tells where a gene is located on a chromosome. In human cells, there are 23 pairs of homologous chromosomes, for a total of 46. Genes located on chromosomes, which get passed on to offspring during reproduction, are the basis for heredity. What Mendel essentially revealed is that it is not the traits that are passed from one generation to the next, but rather the genes that are responsible for those traits.

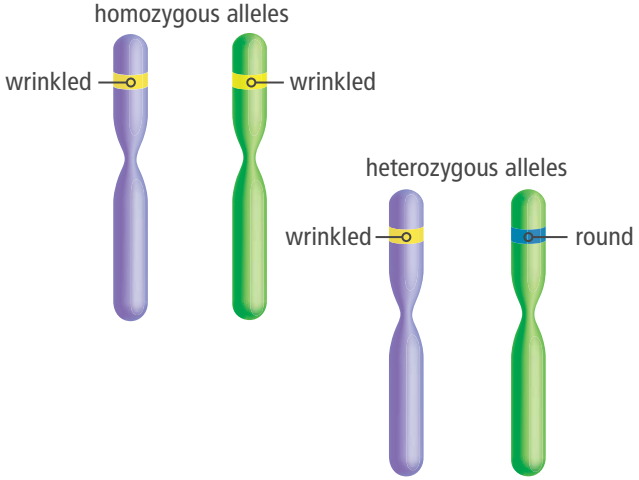
FIGURE 8: Alleles are different forms of a gene. They are located at the same position on homologous chromosomes.



Genes contain genetic information, but this information varies widely from one organism to another due to different alleles. An allele is any of the alternative forms or versions of a gene that may occur at a specific locus. Human cells have two alleles for each gene, which are found on homologous chromosomes. You receive one allele from one parent and one allele from your other parent. The same is true for almost all organisms that reproduce sexually, including pea plants. The traits observed in Mendel's experiments, such as flower color or plant height, resulted from varying alleles.

Explain How is an allele related to a gene?

FIGURE 9: Heterozygous and Homozygous Alleles



Combinations of Alleles

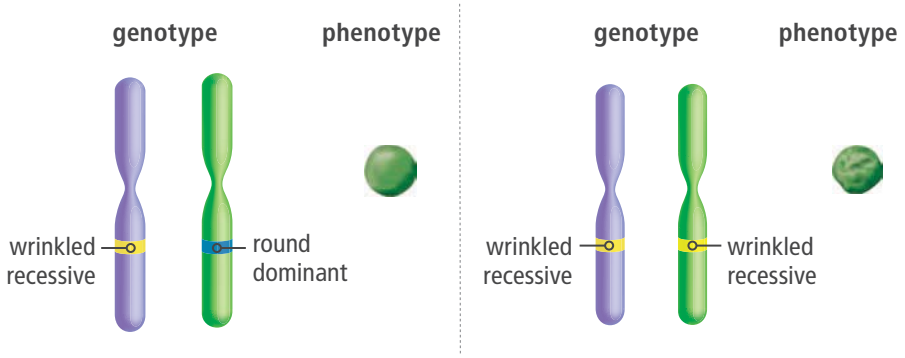
Your body cells contain two alleles for each gene. These alleles may be the same, or they may be different. The term homozygous describes two of the same alleles at a specific locus. The term heterozygous describes two different alleles at the same locus. For example, you may inherit an allele for freckles from one parent and another allele for no freckles from your other parent. The same holds true for pea plants. A pea plant may have a purple flower allele and a white flower allele, making it heterozygous for that trait.

Gather Evidence What is one question you could ask about how traits are expressed when an organism has heterozygous alleles for a trait?

Traits

When describing homozygous or heterozygous pairs of alleles, we are referring to an organism's actual genetic makeup. This is known as its genotype. If a pea plant has one allele for round seeds and one allele for wrinkled seeds, it is said to be heterozygous. Both of these alleles make up its genotype even though one trait will be masked. The actual physical characteristics, or traits, of an individual make up its phenotype. The plant might have an allele for wrinkled seeds, but the phenotype expressed is for round seeds.

FIGURE 10: Only the dominant allele is expressed when two different alleles for a gene are present.



Gather Evidence Based on what you know about Mendel's studies on purple and white flowers, why can genotype be different from phenotype?

Sometimes only one allele in the pair will affect the trait. As Mendel's results demonstrated, in some cases one allele may be dominant over another allele. A dominant allele is the allele that is expressed when two different alleles or two dominant alleles are present. A recessive allele is the allele that is only expressed when two recessive copies occur together.

The allele combination, or genotype, of an organism is often represented by a set of letters. Because each body cell contains two alleles per gene, two letters are needed to represent each allele in the pair. Uppercase letters represent dominant alleles, and lowercase letters represent recessive alleles.

In the chromosomes shown in Figure 10, the dominant allele, *R*, codes for round peas. The recessive allele, *r*, codes for wrinkled peas. The round phenotype will occur if one or two copies of the dominant allele is present. So plants that are homozygous dominant (*RR*) or heterozygous (*Rr*) will have round peas. The wrinkled phenotype, on the other hand, occurs only when two copies of the recessive allele are present. Only plants with the homozygous recessive (*rr*) genotype will have wrinkled peas.

Analyze In pea plants, *T* represents the allele for a tall plant, which is a dominant trait, and *t* represents the allele for a dwarf, or short plant, which is the recessive trait. Identify whether the genotypes *Tt*, *tt*, and *TT* are homozygous dominant, homozygous recessive, or heterozygous. Then identify the phenotype for each.

Explain Use what you have learned about Mendel's contributions to genetics to answer the following questions.

1. When Mendel crossed two purple-flowered plants from the F_1 generation, he found that out of every four flowers, three were purple and one was white. Which of these traits, purple or white, is most likely to be the dominant trait? Explain your reasoning.
2. Write two questions you could ask to learn more about how food preferences, such as distaste for broccoli, are passed from parents to offspring.

Extending Mendelian Genetics

Mendel's use of pea plants ensured that he would be able to follow easily predictable dominant and recessive patterns of inheritance. We now know that most phenotype expression is much more complex. Very few human traits follow the dominant and recessive relationship, or "Mendelian" rules of inheritance.

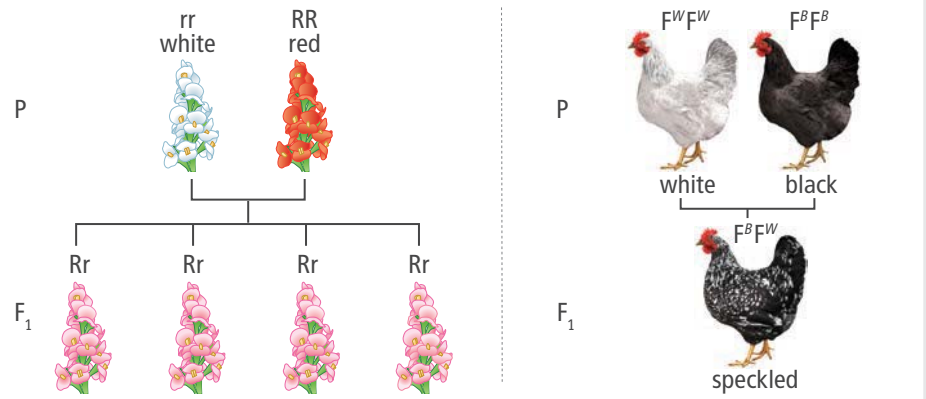
Complex Patterns of Inheritance

Mendel's basic theory of heredity was correct, but his research could not have explained all of the continuous variations for many traits. Many traits result from alleles with a range of dominance, rather than a strict dominant and recessive relationship.

Incomplete Dominance and Codominance

Sometimes alleles show incomplete dominance, in which a heterozygous phenotype is somewhere between the two homozygous phenotypes. This yields a blended result. For example, a cross between a snapdragon with white flowers and a snapdragon with red flowers results in offspring with pink flowers. Sometimes, both alleles of a gene are equally expressed and appear in the phenotype. These alleles show codominance, and both traits are fully and separately expressed. For example, when a certain breed of white-feathered chicken is crossed with the black-feathered phenotype of the same breed, their offspring have feathers that are speckled black and white.

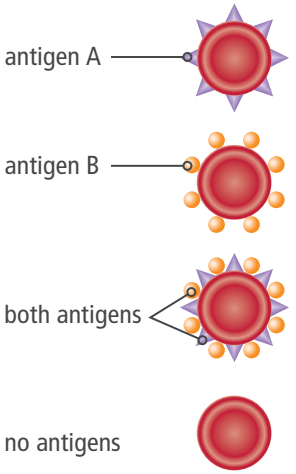
FIGURE 11: Incomplete Dominance and Codominance



Multiple Alleles

In some cases there are more than two alleles possible in a population. Human blood type is an example of multiple alleles. The three alleles are called I^A, I^B, and *i*. Both I^A and I^B result in a protein, called an antigen, on the surface of red blood cells. Allele *i* is recessive and does not result in an antigen. Someone with a genotype of I^Ai will have type A blood, and someone with a genotype of I^Bi will have type B blood. I^A and I^B alleles are also codominant. That means someone with a genotype of I^AI^B will have type AB blood. People with an *ii* genotype have red blood cells without an antigen, and they have type O blood.

FIGURE 12: Human blood type is controlled by multiple alleles, two of which are codominant.



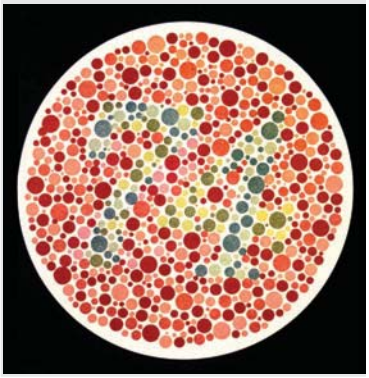
Sex-Linked Traits

Recall that humans have 23 pairs of chromosomes and that the last pair is referred to as sex chromosomes. These chromosomes—X and Y—contain different genes, which make a unique pattern of inheritance. Many of the genes seen on the X chromosome do not have corresponding genes on the Y chromosome, simply because the Y chromosome is so much smaller. Males only have one copy of the Y chromosome, so any recessive gene on a Y chromosome will be expressed. Any recessive gene on an X chromosome also will be expressed in males, because there is no second X chromosome to mask the recessive allele's expression. The genes located on an X or Y chromosome are referred to as sex-linked genes. Red-green colorblindness is an example of a trait caused by a sex-linked gene that occurs more often in males.

Females have double the number of genes located on an X chromosome, but they do not need double the number of their associated proteins. A process known as X inactivation solves this dilemma. Only one X chromosome is active, while the other is silenced or has very few active genes. X inactivation results in more balanced gene expression between males and females.

Explain The gene for red-green colorblindness is located on the X chromosome. Does the mother or father pass the gene for colorblindness to sons? Explain your answer.

FIGURE 13: People with red-green colorblindness cannot distinguish between the colors red and green.



Polygenic Traits

In contrast to the traits studied by Mendel, most plant and animal traits are actually the product of multiple genes. Very few traits in humans are controlled by a single gene. Your height is an example of a polygenic trait, in which multiple genes contribute to the overall phenotype observed. The height genes you inherit from your mother and father accumulate, and the final height that you are likely to reach is due in part to the cumulative effect of these genes. Scientists have discovered over 600 genes that affect height. These complex traits show a continuous range of phenotypes from very short to very tall. Polygenic traits often show a bell-shaped curve when graphed. Many people fall around the average, and very few show one extreme or the other.

Epistasis

Another polygenic trait is fur color in mice and in other mammals. In mice, at least five different genes interact to produce the phenotype. Two genes give the mouse its general color. One gene affects the shading of the color, and another gene determines whether the mouse will have spots. But the fifth gene involved in mouse fur color can overshadow all of the others. In cases such as this, one gene, called an epistatic gene, can interfere with the expression of other genes. Genes that modify the expression of another gene are said to show epistasis.

In albinism, a single epistatic gene interferes with the expression of other genes. Albinism is characterized by a lack of pigment in skin, hair, and eyes. A mouse that is homozygous for the alleles that prevent the coloration of fur will be white, regardless of the phenotypes that would normally come from the other four genes. A person with two recessive alleles for albinism will have very light skin, hair, and eyes, regardless of the other genes he or she has inherited.

FIGURE 14: Albinism in this wallaby is caused by an epistatic gene that blocks the production of pigment.

Cause and Effect

FIGURE 15: Several different genes interact to produce the range of human eye colors.

Genes for Eye Color

Another example of epistasis occurs in human eye color. Two genes thought to be responsible for eye color are called *OCA2* and *HERC2*, both located on chromosome 15. The *OCA2* gene codes for a protein involved in storing pigment in the iris. This protein helps cells store melanin, the pigment that affects eye coloration. More of the protein leads to darker eyes, which may appear brown. Less of the protein leads to lighter eyes, which may appear blue. The expression of the *OCA2* gene, however, can be turned on or off by a mutation in another gene. This gene, called *HERC2*, can reduce the expression of *OCA2*, leading to less melanin being stored in the iris and resulting in blue eyes. Several other genes are known to contribute to eye color, including those that lead to green eyes.

Analyze Draw a simple diagram to model the scenario described in each question. Use your diagram as evidence for your explanations.

1. A child inherits a functional copy of the *OCA2* gene from his mother but a mutated version of this gene from his father. Predict his eye color. Explain your answer.
2. Another child inherits two functional copies of the *OCA2* gene but also inherits two copies of the *HERC2* gene that suppresses the expression of the *OCA2* genes. What would you predict about the color of this child's eyes? Explain your answer.

Genes and the Environment

The environment also interacts with genes and affects their expression. Environmental influences, such as temperature, diet, light, and even pH, all play a role in the expression of countless traits in plants and animals. For example, the sex of sea turtles depends both on genes and on their environment. Female turtles make nests on beaches and bury their eggs in the sand. Eggs that mature in warmer temperatures develop into female turtles. Eggs that mature in cooler temperatures develop into male turtles.

Genes and the environment also interact to determine certain human traits. For example, a person's height is determined by genes, but environmental factors, such as lifestyle and nutrition also affect height. Studies of identical twins have shown that the environment during early development can have long-lasting effects. One twin might get more nutrients than the other because of its position in the mother's uterus. This difference can result in height and size differences that last throughout the twins' lives. Also, twins raised in environments with different diets and health care often differ in height as well as other physical traits.

Gather Evidence How might patterns of inheritance influence taste preferences? What environmental factors might affect this trait?

Careers in Science

Genomics: Studying Genomes

Genomics is a branch of biology that analyzes the DNA sequence of specific organisms and compares it to other organisms with the hope of gaining information about a gene's particular function. Scientists in this field might study the DNA code of an organism, the length of genes, and numbers of genes, or the locations of genes on chromosomes. They are particularly interested in any similarities and differences in the genome of various organisms.

A career in genomics requires a strong background in molecular biology but also a solid foundation in math and statistics. Genomicists often use computers to aid in the analysis and presentation of vast amounts of data. This use of computer databases to organize and analyze biological data is called bioinformatics. A sharp eye for detail and an underlying curiosity about the world are also essential characteristics in this and other fields of science.

One area of genomics called gene mapping got its start with the mapping of a simple virus in 1977. To date, scientists have mapped the genome of hundreds of animals, including mice, frogs, and chimpanzees. Our own genome was sequenced as part of the Human Genome Project completed in 2003.

Plants also have been studied using gene sequencing. Watermelons, sugar beets, rice, and wheat have all had their genomes mapped. Scientists today often use techniques called next-generation sequencing, which are higher-yielding methods than previous techniques, resulting in millions of copies of DNA in a short period. A small flowering plant called *Arabidopsis*, a type of mustard plant, was the first plant to have its genome sequenced in 2000. *Arabidopsis* is still used today as a model organism for research into the processes of all flowering plants. Genomicists and plant biologists are working together to research variant alleles of *Arabidopsis* to improve understanding of other plants, including those used for food. Because the DNA sequence of *Arabidopsis* is already known, scientists can use this information and compare it to other plants. Research on rice and corn genomes is aimed at producing crop varieties that produce higher yields, are less susceptible to disease, or can grow in drought conditions.

FIGURE 16: The field of genomics attempts to understand our genetic code better in order to find out how genes affect our traits, our health, and even our future.

The study of animal genomes gives researchers in many fields of research incredibly valuable information about how our own genes might function and what happens when they do not function properly. Plant genome sequencing provides scientists with information on how to grow crops that are more productive. The insights gained from the field of genomics will undoubtedly have far-reaching effects on industries, such as pharmaceutical research, health care, and agriculture.

Language Arts Connection

Write a brief report answering these questions.

- Do you think you would enjoy a career as a genomicist? Why or why not?
- Which organism would you like to study the DNA of, and why?
- Why do you think studying the genome of other animals might provide valuable information?
- Why might scientists be interested in the genomes of plants?
- In what ways do you think the field of genomics has improved our lives?
- How might changes in technology change the way we study the genomes of organisms?

DISCUSSION: SEQUENCING YOUR OWN GENOME

EVALUATING CLAIMS: EYE COLOR AND OUR ANCESTORS

Go online to choose one of these other paths.

Lesson Self-Check

CAN YOU EXPLAIN IT?

FIGURE 17: Perhaps if you were given different food choices as a young child, you would make different food choices today.

You have explored throughout this lesson about inherited traits. Both genes and the environment play a role in shaping who we are. Eating a healthy diet is an important part of growing up and maintaining good health as we age. Nonetheless, some foods are just not that appealing to some people. Researchers are looking at how our food preferences develop—is it genetic, or is it our environment? This question is an example of the long-standing "nature vs. nurture" debate. Doctors and scientists alike have always maintained that parents should provide a variety of healthy foods to children so that they are familiar with these tastes from a young age. But do we have genes that predispose us to like or not like—as in the case of broccoli—certain foods? Several studies have linked a specific gene to a taste receptor that perceives broccoli and similar vegetables as bitter, whereas people without this gene do not detect the bitterness. This makes sense from a biological standpoint, as taste reception is a biochemical process. However, other studies have suggested that a "food window" of sorts exists when children are as young as four months old. During this sensitive time period, exposure to different foods may influence the child's food choices later in life. Some feel that this critical period is when children should be exposed to as many different foods and flavors as possible.

Explain When it comes to something like food preferences, how do genetics and the environment influence traits? In your answer, discuss the following:

- How are traits generally passed from parents to offspring?
- How can dominant-recessive relationships influence which trait is expressed in the phenotype of the organism?
- How is the expression of genes influenced by other genes and the environment?

CHECKPOINTS

Check Your Understanding

1. Which of the following statements best describes how genes relate to traits?

a. Genes code for the production of specific proteins. These proteins lead to different traits.

b. Genes and traits function together to produce proteins.

c. Traits contain instructions for making proteins, and genes are the observable outcome of such proteins.

d. Genes are expressed according to instructions in traits.

2. Why did Mendel remove the stamens of some pea plants during his first experiments? Select all correct answers.

a. to prevent reproduction from occurring

b. to control which parent plants were allowed to reproduce

c. to prevent self-fertilization of the pea plants

d. to allow the pea plants to reproduce asexually

3. Mendel's F_1 generation of pea plants were heterozygous. What does this mean?

a. All of the offspring plants would have the recessive trait.

b. Half of the offspring plants would have the dominant trait, while the other half would have the recessive trait.

c. The offspring had two identical alleles for the same gene.

d. The plants had two different alleles for the same trait.

4. Which of the following statements best describes why a recessive trait is not observed in the offspring of a cross between a homozygous-dominant and a homozygous-recessive parent?

a. The offspring will be heterozygous, and the dominant allele masks the appearance of the recessive allele.

b. Recessive alleles are blended with dominant alleles to make an intermediate trait.

c. The offspring will likely be homozygous dominant for this trait and therefore show the dominant trait.

d. The dominant alleles will destroy the recessive alleles.

5. Which of the following questions can be answered by Mendel's Law of Segregation?

a. Why do the offspring of a plant that is homozygous tall (*TT*) and homozygous short (*tt*) all appear tall?

b. In what way do traits pass from one generation to the next?

c. How can a plant that is heterozygous for height (*Tt*) have both tall and short offspring?

d. Why don't tall pea plants also all have purple flowers?

6. Use the following words to complete this statement: *phenotype*, *genotype*, *heterozygous*, *homozygous*, *traits*, *alleles*, *genes*

Chromosomes contain _____, which help to determine an organism's _____. Genes come in alternate forms called _____, and both parents may not have the same type of allele. The actual gene combination that an organism receives from its parents is called its _____, while the trait that gets expressed as a result is referred to as its _____. If an offspring receives the same type of allele for a given gene from each parent, it is said to be _____ for that trait. If the alleles differ, it is _____.

7. Explain why a recessive allele can only be expressed when the organism is homozygous.

MAKE YOUR OWN STUDY GUIDE

In your Evidence Notebook, design a study guide that supports the main idea from this lesson:

Both genes and the environment influence the expression of traits passed from parents to offspring.

Remember to include the following information in your study guide:

- Use examples that model main ideas.
- Record explanations for the phenomena you investigated.
- Use evidence to support your explanations. Your support can include drawings, data, graphs, laboratory conclusions, and other evidence recorded throughout the lesson.

Consider how genes function to produce traits, how different genes interact, and how the environment influences genes.