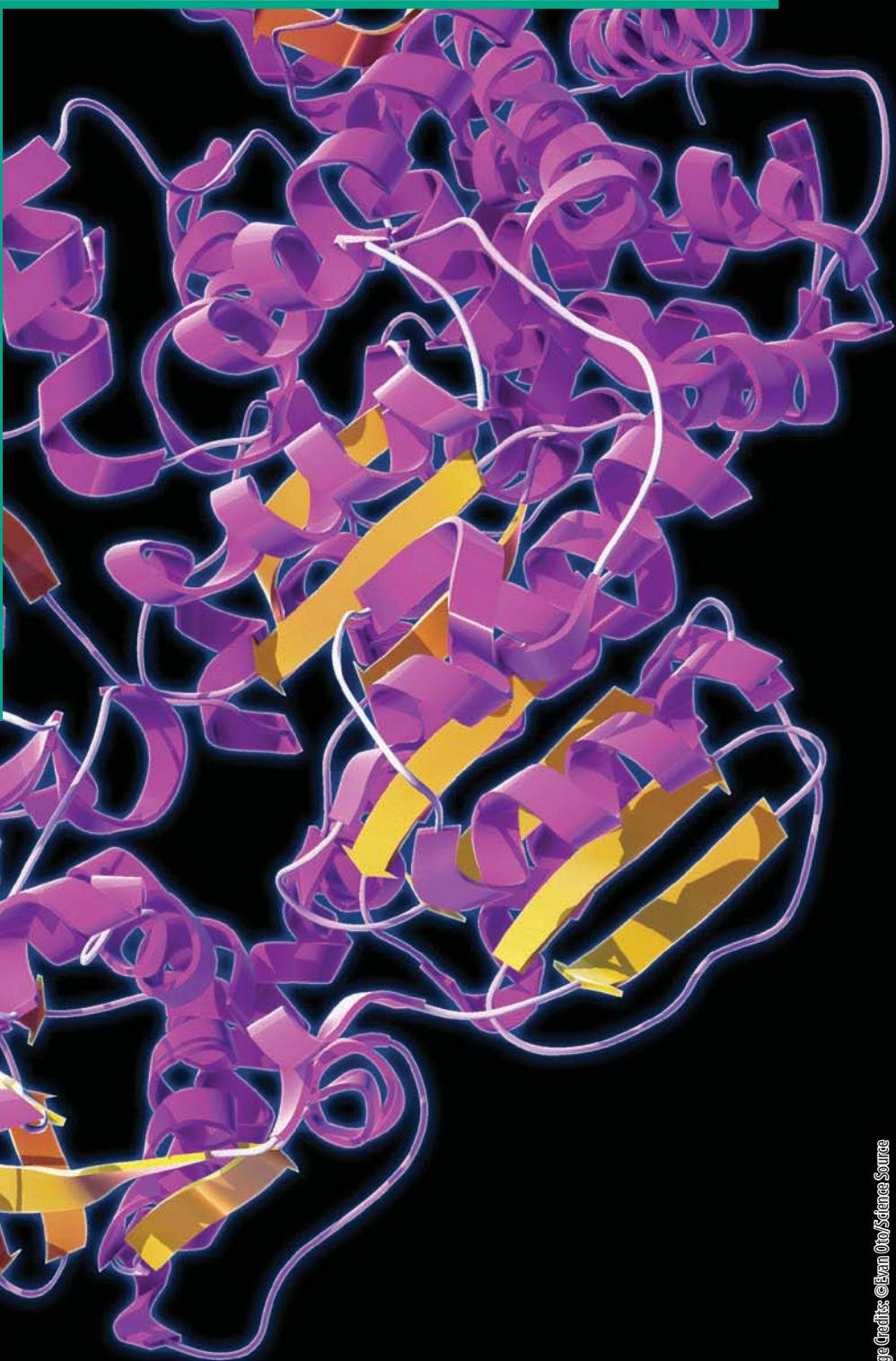


UNIT 7

Genetics and Heredity

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Proteins like Cas9 can be used to edit DNA.

FIGURE 1: Children inherit traits from each of their parents.



In families, children often share similar physical features, such as hair color, skin color, and facial shape, with one or both of their parents. These traits and others are passed from parents to offspring, or inherited. Because of these shared features, it is often possible to tell that two people are closely related, whether siblings, parent and child, or even grandparent and grandchild. However, except in the case of identical twins, even closely related individuals have unique features. Some children may share distinctive features of their parents—a father's chin or a mother's eyes—but not all children inherit the same sets of features.



Predict Why do you think siblings don't look exactly the same if all of their DNA comes from the same mother and father?

DRIVING QUESTIONS

As you move through the unit, gather evidence to help you answer the following questions. In your Evidence Notebook, record what you already know about these topics and any questions you have about them.

1. How are traits passed from parents to offspring?
2. Why do the offspring of two parents all look different from each other?
3. How does diversity in traits arise over generations?
4. How can we determine the probability that an organism's expressed version of a trait will be passed on to its offspring?
5. Can scientists alter the genetic material of other organisms? How might humans use this ability?

UNIT PROJECT

Investigating the Heredity of Disease

Many traits and conditions can arise from either genetic or environmental causes. Explore the heredity of Huntington's disease. Based on your analysis, determine the likely cause or causes of Huntington's disease, and the role that genetic or environmental factors may play.



Go online to download the Unit Project Worksheet to help you plan your project.

Meiosis

Sperm and egg cells are produced during the process of meiosis.

CAN YOU EXPLAIN IT?

FIGURE 1: Could it be possible that everyone has a twin?



Image credits: (t) ©Dr. Yorgos Nikas/Science Source; (b) ©Design Pics Inc./Alamy



Gather Evidence

As you explore the lesson, gather evidence to explain how meiosis and sexual reproduction increase genetic diversity.

Humans have unique versions of traits that cause us to look and act differently from one another. Aside from identical twins, there is great variety in physical traits from one person to the next. However, some people believe there may be an exact copy of themselves somewhere in the world. For example, some have claimed to have found their “twin” on the Internet. Have you ever wondered if there could be a copy of you somewhere else in the world? Do you think it is possible for someone to be born from a different mother and father, yet have the same genetic makeup as you?

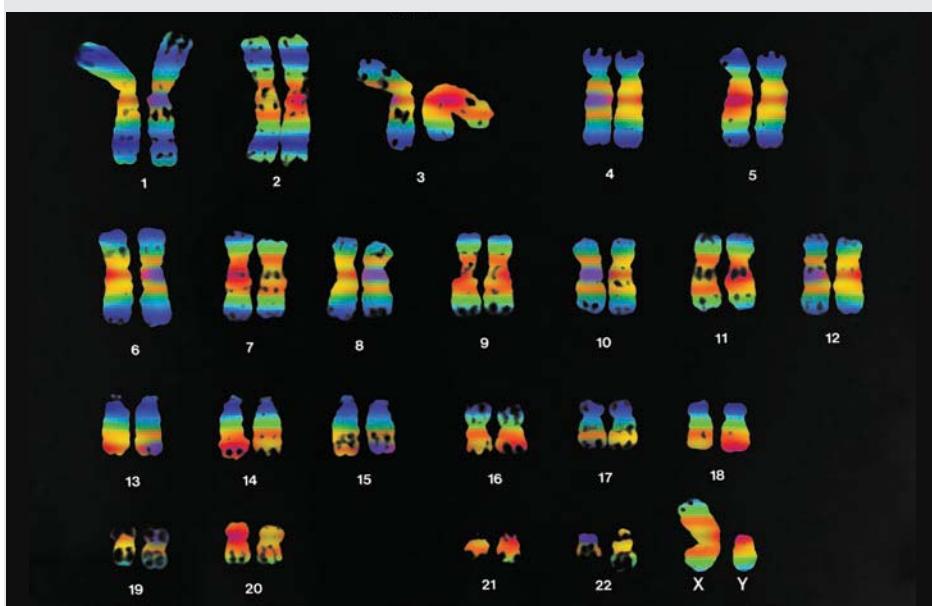


Predict What do you think the chances are that there is someone in the world exactly like you?

Chromosomes and Meiosis

DNA is the genetic material in organisms. DNA codes for proteins and contains the information that determines when proteins are made. In complex organisms, long strands of DNA are packaged together with proteins into chromosomes in the nucleus of the cell. Images like the one in Figure 2 can be analyzed to determine a karyotype, which shows the 23 pairs of chromosomes in a human cell. The brightly colored structures are pairs of highly condensed chromosomes formed during metaphase of mitosis.

FIGURE 2: Chromosomes in a Human Cell



Collaborate Write your answers to the following questions about the image in Figure 2. Compare your answers with a partner, and write down any new information that you had not previously recorded.

1. What patterns do you observe?
2. How many chromosomes do human body cells have?
3. What differences do you see among the different pairs of chromosomes and the chromosomes within a pair?

Chromosome Structure and Function

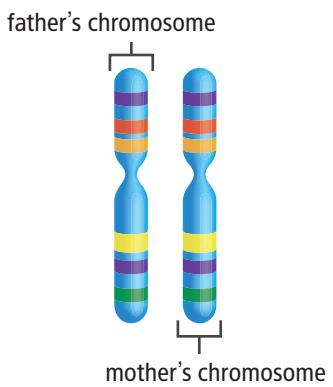
Each pair of chromosomes in your cells is referred to as a homologous pair.

Homologous chromosomes are two chromosomes—one inherited from the mother, one from the father—that have the same length and general appearance. These chromosomes have copies of the same genes, although the two copies may differ.



Analyze What percentage of your genetic material is from your mother and what percentage is from your father?

FIGURE 3: Homologous chromosome pairs include one chromosome inherited from the father and another inherited from the mother.



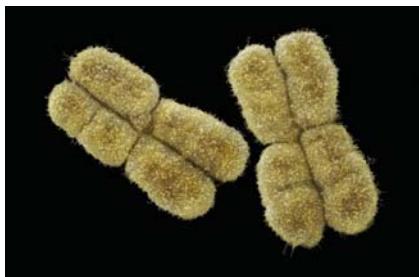
Autosomes and Sex Chromosomes

You may have noticed that all of the chromosome sets in Figure 2 are labeled with a number except for one set, which is labeled with an X and a Y. Together, chromosome pairs 1 through 22 make up your **autosomes**, which are chromosomes that contain genes for characteristics not directly related to the sex of an organism.

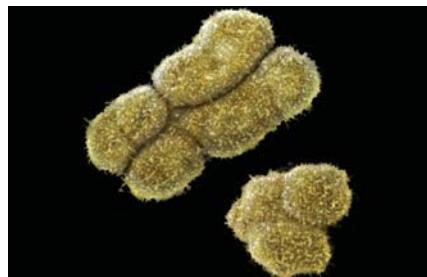
Analyze Do the chromosomes in Figure 2 on the previous page belong to a female or a male? Explain how you know.

Most sexually reproducing species also have sex chromosomes that directly control the development of sexual characteristics. Humans have two very different **sex chromosomes**: X and Y. In most mammals, including humans, an organism's sex is primarily determined by the XY system. An organism with two X chromosomes, or XX, is female. An organism with one X and one Y chromosome, or XY, is male.

FIGURE 4: Sex chromosomes control the development of sexual characteristics.



a Females have two X chromosomes.

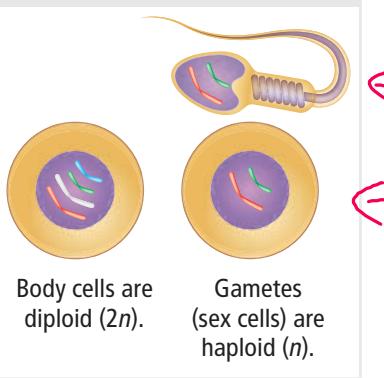


b Males have an X chromosome and a Y chromosome.

Body Cells and Germ Cells

The 23 pairs of chromosomes you analyzed earlier are from a body, or somatic, cell. Your body cells are called diploid cells because they contain two copies of every chromosome. However, the two copies are not exact copies because one is from your mother and one is from your father. Diploid cells can be represented as $2n$. In humans, the diploid chromosome number is 46.

FIGURE 5: Body Cells and Gametes (Cells are not to scale.)



In addition to body cells, you also have germ cells located in your reproductive organs. Germ cells form **gametes**, or sex cells. The male sex cells are sperm and the female sex cells are eggs. Unlike body cells, gametes have only one copy of each chromosome. These cells are called haploid and can be represented as n . Human gametes thus contain 23 chromosomes. Only DNA in gametes is passed down to the organism's offspring. The DNA in body cells is not.

Sexual reproduction involves the fusion of two gametes of different types, resulting in offspring that are a genetic mixture of both parents. The joining of these two gametes is called fertilization. When fertilization occurs, the nuclei of the egg and sperm fuse to form a single nucleus.



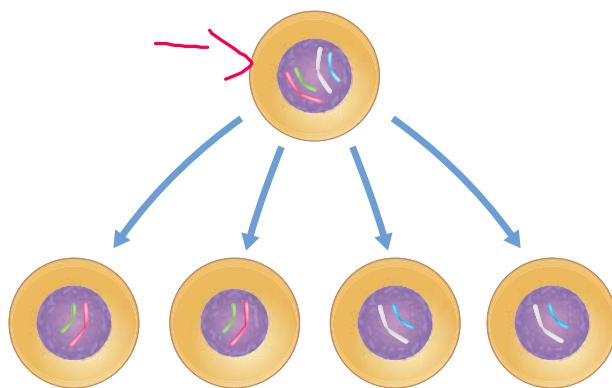
Explain Answer the following questions about body cells and gametes.

1. What is an example of a body cell in your body?
2. Why do gametes have half a set of DNA? What would happen if they had a full set of DNA? Explain your answer.

The Process of Meiosis

Recall that body cells reproduce during part of the cell cycle. During mitosis and cytokinesis, the nucleus and cytoplasm divide, resulting in daughter cells that are genetically identical to the parent cell. Germ cells in your reproductive organs undergo the process of meiosis to form gametes. **Meiosis is a form of nuclear division that divides one diploid cell into four haploid cells.** There are two rounds of cell division—meiosis I and meiosis II. This process divides the DNA and reduces each resulting cell's chromosome number by half.

FIGURE 6: Meiosis has many stages and produces four haploid cells from one diploid cell.



diploid = two copies
haploid = one copy



Predict Meiosis divides one cell into four cells, but each resulting cell has half the amount of DNA as compared to the original cell. How do you think this is possible?

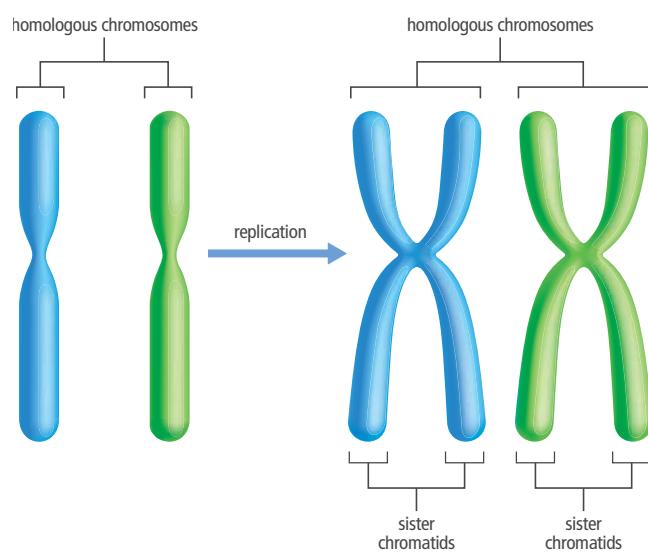
Chromosomes and Replication

To understand meiosis, it is necessary to distinguish between homologous chromosomes and sister chromatids. As Figure 7 shows, homologous chromosomes are two separate chromosomes, one from your mother and one from your father. Homologous chromosomes are similar to each other because they are the same length and carry the same genes. However, they are not exact copies of each other. In contrast, a **chromatid** is one half of a duplicated chromosome. Sister chromatids refers to the duplicated chromosomes that remain attached (by the centromere). Homologous chromosomes divide during meiosis I, and sister chromatids are split and separated into new gametes during meiosis II.



Analyze What is the difference between the genetic material on two sister chromatids and the genetic material on homologous chromosomes?

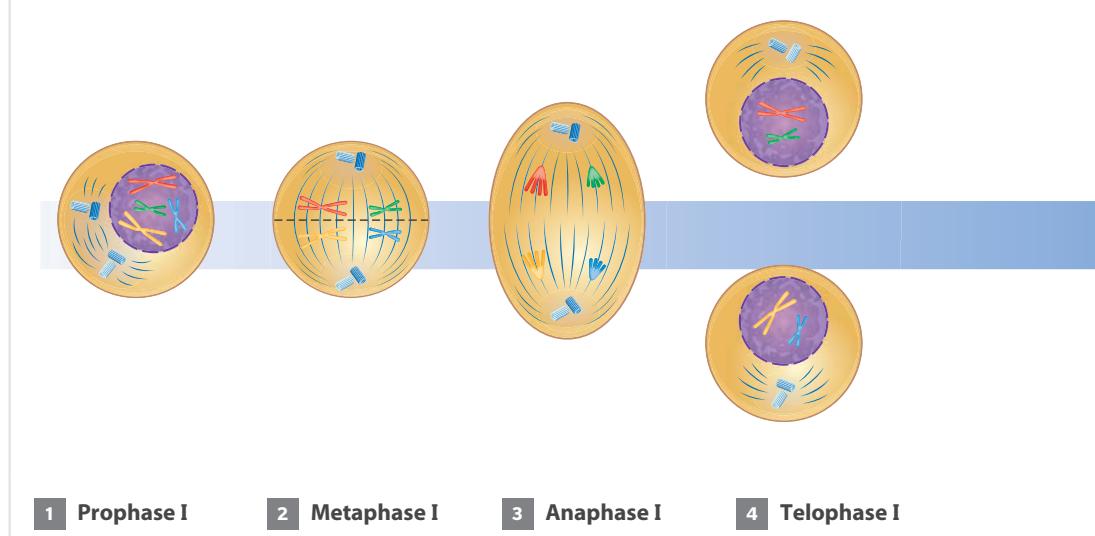
FIGURE 7: Homologous chromosomes are two separate chromosomes, while sister chromatids are duplicated chromosomes that remain attached to one another.



Meiosis I

Before meiosis begins, DNA is copied during S phase. Meiosis I separates homologous chromosomes, producing two haploid cells with duplicated chromosomes. Meiosis I can be described in distinct phases, each of which is a series of gradual changes.

FIGURE 8: Meiosis I and meiosis II are each made up of four phases.



1 Prophase I

2 Metaphase I

3 Anaphase I

4 Telophase I

Explore Online

Hands-On Lab



Modeling Meiosis

Make a model to illustrate how the arrangement and separation of chromosomes during meiosis causes an increase in genetic diversity.

1. Prophase I During this first phase of meiosis, the nuclear membrane breaks down, the centrosomes and centrioles move to opposite sides of the cell, and spindle fibers start to assemble. The duplicated chromosomes condense, and homologous chromosomes pair up. They appear to pair up precisely, gene for gene, down their entire length. The sex chromosomes also pair with each other, and some regions of their DNA appear to line up as well.

2. Metaphase I The homologous chromosome pairs randomly line up along the middle of the cell, or the cell equator, attached to spindle fibers. The result is that 23 chromosomes—some from the father, some from the mother—are lined up along each side of the cell equator. This arrangement mixes up the chromosomal combinations and helps make and maintain genetic diversity.

3. Anaphase I Next, the paired homologous chromosomes separate from each other and move toward opposite sides of the cell. The sister chromatids remain together during this step and throughout meiosis I.

4. Telophase I The cell undergoes cytokinesis.

After telophase I, the nuclear membrane forms again in some species, and the spindle fibers disassemble. These steps occur during a period between meiosis I and meiosis II.

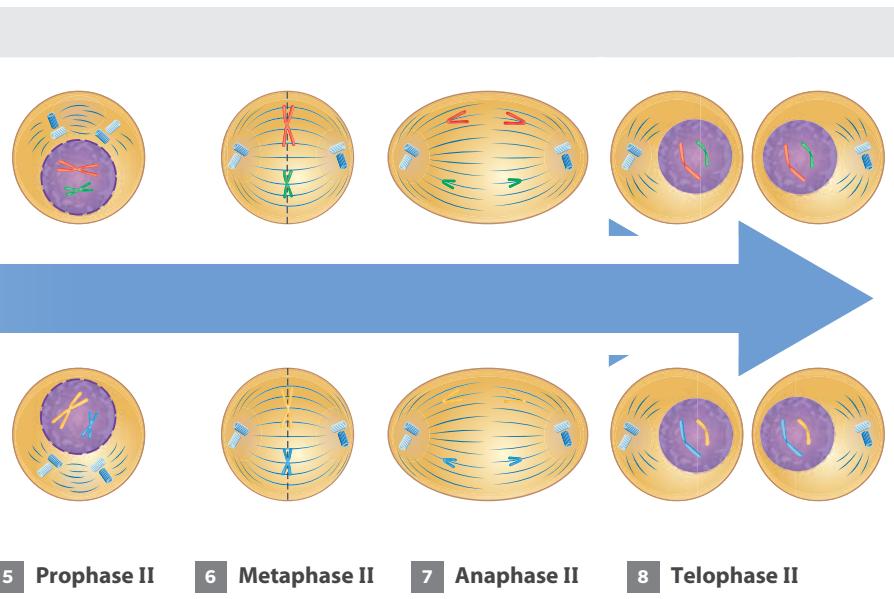


Analyze Observe the model of meiosis I in Figure 8. Use this model to answer the following questions:

1. What are the products of meiosis I? Explain in terms of number of cells and the genetic material contained in those cells.
2. Describe the arrangement of chromosomes in metaphase I. Why do you think chromosomes are arranged in this way?
3. What are some of the strengths and limitations of this model?

Meiosis II

Meiosis II separates sister chromatids, which results in chromosomes that are not doubled. The diagram of this process applies to both of the cells produced in meiosis I. It's important to note that DNA is not copied between meiosis I and meiosis II.



5. Prophase II The nuclear membrane breaks down, centrosomes and centrioles move to opposite sides of the cell, and spindle fibers assemble.

6. Metaphase II Spindle fibers align the 23 chromosomes at the cell equator. Each chromosome still has two sister chromatids at this stage.

7. Anaphase II Next, the sister chromatids are pulled apart from each other and move to opposite sides of the cell.

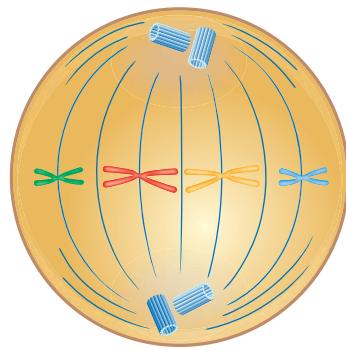
8. Telophase II Finally, nuclear membranes form around each set of chromosomes at opposite ends of the cell, the spindle fibers break apart, and the cell undergoes cytokinesis.

Explain According to this model, do all the gametes produced by an organism have the same genetic material? Use evidence to support your claim.



Cause and Effect

FIGURE 9: Metaphase in Mitosis



Comparing Chromosome Arrangement

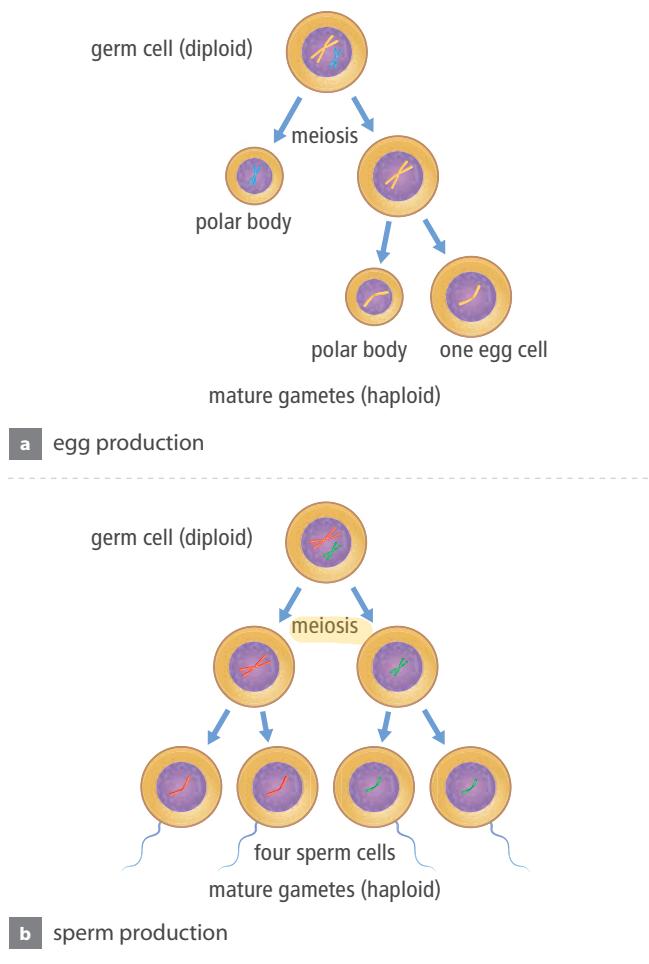
Mitosis, which occurs in body cells, produces two genetically identical cells. Like meiosis, mitosis includes metaphase. However, the alignment of chromosomes differs, which affects the genetic makeup of the final cells.



Explain Answer these questions about metaphase in meiosis and mitosis.

- How do the arrangements of chromosomes in metaphase I and metaphase II of meiosis compare to each other and to the metaphase stage of mitosis?
- What are the final products of mitosis and meiosis? How does the arrangement of chromosomes during metaphase affect the genetic makeup of the final products?

FIGURE 10: Gametogenesis (Cells are not to scale.)



Gametogenesis

The haploid cells produced by meiosis are not able to be fertilized until they go through additional changes to produce mature gametes. The final stages of this process, called gametogenesis, differ between the sexes. The formation of an egg, the female gamete, begins before birth, inside the developing body of a female embryo, and is not finished until a sperm fertilizes that egg many years later. Only one of the four cells produced by meiosis actually makes an egg. The other cells produced are called polar bodies and are not typically able to be fertilized. Nearly all of a zygote's cytoplasm and organelles come from the egg. Since mitochondria carry their own DNA, the mitochondrial DNA in the embryo is identical to the mother's.

The sperm cell, the male gamete, is much smaller than the egg. The sperm cell's main contribution to an embryo is DNA. Yet it must swim to an egg to fertilize it, so the ability to move is critical. Sperm formation starts with a round cell and ends by making a streamlined cell that can move rapidly. During this process, significant changes occur. DNA is tightly packed and much of the cytoplasm is lost, resulting in a compact head. The sperm cell develops a whip-like flagellum and a neck region with mitochondria that provide the energy needed to drive the cell's flagellum. Other changes, such as the addition of proteins to the cell membrane, also take place.



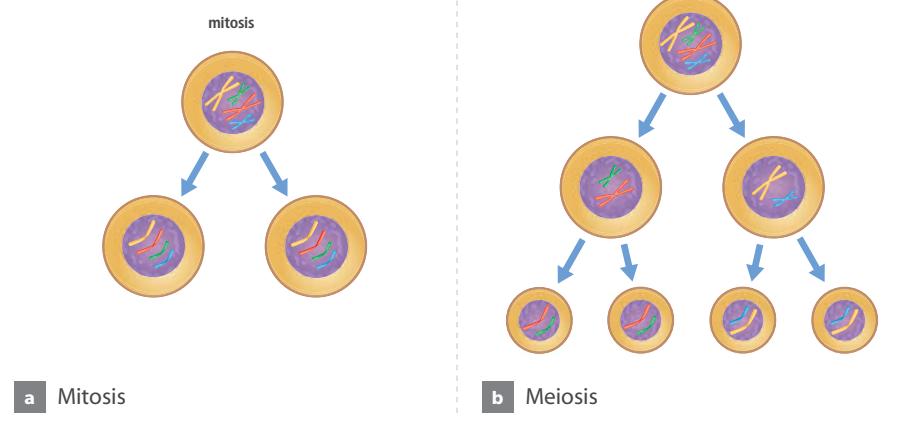
Analyze Make a Venn diagram to compare and contrast egg production and sperm production.

Comparing Mitosis and Meiosis

Mitosis is a process that occurs in body cells. It is essential for the growth and development of an organism. In contrast, meiosis occurs in germ cells.

FIGURE 11: Mitosis and meiosis produce different kinds of cells.

E **Explain** Make a table to compare mitosis and meiosis in terms of the number and type of cells produced, the genetic material in the cells, and the role of each process in the body. Does meiosis or mitosis occur more frequently in your body? Explain your answer.



Meiosis and Genetic Variation

One of the major outcomes of meiosis and sexual reproduction is the resulting increased genetic diversity within a species. **Genetic variation** refers to differences in the genetic material of individuals in a population.

FIGURE 12: Genetic variation is responsible for the different versions of traits you see in this cat's offspring.

Mechanisms of Genetic Variation

Meiosis and sexual reproduction increase genetic diversity, or genetic variation, within a population. Gametes have different combinations of genes than their parent cells due to crossing over and independent assortment, which both occur during meiosis.

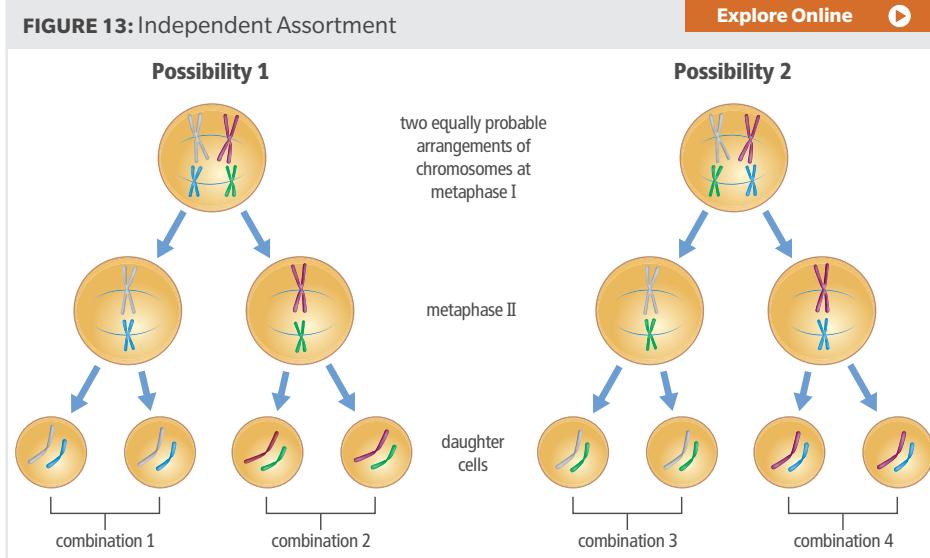
Independent Assortment

When homologous chromosomes pair up in metaphase I of meiosis, the chromosomes from your father and the chromosomes from your mother line up randomly on either side of the cell's equator. This assortment of chromosomes is a matter of chance. The arrangement of any one homologous pair does not depend on the arrangement of any other homologous pair. Therefore, it is referred to as **independent assortment**.



FIGURE 13: Independent Assortment

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Problem Solving

To determine the number of possible chromosome combinations that can result from independent assortment, you can use this formula:

$$\text{Combinations} = 2^n$$

where n = number of different chromosomes.

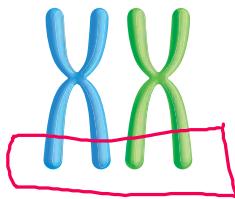
- What is the number of possible chromosome combinations for a human cell with 23 different chromosomes?
- How does your answer to Question 1 support the claim that independent assortment increases variation in an organism's offspring?

Crossing Over

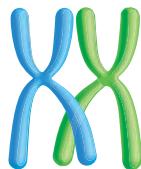
Crossing over is the exchange of chromosome segments between homologous chromosomes. It occurs during prophase I in meiosis I, and it is a regulated process. At this stage of meiosis, each chromosome has been duplicated, the sister chromatids are still connected, and homologous chromosomes have paired up. Some of the chromatids are very close to each other. Part of one chromatid from a chromosome may break off and reattach to the other chromosome. Because crossing over results in new combinations of genes, it is an example of genetic recombination.

FIGURE 14: Crossing Over

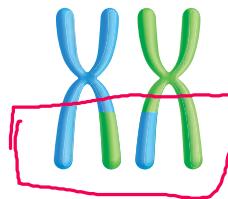
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- 1 Two homologous chromosomes pair up with each other during prophase I in meiosis.



- 2 In this position, some chromatids are very close to each other and segments cross.



- 3 Some of these segments break off and reattach to the other homologous chromosome.

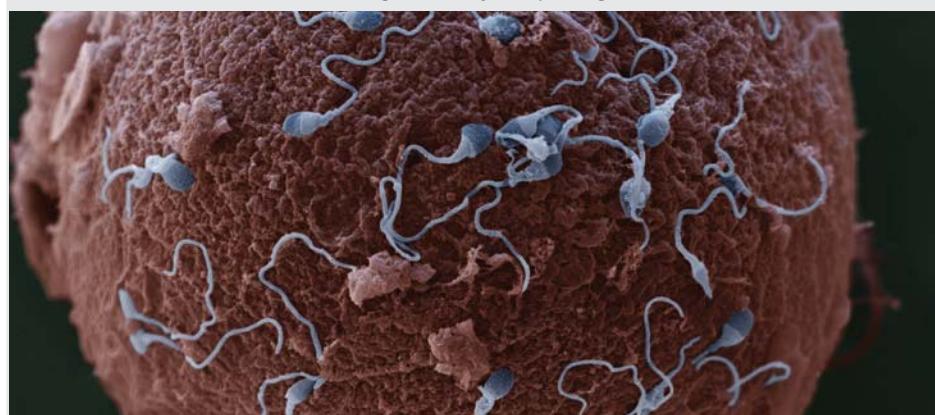


Explain How does crossing over contribute to genetic diversity?

Fertilization

Once mature gametes have formed during the process of gametogenesis, the gametes are ready for fertilization. In fertilization, two gametes of different types fuse, producing a zygote with a complete set of DNA—half from one parent and half from the other. The zygote formed will have a unique combination of genes. The mixing and matching of genetic material during meiosis and fertilization is responsible for the genetic variation in sexually reproducing organisms.

FIGURE 15: Fertilization results in a genetically unique organism.

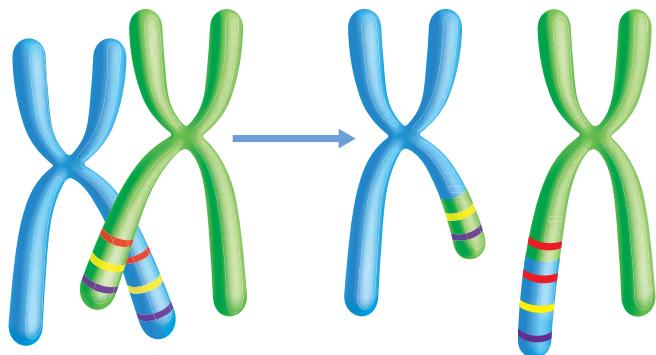


Explain Use what you have learned about meiosis and sexual reproduction to construct an explanation for why offspring are not exact replicas of their parents. In your answer, include a discussion of sexual reproduction, independent assortment, and crossing over.

Guided Research

Gene Duplication and Genetic Variation

FIGURE 16: Gene duplication has influenced the traits of domestic sunflower plants.



In metaphase I of meiosis, homologous chromosomes exchange DNA segments via crossing over. This leads to genetic variation in the offspring of sexually reproducing organisms. Sometimes during crossing over, homologous chromosomes do not align with each other properly. If this happens, the two segments crossing over may be different in size. As a result, one chromosome may have two copies of a gene or genes, which is called gene duplication. The other chromosome may have no copy of the gene or genes, known as a gene deletion.

Gene duplication has occurred many times over millions of years of eukaryotic evolution. For example, domesticated sunflowers have a duplicated gene that lengthens plants' growing period. Interestingly, this gene duplication is not the result of domestication. Evidence shows that the duplication occurred long before Native Americans began breeding the plants as a part of their horticultural practices. This variation of sunflower was simply preferred by Native Americans.



Language Arts Connection Conduct research to construct an argument for how gene duplication increases genetic variation. Start by choosing a specific species to research, and look for materials explaining how gene duplication has affected this species. As you conduct your research, evaluate your sources carefully to be sure they are reliable. Do they present verifiable facts? Are the opinions those of an expert or experts in this field? Is there enough evidence to support the claims being made?

Using your own words, write an argument explaining how gene duplication from unequal crossing over has influenced genetic variation in a certain species. Use these questions to guide your research:

1. Which species will you be researching, and what evidence exists that gene duplication has occurred in this species?
2. How did gene duplication influence the traits of this species?
3. What is the connection between gene duplication and the evolution of this species?

Lesson Self-Check

CAN YOU EXPLAIN IT?

FIGURE 17: How likely is it that there is a genetic copy of you somewhere?



Now that you have learned about meiosis and genetic variation, think again about the possibility of finding a genetic copy of yourself. According to some estimates, the number of possible gene combinations made by meiosis is trillions of times more than the number of people who have ever lived on Earth. Independent assortment alone makes millions of possible combinations of chromosomes. Each chromosome contains anywhere from hundreds to thousands of genes. When those genes are shuffled during meiosis, an astounding number of combinations is possible. Multiply this by the probability that the gametes that formed you would merge, and it's no wonder there is so much variation in the human race.



Explain In general, how likely is it that there is someone in the world who is genetically identical to you? Refer to the notes in your Evidence Notebook to construct an explanation for this question using a claim, evidence, and reasoning. Your explanation should include a discussion of sexual reproduction, meiosis, crossing over, and independent assortment.

1. State your claim.
2. Cite evidence to support your claim. Include models and examples as necessary.
3. Explain how the evidence you cited supports your claim. For example, consider the number of possible chromosome combinations made by independent assortment. How would this evidence support the statement you are making?

CHECKPOINTS

Check Your Understanding

1. Fruit fly gametes each have four chromosomes representing 2^4 , or 16, possible chromosome combinations. How many chromosome combinations could result from fertilization between a fruit fly egg and a sperm cell?

2. A student uses string to model four pairs of homologous chromosomes in a parent cell. Each chromosome pair is a different color. Which model would best show the genetic makeup of a daughter cell produced by meiosis?
 - a. two strings, each a combination of different colors
 - b. two strings, each the same color
 - c. four strings, each a combination of different colors
 - d. four strings, each the same color

3. Which of the following statements describe differences between mitosis and meiosis? Select all correct answers.
 - a. Mitosis produces diploid cells, and meiosis produces haploid cells.
 - b. Mitosis is involved in asexual reproduction, and meiosis is involved in sexual reproduction.
 - c. Only body cells result from mitosis, but both body cells and gametes result from meiosis.
 - d. Mitosis produces genetically unique cells, and meiosis produces genetically identical cells.
 - e. Two daughter cells are produced by mitosis, and four daughter cells are produced by meiosis.

4. Describe two pieces of evidence to support the claim that sexual reproduction increases genetic variation.

5. Identify the process shown in Figure 18. Then explain how the figure provides evidence to support the claim that meiosis increases genetic variation.

6. Make a table categorizing each of the items in the list as a description of diploid or haploid cells.

- contain single chromosomes, each from one parent
- are described as $2n$
- make fertilization possible
- result from meiosis
- contain chromosomes in pairs, one from each parent
- are described as n
- result from mitosis

7. Why is it important that human gametes have half a set of DNA instead of a full set of DNA? Use scientific reasoning to support your claim.

MAKE YOUR OWN STUDY GUIDE



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

Inheritable genetic variations result from new genetic combinations made through meiosis and sexual reproduction.

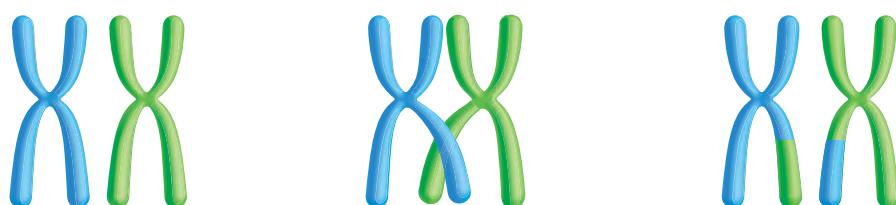
Independent assortment and crossing over are processes that contribute to genetic variation within a species.

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 the models and explanations in this lesson can be used to support a claim for how meiosis and sexual reproduction increase genetic variation.

FIGURE 18: This process occurs during meiosis.



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.

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.

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.

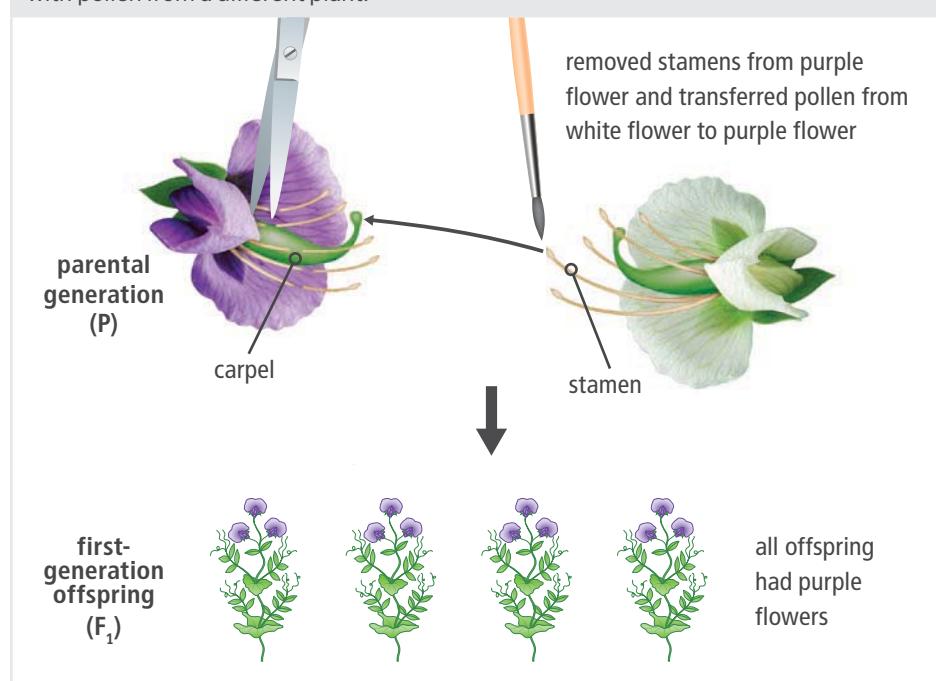


Image Credits: (t) © Andrey Kuzmin/Fotolia

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.

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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.

 **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?

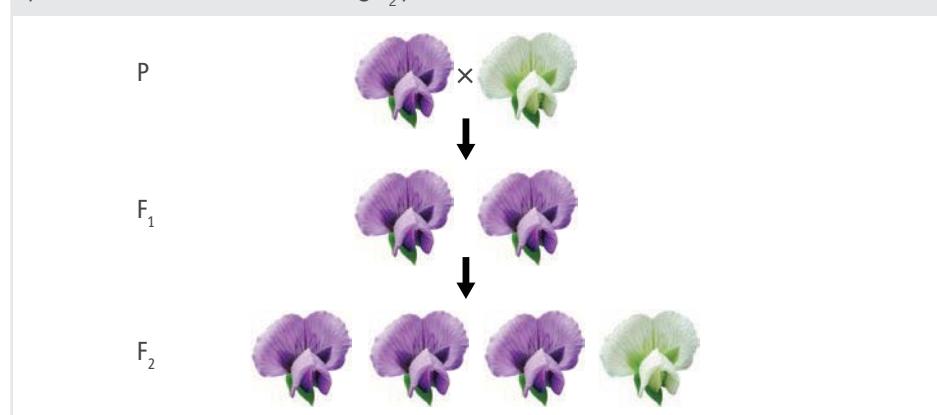
FIGURE 4: Mendel worked with seven traits in pea plants for his experiments.

Flower color	Flower position	Seed color	Seed shape	Pod shape	Pod color	Stem length
						
purple	axial	yellow	round	inflated	green	tall
						
white	terminal	green	wrinkled	constricted	yellow	dwarf

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 F₁—generation. In the second experiment, Mendel let the F₁ generation self-fertilize, meaning he did not control their pollination himself. Recall that both F₁ plants had purple flowers. The offspring from these crosses, referred to as the F₂ generation, showed a different set of traits.

FIGURE 5: Purebred white and purple plants were crossed to make the F₁ generation. F₁ plants then self-fertilized, making F₂ plants.



Collaborate

Discuss these questions with a partner.

1. What pattern occurred when the P generation was crossed?
2. What patterns occurred when the F₁ generation was crossed?
3. What questions do you think Mendel would have asked after seeing these results?

Mendel performed similar crosses with F₁ 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 F₂ 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 F₁ 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

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.



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?



Analyze

Answer the following questions about Mendel's data.

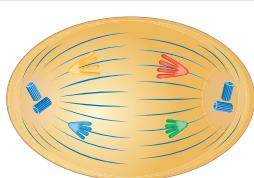
1. What patterns do you notice in the data?
2. What questions might Mendel have asked after seeing this data?



Gather Evidence

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

FIGURE 7: Anaphase I



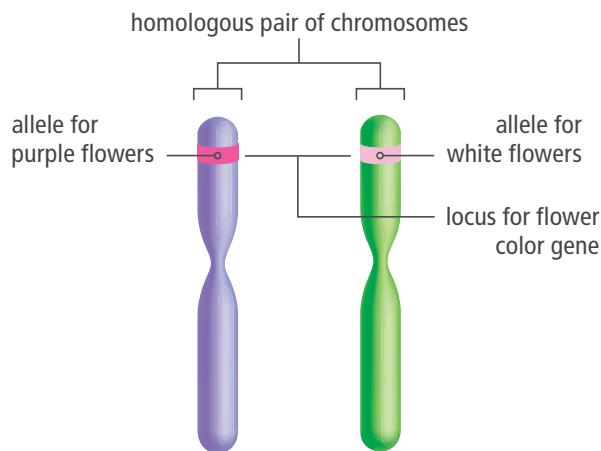
Traits, Genes, and Alleles

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.

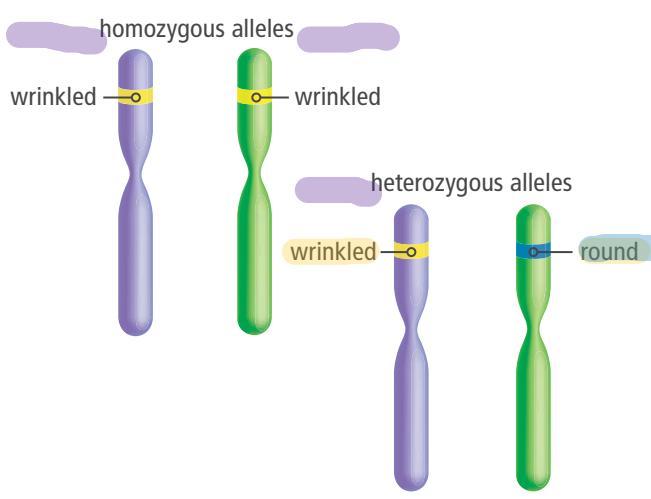
FIGURE 8: Alleles are different forms of a gene. They are located at the same position on homologous chromosomes.



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

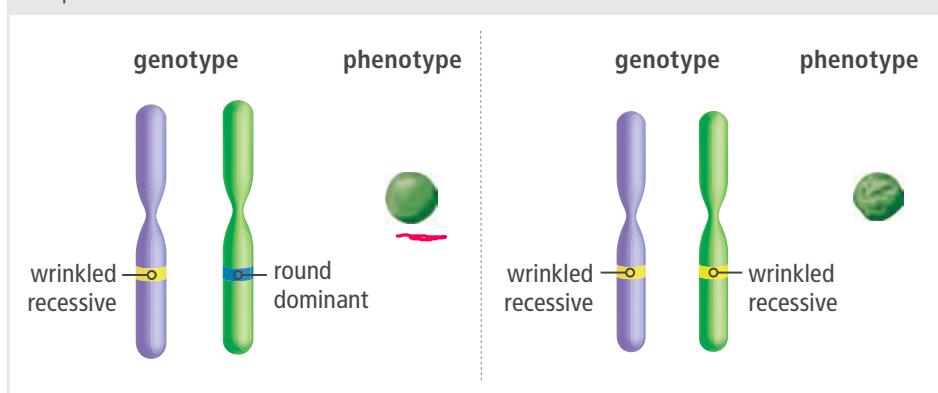
Your body cells contain two alleles for each gene. These alleles may be the same, or they may be different. The term **homozygous** describes two of the same alleles at a specific locus. The term **heterozygous** describes two different alleles at the same locus. For example, you may inherit an allele 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.



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.



Gather Evidence

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



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.

- When Mendel crossed two purple-flowered plants from the *F₁* 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.
- 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

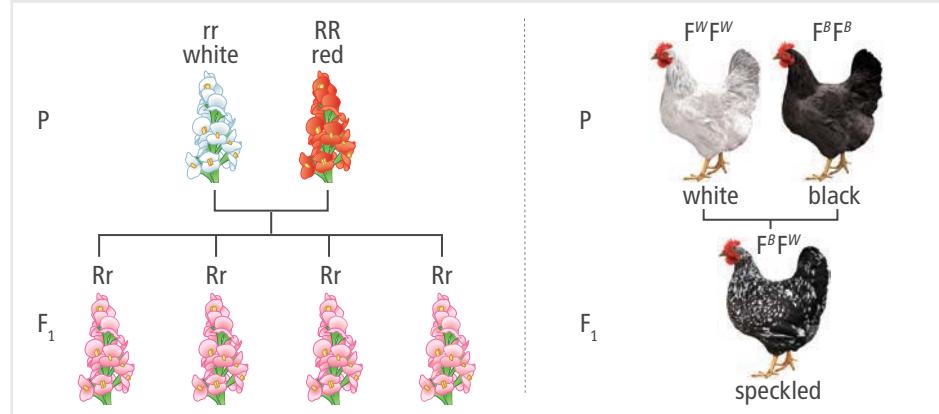
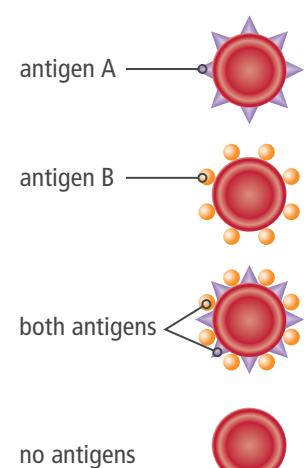


FIGURE 12: Human blood type is controlled by multiple alleles, two of which are codominant.



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 $I^A i$ will have type A blood, and someone with a genotype of $I^B i$ will have type B blood. I^A and I^B alleles are also codominant. That means someone with a genotype of $I^A I^B$ will have type AB blood. People with an ii genotype have red blood cells without an antigen, and they have type O blood.

Sex-Linked Traits

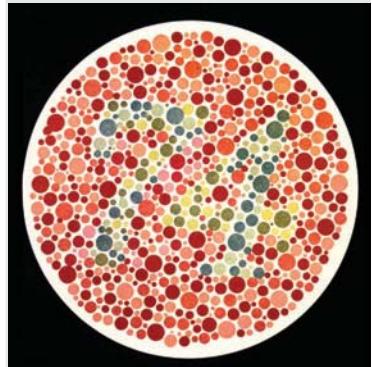
Recall that humans have 23 pairs of chromosomes and that the last pair is referred to as sex chromosomes. These chromosomes—X and Y—contain different genes, which make a unique pattern of inheritance. Many of the genes seen on the X chromosome do not have corresponding genes on the Y chromosome, simply 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.

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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 F₁ 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.

Traits and Probability

The color of corn, *Zea mays*, is an inherited trait.

CAN YOU SOLVE IT?

FIGURE 1: A cat breeder crossed an orange female cat with a black male cat.



a Female cat



b Male cat



c Tortoiseshell kitten from the litter



Gather Evidence

What can you determine from the fact that only male kittens inherited the mother's phenotype?

Animal breeders select animals to cross based on desired characteristics. Imagine a cat breeder who wants a litter of kittens—half with solid orange fur and half with solid black fur. The breeder decides to cross a female orange cat with a male black cat. The resulting litter has three orange male kittens and three tortoiseshell female kittens. Tortoiseshell is a mixture of orange and black fur. The breeder successfully bred solid orange kittens, but there were no black kittens in the litter.



Predict Answer the following questions in your Evidence Notebook:

1. Why was the litter of kittens not half black and half orange?
2. Why were there only female tortoiseshell kittens?

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Predicting Generations

Gregor Mendel's trials with purebred white-flowered (pp) and purple-flowered (PP) pea plants yielded a heterozygous purple (Pp) F_1 generation. When the F_1 plants self-pollinated, the white flowers reappeared. The F_2 plants were one-fourth PP , one-half Pp , and one-fourth pp .



Predict If you crossed two plants from the F_2 generation, what procedure would you follow to determine the genotypes of the next generation?

Modeling Genetic Crosses

In the early 1900s, several British scientists expanded upon Mendel's work. One, R. C. Punnett, explored genetic crosses with chickens and other species. The model he developed tracks the alleles each parent can donate to predict the outcome of crosses.

FIGURE 3: The common vizsla has smooth hair, but the wirehaired vizsla has a wiry coat.



a Smooth vizsla



b Wirehaired vizsla

Coat texture in dogs is a heritable characteristic. Some dogs, like the vizsla, can have a smooth coat or a wiry coat, and this trait is controlled by one gene. The wire-coated allele is dominant, noted as W , and the smooth-coated allele is recessive, noted as w .

Imagine a dog breeder wants to cross two wirehaired vizslas and that both dogs are heterozygous for the trait. Each parent is heterozygous for the wirehaired trait, so each one has two different alleles for coat texture. The alleles are separated into gametes during meiosis. There are two possible gametes for each parent, one for each allele he or she carries.



Analyze What alleles can each heterozygous vizsla parent pass on in his or her gametes?

The genotype of an organism indicates which alleles the organism carries for a certain characteristic. Each gamete contains one allele for each trait in an organism's DNA. Punnett recognized a relationship between parental gametes and the genotypes of offspring. He used this relationship to develop a simple table, now known as a **Punnett square**, that predicts all possible offspring genotypes resulting from a specific cross. This model is a quick and easy way to determine the probable outcome of a cross.

FIGURE 2: Purple plants in the F_1 generation self-pollinated to produce the F_2 generation.

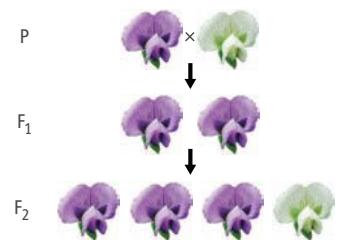
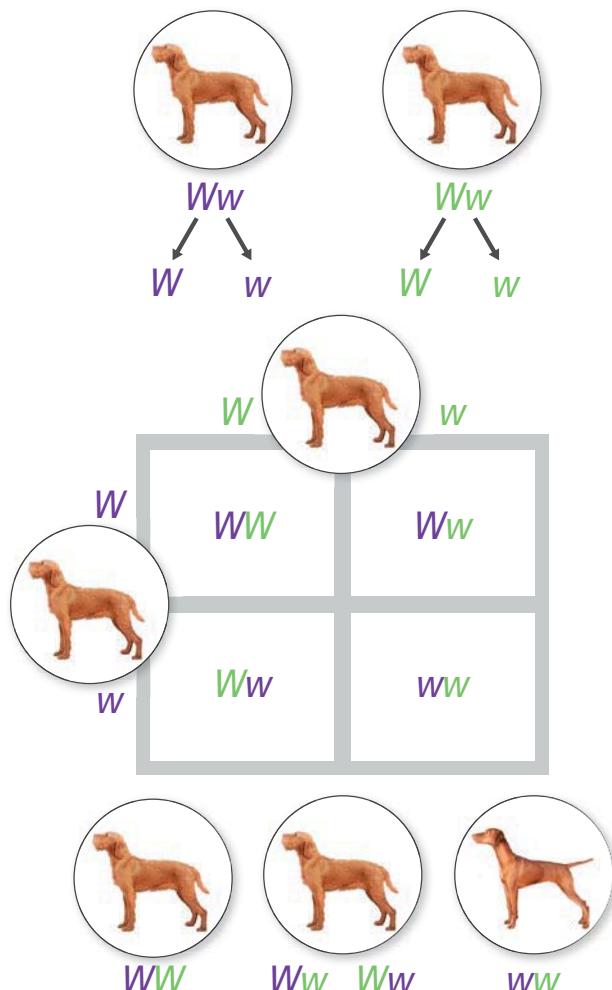


FIGURE 4: A Punnett square is used to model the cross between two parents with known genotypes.

Explore Online 



A model for the cross between two heterozygous, wirehaired vizslas is shown in Figure 4. Because each parent donates one gamete to each offspring, gametes will have either a dominant, wire-coated allele (*W*) or a recessive, smooth-coated allele (*w*).

To complete a Punnett square, divide a square into four equal sections. Write the alleles of each parent on the outside of the square, one set above the columns and one set to the left of the rows. Write the dominant allele first.

Next fill in each box in the Punnett square with the parent allele from the top of the column and the parent allele from the beginning of the row. When complete, each box will contain one allele from each parent.

The completed Punnett square shows three possible genotypes for coat type: homozygous dominant (*WW*), heterozygous (*Ww*), or homozygous recessive (*ww*). From these genotypes, we can predict that there is a one in four chance that the *WW* genotype will occur. There is a two in four chance that the *Ww* genotype will occur. Finally, there is a one in four chance that the *ww* genotype will occur.

In this cross, both the homozygous dominant and heterozygous genotypes will have wire coats. Only the homozygous recessive genotype will have a smooth coat.



Math Connection The wire-coated allele (*W*) is dominant to the smooth-coated allele (*w*). Use the Punnett square to answer the following questions:

1. What percentage of puppies will have the same genotype as the parents, *Ww*?
2. What percentage of puppies will have the wire-coat phenotype?
3. What percentage of puppies will have the smooth-coat phenotype?

A Punnett square models complex processes by focusing on desired traits rather than a genome. Pulling the letters that represent the parental genotype apart and placing them along the outside of the Punnett square shows the segregation of homologous chromosomes and possibly different alleles during meiosis. Each gamete contains only one version of the gene, and there is an equal opportunity for a gamete to contain either allele.

The assignment of alleles to the empty boxes models fertilization. Just as haploid gametes join to make a diploid zygote, the parental alleles join to make letter pairs in the Punnett square. The letter pairs represent potential offspring genotypes. This is the real value of a Punnett square. Modeling these processes makes it possible to predict the genotypes of offspring from a specific cross.



Analyze What do the letters on the top and side of a Punnett square represent?

Calculating Probabilities

Scientists use a branch of mathematics called *probability* to determine the likelihood that offspring will be born with certain characteristics. **Probability** is the chance that an outcome will occur, such as the birth of a dog with a wire coat. The probability of an event occurring can be determined using the following equation:

$$\text{probability} = \frac{\text{number of ways a specific event can occur}}{\text{number of total possible outcomes}}$$

An easy way to explore probability is by flipping a coin. Each flip has two possible outcomes: the coin either lands heads up or it lands tails up. The probability of the coin landing heads up is one out of two, or $\frac{1}{2}$. The probability of the coin landing tails up is also one out of two, or $\frac{1}{2}$. Probability is usually expressed on a scale of 0 to 1, with 0 being an impossible outcome and 1 being a certain outcome.

Now, consider what happens when you flip two coins at the same time, as shown in Figure 5. The results of the two flips are independent, so the result of one coin flip does not impact the result of the other. Both coins are free to land heads up or tails up. Calculate the probability of two independent events occurring together by multiplying the probability of the individual events. The probability of flipping heads is $\frac{1}{2}$. Therefore, the probability of flipping two heads together is $\frac{1}{2} \times \frac{1}{2} = \frac{1}{4}$.

Probabilities are averages, not exact numbers. If you flip a coin twice, you will not always get one heads and one tails. You may get two heads or two tails. The more you repeat an event, the closer you will get to the average described by probability.



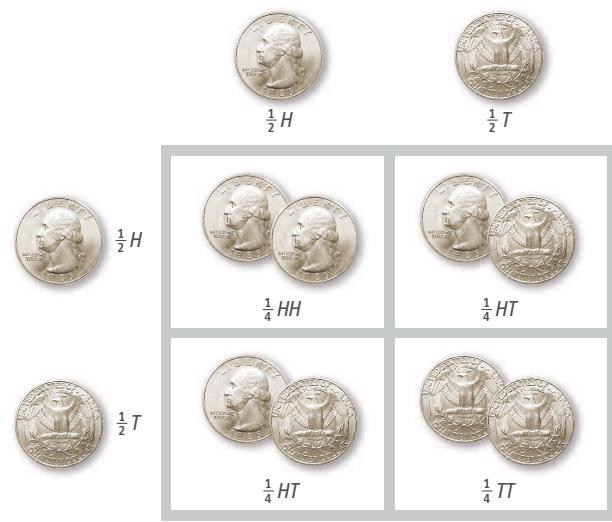
Math Connection Determine each of the following probabilities using Figure 4.

1. What is the probability of a *Ww* genotype? Of a *WW* genotype?
2. What is the probability of a puppy with a smooth coat being born?

In the cross modeled in Figure 4, what events would have to occur to produce a heterozygous puppy? The father could donate the dominant allele (*W*) and the mother could donate the recessive allele (*w*). The reverse could also occur. Both of these events would produce a heterozygous puppy, and both are equally likely to occur.

The probability of an event that can occur in more than one way is equal to the probability of the individual events added together. So, the probability of a sperm with a dominant allele fertilizing an egg with a recessive allele is $\frac{1}{4}$. The probability of a sperm with a recessive allele fertilizing an egg with a dominant allele is also $\frac{1}{4}$. Therefore, the probability of producing a heterozygote can be calculated as $\frac{1}{4} + \frac{1}{4} = \frac{1}{2}$. In other words, there is a one in two chance that a puppy will be born that is heterozygous (*Ww*) for a wire coat.

FIGURE 5: A Punnett square reflects the probability of two independent events occurring at the same time.



Explain How can a Punnett square help you explain the phenotypes of the kittens discussed at the beginning of this lesson? Use your knowledge of meiosis to help support your answer.



Patterns

The pattern of inheritance observed in sexually reproducing organisms is explained by chance. This makes probabilities particularly useful for analyzing some of the mathematics behind inheritance.

Determining Types of Crosses

FIGURE 6: A Curly Bashkir Horse



In most horse breeds, a smooth coat is dominant to a curly coat. The recessive allele is responsible for naturally curly coats that occasionally appear in some horse breeds. Because the gene is recessive, these occurrences are rare. In a few horse breeds, such as the Bashkir horse, the curly-coat allele, C , is dominant and the smooth-coat allele, c , is recessive.

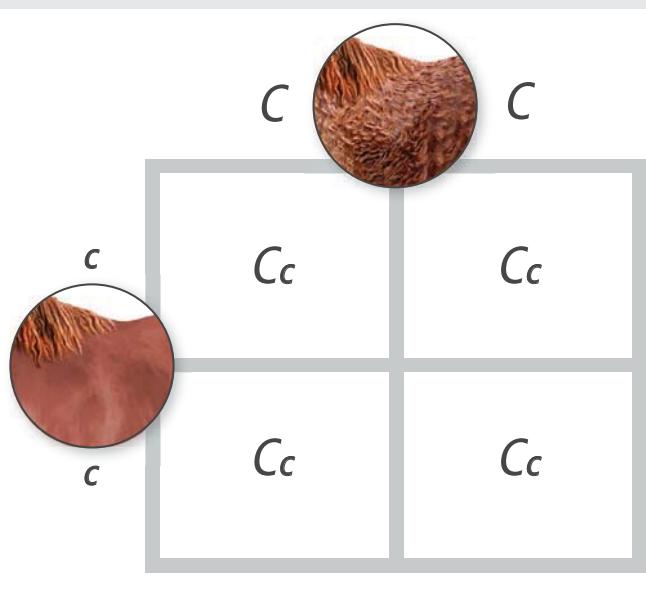


Predict Imagine you crossed a smooth-coated Bashkir horse with a curly-coated Bashkir horse. How could you determine the possible outcomes of this cross?

Analyzing the Inheritance of One Trait

All of the genetic crosses discussed so far have involved one trait, from flower color in pea plants to coat texture in dogs. A cross that examines one trait is a **monohybrid cross**. There are three basic types of monohybrid crosses: a homozygous-homozygous cross, a heterozygous-heterozygous cross, and a heterozygous-homozygous cross.

FIGURE 7: Homozygous-Homozygous Cross



Homozygous-Homozygous Cross

A homozygous-homozygous cross occurs when a homozygous dominant parent crosses with a homozygous recessive parent. Imagine that a Bashkir horse that is homozygous dominant for curly hair (CC) is crossed with a Bashkir horse that is homozygous recessive for smooth hair (cc).

The Punnett square in Figure 7 models the possible outcomes of the cross. As shown, a homozygous-homozygous cross always results in heterozygous offspring because one parent can donate only dominant alleles and the other can donate only recessive alleles. The sole possible outcome of the cross is one dominant allele and one recessive allele, which is a heterozygous combination. For the cross shown in Figure 7, all of the offspring would have the heterozygous genotype, Cc . They would have curly coats because the dominant curly-coat allele, C , is present in all genotypes. Each offspring would also carry the recessive smooth-coat allele, c .



Math Connection Probability is measured on a scale from 0 to 1. For a homozygous-homozygous cross, determine the following probabilities:

1. Probability of homozygous recessive offspring
2. Probability of homozygous dominant offspring
3. Probability of heterozygous offspring

Heterozygous-Heterozygous Cross

Imagine you wish to cross two curly-coated, heterozygous Bashkir horses. Each horse has the genotype Cc and can pass on either the dominant allele for curly hair or the recessive allele for smooth hair. The probability of each parent donating a dominant allele to the offspring is $\frac{1}{2}$. The probability of each parent donating a recessive allele to the offspring is also $\frac{1}{2}$.

Figure 8 shows the Punnett square for this heterozygous-heterozygous cross. From each parent, half the offspring receive a dominant allele (C) and half receive a recessive allele (c).



Math Connection In the heterozygous-heterozygous cross modeled in Figure 8, what is the probability of offspring with homozygous dominant, heterozygous, or homozygous recessive genotypes?

This type of cross for a single trait always results in a genotypic ratio of 1:2:1. This means that $\frac{1}{4}$ of offspring will have the homozygous dominant genotype, $\frac{2}{4}$ will have the heterozygous genotype, and $\frac{1}{4}$ will have the homozygous recessive genotype. The phenotypic ratio is 3:1 of dominant:recessive phenotypes. In other words, of the potential offspring phenotypes, $\frac{3}{4}$ will have the dominant phenotype and $\frac{1}{4}$ will have the recessive phenotype.

Heterozygous-Homozygous Cross

Now, imagine a heterozygous-homozygous cross between a heterozygous Bashkir horse with curly hair (Cc) and a homozygous recessive Bashkir horse with smooth hair (cc). From the homozygous parent, the offspring receive a recessive allele, c . From the heterozygous parent, half the offspring receive a dominant allele, C , and half receive a recessive allele, c .

Figure 9 shows the Punnett square for this heterozygous-homozygous cross. This cross results in two offspring with the genotype Cc and two offspring with the genotype cc .



Math Connection What is the probability the offspring in this cross will have a heterozygous genotype? What about a homozygous-recessive genotype?

A heterozygous-homozygous cross always produces parental genotypes in a 1:1 genotypic ratio. For the cross in Figure 9, the probability of offspring with the heterozygous genotype and the probability of offspring with the homozygous recessive genotype are both $\frac{1}{2}$. The phenotypic ratio in this instance is also 1:1, because the probability that each coat type will occur is $\frac{1}{2}$. So, in this cross, half of the offspring will have curly coats and half will have smooth coats.

FIGURE 8: Heterozygous-Heterozygous Cross

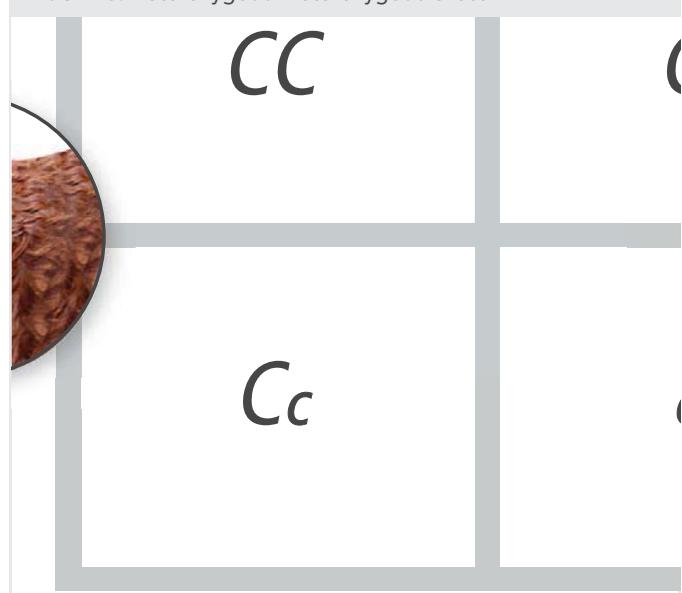
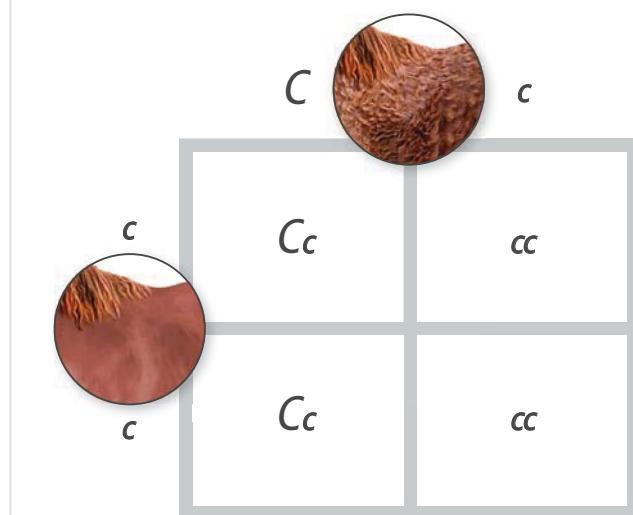


FIGURE 9: Heterozygous-Homozygous Cross



Analyze In your Evidence Notebook, complete a cross between a heterozygous horse (Cc) and a homozygous-dominant horse (CC). Were your results the same?



Hands-On Activity

Determining a Genotype

FIGURE 10: Peaches and nectarines are the same species, *Prunus persica*.



MATERIALS

- paper
- pencil



Peaches have fuzzy skin. A nectarine is a variety of smooth-skinned peach. A dominant allele, *G*, causes fuzzy skin. All peaches have at least one copy of this allele. Nectarines come from trees that are homozygous recessive (*gg*) for fuzz.

Imagine your company sells peach and nectarine seedlings. You developed a new type of peach tree that is very popular. To meet demand, you must learn the genotypes of your breeding stock. You determine them by setting up a **testcross** between an individual that has a dominant phenotype but an unknown genotype and an individual that is homozygous recessive.



Predict How can a testcross help you find the unknown genotype of the plant?

PROCEDURE

1. Plant A produces peaches. You need to determine its genotype. Plant B produces nectarines that have smooth skin and a known genotype of *gg*. You cross Plant A with Plant B.
3. The resulting cross yields twelve plants. Six plants produce peaches upon the first fruiting and six plants produce nectarines upon the first fruiting.
4. Use Punnett squares to determine the genotype of Plant A.

ANALYZE

Answer the following questions in your Evidence Notebook:

1. What is the genotype of Plant A? Explain how you arrived at your answer.
2. Plant A is crossed with a plant that has a genotype of *GG*. What are the possible genotypes and phenotypes of the offspring?
3. Plant A is crossed with a plant that has a genotype of *Gg*. What is the ratio of dominant to recessive phenotypes of the offspring?
4. In terms of genotype, is Plant A the best plant to produce as many peach seedlings as possible? Why or why not? Which genotype would be best?

Analyzing the Inheritance of Two Traits

A **dihybrid cross** examines the inheritance of two traits. Consider the peas shown in Figure 11, which can be yellow or green and round or wrinkled. The yellow allele, *Y*, is dominant to the green allele, *y*. The round allele, *R*, is dominant to the wrinkled allele, *r*. Figure 12 shows a cross between two heterozygous plants (*YyRr*). Each gamete receives one allele for pea color and one allele for pea shape. Each pea color allele has an equal probability of being paired with each pea shape allele. There are four possible combinations of alleles in heterozygous dihybrid gametes. The probability of producing any of the four gametes is one out of four.

FIGURE 11: Phenotypes of Peas



FIGURE 12: A dihybrid cross between two heterozygous pea plants.

		F ₁ generation					
		YR	Yr	<i>YyRr</i>	yR	yr	
YR	YR	YYRR	YYRr	<i>YyRR</i>	<i>YyRr</i>		
	Yr	YYRr	YYrr	<i>YyRr</i>	<i>Yyrr</i>		
	yR	<i>YyRR</i>	<i>YyRr</i>	yyRR	<i>yyRr</i>		
	yr	<i>YyRr</i>	<i>Yyrr</i>	<i>yyRr</i>	<i>yyrr</i>		
F ₂ generation							



Gather Evidence

Determine the number of possible phenotypes in the dihybrid cross. What is the ratio for all the possibilities?



Math Connection Use the Punnett square to answer the following questions:

- What is the probability that the cross will produce a plant that is heterozygous for both traits? What is the probability of producing a plant with yellow and round peas? Why are these two probabilities different?
- Make a Punnett square for the dihybrid cross *YyRr* and *yyrr*. How are the probabilities of this cross different from those in Question 1?

In this cross, the chance of producing offspring that exhibit both dominant traits (yellow and round) is $\frac{9}{16}$. The chance of producing offspring that exhibit one dominant trait and one recessive trait (yellow and wrinkled or green and round) is $\frac{3}{16}$. Finally, the chance of producing offspring that exhibit both recessive traits (green and wrinkled) is $\frac{1}{16}$. Using Figure 12, you can see these possibilities. There are nine yellow and round peas, three yellow and wrinkled peas, three green and round peas, and one green and wrinkled pea. Therefore, a heterozygous-heterozygous dihybrid cross results in a phenotypic ratio of 9:3:3:1.



Explain Why are Punnett squares a useful model for scientists studying traits and genetic disorders? In which other types of careers would this model be useful?

Sex-Linked Inheritance

Human offspring have an equal probability of being male (XY) or female (XX). The mother donates an X chromosome, so the chromosome donated by the father is the one that determines the sex of the offspring. The father could donate either an X chromosome, in which case the child would be female, or a Y chromosome, in which case the child would be male, as shown in Figure 13. The probability of either occurrence is $\frac{1}{2}$.

FIGURE 13: Females donate an X chromosome to offspring while males can donate either an X or a Y chromosome.



a Female Sex Chromosomes

b Male Sex Chromosomes



Predict How would the inherited traits discussed in this lesson be influenced if those alleles were on a sex chromosome? Would the probability of inheritance change?

Expressing Sex-Linked Traits

Explore Online

Hands-On Lab



Sex-Linked Inheritance Use a model to determine the pattern of inheritance for sex-linked traits.

Genes located on sex chromosomes are **sex-linked genes**. These genes follow a pattern of inheritance called *sex-linked inheritance* and are not always connected to sexual characteristics. All other genes occur on autosomes, or non-sex chromosomes, and follow autosomal inheritance patterns. Few genes appear on both the X and Y chromosome, so males, with only one X chromosome, often express X-linked genes.

To prevent the double expression of sex-linked traits in females, female embryos go through the process of X inactivation. During this process, one X chromosome in each cell randomly becomes inactive very early in development. All descendants of these early cells have the same inactive X. This process does not impact the phenotype of homozygous females because both of their X chromosomes have the same allele. Heterozygous females can be impacted by X inactivation, depending upon the genes involved.



Analyze Imagine an X-linked recessive disease. X^A represents the dominant allele and X^a represents the recessive allele. What are the different kinds of gametes a heterozygous female and a male with a dominant allele can produce?

Analyzing the Inheritance of Sex-Linked Traits

Cone cells in the human eye have color-sensing molecules called *photopigments* that normally respond to either red, blue, or green light. The most common type of color blindness, red-green color blindness, involves abnormalities in the photopigments in green or red cone cells. The genes responsible for red-green color blindness are located on the X chromosome, so red-green color blindness is a sex-linked trait.

The dominant allele that produces normal vision is represented by the C superscript (X^C). The recessive allele that is responsible for red-green color blindness is represented by the c superscript (X^c). For heterozygous females, the presence of one dominant allele is enough to overcome the expression of the recessive allele.

When using a Punnett square to perform a sex-linked cross, place the female chromosomes at the top of the square and the male chromosomes to the left of the square. Sex-linked crosses track sex chromosomes *and* the trait of interest simultaneously. These characters are linked and therefore always appear together as a capital letter for the sex chromosome and a superscript for the trait of interest.



Math Connection Using the Punnett square in Figure 14, determine the probabilities that a couple will have a colorblind child, a colorblind son, or a colorblind daughter.



Gather Evidence

Which genotypes for males and females result in normal vision and which result in color blindness?

FIGURE 14: A cross between a female heterozygous for red-green color blindness and a male with normal vision.

X^C	X^c	
X^C	$X^C X^C$	$X^C X^c$
Y	$X^c Y$	$X^c Y$

Most sex-linked traits occur on the X chromosome. Thus, sex-linked inheritance patterns are mostly due to differences in expression of the X chromosome. An affected male offspring requires only a single recessive allele, while an affected female requires two recessive alleles. This decreases the likelihood that a female will be homozygous recessive. She is more likely to be a heterozygous carrier of the recessive trait.

Sex-linked crosses are similar to monohybrid crosses, but there is a key difference—the trait and the sex chromosome are inherited as one unit and cannot be separated. So, in a male unaffected by a sex-linked condition with genotype $X^C Y$, the normal allele, C, and the X chromosome are always inherited together.



Engineering

For those affected by or are carriers of a heritable disorder, the decision of whether to have children can be monumental. Genetic counseling helps inform this decision by predicting the likelihood that a particular couple will have a child with an inherited disease. Genetic counselors use Mendelian genetics, pedigrees, and genetic tests to model the potential outcomes for prospective parents.



Explain How do the genotypic and phenotypic ratios of the sex-linked traits differ from those of a monohybrid cross?

Data Analysis

Pedigrees

Long before DNA testing made it possible to determine genotypes analytically, scientists constructed pedigrees to study inheritance patterns. A pedigree is a family tree that tracks a trait through multiple generations.

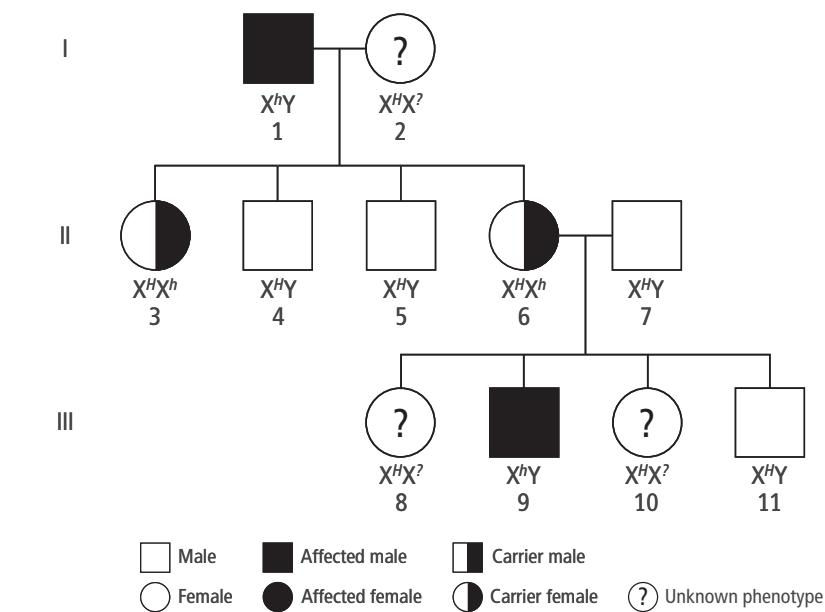
The inheritance pattern of hemophilia can be determined by analyzing a pedigree. Hemophilia is a sex-linked disorder that causes uncontrolled bleeding because the body fails to make one or more clotting factors. It can be fatal if untreated.

Pedigrees are built using symbols to represent relationships between individuals. Figure 15 is a pedigree following hemophilia through three generations. Males are represented by squares and females are represented by circles. A direct line between two individuals indicates a relationship. Siblings are listed from left to right in order from oldest to youngest, connected by a sibling relationship line. Parents and offspring are connected by a line of descent.

Fully shaded shapes represent individuals who are affected by the trait of interest—hemophilia. Affected males must have the hemophilia allele on the X chromosome, represented by X^h . Unaffected males must have a normal allele at this location, represented by X^H .

Half-shaded shapes represent carriers. No females in the second generation have hemophilia. Therefore, they all must have at least one normal allele for this gene. Also, their father can only pass along the hemophilia allele to his

FIGURE 15: A pedigree tracing hemophilia through three generations.



daughters. Therefore, all daughters in the second generation are heterozygous for this condition, $X^H?X^h$. These females are carriers of the hemophilia gene.

It is impossible to determine the genotype of Female 2. She may be a carrier for hemophilia who passed along a normal allele to all of her children. Or she could be homozygous dominant for this gene. This unknown allele on the X chromosome is represented by $X^?$.

It is also impossible to determine the genotypes of Females 8 and 10. The father can donate only a dominant allele. The mother can donate either a dominant or a recessive allele. Therefore, the daughters are either homozygous dominant or heterozygous at this location. Again, the unknown allele on the X chromosome is represented by $X^?$.



Data Analysis

Use the pedigree to answer the following questions.

- Is hemophilia a dominant or recessive trait? Use evidence to support your claim.
- In the second generation, how many females are carriers of the gene? What is their genotype?
- Imagine Male 9 married a female carrier. What is the probability that they will produce a female child with hemophilia? A child who does not have hemophilia? A child who has the parental phenotypes? Use evidence to support your answers.

PRACTICING
GENETIC CROSSES



MODELING MONOHYBRID & DIHYBRID CROSSES

ANIMAL BREEDING

Go online to choose one of these other paths.

Lesson Self-Check

CAN YOU SOLVE IT?

FIGURE 16: Kittens with tortoiseshell fur resulted from crossing an orange cat with a black cat.



a Female cat



b Male cat



c Tortoiseshell kitten from the litter

Recall the cat breeder from the beginning of the lesson. The breeder hoped to produce a litter of kittens in which half the kittens were orange and half were black. To achieve this, the breeder crossed an orange female cat with a black male cat. When the kittens were born, three were male and three were female. As expected, half the kittens had orange fur. However, the remaining kittens had a mixture of orange and black fur called *tortoiseshell*. To complicate things further, the orange kittens were all males and the tortoiseshell kittens were all females.



Explain Refer to the notes in your Evidence Notebook to answer the following questions:

1. Why was the litter of kittens not half black and half orange?
2. Why were there only female tortoiseshell kittens?
3. Which alleles were passed on by each parent cat in this cross? Which alleles did the male offspring receive? Which alleles did the female offspring receive?

Tortoiseshell coloring in cats is usually expressed only in females. This tells us that the gene controlling black and orange color is located on the X chromosome. Males have one X chromosome with either an allele for orange fur (X^B) or one for black fur (X^b).

This gives two possible genotypes for males: $X^B Y$ or $X^b Y$. Because males have only one version of the allele, they will always express that allele. Females, however, have two X chromosomes. Thus, they can be homozygous for orange fur ($X^B X^B$), homozygous for black fur ($X^b X^b$), or heterozygous ($X^B X^b$).

Remember, in females one X chromosome in each cell is inactive. X inactivation does not impact homozygous females ($X^B X^B$ and $X^b X^b$) because the same allele is expressed regardless of which X chromosome is active. X inactivation impacts heterozygous females ($X^B X^b$) because it is random. The color expressed by each cell depends on which of the two chromosomes is active. Black fur occurs on skin patches that have an inactive X^B allele. Orange fur occurs where the X^b allele is inactive. The patches of color occur randomly, giving these females their characteristic mosaic tortoiseshell pattern.

CHECKPOINTS

Check Your Understanding

Use the following information to answer Questions 1–3.

Two heterozygous, wirehaired vizslas were crossed. The genotypes of their potential offspring are shown in the Punnett square in Figure 17.

FIGURE 17: Heterozygous-Heterozygous Cross

	<i>W</i>	<i>W</i>
<i>W</i>	<i>WW</i>	<i>Ww</i>
<i>W</i>	<i>Ww</i>	<i>WW</i>

- What is the phenotypic ratio of wire-coated to smooth-coated offspring?
- What is the genotypic ratio of homozygous dominant to heterozygous to homozygous recessive offspring?
- What genotype has a 100% chance of expressing a recessive allele?

Use the following information to answer Questions 4 and 5.

Duchenne muscular dystrophy is an X-linked recessive disease that causes degeneration and weakness in muscles. The normal condition is represented by the superscript *D*, and the allele that causes Duchenne muscular dystrophy is represented by the superscript *d*.

- Draw a Punnett square to show a cross between a homozygous-dominant female and a male with Duchenne muscular dystrophy.
- Which combination of parental genotypes is most likely to result in carrier daughters?

Use the following information and the Punnett square in Figure 18 to answer Questions 6–8.

The trait for purple flowers in pea plants (*P*) is dominant to the trait for white flowers (*p*).

FIGURE 18: Heterozygous-Homozygous Recessive Cross

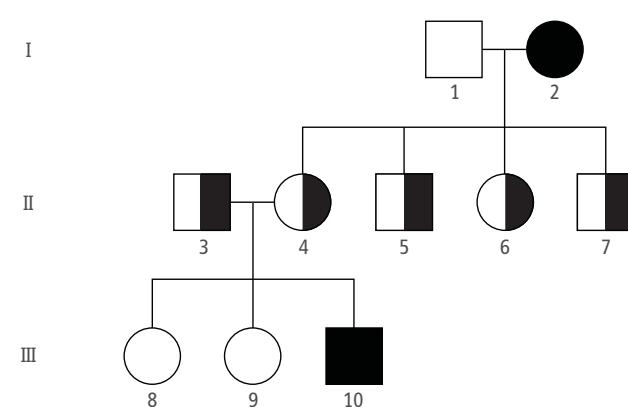
<i>P</i>		<i>p</i>
<i>p</i>	<i>Pp</i>	<i>pp</i>
<i>p</i>	<i>Pp</i>	<i>pp</i>

- What is the probability that the heterozygous parent will donate a recessive *p* allele?
- What is the probability that the homozygous-recessive parent will donate a recessive *p* allele?
- What is the probability of both parents donating a recessive *p* allele?
- Why is the known genotype in a testcross always homozygous recessive? Provide an example to support your claim.
- For each pair, calculate the probability of producing a homozygous recessive genotype. Then place the pairs in order of increasing probability.
 - Aa* × *aa*
 - aa* × *aa*
 - Aa* × *Aa*
- Parents of genotype *AABB* and *aabb* were crossed and produced all heterozygotes with the genotype *AaBb*. Heterozygotes from the *F₁* generation were crossed and produced a phenotypic ratio of 9:3:3:1. How does this sequence of events support the law of independent assortment?

Use the following information and the pedigree in Figure 19 to answer Questions 12–14.

This simple pedigree traces an autosomal-recessive disorder across three generations. This disorder is not sex-linked and follows Mendelian patterns of inheritance. The dominant allele is *A*, and the recessive allele that causes the disorder is *a*.

FIGURE 19: Autosomal-Recessive Pedigree



12. The four siblings in the second generation have the same genotype. What is it?
13. What is the most likely genotype of the father in the first generation?
14. What is the genotype of both affected individuals?
15. Imagine a plant can have striped flower petals or solid flower petals. Solid coloring (*Z*) is dominant to stripes (*z*). Which parental cross would yield the following ratio of offspring: 1 homozygous dominant (*ZZ*): 2 heterozygotes (*Zz*): 1 homozygous recessive (*zz*)?
 - a. homozygous dominant–homozygous recessive
 - b. homozygous dominant–homozygous dominant
 - c. homozygous dominant–heterozygous
 - d. heterozygous–heterozygous

Use following information to answer Questions 16–18. Make a Punnett square for each cross to support your answers.

In pea plants, yellow seed color (*Y*) is dominant to green seed color (*y*); round seeds (*R*) are dominant to wrinkled seeds (*r*).

16. What is the probability that parents with the genotypes *YyRR* and *YYRR* will produce an offspring with the genotype *YYRR*?
17. What is the probability that parents with the genotypes *yyrr* and *YyRr* will produce offspring with the genotype *yyrr*?
18. What is the probability that parents with the genotypes *YYRR* and *yyrr* will produce offspring with the genotype *YyRr*?

MAKE YOUR OWN STUDY GUIDE



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

The expression of genes determines an organism's phenotype.

Punnett squares can be used to determine the probability of offspring expressing certain traits.

If genes are sex-linked, males will express the allele found on the X chromosome while females express the allele on the active X chromosome. If the gene is located on the Y chromosome, it is expressed only in males.

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.

There is a cause-and-effect relationship between an organism's DNA and its phenotype. Consider other cause-and-effect relationships, such as the effect a parent's DNA has on offspring.

Mutations and Genetic Diversity

Mutations can affect the sequence of nucleotides in your DNA. Screening techniques can be used to detect these changes.

CAN YOU EXPLAIN IT?

FIGURE 1: Red blood cells normally have a rounded shape. A mutation in a protein called hemoglobin causes red blood cells to have a “sickled,” or bent, shape.



Image credits: (t) ©Kevin Curtis/Science Source; (b) ©Science Picture Co./Getty Images



Gather Evidence

As you explore the lesson, gather evidence to explain how mutations increase genetic diversity.

When you think of mutations, you may imagine enhanced, superhuman abilities, or you may think of negative effects on the body. Some mutations can be beneficial, while others can be quite harmful. For example, sickle cell anemia is a disease caused by a mutation that affects red blood cells. The result is anemia, or a shortage of healthy red blood cells in the body. Other symptoms include fatigue, pain, swelling of hands and feet, and delayed growth. The sickle cell anemia allele, HbS, causes the disease and can be passed on from parent to offspring. Despite its damaging effects, the HbS allele persists in relatively high frequencies in some parts of the world. These areas are typically near Earth’s equator and include parts of Africa and the Mediterranean.



Predict Why might the HbS allele be more common in some parts of the world than in others? What do you think causes this pattern?

Gene Mutations

What you are made of and how your body functions begins with the instructions from your DNA. Your DNA carries the code from which all the proteins that give your body structure and help your body carry out life-maintaining processes are produced. Changes in DNA, or mutations, may result in diseases like sickle cell anemia. How do mutations occur and what causes them?

Causes of Mutations

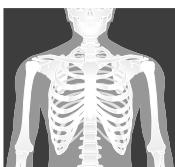
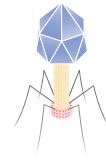
Mutations can be categorized as gene mutations or chromosomal mutations.

Gene mutations are changes in the DNA sequence of a single gene. Typically, gene mutations happen during DNA replication. DNA polymerase has a built-in proofreading function that repairs mutations, but a small number of replication errors do not get fixed. They build up over time, and can eventually affect how the cell works. Many studies suggest that mutations in somatic cells, coupled with a decrease in the body's self-repairing ability, may contribute to the process of aging.

Mutagens are agents in the environment that can change DNA or increase the frequency of mutation in organisms. Some mutagens occur naturally, such as ultraviolet (UV) rays in sunlight. Some chemicals have also been linked to mutations, such as those in food and cosmetics. Biological mutagens include bacteria and viruses.

 **Collaborate** When you get x-rays at the dentist, a lead vest is placed over your body. Write why you think this is necessary, and explain to a partner.

FIGURE 2: Mutagens can change DNA. The main types of mutagens include radiation, chemicals, and infectious agents.

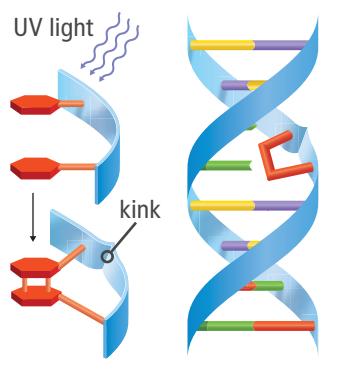
Radiation		Chemicals			Infectious Agents	
						
X-rays (medical uses)	UV (from sunlight)	Processed foods and preservatives	Cleaning products and cosmetics	Carcinogens (e.g., cigarettes)	Viruses (e.g., HPV)	Bacteria (e.g., H. pylori)

One example of a mutation caused by a mutagen is a thymine dimer. Recall that in DNA, adenine always pairs with thymine. UV light can cause neighboring thymine nucleotides to break their hydrogen bonds to adenine and bond together, forming a thymine dimer. The dimer causes the DNA to kink, which interferes with replication. Cells have a process for correcting these mutations. One enzyme removes the thymine dimer, another replaces the damaged section, and a third bonds the new segment in place. Sometimes, this process is not effective. When these mutations are not corrected in genes that regulate cell and tumor growth, they may result in cancer.



Explain Some cancer drugs take advantage of mutagenic properties. One type of drug wedges its way between nucleotides in DNA. Explain how the action of this drug would cause cancer cells to eventually lose their ability to function and reproduce.

FIGURE 3: Thymine Dimer



Point Mutations

A point mutation is a mutation in which one nucleotide is substituted for another. In other words, an incorrect nucleotide takes the place of the correct nucleotide. Very often, such a mistake is caught and fixed by DNA polymerase. If not, the substitution may permanently change an organism's DNA.



Cause and Effect

Let's examine some mutations and determine their effects on the sequence of amino acids. Remember that in protein synthesis, the DNA code is transcribed to make a strand of mRNA, which is then translated into a sequence of amino acids using codons. Some mutations affect the amino acid sequence, which can affect the structure and function of the resulting protein.

FIGURE 4: A codon chart shows which amino acids correspond to each possible combination of mRNA bases.

AUG 344

A blue magnifying glass icon with a white circular search area.

Language Arts

 **Connection** Research a human health condition caused by a mutation, and write a blog post explaining how people are working to address the condition. What has been done to raise awareness of the condition? How are scientists approaching this condition? What kinds of treatments have been proposed so far, and which of them seems most promising?

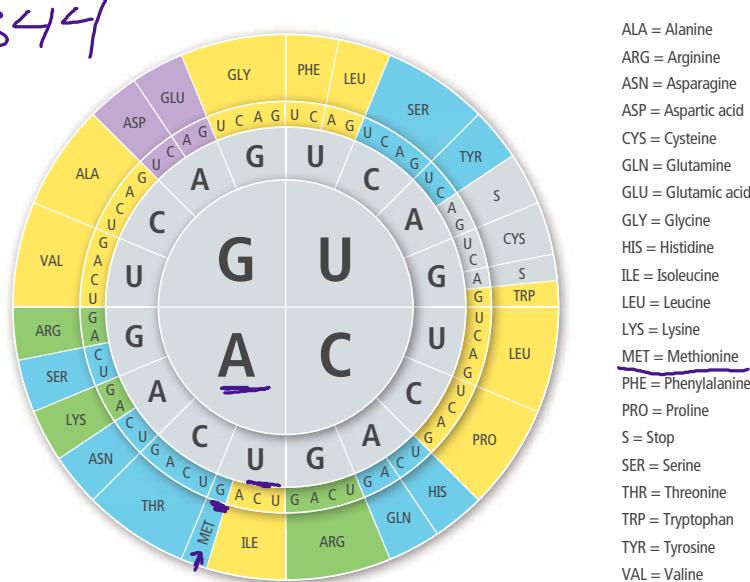


FIGURE 5: Normal and Mutated DNA Sequences

	Normal sequence	Mutation 1	Mutation 2	Mutation 3
DNA	CTC	CAC	ATC	CTT
mRNA	GAG	GUG	UAG	GAA



Analyze Use the chart in Figure 4 to analyze the DNA sequences in Figure 5.

1. For each mRNA sequence, determine the corresponding amino acid.
 2. Which mutations changed the identity of the amino acid as compared to the normal sequence?
 3. If you had to create names for the three types of mutations you analyzed, what would they be?

Mutations that change a codon, but not the identity of an amino acid in a protein, do not affect the amino acid sequence of that protein. This type of mutation is sometimes called a “silent mutation” because it does not change the structure and function of the protein. However, there are times when the substitution of a base results in a change in a codon and consequently in a new amino acid. This is called a “missense” mutation. If a mutation results in a “stop” codon being formed, the protein will not be complete. This is called a “nonsense” mutation. In both types, the amino acid sequence has changed and the protein’s structure and function may be altered.

Sickle cell anemia is caused by a point mutation that alters the gene which codes for the hemoglobin protein in red blood cells. Hemoglobin is made of four subunits with each of the subunits containing iron. This arrangement allows red blood cells to be efficient in transporting oxygen molecules from the lungs to the cells because oxygen molecules bind to the iron atoms. In HbS alleles, glutamic acid is substituted by valine. The protein synthesized using the mutated gene as a template has a different structure than that of a typical hemoglobin protein.

Glutamic acid is a negatively-charged amino acid that is attracted to positively-charged amino acids. This interaction between amino acids helps the protein keep its shape. Unlike glutamic acid, valine is not attracted to positively-charged amino acids. So, instead of grouping together to form the structure in Figure 6, the hemoglobin subunits form long, rigid chains. This results in red blood cells that have a “sickle” shape.

FIGURE 6: Hemoglobin has four subunits, each with an iron atom to which oxygen molecules attach.

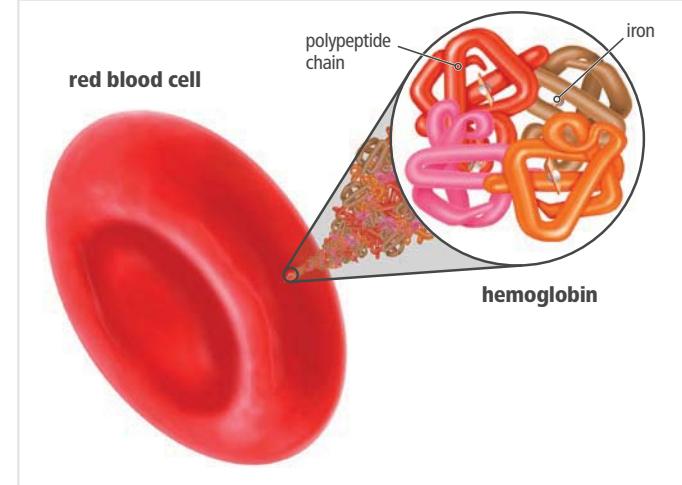
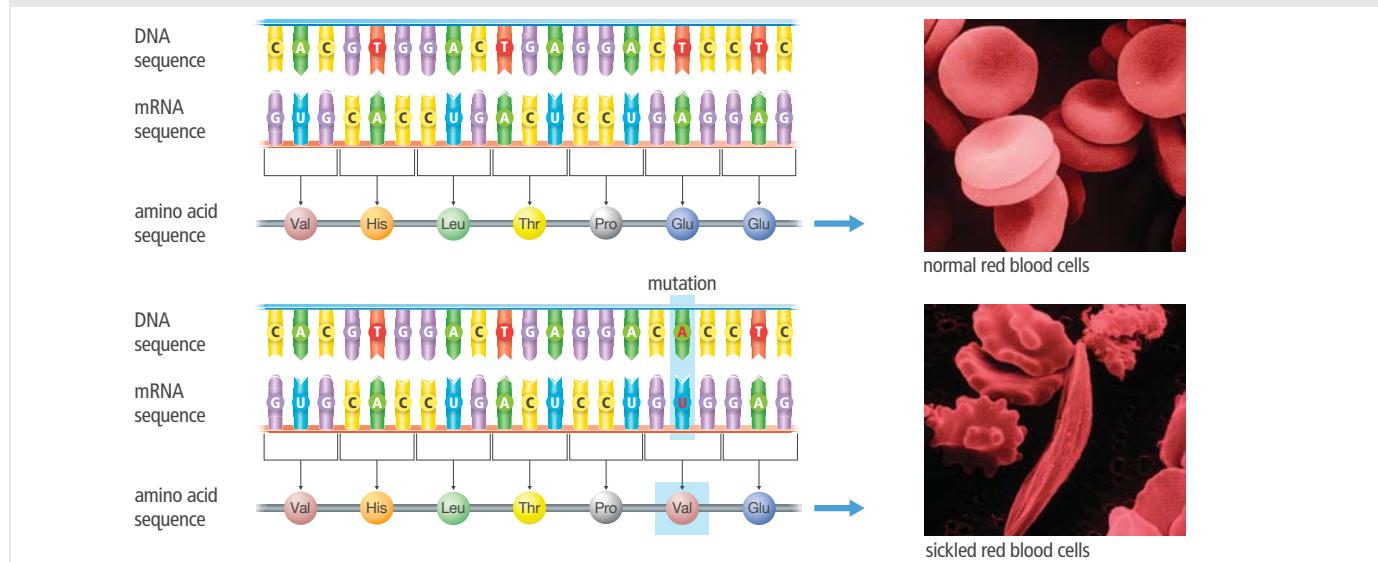


FIGURE 7: Sickle cell anemia results from a mutation that alters the structure of hemoglobin.



When sickle-shaped red blood cells stack on top of each other, they can clog blood vessels. This mutation causes anemia, and consequently fatigue and the other symptoms of sickle cell anemia. The cells do not get enough oxygen to produce the energy the body needs to properly maintain processes that keep the body healthy.



Model Draw a flow chart to illustrate how a change in a nucleotide in a DNA strand leads to symptoms experienced by those with sickle cell anemia.

Frameshift Mutations

A frameshift mutation involves the insertion or deletion of one or more nucleotides in the DNA sequence. This mutation changes the reading frame, or the arrangement of nucleotides into codons. To understand how a frameshift mutation affects an mRNA strand, imagine a short sentence of three-letter “codons”:

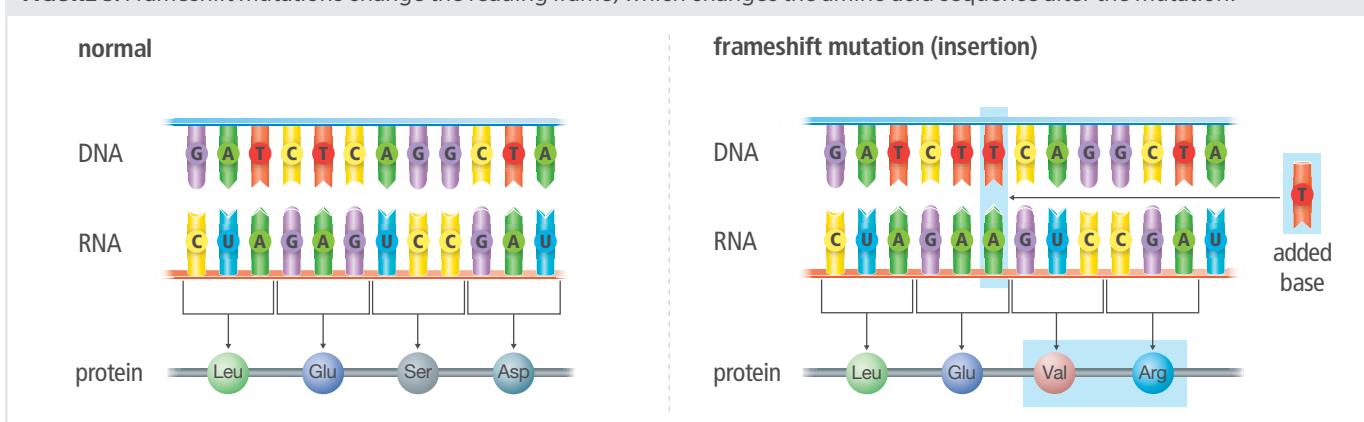
THE CAT ATE THE RAT

If the letter *E* is removed, or deleted, from the first “THE,” all the letters that follow shift to the left. The sentence now reads:

THC ATA TET HER AT...

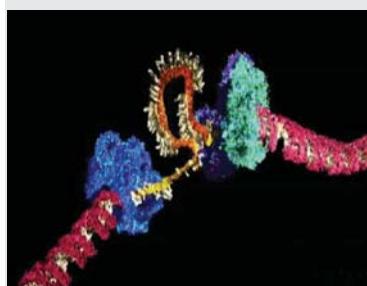
The sentence no longer makes sense. The same would be true if a nucleotide was added, or inserted, and all the letters shifted to the right, as shown in Figure 8.

FIGURE 8: Frameshift mutations change the reading frame, which changes the amino acid sequence after the mutation.



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FIGURE 9: Trinucleotide repeat expansions make a loop of duplicate nucleotides.



A nucleotide sequence loses its meaning when an insertion or deletion shifts all the codons by one nucleotide. This change throws off the reading frame, which results in codons that code for different amino acids.

Trinucleotide Repeat Expansions

Frameshift mutations may also occur in sections of DNA that consist of repeating nucleotides, such as CAG CAG CAG. These repeating segments are known as trinucleotide repeats because they involve three nucleotides. During replication, DNA polymerase may “slip” and make duplicate copies of the repeated sequence. This forms a “hairpin” loop of DNA that sticks out from its complementary strand. When this strand is replicated, the loop becomes part of the DNA, resulting in a longer double strand of DNA. This expansion continues as cells divide and DNA is replicated.



Analyze People with sickle cell anemia have two copies of the HbS allele. People with one copy are carriers and do not have the disease.

1. Is the sickle cell allele dominant or recessive? Explain how you know.
2. If two carriers have children, what is the probability of one of their children having the disease?

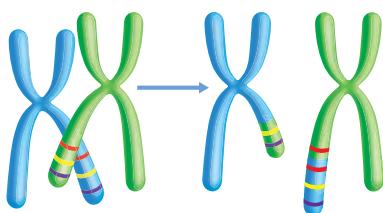
Chromosomal Mutations

Chromosomal mutations are changes in either chromosome segments or whole chromosomes. These mutations may change the amount of genetic material or change the structure of a chromosome, and they usually occur during mitosis and meiosis.

Gene Duplication

During meiosis, homologous chromosomes exchange DNA segments through crossing over. If the chromosomes do not align with each other, a segment of one chromosome may break off and attach itself to the other chromosome, resulting in one chromosome with two copies of a gene or genes. This process is called gene duplication. The chromosome that lost the segment has undergone gene deletion.

FIGURE 10: The douc langur has digestive enzymes that evolved as the result of a gene duplication event. These enzymes allow douc langurs to digest leaves and fruits.



Mutations can have harmful effects, but they can also increase **genetic variation**, or the variety of traits among individuals within a population. Gene duplication has occurred many times in the evolution of eukaryotic organisms. When gene duplication occurs, multiple copies of a gene are present. As a result, one copy of the gene can encode functional proteins, while the other copies are “free” to accumulate mutations. Mutated genes may encode proteins with new structures, which may take on new functions in the organism.

Model Draw a model illustrating how gene duplication and mutations can lead to a gene with a new function over the course of several generations.



Engineering

Sometimes the entire genome is duplicated. This type of error can lead to **polyploidy**, or multiple copies of the genome. Genome duplication has occurred in the evolution of many crop plants, such as strawberry, wheat, and mustard plants.

Scientists can use chemicals to artificially induce polyploidy in cells. These chemicals interfere with the formation of microtubules, disrupting the separation of chromosomes during mitosis. As a result, one daughter cell receives a double set of chromosomes. This technique has been used to manipulate traits such as flower size to make plants more desirable to customers.



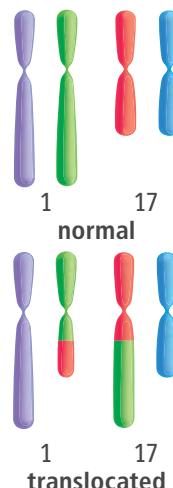
Analyze Suppose you wanted to chemically induce polyploidy to make a plant with larger fruit. Write a list of questions you would ask to define and delimit the problem.

FIGURE 11: Polyploidy in Plants

Common Name	Chromosome Number
Banana	$3N = 33$
Potato	$4N = 48$
Common wheat	$6N = 42$
Boysenberry	$7N = 49$
Strawberry	$8N = 56$

Gene Translocation

FIGURE 12: Gene Translocation



Translocation is another type of chromosomal mutation. In translocation, a segment of one chromosome moves to a nonhomologous chromosome. Translocations are often reciprocal, which means that the two nonhomologous chromosomes exchange segments with each other. In Figure 12, a translocation occurs between chromosome 1 and chromosome 17. This is known as a balanced translocation because the swapping of segments did not break up any genes, and there was no gain or loss of material.

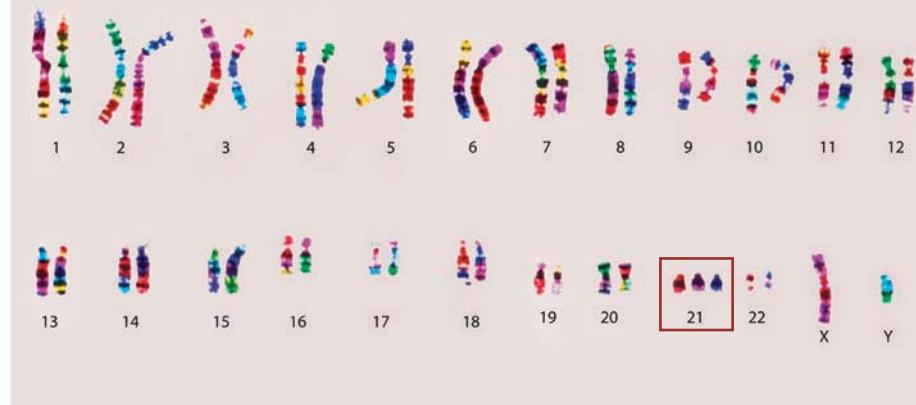


Predict Many people with balanced translocation mutations are not aware they have them until they try to have children. How might this be possible?

Nondisjunction Mutations

Nondisjunction mutations occur when one or more homologous chromosomes do not separate during anaphase of meiosis. The resulting gametes do not have the same number of chromosomes and can have more or fewer chromosomes than the parent cell.

FIGURE 13: A karyotype can be used to identify a nondisjunction mutation.



Examples of human disorders caused by nondisjunction include Down syndrome and Klinefelter disorder. Down syndrome occurs in people with three copies of chromosome 21. Klinefelter disorder is caused by an extra X chromosome in the cells of males. Recall that males have one X and one Y chromosome. A male with this disorder would have three chromosomes: XXY. This mutation affects the learning ability and sexual development of males. Turner syndrome is another example of a disorder caused by nondisjunction. Females with this syndrome have only one X chromosome instead of two. This missing X chromosome interferes with the development of secondary sexual characteristics in females.



Model Draw a model to illustrate how a nondisjunction mutation could occur during either anaphase I or anaphase II of meiosis.



Explain Make a chart to organize and describe the main types of mutations you have learned about so far. Then use your chart to help you write an explanation for these questions: When is a mutation likely to increase genetic variation? When is a mutation likely to have harmful effects?

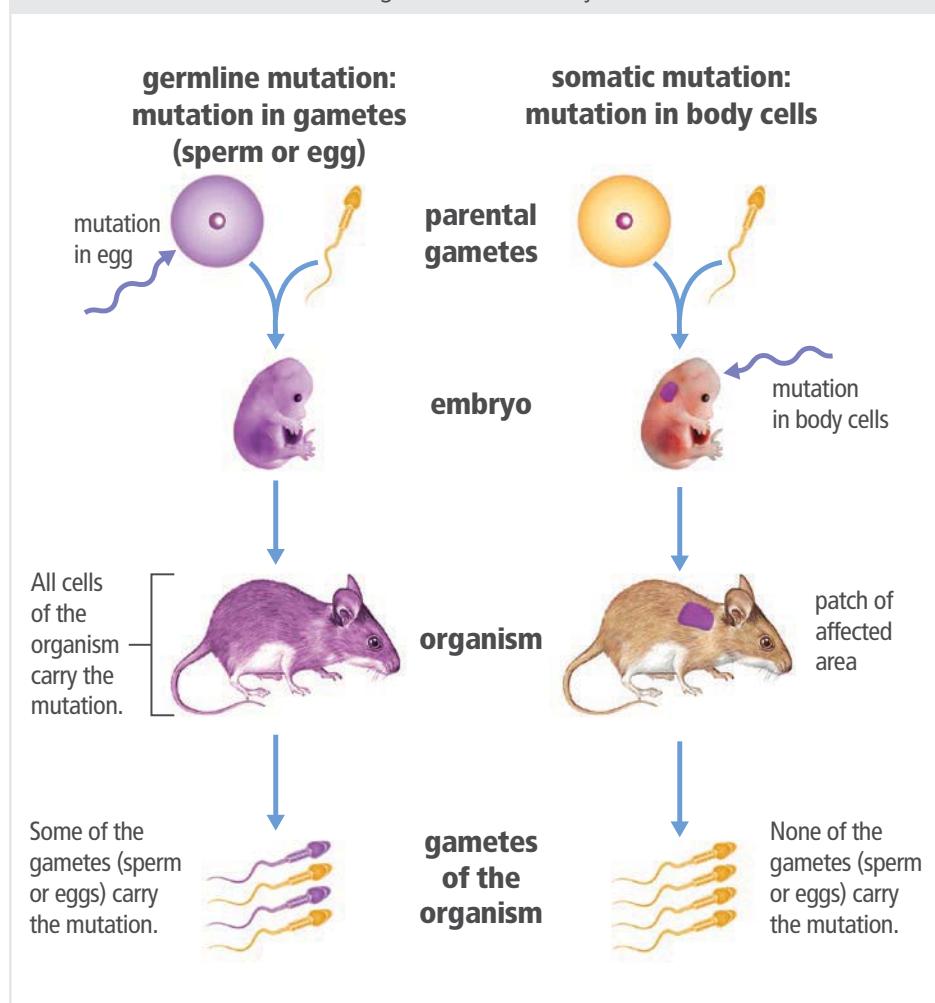
Effects of Mutations

You have seen how HbS, the sickle cell anemia allele, can be passed on from parent to offspring. Whether or not a mutation such as this gets inherited depends on the type of cell in which the mutation occurs. If a mutation is transmitted, it may or may not affect the phenotype, or the physical expression of a trait, in the organism.

Impacts on Offspring

There are two major types of cells in the body: body cells and germ cells. **Germ cells** are involved in the formation of gametes. Body cells, or **somatic cells**, include all other cells of the body. Mutations happen in both of these cell types, but only mutations in germ cells may be passed from parent to offspring. Mutations in the germ line affect the phenotype of offspring. Often, this effect is so harmful that offspring do not develop properly or die before they can reproduce. Other mutations, though less severe, often still result in less adaptive phenotypes. More rarely, a mutation results in a more beneficial phenotype.

FIGURE 15: Mutations can occur in gametes and in body cells.



Explain Would a mutation in one of your muscle cells be passed down to your offspring? Use evidence to support your explanation.

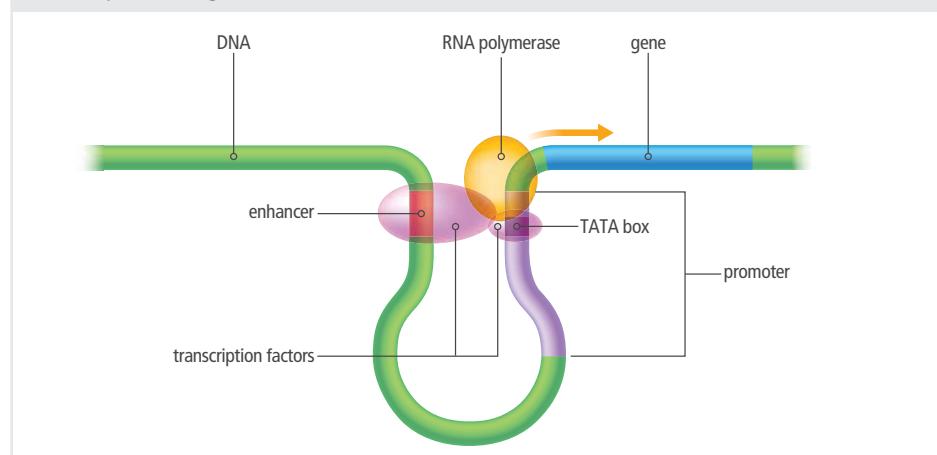
Impacts on Phenotype

Chromosomal mutations affect many genes and have a major impact on the organism. A mutation may break up a gene, inactivating it, or make a new hybrid gene with a new function. Translocated genes may also come under the control of new promoters.

 **Collaborate** How might a mutation that affects a regulatory element, such as a promoter, transcription factor, or enhancer, affect the expression of a gene? Discuss possible outcomes of mutations affecting each of these elements. Would the gene be expressed? If so, how might its expression change?

Gene mutations, though smaller in scale than chromosomal mutations, can also have a big effect on an organism. Even a mutation in a noncoding region can cause problems. Recall that DNA sequences such as promoters and enhancers interact with transcription factors and RNA polymerase to start transcription. Therefore, a mutation that affects any one of these elements could also affect the expression of one or more genes.

FIGURE 16: A promoter is a segment of DNA that binds to proteins that help initiate the transcription of a gene.



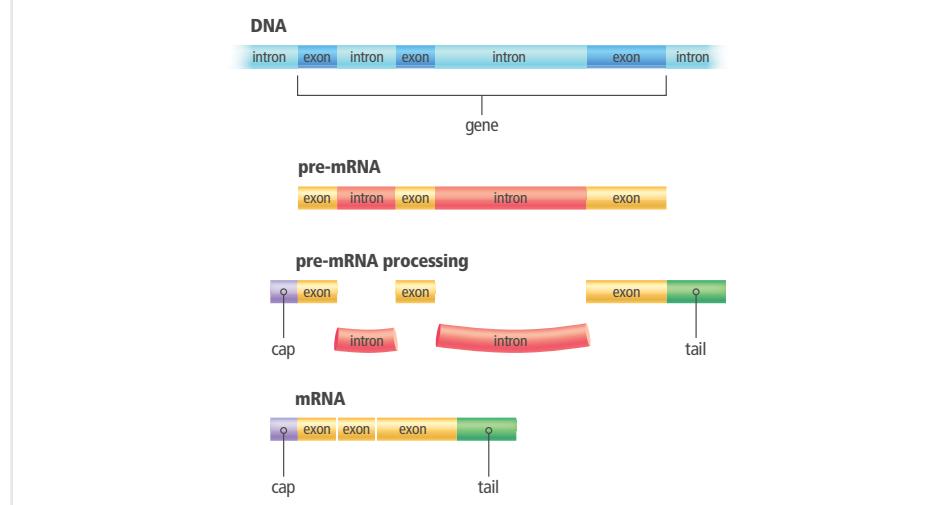
Many gene mutations, however, do not affect an organism's phenotype. Remember that many codons code for the same amino acid. Therefore, some substitutions have no effect, especially those occurring in the third nucleotide of a codon. If AAG changes to AAA, the resulting protein still has the correct amino acid, lysine. Similarly, an incorrect amino acid might have little effect on a protein if it has about the same size or polarity as the original amino acid or if it is far from an active site.

Cause and Effect



Would a mutation in an intron affect the structure and function of the resulting protein? Explain your answer.

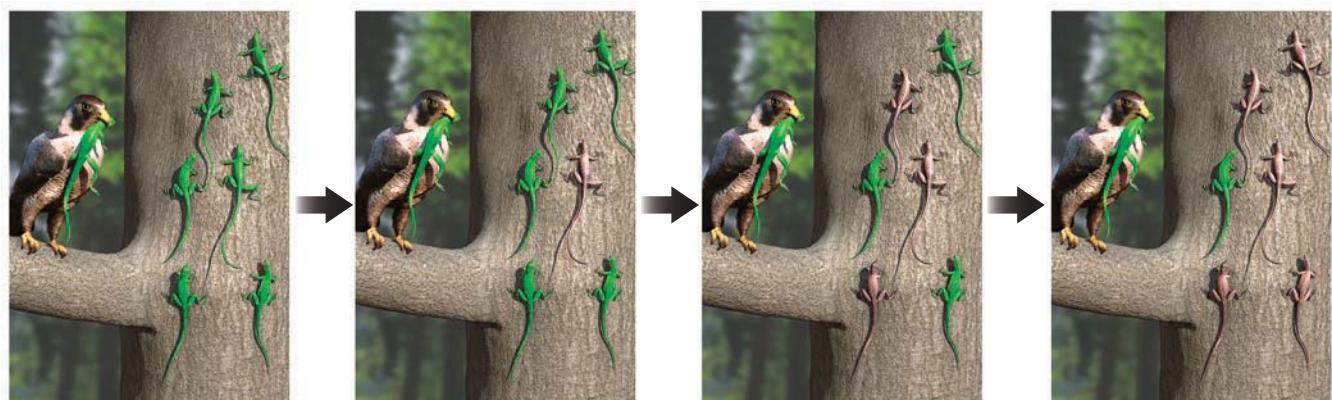
FIGURE 17: In mRNA processing, introns are removed from the pre-mRNA strand.



Impacts on Genetic Diversity

Genetic variation, or genetic diversity, is the variety of genes within a population. While genetic recombination via sexual reproduction is a major source of genetic diversity, mutations in germ cells are the ultimate source of genetic diversity in an organism's genome. Genetic diversity is the basis of a process called natural selection. In natural selection, environmental factors "select for" phenotypes that allow organisms to better survive and reproduce. For example, an individual might have a phenotype that allows the organism to attract more mates than other individuals. This individual would have more opportunities to pass down their genes, and over the course of many generations, this phenotype could become more prevalent in the population.

FIGURE 18: Mutations increase genetic diversity, which is the basis of natural selection.



Analyze Assume that in these lizards, the brown phenotype results from a mutation. Why does this phenotype become more common in the population over time?

When less adaptive phenotypes result from mutations, natural selection typically removes these mutant alleles from the population. Less adaptive phenotypes may make it difficult for organisms to survive or reproduce. These traits are "selected against" by environmental factors and tend to become less prevalent in a population over time.



FIGURE 19: A mutation in humans has been shown to protect against coronary artery disease.

Sometimes, a mutation results in a more beneficial phenotype. These mutations are favored by natural selection. For example, one type of deletion mutation in humans has been shown to protect against coronary artery disease, a condition characterized by the hardening and thickening of artery walls.



Explain Two species of rabbits occupy an area that experiences four seasons. The first type of rabbit has white fur in the winter and brown fur in the spring. The other species has brown fur all year round. Which of these types of rabbits has a more beneficial phenotype? Explain your answer.



Explain In some cases, mutations that have some harmful effects continue to persist in certain human populations. Why might a mutation with detrimental effects persist in a given population?



Engineering

FIGURE 20: UV-protective clothing protects the wearer from skin damage due to radiation from the sun.



MATERIALS

- fabric (3 types)
- plates, paper
- UV beads or paper
- UV light box or sunlight



Testing UV-Protective Fabrics

When our body is exposed to moderate levels of radiation from sunlight, it may respond by tanning or burning. The exposure activates the production and release of a brown pigment called melanin. This pigment acts like a natural sunscreen by helping block *ultraviolet (UV)* light, an invisible type of radiation present in sunlight. Recall that UV light is a mutagen that can damage skin tissues. Prolonged exposure to UV light can lead to skin cancer caused by mutations in the DNA of skin cells. The most common type of damage is the formation of thymine dimers, or pairs of thymine bases bonded together in DNA. These mutations interfere with both replication and translation.

UV-protective clothing is designed to protect people from UV light. In this activity, you will work with your classmates to develop a testing system that could be used to identify fabrics that can be used for UV-protective clothing. The system should be easy to use and cost less than one hundred dollars. The testing system should also allow the user to test up to 100 pieces of fabric in an eight hour work day. Finally, the system should be lightweight and portable, so that one person can carry it.

DEFINE AND DELIMIT

Write a statement identifying the problem you are designing a solution for. What are the criteria and constraints for an effective testing system?

DESIGN

As a team, brainstorm some possible solutions to the problem. Make a decision matrix to choose the solution that best meets the criteria. Once you have chosen a potential solution, make a prototype of your testing system.

TEST

Conduct a test to gather data showing how well your testing system works. If your solution does not fully meet the criteria, return to your design. Continue developing and testing solutions until you feel certain that your solution meets the most important criteria and constraints.

COMMUNICATE

Write an explanation communicating your results. Which type of system is best for testing these fabrics? Give evidence to support your explanation. Include a diagram of the final solution your team selected.

p53: THE TUMOR SUPPRESSOR GENE

MUTATIONS AND HUMAN HEALTH

INVESTIGATING MELANISM

Go online to choose one of these other paths.

Lesson Self-Check

CAN YOU EXPLAIN IT?

FIGURE 21: “Sickling” of red blood cells occurs when deoxygenated HbS molecules form long chains, or polymers. These polymers force the cell to change shape.



The allele for sickle cell anemia is linked with resistance to malaria, a disease caused by a parasite transmitted from one infected person to another by mosquitoes. Individuals who have this disease may experience swelling of the brain, difficulty in breathing, liver and kidney failure, anemia, and low blood sugar. Although modern medical techniques can diagnose and cure malaria through early treatment, if untreated, the complications of malaria can lead to death.

Individuals who have malaria, but are also carriers of the sickle cell anemia gene (HbS) have been observed to not advance to the serious stage of malaria. Thus, in the absence of modern medical treatment, having one of these genes helps protect them from the fatal consequences of malaria. According to the Centers for Disease Control and Prevention, HbS can provide 60% protection against malaria.



Explain Why is the HbS allele more common in some populations than in others?

Answer the following questions in your explanation.

1. How do changes in DNA lead to changes in the structure of red blood cells in people with the HbS allele?
2. Is the phenotype that corresponds to the HbS allele harmful, beneficial, or both? Explain your answer.
3. Why is the frequency of the HbS allele higher in areas near Earth’s equator, such as parts of Africa and the Mediterranean?

CHECKPOINTS

Check Your Understanding

- 1.** The results of a study on the effects of a mutagen on bacteria had the following results. Some bacterial cultures were exposed to the mutagen, some were not. Which culture was most likely exposed to the mutagen?

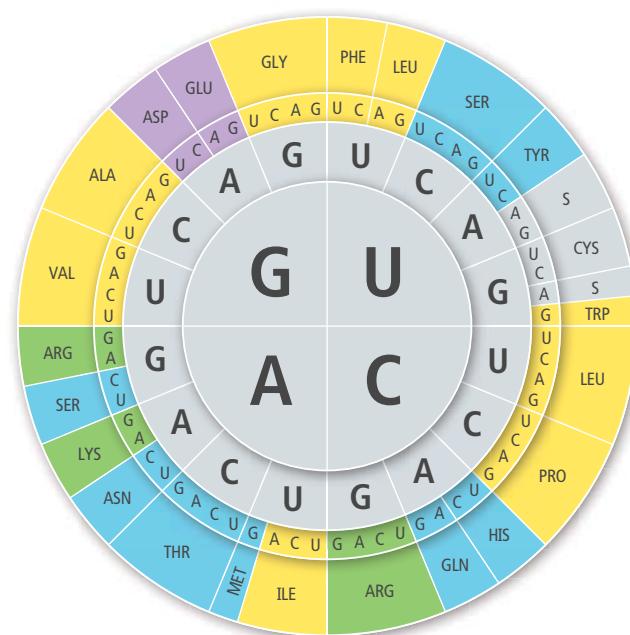
Culture	Number of mutant bacteria
A	0
B	350
C	10
D	4
E	3

- 2.** Epidermolysis bullosa is a disease characterized by very delicate skin that easily blisters upon scratching or being exposed to the slightest friction. The disease is caused by a missense mutation. Which statement describes the mutation that causes epidermolysis bullosa?
- a.** The mutation is a result of the premature completion of a protein.
 - b.** The mutation is caused by a change in one of the amino acids.
 - c.** This mutation is a result of the reading frame being shifted.
 - d.** This mutation is caused by the duplication of the genome.
- 3.** Before the genetic code could be understood, scientists needed to know that a codon is composed of three nucleotides. This situation is an example of the
- a.** cumulative nature of scientific evidence.
 - b.** scientists making inferences based on data.
 - c.** way that theories can lead to scientific laws.
 - d.** ability of scientists to make hypotheses.

- 4.** Individuals with trisomy X have three X chromosomes in their cells. Which statement can be used to describe this condition? Select all correct answers.
- a.** This condition is caused by a chromosomal mutation known as nondisjunction mutation.
 - b.** This condition is a result of the exchange of genetic material between two homologous chromosomes.
 - c.** This mutation is a result of chromosomes not separating during anaphase of mitosis.
 - d.** This mutation is caused by balanced translocation, a type of chromosomal mutation.
- 5.** Which of the following can be changed during meiosis? Select all correct answers.
- a.** base sequence
 - b.** number of amino acids
 - c.** number of chromosomes
 - d.** gene sequence
- 6.** Which processes are involved in the inheritance of mutated genes? Select all correct answers.
- a.** meiosis
 - b.** fertilization
 - c.** mitosis
- 7.** Watermelons are exposed to a mutagen to produce a variety that has four sets of chromosomes. The new variety is then allowed to mate with a normal watermelon to produce seedless watermelons. What type of mutation is involved in the growing of seedless watermelon?
- 8.** Rachel Carlson was one of the first ecologists to warn against the widespread use of pesticides and other potential mutagens and toxins. How might the presence of a chemical mutagen in the environment affect the genetic makeup and size of a population over time?

MAKE YOUR OWN STUDY GUIDE

FIGURE 22: Codon Chart



ALA = Alanine

GLY = Glycine

PRO = Proline

ARG = Arginine

HIS = Histidine

S = Stop

ASN = Asparagine

ILE = Isoleucine

SER = Serine

ASP = Aspartic acid

LEU = Leucine

THR = Tryptophan

CYS = Cysteine

LYS = Lysine

TYR = Tyrosine

GLN = Glutamine

MET = Methionine

VAL = Valine

GLU = Glutamine acid

PHE = Phenylalanine



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

A mutation is a change in the sequence of an organism's DNA, and may occur spontaneously or as the result of exposure to a mutagen.

Mutations contribute to genetic diversity because as the genetic makeup of organisms are changed through mutations, variety is produced.

Mutations may or may not affect an organism's phenotype.

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 mutations can lead to changes in DNA at both the cellular and organismal levels, and develop an explanation as to how each of these types of changes may or may not lead to changes in phenotype in real-world situations.

9. Consider this small part of a DNA sequence:

GTG–GAC–TGA–GGA

Use this sequence and the codon chart in Figure 22 to make a model showing how a frameshift mutation happens and how the amino acid sequence is affected.

10. Huntington's disease affects how the brain functions.

Individuals who have this disease are not able to control the movement of their bodies. Additionally, they experience emotional problems and loss of cognitive ability. This disease is caused by a trinucleotide repeat involving CAG. What causes this mutation and how does it affect the protein synthesized by the mutated gene?

11. Can a parent pass on a mutation in a kidney cell to his child? Why or why not?

12. Explain how mutations contribute to genetic diversity.

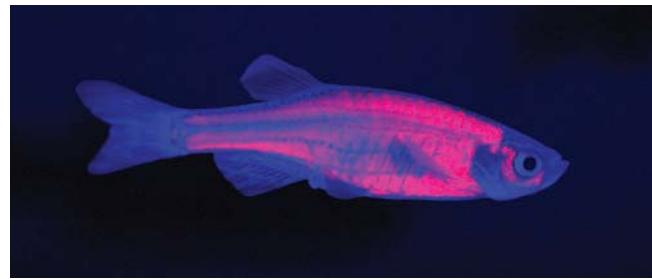
Genetic Engineering

Aspen trees can reproduce by sprouting new, identical trees from their roots.

CAN YOU EXPLAIN IT?

Many organisms, such as jellyfish, fluoresce when exposed to particular wavelengths of light. Fluorescence occurs when an organism absorbs light and then emits it, appearing to glow. Fluorescent zebrafish can be purchased to enjoy in your aquarium. There's just one catch: zebrafish don't naturally fluoresce.

FIGURE 1: Zebrafish were genetically altered to fluoresce in many colors.



Gather Evidence

As you explore the lesson, gather evidence to explain how a gene from one organism can be inserted into the genome of an unrelated organism?

Fluorescent zebrafish are the result of decades of scientific research. Researchers at the National University of Singapore studying the green fluorescent protein (GFP) that causes fluorescence in jellyfish inserted the GFP-coding gene into zebrafish, resulting in a zebrafish that emitted green light.

The U.S. Food and Drug Administration (FDA) approved the sale of fluorescent zebrafish as pets in the United States. The FDA decided not to regulate the altered zebrafish because they were not intended to be part of the food supply. In addition, there was no research to suggest that the fluorescent strains would be more harmful to the environment than the original strains in case of accidental release.



Predict What does it mean to change the genome of an organism? Is this fluorescent zebrafish a new type of animal?

Isolating Genes

Huntington's disease causes nerve cells in the brain to break down. The onset of Huntington's often begins midlife, with no physical hints of the disease before symptoms arise. For those who have a parent with Huntington's disease, a Punnett square or pedigree analysis may provide a probability of having the disease, but not a definitive diagnosis. For Huntington's and many other diseases, genetic material can be tested to determine whether a person has, or is a carrier of, a specific disease.



Gather Evidence Would you undergo tests to determine your likelihood of having certain diseases? Why or why not? If you did, what would you want to happen to your genetic information? Should it be shared with scientific researchers, your health insurer, or your future employers? Explain your reasoning.

Genetic Testing

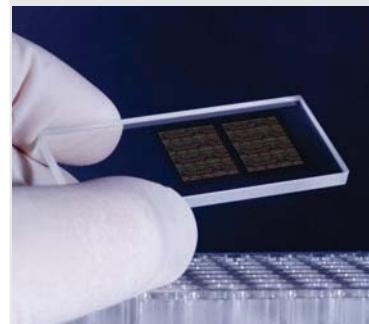
Genetic testing is the analysis of a person's DNA to determine the risk of having or passing on a genetic disorder. Geneticists test for abnormalities in genetic material, from entire chromosomes down to individual genes. It is also possible to test for proteins that indicate a particular disease. Since proteins reflect the DNA patterns of genes, this is an indirect method of testing genetic material. Genetic testing is a powerful tool to screen for genetic disorders. However, not all diseases can be found through genetic testing.



Analyze Why can't genetic testing identify all diseases? How does inheriting cystic fibrosis differ from developing cardiovascular disease due to poor diet and exercise?

There are thousands of genetic tests available, each targeting a specific gene or genomic region. DNA microarrays are tools that allow scientists to study many genes, or their expression, at once. A microarray is a small chip that is dotted with all of the genes being studied. The genes are laid out in a grid pattern. Each block of the grid is so small that a one-square-inch chip can hold thousands of genes. Microarrays, such as the one shown in Figure 2, help researchers find which genes are expressed in which tissues, and under what conditions.

FIGURE 2: DNA microarrays are used in genetic testing.



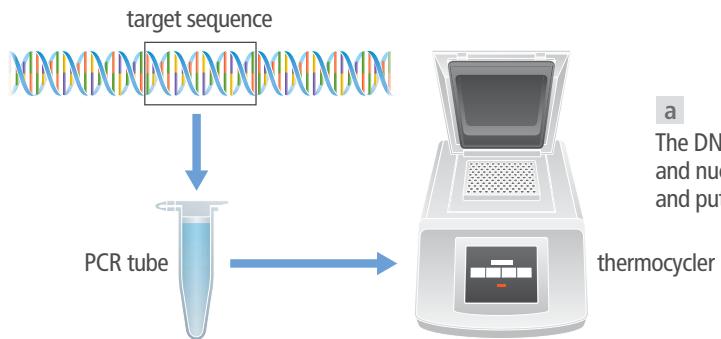
Collaborate In a group, discuss the benefits, risks, and limitations of genetic testing. Why is it important to identify carriers of a genetic disease? How should genetic information be used and safeguarded?

Replicating Genes

Genetic tests are useful for genes that have been linked to a disease, but identifying specific genes that cause disease is not simple. Scientists spend years finding genes that are associated with a particular disease among the 20,000–25,000 genes in the human genome. Small quantities of target sequences collected from patients must be amplified many times to produce the amount needed for testing. The invention of the **polymerase chain reaction (PCR)** was a turning point, making it possible to obtain the large amounts of DNA needed for genetic testing in hours instead of days.

FIGURE 3: The steps of the polymerase chain reaction.

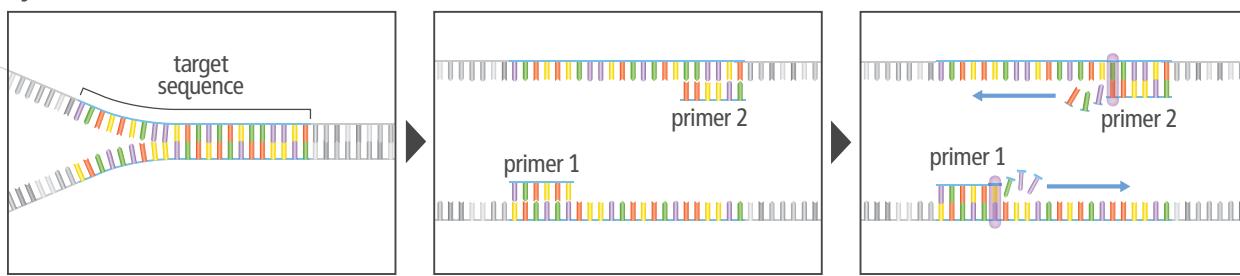
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a

The DNA sample, primers, DNA polymerase, and nucleotides are placed in the PCR tube and put in the thermocycler.

Cycle 1



b Separating

The temperature is raised to 95 °C (203 °F) to separate the DNA strands.

c Binding

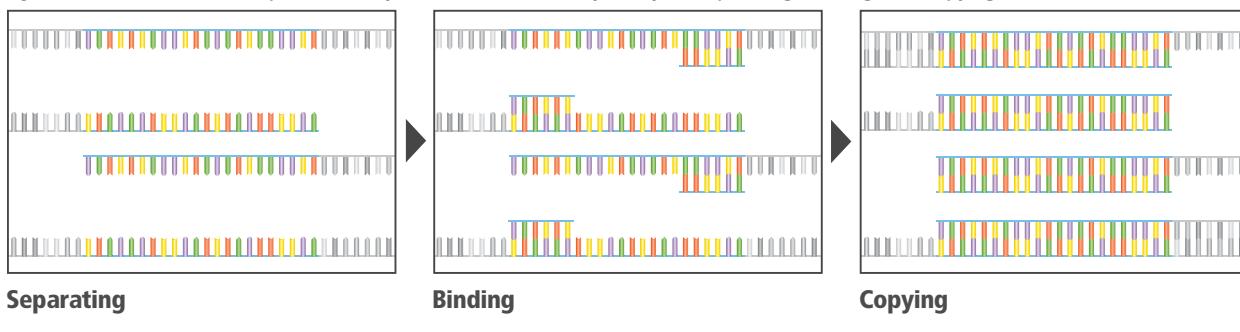
The temperature is cooled to 55 °C (131 °F), and the primers bind to the DNA strands.

d Copying

The temperature is heated to 72 °C (152 °F). DNA polymerase locates the primers and begins synthesizing a complementary strand. It continues to synthesize the DNA strand until it reaches the end of the strand.

Cycle 2

The same three steps occur in Cycle 2 and each subsequent cycle: separating, binding, and copying.



Separating

Binding

Copying

Cycle 3



At the end of Cycle 3, fragments that include only the target DNA have been synthesized.

Cycle 4

By the end of Cycle 4, eight target fragments have been synthesized.

Cycle 30

After 30 cycles, more than a billion fragments have been synthesized.

Figure 3a shows the beginning of a PCR run. DNA is extracted from cell nuclei and added to a PCR tube, along with primers, DNA polymerase, and nucleotides. The tube is placed inside a thermocycler, which automatically regulates the temperature of the solution.

The polymerase chain reaction occurs in three steps:

Separating The thermocycler heats the sample until the complementary strands of DNA separate (Figure 3b). Separation occurs around 95 °C (203 °F).

Binding The thermocycler then cools to around 55 °C (131 °F) (Figure 3c), and primers bind to the separated DNA strands. Primers are short nucleotide segments that allow a specific type of DNA polymerase to attach to the DNA strands. Two primers are required for each reaction. One primer attaches to the beginning of the target segment on one strand of DNA. The other primer attaches to the beginning of the target segment on the complementary strand of DNA.

Copying The thermocycler heats to 72 °C (162 °F) (Figure 3d). At this temperature, DNA polymerase attaches to the primer segments and begins adding complementary nucleotides. The free nucleotides added to the solution act as building materials for the new strands of DNA. DNA synthesis continues until the DNA polymerase reaches the end of the strand and detaches. A complementary strand of DNA is produced, and the first PCR cycle is complete.



Collaborate With a partner, take turns explaining and modeling how the three steps of PCR produce DNA sequences. While you walk your partner through the steps, explain the significance of the following terms: *DNA polymerase*, *nucleotides*, *primers*, *DNA separation*, *primer binding*, *DNA synthesis*, and *thermocycler*. Then, your partner explains and models the process to you. Continue to take turns until both of you feel comfortable with the steps of PCR.

The cycle is repeated a second time. The thermocycler heats to 95 °C and the DNA strands separate. The thermocycler cools to 55 °C and primers bind to the target sites. Finally, the thermocycler heats to 72 °C. DNA polymerase attaches to primer segments and synthesizes a complementary strand of DNA using the free nucleotides.

The thermocycler continues to heat and cool the solution automatically. The first fragment of the target DNA sequence is synthesized after the third cycle. More than one billion fragments of target DNA are synthesized after thirty cycles. PCR cycles continue until an adequate amount of the target DNA is produced.



Analyze Why is it necessary to keep changing the temperature in the PCR process? Use evidence to support your claim.

The polymerase chain reaction was invented by Kary Mullis in 1983, who shared the Nobel Prize in Chemistry in 1993. This invention solved two problems Mullis was facing. First, his lab was trying to create a new use for the oligonucleotides, or short DNA segments, they produced. PCR uses oligonucleotides as primers. Second, genetic testing and other DNA-related tests took weeks to perform. PCR greatly decreased the time required to amplify a DNA sample.



Explain Describe the relationship between genetic testing and the polymerase chain reaction. How has the PCR technique made genetic testing possible on a large scale?



Patterns

DNA replication produces a complementary strand of DNA, while PCR amplifies a target section of DNA by copying just that section. How else are DNA replication and PCR similar?
How are they different?

Cloning and Engineering

FIGURE 4: These cereal plants can grow in soil with little water.



As the world's population increases, so does the demand for food. Long periods of drought in many areas of the world threaten food production because many commercial crops are not adapted to dry climates. To maintain food production as land becomes drier, scientists engineered plants that are drought resistant.



Gather Evidence Other strategies for growing food in dry climates include water conservation, sustainable farming practices, and improved fertilizers. Make a list of possible criteria for evaluating drought-resistant crops along with the other solutions.

FIGURE 5: Some plants produce "pups," or genetically identical offspring.



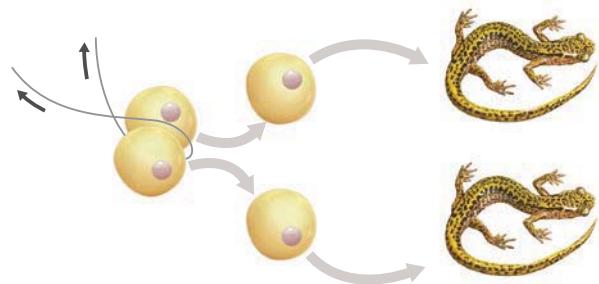
Cloning Organisms

Many plants produce genetically identical offspring, or **clones**, through asexual reproduction. Humans have cloned plants for thousands of years by taking cuttings from one plant and planting them, producing clones. When the offspring, or "pup," of a spider plant, shown in Figure 5, is planted, a genetically identical plant grows. Humans clone plants with desirable traits, such as bigger or more flavorful fruit. Eventually these traits appear more often in the new population.

Bacteria produce clones through binary fission, a type of asexual reproduction. In binary fission, a bacterial chromosome is replicated. The cell splits into two daughter cells that are genetically identical to the mother cell. Making clones ensures beneficial traits, such as resistance to antibiotics, spread quickly in a bacterial population.

Cloning has a low success rate in more complex organisms, such as vertebrate animals. Advances in genetic engineering, though have made it possible to produce artificial mammalian clones. The sections below describe breakthroughs in cloning.

FIGURE 6: The embryo twinning process.



Embryo Twinning

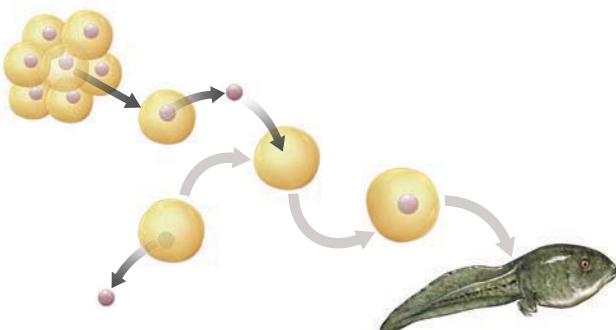
In 1903, Hans Spemann separated the cells of two-celled salamander embryos. The separated cells continued to develop normally, resulting in two salamanders (Figure 6). Spemann determined that vertebrates can be "twinned" to form identical organisms. This experiment showed that embryonic cells have a full set of genetic material. So, each cell has the potential to grow into a complete organism.

Nuclear Transfer

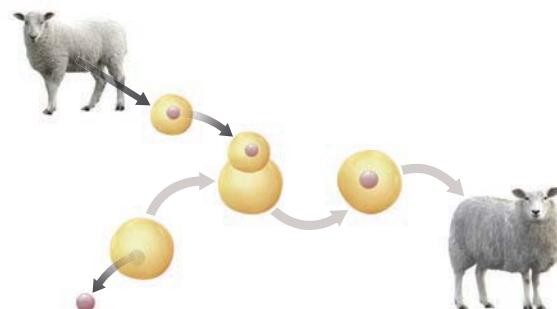
Cloning mammals involves replacing the nucleus of an unfertilized egg with the nucleus of a cell from the animal that is being cloned. The egg cell is implanted into a surrogate mother to develop as it would during a normal pregnancy. The resulting offspring is a clone. Some of the milestones in nuclear transfer are shown in Figure 7.

FIGURE 7: Milestones in the advancement of cloning techniques.

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a Embryonic cell nuclear transfer



b Somatic cell nuclear transfer

In 1952, Robert Briggs and Thomas King performed the first successful nuclear transfer (Figure 7a). The nucleus from an embryonic frog cell was inserted into an egg cell with its nucleus removed. The egg cell then developed into a tadpole. This experiment demonstrated that nuclear transfer could be used to clone organisms.

Scientists later adapted nuclear transfer methods to produce clones of other animals, including mammals. Further research led to new techniques which allowed the use of other cell types as nuclear donors, eliminating the need to use embryos.

In 1996, Dolly the sheep became the first mammal cloned from an adult somatic, or body, cell (Figure 7b). Somatic cells are differentiated, so many genes not necessary for the cell's function are deactivated. These genes must be reactivated for cloning to succeed. Of 277 attempts in this experiment, only Dolly survived.

Cloning After Dolly

Milestones in cloning after Dolly include cloning primates, producing sheep from genetically engineered cells, cloning endangered animals, and creating stem cells from somatic cell nuclear transfer. New advances in cloning have raised ethical concerns, such as concerns regarding human cloning.

Pet cloning is one of these advancements. Several companies offer cloning services that will produce an exact genetic copy of a pet. Though they are genetically identical, these animals often look and act differently than the original pet.



Gather Evidence Why is a clone not an exact copy of a donor animal? Consider the effect of genetics and environmental conditions. Use evidence to support your answer.

FIGURE 8: A cloned puppy with the genetic father.



Cloning Ethics

Henrietta Lacks died of cervical cancer in 1951. Before she died, a researcher took a sample of her tumor. From this sample, scientists made the first "immortal" cell line, named HeLa for the first two letters of Henrietta's first and last names. Unlike other cells, HeLa cells did not die when cultured in the lab. The cells divided indefinitely, providing a never-ending source of cells for scientific research. From the polio vaccine and cloning to AIDS research and experiments in space, HeLa cells have been a cornerstone of science for more than half a century.

FIGURE 9: Henrietta Lacks



Most of this research took place without the knowledge or permission of Henrietta Lacks or her family. This raises the issue of cloning ethics. Ethics are principles that set standards of right or wrong for a person or group. As advances in genetics continue, discussions about ethics and treatment of genetic material become more important.



Language Arts Connection Further research the story of Henrietta Lacks.

Should individuals have control over their genetic material? How would you feel if your genetic material was taken without permission? Use evidence to support your claims.

Engineering Genes

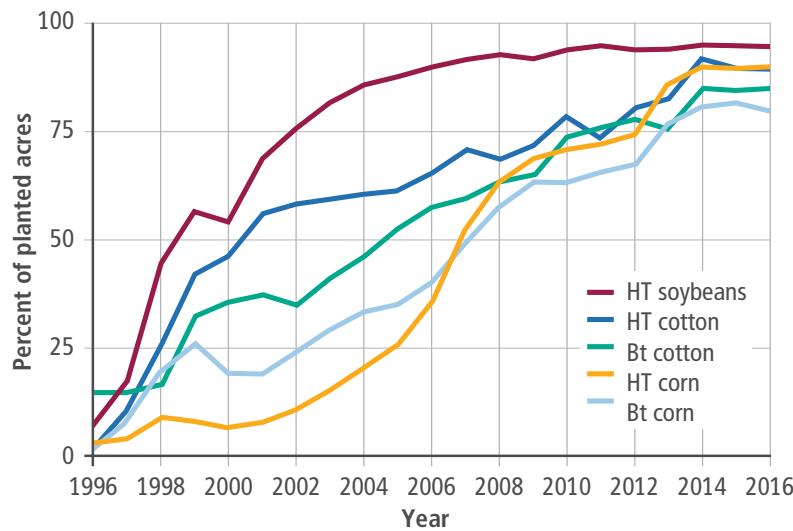
Genetic engineering is the process of altering the genetic material of an organism, changing its traits or introducing a new, desirable trait. Once a desirable trait has been successfully inserted into a genome, the new genome—and trait—can be passed on to future generations using cloning. An organism with one or more genes from another organism inserted into its genome is called a **transgenic** organism.

Genetically modified (GM) crops are becoming more widely used by farmers. If a farmer plants clones of GM crops, then he or she knows the desired trait is present in the entire population. However, this would also decrease genetic diversity, a necessary feature for a robust and flexible population.

Adoption of Genetically Engineered Crops in the U.S., 1996-2016

FIGURE 10: The usage of genetically engineered crops in the United States.

Analyze Compare the risks versus the benefits of using cloned, GM plants instead of GM plants propagated through sexual reproduction.



Sources: USDA, Economic Research Service using data from Fernandez-Comejo and McBride (2002) for the years 1996–99 and USDA, National Agricultural Statistics Service, June Agricultural Survey for the years 2000–16.

In the early 1990s, the FDA approved genetically engineered plants for human consumption in the United States. Insect resistance and herbicide resistance are among the most common genetic modifications in crops, as shown in Figure 12. Much of the genetically modified corn produced is fed to livestock, but GM corn does appear in the human food supply as ingredients such as high-fructose corn syrup and corn starch. No long-term studies have found negative side effects from eating GM plants.

Genetic Engineering in Bacteria

Recombinant DNA technology, combining the genes from more than one organism, is a key element of genetic engineering. The organisms can be from the same species or different species. One method of producing recombinant DNA is to add foreign DNA to a plasmid. In bacteria, a plasmid is a small, circular segment of DNA that is separate from the bacterial chromosome. The foreign DNA that is inserted into the plasmid is then expressed by the bacteria.

Bacteria naturally recombine their DNA by absorbing plasmids from the environment or by exchanging plasmids between two bacteria. There can be multiple plasmids within a bacterium, and each one is able to replicate independently from the bacterial chromosome. Genetically modified bacteria are able to produce antibiotics, insulin, therapeutic proteins, and other types of proteins.

Imagine foreign DNA containing a gene for producing human insulin is inserted into a plasmid. Because plasmids self-replicate, numerous copies of a plasmid can exist within a bacterium. Plasmids are shared with daughter cells during binary fission, and bacteria divide at relatively fast speeds. A handful of bacteria with a plasmid coding for human insulin can quickly become a manufacturing center for a protein.



Collaborate Genetically engineering bacteria to produce drugs can be cheaper than producing the drugs in a lab. Discuss the impacts cheaper drugs may have on society and science.



Engineering

Editing Genes with CRISPR

Genetically engineering organisms requires the ability to cut DNA strands in specific places. Precisely cutting DNA can be difficult, time-consuming, and costly work. To solve this problem, genetic engineers needed to find an easier, faster, and cheaper method for precisely cutting DNA.

As it turns out, bacteria use a mechanism for precise DNA cuts called CRISPR, named for the clustered regularly interspaced palindromic repeats (CRISPRs) in bacterial DNA. These repeated sequences surround segments of viral DNA that bacteria have been exposed to. An enzyme uses the information in this viral library to target and cut viral DNA, preventing viral replication.

CRISPR is exciting for genetic engineers because it provides a very precise method for cutting DNA at a specific point. Cutting DNA easily and accurately simplifies the process of replacing defective genes with functional genes. This is one of the more difficult tasks in gene therapy, but one with the greatest potential benefits to humans. New ways to apply the CRISPR system to scientific problems are still being discovered. As with most genetic advances, the excitement surrounding the prospective benefits of CRISPR is tempered by the ethical concerns raised by such a powerful gene-editing tool.



Gather Evidence In what ways do you think CRISPR can advance the field of genetic engineering? What concerns do you think people might have about CRISPR?

FIGURE 11: Bacterial plasmid

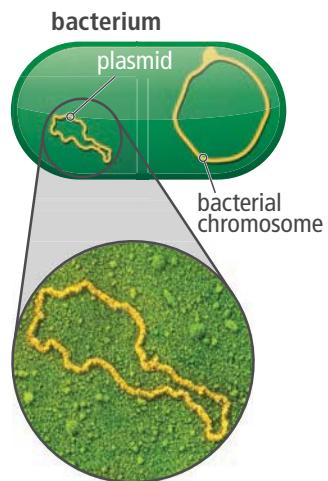


FIGURE 12: Recombinant DNA.



Genetic Engineering in Plants

One of the most common methods for genetic modification in plants is the use of bacterial plasmids. A gene for a desired trait is inserted into a plasmid, and the plasmid is added to a plant cell. When the plant cell is infected, the recombinant DNA is inserted directly into the plant genome, modifying the plant. The plant expresses the bacterial DNA as well as its own.

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Hands-On Lab



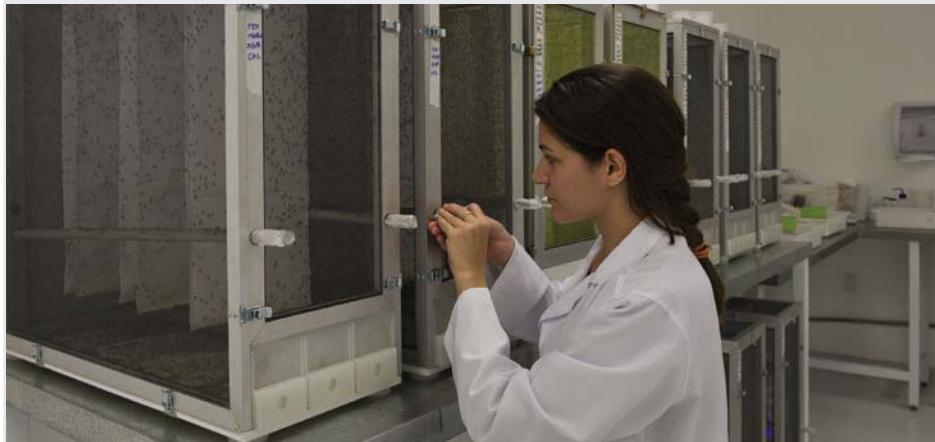
Modeling Genetic Engineering

Simulate the techniques used by genetic engineers to modify genes in humans using recombinant DNA technology.

Genetic Engineering in Animals

Animal models of human diseases are valuable tools in medical research. These models allow scientists to study the disease process, from the genetic basis of a disease to how it responds to chemical substances. Through genetic engineering, scientists have been able to develop more and better models to study disease.

FIGURE 13: A scientist studies genetically modified mosquitoes.



 **Model** Draw a flow chart that demonstrates how genetically engineering mosquitoes can reduce the risk of illness in humans.

Consider the use of genetically modified mosquitoes to prevent the spread of disease. Mosquitoes act as vectors for many diseases. A vector carries foreign DNA into another cell or organism. One species, *Aedes aegypti*, is known to transmit the viruses for yellow fever, chikungunya, dengue, and Zika. Dengue is one of the leading causes of illness and death in tropical and subtropical regions. There is no vaccine for dengue, and the best way to minimize dengue cases is to minimize bites from infected mosquitoes.

To solve this problem, scientists engineered mosquitoes so they required a human-made drug to survive. When modified male mosquitoes are released into wild populations, they breed with wild females, passing the drug-dependency gene to their offspring. The affected males die soon after breeding, and any offspring die before maturity without access to the drug. Several field trials demonstrated that release of mosquitoes modified in this way can effectively control mosquito populations.

The possibility of unintended effects is a big constraint to this solution. The potential unintended effects of releasing genetically engineered mosquitoes into the wild is not fully understood. There may be tradeoffs for scientists and society between the risks of unintended effects and the benefits of smaller mosquito populations.



Explain There typically are tradeoffs when selecting a solution to a problem.

Genetically engineered crops may be able to help farmers produce greater yields, but a tradeoff is the reduction in the genetic variation in crops, making crops more susceptible to disease. What other tradeoffs exist for this solution?



Engineering

Genetically Engineering Salmon

Demand for Atlantic salmon has increased, and wild populations of salmon have decreased, mainly due to overfishing and other environmental impacts. Struggling wild populations suggest that commercial fishing is not sustainable at current rates. Farm-raised Atlantic salmon is an alternative to wild-caught fish and reduces pressure on wild populations by providing a reliable salmon source. Salmon can be farmed in ocean pens or in land-based facilities. Some farm-raised salmon are bred for advantageous traits, such as disease resistance.

Farming has drawbacks, though. Farms require space and resources to feed, house, and maintain the fish as they grow. The salmon typically take 28–36 months to reach market weight, and production costs can drive up the price.



Analyze Define the engineering problem outlined for salmon production. What are the important criteria? What constraints might exist for a solution that reduces the stress on wild-caught salmon and on farmers raising these fish?

To solve some of the problems with farming salmon, scientists produced genetically engineered Atlantic salmon. They inserted a growth hormone from Chinook salmon and a promoter from ocean pout into the genome. The promoter allows the growth hormone to be active all year, instead of only part of the year as in normal salmon.

Transgenic Atlantic salmon grow to twice the size of normal Atlantic salmon in the same amount of time. This decreases the time to market weight to as few as 18 months, compared to up to 36 months in normal salmon. So, farmers are able to grow and sell more salmon in a given time period. There are also environmental benefits such as decreased usage and contamination of water resources. The genetically modified salmon are raised in land-based facilities with pollution management and water recycling systems. Genetically modified salmon reduce the impact on wild populations and aquatic ecosystems.

There is still public resistance to eating genetically modified organisms. This represents a social challenge to the success of farming genetically engineered salmon. One of the biggest environmental concerns is the possibility that a GM individual may escape and breed with wild individuals. This could introduce the modified gene into wild populations through any offspring produced, with unknown long-term effects on wild Atlantic salmon or other species.



Explain Design a decision matrix and use it to analyze criteria for the use of commercial fishing, normal salmon farming, and GM salmon farming in meeting the demand for salmon. Weight the criteria on a scale of 0 to 5. What is the best solution based on your criteria? Are there any problems with this solution that can be anticipated and avoided?

FIGURE 14: A normal Atlantic salmon compared to a GM Atlantic salmon of the same age.



Further Applications of Genetic Engineering



Collaborate With a partner, discuss the benefits and risks of transgenic mosquitoes for humans and ecosystems.

New technologies can have unforeseen impacts on society and the environment. The positive effects of controlling mosquito populations with transgenic mosquitoes are clear: reduced illness and death due to infections from mosquito-borne viruses. There are also negative effects to this solution, though, that may be hard to believe. Mosquitoes may be pests for humans, but they are a food source for other animals.

Impacts on Conservation

In the future, ecosystems may undergo rapid change due to climate change, habitat destruction, and human influence. Populations may be forced to adapt or move to new habitats to survive. This is a problem because natural selection, the mechanism by which populations adapt, is not a rapid process and works over many generations. Scientists are looking for ways to help threatened species.

FIGURE 15: The ‘i’iwi.



Hawaii had no mosquitoes until the early 1800s when a whaling vessel carrying water from Mexico brought them to the islands. Today, avian malaria, carried by these invasive mosquitoes, has decimated the native bird population. The ‘i’iwi, or Hawaiian honeycreeper, and other birds native to Hawaii are going extinct. Many scientists think the only way to save these birds is to wipe out the mosquito population. Scientists are considering releasing GM mosquitoes that will die prematurely, reducing the mosquito population and hopefully saving Hawaii’s native birds.

For species threatened by climate change or low genetic diversity, scientists are investigating a process known as **facilitated adaptation**. Facilitated adaptation involves humans

guiding adaptations in threatened populations by changing the species’ genome. Advantageous genes can be added to a genome through hybridization, selective breeding, or genetic engineering using recombinant DNA technology. For example, scientists are considering inserting genes from species that can tolerate higher temperatures into different species suffering from global warming.

One drawback of facilitated adaptation is the possibility of unintended effects related to changing genomes that have evolved over millions of years. Scientists may be able to identify the main function of a gene, but they cannot determine all the ways a gene interacts with the rest of the genome. Loss of function, or an unintended new function, may occur by changing an organism’s genome. Facilitated adaptation could also lead to an unintended loss of genetic diversity. If the genetically engineered individuals are much more successful than normal individuals, that single gene could become widespread in the population.



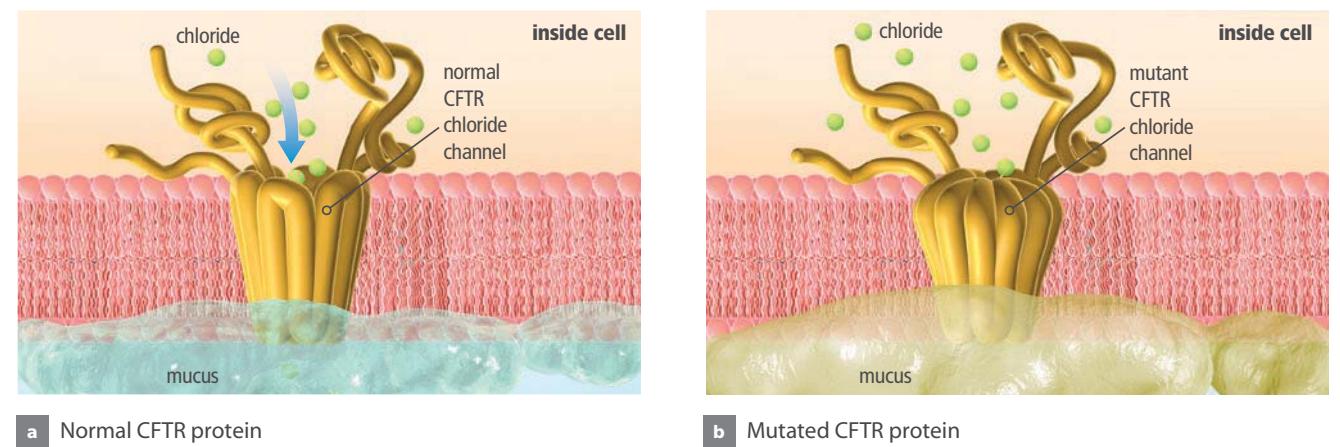
Engineering Define a problem facing conservation. Explain what role genetic engineering could play in solving that problem. Use evidence to support your claims.

Gene Therapy

Gene therapy uses genetic engineering to treat or prevent the genetic basis of disease. A common gene therapy technique uses a delivery mechanism, or vector, such as a bacterium or virus, to deliver a new gene to target cells. Once the gene enters the cells, the new DNA is transcribed and the new protein is expressed.

Not all diseases are good candidates for gene therapy. For example, a disease caused by the interaction of multiple genes is not a good candidate because the necessary modification of genes would be too complex. Also, if the genetic basis for a disease is not understood, it is not a good candidate. Scientists need to know which gene to modify to combat the disease. If the biology of the disorder is not understood, the disease is also not a good gene therapy candidate. Finally, if there is no way to get new genetic information to affected cells, the disease is not a good candidate for gene therapy.

FIGURE 16: Cystic fibrosis is caused by a mutated CFTR protein.



Cystic fibrosis (CF) is an inherited disease that affects the respiratory and digestive systems. Airways and some organs are naturally lined and protected by a layer of mucus. Cystic fibrosis causes abnormal, sticky mucus secretions in these areas. Symptoms include coughing and wheezing, digestive problems, and increased probability of infections. The most common cause of death in untreated CF patients is a fatal lung infection.

The protein that regulates mucus secretion in the respiratory, reproductive, and digestive systems is encoded by the Cystic Fibrosis Transmembrane Conductance Regulator (*CFTR*) gene. A normal version of the *CFTR* gene produces a protein that acts as a channel to move chloride ions across the cell membrane in mucus-producing cells. This helps regulate the water content of surrounding tissues, leading to normal, moist mucus. A mutated gene leads to disruption of the chloride channels, lowering the water content of nearby cells. This causes the thick, sticky mucus characteristic of cystic fibrosis.

Cellular functions are highly related to the structure of DNA. In the case of cystic fibrosis, the change in the DNA sequence of the mutated *CFTR* gene results in a different amino acid sequence in the CFTR protein. Typically, a phenylalanine amino acid is missing from the protein sequence. When this protein is expressed, the abnormal structure leads to a loss of protein function.

As shown in Figure 16a, with normal CFTR function, chloride ions move across the cell membrane and congregate on the outside of the cell, making an ionic gradient. The hypertonic solution outside of the cell attracts more water and maintains mucus of a normal consistency. A healthy, watery mucus layer traps particulates and bacteria before they can harm the cell. The cilia of the cell are free to move and sweep away the foreign matter.



Gather Evidence

Does cystic fibrosis meet the criteria to be considered for gene therapy? Use evidence to support your claims.

In a person affected by cystic fibrosis, the irregular protein produced by the mutated *CFTR* gene cannot transport chloride ions across the cell membrane, as shown in Figure 16b. This loss of protein function results in a higher concentration of chloride and sodium ions inside the cell and a lower concentration of these ions outside of the cell. The hypotonic solution causes water to move into the cell, drying out the mucus layer. The thick, sticky mucus prevents the cilia from moving and clearing debris. The increased presence of debris and pathogens causes increased infections in individuals with cystic fibrosis.



Engineering

Developing Approaches to Gene Therapy



Analyze

A loss-of-function mutation results in a mutated protein that does not function correctly. How could gene therapy treat this type of genetic disorder?

The problems gene therapy attempts to solve are broad and span many kinds of diseases, from genetic immune disorders to cancers. Many different approaches are required to solve these problems. To alleviate respiratory symptoms of cystic fibrosis (CF), for example, scientists need to deliver a functioning copy of the *CFTR* gene to lung cells. However, it is hard to access and modify every lung cell.

A solution to this problem is to deliver the gene therapy through an aerosol that patients inhale. Affected cells that receive a functioning copy of the gene will begin to show normal gene expression, which alleviates the symptoms of cystic fibrosis.

Gene therapy is not always this straightforward. For example, some mutations produce a dominant-negative protein. This type of mutated protein does not do its job correctly and also blocks normal proteins from functioning. Simply delivering a working copy of the gene to affected cells won't work because the dominant-negative protein would still block the function of normal proteins. A solution to this problem is to "silence," or turn off, the mutated gene so that no protein is produced. Huntington's disease produces a dominant-negative protein and is a promising candidate for gene-silencing therapies.

Gene therapy relies on many different biotechnologies. Without genetic testing, it would be harder to determine which patients would benefit from gene therapy. The genes required for insertion into affected cells are produced through PCR. Without the rapid amplification of DNA through PCR, gene therapies would take much longer to produce. CRISPR is a relatively new tool, but it is already affecting gene therapy by making it easier to cut and edit DNA segments of a mutated gene.



Explain Think back to the fluorescent zebrafish from the beginning of this lesson. Using this example, explain some implications of being able to edit genes. Where do you think science will go from here?

Language Arts Connection

Knockout Mice

Knockout mice have a gene of interest knocked out, which means the gene is turned off. Knockout mice are often used in genetic engineering, allowing researchers to study structure and function in gene expression. Many knockout mice are named for the gene that has been deactivated. For example, the p53 knockout mouse does not have the *p53* gene, which produces a protein that stops tumor growths. This line of mice is susceptible to cancer. Other mice have genes knocked out that affect obesity, anxiety, and other traits.

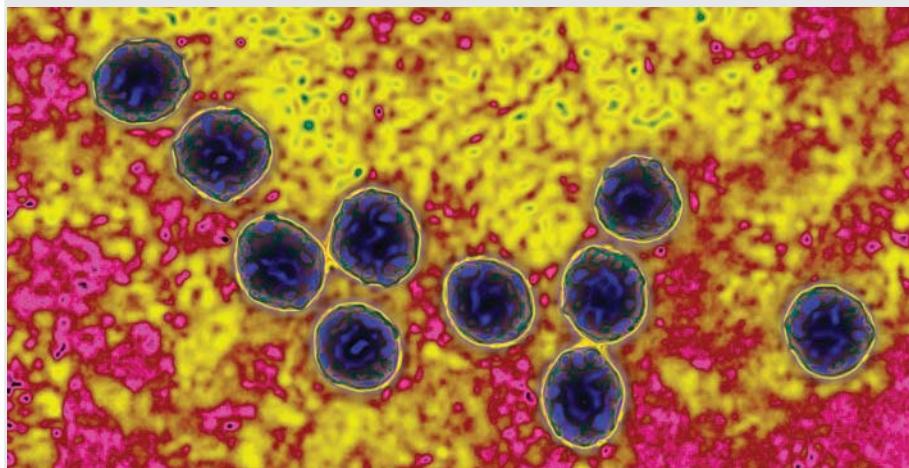
Knockout mice have been used in thousands of experiments studying many different diseases. Recently, knockout mice helped scientists confirm the link between Zika infections in pregnant women and birth defects.

The Zika virus was identified in humans in 1952. The first major outbreak of Zika occurred in 2007. Another major outbreak occurred in 2016 and scientists started to study the effects of Zika infections in more detail. Of particular interest were reports that the Zika virus causes microcephaly, a birth defect characterized by a small head and abnormal brain development. Scientists needed to learn more about the link between Zika and birth defects to accurately advise the public on the risks of Zika infections.

Mice are not ideal models for testing the effects of Zika because the mouse immune system prevents a sustained Zika infection. To solve this problem, a group of scientists knocked out a key

Image credits: ©James Cavallini/Science Source

FIGURE 17: The Zika Virus



immune system gene. When the gene was not expressed, the Zika virus could replicate within pregnant mice. None of the fetuses survived, but scientists did find concentrations of the Zika virus in the placenta that were 1000 times higher than the concentration of Zika in the mother's blood. The placenta is responsible for supplying blood to the fetus. A high viral concentration in the placenta supports the hypothesis that Zika affects the placenta, thereby harming the fetus.

The Zika virus was also found in the heads of the fetuses. This suggests that Zika directly affects brain development. Scientists have continued the Zika research using knockout mice and other techniques, and there is now a confirmed link between Zika and birth defects in humans.

Knockout mice provide a valuable model for studying the effects of gene expression, but there are limitations. Some genes behave differently in mice than in humans. A knocked-out gene

may fail to produce a response in mice when the gene is known to cause a response in humans. Or the gene may cause a different response in humans than is seen in mice. These constraints must be considered when developing or selecting a knockout mouse model for an experiment. Knockout mice are imperfect models in these cases, though they still may provide some information about the function of genes.



Language Arts Connection

Answer the following questions in your Evidence Notebook. Use evidence from the text to support your answers:

- What happens when a gene is knocked out in a mouse?
- How are structure and function related in the development of knockout mice?
- What is a limitation of using knockout mice for disease models?

LABELING GMOs



MODELING GENETIC
ENGINEERING

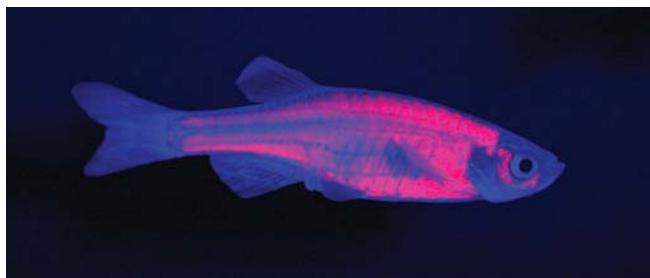
BACK FROM EXTINCTION

Go online to choose one
of these other paths.

Lesson Self-Check

CAN YOU EXPLAIN IT?

FIGURE 18: Genetically modified zebrafish.



Fluorescent zebrafish are genetically modified. Originally, fluorescent color genes from jellyfish and sea anemones were inserted into zebrafish eggs. The color genes became part of the zebrafish DNA. It is now a heritable trait that is passed to offspring. Current generations of fluorescent zebrafish are born, not modified, but their roots lie in genetic modification.



Explain Refer to the notes in your Evidence Notebook to answer the following questions:

1. How can a gene from one organism be inserted into the genome of an unrelated organism?
2. Does genetically altering an organism make a new species?
3. What are the implications of genetic engineering?

The green fluorescent protein (GFP) is used for more than creating glow-in-the-dark pets. When a GFP sequence is added to a gene, the translated protein will include the green fluorescent protein, which glows. This glowing tag allows scientists to track the protein in the organism. Knowing where, when, and how often a protein is made is important for understanding what abnormal expression of a protein looks like. GFP has been modified to produce a range of colors. The different colors are used by scientists to track multiple proteins at the same time.

Research performed using GFP tags includes exploring cell behavior during embryonic development, monitoring cell death during apoptosis, and studying insulin cells in the pancreas. Processes that are difficult to monitor directly, such as the growth of a neuron or tumor, can be tracked using GFP-tagged proteins.

CHECKPOINTS

Check Your Understanding

1. What is the difference between genetic engineering and cloning?
 - a. Genetic engineering is governed by an international ethics committee. Cloning does not have any formal ethics oversight.
 - b. Genetic engineering uses PCR and CRISPR. Cloning does not use PCR or CRISPR.
 - c. Genetic engineering focuses on changing an organism's genome, while cloning focuses on exactly copying genetic material.
 - d. Genetic engineering refers to gene manipulation in humans. Cloning refers to gene manipulation in all other species.
2. Place the elements in order to model how mosquito populations can be controlled using genetic engineering.
 - a. affected males and affected offspring die
 - b. insertion of gene into mosquito embryo
 - c. release of affected male mosquitos to the wild
 - d. development of drug-dependency gene
 - e. breeding of genetically modified mosquitoes
 - f. affected males mate with wild females
3. What would happen if a thermocycler malfunctioned during a PCR run and never heated the solution?
 - a. The DNA polymerase used to separate the DNA strands would not be activated.
 - b. The PCR would proceed at a slower rate.
 - c. The primers would not bind to the target DNA sites, and DNA synthesis would not occur.
 - d. The DNA strands would never separate, and the PCR would never begin.
4. Which of the following is not a criterion for a disease being a good candidate for gene therapy?
 - a. genetic information can be distributed to new cells
 - b. biology is understood
 - c. genetic basis of disease is identified
 - d. controlled by one gene up to a handful of genes
 - e. none of the above
5. How does the mutated CFTR protein contribute to cystic fibrosis?
 - a. The mutation prevents the channel protein from moving chloride across the membrane, resulting in a thick, sticky mucus.
 - b. The mutation prevents the channel protein from moving sodium across the membrane, resulting in a thick, sticky mucus.
 - c. The mutation causes the channel protein to produce the sticky, thick mucus.
 - d. The mutation attracts more mucus to the channel protein.
6. How is CRISPR used in genetic engineering?
 - a. to clone cells
 - b. to cut DNA
 - c. to insert foreign DNA in a chromosome
 - d. to test for genetic conditions

MAKE YOUR OWN STUDY GUIDE



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

Genetic engineering is used to solve many societal and environmental problems, but there are benefits and risks associated with genetic engineering.

The ethical considerations of cloning and genetic engineering are complex. Scientists must balance scientific progress with the concerns of the public.

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 genetics, engineering, technology, and society influence and affect each other.

A BOOK EXPLAINING
COMPLEX IDEAS USING
ONLY THE 1,000 MOST
COMMON WORDS

BOOKS INSIDE US

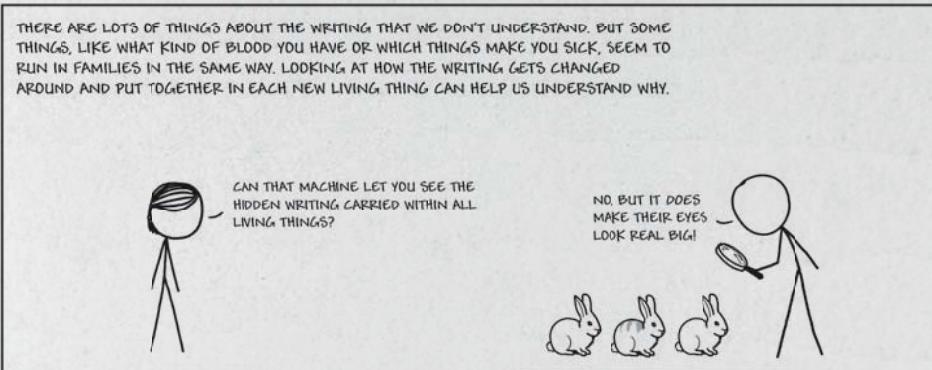
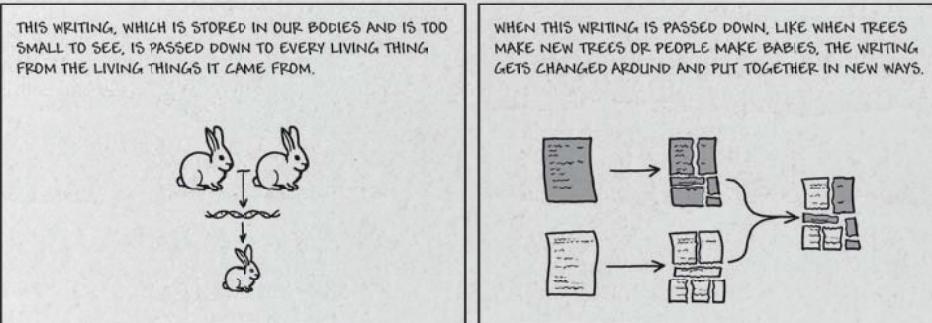
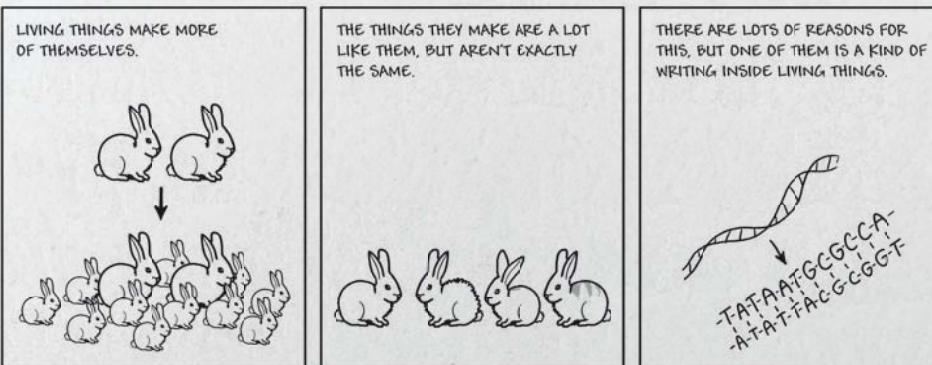
Why people's children are sort of like them but not exactly the same



You know that genetic material from two parents results in offspring with traits that follow a pattern of inheritance. Mendel found that these patterns can be predicted by using mathematical probabilities. Here's an overview.

RANDALL MUNROE
XKCD.COM

THE STORY OF HIDDEN WRITING AND FAMILY TREES

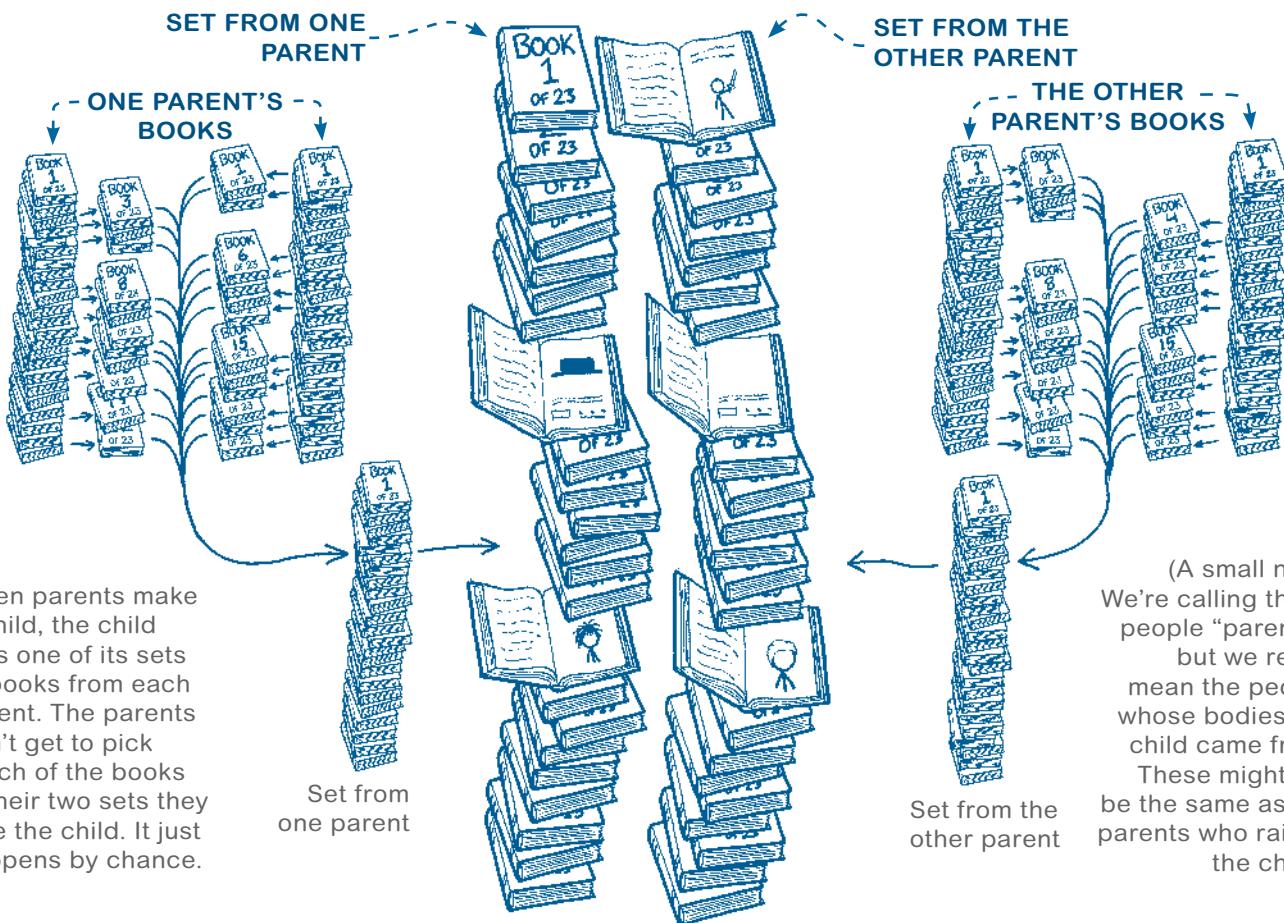


WRITING INSIDE YOUR BODY

Living things are full of a kind of writing telling them how to grow. It's not written using letters and words; it uses four different tiny pieces, like letters, stuck together in long lines. We can think of them like words in a set of books, but remember, they're not really "words" like we're used to. People, like a lot of animals, have two different full sets of books in their bodies.

One set is from each parent. The two sets match; each book in a set has a matching book in the other set that is about the same size, as if they have the same number of pages, even if each page doesn't have the same words. We won't worry about how to read what the writing says. After all, in real life, we don't really know what most of the writing is for!

THE TWO SETS OF WRITING IN YOUR BODY THAT TELL IT HOW TO GROW



When parents make a child, the child gets one of its sets of books from each parent. The parents don't get to pick which of the books in their two sets they give the child. It just happens by chance.

LEFT AND RIGHT

To learn a little more about how the left and right books come together, let's imagine that there's a piece of writing in a person's book that decides whether they will have a hat or not. (We'll pretend hats are part of our bodies.)

In the part of the book about hats, let's say some books have a piece of writing that means "HAT" and other books don't.

Here's the Hat Law: If either of your books says "HAT," then you will have a hat.



This person has a hat because there's a hat in one of their books.



This person has a hat because there's a hat in both of their books.



This person has a hat because there's a hat in one of their books.



This person doesn't have a hat because there's no hat in either one of their books.

WHY CHILDREN ARE DIFFERENT

TWO PARENTS WHO ARE THE SAME

Let's suppose two parents with hats have a baby. These parents both have the same thing in their books, as far as hats go—a hat in their left book and no hat in their right.

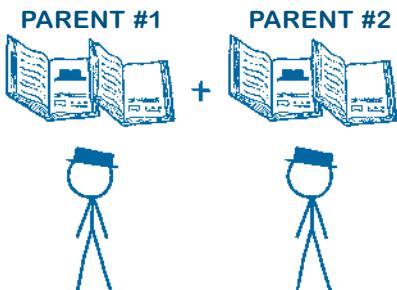
WHAT HAPPENS TO THE KIDS?

Since the parents both have hats, you might think the kids will have hats too. But it's not that simple.

CHANCE SQUARE

Each parent gives the child one of its two books. Since each parent has one "hat" book and one "no hat" book, the child could end up with two "hat" books, two "no hat" books, or one of each!

You can draw a square like this one to figure out the chances of each thing happening.

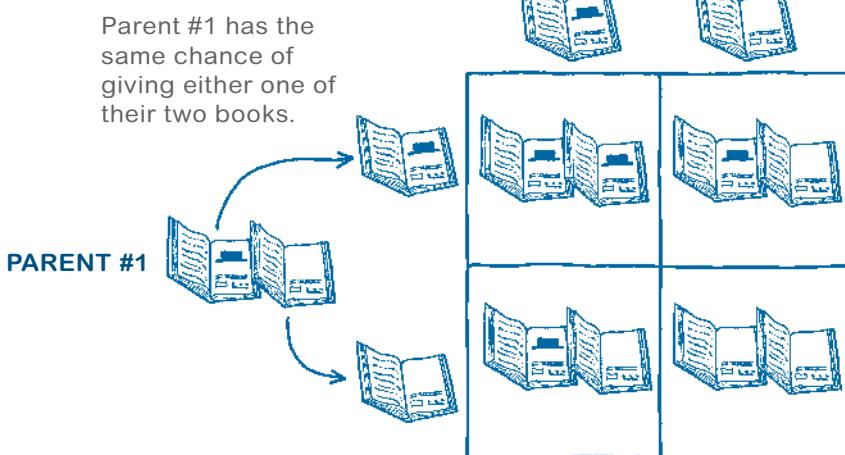


A NOTE ON HATS

In real life, hats aren't passed down in families like this, but lots of real things are.

Humans have yellow stuff in their ears, which is either wet or dry. "Having wet yellow ear stuff" is passed down in families like these hats are.

In cats, short hair is passed down in the same way. Having long hair is like having a hat and follows the same rules as hats do in these pictures.

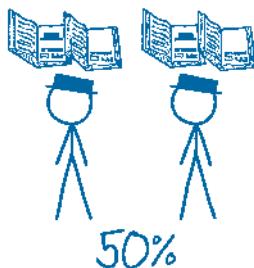


These four squares show the four things that can happen. Since each square has an equal chance of happening, we can use it to figure out the child's chance of getting each pair of books.

The child has a one-in-four chance of getting the "no hat" book from both parents. If that happens, the child will not have a hat, even though both parents do.



The child has a two-in-four chance of getting the "no hat" book from one parent and the "hat" book from the other. That means the child will have one "hat" and one "no hat" book, just like both parents do. Like their parents, these children will have hats.



The child has a one-in-four chance of getting the "hat" book from both parents. This child will have a hat, and they are different from their parents because they don't have a "no hat" book.





Go online for more
about *Thing Explainer*.

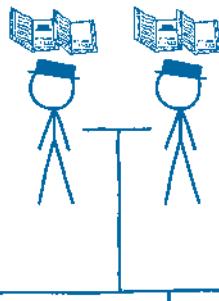
A HAT FAMILY TREE

Let's follow a pretend family tree to see how different branches end up with different sets of hat and no hat books.

This person, who has one hat book like their parents, meets someone with no hat and has children.

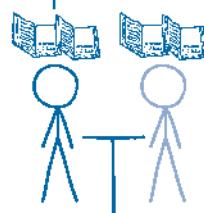


The family tree starts with two parents. Each has one hat book and one no hat book.

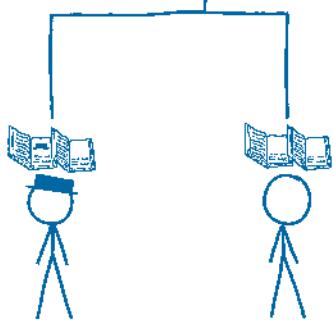
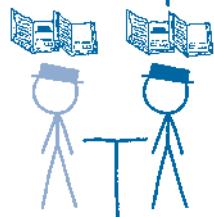


The parents have three children. Two of them have one hat book, like their parents (there's a two-in-four chance of this) and the third has no hat books (there's a one-in-four chance of this).

This person, who has no hat, meets someone else with no hat and has children.



This person, who has one hat book, meets someone else with one hat book and has children.



This person has a hat, since they got the hat book from their hat-wearing parent. There was a one-in-two chance of this.

This person has no hat, which means hat books are gone from this branch of the family until someone with a hat shows up.

These people have one hat book and one no hat book, like their parents and grandparents.

This person got a hat book from both parents, which means all children this child has will have hats.

Since neither of the parents in this branch have hats, neither of the children have hats either, since no one has a hat book.

Engineering Connection

Curing Blindness with Gene Therapy Certain diseases of the retina and eye, including some that can cause blindness, have genetic causes. With recent advances in technology, genetic engineers now have tools that may allow them to reverse the genetic damage and perhaps restore patients' sight. Clinical trials have shown promise in this area, with more research emerging every day.



Using library and Internet resources, research the factors scientists must consider when developing a gene therapy for blindness. Write a report to discuss the pros and cons of medical advances, using gene therapy as the example. Reference information from a range of sources to develop a clear dialogue that explains the technical, safety, social or other factors related to medical advances such as gene therapy.

FIGURE 1: Scientists are developing therapies for genetic eye diseases.



Music Connection

Does Practice Make Perfect? Scientific studies have found that when it comes to musical talent, genetics may play an important role. Practice or no practice, it's possible that some aspects of musical ability may be hard-wired into our genes.



Using library and Internet resources, research studies that have examined the role genetics may play in the development of musical talent. Form your own opinion about whether practice, genetics, or both are the key to becoming an expert musician. Write a blog post stating your opinion. Cite evidence and examples from credible sources to support your claim.

FIGURE 2: Musicians practice many hours every week.



Life Science Connection

Beneficial Mutations Small changes, or mutations, in DNA can result in new or modified phenotypes. If those mutations occur in germ cells, they may be passed on to future generations. Some scientists think that changes in environmental conditions, such as global warming, may cause an increase in the rate of mutations. Scientists have found evidence that the genetics of several species, such as brown-lipped snails and red squirrels, are changing in response to higher temperatures.



Using library and Internet resources, research at least three organisms scientists claim have mutations that became more common due to changing environmental conditions. Make an infographic explaining what mutations are and how they happen. Then, describe why scientists think certain mutations are helping some species survive in environments with higher temperatures. Not all scientists may agree with the causes for the changes observed. If you find evidence supporting a different claim, explain the counterargument and why scientists may draw different conclusions. Use evidence from credible resources to support your claims.

FIGURE 3: Shell color and banding pattern in brown-lipped snails are dependent on temperature.



SYNTHESIZE THE UNIT



In your Evidence Notebook, make a concept map, graphic organizer, or outline using the Study Guides you made for each lesson in this unit. Be sure to use evidence to support your claims.

When synthesizing individual information, remember to follow these general steps:

- Find the central idea of each piece of information.
- Think about the relationships between the central ideas.
- Combine the ideas to come up with a new understanding.

DRIVING QUESTIONS

Look back to the Driving Questions from the opening section of this unit. In your Evidence Notebook, review and revise your previous answers to those questions. Use the evidence you gathered and other observations you made throughout the unit to support your claims.

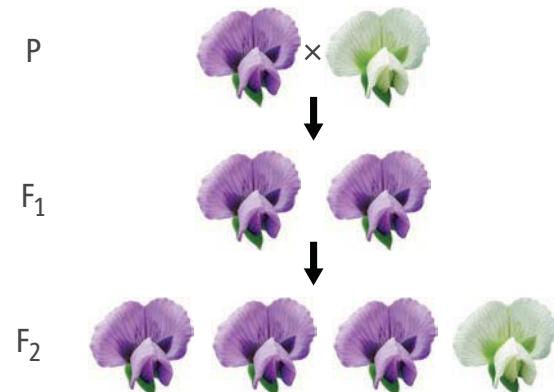
PRACTICE AND REVIEW

1. What processes that occur during meiosis contribute to genetic diversity in offspring? Select all correct answers.
 - a. crossing over
 - b. cytokinesis
 - c. gametogenesis
 - d. independent assortment

2. If meiosis produces four daughter cells, why does gametogenesis in females produce only a single egg cell?
 - a. Egg cells contain four times the amount of DNA as sperm gamete cells.
 - b. Egg cells in females are not produced via meiosis.
 - c. Gametogenesis involves steps in addition to meiosis, in which a single egg cell is retained.
 - d. The process of meiosis does not fully complete when producing an egg cell.

3. The human genome contains roughly 1,000 olfactory receptor genes, which allow us to detect and distinguish different odors. While only about one-third of these genes are functional, all of the genes may have arisen as duplications of a single ancestral gene. Put these steps in order to illustrate how this process could have occurred.
 - a. mutations accumulate over time in the duplicate copy of the gene
 - b. chromosomes exchange uneven amounts of DNA
 - c. the duplicate gene encodes a protein with a slightly different function
 - d. homologous chromosomes line up in the middle of the cell during meiosis
 - e. a chromosome obtains multiple copies of the same gene

FIGURE 4: A purebred purple and purebred white flower were crossed. The F₁ generation self-pollinated, resulting in the F₂ generation.



4. Examine the image shown in Figure 4 representing the cross of purebred plants and the self-pollination of the F₁ generation. Use these terms below to complete the statement explaining why some plants in the F₂ generation display white flowers.

dominant, recessive, heterozygous, homozygous

The F₁ plants are _____, with one _____ allele coding for white flowers. The plants in the F₂ generation with white flowers are _____, with two alleles for white flowers. The plants in the F₂ generation with purple flowers may have one or two copies of the purple flower allele, because it is _____ and masks the white trait.

- 5.** Imagine that a species of mouse has a gene controlling fur color, with the dominant allele associated with black fur and the recessive allele associated with white fur. A second, epistatic gene also influences fur color in these animals. What can you say about the color of a mouse that is heterozygous for the first fur-color gene?
- The mouse's fur color will be black.
 - The mouse's fur color will be white.
 - The mouse's fur color will be gray, or a mix of black and white.
 - The mouse's fur color cannot be determined without knowing the genotype of the epistatic gene

Use the information below to answer Questions 6–8.

The trait for red coloring (*R*) is dominant to the trait for white coloring (*r*) in birds. Imagine two heterozygous birds were crossed.

- Make a Punnett square for the cross between two heterozygous birds.
- What is the probability that the offspring of this cross will have red coloring?
- What percentage of possible offspring will have *RR*, *Rr*, or *rr* genotypes?
- In what ways can a recessive X-linked disease such as red-green color blindness be inherited by male offspring?
 - From the father, but only if the father has the colorblind phenotype.
 - From the father, even if the father is unaffected.
 - From the mother, but only if the mother has the colorblind phenotype.
 - From the mother, even if the mother is unaffected.
- Would you expect a mutation that deletes one base in a protein-coding region of DNA to be more or less harmful than a mutation that deletes three bases in a coding region? Explain your answer.

- 11.** Imagine that a pea plant develops a somatic cell, or body cell, mutation that allows the plant to grow twice as tall as other pea plants. What will be true of the plant's offspring?
- All offspring will be taller than other plants.
 - Some offspring will be taller than other plants.
 - Some offspring may be taller, but only if the plant self-pollinates.
 - None of the offspring are likely to be taller than other plants.
- 12.** The polymerase chain reaction, or PCR, allows scientists to amplify target regions of genetic material for further study. What are some of the possible applications of PCR? Select all correct answers.
- Amplify the sequence of a gene from a human blood sample to determine if a mutation is present.
 - Amplify a protein sequence from a salamander egg to inject into another egg.
 - Amplify a DNA region from a herbicide-resistant plant to insert into the DNA of another plant species.
 - Amplify a region of human DNA to insert into a bacterial plasmid.

UNIT PROJECT

Return to your unit project. Prepare your research and materials into a presentation to share with the class. In your final presentation, include an evaluation of your predictions, analysis, and conclusions.

Remember these tips while evaluating:

- Look at the empirical evidence—evidence based on observations and data. Does the evidence support your explanation of the cause or causes of progeria?
- Consider if the explanation is logical. Does it contradict any evidence you have seen?
- Is there enough evidence from credible sources to support your conclusions?

Analyzing Traits in Tomato Plants

In 2012, the tomato genome was fully sequenced. This knowledge allowed geneticists to study tomato traits and their genetic basis. For example, several genes affect the color of the fruit. Tomatoes can come in many colors. Interactions between the tomato's skin and the fleshy material result in the color we see. Tomato skin can either be yellow or clear.

Imagine you're a farmer and want to know the genotypes of your tomato plants regarding tomato skin color. You ran several testcrosses, shown in Figure 6, to determine the genotypes of your plants (the P generation).

1. DEFINE THE PROBLEM

With your team, write a statement outlining the problem you've been asked to solve. Record any questions you have about the problem and the information you need to solve it.

2. ANALYZE DATA

Make a plan for organizing the data and approaching the problem. How will you determine which allele is dominant and which allele is recessive? What assumptions can you make based on the data?

3. DEVELOP A MODEL

With your team, develop a system for modeling the yellow and clear alleles. When you have decided on a system, indicate the genotypes for each phenotype in the P-generation crosses. Then, use a Punnett square to calculate the probability of producing tomatoes with clear or yellow skin for each type of possible cross between heterozygous, homozygous-recessive, and homozygous-dominant plants. Determine the probability of producing each genotype and phenotype for each possible type of cross.

4. CONDUCT RESEARCH

Research ways that geneticists might alter or enhance this trait. Why might they want to do so?

5. COMMUNICATE

Write a report describing your findings and the process you used to determine how this trait is influenced by different alleles. In your report explain which allele is dominant and recessive, describe the model you used, and give the genotypes of the plants in the P generation.

FIGURE 5: Tomatoes can come in many colors including red, yellow, green, and even white.

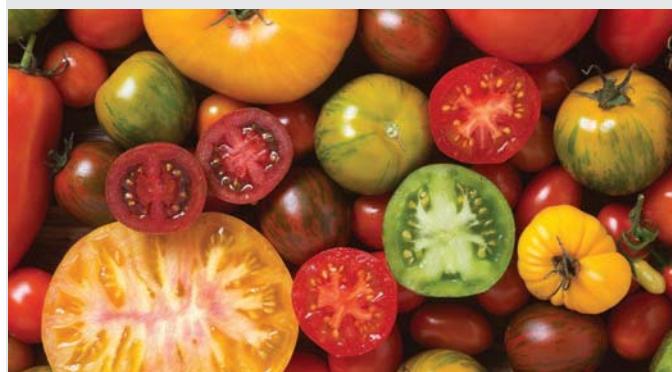


FIGURE 6: Number of tomato plants resulting from crosses of parental plants with yellow or clear skin.

P generation phenotypes	F ₁ generation phenotypes	
	number of yellow	number of clear
yellow × clear	16	17
yellow × yellow	25	8
yellow × yellow	32	0
yellow × clear	23	21
yellow × clear	26	0
clear × clear	0	29
yellow × yellow	33	11



CHECK YOUR WORK

A complete presentation should include the following information:

- an explanation based on evidence explaining how alleles are related to this trait in tomato plants
- a valid model for the alleles involved in this trait and correct Punnett squares showing the probability of each genotype and phenotype from different types of crosses
- a description of techniques geneticists use to genetically engineer plants and reasons for using technology in this way