Genetics: Content Background Document

1. Introduction

As we begin our exploration of genetics, consider what you already know about this topic. You may recognize that the offspring of two parents can be different from another depending on the combination of genetic material they inherit from each parent. You may also have a basic mental picture of the key players involved in heredity, from DNA to genes to chromosomes. If someone asked you why differences exist among individuals in a population, you might point to either genetic factors (such as genes and chromosomes) or environmental factors (such as food, climate, sunlight, and the presence of predators) that can impact an individual's growth or development.

But how well do you actually understand genetic and reproductive processes and environmental influences on individual differences?

The content that follows will challenge you to broaden and deepen your understanding of genetic structures and processes and the underlying factors that influence trait inheritance and variations in living things. To a lesser extent, we'll explore how genetics and environment interact and influence trait development. A strong conceptual understanding of genetics will also help you better understand student ideas and misconceptions so that you can teach genetics content more confidently and effectively. Learning to identify and address common student misconceptions will enable you to help students develop scientifically accurate understandings of genetics that will become building blocks for further study.

This document was written with you, the teacher, in mind. The subject matter is tied to the lessons you'll be teaching, but the concepts are presented at a higher level to equip you with the tools and background you'll need to guide student learning.

2. Getting Started: Students' Conceptions of Living Things

First, let's begin with a fundamental science concept: life. What does it mean to be a living thing, and how does one distinguish between living and nonliving things? This concept is essential for studying biology and developing accurate scientific understandings of how living things reproduce and survive on Earth.

Before you begin teaching this unit, you'll want to make sure your students have an accurate conception of living things and their characteristics:

- 1. They are composed of cells.
- 2. They maintain homeostasis—a constant state. Mammals, for example, maintain a constant temperature.
- 3. They use energy and chemicals for growth and maintenance.
- 4. They respond to stimuli—from a single contraction of a unicellular organism to the complex reactions of multicellular organisms that involve all the senses.

5. They reproduce, which means they have the ability to produce new individual organisms either asexually from a single-parent organism or sexually from two parents.

Understanding which processes characterize living things and which do not is essential for developing biological knowledge. According to the Disciplinary Core Ideas progression in the *Next Generation Science Standards*, students in grades K–12 are expected to "[increase] sophistication of student thinking." By grades 6–8, students are expected to understand how all living things are engaged in life processes. Equipped with these insights, students will be ready to examine genetic processes and the roles inheritance and environment play in trait variations within populations.

3. What Are Traits?

To develop a scientifically accurate understanding of genetics, we need to make sure we're using words correctly. Let's start with a common term scientists use: *traits*.

Think of a dog. How do you know that the organism you have in mind is actually a dog? You might say that a dog has four legs, two ears, fur, and a tail that wags. These and many other features are the traits of a dog. All species, or organisms of the same kind, have a unique set of traits that set them apart from other species and help us recognize them.

Organisms have many different kinds of traits. *Physical traits* include such characteristics as hair or fur color, eye color, the presence or absence of wings, and many other observable characteristics. *Behavioral traits* may include mating rituals or methods of caring for young. For example, bees do a waggle dance to point other bees to food or water.

Another kind of trait involves DNA, a complex molecule called *deoxyribonucleic acid* that is found in most body cells. All of the biological information for an organism is found in its DNA. Genes, the functional units of DNA, carry genetic information that determines the traits of an organism. The specific sequence of DNA that an organism inherits is called a *molecular trait*. In addition to studying physical traits, scientists explore the molecular basis for a trait, or the molecular traits themselves.

A series of chemical reactions in an organism, or a *chemical pathway*, is also considered a trait. One example is the series of chemical reactions that break down sugars in the body to release energy. *Developmental pathways* are yet another category of trait. Tissue specialization in various parts of the body, such as heart tissue and other muscle tissue, is an example of a developmental pathway.

The sum and interaction of individual traits are what make an organism unique. All organisms of the same kind have similar traits. These shared traits help us identify a species. But not all individuals of the same species are exactly the same. In other words, some traits vary among individuals within a species. Scientists who study genetics seek to understand the processes that lead to trait development and whether trait variations can be passed from one generation to the next.

Passing genetic information for trait development to subsequent generations through reproduction is a fundamental characteristic of living things. This is the focus of the next section.

4. Reproduction: Asexual and Sexual

Reproduction is defined as "the ability to produce new individual organisms either asexually from a single-parent organism or sexually from two parent organisms." This characteristic of living organisms is necessary for the survival of a species.



STOP AND THINK

What are the differences between asexual and sexual reproduction? What are some examples of organisms that engage in each type of reproduction?

4.1 Asexual Reproduction—Duplicating Genetic Material

Asexual reproduction commonly occurs in single-celled organisms. You may remember viewing amoeba and paramecia under a microscope in your high school biology class. These single-celled protists are prime examples of organisms that reproduce asexually. They duplicate themselves simply by splitting a single parent cell into two daughter cells. Bacteria also divide in this way and reproduce relatively quickly. Asexual reproduction doesn't involve recombining the genes of two individuals of a species. Rather, the newly formed organism is an exact *copy*, or clone, genetically identical to its parent. Some single-celled organisms reproduce asexually through *budding*, as in yeast, where the bud, or daughter cell, is smaller than the larger mother cell but still contains identical genetic material. Life-science teachers often demonstrate another form of asexual reproduction called *vegetative propagation*. Plants like potatoes, spider plants, or various plant bulbs are used to produce new, genetically identical offspring without the creation of seeds.

Many additional modes of asexual reproduction occur in nature, such as *parthenogenesis*, in which an unfertilized egg develops into a fully functioning individual. This type of reproduction is common in some invertebrates, such as stick insects, aphids, and rotifers, and in vertebrates like hammerhead and blacktip sharks. To complicate matters, some organisms can reproduce either asexually or sexually depending on environmental conditions. These examples, and many more, attest to the fact that organisms have developed a multitude of asexual reproductive modes.

Reproducing asexually has some advantages and disadvantages. One advantage is that since an organism doesn't need to find a mate, reproduction requires less energy and can occur more quickly, which in turn results in rapid population growth. Additionally, the genes are successfully combined in the parent organism. The parent is alive! The major disadvantage of asexual reproduction is that organisms are particularly vulnerable to environmental changes. With so little variation among organisms, a detrimental change could wipe out the entire population.

Although offspring of organisms that reproduce asexually are generally clones of the parent, rare errors can occur when the genetic material is copied, resulting in a limited number of variations in the population. These errors are mutations—permanent changes in the DNA sequence—that occur randomly in cells. Mutations have varying effects on a population. In some instances, they may be lethal, resulting in a failure of the organism to survive. In other instances, they may confer a survival advantage that enables a cell, through a process of natural selection, to have greater reproductive success so the new mutation appears more frequently in the population. Or the mutation may have a neutral effect on reproductive success but result in trait variation.

The environment plays a significant role in the variations observed among organisms that reproduce asexually. Factors such as sunlight and the availability of nutrients or water can influence traits in these organisms. For example, differing amounts of sunlight or water can affect a plant's color, height, size, and number of leaves.

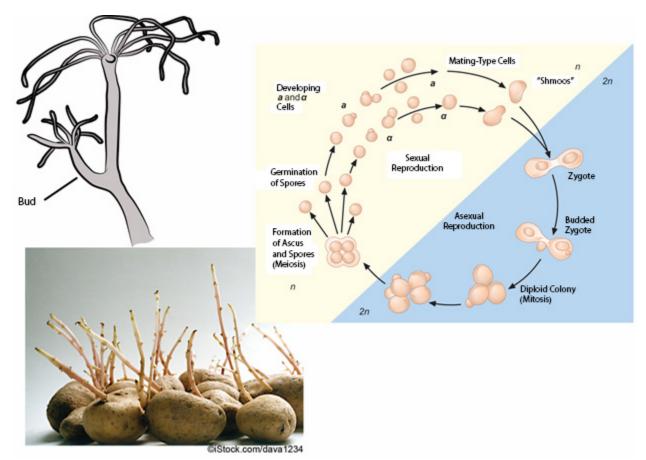


Figure 1. Modes of asexual reproduction

The association of mutations with harmful change may have significant teaching implications. Although the Genetics lesson series doesn't address mutations, students may bring this up in class. If they do, it's important to emphasize that not all mutations in the DNA sequence are harmful; some changes may be neutral or even beneficial.

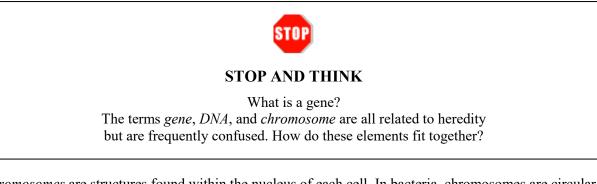
4.2 Sexual Reproduction—Combining Genetic Material

Because this unit on genetics is about variations in individuals of the same species, the focus is on organisms that reproduce sexually, combining genetic material from two parents into a unique, new individual. Sexual reproduction offers an advantage for species continuity by increasing variation through gene recombination, which enables organisms to adapt more effectively to environmental change. However, compared to asexual reproduction, sexual reproduction is slower, requires more energy, and yields offspring with new combinations of genes, some of which will be unsuccessful for survival or reproduction.

How can the genetic makeup of a species be maintained when genes from *both* parents combine to produce a single new offspring? Wouldn't the offspring inherit *double* the amount of genetic material from the parents? So how does sexual reproduction work? Let's begin with some basic concepts.

5. Genetics: What Is a Gene?

Many of your students may be familiar with terms like *gene*, *chromosome*, or *DNA* without knowing what they actually mean. For this reason, it's essential to help students develop scientifically accurate understandings of these and other terms associated with genetics. Let's focus for a moment on these basic genetics terms.



Chromosomes are structures found within the nucleus of each cell. In bacteria, chromosomes are circular, but for this discussion, we're referring to the long, linear chromosomes in most organisms. In general, chromosomes are composed of a long, double-stranded molecule of DNA (deoxyribonucleic acid) wrapped around many protein molecules. Found in most body cells, this complex DNA molecule is copied faithfully and passed on from one generation to the next. You may remember constructing models of DNA in school, creating a double helix twisted like a spiral staircase.

Most of the time, chromosomes can't be seen inside the nucleus of a cell, not even under a microscope. However, as cells divide, the chromosomes condense and become thicker, which makes it possible to observe them if the cells are stained and viewed microscopically. Each species has a characteristic number of chromosomes that may range from as few as two to more than a thousand. The number of chromosomes seems to be unrelated to either the number of genes or the complexity of the species. Organisms that reproduce sexually have two sets of chromosomes, one set inherited from each parent. For example, humans have two sets of 23 chromosomes, or a total of 46 chromosomes. Dogs have a total of 78 chromosomes, two sets of 39 chromosomes. In all organisms of a given species, each cell has the same number of chromosomes (two copies of each chromosome), except for sex cells (sperm and eggs), which have half the number of chromosomes compared to other cells (one copy of each chromosome).

Genes are sequences of nucleotide bases—adenine, thymine, guanine, and cytosine—at specific locations on a strand of DNA in a chromosome. While the chemical makeup and function of genes is beyond the scope of these lessons and the expectations of the NGSS for 6th-grade students, it's important that you understand how genes can shape an organism's traits. The nucleotides in a gene code for the specific amino acids that make up proteins. Proteins carry out instructions or tasks, such as building a new copy of a cell or repairing cellular damage. Most proteins have very specific functions, so if a cell needs to perform a new task, it must make a new protein. Similarly, if a cell needs to do something faster or slower than before, it makes more or less of the responsible protein. Genes contain information about which proteins to make and in what amounts. Interestingly, only one sixtieth of our entire genome codes for making proteins. The rest of the DNA helps direct when and where in the body each gene will be used. So the collection of all of the DNA in a cell (often called its *genome*) gives biological instructions for an organism's distinct traits, passed from parent to offspring. Genomes and the genes they contain are the blueprint for making new individuals, including human beings!

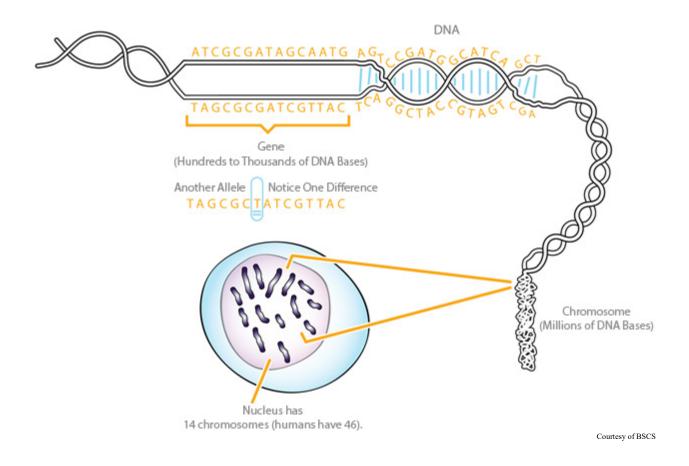


Figure 2. A *chromosome* is a long, double-stranded molecule of *DNA*. Each species has a specific number of chromosomes. Chromosomes come in matched pairs—one set from each parent. *Genes* exist at specific locations on a strand of DNA in a chromosome and contain a code for certain traits. *Alleles* are different forms or variations of a gene. An individual might get the same allele of a gene from each parent or different alleles from each parent.



STOP AND THINK

How many copies of each chromosome do you have in most of your body cells? How many copies of each gene do you have? Whom did you inherit each copy from?

Because genes are located on DNA strands in chromosomes, and each individual inherits one chromosome from each parent, genes in sexually reproducing organisms exist in pairs. When we think about traits we inherited from our parents, we see that we may have a trait from one parent but not the other. For instance, why do you have your mother's dimples? Why do you have your father's curly hair and not your mother's straight hair? Think about the genes that code for a certain trait. Different forms of that gene determine the trait you received, such as a gene that determined the curliness of your hair. These variations are called *alleles*, alternative forms of a gene located at a fixed position on a chromosome. The

two alleles you have for a gene (one on each chromosome) may be the same or different. The trait you exhibit depends on which alleles of the gene you inherited from your parents.

STOP

STOP AND THINK

If each individual of a species has a certain number of chromosomes, and each parent contributes genetic material to their offspring, why does the offspring not have *double* the number of the parents' chromosomes?

Most cells in the body contain *all* the chromosomes for that individual. In humans, all 23 pairs of chromosomes, 46 chromosomes in all, are found in the nucleus of cells (unlike blood cells that have abandoned their nucleus). These cells reproduce through the process of *mitosis*. Before mitosis, a cell copies each DNA molecule on each chromosome. During mitosis, a copy of each chromosome is distributed to the newly formed daughter cells. However, sexually reproducing organisms produce special cells involved in reproduction: sperm and eggs. These specialized sex cells are formed through a process called *meiosis*. In this process, each new cell receives just *one* chromosome from each chromosome pair. Thus, the sperm and egg cells created have just one set of chromosomes (a total of 23 in humans). When a sperm and egg unite in the process of fertilization, a new individual is formed with two copies of each chromosome, restoring the original number of chromosomes (46 in humans), half from each parent. Logical! But there's more.

6. Mendelian Inheritance

To better understand the origin of genetics, we need to travel back to 1866 to a monastery in what is now the Czech Republic. There in the monastery garden, a monk named Gregor Mendel conducted a series of experiments crossing pea plants with different characteristics. Carefully observing and recording variations in their structures and color, Mendel developed a theory of inheritance that forms the basis of our understanding of genetics today. Before Mendel's experiments, it was thought that organisms inherited characteristics through a *blend* of traits from their parents. But Mendel's experiments introduced completely new ideas about trait inheritance.

Plants are sexually reproducing organisms. In flowering plants, the sperm is located in the pollen produced inside a flower. Eggs are found in the ovary at the base of the flower. Mendel grew some pea plants with purple flowers and some with white flowers. When the purple-flowered plants were crossed with other purple-flowered plants, the seeds they produced always yielded purple-flowered plants. Similarly, when Mendel crossed white-flowered plants with other white-flowered plants, they always produced seeds that yielded white-flowered plants. These are called *true-breeding plants*.

When Mendel crossed true-breeding purple-flowed plants with white-flowered plants, however, he discovered that the seeds yielded pea plants of a single color (purple). He called these new plants the F1 or *first filial generation (filial* means "relating to a son or a daughter"). When Mendel fertilized these first-generation plants with pollen from their own group, both traits appeared in the F2 or *second filial generation* of pea plants in a consistent mathematical ratio: three purple-flowered plants to one white-flowered plant. Mendel then reasoned that because the colors didn't blend together and the white trait reappeared in the second generation even though the first-generation parents didn't have white flowers,

the "instructions" for both flower colors had to exist in the first-generation parents. The secondgeneration plants must have received instructions for white flowers from their parents, even though none of the parents exhibited the white-flowered trait.

The same pattern was evident when he examined tall and short pea plants, round and wrinkled peas, and four other traits. Mendel surmised that different units, called *factors*, must exist for each trait, and each factor must have two versions. For example, purple and white are factors for flower color, and round and wrinkled are factors for pea-plant seeds. We now call these factors *genes*. Earlier in our discussion, we introduced the term *alleles*, which describes the variations of these factors.

Using what he called the *principle of segregation*, Mendel hypothesized that alleles exist in pairs that separate (or segregate) when sex cells are formed. When a male sex cell from one parent is united with a female sex cell from the other parent, the offspring inherit one allele from each parent. Which allele a parent contributes to a sperm or egg cell is entirely random.

Let's review what happened when Mendel crossed purple- and white-flowered pea plants. All of the F1 or first filial generation of pea plants had purple flowers. Mendel then noted that in the F2 or second filial generation of plants that resulted from crossing or mating F1 plants, the ratio of purple-flowered plants to white-flowered plants was 3:1.

What a surprising outcome! In the first generation of pea plants, all the flowers were purple, even though they had one white-flowering parent. In the second generation, three quarters of the flowers were purple, and one quarter were white, even though all their parents had purple flowers.

Mendel then used his understanding of math to consider how such a result might have occurred. He postulated that each parent had two different alleles but randomly contributed only one allele to each offspring. That means in the F1 generation, each plant would have received a purple-flower allele from one parent and a white-flower allele from the other. The purple-flowered trait must have "covered up" the white-flowered trait so that each F1 plant had purple flowers despite having alleles for both white and purple flowers. Consequently, each F1 parent might have randomly contributed either a white- or purple-flower allele to the F2 generation plants. If that were the case, the F2 generation plants could have inherited a purple-flower allele from each parent, a purple-flower allele from one parent and a white-flower allele from the other. If a plant had even one purple-flower allele, it would exhibit the purple-flowered trait. A plant would exhibit a white-flowered trait only if it inherited a white-flower allele from each parent.

To describe this phenomenon, Mendel developed the *principle of dominance*, the idea that an organism with at least one dominant allele will display the dominant trait. So when there are two different alleles, the trait that appears is *dominant*, and when there are two recessive alleles, the trait that appears is *recessive*. We now know that the principle of dominance doesn't apply to the action of many genes, and a dominant-recessive relationship between alleles is more the exception than the rule. Nevertheless, it's common practice to represent a dominant allele with a capital letter and a recessive allele with a lowercase letter. As you examine figure 3, think about what the letters represent: strands of DNA that code for a particular protein. So we might represent the passing of alleles from parent to offspring using letters to show the allele combinations of the parents and the possible allele combinations of the offspring.





Photograph by Maksym Surovts

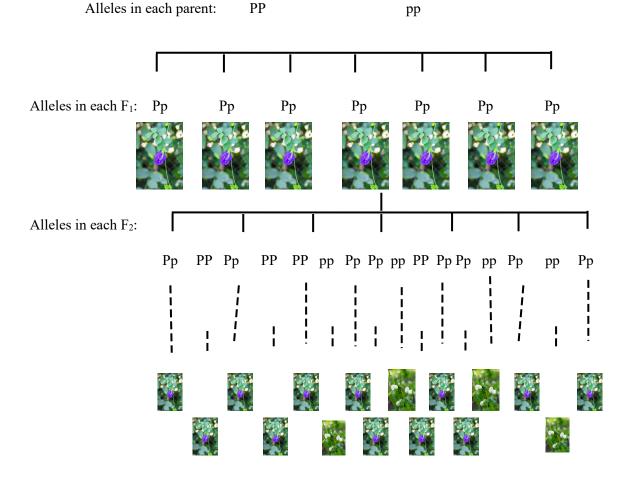


Figure 3. Three generations of pea plants. Parents are true-breeding purple-flowered plants and true-breeding white-flowered plants. In the F_1 generation, all the plants have purple flowers, but each has a dominant allele for purple flowers and a recessive allele for white flowers. The plants in the F_2 generation show a 3:1 ratio of purple to white flowers. The purple flowers may have either two purple alleles or a purple and a white allele. The white flowers must have received a white allele from each of the parent plants.

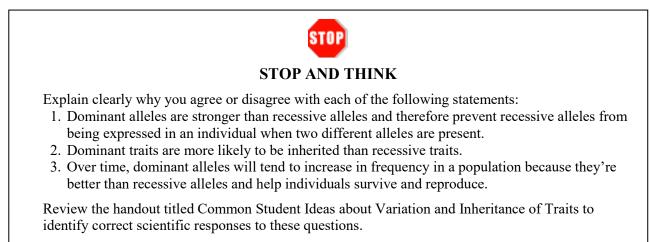
7. Key Terms

Now let's review some key terms describe the combination of alleles an individual inherits from the parents. If each parent contributes the same allele of a gene, the offspring is called *homozygous* for that particular gene. The Greek prefix *homo* means "same," and *zygous* refers to a zygote, the cell formed when egg and sperm unite. *Zygote* comes from another Greek word that means "yoked" or "to yoke together." So a homozygous individual has two of the same alleles for a particular gene. If each parent contributes a different allele for a particular gene, the offspring is called *heterozygous* for that gene. In Greek, the word *hetero* means "different."

The term *genotype* refers to an organism's genetic makeup, or the combination of two alleles received from the parents. The physical appearance of an individual, or the visible trait, is the *phenotype*. Think of how genotype and phenotype relate to the principle of dominance. Purple flowers appear even if a white, recessive allele is present.

It's important to mention here that the word *dominant* is often interpreted as strong and powerful, which can lead to the following misconceptions regarding heredity. Being aware of these misconceptions will heighten your awareness of inaccuracies in students' ideas so you can support your students in developing more scientific understandings.

- *Dominant alleles are more likely to be inherited than recessive alleles.* The fact is that alleles are equally likely to be inherited. The dominant phenotype is more likely to be observed among offspring because two possible allele combinations lead to a dominant phenotype, but only one combination results in a recessive phenotype.
- Dominant alleles are found more frequently in populations than recessive alleles. A dominant allele may actually be rare in a population. It's true that if a parent is homozygous dominant for a trait, all offspring will inherit a dominant allele and exhibit the dominant trait. But very few parents may have the dominant trait. Furthermore, if a dominant allele is rare in a population, most individuals who carry it will be heterozygous. In that case, only half of the person's offspring will receive the dominant allele. An example in humans is the achondroplasia form of dwarfism. Although this is the most common type of dwarfism and has a dominant allele, it is quite rare in the population.
- *Dominant alleles are inherited from the stronger parent*. Any individual, male or female, robust or fragile, can carry dominant alleles.



Diagrams can often make it easier to understand the process of alleles separating and recombining. A *Punnett square* is a helpful tool used to identify all possible gene combinations (genotypes) and variation in traits (phenotypes) from a cross between two individuals. Punnett squares also predict the probability of offspring exhibiting a certain genotype and phenotype. As we discussed earlier, the principle of segregation states that pairs of alleles are segregated when chromosomes separate to form gametes, and the principle of dominance states that the trait associated with the dominant allele will show up when a dominant allele is paired with a recessive allele.

Punnett square 1 (table 1) shows what happens when a pea plant homozygous for the dominant allele is crossed with a pea plant homozygous for the recessive allele. As in Mendel's F1 generation of purple-flowered and white-flowered pea plants, all the offspring in this diagram are 100% heterozygous.

In this example, we'll focus on the trait of seed shape. A capital R signifies the dominant allele for round seeds from one parent, and a lowercase r signifies the recessive allele for wrinkled seeds from the other parent.

Punnett Square 1					
		Parent 2 (rr)			
		r	r		
Parent 1	R	Rr	Rr		
(RR)	R	Rr	Rr		

Table 1. Crossing a homozygous, dominant individual and a homozygous, recessive individual

Notice that the allele for round seeds from Parent 1 is dominant, concealing or masking the recessive allele for wrinkled seeds from Parent 2. Based on the results, we can confidently predict that all offspring will be heterozygous for the dominant allele and will therefore have round seeds.

Punnett square 2 (table 2) shows another example of the principle of dominance. In this case, a homozygous, dominant pea plant (RR) is crossed with a heterozygous pea plant (Rr). As in the previous example, this results in 100% of the offspring having the dominant phenotype, though 50% of them are RR and 50% are Rr. All the offspring will exhibit the dominant trait, with no probability of the recessive trait appearing, even though half of the offspring have both the round and wrinkled alleles.

Punnett Square 2						
		Mother (RR)				
		R	R			
Father	R	RR	RR			
(Rr)	r	Rr	Rr			

Table 2. Crossing a homozygous dominant female and a heterozygous male

Punnett square 3 (table 3) depicts a scenario similar to Mendel's F2 generation of white- and purpleflowered pea plants. Each parent exhibits wrinkled seeds in spite of the fact that each is heterozygous, with both a dominant round-seed allele and a recessive wrinkled-seed allele. With this type of cross, on average, one fourth of the offspring will be homozygous recessive (rr) and exhibit the recessive phenotype (wrinkled seeds); one fourth of the offspring is expected to be homozygous dominant (RR) and display the dominant phenotype (round seeds); and one half will be heterozygous dominant (Rr) and exhibit the dominant phenotype (round seeds).

Punnett Square 3					
		Mother (Rr)			
		R	r		
Father	R	RR	Rr		
(Rr)	r	Rr	rr		

Table 3. F₂ generation resulting from crossing two F₁ heterozygous individuals

When using Punnett squares in your teaching, make sure you refer to *probability* or *expected ratios* of the different phenotypes and genotypes rather than a specific number of each type of offspring. This emphasizes the random selection of alleles that will appear in each gamete during sexual reproduction. With small numbers of offspring, the expected ratios may not be observed, but the probability of having offspring with a particular trait is the same for each individual. The greater the number of offspring, the more likely it will be that the predicted ratios of dominant to recessive phenotypes will be observed. So even though you might predict that one quarter of the F1 offspring they produce. For example, if F1 parents produce four offspring, it wouldn't be surprising if the recessive trait appeared in none of them, all of them, or two of them. It would be surprising, however, if they have 100 children, and *none* of them have the recessive trait!

Assuming that one quarter of the offspring will exhibit the recessive trait is another misconception students often have. For example, if both parents have straight hair, but each of them carries a recessive allele for curly hair, you might predict, based on a Punnett square diagram, that one quarter of their children will have curly hair. If their first child has curly hair, you might predict that the next three children will have straight hair. But the probability of having a child with curly hair is the same with *each* pregnancy, so it's possible for two, three, or more children to have curly hair.

8. Pedigree Analysis: Following Inheritance

Pedigree analysis is a tool that allows geneticists to track patterns of trait expression across multiple generations of a family and make inferences about whether an allele associated with a particular trait is dominant or recessive. This tool is often used to analyze traits in organisms, such as humans, that don't produce large numbers of offspring.

As noted earlier, Punnett squares predict the *ratios* of offspring with dominant and recessive phenotypes, but these ratios are more likely to be observed when hundreds of offspring are produced than with ten or fewer offspring.

The basic rules of pedigree analysis are as follows:

- Females are represented by circles and males by squares.
- Parents are connected by a horizontal line.
- A vertical line connects parents to their offspring.
- Siblings are connected by a horizontal line above them.
- Individuals that show the form of the trait being followed are indicated by a shaded symbol.

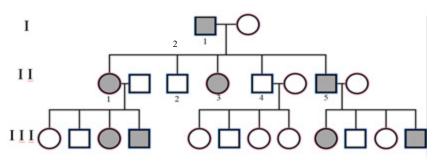


Figure 4. A typical pedigree

In this example (figure 4), the recessive trait is shaded and the dominant trait is not. By examining this pedigree chart, you can determine which individuals have different traits. Can you also determine the alleles of each individual? Consider the father (I-1) with the recessive trait. What two alleles does he need to have to exhibit the recessive trait?

Now consider the mother (I-2). Because a dominant trait is expressed whenever an individual has at least one dominant allele, she might have either two dominant alleles or one dominant and one recessive allele. However, if the mother had two dominant alleles, would it be possible for her to have offspring that exhibit the recessive trait? No, she would have to pass along one dominant gene to each offspring, and the offspring would each have the dominant trait. In this example, some of the children (II-1, II-3, and II-5) exhibit the recessive trait, so they must have received one recessive gene from their mother *and* one from their father. So the mother must be heterozygous, which means she must have both a dominant and a recessive gene.



STOP AND THINK

Based on this sample pedigree, can you figure out the genotype (the allele combinations) of each individual in the three generations of this family?

9. Other Patterns of Inheritance, or Non-Mendelian Inheritance

It would be wonderful if all inheritance patterns could be explained as neatly as Mendelian genetics presents them. However, as we've learned, there are no absolutes in nature. It turns out that purely Mendelian traits represent a small minority of all traits, and for most sexually reproducing organisms, inheritance patterns are much more complex. For example, as you may recall, when Mendel recorded the colors of his pea-plant blossoms, he theorized that the factor, or gene, for flower color was composed of two discrete alleles, purple or white. All the crosses Mendel performed resulted in either the white or purple phenotypes of the parents.

However, had Mendel used four-o'clock plants, he would have seen pink flowers—a blending of red and white parent flower colors—in the F1 generation. This is an example of *incomplete dominance*, which produces an F2 generation of red, pink, and white flowers in a ratio of 1:2:1. The heterozygous (WR) flowers are a third phenotype, a blending of red (RR) and white (WW) flowers that results in pink (WR)

flowers. In incomplete dominance, the trait you see when an individual has both allele variations (heterozygous) lies somewhere between the two traits of the homozygous individuals.

A similar situation arises when the phenotypes produced by both alleles are clearly, rather than partially, expressed. For example, in certain varieties of chickens, the allele for black feathers is *codominant* with the allele for white feathers. Heterozygous chickens are speckled with black and white feathers. Instead of the color blending as we saw in the previous example of incomplete dominance, both black and white colors appear distinctly and separately in individual chickens of the F1 generation. Many human genes, including one for a protein that controls cholesterol levels in the blood, also show codominance. People with the heterozygous form of this gene produce two different forms of the protein, each with a different effect on cholesterol levels.

Based on Mendel's theory that factors, or genes, for traits exist as one of two alleles, we may conclude that every trait is represented by one gene, and that each gene has only two alleles. In nature, however, most genes have *multiple alleles* (more than two). An individual, of course, can have at most only two allele variations for each gene, but many different alleles may be seen within a population. One example is coat color in rabbits. A single gene with at least four different alleles determines a rabbit's coat color. These four known alleles display a pattern of simple dominance that can produce four coat colors.

Another interesting type of non-Mendelian genetics is the production of a single trait from the interaction of several genes. For example, at least three genes are involved in producing the reddish-brown pigment in the eyes of fruit flies. Traits controlled by two or more genes are said to be *polygenic*, meaning "many genes." In fact, most inherited traits fall into this category. A wide range of phenotypes are often represented in polygenic traits, which display continuous or nearly continuous variation. For example, at least six genes control coat color in cats, and the variety of skin, hair, and eye colors in humans can be attributed, at least in part, to the many genes that control these traits.

10. Summary

In this document, we explored how the random combination of genes from each parent in sexually reproducing organisms can result in trait variations in the offspring of a species. Mendelian genetics (concepts based on Mendel's initial ideas about heredity) describes the basic mechanisms and predicted outcomes of sexual reproduction, but genetics is a long, complicated process involving a range of interactions, from molecules to multicellular organisms. It remains a subject of intense scientific study. For students, understanding the basic concepts of heredity is an important step in learning about themselves and their world.