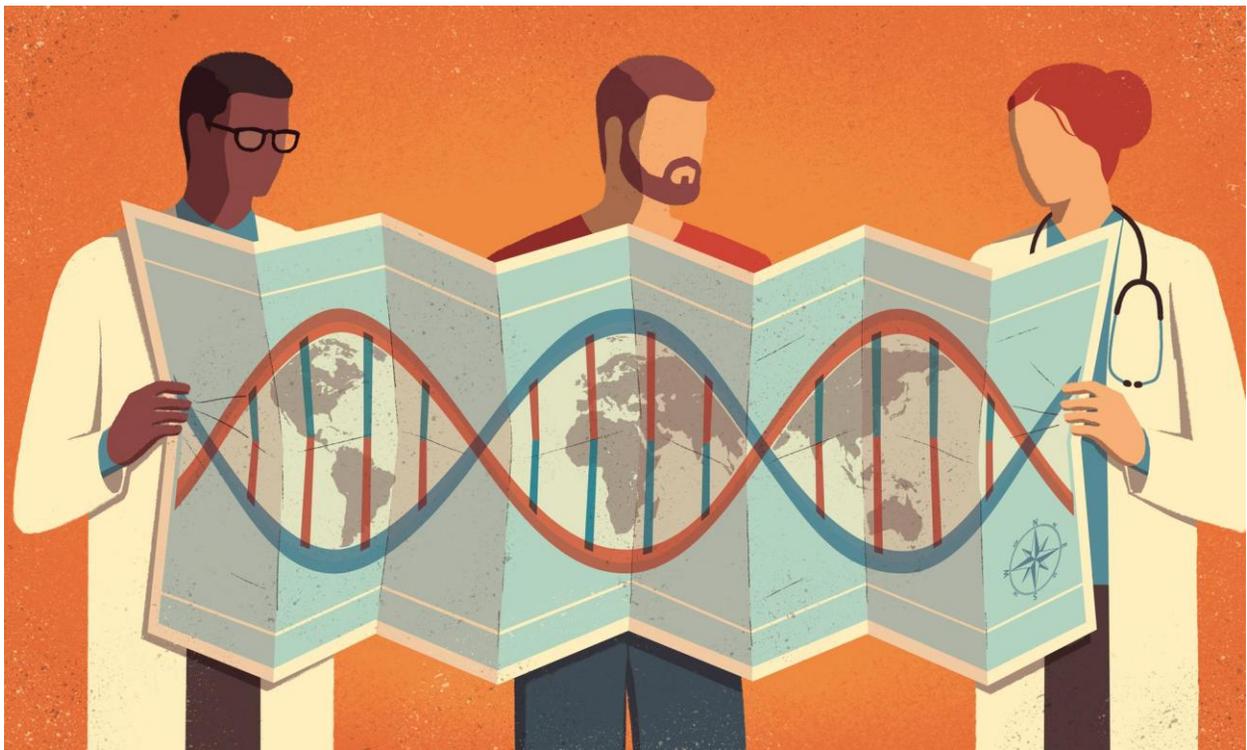# Genetics

Reading

Packet

5.4 Mendel and His Peas

Learning Objectives
- Describe Mendel's first genetics experiments.

Introduction

Why do you look like your family?

For a long time people understood that traits are passed down through families. The rules of how this worked were unclear, however. <mark>The work of Gregor Mendel was crucial in explaining how traits are passed down to each generation.</mark>

Guided Learning

What is Heredity?

What does the word "inherit" mean? You may have inherited something of value from a grandparent or another family member. <mark>To inherit is to receive something from someone who came before you</mark>. You can inherit objects, but you can also inherit traits. For example, you can inherit a parent's eye color, hair color, or even the shape of your nose and ears! **Heredity** <mark>is the passing of traits from parent to offspring.</mark>

<mark>**Genetics** is the study of heredity. The field of genetics seeks to explain how traits are passed on from one generation to the next</mark>.

Gregor Mendel

<mark>In the late 1850s, an Austrian monk named Gregor Mendel (**Figure** below) performed the first genetics experiments. Because of his work, Mendel is considered the "Father of Genetics</mark>."

¹

Gregor Mendel

Mendel studied the inheritance patterns for many different **traits**, or forms of characteristics, in pea plants, including round seeds versus wrinkled seeds, white flowers versus purple flowers, and tall plants versus short plants. He noticed that sometimes a trait would be present in one generation of pea plants but would disappear in the offspring of those plants. Mendel performed a variety of experiments on pea plants to try to understand what was happening. Pea plants were a great organism for Mendel to study because they have easily identifiable traits (**Figure** below). For example, pea plants are either tall or short, which is an easy trait to observe. Furthermore, pea plants grow quickly, so he could complete many experiments in a short period of time.

Seed		Flower	Pod		Stem	
Form	Cotyledon	Color	Form	Color	Place	Size
Round	Yellow	White	Full	Green	Axial pods	Tall
Wrinkled	Green	Violet	Constricted	Yellow	Terminal pods	Short
1	2	3	4	5	6	7

Characteristics of pea plants.

Pea plants were also a good choice for study because they can either self-pollinate or be cross-pollinated. **Self-pollination** means that only one flower is involved; the flower's pollen can fertilize its own ovule, or egg. **Cross pollination** occurs when pollen from one flower fertilizes the ovule of another plant. As a result, one plant's sex cells combine with another plant's sex cells. This is called a "cross." These crosses produce **offspring** (or "children"), just like when male and female animals mate. Since Mendel could move pollen between plants, he could carefully control and then observe the results of crosses between two different types of plants.

Mendel's Experiments

In one of Mendel's early experiments, he crossed a short plant and a tall plant. What do you predict the offspring of these plants were? Medium-sized plants? Most people during Mendel's time would have said medium-sized. But an unexpected result occurred.

Mendel observed that the offspring of this cross (called the **F1 generation**) were all tall plants! Next, Mendel let the F1 generation self-pollinate. That means the tall plant offspring were crossed with

each other. He found that most of their offspring (the **F2 generation**) were tall, but a few were short. Shortness skipped the F1 generation, but reappeared in the F2 generation!

Mendel then repeated the same experiment for purple flowered-plants and white flowered-plants. When he crossed the purple and white flowered plants, do you think the colors blended? No, they did not. Just like the previous experiment, all offspring in this cross (the F1 generation) were one color: purple. In the F2 generation, most of plants had purple flowers, and a few had white flowers. There was no blending of traits in any of Mendel's experiments. In all, Mendel studied seven characteristics, with almost 20,000 F2 plants analyzed. All of his results were similar to the first experiment —about three out of every four plants had one trait, while just one out of every four plants had the other. Mendel called the trait that was seen in the F1 generation a **dominant trait**. The trait that disappeared in the F1 generation and then reappeared was called a **recessive trait**.

Review

- Gregor Mendel was the father of the field of genetics, which seeks to explain how traits are passed on from one generation to the next.
- To study genetics, Mendel chose to work with pea plants because they have easily identifiable traits.
- Mendel discovered that some traits, which he called recessive, can skip a generation while other traits, which he called dominant, are present in all generations.

Vocabulary

Cross-Pollination

Movement of pollen from one flower to the stigma of another so that one plant's sex cells combine with another plant's sex cells.

Dominant Trait

Trait that was present in the F1 generation.

F1 Generation

Offspring of a cross between individuals that have contrasting traits.

F2 Generation

Offspring from the self-pollination of the F1 generation.

Genetics

Study of heredity.

Heredity

The passing of traits from parent to offspring.

Offspring

Result of a reproductive process; children.

Self-Pollination

Fertilization that occurs when a flower's pollen lands on it's own stigma and pollinates itself.
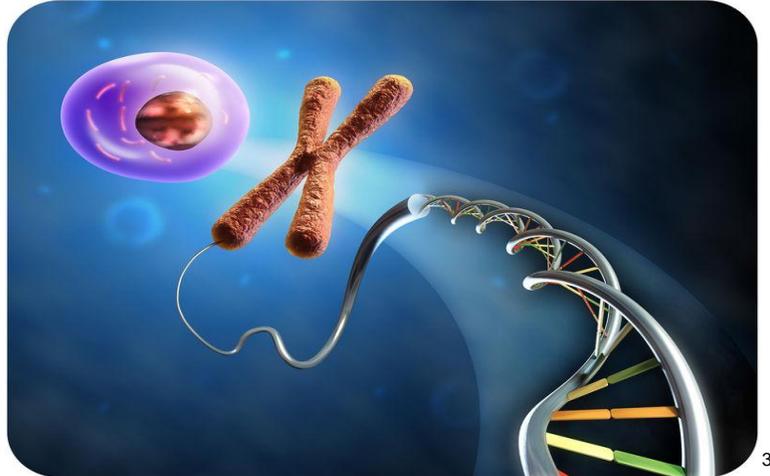
Recessive Trait

Trait that is hidden in the F1 generation.

Trait

The different forms of a characteristic in a population, such as brown hair or red hair.

5.5 Modern Genetics

Learning Objectives

- Compare Mendel's experiments with our modern understanding of genes.
- Explain how genes and alleles are related to an organism's genotype.
- Explain how an organism's genotype is related to its phenotype.

Introduction

Did Mendel know about DNA?

No, people did not understand that DNA is our hereditary material until long after Mendel's time. Our modern understanding of DNA and chromosomes helped to explain how Mendel's rules worked.

Guided Learning

Unraveling the Mystery

Mendel laid the foundation for modern genetics, but there were still a lot of questions he left unanswered. What exactly are the dominant and recessive factors that determine how all organisms look? And how do these factors work?

Since Mendel's time, scientists have discovered the answers to these questions. Genetic material is made out of **DNA**. It is the DNA that makes up the hereditary factors that Mendel identified. By applying our modern knowledge of DNA and chromosomes, we can explain Mendel's findings and build on them. In this concept, we will explore the connections between Mendel's work and modern genetics.

Traits, Genes, and Alleles

Recall that our DNA is wound into **chromosomes**. Each of our chromosomes contains a long chain of DNA that encodes hundreds, if not thousands, of genes. A **gene** codes for a specific characteristic in an organism. Each of these genes can have slightly different versions from individual to individual. These different forms of genes are called **alleles**.

In describing genetic crosses, letters are used to represent alleles. For example, remember that for the height gene in pea plants there are two possible factors. These factors are alleles. The **dominant** allele is represented by a capital letter (*T* for tall) while the **recessive** allele is represented by a lowercase letter (*t*).

Genotype and Phenotype

Genotype is a way to describe the combination of alleles that an individual has for a certain gene (**Table** below). For each gene, an organism has two alleles, one from Mom and one from Dad. The genotype is represented by letter combinations, such as *TT*, *Tt*, and *tt*.

When an organism has two of the same alleles for a specific gene, it is **homozygous** (homo- means "same") for that gene. An organism can be either homozygous dominant (*TT*) or homozygous recessive (*tt*). If an organism has two different alleles (*Tt*) for a certain gene, it is known as **heterozygous** (hetero- means different).

Example	Description	Definition
TT or tt	homozygous	two of the same allele
Tt	heterozygous	one dominant allele and one recessive allele
TT	homozygous dominant	two dominant alleles
tt	homozygous recessive	two recessive alleles

Phenotype is a way to describe the traits you can see. The genotype is like a recipe for a cake, while the phenotype is like the cake made from the recipe. The genotype expresses the phenotype. For example, the phenotypes of Mendel's pea plants were either tall or short, or they were purple-flowered or white-

flowered.

<mark>Can organisms with different genotypes have the same phenotypes? Let's see.</mark>

<mark>Recall that, in his experiments, Mendel crossed plants with two contrasting traits. For example, he crossed a tall plant with a short plant. We now know that the tall plant had a genotype of *TT* and the short plant had a genotype of *tt*. The offspring receives one allele from the tall parent (*T*) and one allele from the short parent (*t*), resulting in a genotype of *Tt* for the F1 generation. Also recall that the F1 generation expressed only the dominant phenotype; all of the plants were tall. In this case, the genotypes of *TT* and *Tt* both result in the tall phenotype because the dominant allele masks the recessive allele when they are present together. So yes, different genotypes can result in the same phenotype. Remember, though, that the recessive phenotype will only be expressed when the dominant allele is absent, or when an individual is homozygous recessive (*tt*)</mark>

Environmental Influences

Sometimes, even though your genes code for a specific trait, your environment or lifestyle choices can alter a person's phenotype.<mark> For instance, your DNA may code for blonde hair. If you choose to dye your hair brown, you are altering the color of your hair through outside chemicals, *not* through your DNA. Another example would be if your DNA puts you at risk for heart disease</mark>. Through diet and exercise, you can minimize that risk.

Review

- Mendel's hereditary "factors" are variants of genes called alleles.
- Genotype describes the combination of alleles that an individual has for a certain gene, while phenotype describes the traits that you can see.

Vocabulary

Alleles

Different forms of a gene.

Chromosome

Structure composed of DNA wrapped around proteins.

DNA (deoxyribonucleic acid)

Hereditary material of a cell.

Dominant Trait

Trait that is expressed if at least one copy of the allele is present. Represented by a capital letter.

Gene

A portion of DNA that codes for a specific trait.

Genotype

The combination of alleles an individual has for a certain gene. The alleles are inherited from both parents and code for a phenotype.

Homozygous

Having two of the same alleles for a specific gene.

Heterozygous

Having two different alleles for a specific gene.

Phenotype

An organism's appearance as coded for by the genotype.

Recessive Trait

Trait that is expressed only when two copies of the allele (one contributed by each parent) are present; the recessive trait is hidden when the dominant allele is present, such as in the F1 generation. Represented by a lowercase letter.

5.6 Probability and Punnett Squares

Learning Objectives

- Draw a Punnett square to make predictions about the traits of the offspring of a simple genetic cross.

Introduction

What's the chance of the coin landing on heads?

Probability is the likelihood that an event will occur. For instance, there is always a 50-50 chance that a coin will land on heads. Half the time it will land on heads and half the time it will land on tails. These rules

---

of probability also apply to genetics. If a parent has one dominant and one recessive factor for a trait, than half the time the dominant factor will be passed on, and half the time the recessive factor will be passed on.

Guided Learning

Probability and Punnett Squares

A **Punnett square** is a special tool derived from the laws of probability. It is used to predict the offspring from a cross, or mating between two parents.

An example of a Punnett square (**Figure** below) shows the results of a cross between two heterozygous purple flowers (*Bb*).
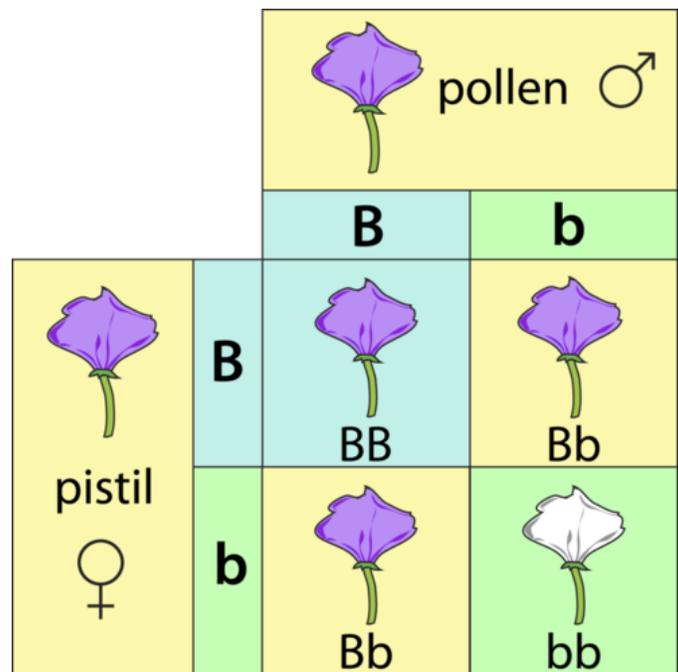
The Punnett square of a cross between two purple flowers (Bb).

To create a Punnett square, perform the following steps:
1. Take the factors from the first parent and place them at the top of the square (*B* and *b*).
2. Take the factors from the second parent and line them up on the left side of the square (*B* and *b*).
3. Pull the factors from the top into the boxes below.
4. Pull the factors from the side into the boxes next to them.

The possible offspring are represented by the letters in the boxes, with one factor coming from each

parent.

Predicted Results from Punnett square:

- Top left box: *BB*, or Purple flowers
- Top right box: *Bb*, or Purple flowers
- Lower left box: *Bb*, or Purple flowers
- Lower right box: *bb*, or White flowers

Only one out of the four, or 25% of the boxes, shows white flowers (*bb*). The other 75% have purple flowers (*BB*, *Bb*), because the purple factor (*B*) is the dominant factor.

Remember that Punnett squares are used to *predict* results of a cross. *Actual* results may differ slightly from those predicted through using a Punnett square, just as actual results from flipping a coin may differ slightly from the prediction or probability of getting half heads and half tails.

Now imagine you cross one of the white flowers (*bb*) with a purple flower that is heterozygous (*Bb*). The only possible allele in the white flower is recessive (*b*), while the purple flower can pass on either dominant (*B*) or recessive (*b*) alleles.

Practice using a Punnett square with this cross (see **Table** below).

	b	b
B		
b		

Did you find that 50% of the offspring will be purple, and 50% of the offspring will be white?

Review

- A Punnett square is a special tool used to predict the offspring from a cross, or mating between two parents.
- In a Punnett square, the parent genotypes are shown outside the boxes and the possible offspring are represented by the letters in the boxes, with one factor coming from each parent.
- Each box in a Punnet square represents one out of four possible offspring, or 25%.
- The predicted genotypes and phenotypes may not match actual genotypes and phenotypes of a cross.

Vocabulary

Probability

> The likelihood of something happening.

Punnett square

> Chart that is used to predict the outcome of a particular cross.

5.7 Exceptions to Mendel's Rules

Learning Objectives

- Describe how some patterns of inheritance violate Mendel's rules.

Introduction

Are all people either short or tall?

Unlike Mendel's peas, people do not all fall into two categories: short or tall. Most people, in fact, are somewhere in between. This is true for many human traits. In some cases, Mendel's rules are too simple to explain the inheritance of traits. Each characteristic Mendel investigated was controlled by one gene that had only two possible alleles, one of which was completely dominant over the other. We now know that inheritance is often more complicated than this. In blood types, for example, there are actually three alleles instead of two.

Guided Learning

Exceptions to Mendel's Rules

In all of Mendel's experiments, he worked with traits where a single gene controlled the trait. Each also had one allele that was always dominant over the recessive allele. But this is not always true. There are exceptions to Mendel's rules, and these exceptions usually have something to do with the dominant allele. If you cross a homozygous red flower with a homozygous white flower, according to Mendel's laws, what color flower should result from the cross? Either a completely red or completely white flower, depending on which allele is dominant. But since Mendel's time, scientists have discovered this is not always the case.

Incomplete Dominance

<mark>One allele is **not** always completely dominant over another allele. Sometimes an individual has a phenotype between the two parents because one allele is not dominant over another. This pattern of inheritance is called **incomplete dominance**</mark>. For example, snapdragon flowers show incomplete dominance. One of the genes for flower color in snapdragons has two alleles, one for red flowers and one for white flowers.

A plant that is homozygous for the red allele (*RR*) will have red flowers, while a plant that is homozygous for the white allele will have white flowers (*WW*). But the heterozygote will have pink flowers (*RW*) (**Figure** below). Neither the red nor the white allele is dominant, so the phenotype of the offspring is a blend of the two parents.

Pink snapdragons are an example of incomplete dominance.

Codominance and Multiple Alleles

<mark>Another exception to Mendel's laws is a phenomenon called **codominance**</mark>. For example, our blood type shows codominance. Do you know what your blood type is? Are you A? B? O? AB? Those letters actually represent alleles. Unlike other traits, your blood type has three alleles (A, B, and O), instead of two! The A allele is dominant, as is the B allele. They are both dominant over the allele that codes for type O blood, but when they are present together (AB), <mark>they are both expressed. This is called codominance because the prefix *co-* means *both or together*. This pattern of inheritance is significantly different than Mendel's rules for inheritance, because both alleles are expressed completely, and one does not mask</mark>

---

6 *Incomplete Dominance* by ROCHELLE HARTMAN / CK-12 / CC-BY-SA 3.0.

Polygenic Traits

Another exception to Mendel's rules is **polygenic inheritance**, which occurs when a trait is controlled by more than one gene. This means that each dominant allele "adds" to the expression of the next dominant allele.

Usually, traits are polygenic when there is wide variation in the trait. For example, humans can be many different sizes. Height is a polygenic trait, controlled by at least three genes with six alleles. If you are dominant for all of the alleles for height, then you will be very tall. There is also a wide range of skin color across people. Skin color is also a polygenic trait.

Sex-Linked Traits

One special pattern of inheritance that doesn't fit Mendel's rules is sex-linked inheritance, referring to the inheritance of traits that are located on genes on the sex chromosomes. Since males and females do not have the same sex chromosomes, there will be differences between genders in how these **sex-linked traits**—traits linked to genes located on the sex chromosomes—are expressed.

One example of a sex-linked trait is red-green colorblindness. People with this type of color blindness cannot tell the difference between red and green. They often see these colors as shades of brown (**Figure** below). Boys are much more likely to be colorblind than girls. This is because colorblindness is a sex-linked, recessive trait, and girls would need to inherit two copies of the gene whereas boys only need to inherit one.

Boys only have one X chromosome, so if that chromosome carries the gene for color blindness, they will be colorblind. As girls have two X chromosomes, a girl can have one X chromosome with the colorblind gene and one X chromosome with a normal gene for color vision. Since colorblindness is recessive, the dominant normal gene will mask the recessive color blind gene. Females with one color blindness allele and one normal allele are referred to as **carriers**. They carry the allele but do not express it.

How would a female become color-blind? She would have to inherit two genes for color blindness, which is very unlikely. Many sex-linked traits are inherited in a recessive manner.

Asexual Reproduction

Mendel's rules also assume that there are two parents contributing to the genotype of the offspring. In **asexual reproduction**, however, there is only one parent, and an exact copy of the parent's DNA is passed onto the offspring. As a result, the offspring are **clones** of the parent. A clone is an organism that has identical DNA to another individual.

Review

- In polygenic inheritance, a trait is controlled by more than one gene.
- Examples of polygenic inheritance include height or skin color.
- Incomplete dominance, as seen in snapdragon flower color, is a form of inheritance in which one allele is only partly dominant over the other allele, resulting in an intermediate phenotype.
- Codominance, as in human blood type, is a form of inheritance in which both alleles are expressed equally in the phenotype of the heterozygote.
- Organisms that reproduce asexually pass on all of their DNA, and therefore all of their traits, onto their offspring.
- Each individual has two sex chromosomes; females have two X chromosomes (XX), while males have one X chromosome and one Y chromosome (XY).
- Sex-linked traits are located on genes on the sex chromosomes.

Vocabulary

Asexual Reproduction

Reproduction in which there is only one parent and all genetic material is passed on to offspring.

Carrier

Person who carries the recessive allele for a trait but does not express the trait.

Clone

An exact copy of another organism; contains identical DNA.

Codominance

Form of inheritance in which two alleles are expressed equally in the phenotype of the heterozygote.

Incomplete Dominance

Form of inheritance in which one allele for a specific trait is not completely dominant over the other allele, resulting in an intermediate phenotype.

Polygenic Trait

Trait that is controlled by more than one gene.

Sex-Linked Trait

Trait linked to genes located on the sex chromosomes.