# Modern Genetics

**Say Thanks to the Authors**
Click http://www.ck12.org/saythanks
*(No sign in required)*

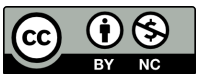
To access a customizable version of this book, as well as other interactive content, visit www.ck12.org

CK-12 Foundation is a non-profit organization with a mission to reduce the cost of textbook materials for the K-12 market both in the U.S. and worldwide. Using an open-content, web-based collaborative model termed the **FlexBook®**, CK-12 intends to pioneer the generation and distribution of high-quality educational content that will serve both as core text as well as provide an adaptive environment for learning, powered through the **FlexBook Platform®**.



Printed: August 11, 2014

flexbook
next generation textbooks



CHAPTER 1

# Modern Genetics

## Lesson Objectives

- Compare Mendel's laws with our modern understanding of chromosomes.
- Explain how codominant traits are inherited.
- Distinguish between phenotype and genotype.
- Explain how polygenic traits are inherited.

## Check Your Understanding

- What is a visual representation of a genetic cross?
- What is stated in Mendel's law of segregation?

## Vocabulary

- codominance
- genotype
- heterozygous
- homozygous
- phenotype
- polygenic inheritance

## Mendel and Modern Genetics

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 lesson, we will explore the other 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. Each of these genes can have slightly different versions from individual to individual. These variants of genes are called alleles.

For example, remember that for the height gene in pea plants there are two possible factors. These factors are alleles. There is a dominant allele for tallness ($T$) and a recessive allele for shortness ($t$).

## Genotype and Phenotype

**Genotype** is a way to describe the combination of alleles that an individual has for a certain gene ( **Table** 1.1). For each gene, an organism has two alleles, one on each chromosome of a homologous pair of chromosomes (think of it as one allele from mom, one allele 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).

**TABLE 1.1: Genotypes**

| genotype | definition | example |
|---|---|---|
| homozygous | two of the same allele | *TT* or *tt* |
| heterozygous | one dominant allele and one recessive allele | *Tt* |
| homozygous dominant | two dominant alleles | *TT* |
| homozygous recessive | two recessive alleles | *tt* |

**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 were purple-flowered or white-flowered.

Can organisms with different genotypes have the same phenotypes? Let's see.

What is the phenotype of a pea plant that is homozygous dominant (*TT*) for the tall trait? Tall. What is the phenotype of a pea plant that is heterozygous (Tt)? It is also tall. The answer is yes, two different genotypes can result in the same phenotype. Remember, the recessive phenotype will be expressed only when the dominant allele is absent, or when an individual is homozygous recessive (tt) ( **Figure** 1.1).

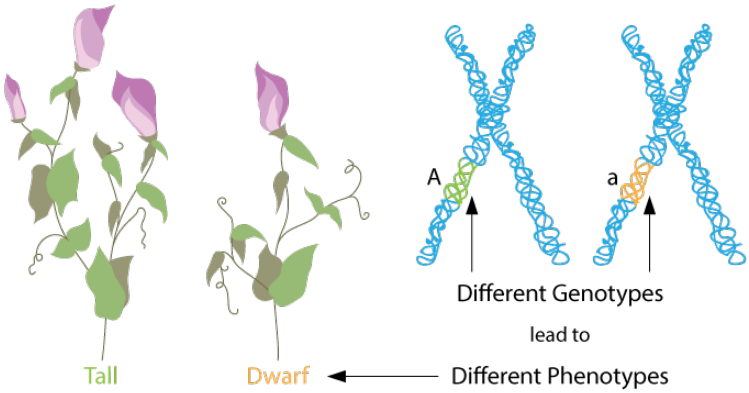



FIGURE 1.1

Different genotypes (AA, Aa, aa or TT, Tt, tt) will lead to different phenotypes, or different appearances of the organism

## Exceptions to Mendel's Laws: Incomplete Dominance and Codominance

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 to 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

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.** 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** 1.2). Neither the red nor the white allele is dominant, so the phenotype of the offspring is a blend of the two parents.

FIGURE 1.2

Pink snapdragons are an example of incomplete dominance.

Another example of incomplete dominance is sickle cell anemia, a disease in which a blood protein called hemoglobin is produced incorrectly, causing the red blood cells to have a sickle shape. A person that is homozygous recessive (ss) for the sickle cell trait will have red blood cells that all have the incorrect hemoglobin. A person who is homozygous dominant (SS) will have normal red blood cells.

What type of blood cells do you think a person who is heterozygous (Ss) for the trait will have? They will have some misshapen cells and some normal cells ( **Figure** 1.3). Both the dominant and recessive alleles are expressed, so the result is a phenotype that is a combination of the recessive and dominant traits.

## Codominance

Another exception to Mendel's laws is a called **codominance.** For example, our blood type shows codominance. Do you know what your blood type is? Are you A? O? AB? Those letters actually represent alleles. Unlike other traits, your blood type has 3 alleles, instead of 2!

The ABO blood types ( **Figure** 1.4) are named for the protein, or antigen, attached to the outside of the blood cell. An antigen is a substance that provokes an immune response, your body's defenses against disease, which will be discussed further in the *Diseases and the Body's Defenses* chapter.

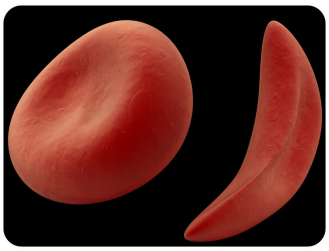


FIGURE 1.3

Sickle cell anemia causes red blood cells to become misshapen and curved (right cell) unlike normal, rounded red blood cells (left cell).

In this case, two alleles are dominant and completely expressed ($I^A$ and $I^B$), while one allele is recessive ($i$). The $I^A$ allele encodes for red blood cells with the A antigen, while the $I^B$ allele encodes for red blood cells with the B antigen. The recessive allele ($i$) doesn't encode for any proteins. Therefore a person with two recessive alleles ($ii$) has type O blood. As no dominant ($I^A$ and $I^B$) allele is present, the person cannot have type A or type B blood.

There are two possible genotypes for type A blood, homozygous ($I^AI^A$) and heterozygous ($I^Ai$), and two possible genotypes for type B blood, ($I^BI^B$ and $I^Bi$). If a person is heterozygous for both the $I^A$ and $I^B$ alleles, they will express both and have type AB blood with both antigens on each red blood cell.

This pattern of inheritance is significantly different than Mendel's rules for inheritance because both alleles are expressed completely and one does not mask the other.

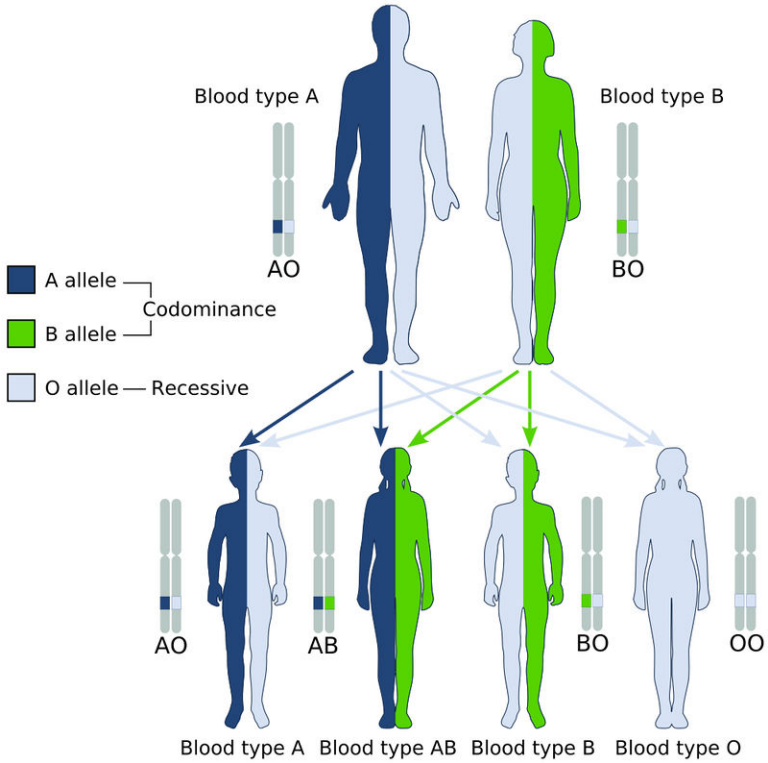



FIGURE 1.4

An example of codominant inheritance is ABO blood types.

## Polygenic Traits and Environmental Influences

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. 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.

Polygenic inheritance often results in a bell shaped curve when you analyze the population ( **Figure** 1.5). That means that most people fall in the middle of the phenotypic range, such as average height, while very few people are at the extremes, such as very tall or very short.

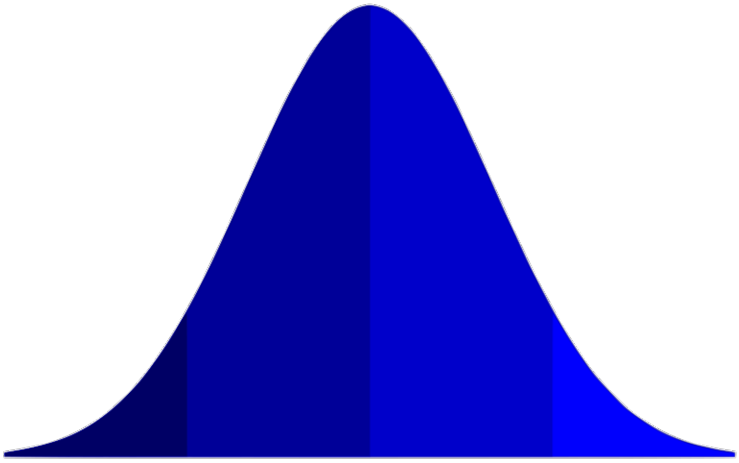


FIGURE 1.5

Polygenic traits tend to result in a distribution that resembles a bell-shaped curve, with few at the extremes and most in the middle. There may be 4 or 6 or more alleles involved in the phenotype. At the left extreme, individuals are completely dominant for all alleles, and at the other extreme, individuals are completely recessive for all alleles. Individuals in the middle have various combinations of recessive and dominant alleles.

## Lesson Summary

- Variants of genes are called alleles.
- Genotype is the combination of alleles that an individual has for a certain gene, while phenotype is the appearance caused by the expression of the genotype.
- Incomplete dominance and codominance do not fit Mendel's rules because one allele does not entirely mask the other.
- In polygenic inheritance, many genes control a trait. Each dominant allele adds to the next dominant allele.

## Review Questions

### Recall

1. What is a variant of a gene that occurs at the same place on homologous chromosomes?
2. What is the type of allele that only affects the phenotype in the homozygous condition?
3. What type of allele masks the expression of the recessive allele?

4. What is the term for the specific alleles of an individual for a particular trait?

5. What is the term for the appearance of the organism, as determined by the genotype?

### Apply Concepts

6. If two individuals have a certain phenotype, such as tall pea plants, does that mean they must have the same genotype?

7. What is the term for the pattern of inheritance where an individual has an intermediate phenotype between the two parents?

8. What is the inheritance pattern where both alleles are expressed?

### Think Critically

9. IQ in humans varies, with most people having an IQ of around 100, and with a few people at the extremes, such as 50 or 150. What type of inheritance do you think this might describe?

10. A dark purple flower is crossed with a white flower of the same species and the offspring have light purple flowers. What type of inheritance does this describe? Explain.

## Further Reading / Supplemental Links

- http://staff.jccc.net/pdecell/evolution/polygen.html
- http://www.estrellamountain.edu/faculty/farabee/BIOBK/BioBookgenintro.html
- http://www.physorg.com/news188148947.html/

## Points to Consider

- Hypothesize about the genetic differences between males and females.
- Can you name any human genetic disorders?
- If a baby inherits an extra chromosome, what might the result be?

## References

1. Zachary Wilson. CK-12 Foundation . CC BY-NC 3.0
2. Rochelle Hartman. http://www.flickr.com/photos/tinfoilraccoon/1463287867/ . CC BY 2.0
3. Image copyright Sebastian Kaulitzk, 2013. http://www.shutterstock.com . Used under license from Shutterstock.com
4. User:YassineMrabet/Wikimedia Commons, based on image by the NIH. http://commons.wikimedia.org/wiki/File:ABO_system_codominance.svg . Public Domain
5. David Remahl, User:Nae'blis/Wikimedia Commons. http://commons.wikimedia.org/wiki/File:Bellcurve.svg . The copyright holder of this work allows anyone to use it for any purpose including unrestricted redistribution, commercial use, and modification