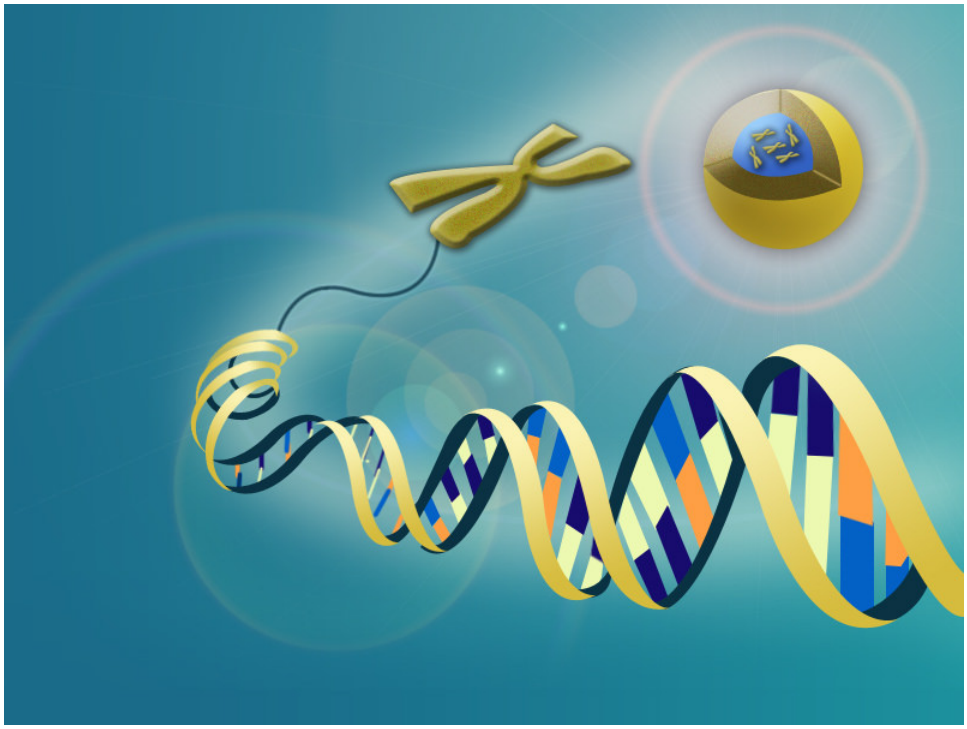
CHAPTER 4 Instructional Segment 4

Chapter Outline

4.1 Molecular Genetics Overview

Learning Objectives

- Define allele.
- Compare heterozygous to homozygous.
- Distinguish genotype from phenotype.
- Compare Mendel's laws with the modern understanding of chromosomes.



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.

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 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. Each of these genes can have slightly different versions from individual to individual. These variants of genes are called **alleles**. Each parent only donates one allele for each gene to an offspring.

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** 4.1). 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 4.1: 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 they 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** 4.1).

Summary

- 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.

Explore More

Use the resources below to answer the questions that follow.

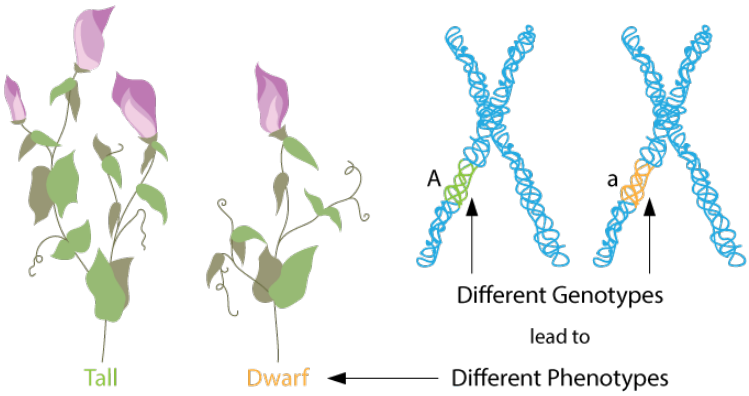


FIGURE 4.1

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

Explore More I

- **Link Between Genotype and Phenotype** at http://www.sciencelearn.org.nz/Contexts/Uniquely-Me/Sci-Media/Video/Researching-the-link-between-genotype-and-phenotype

1. When geneticists look at genotype, what are they really studying?
2. Why do geneticists like to turn genes off? What question(s) do they ask?

Explore More II

- **iPlant Genotype to Phenotype** at http://www.youtube.com/watch?v=nIh0Qy_CZsU (3:49)

MEDIA

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1. Do most of the complex phenotypes we observe come from a single gene?
2. What has led to the rapid analysis of DNA? Where do scientists now hope to apply these tools?
3. What are some of the phenotypic plant traits that scientists are investigating? Why do you think these traits were chosen?

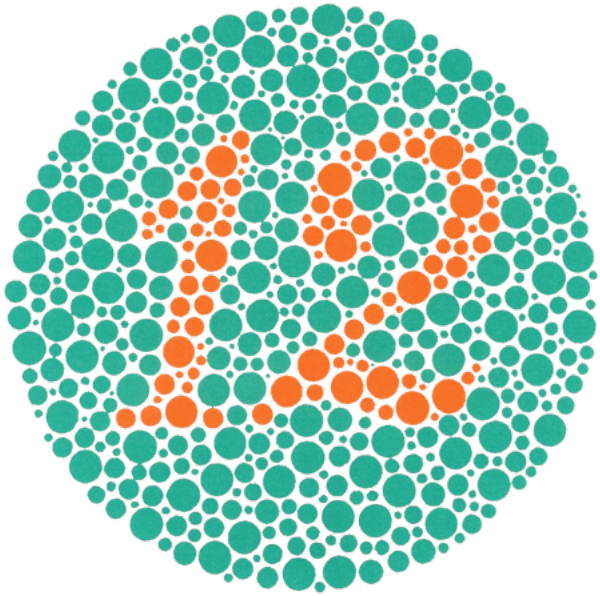
Review

1. What is an allele?
2. What is the type of allele that only affects the phenotype in the homozygous condition?
3. If two individuals have a certain phenotype, does that mean they must have the same genotype?
4. A tall, green plant is homozygous for each trait. If *T* is the tall allele, and *G* is the green allele, what is the genotype and the phenotype of this plant?

4.2 Mendelian Inheritance

Learning Objectives

- Define genetic trait.
- Distinguish autosomal traits from X-linked traits.
- Use a pedigree to determine the mode of inheritance.
- Summarize the inheritance of red-green color blindness.



What number can you see?

Red-green colorblindness is a common inherited trait in humans. About 1 in 10 men have some form of color blindness, however, very few women are color blind. Why?

Mendelian Inheritance in Humans

Characteristics that are encoded in DNA are called **genetic traits**. Different types of human traits are inherited in different ways. Some human traits have simple inheritance patterns like the traits that Gregor Mendel studied in pea plants. Other human traits have more complex inheritance patterns.

Mendelian inheritance refers to the inheritance of traits controlled by a single gene with two alleles, one of which may be dominant to the other. Not many human traits are controlled by a single gene with two alleles, but they are

a good starting point for understanding human heredity. How Mendelian traits are inherited depends on whether the traits are controlled by genes on autosomes or the X chromosome.

Autosomal Traits

Autosomal traits are controlled by genes on one of the 22 human autosomes. Consider earlobe attachment. A single autosomal gene with two alleles determines whether you have attached earlobes or free-hanging earlobes. The allele for free-hanging earlobes (F) is dominant to the allele for attached earlobes (f). Other single-gene autosomal traits include widow's peak and hitchhiker's thumb. The dominant and recessive forms of these traits are shown in **Figure** 4.2. Which form of these traits do you have? What are your possible genotypes for the traits?

The chart in **Figure** 4.2 is called a **pedigree**. It shows how the earlobe trait was passed from generation to generation within a family. Pedigrees are useful tools for studying inheritance patterns.

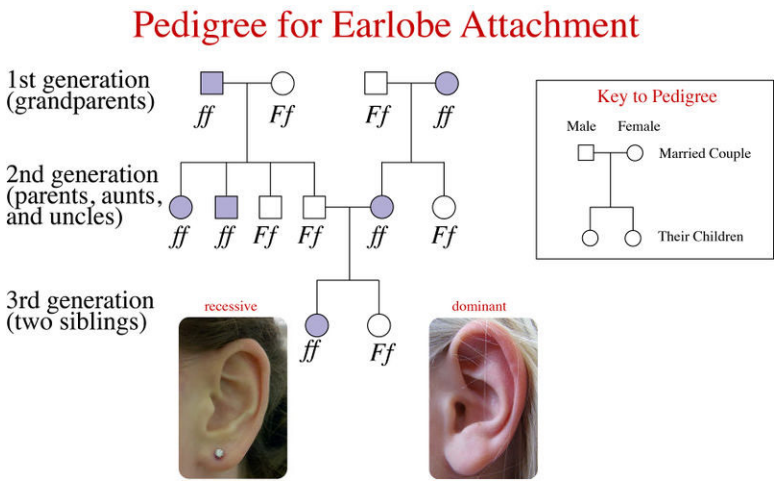


FIGURE 4.2

Having free-hanging earlobes is an autosomal dominant trait. This figure shows the trait and how it was inherited in a family over three generations. Shading indicates people who have the recessive form of the trait. Look at (or feel) your own earlobes. Which form of the trait do you have? Can you tell which genotype you have?

Other single-gene autosomal traits include widow's peak and hitchhiker's thumb. The dominant and recessive forms of these traits are shown in **Figure** 4.3. Which form of these traits do you have? What are your possible genotypes for the traits?

Sex-Linked Traits

Traits controlled by genes on the sex chromosomes are called **sex-linked traits**, or **X-linked traits** in the case of the X chromosome. Single-gene X-linked traits have a different pattern of inheritance than single-gene autosomal traits. Do you know why? It's because males have just one X chromosome. In addition, they always inherit their X chromosome from their mother, and they pass it on to all their daughters but none of their sons. This is illustrated in **Figure** 4.4.

Because males have just one X chromosome, they have only one allele for any X-linked trait. Therefore, a recessive X-linked allele is always expressed in males. Because females have two X chromosomes, they have two alleles for any X-linked trait. Therefore, they must inherit two copies of the recessive allele to express the recessive trait. This explains why X-linked recessive traits are less common in females than males. An example of a recessive X-linked trait is **red-green color blindness**. People with this trait cannot distinguish between the colors red and green. More than one recessive gene on the X chromosome codes for this trait, which is fairly common in males but relatively rare in females (**Figure** 4.5).

FIGURE 4.3
Widow's peak and hitchhiker's thumb are dominant traits controlled by a single autosomal gene.

Summary

- A minority of human traits are controlled by single genes with two alleles.
- They have different inheritance patterns depending on whether they are controlled by autosomal or X-linked genes.

Review

1. Describe the inheritance pattern for a single-gene autosomal dominant trait, such as free-hanging earlobes.
2. Draw a pedigree for hitchhiker's thumb. Your pedigree should cover at least two generations and include both dominant and recessive forms of the trait. Label the pedigree with genotypes, using the letter *H* to represent the dominant allele for the trait and the letter *h* to represent the recessive allele.
3. Why is a recessive X-linked allele always expressed in males?
4. What is necessary for a recessive X-linked allele to be expressed in females?
5. What is an example of a recessive X-linked trait?

Resources

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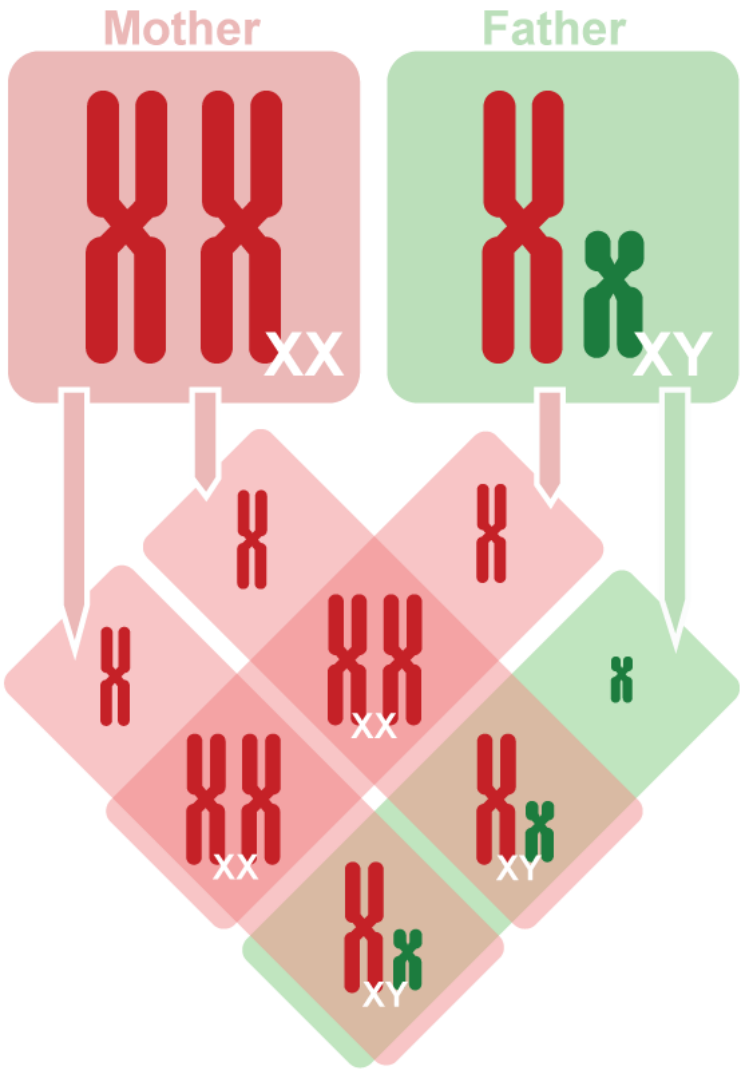


FIGURE 4.4

Inheritance of Sex Chromosomes. Mothers pass only X chromosomes to their children. Fathers always pass their X chromosome to their daughters and their Y chromosome to their sons. Can you explain why fathers always determine the sex of the offspring?

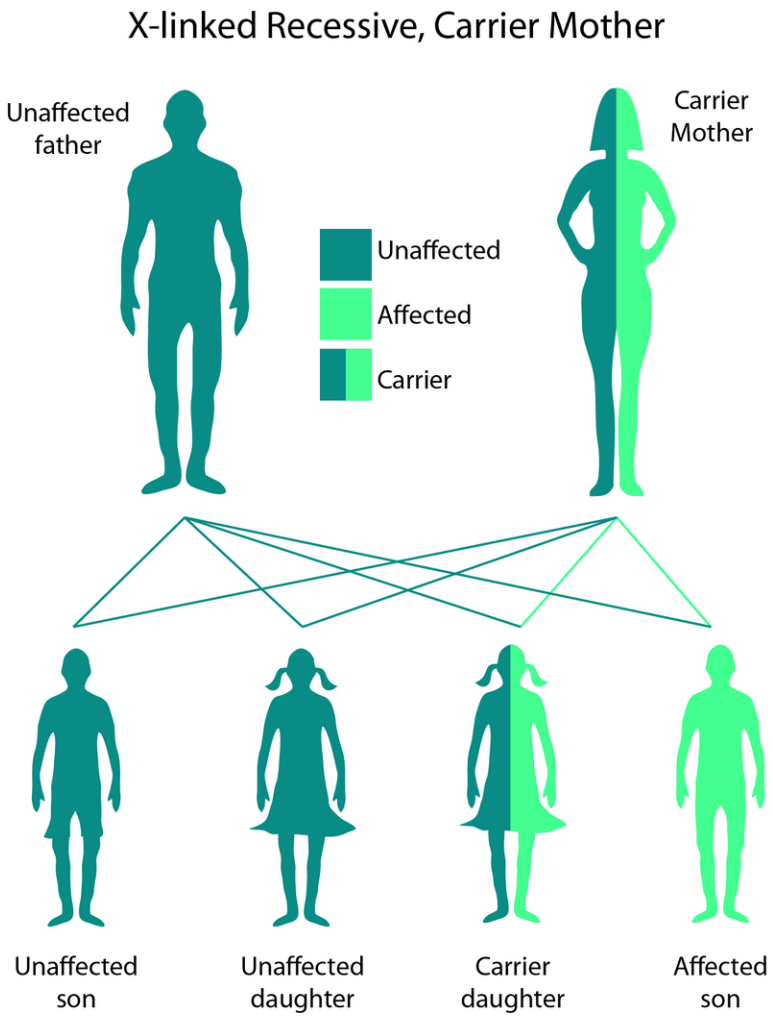


FIGURE 4.5

Pedigree for Color Blindness. Color blindness is an X-linked recessive trait. Mothers pass the recessive allele for the trait to their sons, who pass it to their daughters.

4.3 Mendel's Laws

Learning Objectives

- Distinguish between dominant and recessive traits.
- Explain the law of segregation.

What does it mean to be dominant?

The most powerful or influential individual in a group is sometimes called dominant. In genetics, a dominant trait means nearly the same thing. A dominant trait is the most influential trait and masks the other trait.

Dominance

Do you remember what happened when Mendel crossed purple flowered-plants and white flowered-plants? All the offspring had purple flowers. There was no blending of traits in any of Mendel's experiments. Mendel had to come up with a theory of inheritance to explain his results. He developed a theory called the **law of segregation**.

The Law of Segregation

Mendel proposed that each pea plant had two hereditary factors for each trait. There were two possibilities for each hereditary factor, such as a purple factor or white factor. One factor is **dominant** to the other. The other trait that is masked is called the **recessive** factor, meaning that when both factors are present, only the effects of the dominant factor are noticeable (**Figure** 4.6). Although you have two hereditary factors for each trait, each parent can only pass on one of these factors to the offspring. When the sex cells, or **gametes** (sperm or egg), form, the heredity factors must separate, so there is only one factor per gamete. In other words, the factors are "segregated" in each gamete. Mendel's law of segregation states that the two hereditary factors separate when gametes are formed. When

fertilization occurs, the offspring receive one hereditary factor from each gamete, so the resulting offspring have two factors.

The law of segregation predates our understanding or meiosis. Mendel developed his theories without an understanding of DNA, or even the knowledge that DNA existed. Quite a remarkable feat!

FIGURE 4.6

In peas, purple flowers are dominant to white. If one of these purple flowers is crossed with a white flower, all the offspring will have purple flowers.

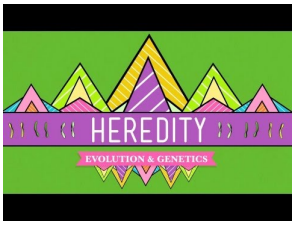
Example Cross

This law explains what Mendel had seen in the F1 generation when a tall plant was crossed with a short plant. The two heredity factors in this case were the short and tall factors. Each individual in the F1 would have one of each factor, and as the tall factor is dominant to the short factor (the recessive factor), all the plants appeared tall.

In describing genetic crosses, letters are used. The dominant factor is represented with a capital letter (*T* for tall) while the recessive factor is represented by a lowercase letter (*t*). For the *T* and *t* factors, three combinations are possible: *TT*, *Tt*, and *tt*. *TT* plants will be tall, while plants with *tt* will be short. Since *T* is dominant to *t*, plants that are *Tt* will be tall because the dominant factor masks the recessive factor.

In this example, we are crossing a *TT* tall plant with a *tt* short plant. As each parent gives one factor to the F1 generation, all of the F1 generation will be *Tt* tall plants.

When the F1 generation (*Tt*) is allowed to self-pollinate, each parent will give one factor (*T* or *t*) to the F2 generation. So the F2 offspring will have four possible combinations of factors: *TT*, *Tt*, *tT*, or *tt*. According to the laws of probability, 25% of the offspring would be *tt*, so they would appear short. And 75% would have at least one *T* factor and would be tall.



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Summary

- One hereditary factor is dominant to the other. The dominant trait masks the recessive factor, so that when both factors are present, only the effects of the dominant factor are noticeable.

- According to Mendel's law of segregation, there are two hereditary factors for each trait that must segregate during gamete (egg and sperm) production. As a result, offspring receive one factor from each parent, resulting in two factors for each trait in the offspring.

Explore More

Use the resource below to answer the questions that follow.

- **Mendel's Experiment** at http://www.sumanasinc.com/webcontent/animations/content/mendel/mendel.html

1. In Mendel's experiments, did it matter if the dominant trait came from the seed plant or the pollen plant?
2. Yellow is a dominant trait in peas. You breed two plants with yellow peas, and some of the offspring's peas are green? How can this be? Explain your answer fully.
3. For some of his experiments Mendel saw a 9:3:3:1 ratio, consisting of 9 yellow/smooth, 3 yellow/wrinkled, 3 green/smooth, and 1 green/wrinkled. What did he conclude from this ratio? Explain where these ratios came from.

Review

1. What is the difference between a dominant trait and a recessive trait?
2. Explain the law of segregation.
3. When Mendel crossed a *TT* tall plant with a *tt* short plant, what did he observe in the F1 generation? Why?
4. If *PP* purple plants are crossed with *pp* white plants, what will be the possible combinations of factors if the F1 generation is allowed to self-pollinate?

4.4 Punnett Squares

Learning Objectives

- Explain the relationship between probability and genetics.
- Use a Punnett square to make predictions about the traits of the offspring of a genetic cross.

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

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. What is the chance of it landing heads twice in a row? Or three times? These rules of probability also apply to genetics.

A parent only gives one factor for each trait to an offspring. If a parent has one dominant and one recessive factor for a trait, then, on average, half the time the dominant factor will be passed on, and half the time the recessive factor will be passed on.

Probability and Punnett Squares

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

An example of a Punnett square (**Figure** 4.7) shows the results of a cross between two purple flowers that each have one dominant factor and one recessive factor (*Bb*).

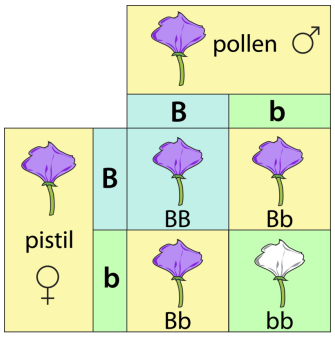


FIGURE 4.7

The Punnett square of a cross between two purple flowers (*Bb*). A Punnett square can be used to calculate what percentage of offspring will have a certain trait.

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.

Results:

- 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 of the plants out of the four, or 25% of the plants, has white flowers (*bb*). The other 75% have purple flowers (*BB*, *Bb*), because the purple factor (*B*) is the dominant factor. This shows that the color purple is the **dominant trait** in pea plants.

Now imagine you cross one of the white flowers (*bb*) with a purple flower that has both a dominant and recessive factor (*Bb*). The only possible gamete in the white flower is recessive (*b*), while the purple flower can have gametes with either dominant (*B*) or recessive (*b*).

Practice using a Punnett square with this cross (see **Table** 4.2).

TABLE 4.2: White Flower (bb) Crossed with Purple Flower (Bb)

	b	**b**
B	**Bb**	**Bb**
b	**bb**	**bb**

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

Summary

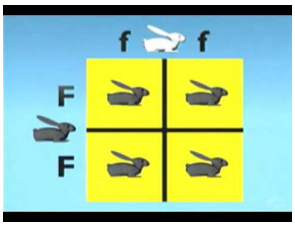
- A Punnett square is a special tool used to predict the offspring from a cross, or mating between two parents.
- In a Punnett square, the possible offspring are represented by the letters in the boxes, with one factor coming from each parent.

Explore More

Use the resources below to answer the questions that follow.

Explore More I

- **Gregor Mendel's Punnett Square** at http://www.youtube.com/watch?v=d4izVAkhMPQ (5:03)



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1. What are Mendel's two rules of heredity?
2. If a pure bred black rabbit and a white rabbit mate, what will their potential offspring look like?
3. If two hybrid *Ff* rabbits mate, what will the offspring look like?

Explore More II

- **Punnett Square Calculator** at http://scienceprimer.com/punnett-square-calculator

1. If you cross an *Aa* individual with another *Aa* individual, what will the genotype ratio be in the next generation? What will be the phenotype ratio?
2. If you cross an *AABb* individual with an *Aabb* individual, what will the genotype ratio be in the next generation? What will be the phenotype ratio?
3. If you cross an *AAbb* individual with an *aabb* individual, what will the genotype ratio be in the next generation? What will be the phenotype ratio?

Review

1. In peas, yellow seeds (*Y*) are dominant over green seeds (*y*). If a *yy* plant is crossed with a *YY* plant, what ratio of plants in the offspring would you predict?
2. What ratio of plants in the offspring would you predict from a *Yy* x *Yy* cross?
3. In guinea pigs, smooth coat (*S*) is dominant over rough coat (*s*). If an *SS* guinea pig is crossed with an *ss* guinea pig, what ratio of guinea pigs in the offspring would you predict?
4. What ratio of guinea pigs in the offspring would you predict from a *Ss* x *ss* cross?

4.5 Sex-linked Inheritance

Learning Objectives

- Define sex-linked trait.
- Explain the genetics of sex determination in humans.
- Explain sex-linked inheritance.

Male or female?

One of the exciting things about expecting a child is wondering if the baby will be a boy or a girl. There are many superstitions about how one might influence or predict the outcome. But what really determines if a baby is male or female? We now know that the gender of a baby is determined by a special pair of chromosomes known as the sex chromosomes.

Sex-linked Inheritance

What determines if a baby is a male or female? Recall that you have 23 pairs of chromosomes—and one of those pairs is the **sex chromosomes**. Everyone has two sex chromosomes. Your sex chromosomes can be X or Y. Females have two X chromosomes (XX), while males have one X chromosome and one Y chromosome (XY).

If a baby inherits an X chromosome from the father and an X chromosome from the mother, what will be the child's sex? The baby will have two X chromosomes, so it will be female. If the father's sperm carries the Y chromosome, the child will be male. Notice that a mother can only pass on an X chromosome, so the sex of the baby is determined by the father. The father has a 50 percent chance of passing on the Y or X chromosome, so there is a 50 percent chance that a child will be male, and there is a 50 percent chance a child will be female. This 50:50 chance occurs for each baby. A couple's first five children could all be boys. The sixth child still has a 50:50 chance of being a girl.

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 the sexes in how these **sex-linked traits**—traits linked to genes

located on the sex chromosomes—are expressed. Sex-linked traits usually refer to traits due to genes on the X chromosome.

One example of a sex-linked trait is red-green colorblindness. People with this type of colorblindness cannot tell the difference between red and green. They often see these colors as shades of brown (**Figure** 4.8). Boys are much more likely to be colorblind than girls (**Table** 4.3). This is because colorblindness is a sex-linked, recessive trait.

Boys only have one X chromosome, so if that chromosome carries the gene for colorblindness, 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 colorblind gene. Females with one colorblindness allele and one normal allele are referred to as **carriers**. They carry the allele but do not express it.

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

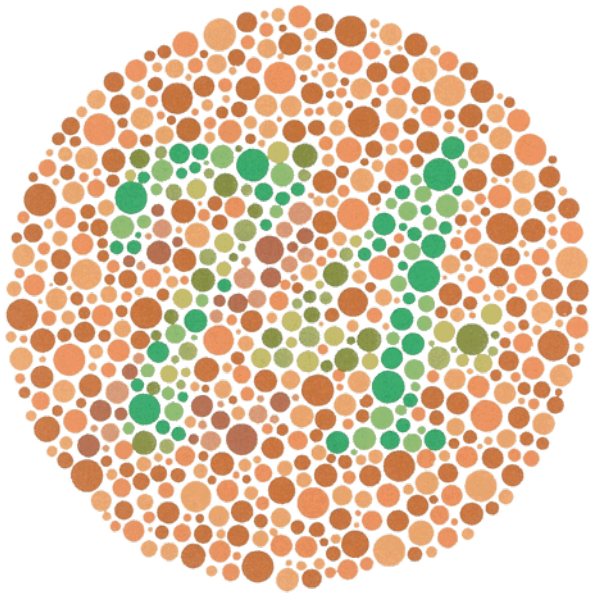


FIGURE 4.8

A person with red-green colorblindness would not be able to see the number.

TABLE 4.3: Cross Between a Female Carrier for Colorblindness and a Male with Normal Vision

	$\mathbf{X}^c$	**X**
X	$\mathbf{X}^c\mathbf{X}$ (carrier female)	**XX** (normal female)
Y	$\mathbf{X}^c\mathbf{Y}$ (colorblind male)	**XY** (normal male)

According to this Punnett square (**Table** 4.3), the son of a woman who carries the colorblindness trait and a male with normal vision has a 50% chance of being colorblind.

Summary

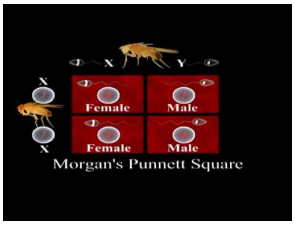
- Each individual has two sex chromosomes; females have two X chromosomes (XX), while males have one X chromosome and one Y chromosome (XY).
- Sex-linked traits are located on genes on the sex chromosomes.

Explore More

Use the resources below to answer the questions that follow.

Explore More I

- **Sex-linked Traits** at http://www.youtube.com/watch?v=H1HaR47Dqfw (5:16)



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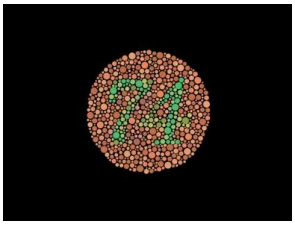
1. What was unusual about the F_2 generations in Morgan's crosses?
2. According to Morgan, where is the gene for eye color located?
3. How did Morgan test his hypothesis on the location of the eye color gene?
4. What are three traits that humans have that are related to genes exclusive to the X-chromosome?

The "Morgan" referred to in the above clip is Thomas Hunt Morgan. You can find out more about him and his work here: http://www.nature.com/scitable/topicpage/thomas-hunt-morgan-and-sex-linkage-452 .

Explore More II

- **Inheritance of Sex-linked Traits** at http://www.youtube.com/watch?v=IJqFk-28G08 (4:49)



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1. What are the three types of color blindness? How are they caused?
2. What is the "Law of Dominance"?
3. Can a woman have colorblindness if her father does not? Explain your answer fully.
4. A woman is color blind but her sister isn't. What does that tell you about their parents

If you're still puzzled by sex-linked traits you can go to this site for more practice solving problems. Make sure you make good use of the "hints" on the site.

- **Sex-linked genes** at http://www.ksu.edu/biology/pob/genetics/xlinked.htm

Review

1. What are the sex chromosomes of a male and a female?
2. Explain why the father determines the sex of the child.
3. What is sex-linked inheritance?
4. A son cannot inherit colorblindness from his father. Why not?

4.6 Mutation Effects

Learning Objectives

- Describe beneficial mutations.
- Give examples of harmful mutations.

Is this rat hairless?

Yes. Why? The result of a mutation, a change in the DNA sequence. The effects of mutations can vary widely, from being beneficial, to having no effect, to having lethal consequences, and every possibility in between.

Effects of Mutations

The majority of mutations have neither negative nor positive effects on the organism in which they occur. These mutations are called **neutral mutations**. Examples include silent point mutations. They are neutral because they do not change the amino acids in the proteins they encode.

Many other mutations have no effect on the organism because they are repaired before protein synthesis occurs. Cells have multiple repair mechanisms to fix mutations in DNA. One way DNA can be repaired is illustrated in the **Figure** 4.9. If a cell's DNA is permanently damaged and cannot be repaired, the cell is likely to be prevented from dividing.

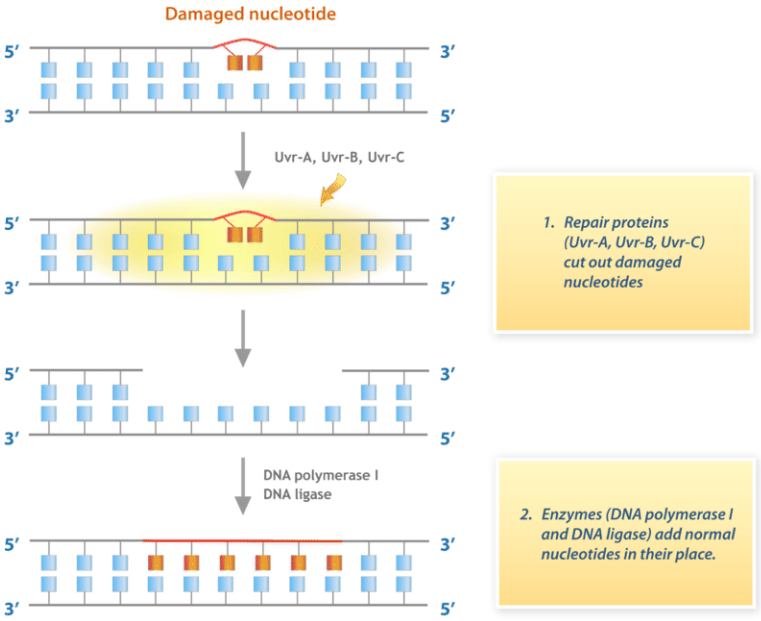

FIGURE 4.9

DNA Repair Pathway. This flow chart shows one way that damaged DNA is repaired in *E. coli* bacteria.

Beneficial Mutations

Some mutations have a positive effect on the organism in which they occur. They are called **beneficial mutations**. They lead to new versions of proteins that help organisms adapt to changes in their environment. Beneficial mutations are essential for evolution to occur. They increase an organism's changes of surviving or reproducing, so they are likely to become more common over time. There are several well-known examples of beneficial mutations. Here are just two:

1. Mutations in many bacteria that allow them to survive in the presence of antibiotic drugs. The mutations lead to **antibiotic-resistant** strains of bacteria.
2. A unique mutation is found in people in a small town in Italy. The mutation protects them from developing atherosclerosis, which is the dangerous buildup of fatty materials in blood vessels. The individual in which the mutation first appeared has even been identified.

Harmful Mutations

Imagine making a random change in a complicated machine such as a car engine. The chance that the random change would improve the functioning of the car is very small. The change is far more likely to result in a car that does not run well or perhaps does not run at all. By the same token, any random change in a gene's DNA is likely to result in a protein that does not function normally or may not function at all. Such mutations are likely to be harmful. Harmful mutations may cause genetic disorders or cancer.

- A **genetic disorder** is a disease caused by a mutation in one or a few genes. A human example is cystic fibrosis. A mutation in a single gene causes the body to produce thick, sticky mucus that clogs the lungs and blocks ducts in digestive organs.

- **Cancer** is a disease in which cells grow out of control and form abnormal masses of cells. It is generally caused by mutations in genes that regulate the cell cycle. Because of the mutations, cells with damaged DNA are allowed to divide without limits. Cancer genes can be inherited.

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Summary

- Mutations are essential for evolution to occur because they increase genetic variation and the potential for individuals to differ.
- The majority of mutations are neutral in their effects on the organisms in which they occur.
- Beneficial mutations may become more common through natural selection.
- Harmful mutations may cause genetic disorders or cancer.

Review

1. Why are mutations essential for evolution to occur?
2. What is a genetic disorder?
3. What is cancer? What usually causes cancer?

4.7 Gametogenesis

Learning Objectives

- Define gametogenesis.
- Compare and contrast spermatogenesis and oogenesis.
- Explain the importance of a polar body.

What's the biggest cell on Earth?

The ostrich egg - unfertilized, of course. Yes, this egg, just like a human ovum, is just one cell. The egg shell membrane encloses the nucleus containing the genetic material and the cytoplasm.

Gametogenesis

At the end of meiosis, four haploid cells have been produced, but the cells are not yet gametes. The cells need to develop before they become mature gametes capable of fertilization. The development of haploid cells into gametes is called **gametogenesis**.

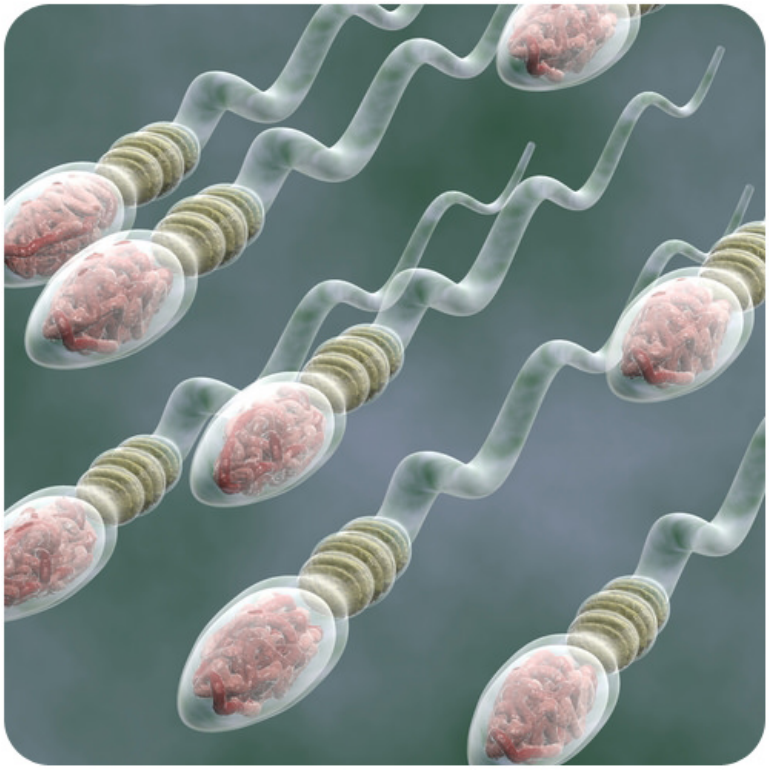


FIGURE 4.10

How much DNA is in a gamete? The sperm cell forms by meiosis and spermatogenesis. Because it forms by meiosis, the sperm cell has only half as much DNA as a body cell. Notice the three distinct segments: a head piece, a flagella tail and a midpiece of mostly mitochondria. What is the role of each section?

Gametogenesis may differ between males and females. Male gametes are called **sperm**. Female gametes are called **eggs**. In human males, for example, the process that produces mature sperm cells is called **spermatogenesis**. During this process, sperm cells grow a tail and gain the ability to "swim," like the human sperm cell shown in **Figure** 4.11. In human females, the process that produces mature eggs is called **oogenesis**. Just one egg is produced from the four haploid cells that result from meiosis. The single egg is a very large cell, as you can see from the human egg in **Figure** 4.11.

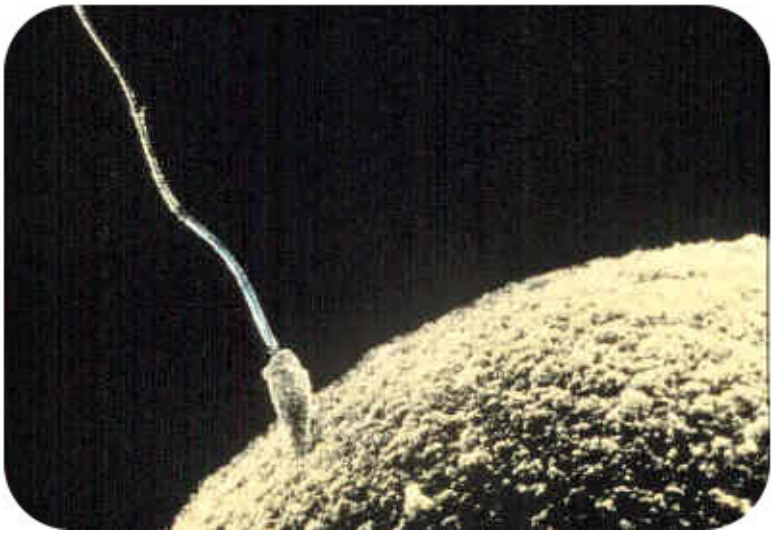


FIGURE 4.11

A human sperm is a tiny cell with a tail. A human egg is much larger. Both cells are mature haploid gametes that are capable of fertilization. What process is shown in this photograph? Notice the sperm with the head piece containing the genetic material, a flagella tail that propels the sperm, and a midpiece of mostly mitochondria, supplying ATP.

Spermatogenesis and Oogenesis

During spermatogenesis, primary **spermatocytes** go through the first cell division of meiosis to produce secondary spermatocytes. These are haploid cells. Secondary spermatocytes then quickly complete the meiotic division to become **spermatids**, which are also haploid cells. The four haploid cells produced from meiosis develop a flagellum tail and compact head piece to become mature sperm cells, capable of swimming and fertilizing an egg. The compact head, which has lost most of its cytoplasm, is key in the formation of a streamlined shape. The middle piece of the sperm, connecting the head to the tail, contains many mitochondria, providing energy to the cell. The sperm cell essentially contributes only DNA to the zygote.

On the other hand, the egg provides the other half of the DNA, but also organelles, building blocks for compounds such as proteins and nucleic acids, and other necessary materials. The egg, being much larger than a sperm cell, contains almost all of the cytoplasm a developing embryo will have during its first few days of life. Therefore, oogenesis is a much more complicated process than spermatogenesis.

Oogenesis begins before birth and is not completed until after fertilization. Oogenesis begins when **oogonia** (singular, oogonium), which are the immature eggs that form in the ovaries before birth and have the diploid number of chromosomes, undergo mitosis to form primary **oocytes**, also with the diploid number. Oogenesis proceeds as a primary oocyte undergoes the first cell division of meiosis to form secondary oocytes with the haploid number of chromosomes. A secondary oocyte only undergoes the second meiotic cell division to form a haploid ovum if it is fertilized by a sperm. The one egg cell that results from meiosis contains most of the cytoplasm, nutrients, and organelles. This unequal distribution of materials produces one large cell, and one cell with little more than DNA. This other cell, known as a **polar body**, eventually breaks down. The larger cell undergoes meiosis II, once again producing a large cell and a polar body. The large cell develops into the mature gamete, called an **ovum** (**Figure** 4.12). The unequal distribution of the cytoplasm during oogenesis is necessary as the zygote that results from fertilization receives all of its cytoplasm from the egg. So the egg needs to have as much cytoplasm as possible.

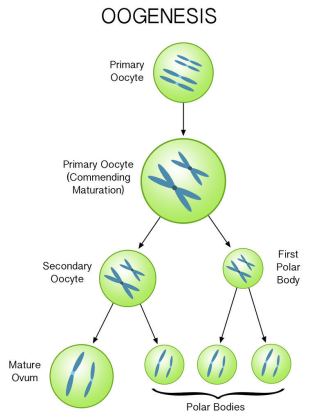
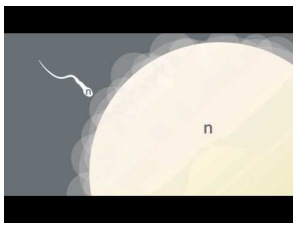


FIGURE 4.12

Maturation of the ovum. Notice only one mature ovum, or egg, forms during meiosis from the primary oocyte. Three polar bodies may form during oogenesis. These polar bodies will not form mature gametes. Conversely, four haploid spermatids form during meiosis from the primary spermatocyte.



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Summary

- Meiosis is a step during spermatogenesis and oogenesis.
- Spermatogenesis produces four haploid sperm cells, while oogenesis produces one mature ovum.

Review

1. What is gametogenesis, and when does it occur?
2. What are the main differences between oogenesis and spermatogenesis?
3. How many chromosomes are in a human oogonia?
4. Why is there unequal distribution of the cytoplasm during oogenesis?

4.8 Meiosis

Learning Objectives

- Give an overview of sexual reproduction.
- Summarize meiosis.
- Outline the stages of meiosis.
- Describe how chromosomes separate in meiosis I and meiosis II.

How do you make a cell with half the DNA?

Meiosis. This allows cells to have half the number of chromosomes, so two of these cells can come back together to form a new organism with the complete number of chromosomes. This process not only helps produce gametes, it also ensures genetic variation.

Meiosis

The process that produces haploid gametes is meiosis. **Meiosis** is a type of cell division in which the number of chromosomes is reduced by half. It occurs only in certain special cells of the organisms. During meiosis, homologous chromosomes separate, and **haploid** cells form that have only one chromosome from each pair. Two cell divisions occur during meiosis, and a total of four haploid cells are produced. The two cell divisions are called meiosis I and meiosis II. The overall process of meiosis is summarized in **Figure** 4.13.

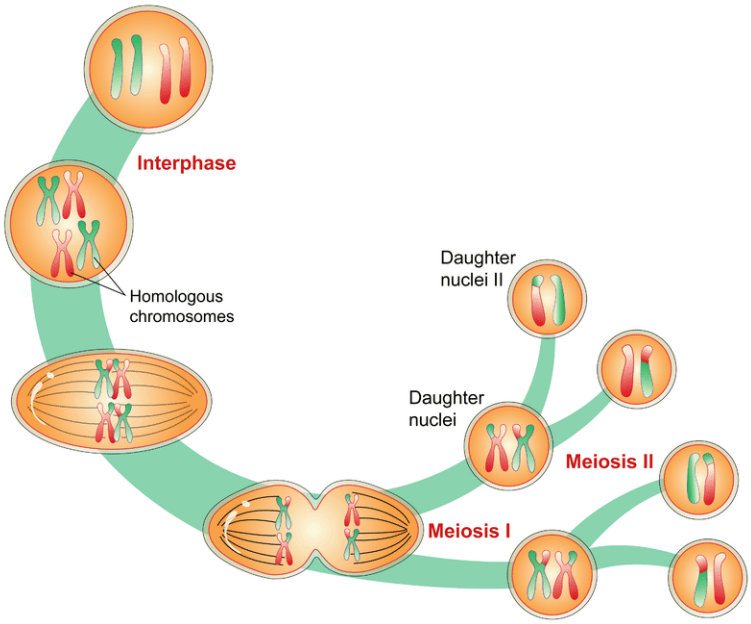

FIGURE 4.13

Overview of Meiosis. During meiosis, homologous chromosomes separate and go to different daughter cells. This diagram shows just the nuclei of the cells. Notice the exchange of genetic material that occurs prior to the first cell division.

Multimedia

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Phases of Meiosis

Meiosis I begins after DNA replicates during interphase of the cell cycle. In both meiosis I and meiosis II, cells go through the same four phases as mitosis - prophase, metaphase, anaphase and telophase. However, there are important differences between meiosis I and mitosis. The flowchart in **Figure** 4.14 shows what happens in both meiosis I and II.

Compare meiosis I in this flowchart with the figure from the *Mitosis and Cytokinesis* concept. How does meiosis I differ from mitosis? Notice at the beginning of meiosis (prophase I), homologous chromosomes exchange segments of DNA. This is known as **crossing-over**, and is unique to this phase of meiosis.

Meiosis I

1. Prophase I: The nuclear envelope begins to break down, and the chromosomes condense. Centrioles start moving to opposite poles of the cell, and a spindle begins to form. Importantly, **homologous chromosomes** pair up, which is unique to prophase I. In prophase of mitosis and meiosis II, homologous chromosomes do not form pairs in this way. Crossing-over occurs during this phase (see the *Genetic Variation* concept).
2. Metaphase I: Spindle fibers attach to the paired homologous chromosomes. The paired chromosomes line up along the equator (middle) of the cell. This occurs only in metaphase I. In metaphase of mitosis and meiosis II, it is sister chromatids that line up along the equator of the cell.

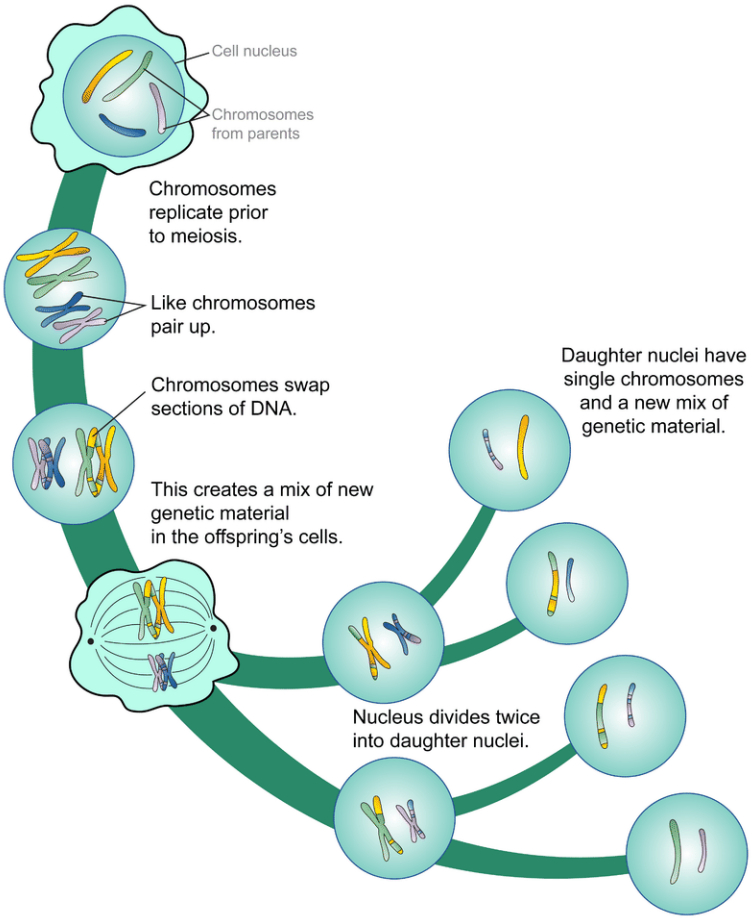

FIGURE 4.14

Phases of Meiosis. This flowchart of meiosis shows meiosis I in greater detail than meiosis II. Meiosis I—but not meiosis II—differs somewhat from mitosis. Compare meiosis I in this flowchart with the earlier figure featuring mitosis. How does meiosis I differ from mitosis?

3. Anaphase I: Spindle fibers shorten, and the chromosomes of each homologous pair start to separate from each other. One chromosome of each pair moves toward one pole of the cell, and the other chromosome moves toward the opposite pole.
4. Telophase I and Cytokinesis: The spindle breaks down, and new nuclear membranes form. The cytoplasm of the cell divides, and two haploid daughter cells result. The daughter cells each have a random assortment of chromosomes, with one from each homologous pair. Both daughter cells go on to meiosis II. The DNA does not replicate between meiosis I and meiosis II.

Meiosis II

1. Prophase II: The nuclear envelope breaks down and the spindle begins to form in each haploid daughter cell from meiosis I. The centrioles also start to separate.
2. Metaphase II: Spindle fibers line up the sister chromatids of each chromosome along the equator of the cell.
3. Anaphase II: Sister chromatids separate and move to opposite poles.
4. Telophase II and Cytokinesis: The spindle breaks down, and new nuclear membranes form. The cytoplasm of each cell divides, and four haploid cells result. Each cell has a unique combination of chromosomes.

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Summary

- Meiosis is the type of cell division that produces gametes.
- Meiosis involves two cell divisions and produces four haploid cells.
- Sexual reproduction has the potential to produce tremendous genetic variation in offspring. This is due in part to crossing-over during meiosis.

Review

1. What is meiosis?
2. Compare the events of metaphase I to metaphase II?
3. Create a diagram to show how crossing-over occurs and how it creates new gene combinations on each chromosome.
4. Explain why sexual reproduction results in genetically unique offspring.
5. Explain how meiosis I differs from mitosis.

4.9 Population Genetics

Learning Objectives

- Distinguish between microevolution and macroevolution.
- Define gene pool.
- Explain how to calculate allele frequencies.

Jeans vs. Genes. What's the difference?

Plenty. One you have for life, the other just lasts a few years. One is the basis for the passing of traits from one generation to the next. Some jeans you change frequently. But what happens when you change a gene's frequency? Essentially, evolution is a change in gene frequencies within a population.

Genes in Populations

Darwin knew that heritable variations are needed for evolution to occur. However, he knew nothing about Mendel's laws of genetics. Mendel's laws were rediscovered in the early 1900s. Only then could scientists fully understand the process of evolution. We now know that variations of traits are heritable. These variations are determined by different **alleles**. We also know that evolution is due to a change in alleles over time. How long a time? That depends on the scale of evolution.

- **Microevolution** occurs over a relatively short period of time within a population or species. The Grants observed this level of evolution in Darwin's finches (see the "Biogeography" concept).
- **Macroevolution** occurs over geologic time above the level of the species. The fossil record reflects this level of evolution. It results from microevolution taking place over many generations.

Remember that individuals do not evolve. Their **genes** do not change over time. The unit of evolution is the population. A **population** consists of organisms of the same species that live in the same area. In terms of evolution, the population is assumed to be a relatively closed group. This means that most mating takes place within the population. The science that focuses on evolution within populations is **population genetics**. It is a combination of evolutionary theory and Mendelian genetics.

Gene Pool

The genetic makeup of an individual is the individual's **genotype**. A population consists of many genotypes. Altogether, they make up the population's gene pool. The **gene pool** consists of all the genes of all the members of the population. For each gene, the gene pool includes all the different alleles for the gene that exist in the population. For a given gene, the population is characterized by the frequency of the different alleles in the gene pool.

Allele Frequencies

Allele frequency is how often an allele occurs in a gene pool relative to the other alleles for that gene. Look at the example in the **Table** 4.4. The population in the table has 100 members. In a sexually reproducing species, each member of the population has two copies of each gene. Therefore, the total number of copies of each gene in the gene pool is 200. The gene in the example exists in the gene pool in two forms, alleles *A* and *a*. Knowing the genotypes of each population member, we can count the number of alleles of each type in the gene pool. The table shows how this is done.

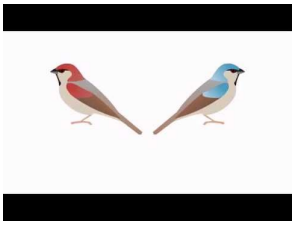
TABLE 4.4: Number of Alleles in a Gene Pool

Genotype	Number of Individuals in the Population with that Genotype	Number of Allele *A* Contributed to the Gene Pool by that Genotype	Number of Allele *a* Contributed to the Gene Pool by that Genotype
AA	50	$50 \times 2 = 100$	$50 \times 0 = 0$
Aa	40	$40 \times 1 = 40$	$40 \times 1 = 40$
aa	10	$10 \times 0 = 0$	$10 \times 2 = 20$
Totals	100	140	60

Let the letter *p* stand for the frequency of allele *A*. Let the letter *q* stand for the frequency of allele *a*. We can calculate *p* and *q* as follows:

- *p* = number of *A* alleles/total number of alleles = 140/200 = 0.7
- *q* = number of *a* alleles/total number of alleles = 60/200 = 0.3
- Notice that $p + q = 1$.

Evolution occurs in a population when allele frequencies change over time. What causes allele frequencies to change? That question was answered by Godfrey Hardy and Wilhelm Weinberg in 1908 (see the *Hardy-Weinberg* Theorem concept).



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Further Reading

Hardy-Weinberg

Summary

- Microevolution occurs over a short period of time in a population or species. Macroevolution occurs over geologic time above the level of the species.
- The population is the unit of evolution.
- A population's gene pool consists of all the genes of all the members of the population.
- For a given gene, the population is characterized by the frequency of different alleles in the gene pool.

Review

1. Compare microevolution to macroevolution.
2. Why are populations, rather than individuals, the units of evolution?
3. What is a gene pool?
4. Assume that a population of 50 individuals has the following numbers of genotypes for a gene with two alleles, *B* and *b*: *BB* = 30, *Bb* = 10, and *bb* = 10. Calculate the frequencies of the two alleles in the population's gene pool.

4.10 Descriptive Statistics

Learning Objectives

- State why descriptive statistics are useful.
- Identify the mean, median, and mode of a sample.
- Describe the range of a sample.

The girls in this picture vary in height. The shortest girl has a height of 52 cm, and the tallest girl has a height of 64 cm. The other two girls fall in between these two extremes. How could you describe the heights of all four girls with a single number? How could express how they vary in height with another number?

Using Statistics to Describe a Sample

The girls in the picture above make up a small sample—there are only four of them. In scientific investigations, samples may include hundreds or even thousands of people or other objects of study. Especially when samples are very large, it's important to be able to summarize their overall characteristics with a few numbers. That's where descriptive statistics come in. Descriptive statistics are measures that show the central tendency, or center, of a sample or the variation in a sample.

Describing the Center

The central tendency of a sample can be represented by the mean, median, or mode.

- The **mean** is the average value. It is calculated by adding the individual measurements and dividing the sum by the total number of measurements.

- The median is the middle value. To find the median, rank all the measurements from smallest to largest and then find the measurement that is in the middle.
- The mode is the most common value. It is the value that occurs most often.

Q: A sample of five children have the following heights: 60 cm, 58 cm, 54 cm, 62 cm, and 58 cm. What are the mean, median, and mode of this sample?

A: The mean is (60 cm + 58 cm + 54 cm + 62 cm + 58 cm) ÷ 5 = 58 cm. The median and mode are both 58 cm as well. The mean, median, and mode are not always the same, as they are for this sample. In fact, sometimes these three statistics are very different from one another for the same sample.

Describing the Range

Many samples have a lot of variation in measurements. Variation can be described with a statistic called the range. The **range** is the total spread of values in a sample. It is calculated by subtracting the smallest value from the largest value.

Q: What is the range of heights in the sample of children in the previous question?

A: The range is 62 cm - 54 cm = 8 cm.

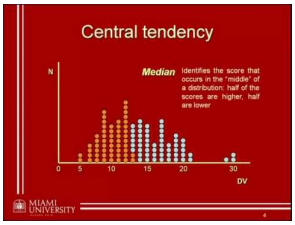
Summary

- Descriptive statistics are measures that summarize the characteristics of a sample.
- The central tendency, or center, of a sample can be represented by the mean, median, or mode.
- The variation in a sample can be represented by the range, or the total spread of values.

Review

1. What are descriptive statistics, and why are they useful?
2. Find the mean, median, and mode of this set of values: 12 g, 9 g, 13 g, 12 g, 20 g, 17 g, 15 g.
3. What is the range of the set of values in question 2?

Resources



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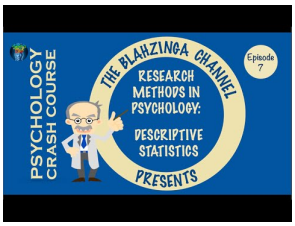

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