Biology - Student Reader & Workbook
Unit 3, Chapter 3: Human Genetics

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Do these colorful structures look familiar? There are 46 of them, and they occur in pairs. Can you guess what they are? Here's a hint: you have 46 structures like these in each of your cells.

The picture is an actual image of a complete set of human chromosomes. Chromosomes are the structures that contain genes and consist of DNA. They are responsible for your inherited characteristics. In this chapter, you will learn how human chromosomes and genes are inherited and how they control the traits that make each of us unique.

Lesson 3.1Human Chromosomes and Genes

Lesson Objectives

- Define the human genome.
- Describe human chromosomes and genes.
- Explain linkage and linkage maps.

Lesson Vocabulary

human genome
Human Genome Project
autosome
sex chromosome
linked genes
sex-linked gene
X-linked gene
linkage map

Lesson Introduction

Nobody else in the world is exactly like you. What makes you different from everyone else? Genes have a lot to do with it. Unless you have an identical twin, no one else on Earth has exactly the same genes as you. What about identical twins? Are they identical in every way? They develop from the same fertilized egg, so they have all same genes, but even they are not completely identical. Why? The environment also influences human characteristics, and no two people have exactly the same environment.

Before Reading: Think - Pair - Share

*What is the relationship between **DNA**, **genes**, and **chromosomes**?*

_____

_____

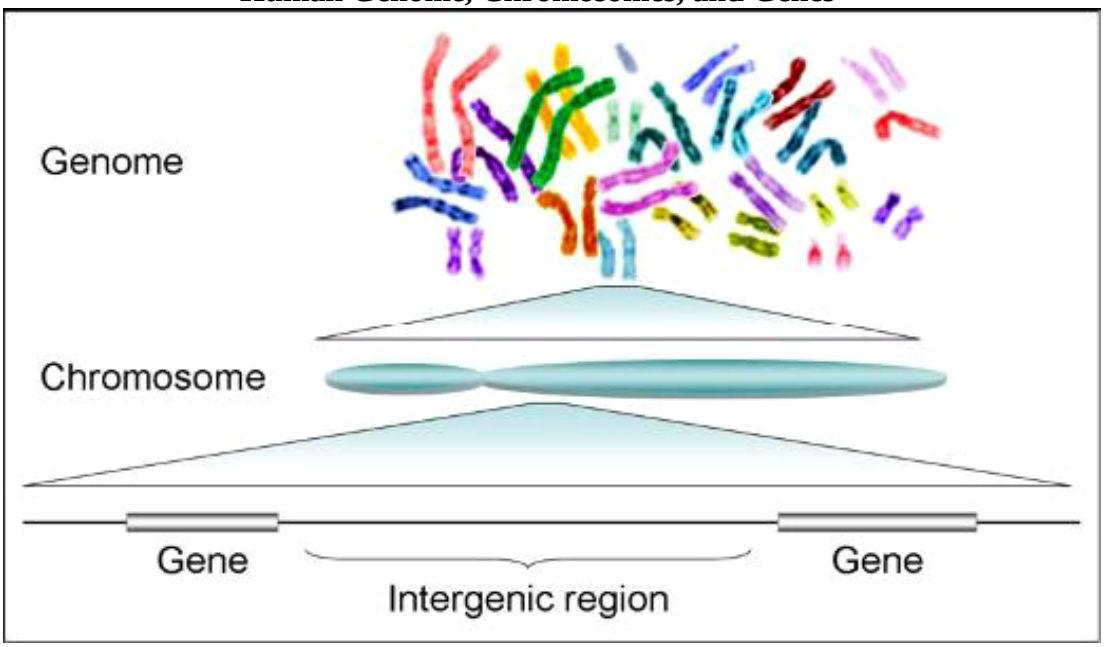
_____

The Human Genome

All of the DNA of the human species makes up the human genome. This DNA consists of about 3 billion base pairs and is divided into thousands of genes on many different chromosomes. Genes are sections of DNA within the chromosome that code for protein. Not all of your DNA codes for protein. The human genome also includes noncoding sequences of DNA, as shown in Figure 3.1. Why do you think the noncoding sequences of DNA exist?

Figure 3.1: Each chromosome of the human genome contains many genes as well as noncoding intergenic ("between genes") regions.

Human Genome, Chromosomes, and Genes



Thanks to the **Human Genome Project**, scientists now know the DNA sequence of the entire human genome. The Human Genome Project is an international project that includes scientists from around the world. It began in 1990, and by 2003, scientists had sequenced all 3 billion base pairs of human DNA. Now they are trying to identify all the genes in the sequence.

Reading Check:

1. A _____ is a section of DNA that codes for a protein. Thousands of genes can be found on just one _____. We have a total of 46 _____ in each of our cells. All of your DNA together makes up your _____.

2. The Human Genome Project is a big breakthrough in our understanding of heredity. What is the purpose of identifying genes? Can you think of two potential uses for identified genes?

_____

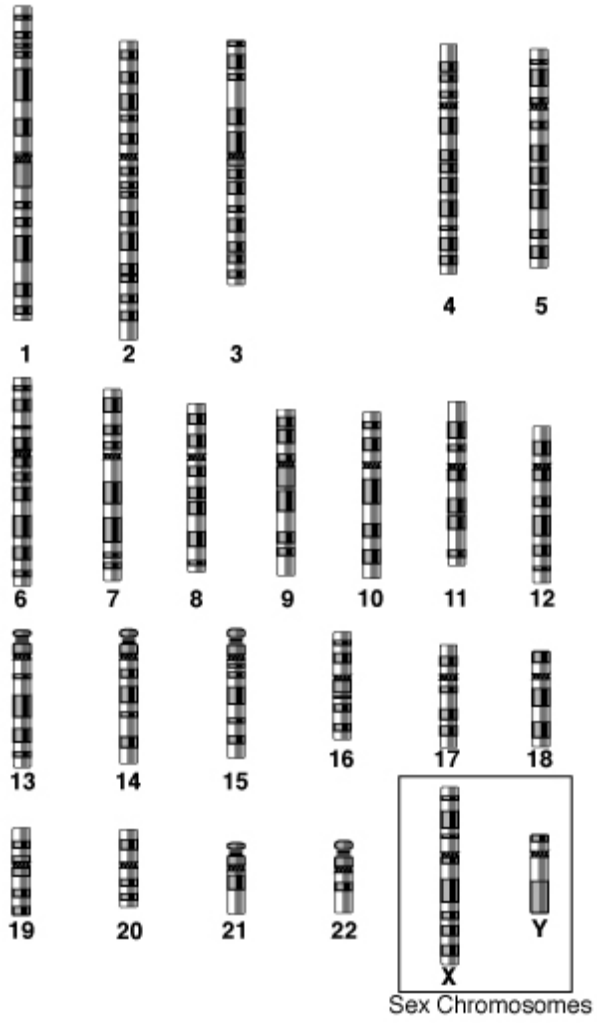
_____

Chromosomes and Genes

Each species has a characteristic number of chromosomes. The human species is characterized by 23 pairs of chromosomes, as shown in Figure 3.2.

Figure 3.2: Human chromosomes, except for the sex chromosomes X and Y, are shown here arranged by size. Chromosome 1 is the largest, and chromosome 22 is the smallest. All normal human cells (except gametes) have two of each chromosome, for a total of 46 chromosomes per cell.

Human Chromosomes



Autosomes

Of the 23 pairs of human chromosomes, 22 pairs are autosomes (numbers 1–22 in Figure 3.2). **Autosomes** are chromosomes that contain genes for characteristics that are unrelated to sex. These chromosomes are the same in males and females. The great majority of human genes are located on autosomes.

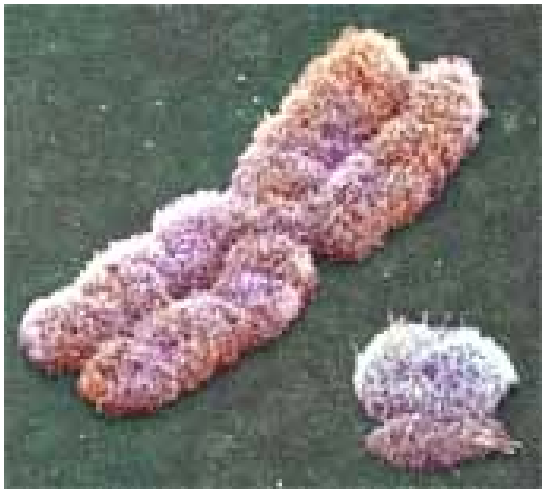
Sex Chromosomes

The remaining pair of human chromosomes consists of the **sex chromosomes**, X and Y. Females have two X chromosomes, and males have one X and one Y chromosome. In females, one of the X chromosomes in each cell is inactivated and known as a Barr body. This ensures that females, like males, have only one functioning copy of the X chromosome in each cell.

As you can see from Figure 3.3, the X chromosome is much larger than the Y chromosome. The X chromosome has about 2000 genes, whereas the Y chromosome has fewer than 100, none of which are essential to survival. Virtually all of the X chromosome genes are unrelated to sex. Only the Y chromosome contains genes that determine sex. A single Y chromosome gene, called SRY, triggers an embryo to develop into a male. Without a Y chromosome, an individual develops into a female, so you can think of female as the default sex of the human species. Can you think of a reason why the Y chromosome is so much smaller than the X chromosome?

Figure 3.3: The human X chromosome (left) and Y chromosome (right) differ considerably in size and in the number of genes they carry.

Human X and Y Chromosomes



Reading Check:

1. *Which chromosomes contain the great majority of genes in humans? (circle one)*
   autosomes *sex chromosomes*

2. *Which gender has two X chromosomes? (circle one)* *male* *female*

3. *Which chromosome is larger? (circle one)* *X* *Y*

4. *What is a Barr body and what is its purpose?* _____
   _____

5. *Where is the SRY gene found and what does it do?* _____
   _____

6. *How many total chromosomes are in each normal cell (except for gametes) in a human?*
   _____

7. *How many total chromosomes does a human gamete (sex cell) have?* _____

Human Genes

Humans have an estimated 20,000 to 25,000 genes. This may sound like a lot, but it really isn't. Far simpler species have almost as many genes as humans. However, human cells use different processes to make multiple proteins from the instructions encoded in a single gene. Of the 3 billion base pairs in the human genome, only about 25 percent make up genes and their regulatory elements. The functions of many of the other base pairs (noncoding DNA) are still unclear.

Do you remember Mendel's experiments with pea plants? He was able to arrive at a few key laws of genetics because he was working with plant characteristics that were on different chromosomes, were dominant or recessive, and had only two different alleles - like purple or white for flower color. Human genetics is far more complicated. The majority of human genes have two or more possible alleles. Differences in alleles account for the considerable genetic variation among people. In fact, most human genetic variation is the result of differences in individual DNA bases within alleles.

Think/Pair/Share

Mendel's Law of Independent Assortment stated that factors controlling different characteristics are inherited independently of each other. However, sometimes certain characteristics are often inherited together, like red hair and light skin. Why do you think this might happen?

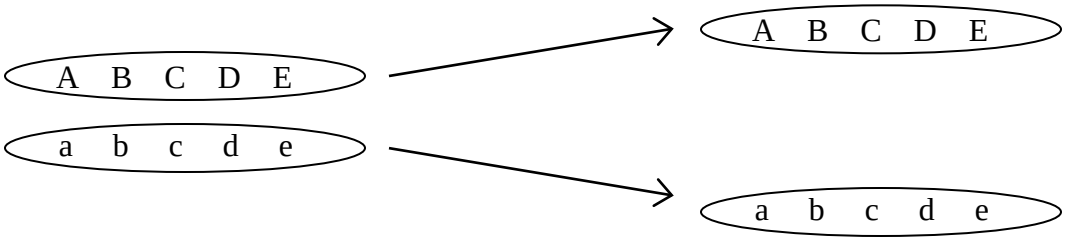
Linkage

Mendel studied characteristics of pea plants that were located on different chromosomes. Genes that are located on the same chromosome are called **linked genes**. Alleles for these genes tend to be inherited together during meiosis, unless they are separated by crossing-over. Crossing-over occurs when two homologous chromosomes exchange genetic material during meiosis I. The closer together two genes are on a chromosome, the less likely that the cross-over will occur between the two genes causing their alleles to be separated.

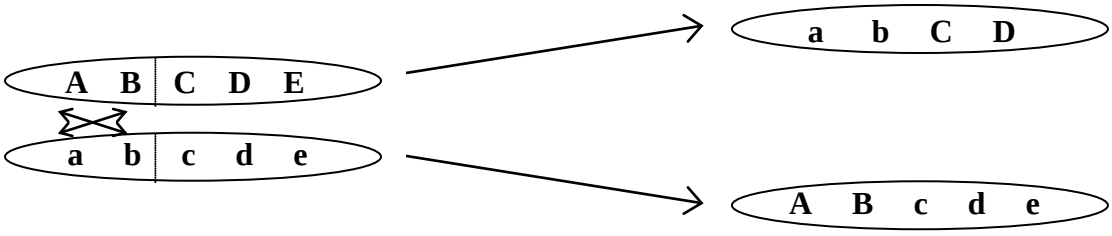
Linkage explains why certain characteristics are frequently inherited together. For example, genes for hair color and eye color are linked, so certain hair and eye colors tend to be inherited together, such as blonde hair with blue eyes and brown hair with brown eyes. What other human traits seem to occur together? Do you think they might be controlled by linked genes?

Figure 3.4: Diagram A below shows two homologous chromosomes separating into different gametes. The letters represent different genes. Genes located on the same chromosome are considered linked because they are often inherited together. However, if crossing-over occurs during meiosis such as in diagram B, genes can become unlinked. Because genes A and E are further away from each other on the chromosome, they are more likely to becomes unlinked than two close together, like A and B.

A – Homologous chromosomes separating into different gametes with no crossing-over. Genes located on the same chromosome are linked.



B – Homologous chromosomes separating into different gametes after crossing-over has occurred between genes B and C. Alleles A and B have become unlinked from C, D, and E.



Sex-Linked Genes

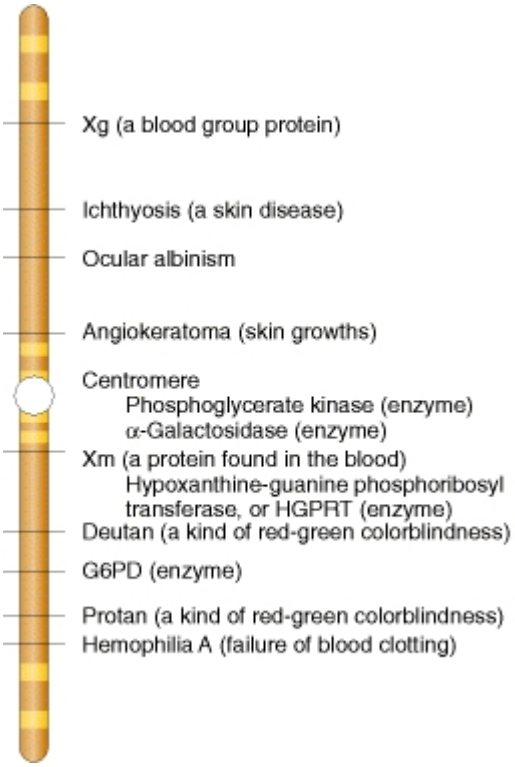
Genes located on the sex chromosomes are called **sex-linked genes**. Most sex-linked genes are on the X chromosome because the Y chromosome has relatively few genes. Strictly speaking, genes on the X chromosome are **X-linked genes**, but the term *sex-linked* is often used to refer to them. Can you think of some traits that might be sex-linked?

Mapping Linkage

Linkage can be assessed by determining how often crossing-over occurs between two genes on the same chromosome. Genes on different (nonhomologous) chromosomes are not linked. They assort independently during meiosis, so they have a 50 percent chance of ending up in different gametes. If genes show up in different gametes less than 50 percent of the time (that is, they tend to be inherited together), they are assumed to be on the same (homologous) chromosome. They may be separated by crossing-over, but this is likely to occur less than 50 percent of the time. The less often crossing-over occurs, the closer together on the same chromosome the genes are presumed to be. Frequencies of crossing-over can be used to construct a **linkage map** like the one in Figure 3.5. A linkage map shows the locations of genes on a chromosome.

Figure 3.5: This linkage map shows the locations of several genes on the X chromosome. Some of the genes code for normal proteins. Others code for abnormal proteins that lead to genetic disorders.

Linkage Map for the Human X Chromosome



Reading Check:

*1. What are **linked genes**? _____*

2. How can you tell if genes are linked? _____
_____

3. In Figure 3.5, which pair of genes would you expect to have a lower frequency of crossing-over: the genes that code for hemophilia A and G6PD deficiency, or the genes that code for protan and Xm? Why? _____

Lesson Summary

- The human genome consists of about 3 billion base pairs of DNA. In 2003, the Human Genome Project finished sequencing all 3 billion base pairs.
- Humans have 23 pairs of chromosomes. Of these, 22 are autosomes and 2 are sex chromosomes (X, Y). Females have two X chromosomes, and males have one X and one Y. Human chromosomes contain a total of 20,000 to 25,000 genes, the majority of which have two or more alleles.
- Linked genes are located on the same chromosome. Sex-linked genes are located on a sex chromosome, and X-linked genes are located on the X chromosome. The frequency of crossing-over between genes is used to construct linkage maps that show the locations of genes on chromosomes.

Lesson Review Questions

Recall

1. Describe the human genome.
2. What has the Human Genome Project achieved?
3. What are linked genes?
4. Describe human genetic variation.

Apply Concepts

5. Explain how you would construct a linkage map for a human chromosome. What data would you need?

Think Critically

6. Compare and contrast human autosomes and sex chromosomes.
7. People with red hair usually have very light skin. What might be a genetic explanation for this observation?

Points to Consider

You read in this lesson about the chromosomes and genes that control human traits. Most traits are controlled by genes on autosomes, but many are controlled by genes on the X chromosome.

- Do you think it matters whether a gene is on an autosome or the X chromosome when it comes to how it is inherited?
- How do mothers and fathers pass their sex chromosomes to their sons and daughters? Their autosomes?

Multimedia Links

You can watch a video about the Human Genome Project and how it cracked the code of life at this link:
http://www.pbs.org/wgbh/nova/genome/program.html

You can watch a short animation about human chromosomes at this link:
http://www.dnalc.org/view/15520-DNA-is-organized-into-46-chromosomes-including-sex-chromosomes-3D-animation.html

At the link below, you can click on any human chromosome to see which traits its genes control.
http://www.ornl.gov/sci/techresources/Human_Genome/posters/chromosome/chooser.shtml

At the link below, you can watch an animation that explains why the Y chromosomes is so much smaller than the X chromosome.
http://www.hhmi.org/biointeractive/gender/Y_evolution.html

To learn more about the coding and noncoding sequences of human DNA, watch the animation at this link:
http://www.hhmi.org/biointeractive/dna/DNAi_coding_sequences.html

At the link below, you can watch an animation showing how genes on the same chromosome may be separated by crossing-over.
http://www.biostudio.com/d_%20Meiotic%20Recombination%20Between%20Linked%20Genes.htm

Lesson 3.2: Human Inheritance

Lesson Objectives

- Describe inheritance in humans for autosomal and X-linked traits.
- Identify complex modes of human inheritance.
- Describe genetic disorders caused by mutations and abnormal numbers of chromosomes.

Lesson Vocabulary

genetic trait
pedigree
sex-linked trait
X-linked trait
multiple allele trait
pleiotropy
epistasis
nondisjunction
gene therapy

Lesson Introduction

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.

Before Reading: Think Aloud

RECALL: In chapter 6 you learned that Gregor Mendel was lucky in choosing pea plants because of their simple inheritance patterns. The characteristics he looked at were dominant or recessive, only had two alleles, and were found on different chromosomes. What are some things that make inheritance patterns more complex?

_____

_____

_____

Mendelian Inheritance in Humans

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 (chromosomes 1-22) or the X chromosome.

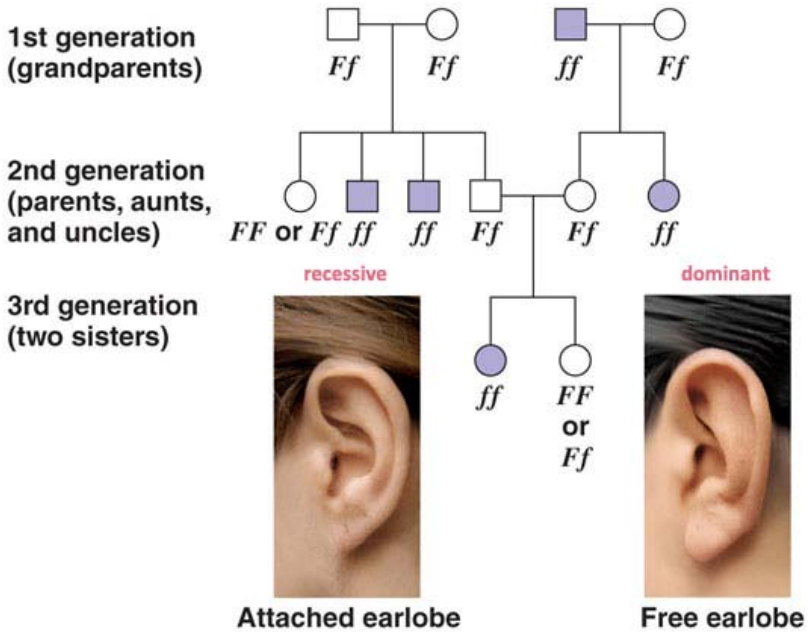
Autosomal Traits

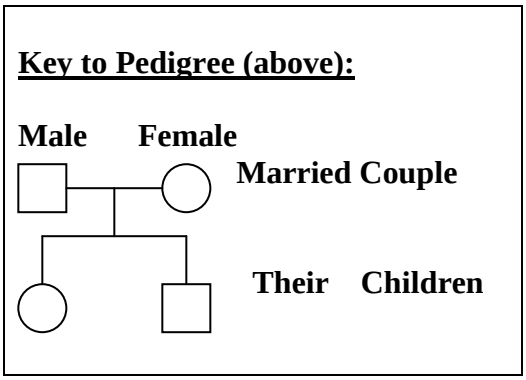
Autosomal traits are controlled by genes on one of the 22 human autosomes. Consider earlobe attachment, which is shown in Figure 3.6. A single autosomal gene with two alleles determines whether you have attached earlobes or free-hanging earlobes. The allele for free-hanging earlobes (*F*) is dominant to the allele for attached earlobes (*f*).

The chart in Figure 3.6 is called a **pedigree**. It shows how the earlobe trait was passed from generation to generation within a family. Squares represent males and circles represent females. Pedigrees are useful tools for studying inheritance patterns.

Figure 3.6: 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?

Pedigree for Earlobe Attachment





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 3.7. Which form of these traits do you have? What are your possible genotypes for the traits?

Figure 3.7: Widow's peak and hitchhiker's thumb are dominant traits controlled by a single autosomal gene.

Single-Gene Autosomal Traits

Reading Check:

1. You may have heard of the word "pedigree" in the context of a purebred animal. In general, pedigrees show a record of ancestry. How is the pedigree in Figure 8.5 similar to a family tree? How is it different?

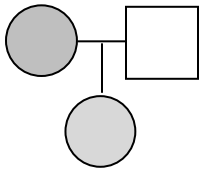
_____

_____

2. A) Hitchhiker's thumb (T) is dominant to a straight thumb (t). What is the genotype for someone with a straight thumb? _____

B) What are the two possible genotypes for someone who has a hitchhiker's thumb? _____

Use the pedigree to the right for hitchhiker's thumb to answer the following questions:



3. What is the genotype of the mother? _____

4. Does the daughter have a hitchhiker's thumb? _____

5. A) What MUST the genotype of the father be? _____

B) How do you know? _____

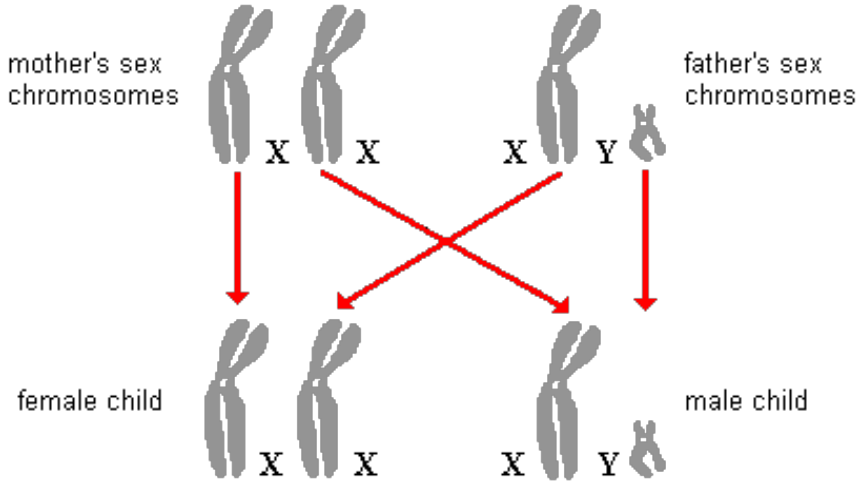
Sex-Linked Traits

Traits controlled by genes on the sex chromosomes are called **sex-linked traits**. Remember the sex chromosomes X and Y? Females have two X chromosomes and males have one X and one Y chromosome. The Y chromosome is much smaller than the X chromosome and contains far fewer genes. However, it does contain the SRY gene, which is what causes the gender to be male. Since the X chromosome contains many more genes than the Y chromosome, most sex-linked traits are **X-linked traits**.

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 pass it on to all their daughters but none of their sons. This is illustrated in Figure 3.8.

Figure 3.8: Mothers pass only X chromosomes to their children. Fathers always pass their X chromosome to their daughters and their Y chromosome to their sons.

Inheritance of Sex Chromosomes



PREDICT:

If a recessive condition is X-linked, would you expect it to occur more often in females or males? Why?

_____

_____

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 a copy of the recessive allele from both their mother and father to express the trait. If they are heterozygous for the condition and have only one recessive allele, the trait is not expressed. They are considered "carriers" for that trait instead. This explains why X-linked recessive traits are less common in females than males.

Reading Check:

1. Why do fathers always determine the sex of the offspring? _____

_____

2. Why do X-linked recessive traits appear more often in males than females?

_____

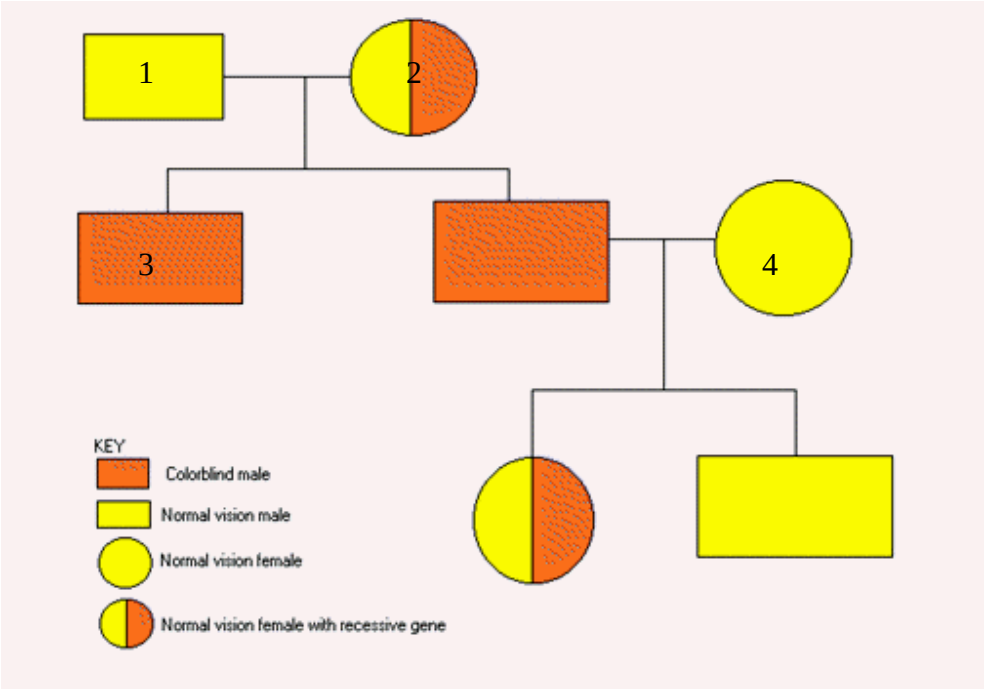
_____

3. If a male inherits an X-linked disease, which parent did he inherit it from? _____

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 3.9: 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.

Pedigree for Color Blindness



Pedigree Practice:

To distinguish an X-linked trait from an autosomal trait, the genotype is often written with the X or Y chromosome included. For instance, a homozygous dominant female for color vision would be written as X^BX^B. A male with the dominant allele for color vision would be X^BY.

1. *Can you explain why there is no allele written next to the Y in the genotype for the male?*

   _____

2. *Try writing the genotypes for the following individuals using the pedigree in Figure 8.8 above. Use B for the dominant allele and b for the recessive allele.*

   A) Person #1 _____ *B) Person #2 _____*
   C) Person #3 _____ *D) Person #4 _____*

Non-Mendelian Inheritance

Most human traits have more complex modes of inheritance. For example, the traits may be controlled by multiple alleles or multiple genes.

Before Reading: Think Aloud

RECALL: So far we've only been discussing traits that are dominant or recessive, but one of the reasons why human genetics is more complicated than the pea plants that Mendel studied is because some traits are codominant or incompletely dominant instead.

1. How is a heterozygous genotype expressed if the alleles are ***codominant****?*

_____

2. How is a heterozygous genotype expressed if the alleles are ***incompletely dominant****?*

_____

Multiple Allele Traits

The majority of human genes are thought to have more than two alleles. Traits controlled by a single gene with more than two alleles are called **multiple allele traits**. An example is ABO blood type. There are three common alleles for this trait, which can be represented by the letters *A*, *B*, and *O*. As shown in Table 3.1, there are six possible ABO genotypes but only four phenotypes. This is because alleles *A* and *B* are codominant to each other and both are dominant to *O*. In other words, if someone is has both alleles A and B, both proteins A and B will be expressed on their red blood cells. However, if someone has alleles A and O or alleles B and O, only protein A or protein B will be expressed. Someone who is blood type O does not express proteins A or B.

Table 3.1: ABO blood type is controlled by one gene with three common alleles.

ABO Blood Type	
Genotype	**Phenotype**
AA	A
AO	A
AB	AB
BB	B
BO	B
OO	O

Reading Check:

1. *Why is blood type a **multiple allele** trait?* _____
_____

2. *Why are there only four phenotypes for blood type if there are six possible genotypes?*
_____
_____

3. *Do you know your blood type? What is it? What are your possible genotypes?*
_____

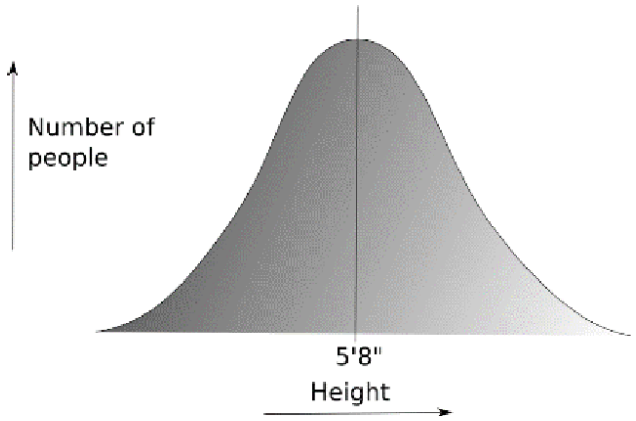
Polygenic Traits

Many human traits are controlled by more than one gene. These traits are called polygenic traits (or characteristics). The alleles of each gene have a minor additive effect on the phenotype. This means for one characteristic, there may be several different genes contributing a small amount to the overall phenotype. There are many possible combinations of alleles, especially if each gene has multiple alleles. Therefore, a whole continuum (range) of phenotypes is possible.

An example of a human polygenic trait is adult height. Several genes, each with more than one allele, contribute to this trait, so there are many possible adult heights. For example, one adult's height might be 1.655 m (5.430 feet), and another adult's height might be 1.656 m (5.433 feet) tall. Adult height ranges from less than 5 feet to more than 6 feet, but the majority of people fall near the middle of the range, as shown in Figure 3.10.

Figure 3.10: Like many other polygenic traits, adult height has a bell-shaped distribution.

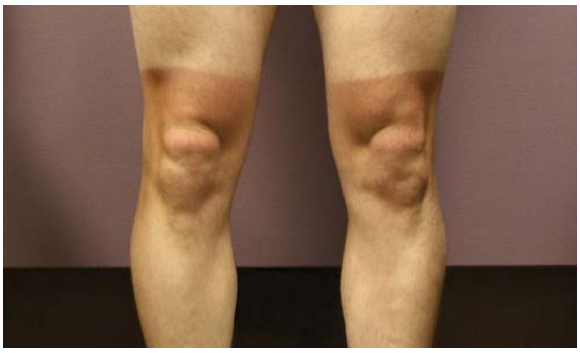
Human Adult Height



Many polygenic traits are affected by the environment. For example, adult height might be negatively impacted by poor diet or illness during childhood. If someone is missing key nutrients as a child, they may not grow as tall as the height determined by their genes. Skin color is another polygenic trait. There is a wide range of skin colors in people worldwide. In addition to differences in skin color genes, differences in exposure to UV light explain most of the variation. As shown in Figure 3.11, exposure to UV light darkens the skin.

Figure 3.11: This picture shows clearly how exposure to UV light can affect skin color. UV light causes skin cells to produce more of a brown pigment called melanin, which makes skin darker.

Effects of UV Light on Skin Color



Reading Check:

1. *What is a **polygenic trait**? Use the familiar prefix "poly-", meaning many, to help you remember.*

_____

2. *What are two examples of how the environment can affect polygenic traits?*

_____

_____

Pleiotropy

Sometimes a single gene may affect more than one trait. This is called **pleiotropy**. An example is the gene that codes for the main protein in collagen, a substance that helps form bones. The gene for this protein also affects the ears and eyes. This was discovered from mutations in the gene. They result in problems not only in bones but also in these sensory organs.

Epistasis

In other cases, one gene affects the expression of another gene. This is called **epistasis**. Epistasis is similar to dominance, except that it occurs between different genes rather than

between different alleles for the same gene. An example is the gene coding for widow's peak. A gene that codes for baldness would "hide" the widow's peak trait if it occurred in the same person.

Word Work

"Pleio-" is a version of the prefix "pleo-", which means more. So pleiotropy is a single gene affecting more than one trait.

The prefix "epi-" means over or above. Epistasis is when the expression of one gene dominates, or is expressed over, the expression of other genes (rather than alleles for the same gene).

Graphic Organizer: Non-Mendelian Genetics

Mode of Inheritance	Definition	Example
Multiple Allele Traits		
Polygenic Traits		
Pleiotropy		
Epistasis		

Reading Check:

Read the following examples of genetic traits and pick the mode of inheritance that is responsible for that trait.

1. PKU is a genetic disorder caused by a mutation in a single gene that creates an enzyme that breaks down the amino acid phenylalanine. As a result, it can cause mental retardation and a reduction of skin and hair pigmentation.

Multiple alleles Polygenic Pleiotropy Epistasis

2. Human eye color has many variations because eye color is determined by what pigments you have and how much of each pigment you have. Many different genes control these pigments.

Multiple alleles Polygenic Pleiotropy Epistasis

3. Someone with the Bombay phenotype cannot make the H protein (antigen). The H antigen is used to help form the A and B proteins (anitgens) that appear on red blood cells. Therefore even though they may have the A or B genes, they will appear as blood type O (lacking A or B antigens).

Multiple alleles Polygenic Pleiotropy Epistasis

Genetic Disorders

Many genetic disorders are caused by mutations in one or a few genes. Other genetic disorders are caused by abnormal numbers of chromosomes.

Before Reading: Think - Pair - Share

PREDICT: In general, which do you think would cause the most harm - mutations in a few genes, or an abnormal number of chromosomes? Explain why.

_____

_____

Genetic Disorders Caused by Mutations

Have you ever seen an animal or person with albinism? Not sure? If you've seen a white rat with pink eyes, you have. An albino organism will not have any skin, hair, or eye color because a genetic mutation has resulted in the organism's inability to produce the protein melanin, which is responsible for pigmentation. Table 3.2 lists several other genetic disorders caused by mutations. Mutations are a change in the DNA of a gene, resulting in a different protein or no

protein at all. Some of the disorders are caused by mutations in autosomal genes, others by mutations in X-linked genes.

Table 3:2: This table describes several genetic disorder caused by mutations in just one gene. Which disorder would you expect to be more common in males than females?

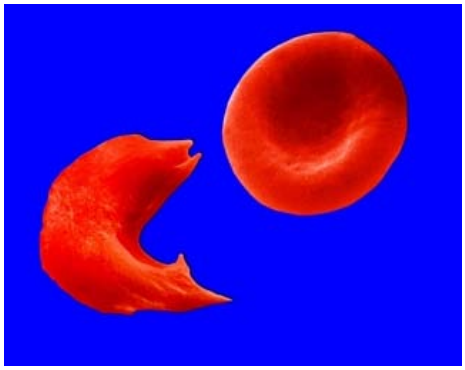
Genetic Disorders Caused by Mutations			
Genetic Disorder	**Direct Effect of Mutation**	**Signs and Symptoms of the Disorder**	**Mode of Inheritance**
Marfan syndrome	defective protein in connective tissue	heart and bone defects and unusually long, slender limbs and fingers	autosomal dominant
Sickle cell anemia	abnormal hemoglobin protein in red blood cells	sickle-shaped red blood cells that clog tiny blood vessels, causing pain and damaging organs and joints	autosomal recessive
Vitamin D-resistant rickets	lack of a substance needed for bones to absorb minerals	soft bones that easily become deformed, leading to bowed legs and other skeletal deformities	X-linked dominant
Hemophilia A	reduced activity of a protein needed for blood clotting	internal and external bleeding that occurs easily and is difficult to control	X-linked recessive

Few genetic disorders are controlled by dominant alleles. Can you guess why? A mutant dominant allele is expressed in every individual who inherits even one copy of it. If it causes a serious disorder, affected people may die young and fail to reproduce. Therefore, the mutant dominant allele is likely to die out of the population. One example of a serious genetic disorder that is controlled by a dominant allele is Huntington's disease. Huntington's disease causes increasingly impaired physical and cognitive ability. How does it stay in the population? The symptoms of Huntington's do not usually appear until mid-life (between ages 35-45), so someone with the disease is able to reproduce and pass on the gene.

A mutant recessive allele, such as the allele that causes sickle cell anemia (see Figure 3.12 and the link below), is not expressed in people who inherit just one copy of it. If they are heterozygous for the condition (they have one normal allele and one sickle cell allele) they are called **carriers**. They do not have the disorder themselves, but they carry the mutant allele and can pass it to their offspring. Because the allele can "hide" in a person, the allele is likely to pass on to the next generation rather than die out.

Figure 3.12: Sickle cell anemia is an autosomal recessive disorder. The mutation that causes the disorder affects just one amino acid in a single protein, but it has serious consequences for the affected person. This photo shows the sickle shape of red blood cells in people with sickle cell anemia.

Sickle-Shaped and Normal Red Blood Cells



Reading Check:

1. Why are most genetic disorders caused by recessive alleles rather than dominant alleles?

_____

_____

*2. If someone is a **carrier** of a disorder, do they show symptoms of the disorder? Explain why or why not.* _____

_____

Chromosomal Disorders

Chromosomal disorders are disorders caused by having the incorrect number of chromosomes. Instead of having 46 chromosomes someone might have 47 or 45. How could this happen? Mistakes may occur during meiosis that result in **nondisjunction**. This is the failure of chromosomes to separate correctly during meiosis I or II. Some of the resulting gametes will be missing a chromosome, while others will have an extra copy of the chromosome. If such gametes are fertilized and form zygotes, they usually do not survive. If they do survive, the individuals are likely to have serious genetic disorders. Table 3.3 lists several genetic disorders that are caused by abnormal numbers of chromosomes.

Word Work

*Let's break down the word "**nondisjunction**". The word "junction" means the state of being joined. The prefix "dis-" reverses the meaning of the root word. For example: dislike, discover, disinterest. What could a synonym for "**disjunction**" (the opposite of being joined) be?*

_____

The prefix "non-" means not. When you put it all together, how would you define the word "nondisjunction"? _____

Table 3.3: Having the wrong number of chromosomes causes the genetic disorders described in this table. Most chromosomal disorders involve the X chromosome. Look back at the X and Y chromosomes in Figure 3.3, and you will see why. The X and Y chromosomes are very different in size, so nondisjunction of the sex chromosomes occurs relatively often.

Genetic Disorders Caused by Abnormal Numbers of Chromosomes		
Genetic Disorder	**Genotype**	**Phenotypic Effects**
Down syndrome	extra copy (complete or partial) of chromosome 21	developmental delays, distinctive facial appearance, and other abnormalities
Turner's syndrome	one X chromosome but no other sex chromosome (XO)	female with short height and infertility (inability to reproduce)
Triple X syndrome	three X chromosomes (XXX)	female with mild developmental delays and menstrual irregularities
Klinefelter's syndrome	one Y chromosome and two or more X chromosomes (XXY, XXXY)	male with problems in sexual development and reduced levels of the male hormone testosterone

Reading Check:

1. *What can cause someone to have an extra or missing chromosome?*

_____

2. *Having a **chromosomal disorder** is most often so damaging that the zygote cannot survive. The effect of a **genetic mutation** can range greatly from a life-threatening disorder to a harmless change in appearance. Why would a chromosomal disorder usually have a much greater impact than a genetic mutation? (Hint: Think about how many genes can be found on one chromosome.)*

_____

_____

Diagnosing Genetic Disorders

A genetic disorder that is caused by a mutation can be inherited. Therefore, people with a genetic disorder in their family may be concerned about having children with the disorder. Professionals known as genetic counselors can help them understand the risks of their children being affected. If they decide to have children, they may be advised to have prenatal ("before birth") testing to see if the fetus has any genetic abnormalities. This is frequently done for Down syndrome for pregnant women over the age of 35. One method of prenatal testing is amniocentesis. In this procedure, a few fetal cells are extracted from the fluid surrounding the fetus, and the fetal chromosomes are examined.

Treating Genetic Disorders

The symptoms of genetic disorders can sometimes be treated, but cures for genetic disorders are still in the early stages of development. One potential cure that has already been used with some success is **gene therapy.** This involves inserting normal genes into cells with mutant genes. Then the cell can use the normal gene to produce the correct protein and avoid or correct the symptoms of the disorder.

Lesson 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.
- Most human traits have complex modes of inheritance. They may be controlled by one gene with multiple alleles or by multiple genes. More complexity may be introduced by pleiotropy (one gene, multiple effect) and epistasis (gene-gene interactions).
- Many genetic disorders are caused by mutations in one or a few genes. Other genetic disorders are caused by abnormal numbers of chromosomes.

Lesson Review Questions

Recall

1. Describe the inheritance pattern for a single-gene autosomal dominant trait, such as free-hanging earlobes.
2. Give an example of a multiple allele trait and a polygenic trait.
3. Identify factors that influence human skin color.
4. Describe a genetic disorder caused by a mutation in a single gene.
5. What causes Down syndrome?
6. What is gene therapy?

Apply Concepts

7. 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.

Think Critically

8. How might red-green color blindness might affect the health of a person with this trait?
9. Compare and contrast dominance and epistasis.
10. Explain why genetic disorders caused by abnormal numbers of chromosomes most often involve the X chromosome.

Points to Consider

Technology has been developed to cure some genetic disorders with gene therapy. This involves inserting normal genes into cells with mutations. Scientists use genetic technology for other purposes as well.

- What other genetic problems might scientists try to solve with genetic technology? What about problems in agriculture?
- Why might scientists want to alter the genes of other organisms? How might this be done?

Multimedia Links

You can watch a video explaining how pedigrees are used and what they reveal at this link:
http://www.youtube.com/watch?v=HbIHjsn5cHo

At the link below, you can watch an animation about another X-linked recessive trait called hemophilia A.
http://www.dnalc.org/view/16315-Animation-13-Mendelian-laws-apply-to-human-beings-.html

You can learn more about ABO blood type by watching the video at this link:
http://www.youtube.com/watch?v=CduLOqZb_XQ

You can watch a video about genetic disorders caused by mutations at this link:
http://www.pbs.org/wgbh/nova/programs/ht/rv/2809_03.html

You can click on any human chromosome at this link to see the genetic disorders associated with it:
http://www.ornl.gov/sci/techresources/Human_Genome/posters/chromosome/chooser.shtml

You can find out more information about sickle cell anemia at the website below:
http://www.dnalc.org/resources/3d/17-sickle-cell.html

The animation at the link below shows how replicated chromosomes can fail to separate during meiosis II:
http://learn.genetics.utah.edu/content/begin/traits/predictdisorder/index.html

At the link below, you can watch the video *Sickle Cell Anemia: Hope from Gene Therapy* to learn how scientists are trying to cure sickle-cell anemia with gene therapy.
http://www.pubinfo.vcu.edu/secretsofthesequence/playlist_frame.asp