In Lesson 7.2 we began our exploration of genetics. Mendel set the stage with his three “laws”: Law of Segregation, Law of Independent Assortment, and Law of Dominance. All of Mendel’s studies were conducted with genes that were located on different chromosomes, thus they could easily segregate, sort independently and express clear dominance. Today we know that certain genes are located on the same chromosomes and are inherited together. This knowledge opened the doors for scientists to explore genetics in ways we only dreamed were possible in the past.

This section of your textbook will begin by introducing the human genome project, explaining more about chromosomes and genes, discussing gene linkage and gene maps and reviewing what Mendel discovered. From there we will discuss Non-Mendelian types of inheritance, as seen in the picture above.

Lesson Objective

• Define the human genome.
• Describe human chromosomes and genes.
• Explain linkage and linkage maps.
• Describe inheritance in humans for autosomal and X-linked traits.
• Develop an understanding of pedigree charts.
• Identify complex Non-Mendelian modes of human inheritance.

Vocabulary

• autosome
• carrier
• genetic trait
• Human Genome Project
• linkage map
• linked genes
• multiple allele trait
• pedigree
• sex chromosome
• sex-linked gene
• sex-linked trait
• X-linked gene
• X-linked trait
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 then they are not completely identical. Why? The environment also influences human characteristics, and no two people have exactly the same environment. Recall we covered this in Lesson 7.2 under ‘Effects of Environment on Phenotypes’.

Human Inheritance

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 much more complex inheritance patterns. As scientists learn more about the human genome and our genes they are gaining more and more understanding of how they work.

The Human Genome Project

All 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 23 pairs of chromosomes. The human genome also includes noncoding sequences of DNA, as shown in Figure 7.23.

Figure 7.23: Human Genome, Chromosomes, and Genes. Each chromosome of the human genome contains many genes as well as noncoding intergenic (between genes) regions. Each pair of chromosomes is shown here in a different color.

Thanks to the Human Genome Project, scientists now know the DNA sequence of almost the entire human genome; approximately 92% to be exact. 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 the functionality of all the genes in the sequence. A summary of their findings to date is shown in Table 7.2 on the next page.

Our Molecular Selves video discusses the human genome, and is available at the following link:
http://www.genome.gov/25520211
Cracking the Code video was presented by NOVA and explains how the Human Genome Project mapped our genome, it is available at the following link:
https://www.youtube.com/watch?v=_lgSDVD4QEc
Table 7.2: The Human Genome Project started in 1990 and completed in 2003 has mapped all three billion base pairs of the human genome and identified approximately 92% of the functionality of human genes. The number of genes and base pairs per chromosome can be seen in the table below.

<table>
<thead>
<tr>
<th>Chromosomes</th>
<th>Number of genes</th>
<th>Number of base pairs</th>
</tr>
</thead>
<tbody>
<tr>
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<td>3000</td>
<td>240 million ~90% known</td>
</tr>
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<tr>
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<td>140 million ~95% known</td>
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<tr>
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<td>130 million ~85% known</td>
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<tr>
<td>Chromosome 10</td>
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<td>130 million ~95% known</td>
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<tr>
<td>Chromosome 11</td>
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<tr>
<td>Chromosome Y</td>
<td>200</td>
<td>50 million ~50% known</td>
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</table>
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 7.24.

Humans have an estimated 20,000 to 30,000 genes on their 23 pairs of chromosomes. This may sound like a lot, but it really is not. Far simpler species have almost as many genes as humans. However, human cells use splicing and other 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 are still unclear. 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](http://www.hhmi.org/biointeractive/dna/DNAi_coding_sequences.html)

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.

Figure 7.24: Human chromosomes 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. Humans have 23 pairs of chromosomes. Pairs 1-22 are autosomes. Pair 23 is the sex chromosomes. Females have two X chromosomes, and males have an X and a Y chromosome.
**Gene Linkage**

Genes that are located on the same chromosome are called linked genes. Alleles for these genes tend to segregate together during meiosis, unless they are separated by crossing-over. Recall 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 their alleles will be separated by crossing-over.

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?

**Mapping Linkage**

Linkage can be assessed by determining how often crossing-over occurs between two genes on the same chromosome. Genes on different (non-homologous) 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 lower the frequency of crossing-over, 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 7.25. A linkage map shows the locations of genes on a chromosome.

![Linkage Map](image)

**Figure 7.25:** 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. 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?

**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. The modes of Mendelian inheritance are autosomal dominant, autosomal recessive, X-linked dominant, and X-linked recessive. How Mendelian traits are inherited depends on whether the traits are controlled by genes on autosomes or the X chromosome.
Autosomal Traits

Of the 23 pairs of human chromosomes, 22 pairs are autosomes (numbers 1–22 in Figure 7.24). Autosomal traits are controlled by genes on one of the 22 human autosomes. 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. 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

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 7.26. Which form of these traits do you have? What are your possible genotypes for the traits? The chart in Figure 7.26 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.

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 7.27 on the next page. Which form of these traits do you have? What are your possible genotypes for the traits?

![Pedigree for Earlobe Attachment](image_url)

**Figure 7.26:** 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?

![Single Gene Autosomal Traits](image_url)

**Figure 7.27:** Widow’s peak and hitchhiker’s thumb are dominant traits controlled by a single autosomal gene.
X-Linked Traits—“Sex-Linked Traits”

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 7.24, the X chromosome is much larger than the Y chromosome. The X chromosome has about 1,400 genes, whereas the Y chromosome has about 200, 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 (which stands for sex-determining region Y gene), 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? At the link that follows, you can watch an animation that explains why:

http://www.hhmi.org/biointeractive/gender/Y_evolution.html

Traits controlled by genes on the sex chromosomes are called X-linked traits, or sex-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 7.28.

![Figure 7.28: 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?](image)

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. Since males only have one X chromosome they only need to inherit one recessive gene on their only X chromosome to have this trait, which is fairly common in males but relatively rare in females who have two X chromosomes and would need to inherit two recessive genes on both of their X chromosomes to have this trait (Figure 7.29).
Pedigree Charts

Earlier in this lesson under the topic of autosomal traits you were shown a pedigree chart for ear lobe attachment traits. A pedigree chart is a diagram that traces the phenotypes of an organism and its ancestors or a particular gene as it appears from one generation to the next. These type of charts are most commonly used when tracing lineage of humans, show dogs, and race horses. Humans use them mostly to predict the probability of the appearance of a genetic disorder and dog breeders and owners of race horses use pedigree charts to selectively breed animals with desirable traits.

Pedigree charts use a standardized set of symbols, squares to represent males and circles to represent females. The organism that are homozygous for the phenotype or gene that is being traced is represented by a filled-in (darkened) square or circle depending on their gender. Organisms that are heterozygous for the phenotype or gene being traced are known as carriers and are represented by a square or circle that is only half filled-in (darkened) depending on their gender.

Generational relationships are shown in the pedigree through connecting lines. Parents are connected by horizontal lines and a vertical line leads to an offspring. Each generation is usually identified by a Roman numeral (I, II, III, IV, etc.). Analysis of a pedigree chart is done using Mendelian principles of inheritance and establish whether a trait follows a dominant or recessive pattern of inheritance.

Most often humans look at pedigree charts to see whom within a family has been afflicted with a genetic disorder and to predict the likelihood of future generations inheriting the genetic disorder. If a pedigree shows a condition appearing in a 50:50 ratio between men and women it is considered an autosomal disorder. Examples of autosomal dominant conditions include baldness, astigmatism, and dwarfism. Autosomal recessive conditions include small eyes, little body hair, and tall stature. However if the condition predominantly affects men in the pedigree chart it is considered an X-linked (sex-linked) disorder. Examples of X-linked disorders include color blindness and hemophilia.

Figure 7.29: 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.
Pedigree Chart Analysis

Pedigree chart analysis can be a very useful tool when exploring autosomal dominant, autosomal recessive, and sex-linked recessive inheritance in a group of related individuals to determine the pattern of inheritance and characteristics of the trait. Let’s look at some rules of inheritance and sample pedigree charts.

General Rules of Inheritance
If the disorder is dominant, one of the parents must have the disorder.
If the disorder is recessive, neither parent has to have the disorder because they can be heterozygous.

Rules of Inheritance for Autosomal Recessive Conditions
Appears in both sexes with equal frequency
Trait tend to skip generations
Affected offspring are usually born to unaffected parents
When both parents are heterozygous, approx. 1/4 of the progeny will be affected
Appears more frequently among the children of marriages between related persons

Rules of Inheritance for Autosomal Dominant Conditions
Appears in both sexes with equal frequency
Both sexes transmit the trait to their offspring
Does not skip generations
Affected offspring must have an affected parent unless they possess a new mutation
When one parent is affected (heterozygous) and the other parent is unaffected, approx. 1/2 of the offspring will be affected
Unaffected parents do not transmit the trait
Rules of Inheritance for X-Linked Dominant
Both males and females are affected; often more females than males are affected
Does not skip generations
Affected sons must have an affected mother; affected daughters must have either an affected mother or an affected father
Affected fathers will pass the trait on to all their daughters
Affected mothers if heterozygous will pass the trait on to 1/2 of their sons and 1/2 of their daughters

Rules of Inheritance for X-Linked Recessive
More males than females are affected
Affected sons are usually born to unaffected mothers, thus the trait skips generations
Approximately 1/2 of carrier mothers’ sons are affected
It is never passed from father to son
All daughters of affected fathers are carriers

Non-Mendelian Inheritance
Most human traits have more complex modes of inheritance than simple Mendelian inheritance. Each characteristic Mendel investigated was controlled by one gene that had two possible alleles, one of which was completely dominant to the other. This resulted in just two possible phenotypes for each characteristic. Each characteristic Mendel studied was also controlled by a gene on a different (non-homologous) chromosome. As a result, each characteristic was inherited independently of the other characteristics. Geneticists now know that inheritance is often more complex than this.

In Lesson 7.2 we briefly looked at two types of non-Mendelian inheritance: codominance and incomplete dominance, see Figure 7.34. Each of these types of inheritance characteristics may be controlled by one gene with two alleles, but the two alleles may have a different relationship than the simple dominant-recessive relationship, both of the traits may blend (incomplete dominance) or both traits may be expressed (codominance).
Other types of non-Mendelian inheritance are controlled by multiple alleles or multiple genes. Traits controlled by multiple genes are called polygenic traits. 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. 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.

**Figure 7.34:** Incomplete Dominance: The flower has pink petals because of incomplete dominance of a red-petal allele and a recessive white-petal allele. Codominance: The flower has red and white petals because of codominance of red-petal and white-petal alleles.

**Codominance**

Codominance occurs when both alleles are expressed equally in the phenotype of the heterozygote. The red and white flower in the **Figure 7.35** below has codominant alleles for red petals and white petals.

**Figure 7.35:** Codominance. The flower has red and white petals because of codominance of red-petal and white-petal alleles.
Incomplete Dominance

Incomplete dominance occurs when the dominant allele is not completely dominant. Expression of the dominant allele is influenced by the recessive allele, and an intermediate phenotype results. The pink flower in Figure 7.36 has an incompletely dominant allele for red petals and a recessive allele for white petals.

![Figure 7.36: Incomplete Dominance. The flower has pink petals because of incomplete dominance of a red-petal allele and a recessive white-petal allele.]

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: \( I^A \), \( I^B \), and \( i \), which can be represented by the letters A, B, and O for blood phenotype. As shown in Table 7.3, 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 Figure 7.37 you can see the possible parental and alleles and offspring genotypes from when different blood types are combined. For example a Father whose gamete has blood type B and contributes an \( I^B \) allele who mates with a Mother who has blood type A and contributes an \( I^A \) can only produce a child who has blood type AB with the alleles \( I^A I^B \). Both \( I^A \) and \( I^B \) are dominant alleles, so the trait is also called codominance. The allele for blood type O (i) is a recessive trait. In Figure 7.38 you can see a Punnett Square cross between a parent with a heterozygous type B blood and a parent with codominant type AB blood and all of the possible offspring genotypes that they could produce.

You can learn more about ABO blood type by watching the video at this link: [https://www.youtube.com/watch?v=9O5JQqlngFY](https://www.youtube.com/watch?v=9O5JQqlngFY) (7:04).

<table>
<thead>
<tr>
<th>Genotype</th>
<th>Phenotype</th>
</tr>
</thead>
<tbody>
<tr>
<td>( I^A I^A ); OR ( I^A i )</td>
<td>A</td>
</tr>
<tr>
<td>( I^B I^B ); OR ( I^B i )</td>
<td>B</td>
</tr>
<tr>
<td>( I^A I^B )</td>
<td>AB</td>
</tr>
<tr>
<td>( ii )</td>
<td>O</td>
</tr>
</tbody>
</table>
Figure 7.38  Punnett squares can be used to determine the potential blood groups of children based on their parents' blood types.

Polygenic Characteristics

Polygenic characteristics are controlled by more than one gene, and each gene may have two or more alleles. The genes may be on the same chromosome or on non-homologous chromosomes.

- If the genes are located close together on the same chromosome, they are likely to be inherited together. However, it is possible that they will be separated by crossing-over during meiosis, in which case they may be inherited independently of one another.
- If the genes are on non-homologous chromosomes, they may be recombined in various ways because of independent assortment.

For these reasons, the inheritance of polygenic characteristics is very complicated. Such characteristics may have many possible phenotypes. Skin color and adult height are examples of polygenic characteristics in humans. Do you have any idea how many phenotypes each characteristic has?
Polygenic Traits

Many human traits are controlled by more than one gene. These traits are called polygenic traits (or characteristics). The genes may be on the same chromosome or on non-homologous chromosomes.

- If the genes are located close together on the same chromosome, they are likely to be inherited together. However, it is possible that they will be separated by crossing-over during meiosis, in which case they may be inherited independently of one another.
- If the genes are on non-homologous chromosomes, they may be recombined in various ways because of independent assortment.

For these reasons, the inheritance of polygenic characteristics is very complicated. Such characteristics may have many possible phenotypes. Skin color, hair color, eye color, and adult height are examples of polygenic characteristics in humans. Do you have any idea how many phenotypes each characteristic has?

For example, the human polygenic trait of adult height has several genes (each with more than one allele), so there are many possible adult heights. 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 7.39. Skin color is another polygenic trait, it is controlled by six different genes as can be seen in the Punnett Square in Figure 7.40 below.

![Height of North American Men](image)

**Figure 7.39:** Like many other polygenic traits, adult height has a bell-shaped distribution.

<table>
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**Figure 7.40** Punnett Square showing the variety of possible combinations from the six genes that contribute to human skin color.
Effects of Environment on Phenotype

Genes play an important role in determining an organism’s characteristics. However, for many characteristics, the individual’s phenotype is influenced by other factors as well. Environmental factors, such as sunlight and food availability, can affect how genes are expressed in the phenotype of individuals. Here are just two examples:

- Genes play an important part in determining our adult height. However, factors such as poor nutrition can prevent us from achieving our full genetic potential.
- Genes are a major determinant of human skin color. However, exposure to ultraviolet radiation can increase the amount of pigment in the skin and make it appear darker.

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 7.41, exposure to UV light darkens the skin.

![Figure 7.41: Effects of UV Light on Skin Color. 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.](image_url)

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 pairs are autosomes. The X and Y chromosomes are the sex chromosomes. Females have two X chromosomes, and males have one X and one Y. Human chromosomes contain a total of 20,000 to 22,000 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.
- 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 effects) and epistasis (gene-gene interactions).
- Many characteristics have more complex inheritance patterns than those studied by Mendel. They are complicated by factors such as codominance, incomplete dominance, multiple alleles, and environmental influences.
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