8.4: Mendelian Inheritance

Dimples

This man is exhibiting a genetic trait — the dimples in his cheeks when he smiles. Genetic traits are characteristics that are encoded in DNA. Some genetic traits, like dimples, have a simple inheritance pattern like the traits that Gregor Mendel studied in pea plants. The way these traits are inherited by offspring from their parents is called Mendelian inheritance.

What Is Mendelian Inheritance?

Mendelian inheritance refers to the inheritance of traits controlled by a single gene with two alleles, one of which may
be completely dominant to the other. The pattern of inheritance of Mendelian traits depends on whether the traits are controlled by genes on autosomes or by genes on sex chromosomes.

- **Autosomal traits** are controlled by genes on one of the 22 pairs of human autosomes. Autosomes are all the chromosomes except the X or Y chromosome, and they do not differ between males and females, so autosomal traits are inherited in the same way regardless of the sex of the parent or offspring.

- Traits controlled by genes on the sex chromosomes are called **sex-linked traits**. Because of the small size of the Y chromosome, most sex-linked traits are controlled by genes on the X chromosome. These traits are called **X-linked traits**. Single-gene X-linked traits have a different pattern of inheritance than single-gene autosomal traits because males have just one X chromosome. Males always inherit their X chromosome from their mother, and they pass on their X chromosome to all of their daughters but none of their sons.

### Studying Inheritance Patterns

There are two very useful tools for studying how traits are passed from one generation to the next. One tool is a pedigree, the other is a Punnett square.

#### Pedigree

The chart below is called a **pedigree**. A pedigree shows how a trait is passed from generation to generation within a family. A pedigree can show, for example, whether a Mendelian trait is an autosomal dominant, autosomal recessive, or X-linked trait. Pedigrees show relationships and identify individuals with a given trait.

The trait represented by the chart below is a hypothetical autosomal trait that is controlled by a dominant allele. At the top of the pedigree are symbols representing a married couple. The husband has the trait (affected male), but the wife does not (unaffected female). The next row of the pedigree shows the couple's children, as well as the spouses of three of the children. For example, the first child on the left is an affected male married to an unaffected female. The third row of the pedigree shows the next generation (the grandchildren of the couple at the top of the pedigree). One of the children in this generation, the affected female on the left, is married to an unaffected male. The other children are not married.
Figure \(\PageIndex{2}\): A pedigree chart is similar to a family tree. It shows how a trait is passed from parents to offspring in a family. The trait represented by this pedigree is an autosomal dominant trait. (CC BY-SA 3.0; Jerome Walker via Wikimedia.org)

Figure \(\PageIndex{3}\): Notice that neither of the parents at the top shows the trait because they are heterozygous and therefore show the dominant trait. However, their youngest son is homozygous recessive and therefore does show the trait (CC BY-SA 3.0; Jerome Walker via Wikimedia.org).

In an autosomal dominant trait, a child that has the trait will always have at least one parent with the trait. In an autosomal recessive trait, two individuals without the trait can have a child with the trait. The pedigree below shows an autosomal recessive trait.

**Punnett Square**

A **Punnett square** is a chart that allows you to easily determine the expected ratios of possible genotypes in the offspring of two parents. The mating between two parents is called a cross. The Punnett square is named after its developer, British geneticist Reginald C. Punnett. You can see a hypothetical example below. In this case, the gene is autosomal, and both parents are heterozygotes \((Aa)\) for the gene. Half the gametes produced by each parent will have
the $A$ allele and half will have the $a$ allele. That's because the two alleles are on homologous chromosomes, which always separate and go to separate gametes during meiosis. According to Mendel's law of segregation, the alleles in the gametes from each parent are written down the side and across the top of the Punnett square. Filling in the cells of the Punnett square gives the possible genotypes of their children. It also shows the most likely ratios of the genotypes, which in this case is 25 percent $AA$, 50 percent $Aa$, and 25 percent $aa$.

![Punnett Square](https://bio.libretexts.org/Bookshelves/Human_Biology/Book%3A_Human_Biology_(Wakim_and_Grewal)/08%3A_Inheritance/…)

**Predicting Genotypes and phenotype with Punnett Squares**

![Punnett Square](https://bio.libretexts.org/Bookshelves/Human_Biology/Book%3A_Human_Biology_(Wakim_and_Grewal)/08%3A_Inheritance/…)

Mendel developed the law of segregation by following only a single characteristic, such as pod color, in his pea plants. In a monohybrid cross, such as the one in Figure `PageIndex(5)`, the Punnett square shows every possible...
combination when combining one maternal (mother) allele with one paternal (father) allele. In this example, both organisms are heterozygous for flower color Bb (purple). Both plants produce gametes that contain both the B and b alleles. The probability of any single offspring showing the dominant trait is 3:1, or 75%. To develop a Punnett square, possible combinations of alleles in a gamete are placed on the top and left side of a square.

**Dihybrid cross**

For a monohybrid cross (above), we are only looking at a single gene. Therefore, the outside of the Punnett square will only have single letters (single alleles). For a dihybrid cross, pairs of alleles are used. This means the outside of the square will have pairs of letters. A Punnett square for a monohybrid cross is divided into four squares, whereas a Punnett square for a dihybrid cross is divided into 16 squares. How many boxes would a Punnett square need if three traits were examined? The squares are filled in with the possible combinations of alleles formed when gametes combine, such as in a zygote.

These types of crosses can be challenging to set up, and the square you create will be 4x4. This simple guide will walk you through the steps of solving a typical dihybrid cross common in genetics. The method can also work for any cross that involves two traits.

**Consider this cross**

A pea plant that is heterozygous for round, yellow seeds is self-fertilized, what are the phenotypic ratios of the resulting offspring?

**Step 1:** Determine the parental genotypes from the text above, the word "heterozygous" is the most important clue, and you would also need to understand that self-fertilized means you just cross it with itself.

\[RrYy \; x \; RrYy\]

**Step 2:** Using the figure below determine the gamete. Combine the R's and Ys of each parent to represent sperm and egg. Do this for both parents

![Diagram](https://bio.libretexts.org/Bookshelves/Human_Biology/Book%3A_Human_Biology_(Wakim_and_Grewal)/08%3A_Inheritance/...

Figure \(\PageIndex{6}\)): To determine the gametes for a dihybrid cross using this diagram. (CC BY-NC 3.0; Suzanne...
You should have come up with the following:

\[ \text{RY, Ry, rY, ry (parent 1) and RY, Ry, rY, ry (parent 2)} \]

Step 3: Set up a large 4x4 Punnett square, place one gamete set from the parent on the top, and the other on the side. See Figure \(\PageIndex{7}\).

Step 4: Write the genotypes of the offspring in each box and determine how many of each phenotype you have. In this case, you will have 9 round, yellow; 3 round, green; 3 wrinkled, yellow; and 1 wrinkled green (Figure \(\PageIndex{7}\)).

Sex 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. A Punnett square can also be used to show how the X and Y chromosomes are passed from parents to their children. This is illustrated in the Punnett square below. It may help you understand the inheritance pattern of sex-linked traits.

**Autosomal Mendelian Traits in Humans**

Single-gene autosomal traits include widow's peak and freckles, both of which are illustrated below. Widow's peak refers to a point in the hairline at the center of the forehead.

- Assume that the dominant and recessive alleles for the widow's peak gene are represented by \( W \) and \( w \), respectively. Because this is a dominant trait, people with the genotype \( WW \) and the genotype \( Ww \) will have a widow's peak, and only people with the genotype \( ww \) will not have the trait.
Figure \(\PageIndex{9}\): Widow's peak is a dominant trait that is controlled by a gene located on an autosomal chromosome. (Created by Mandeep Grewal from the pictures taken by (left: CC BY-NC 3.0; Jmblock2 via Wikimedia.org and right: CC BY-SA; Omer Mor via Wikimedia.org)

• Assume that the dominant and recessive alleles for freckles are represented by \(F\) and \(f\), respectively. Because it is a dominant trait, people with the genotype \(FF\) and the genotype \(Ff\) will have freckles, and only people with the genotype \(ff\) will not have the trait.

Which forms of these two traits do you have? What are your possible genotypes for the traits?

Figure \(\PageIndex{10}\): Having freckles is a single-gene autosomal dominant trait. (CC0; jameslockett via pixabay.com)

X-Linked Mendelian Traits in Humans

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 \(\PageIndex{11}\)). Boys are much more likely to be colorblind than girls (Table below). 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.

\[
\begin{align*}
\text{X} & : x^Cx^C \\
x & : x^C x
\end{align*}
\]
According to this Punnett square (Table above), the son of a woman who carries the colorblindness trait and a male with normal vision has a 50% chance of being colorblind. Figure \(\PageIndex{11}\) shows a simple pedigree for this trait.

![Pedigree for Color Blindness](https://bio.libretexts.org/Bookshelves/Human_Biology/Book%3A_Human_Biology_(Wakim_and_Grewal)/08%3A_Inheritance/...)

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 an X-linked recessive trait. This explains why X-linked recessive traits are less common in females than males and why they show a different pattern of inheritance than autosomal traits.

Another example of a recessive X-linked Mendelian trait is hemophilia. This is a disorder characterized by the inability of
blood to clot normally. England’s Queen Victoria, pictured below, was a carrier of the disorder. Two of Queen Victoria’s five daughters inherited the hemophilia allele from their mother and were carriers. When they married royalty in other European countries, they spread the allele across Europe, including the royal families of Spain, Germany, and Russia. Victoria’s son Prince Leopold also inherited the hemophilia allele from his mother and actually suffered from the disease. For these reasons, hemophilia was once popularly called “the royal disease.”

Feature: My Human Body

Are you color blind or think you might be? If you inherited this X-linked recessive disorder, a world without clear differences between certain colors seems normal to you. It’s all you have ever known. That’s why some people who are color blind are not even aware of it. Simple tests have been devised to determine whether a person is color blind and the degree of this visual deficit. An example of such a test is pictured below. What do you see when you look at this circle? Can you clearly perceive the number 74? If so, you probably have normal red-green color vision. If you cannot see the number, you may have red-green color blindness.

Figure \(\PageIndex{12}\): This circle of colors containing the number 74 is part of the Ishihara color blindness test. (Public domain via Wikimedia Commons)

Being color blind may cause a number of problems. These may range from minor frustrations to outright dangers. For example:

- If you are color blind, it may be difficult to color-coordinate clothing and furnishings. You may end up wearing color combinations that people with normal color vision think are odd or clashing.
- Many LED indicator lights are red or green. For example, power strips and electronic devices may have indicator lights to show whether they are on (green) or off (red).
- Test strips for pH, hard water, swimming pool chemicals, and other common tests are also often color coded. Litmus paper for testing pH, for example, turns red in the presence of an acid, but if you are color blind, you may not be able to read the test result.
• Do you like your steak well done? If you are color blind, you may not be able to tell if the meat is still undercooked (red) or grilled just right. You also may not be able to distinguish ripe (red) from unripe (green) fruits and vegetables such as tomatoes. And some foods, such as dark green spinach, may look more like mud than food and be totally unappetizing.

• Weather maps often are color coded. Is that rain (green) in your forecast or a wintry mix of sleet and freezing rain (pink or red)? If you can't tell the difference, you may go out on the roads when you shouldn't and put yourself in danger.

• Being able to distinguish red from green traffic lights may be a matter of life or death. This can be very difficult for someone with red-green color blindness. That's why in some countries, people with this vision defect are not allowed to drive.

Summary

• Mendelian inheritance refers to the inheritance of traits controlled by a single gene with two alleles, one of which may be completely dominant to the other. The pattern of inheritance of Mendelian traits depends on whether the traits are controlled by genes on autosomes or by genes on sex chromosomes.

• Two tools for studying inheritance are pedigrees and Punnett squares. A pedigree is a chart that shows how a trait is passed from generation to generation within a family. A Punnett square is a chart that shows the expected ratios of possible genotypes in the offspring of two parents.

• Examples of human autosomal Mendelian traits include dimples and earlobe attachment. Examples of human X-linked traits include red-green color blindness and hemophilia.

Review

1. Define genetic traits and Mendelian inheritance.
2. Explain why autosomal and X-linked Mendelian traits have different patterns of inheritance.
3. What is a pedigree, and why is it useful for studying how traits are passed from one generation to the next?
4. What is a Punnett square, and what does it show?
5. Identify examples of human autosomal and X-linked Mendelian traits.
6. Imagine a hypothetical human gene that has two alleles, Q and q. Q is dominant to q and the inheritance of this gene is Mendelian. Answer the following questions about this gene.
   a. If a woman has the genotype Qq and her husband has the genotype QQ, list each of their possible gametes and what proportion of their gametes will have each allele.
   b. What are the likely proportions of their offspring being QQ, Qq, or qq?
   c. Is this an autosomal trait or an X-linked trait? How do you know?
   d. What are the chances of their offspring exhibiting the dominant Q trait? Explain your answer.
7. Explain why fathers always pass their X chromosome down to their daughters.
8. True or False. Women are more likely to have X-linked diseases than men.
9. True or False. Most human autosomal traits are controlled by a single gene with two alleles, similar to Mendel’s pea plants.

10. For each of the scenarios below, choose whether you would use a Punnett square or a pedigree. Choose only the one that best fits the scenario.
   
a. A man and a woman have known genotypes and you want to predict the possible genotypes of their offspring.

b. You want to document which members of your family had or have breast cancer.

Explore More

https://bio.libretexts.org/link?16763#Explore_More

Hemophilia is a rare genetic disorder resulting in uncontrolled bleeding. Learn more about the genetic inheritance of this disease here:

https://www.youtube.com/watch?v=0_Em...ature=youtube

Explore more on inheritance and sex-linked traits here:
Media, iframe, embed and object tags are not supported inside of a PDF.