Introduction to Genetics

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CHAPTER 1 Introduction to Genetics

Lesson Objectives

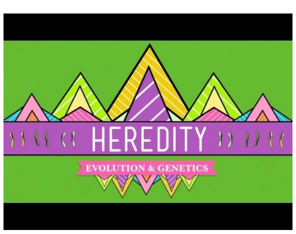
- Define gene and allele.
- Describe the relationship between genotype and phenotype.
- Show how to predict genotype and phenotype ratios in offspring for simple traits.
- Identify ways traits may be more complex than those studied by Mendel.
- Explain how sex-linked traits are inherited.

Lesson Vocabulary

- allele
- autosome
- genotype
- heterozygote
- homozygote
- phenotype
- Punnett square
- sex chromosome
- sex-linked trait

Introduction

When Mendel's laws were rediscovered in 1900, scientists were starting to learn about the molecules of heredity. They had already observed chromosomes and seen cells undergoing meiosis. Within a few decades they would learn the structure of DNA and how proteins are made. They would also learn that Mendel's "factors" consist of DNA. We now call these factors genes. For a great review of Mendel's work and an excellent introduction to this lesson, watch this entertaining video: https://www.youtube.com/watch?v=CBezq1fFUEA .



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Genes and Alleles

Today we know that the traits of organisms are controlled by genes on chromosomes. A gene can be defined as a section of a chromosome that contains the genetic code for a particular protein. The position of a gene on a chromosome is called its locus. Each gene may have different versions. The different versions are called **alleles**. **Figure** 1.1 shows an example in pea plants. It shows the gene for flower color. The gene has two alleles. There is a purple-flower allele and a white-flower allele. Different alleles account for most of the variation in the traits of organisms within a species.

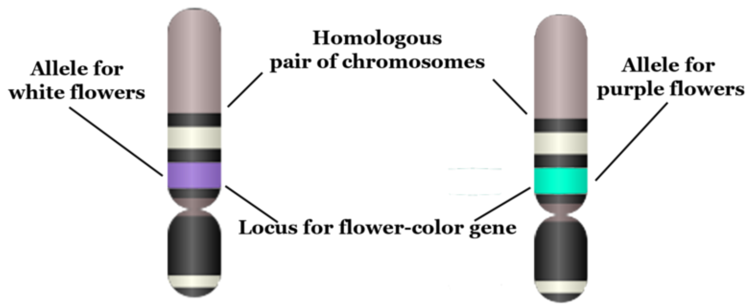

FIGURE 1.1
This diagram shows how genes and alleles are related.

In sexually reproducing species, chromosomes are present in cells in pairs. Chromosomes in the same pair are called homologous chromosomes. They have the same genes at the same loci. These may be the same or different alleles. During meiosis, when gametes are produced, homologous chromosomes separate. They go to different gametes. Thus, the alleles for each gene also go to different gametes.

Genotype and Phenotype

When gametes unite during fertilization, the resulting zygote inherits two alleles for each gene. One allele comes from each parent.

Genotype

The two alleles that an individual inherits make up the individual's **genotype**. The two alleles may be the same or different. Look at **Table** 1.1. It shows alleles for the flower-color gene in peas. The alleles are represented by the letters B (purple flowers) and b (white flowers). A plant with two alleles of the same type (BB or bb) is called a **homozygote**. A plant with two different alleles (Bb) is called a **heterozygote**.

TABLE 1.1: Genotypes and phenotypes for alleles B and b, with B dominant to b

Genotypes	Phenotypes
BB (homozygote)	purple flowers
Bb (heterozygote)	purple flowers
bb (homozygote)	white flowers

Phenotype

The expression of an organism's genotype is called its **phenotype**. The phenotype refers to the organism's traits, such as purple or white flowers. Different genotypes may produce the same phenotype. This will be the case if one allele is dominant to the other. Both BB and Bb genotypes in Table 6.1 have purple flowers. That's because the B allele is dominant to the b allele, which is recessive. The terms *dominant* and *recessive* are the terms Mendel used to describe his "factors." Today we use them to describe alleles. In a Bb heterozygote, only the dominant B allele is expressed. The recessive b allele is expressed only in the bb genotype.

Mendelian Inheritance

Each trait Mendel studied was controlled by one gene with two alleles. In each case, one of the alleles was dominant to the other. This resulted in just two possible phenotypes for each trait. Each trait Mendel studied was also controlled by a gene on a different chromosome. As a result, each trait was inherited independently of the others. With traits like these, it's easy to predict which forms of a trait will show up in the offspring of a given set of parents.

Predicting Alleles in Gametes

Consider a purple-flowered pea plant with the genotype Bb. Half the gametes produced by this parent will have a B allele. The other half will have a b allele. You can see this in **Figure** 1.2. This is similar to tossing a coin. There is a 50 percent chance of a head and a 50 percent chance of a tail. Like a head or tail, there is a 50 percent chance that any gamete from this parent will have the B allele. There is also a 50 percent chance that any gamete will have the b allele.

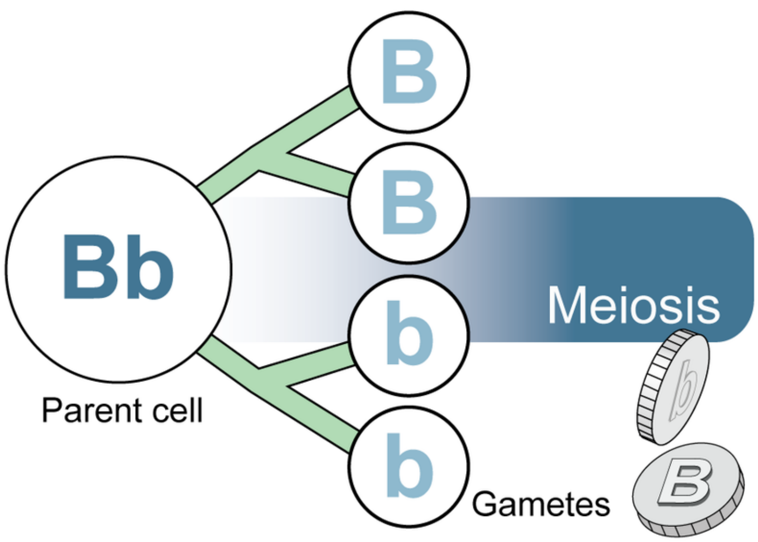

FIGURE 1.2
Gametes from a heterozygote parent (Bb)

Predicting Genotype Ratios

Now let's see what happens if two parent pea plants have the Bb genotype. What genotypes are possible for their offspring? And what ratio of genotypes would you expect? The easiest way to find the answer to these questions is with a Punnett square.

A **Punnett square** is a chart that makes it easy to find the possible genotypes in offspring of two parents. **Figure** 1.3 shows a Punnett square for the two parent pea plants. The gametes produced by the male parent are at the top of the chart. The gametes produced by the female parent are along the left side of the chart. The different possible combinations of alleles in their offspring can be found by filling in the cells of the chart.

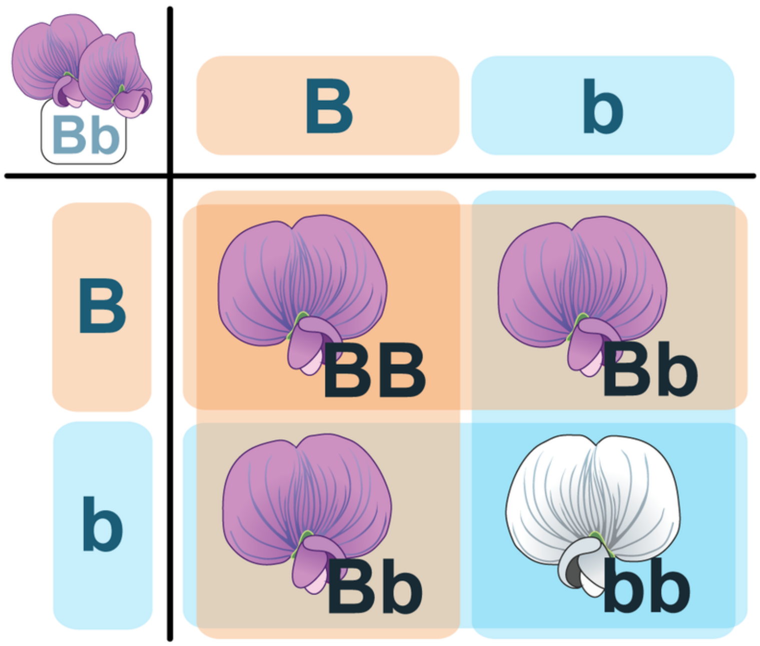

FIGURE 1.3
Punnett square for two Bb parents

If the parents had four offspring, their most likely genotypes would be one BB, two Bb, and one bb. But the genotype ratios of their actual offspring may differ. That's because which gametes happen to unite is a matter of chance, like a coin toss. The Punnett square just shows the possible genotypes and their most likely ratios.

Predicting Phenotype Ratios

You know that the B allele is dominant to the b allele. Therefore, you can also use the Punnett square in **Figure** 1.3 to predict the most likely offspring phonotypes. If the parents had four offspring, their most likely phenotypes would be three with purple flowers (1 BB + 2 Bb) and one with white flowers (1 bb).

Non-Mendelian Inheritance

Inheritance is often more complex than it is for traits like those Mendel studied. Several factors can complicate it.

Codominance and Incomplete Dominance

If a gene has two alleles, one may not be dominant to the other. There are other possibilities. One possibility is called codominance. Another is called incomplete dominance.

- With codominance, both alleles are expressed equally in heterozygotes. The red and white flower in **Figure** 1.4 has codominant alleles for red petals and white petals.

- With incomplete dominance, a dominant allele is not completely dominant. Instead, it is influenced by the recessive allele in heterozygotes. The pink flower in **Figure** 1.4 is an example. It has an incompletely dominant allele for red petals. It also has a recessive allele for white petals. This results in a flower with pink petals.

FIGURE 1.4
Codominance (left) and incomplete dominance (right)

Multiple Alleles

Many genes have more than two alleles. An example is ABO blood type in people. There are three common alleles for the gene that controls this trait. The allele for type A is codominant with the allele for type B. Both of these alleles are dominant to the allele for type O. The possible genotypes and phenotypes for this trait are shown in **Table** below

TABLE 1.2: ABO genotypes and phenotypes

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

Polygenic Traits

Some traits are controlled by more than one gene. They are called polygenic traits. Each gene for a polygenic trait may have two or more alleles. The genes may be on the same or different chromosomes. Polygenic traits may have many possible phenotypes. Skin color and adult height are examples of polygenic traits in humans. Think about all the variation in the heights of adults you know. Normal adults may range from less than 5 feet tall to more than 7 feet tall. There are people at every gradation of height in between these extremes.

Environmental Influences

Genes play an important role in determining an organism's traits. However, for many traits, phenotype is influenced by the environment as well. For example, skin color is controlled by genes but also influenced by exposure to sunlight. You can see the effect of sunlight on skin in **Figure** 1.5.

FIGURE 1.5
Skin color darkens when exposed to the sun.

Sex Chromosomes and Sex-Linked Traits

Animals and most plants have two special chromosomes. They are called **sex chromosomes**. These are chromosomes that determine the sex of the organism. All of the other chromosomes are called **autosomes**. Genes on sex chromosomes may be inherited differently than genes on autosomes.

Human Sex Chromosomes

In people, the sex chromosomes are called X and Y chromosomes. Individuals with two X chromosomes are normally females. Individuals with one X and one Y chromosome are normally males. As you can see in **Figure** 1.6, mothers pass an X chromosome to each of their children. Fathers pass an X to their daughters and a Y to their sons.

Sex-Linked Traits

Traits controlled by genes on the sex chromosomes are called **sex-linked traits**. One gene on the Y chromosome determines male sex. There are very few other genes on the Y chromosome, which is the smallest human chromosome. There are hundreds of genes on the much larger X chromosome. None is related to sex. Traits controlled by genes on the X chromosome are called X-linked traits.

X-linked traits have a different pattern of inheritance than traits controlled by genes on autosomes. With just one X chromosome, males have only one allele for any X-linked trait. Therefore, a recessive X-linked allele is always expressed in males. With two X chromosomes, females have two alleles for any X-linked trait, just as they do for autosomal traits. Therefore, a recessive X-linked allele is expressed in females only when they inherit two copies of it. This explains why X-linked recessive traits show up less often in females than males.

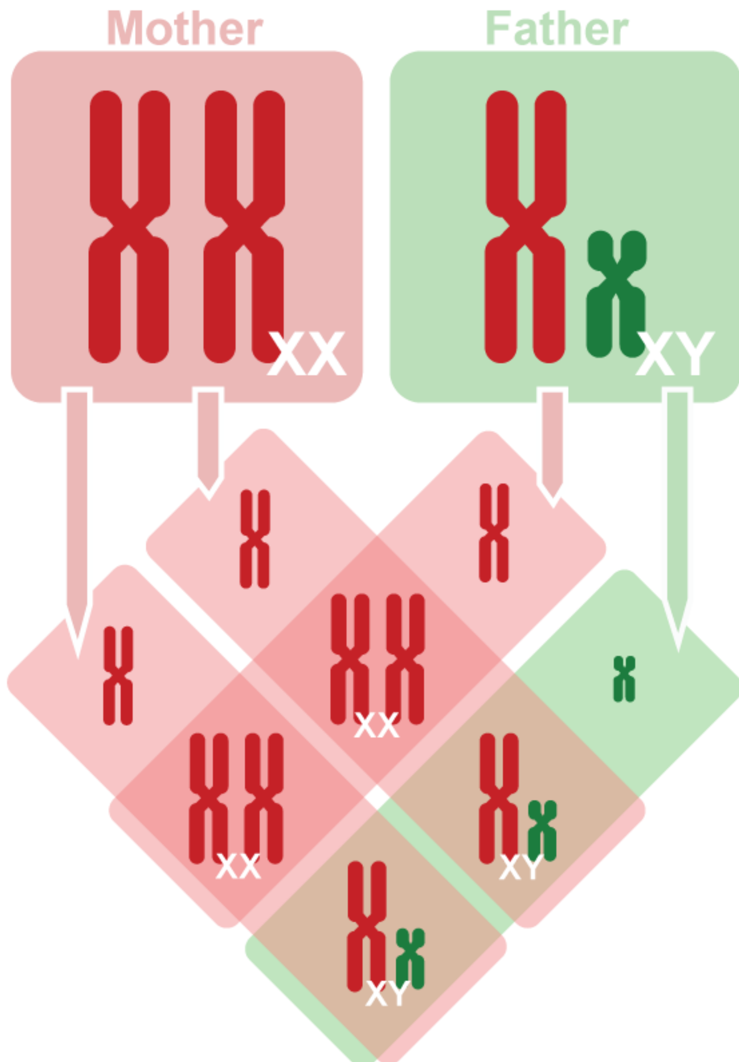

FIGURE 1.6
Inheritance of sex chromosomes

Inheritance of Color Blindness

An example of a recessive X-linked trait is red-green color blindness. People with this trait can't see red or green colors. This trait is fairly common in males but rare in females. **Figure** 1.7 is a pedigree for this trait. A pedigree is a chart that shows how a trait is inherited in a family. The mother has one allele for color blindness. She doesn't have color blindness because she also has a dominant normal allele for the gene. Instead, she is called a carrier for the trait. She passes the allele to half of her children. One daughter is a carrier, and one son has the color blindness trait. No matter how many children this couple has, none of the daughters will have color blindness, but half of the sons, on average, will have the trait. Can you explain why?

Lesson Summary

- Traits are controlled by genes on chromosomes. A gene may have different versions called alleles.
- The two alleles for a gene that an individual inherits make up the individual's genotype. The expression of the genotype as a trait is the individual's phenotype.

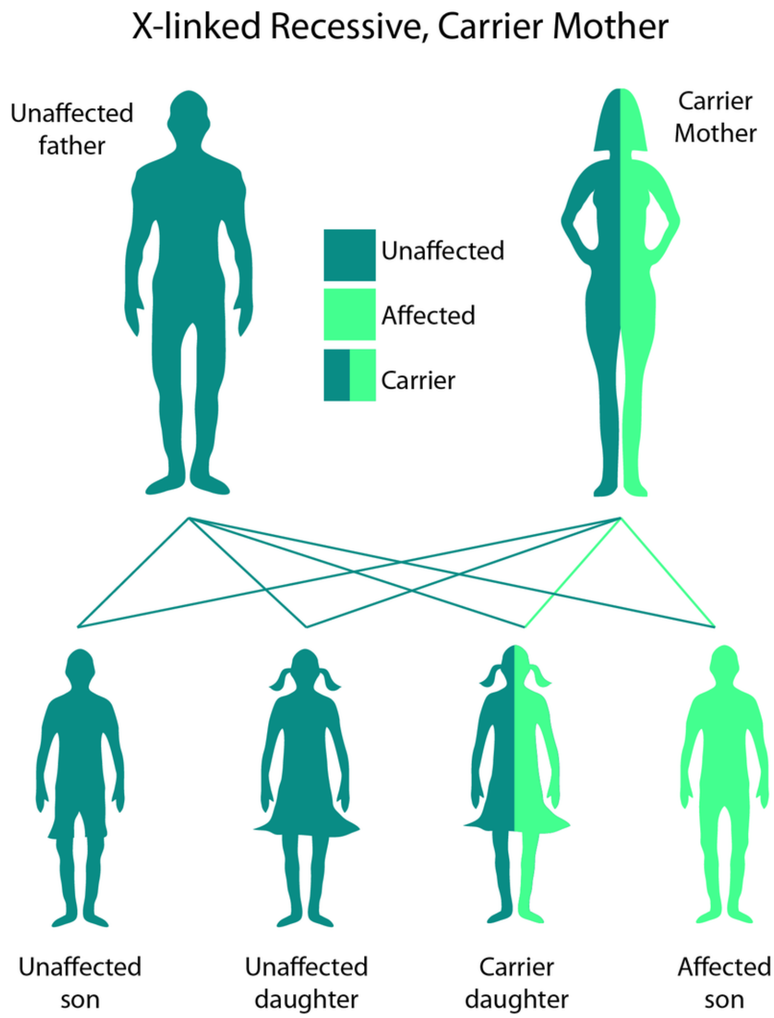

FIGURE 1.7
Pedigree for color blindness

- Mendel studied simple traits controlled by one gene with two alleles and dominance. For traits like these, Punnett squares can be used to predict possible genotypes and phenotypes and their likely ratios in offspring.
- Inheritance is more complex for traits in which there is codominance or incomplete dominance. Traits may also be controlled by multiple alleles or multiple genes. Many traits are influenced by the environment as well.
- Sex chromosomes determine sex in animals and many plants. Other chromosomes are called autosomes. Sex-linked traits are controlled by genes on sex chromosomes. They may be inherited differently than autosomal traits.

Lesson Review Questions

Recall

1. Write a short paragraph in which you correctly use the concepts chromosome, gene, allele, locus, and trait.
2. What are codominance and incomplete dominance? Give an example of each.
3. What is the difference between a multiple allele trait and a polygenic trait?

Apply Concepts

4. Use a Punnett square to determine the possible offspring genotypes of parents with the genotypes Bb and bb. Assume that B is the dominant allele for violet flower color in peas and b is the recessive allele for white flower color. What is the expected ratio of violet-flowered to white-flowered offspring based on your Punnett square?

Think Critically

5. Compare and contrast genotype and phenotype.
6. Explain why it is the father rather than the mother who determines the sex of their offspring.

Points to Consider

Genetics began with the rediscovery of Mendel's laws in 1900. There have been many advances in genetics since then.

1. What are some recent advances in genetics?
2. What do we now know about human genes?

References

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