Chapter 7

Mendelian Genetics

7.1 Lesson 7.1: Mendel's Investigations

Lesson Objectives

- Identify how Mendel's study of science and math was important to his success in research.
- Distinguish between characteristics and traits.
- Explain how Mendel was able to control pollination of the pea plants.
- Identify the terms used to describe the three generations in Mendel's studies.
- State one reason for carrying out a monohybrid cross.
- Identify the traits that appeared in Mendel's F_2 generation.
- Identify the actions of dominant alleles and recessive alleles for a trait.
- Outline the Law of Segregation.
- Outline the Law of Independent Assortment.
- Explain Mendel's results in relation to genes and chromosomes.
- Distinguish between genotype and phenotype.

Introduction

For thousands of years, humans have understood that characteristics such as eye color or flower color are passed from one generation to the next. The passing of characteristics from parent to offspring is called **heredity**. Humans have long been interested in understanding heredity. Many hereditary mechanisms were developed by scholars but were not properly tested or quantified. The scientific study of genetics did not begin until the late 19th century. In experiments with garden peas, Austrian monk Gregor Mendel described the patterns of inheritance.

Gregor Mendel: Teacher and Scientist

Gregor Johann Mendel was an Augustinian monk, a teacher, and a scientist (**Figure** 7.1). He is often called the "father of modern genetics" for his study of the inheritance of traits in pea plants. Mendel showed that the inheritance of traits follows particular laws, which were later named after him. The significance of Mendel's work was not recognized until the turn of the 20th century. The rediscovery of his work led the foundation for the era of modern **genetics**, the branch of biology that focuses on heredity in organisms.

Figure 7.1: Gregor Johann Mendel "The Father of Modern Genetics." 1822-1884. (3)

Johann Mendel was born in 1822 and grew up on his parents' farm in an area of Austria that is now in the Czech Republic. He overcame financial hardship and ill health to excel in school. In 1843 he entered the Augustinian Abbey in Brünn (now Brno, Czech Republic.) Upon entering monastic life, he took the name Gregor. While at the monastery, Mendel also attended lectures on the growing of fruit and agriculture at the Brünn Philosophical Institute. In 1849 he accepted a teaching job, but a year later he failed the state teaching examination. One of his examiners recommended that he be sent to university for further studies. In 1851 he was sent to the University of Vienna to study natural science and mathematics. Mendel's time at Vienna was very important in his development as a scientist. His professors encouraged him to learn science through experimentation and to use mathematics to help explain observations of natural events. He returned to Brünn in 1854 as a natural history and physics teacher.

Mendel's Experiments

In 1853 and 1854, Mendal published two papers on crop damage by insects. However, he is best known for his later studies of the pea plant *Pisum sativum*. Mendel was inspired by both his professors at university and his colleagues at the monastery to study variation in plants. He had carried out artificial fertilization on plants many times in order to grow a plant with a new color or seed shape. **Artificial fertilization** is the process of transferring pollen from the male part of the flower to the female part of another flower. Artificial fertilization is done in order to have seeds that will grow into plants that have a desired trait, such as yellow flowers.

During Mendel's time, the popular **blending inheritance** hypothesis stated that offspring were a "mix" of their parents. For example, if a pea plant had one short parent and one tall parent, that pea plant would be of medium height. It was believed that the offspring would then pass on heritable units, or factors, for medium sized offspring. (Today we know these heritable units are genes; however, Mendel did not know of the concept of a gene.) Mendel noted that plants in the monastery gardens sometimes gave rise to plants that were not exactly like the parent plants, nor were they a "mix" of the parents. He also noted that certain traits reappeared after "disappearing" in an earlier generation. Mendel was interested in finding out if there was a predictable pattern to the inheritance of traits. Between 1856 and 1863 he grew and tested about 29,000 pea plants in the monastery garden.

Mendel may have chosen to study peas because they are fast-growing plants that are available in different varieties. For example, one variety of pea plant has purple flowers, as shown in **Figure** 7.2, while another variety has white flowers.

Mendel chose to study seven characteristics of pea plants. A **characteristic** is a heritable feature, such as flower color. Each characteristic Mendel chose to study occurred in two contrasting traits. A **trait** is a heritable variant of a characteristic, such as purple or white flower color. Table 7.1 lists the seven characteristics Mendel studied, and their two contrasting traits.

Table 7.1: **The Seven Characteristics Mendel Studied and Their Contrasting Traits**

Flower Color	Flower Position on Stem	Stem Length	Pod Shape	Pod Color	Seed Shape	Seed Color
violet-red (purple)	axial	tall	inflated	green	round	green
white	terminal	short	constricted	yellow	wrinkled	yellow

Figure 7.2: *Pisum sativum,* the pea plant species that Mendel studied. (2)

Pea Plant Pollination

In order to study these characteristics, Mendel needed to control the pollination of the pea plants. Pollination occurs when the pollen from the male reproductive part of a flower, called the anthers, is transferred to the female reproductive part of a flower, called the stigma. Pea plants are **self-pollinating**, which means the pollen from a flower on a single plant transfers to the stigma of the same flower or another flower on the same plant. In order to avoid self-pollination, Mendel removed the anthers from the flowers on a plant. He then carefully transferred pollen from the anthers of another plant and dusted the pollen onto the stamen of the flowers that lacked anthers. This process caused cross-pollination. **Cross-pollination** occurs when pollen from one flower pollinates a flower on a different plant. In this way, Mendel controlled the characteristics that were passed onto the offspring. **Figure** 7.3 shows the location of the male and female parts of *P. sativum.*

Mendel's First Experiment

Mendel began his studies by growing plants that were true-breeding for a particular trait. A **true-breeding** plant will always produce offspring with that trait when they self-pollinate. For example, a true-breeding plant with yellow seeds will always have offspring that have yellow seeds. In his first experiment, Mendel cross-pollinated two true-breeding plants of contrasting traits, such as purple and white flowered plants. The true-breeding parent plants are referred to as the **P generation** (parental generation). The hybrid offspring of the P

Figure 7.3: The location of the anthers in the pea flower. The anthers are illustrated alone in the image to the left of the transected flower (at right). Mendel controlled pollination of the plants by removing the immature anthers of certain plants. (8)

generation are called the **F_1 generation** (filial generation). The hybrid offspring of the F_1 generation are called the F_2 generation (filial generation).

Monohybrid Crosses

Mendel first worked with plants that differed in a single characteristic, such as flower color. A hybridization is a cross between two individuals that have different traits. A hybridization in which only one characteristic is examined is called a **monohybrid cross**. The offspring of such a cross are called **monohybrids**. Mendel noted that hybridizing true-breeding (P-generation) plants gave rise to an F_1 generation that showed only one trait of a characteristic. For example, a true-breeding purple-flowering plant crossed with a true-breeding white-flowering plant always gave rise to purple-flowered hybrid plants. There were no white-flowered hybrids! Mendel wanted to know what happened to the white-flowered plants' "heritable factors." If indeed the white-flower "heritable factor" had disappeared, all future offspring of the hybrids would be purple-flowered. To test this idea, Mendel let the F_1 generation plants self-pollinate and then planted the resulting seeds.

Mendel's Results

The F_2 generation plants that grew included white-flowered plants! Mendel noted the ratio of white flowered plants to purple-flowered plants was about 3:1. That is, for every three purple-flowered plants, there was one white flowered plant. **Figure** 7.4 shows Mendel's results for the characteristic of flower color.

Mendel carried out identical studies over three generations, (P, F_1, and F_2), for the other six characteristics and found in each case that one trait "disappeared" in the F_1 generation, only to reappear in the F_2 generation. Mendel studied a large number of plants, as shown in **Table** 7.2, so he was confident that the ratios of different traits in the F_2 generation were representative.

Table 7.2: **Results of F1 Generation Crosses for Seven Characteristics in**

Characteristic	Dominant Trait	Recessive Trait	F2 Generation Dominant:Recessive	Ratio
Flower color	Purple	White	705:224	3.15:1
Flower position on stem	Axial	Terminal	651:207	3.14:1
Stem length	Tall	Short	787:277	2.84:1
Pod shape	Inflated	Constricted	882:299	2.95:1
Pod color	Green	Yellow	428:152	2.82:1

Table 7.2: (continued)

Characteristic	Dominant Trait	Recessive Trait	F2 Generation Dominant:Recessive	Ratio
Seed shape	Round	Wrinkled or angular	5474:1850	2.96:1
Seed color	Yellow	Green	6022:2001	3.01:1

Mendel's Theory of Heredity

Based on his observations, Mendel developed four hypotheses. These hypotheses are known as Mendel's theory of heredity. The hypotheses explain a simple form of inheritance in which two alleles of a gene are inherited to result in one of several traits in offspring. In modern terms, these hypotheses are:

1. **There are different versions of genes**. These different versions account for variations in characteristics. Different versions of a gene are called **alleles.** For example, there is a "yellow-pod" allele and a "green pod" allele. The blending inheritance hypothesis was discredited by Mendel's allele hypothesis.
2. **When two different alleles are inherited together, one may be expressed, while the effect of the other may be "silenced."** In the case of pod color, the allele for green pods is always expressed and is **dominant**. The allele for yellow pods, which is not expressed, is **recessive**. For instance, if a plant inherits a "yellow-pod" gene and a "green pod" gene, it will have only green pods.
3. **For each characteristic, an organism inherits two alleles, one from each parent**. Mendel noted that offspring could inherit their traits from either parent. In the case of the expressed trait, it did not matter whether it was the male gamete or female gamete that supplied the gene.
4. **When gametes are formed, the two alleles of each gene are separated** (**Figure** 7.5). During meiosis, each male or female gamete receives one allele for a trait. When the male and female gametes are fused at fertilization, the resulting zygote contains two alleles of each gene.

Random Segregation of Alleles

The **Law of Segregation** states that a pair of alleles is separated, or segregated, during the formation of gametes. During meiosis, homologous chromosomes are randomly separated. Each resulting gamete has an equal probability or chance of receiving either of the two alleles.

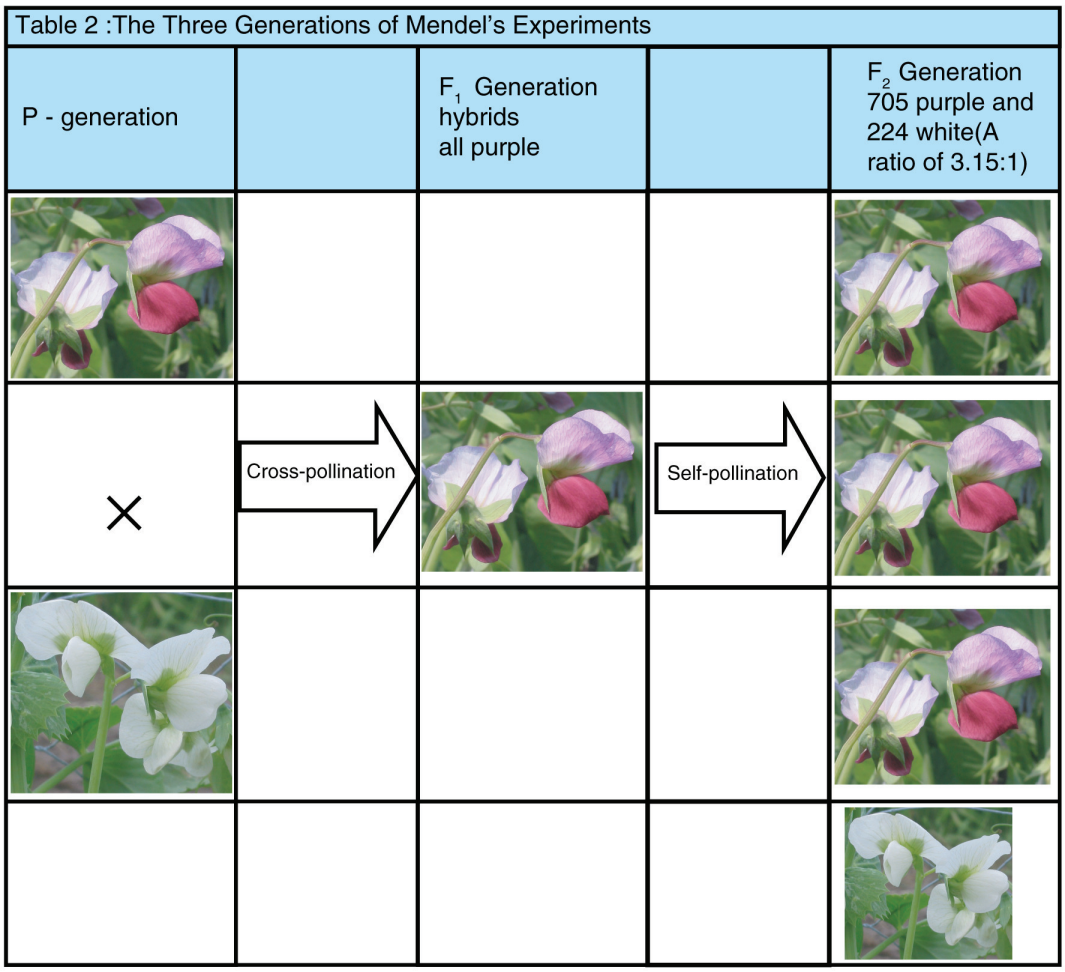
Table 2 :The Three Generations of Mendel's Experiments
P - generation
F1 Generation hybrids all purple
F2 Generation 705 purple and 224 white(A ratio of 3.15:1)
Cross-pollination
Self-pollination
×

Figure 7.4: (6)

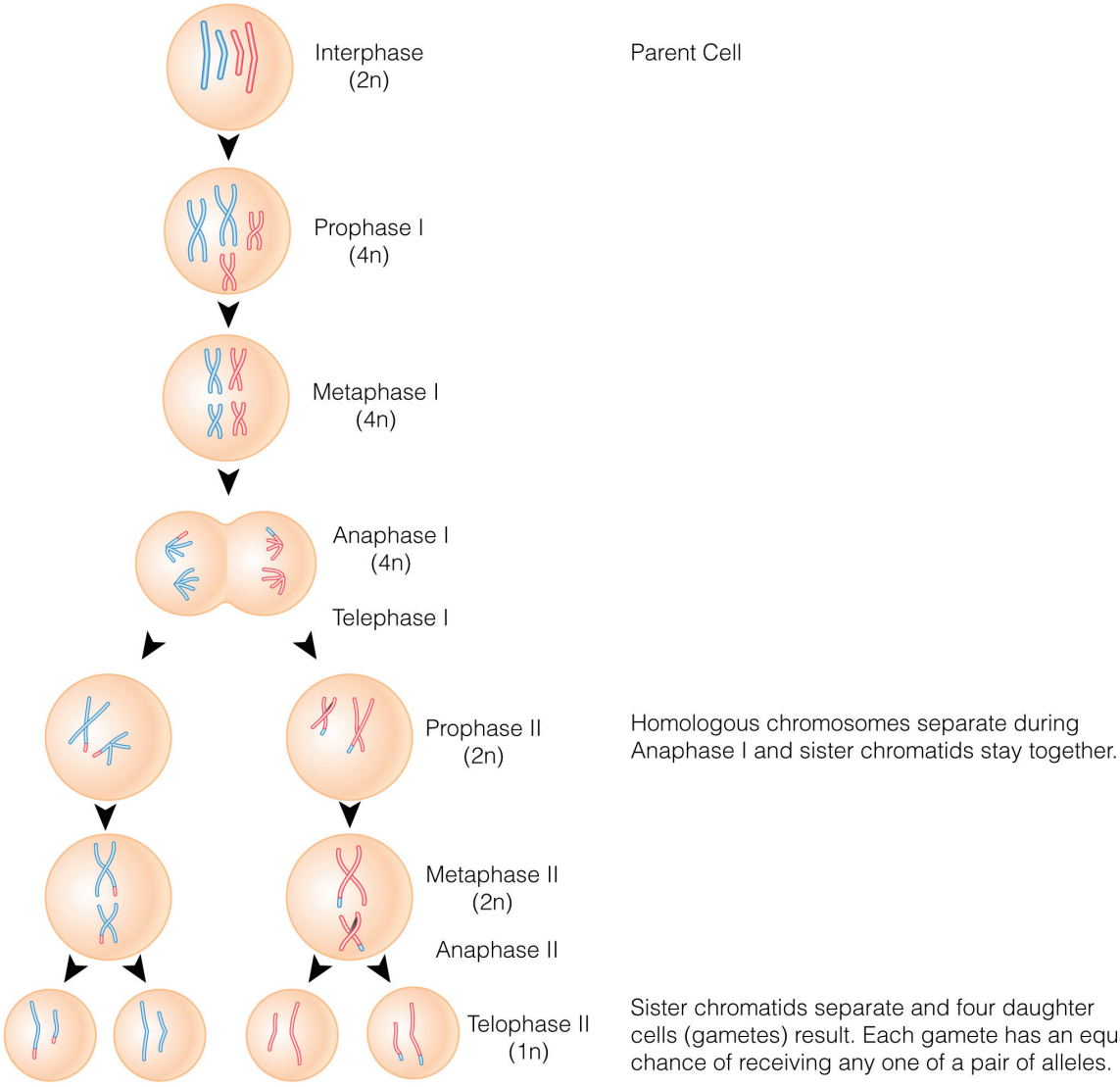

Figure 7.5: Alleles on homologous chromosomes are randomly separated during gamete formation. Upon fertilization, the fusion of a male and female gametes results in new combinations of alleles in the resulting zygote. (15)

Mendel's Second Experiment

Mendel also crossed pea plants that differed in two characteristics, such as seed color and shape. A **dihybrid cross** is a cross in which the inheritance of two characteristics are tracked at the same time. The offspring of such a cross are called **dihybrids**. Mendel wanted to see if the inheritance of characteristics were dependent. He concluded that characteristics were inherited independently of each other.

The Law of Independent Assortment

The **Law of Independent Assortment**, also known as or Mendel's Second Law, states that the inheritance of one trait will not affect the inheritance of another. Mendel concluded that different traits are inherited independently of each other, so that there is no relationship, for example, between seed color and seed shape. In modern terms, alleles of each gene separate independently during gamete formation.

Linked Genes on Chromosomes

We now know that the only alleles that are inherited independently are ones that are located far apart on a chromosome or that are on different chromosomes. There are many genes that are close together on a chromosome, and are packaged into the gametes together. Genes that are inherited in this way are called **linked genes**. Linked genes tend to be inherited together because they are located on the same chromosome. Genetic linkage was first discovered by the British geneticists William Bateson and Reginald Punnett shortly after Mendel's laws were rediscovered.

Mendelian Theory and Molecular Genetics

Mendel was perhaps lucky in that the characteristics he chose to study in the pea plants had a relatively simple pattern of inheritance. These characteristics were determined by one gene for which there were exactly two alleles. One of these alleles was dominant and the other recessive. Had any of these characteristics been determined by more than one gene, he may not have been able to develop such amazing insight into inheritance. In many instances, the relationship between genes and inheritance is more complex than that which Mendel found. Nevertheless, geneticists have since found that Mendel's findings can be applied to many organisms. For example, there are clear patterns of Mendelian inheritance in humans. Albinism (**Figure** 7.6), is a genetic disorder that is inherited as a simple Mendelian trait.

Figure 7.6: Albinism is a recessively inherited disorder in which the body does not produce enough of the pigment melanin. The skin, hair, and eyes of a person with albinism appear white or pale. (11)

Dominant and Recessive Alleles

Mendel used letters to represent dominant and recessive factors. Likewise, geneticists now use letters to represent alleles. Capital letters refer to dominant alleles, and lowercase letters refer to recessive alleles. For example, the dominant allele for the trait of green pod color is indicated by G. The recessive trait of yellow pod color is indicated by g. A true-breeding plant for green pod color would have identical alleles GG in all its somatic cells. Likewise, a true-breeding plant for yellow pod color would have identical alleles gg in all of its somatic cells. During gamete formation, each gamete receives one copy of an allele. When fertilization occurs between these plants, the offspring receives two copies of the allele, one from each parent. In this case, all of the offspring would have two different alleles, Gg, one from each of its parents.

An organism that has an identical pair of alleles for a trait is called **homozygous**. The true-breeding parents GG and gg are homozygous for the pod color gene. Organisms that have two different alleles for a gene are called **heterozygous**. The offspring of the cross between the GG and gg plants are all heterozygous for the pod color gene. Due to dominance and recessiveness of alleles, an organism's traits do not always reveal its genetics. Therefore, geneticists distinguish between an organism's genetic makeup, called its **genotype**, and its physical traits, called its **phenotype**. For example, the GG parent and the Gg offspring have the same phenotype (green pods) but different genotypes.

Lesson Summary

- Genetics is the branch of biology that focuses on heredity in organisms.
- Modern genetics is based on Mendel's explanation of how traits are passed from generation to generation.
- Mendel's use of mathematics in his pea plant studies was important to the confidence he had in his results.
- Mendel carried out his first experiments with true-breeding plants and continued them over a span of three generations.
- For each of the seven characteristics Mendel studied, he observed a similar ratio in the inheritance of dominant to recessive traits (3:1) in the F_2 generation.
- Mendel developed a theory that explained simple patterns of inheritance in which two alleles are inherited to result in one of several traits in offspring.
- The law of segregation states that a pair of alleles is segregated during the formation of gametes and that each gamete has an equal chance of getting either one of the allele.
- The law of independent assortment states that the inheritance of one trait will not affect the inheritance of another. That is, genes are inherited independently of each other.
- Linked genes are genes that are close together on the same chromosome. Linked genes are inherited together.
- Mendelian inheritance patterns can be seen in humans. Albinism is a genetic disorder that is inherited as a simple Mendelian trait.
- Genotype determines phenotype. A homozygous dominant or a heterozygous genotype will always show a dominant phenotype. A homozygous recessive genotype can only show a recessive phenotype.

Review Questions

1. Why was Mendel's understanding of mathematics and science important for his research?
2. What did Gregor Mendel contribute to the science of genetics?
3. What is a true-breeding plant?
4. How was Mendel able to control the pollination of his pea plants?
5. How does cross-pollination differ from self-pollination?
6. How did the appearance of Mendel's F_1 generation differ from the appearance of the P generation?
7. Identify the relationship between genes and alleles.
8. Summarize the law of segregation.
9. Summarize the law of independent assortment.
10. Relate the term homozygous to heterozygous by using an example from Mendel's experiments.
11. Relate the term genotype to phenotype by using an example from Mendel's experi-

ments.

12. Why can't you always identify the genotype of an organism from its phenotype?

Further Reading / Supplemental Links

- http://www.mendelweb.org/MWtime.html
- http://www1.umn.edu/ships/updates/mendel.htm
- http://www.macalester.edu/psychology/whathap/UBNRP/visionwebsite04/twotypes.html
- http://www.mendelweb.org
- http://www1.umn.edu/ships/updates/mendel2.htm
- http://anthro.palomar.edu/mendel/mendel_1.htm
- http://www.mendel-museum.org/eng/1online/experiment.html
- http://evolution.berkeley.edu/evosite/history/discretegenes.shtml
- http://en.wikipedia.org

Vocabulary

allele Different versions of a gene.

anther The male reproductive part of a flower.

artificial fertilization The process of transferring pollen from the male part of the flower to the female part of another flower; done in order to have seeds that will grow into plants that have a desired trait.

blending inheritance hypothesis Hypothesis that stated that offspring were a "mix" of their parents.

characteristic A heritable feature, such as flower color.

cross-pollination Fertilization in which pollen from one flower pollinates a flower on a different plant.

dihybrid cross A cross in which the inheritance of two characteristics are tracked at the same time.

dominant The allele that is expressed when two separate alleles are inherited.

F_1 generation The hybrid offspring of the P (parental) generation; first filial generation.

genetics The branch of biology that focuses on heredity in organisms.

genotype An organism's genetic makeup.

heredity The passing of characteristics from parent to offspring.

heterozygous Organisms that have two different alleles for a gene.

homozygous An organism that has an identical pair of alleles for a trait.

hybridization A cross between two individuals that have different traits.

Law of Independent Assortment States that the inheritance of one trait will not affect the inheritance of another.

Law of Segregation States that a pair of alleles is separated, or segregated, during the formation of gametes.

linked genes Genes that are close together on a chromosome, and are packaged into the gametes together.

monohybrid cross A hybridization in which only one characteristic is examined.

phenotype An organism's physical traits.

recessive The allele that is expressed only in the absence of a dominant allele.

self-pollinating Fertilization in which the pollen from a flower on a single plant transfers to the stigma of the same flower or another flower on the same plant.

stigma The female reproductive part of a flower.

trait A heritable variant of a characteristic, such as purple or white flower color.

true-breeding A plant that will always produce offspring with the parental trait when it self-pollinates.

Points to Consider

Next we will examine Mendelian Inheritance in further detail.

- Do you think all inheritance is as straightforward as the inheritance in pea plants?
- Is there a relationship between inheritance and probability? What might that relationship be?

7.2 Lesson 7.2: Mendelian Inheritance

Lesson Objectives

- Identify how probability is used to predict outcomes of genetic crosses.
- Outline how a Punnett Square helps predict outcomes of genetic crosses.
- Identify how probability can help determine the alleles in a gamete.
- Identify how a testcross is used to determine the genotype of an organism.
- Describe how monohybrid and dihybrid crosses differ.
- Identify the ratio of phenotypes that appeared in Mendel's dihybrid crosses.
- Examine how a pedigree is used in the study of human inheritance.
- Describe how codominance does not follow Mendelian Inheritance.
- Describe how incomplete dominance does not follow Mendelian Inheritance.
- Identify examples of polygenic traits in humans.
- Outline how heredity and environment can interact to affect phenotype.

Introduction

A **Mendelian trait** is a trait that is controlled by a single gene that has two alleles. One of these alleles is dominant and the other is recessive. Several inheritable conditions in humans are passed to offspring in a simple Mendelian fashion. Medical professionals use Mendel's laws to predict and understand the inheritance of certain traits in their patients. Also, farmers, animal breeders, and horticulturists who breed organisms can predict outcomes of crosses by understanding Mendelian inheritance.

Calculating Probability

The rules of probability that apply to tossing a coin or throwing a dice also apply to the laws of segregation and independent assortment. **Probability** is the likelihood that a certain event will occur. It is expressed by comparing the number of events that occur to the total number of possible events. The equation is written as:

Probability = (number of times an event is expected to occur/total number of times an event could happen)

For example, in Mendel's F_2 hybrid generation, the dominant trait of purple flower color appeared 705 times, and the recessive trait appeared 224 times. The dominant allele appeared 705 times out of a possible 929 times (705+224=929).

Probability = (705/929)

(705/929)= 0.76

Probability is normally expressed in a range between 0 and 1, but it can also be expressed as a percentage, fraction, or ratio. Expressed as a percentage, the probability that a plant of the F_2 generation will have purple flowers is 76%. Expressed as a fraction it is about ¾,and as a ratio it is roughly 3:1. The probability of the expression of the dominant allele for other characteristics can also be calculated the same way. In fact, Mendel found that all the other dominant "factors" had approximately a ¾ probability of being expressed in the F_2 hybrid generation. Review Table 7.1 for the results for the other six characteristics.

The probability the recessive trait will appear in the F_2 hybrid generation is calculated in the same way.

Probability = (224/929)

(224/929) = 0.24

The probability of the recessive trait appearing in the F_2 generation is 24% or about ¼.

Results predicted by probability are most accurate when many trials are done. The best way to illustrate this idea is to toss a coin. Because a coin has two sides, every time you toss it the chance of tossing heads or tossing tails is 50%. The outcome of each separate toss is unaffected by any previous or future result. For example, imagine you tossed seven heads in a row. You would think that the next toss is more likely to be a tail, but the possibility of tossing another head is still 50%. If you tossed the coin a total of ten times, a total of seven heads and three tails, you would calculate the probability of tossing heads is 70%. The fact that you carried out only a small number of trials has affected your results. If Mendel had grown only 10 plants, he would have gotten different probabilities for the appearance of dominant and recessive traits. However, Mendel carried out many thousands of trials. He was therefore sure that his results were due to probability, and not to chance.

Probability and the Law of Segregation

Each coin toss is a separate event. Likewise, gamete formation is a separate event. The probability that a Pp heterozygote produces gametes with a P allele or a p allele is 50% for each gamete cell. In a fertilization involving two such plants (as in the F_1 generation self-pollination experiment), each pollen cell and each egg cell have a 50% chance of having the P or p allele.

Predicting Genotypes with Punnett Squares

Mendel developed the law of segregation by following only a single characteristic, such as pod color, in his pea plants. Biologists use a diagram called a **Punnett Square**, to help predict the probable inheritance of alleles in different crosses. In a monohybrid cross, such as the one in **Figure** 7.7, 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 Pp (purple). Both plants produce gametes that contain both the P and p alleles. The probability of any single offspring showing the dominant trait is 3:1, or 75%.

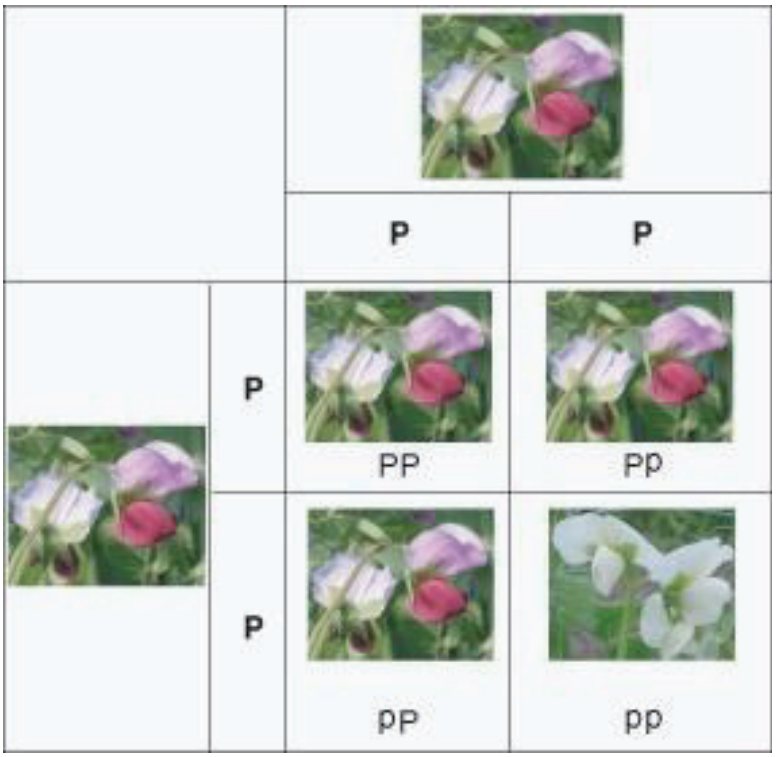

Figure 7.7: A Punnett square helps determine the genotype of this heterozygous cross. Two pea plants, both heterozygous for flower color, are crossed. The offspring will show the dominant purple coloration in a 3:1 ratio. Or, about 75% of the offspring will be purple. (1)

Using Probability to Determine Alleles in Gametes

In the monohybrid cross shown in **Figure** 7.7, two heterozygous plants are crossed. Both plants produce gametes, all of which contain either a P or p allele for flower color. The likelihood that any particular gamete contains the allele for a white flower can be calculated. Because a gamete can only carry one out of two alleles, there are only two possible outcomes

for a gamete. The probability that a gamete will carry the allele for white flower color is ½, 0.5, or 50%. The probability that a gamete will carry the allele for purple flower color is also ½.

Using Probability in a Heterozygous Cross

We can calculate the probability of any one of the offspring being heterozygous (Pp) or homozygous (PP or pp) for flower color. The probability of a plant inheriting the P or p allele from a heterozygous parent is ½. Multiply the probabilities of inheriting both alleles to find the probability that any one plant will be a pp homozygote.

½ × ½ = ¼ or 0.25

Only 25 %, or one outcome out of four, will result in a plant homozygous for white flower color (pp). The possibility that any one plant will be a PP homozygote is also 1/4. The heterozygous allele combination can happen twice (Pp or pP), so the two probabilities are added together ¼ + ¼ = 2/4, or ½. The probability that an offspring plant will be Pp heterozygous is ½.

Testcross and Punnett Squares

Suppose you have a purple and white flower and, as discussed above, purple color is dominant to white. The white flower must be homozygous for the recessive allele, but the genotype of the purple flower is unknown. It could be either PP or Pp. A testcross will determine the organism's genotype. In a **testcross**, the individual with the unknown genotype is crossed with a homozygous recessive individual (**Figure** 7.8). The unknown genotype can be determined by observing the phenotypes of the resulting offspring.

Dihybrid Crosses and Punnett Squares

Dihybrid crosses are more complicated than monohybrid crosses because more combinations of alleles are possible. For example, tracking the inheritance of seed color and pod color in a Punnett square requires that we track four alleles. G is the dominant allele for green pod color and g is the recessive allele for yellow pods. Y is the dominant allele for yellow seed color and y is the recessive allele for green seed color.

Two plants are crossed, one is true-breeding for green pods and yellow seeds (GGYY), the other is true-breeding for yellow pods and green seeds (ggyy). All of the F_1 generation will be heterozygous for both traits (GgYy). **Figure** 7.9, shows the dihybrid cross of the dihybrid P generation and the F_1 generation.

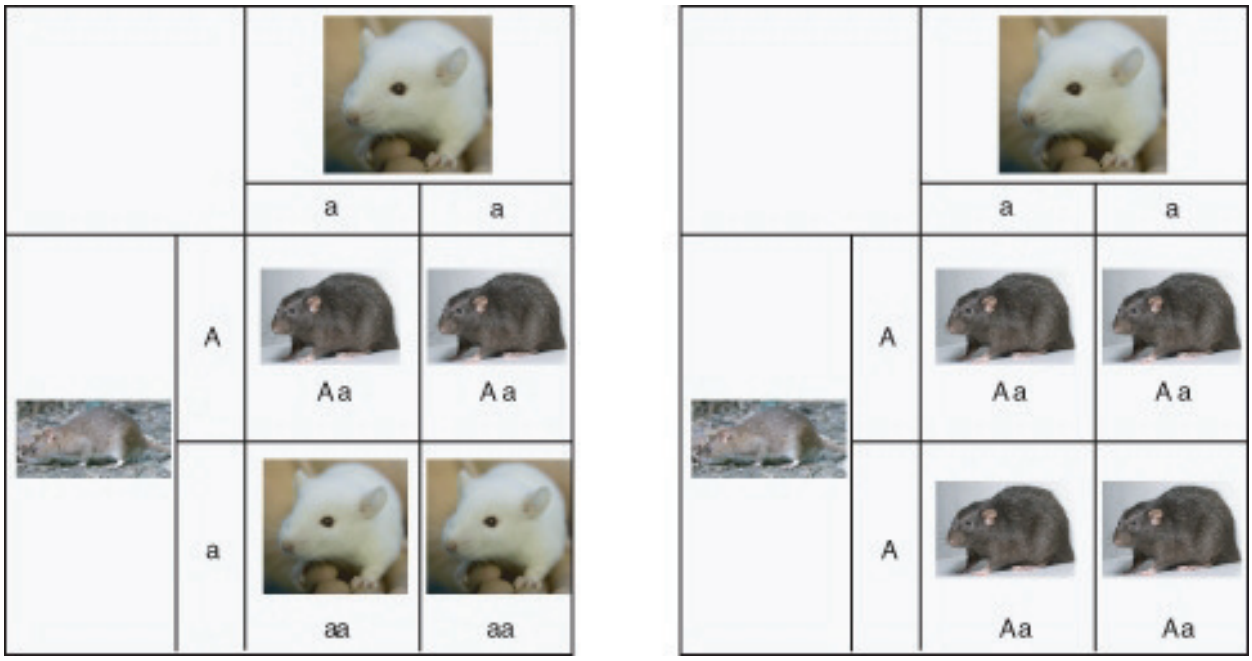

Figure 7.8: A testcross helps reveal the genotype of an organism when that organism shows the dominant trait, such as agouti coat color in rats. Such an organism could be homozygous dominant or heterozygous. Agouti is the common color of the Norway rat, *Rattus norvegicus*. (12)

Heterozygous Dihybrid Cross

In a dihybrid cross, four alleles can be inherited from any one parent at one time. When two heterozygous individuals are crossed, there are a total of 16 possible combinations of the four alleles. The phenotypes of the offspring with two independent traits show a 9:3:3:1 ratio. In a cross involving pea plants heterozygous for round, yellow seeds (GgYy), 9/16 plants have round, yellow seeds, 3/16 have round, green seeds, 3/16 have wrinkled, yellow seeds, and 1/16 has wrinkled, green seeds.

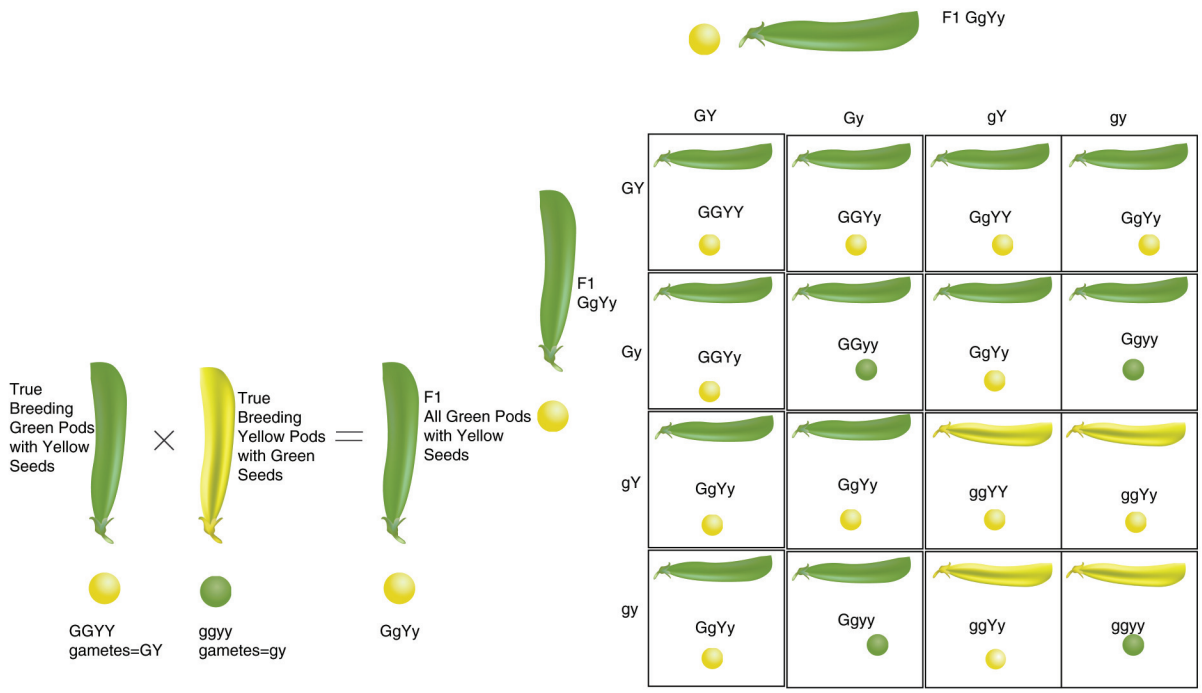

Figure 7.9: The dihybrid crosses were started by crossing two true-breeding plants, just as the monohybrid crosses were. The ratio of phenotypes (9:3:3:1) can be determined from the dihybrid Punnett square on the right. The genotype of the F_2 generation can also be determined. (9)

Mendelian Inheritance in Humans

A **pedigree** is a chart which shows the inheritance of a trait over several generations. A pedigree is commonly created for families, and it outlines the inheritance patterns of genetic disorders. **Figure** 7.10 shows a pedigree depicting recessive inheritance of a disorder through three generations. Scientists can tell the genetics of inheritance from studying a pedigree, such as whether the trait is sex-linked (on the X or Y chromosome) or autosomal (on a chromosome that does not determine sex), whether the trait is inherited in a dominant

or recessive fashion, and possibly whether individuals with the trait are heterozygous or homozygous.

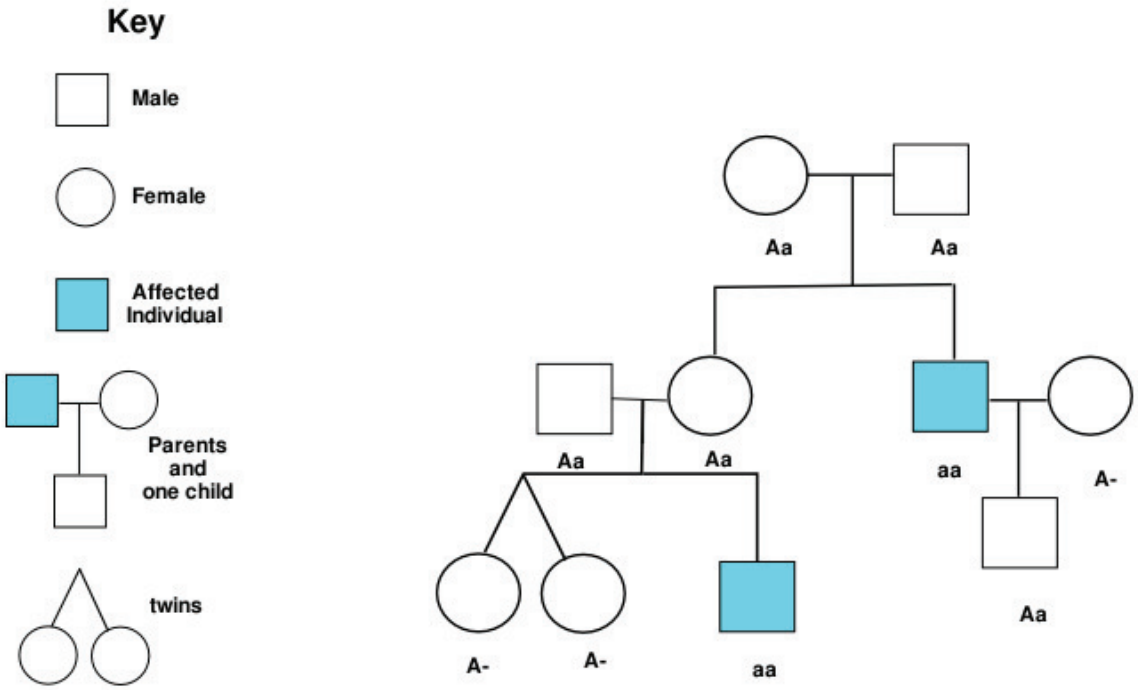

Figure 7.10: In a pedigree, squares symbolize males, and circles represent females. A horizontal line joining a male and female indicates that the couple had offspring. Vertical lines indicate offspring which are listed left to right, in order of birth. Shading of the circle or square indicates an individual who has the trait being traced. The inheritance of the recessive trait is being traced. A is the dominant allele and a is recessive. (4)

Is the trait sex-linked or autosomal? A **sex chromosome** is a chromosome that determines the sex of an organism. Humans have two sex chromosomes, X and Y. Females have two X chromosomes (XX), and males have one X and one Y (XY). An **autosome** is any chromosome other than a sex chromosome. If a trait is autosomal it will affect males and females equally.

A **sex-linked trait** is a trait whose allele is found on a sex chromosome. The human X chromosome is significantly larger than the Y chromosome; there are many more genes located on the X chromosome than there are on the Y chromosome. As a result there are many more X-linked traits than there are Y-linked traits. Most sex-linked traits are recessive. Because males carry only one X chromosome, if they inherit a recessive sex-linked gene they will show a sex-linked condition.

Because of the recessive nature of most sex-linked traits, a female who shows a sex-linked condition would have to have two copies of the sex-linked allele, one on each of her X

chromosomes. **Figure** 7.11 shows how red-green colorblindness, a sex-linked disorder, is passed from parent to offspring.

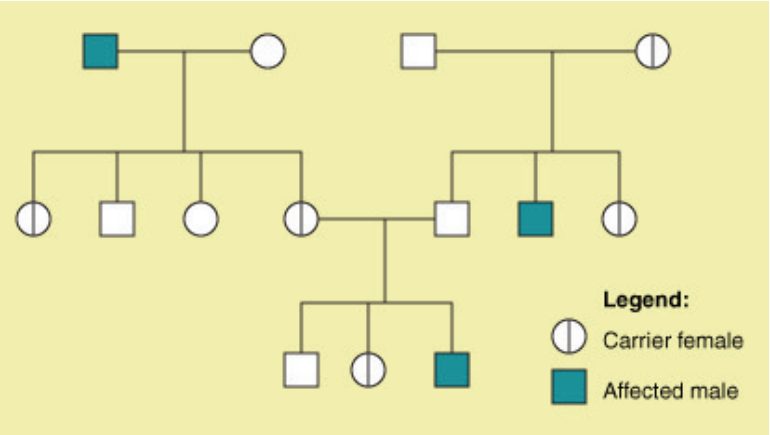

Figure 7.11: An X-linked disorder such as red-green colorblindness is normally passed onto the son of a carrier mother. Usually, females are unaffected as they have a second, normal copy of the allele on the second X chromosome. However, if a female inherits two defective copies of the allele, she will be colorblind. Therefore, every son of a colorblind woman will be colorblind. (13)

Is the Trait Dominant or Recessive? If the trait is autosomal dominant, every person with the trait will have a parent with the trait. If the trait is recessive, a person with the trait may have one, both or neither parent with the trait. An example of an autosomal dominant disorder in humans is Huntington's disease (HD). Huntington's disease is a degenerative disease of the nervous system. It has no obvious effect on phenotype until the person is aged 35 to 45 years old. The disease is non-curable and, eventually, fatal. Every child born to a person who develops HD has a 50% chance of inheriting the defective allele from the parent.

Are the Individuals with the Trait Heterozygous or Homozygous? If a person is homozygous or heterozygous for the dominant allele of a trait, they will have that trait. If the person is heterozygous for a recessive allele of the trait, they will not show the trait. A person who is heterozygous for a recessive allele of a trait is called a **carrier**. Only people who are homozygous for a recessive allele of a trait will have the trait.

Non-Mendelian Modes of Inheritance

The relationship between genotype and phenotype is rarely as simple as the examples Mendel studied. Each characteristic he studied had two alleles, one of which was completely dominant and the other completely recessive. Geneticists now know that alleles can be codominant, or incompletely dominant.

Codominance

Codominance occurs when both traits appear in a heterozygous offspring. Neither allele is completely dominant nor completely recessive. For example, roan shorthorn cattle have codominant genes for hair color. The coat has both red and white hairs. The letter R indicates red hair color, and R' white hair color. In cases of codominance, the genotype of the organism can be determined from its phenotype. The heifer in **Figure** 7.12 is RR' heterozygous for coat color.

Figure 7.12: The roan coat of this shorthorn heifer is made up of red and white hairs. Both the red and white hair alleles are codominant. Therefore cattle with a roan coat are heterozygous for coat color (RR'). (7)

Incomplete Dominance

Incomplete dominance occurs when the phenotype of the offspring is somewhere in between the phenotypes of both parents; a completely dominant allele does not occur. For example, when red snapdragons (C^RC^R)</math> are crossed with white snapdragons (C^WC^W), the F_1 hybrids are all pink hetrozygotes for flower color (C^RC^W). The pink color is an intermediate between the two parent colors. When two F_1 (C^RC^W) hybrids are crossed they will produce red, pink, and white flowers. The genotype of an organism with incomplete dominance can be determined from its phenotype (**Figure** 7.13).

		White Flower	
		C^W	C^W
Red Flower	C^R	C^RC^W	C^RC^W
	C^R	C^RC^W	C^RC^W

Figure 7.13: Snapdragons show incomplete dominance in the traits for flower color. The offspring of homozygous red-flowered and homozygous white-flowered parents are heterozygous pink-flowered. (5)

Complex Forms of Heredity

Traits that are affected by more than one gene are called **polygenic traits**. The genes that affect a polygenic trait may be closely linked on a chromosome, unlinked on a chromosome, or on different chromosomes. Polygenic traits are often difficult for geneticists to track because the polygenic trait may have many alleles. Also, independent assortment ensures the genes combine differently in gametes. Therefore, many different intermediate phenotypes exist in offspring. Eye color (**Figure** 7.14), and skin color are examples of polygenic traits in humans.

Figure 7.14: Eye color and skin color are examples of polygenic traits; they are influenced by more than one gene. (14)

When three or more alleles determine a trait, the trait is said to have **multiple alleles**. The human ABO blood group is controlled by a single gene with three alleles: i, I^A, I^B, and the recessive i allele. The gene encodes an enzyme that affects carbohydrates that are found on the surface of the red blood cell. A and B refer to two carbohydrates found on the surface of red blood cells. There is not an O carbohydrate. Type O red blood cells do not have either

type A or B carbohydrates on their surface.

The alleles I^A and I^B are dominant over i. A person who is homozygous recessive ii has type O blood. Homozygous dominant I^AI^A or heterozygous dominant I^Ai have type A blood, and homozygous dominant I^BI^B or heterozygous dominant I^Bi have type B blood. I^AI^B people have type AB blood, because the A and B alleles are codominant. Type A and type B parents can have a type AB child. Type A and a type B parent can also have a child with Type O blood, if they are both heterozygous (I^Bi,I^Ai). The table (7.3) shows how the different combinations of the blood group alleles can produce the four blood groups, A, AB, B, and O.

Table 7.3: **Bloodtype as Determined by Multiple Alleles**

	I^A	**I^B**	**i**
I^A	I^AI^A TYPE A	I^AI^B TYPE AB	I^Ai TYPE A
I^B	I^A I^B TYPE AB	I^B I^B TYPE B	I^B i TYPE B
i	i I^A TYPE A	i I^B TYPE B	ii TYPE O

Effects of Environment on Phenotype

Genes play an important part in influencing phenotype, but genes are not the only influence. Environmental conditions, such as temperature and availability of nutrients can affect phenotypes. For example, temperature affects coat color in Siamese cats.

The pointed pattern is a form of partial albinism, which results from a mutation in an enzyme that is involved in melanin production. The mutated enzyme is heat-sensitive; it fails to work at normal body temperatures. However, it is active in cooler areas of the skin. This results in dark coloration in the coolest parts of the cat's body, such as the lower limbs and the face, as shown in **Figure** 7.15. The cat's face is cooled by the passage of air through the nose. Generally adult Siamese cats living in warm climates have lighter coats than those in cooler climates.

Height in humans is influenced by many genes, but is also influenced by nutrition. A person who eats a diet poor in nutrients will not grow as tall as they would have had they eaten a more nutritious diet. Scientists often study the effects of environment on phenotype by studying identical twins. Identical twins have the same genes, so phenotypic differences between twins often have an environmental cause.

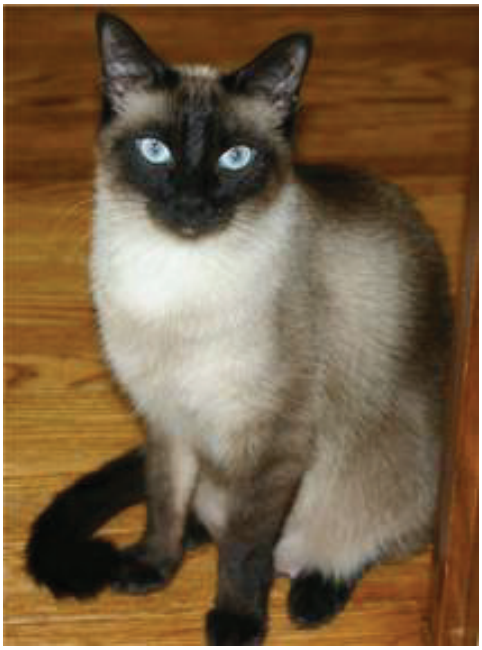

Figure 7.15: The dark "points," on this Siamese cat are caused by a gene that codes for a temperature-sensitive enzyme. The enzyme, which causes a darkening of the cat's fur, is active only in the cooler parts of the body such as the tail, feet, ears, and area around the nose. (10)

Lesson Summary

- Probability is the likelihood that a certain event will occur. It is expressed by comparing the number of events that actually occur to the total number of possible events. Probability can be expressed as a fraction, decimal, or ratio.
- A Punnett square shows all the possible genotypes that can result from a given cross.
- A testcross examines the genotype of an organism that shows the dominant phenotype for a given trait. The heterozygous organism is crossed with an organism that is homozygous recessive for the same trait.
- A dihybrid cross-examines the inheritance of two traits at the same time.
- A pedigree can help geneticists discover if a trait is sex-linked, if it is dominant or recessive, and if the person (or people) who have the trait are homozygous or heterozygous for that trait.
- The Mendelian pattern of inheritance and expression does not apply to all traits. Codominant traits, incompletely dominant traits, and polygenic traits do not follow simple Mendelian patterns of inheritance. Their inheritance patterns are more complex.
- An organism's phenotype can be influenced by environmental conditions.

Review Questions

1. What does the probability equation help to determine?
2. How can probability be expressed?
3. Outline how Punnett squares are useful.
4. Identify the purpose of a testcross.
5. How do the Punnett squares for a monohybrid cross and a dihybrid cross differ?
6. Mendel carried out a dihybrid cross to examine the inheritance of the characteristics for seed color and seed shape. The dominant allele for yellow seed color is Y, and the recessive allele for green color is y. The dominant allele for round seeds is R, and the recessive allele for a wrinkled shape is r. The two plants that were crossed were F1 dihybrids RrYy. Identify the ratios of traits that Mendel observed in the F2 generation, and explain in terms of genotype what each number means. Create a Punnett square to help you answer the question.
7. Draw a pedigree that illustrates the passing of the dominant cleft chin trait through three generations. A person who has two recessive alleles does not have a cleft chin. Let us say that C is the dominant allele, c is the recessive allele.
8. A classmate tells you that a person can have type AO blood. Do you agree? Explain.
9. Mendelian inheritance does not apply to the inheritance of alleles that result in incomplete dominance and codominance. Explain why this is so.
10. Outline the relationship between environment and phenotype.

Further Reading / Supplemental Links

- http://www.nbii.gov/portal/server.pt?open=512&objID=405&PageID=581&mode=2&in_hi_userid=2&cached=true
- http://omia.angis.org.au/retrieve.shtml?pid=417
- http://www.curiosityrats.com/genetics.html
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- http://www.hhmi.org/biointeractive/vlabs/cardiology/content/dtg/pedigree/pedigree.html
- http://www.ndsu.nodak.edu/instruct/mcclean/plsc431/mendel/mendel9.htm
- http://www.emc.maricopa.edu/faculty/farabee/BIOBK/BioBookgeninteract.html

Vocabulary

autosome Any chromosome other than a sex chromosome.

carrier A person who is heterozygous for a recessive allele of a trait.

codominance Occurs when both traits appear in a heterozygous individual.

incomplete dominance Occurs when the phenotype of the offspring is somewhere in between the phenotypes of both parents; a completely dominant allele does not occur.

Mendelian trait A trait that is controlled by a single gene that has two alleles.

multiple alleles When three or more alleles determine a trait, such as with the human ABO blood group.

probability The likelihood that a certain event will occur.

pedigree A chart which shows the inheritance of a trait over several generations.

polygenic traits Traits that are affected by more than one gene.

Punnett square A diagram that helps predict the probable inheritance of alleles in different crosses.

sex chromosome A chromosome that determines the sex of an organism.

sex-linked trait A trait whose allele is found on a sex chromosome.

testcross A cross used to determine an unknown genotype.

Points to Consider

The next chapter is *Molecular Genetics.*

- What do you think Molecular Genetics refers to?
- How can DNA contain all the genetic information?
- If DNA is in the nucleus, and proteins are made on ribosomes in the cytoplasm, how do you think this happens?

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