Unit 7: Genetics Unit

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South Dakota State Science Standards

9-12.N.1.1. Students are able to evaluate a scientific discovery to determine and describe how societal, cultural, and personal beliefs influence scientific investigations and interpretations.

9-12.L.2.1. Students are able to predict inheritance patterns using a single allele.

9-12.S.1.1. Students are able to explain ethical roles and responsibilities of scientists and scientific research.

Prefix/Suffix	Definition
Homo	Same, Similar
Hetero	Different
Со	With, Together
Mono	One
Di	Two

Key Vocabulary Terms

- 1. Allele
- 2. Autosomal Trait
- 3. Classical Inheritance
- 4. Codominance
- 5. Gene
- 6. Heterozygous
- 7. Homozygous
- 8. Incomplete Dominance
- 9. Sex-linked Trait

I. Gregor Mendel: Father of Genetics

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**. While humans have long been interested in understanding heredity, 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. 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. While Mendel did his work in the 1860s, his results were not widely shared with other scientists so he never gain recognition for his discoveries during his lifetime. Nearly 50 years later scientists rediscovered his work, leading to the era of modern **genetics**, the branch of biology that focuses on heredity in organisms.

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

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 while another variety has white flowers.

Mendel chose to study seven traits of pea plants. Each characteristic Mendel chose to study occurred in two contrasting traits. A **trait** is a heritable variant of a genetic character.

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.

Calculating Probability

Mendel was one of the first scientist to apply mathematics to the study of biology. He understood the rules of probability that apply to tossing a coin or throwing a dice also apply to the laws of genetics. **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 one of Mendel's experimental sets, 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 F2 generation will have purple flowers is 76% (move decimal two spaces to the right). Expressed as a ratio it is roughly 3:1. To calculate a ratio, divide by the smallest number to ensure one side is a whole number.

Calculating Ratio: (705/224: 224/224 = 3.15:1)

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 *nearly 30,000 trials!* He was therefore sure that his results were due to probability, and not to chance.

Comprehension Questions:

When Mendel began his work, what was the prevailing view of how traits were inherited?

What is the difference between self-pollination and cross-pollination?

What are two features of pea plants that make it an ideal research specimen for Mendel's work?

A sample of peas contains 800 green peas and 345 yellow peas,

- a. What proportion are green?
- b. What is the ratio of green to yellow peas?

II. Mendel's First Experiment (Monohybrid Cross)

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 crosspollinated two true-breeding plants of contrasting traits, such as purple and white flowered plants. The truebreeding parent plants are referred to as the **P generation** (parental generation). The hybrid offspring of the F1 generation (filial generation). The hybrid offspring of the F1 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 F1 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 whiteflower "heritable factor" had disappeared, all future offspring of the hybrids would be purple-flowered. To test this idea, Mendel let the F1 generation plants self-pollinate and then planted the resulting seeds.

Mendel's Results

The F2 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. Mendel carried out identical studies over three generations, (P, F1, and F2), for the other six characteristics and found in each case that one trait "disappeared" in the F1 generation, only to reappear in the F2 generation. Mendel studied a large number of plants, as shown in Table 1, so he was confident that the ratios of different traits in the F2 generation were representative.

Table 1: Results of F1 Generation Crosses for Seven Characteristics in P. sativum Characteristic Dominant Trait Recessive Trait F2 Generation Dominant:Recessive Ratio				
Trait	Actual Numbers	Ratio		
Flower Color (Purple vs White)	705: 224	3.15:1		
Flower Position (Axial vs Terminal)	651:207	3.14:1		
Stem Length (Tall vs Short)	787: 277	2.84: 1		
Pod Shape (Inflated vs Constricted)	882: 299	2.95: 1		
Pod Color (Green vs Yellow)	428:152	2.82: 1		
Seed Shape (Round vs Wrinkled)	5474:1850	2.96: 1		
Seed Color (Yellow vs Green)	6022:2001	3.01: 1		

Letters are now used to represent alleles. Capital letters are used to represent dominant alleles (those traits that don't disappear in the F1, such as purple flower color). The recessive alleles are represented by a lower-case letter. Mendel began his experiment with purple-flowering plants (FF) and white-flowering plants (ff). These plants each had two identical alleles, either FF or ff. Organisms with two identical alleles for a trait are termed **homozygous dominant** (FF) or **homozygous recessive** (ff). Organisms with two different alleles for a trait are called **heterozygous** (Ff).

Due to the dominant allele, a homozygous dominant plant (FF) will look identical to a heterozygous plant (Ff). The physical expression of a gene, such as flower color or plant height, is known as its **phenotype**. The genetic makeup of the organism, such as FF or Ff, is known as its **genotype**.

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. 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. This came to be known as the **law of segregation**, which 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 chance of receiving either of the two alleles. Although the actions of chromosomes wouldn't be understood for decades, Mendel was describing the results of meiosis (see Figure 1).



Figure 1: Alleles on homologous chromosomes are randomly separated during gamete formation.

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 <u>Figure 2</u>, 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 2: 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.

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. The unknown genotype can be determined by observing the phenotypes of the resulting offspring.

Using Probability to Determine Alleles in Gametes

In the monohybrid cross shown in <u>Figure 2</u>, 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 purple flower color is $\frac{1}{2}$, 0.5, or 50%. The probability that a gamete will carry the allele for purple flower color is also $\frac{1}{2}$.

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 $\frac{1}{2}$. Multiply the probabilities of inheriting both alleles to find the probability that any one plant will be a pp homozygote.

 $\frac{1}{2} \times \frac{1}{2} = \frac{1}{4}$ 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 $\frac{1}{4} + \frac{1}{4} = \frac{2}{4}$, or $\frac{1}{2}$. The probability that an offspring plant will be Pp heterozygous is $\frac{1}{2}$.

Comprehension Questions:

- 1. Which of the following represents a heterozygous individual? a. AA b. Aa c. aa d. AB e. Ab
- 2. Different forms of a trait are called a. variants b. alleles c. tetrads d. heterozygous e. Punnetts

3. In pea plants, tall height is dominant and short height is recessive. Which option below represents the

recessive genotype? a. tall b. short c. TT d. Tt e. tt

- 4. An individual is homozygous recessive for a trait. Write her genotype:
- 5. An individual is heterozygous for a trait. Write her genotype:
- 6. In pea plants, the allele for green pea pod color is dominant to the allele for yellow pea pod color. If a plant is heterozygous for this trait, what will be the plant's phenotype?

7. A purple-flowering plant (AA) is crossed with a white-flowering plant (aa). Complete the following Punnett square:



8.

If one parent's genotype is Bb and the other's is bb, what is the probability that the offspring will have black eyes if B = black eyes and b = red eyes?

- A.~0%
- B. 25%
- C. 50%
- D. 100%
- 8. What is a testcross? In peas, purple flower color is dominant. If you wanted to know if a purpleflowered plant was homozygous (AA) or heterozygous (Aa), describe a procedure you could follow to determine the plant's genotype.

III. Mendel's Second Experiment (Dihybrid Cross)

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.

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 truebreeding for yellow pods and green seeds (ggyy). All of the F1 generation will be heterozygous for both traits (GgYy). Figure 3 below, shows the dihybrid cross of the dihybrid P generation and the F1 generation.

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 3: 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 at right. The genotype of the F2 generation can also be determined.

Comprehension Questions:

1. A pea plant is tall and produces purple flowers. If the plant is heterozygous for each trait (AaBb), the plant will pass on one allele for each trait during gamete formation.

List the different gametes that could be formed?

2. Another pea plant is tall and produces white flowers. Its genotype is Aabb.

List the different gametes that could be formed by this plant?

3. Dihybrid crosses are solved using a 16-square Punnett matrix. Below a problem is set up for the cross between two pea plants: a dwarf, purple-flowered plant (aaBb) and a tall, purple-flowered plant (AaBb).

	AB	Ab	aB	ab
aB	AaBB	AaBb	aaBB	aaBb
ab	AaBb	Aabb	aaBb	aabb
aB	AaBB	AaBb	aaBB	aaBb
ab	AaBb	Aabb	aaBb	aabb

- *a. Out of the sixteen offspring, how many are predicted to be dominant for both traits (tall, purple)?*
- b. Out of the sixteen offspring, how many are predicted to be dwarf?
- *c.* What are the four possible phenotypes and the probability of each (# predicted divided by 16)?

4. Which genotypes complete the following Punnett square?

	RY	Ry	RY	Ry
RY	RRYY	RRYy	RRYY	RRYy
Ry	1	RRyy	RRYy	2
rY	RrYY	RrYy	RrYY	RrYy
ry	RrYy	Rryy	RrYy	RrYY

A. 1. RrYy

- 2. RRyy B. 1. RRYy 2. RRyy C. 1. RRYY 2. rrYY D. 1. RrYY
 - 2. Rryy
- 5. Using a dihybrid cross and the information below, what is the probability of an offspring having white flowers and a purple stem?

One parent's genotype is ggWw and the other's is Ggww. $G = green stem \qquad W = white flowers$ $g = purple stem \qquad w = pink flowers$ A. $\frac{1}{16}$ B. $\frac{1}{4}$ C. $\frac{1}{8}$ D. $\frac{3}{16}$

6. Using a dihybrid cross and the information below, what is the probability of an offspring being short and yellow?

One parent's genotype is TtGg and the other's is Ttgg. T = tall G = green t = short g = yellowA. 1/4 B. 1/16 C. 1/8 D. 6/16

IV. Mendelian Inheritance: Pedigrees

A **pedigree** is a chart which shows the inheritance of a trait over several generations. A pedigree is commonly created for families, and outlines the inheritance patterns of genetic disorders. Figure 4 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 4: 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.

Is the Trait Dominant or Recessive?

If the trait is autosomal dominant (inherited the same by males and females), 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.

VI. Mendelian Inheritance: Sex-linked Traits

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 5 shows how red-green colorblindness, a sex-linked disorder, is passed from parent to offspring.



Figure 5: 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.

Sex-linked Punnett squares are completed by using X^B , X^b or Y. Females inherit two X chromosomes, so they have two of any allele carried on the X chromosome ($X^B X^B, X^B X^b$, or $X^b X^b$). Males only inherit one X, thus carry only one allele if carried on the X chromosome ($X^B Y$ or $X^b Y$). When setting up a Punnett square, you can determine the male's genotype based on their phenotype (if they have a recessive trait, such as red-green colorblindness, they are $X^b Y$). Females can be carriers ($X^B X^b$) even if they don't display the recessive trait. See Figure 6 as an example.

	X ^B	X ^b
X ^B	X ^B X ^B	X ^B X ^b
Y	ХВҮ	X ^b Y

Figure 6. A Punnett Square for a sexlinked recessive trait. This cross is between a female carrier and a male not displaying the trait.

VI. Exceptions to Mendellian 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.

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 ($C^{R}C^{R}$) are crossed with white snapdragons ($C^{W}C^{W}$), the F1 hybrids are all pink hetrozygotes for flower color ($C^{R}C^{W}$). The pink color is an intermediate between the two parent colors. When two F1 ($C^{R}C^{W}$) 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, as is shown in Figure 7.

		White Flower	
		Cw	cw
Red Flower	C ^H	c ^a c ^w	c ^a c ^w
	C ^R	C [#] C [₩]	C [∺] C [₩]

Figure 7: 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.

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 and skin color are examples of polygenic traits in humans.

Blood Types: Multiple Alleles and Codominance

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^A have type A blood, and homozygous dominant I^BI^B or heterozygous dominant I^B have type A blood, and homozygous dominant I^BI^B or heterozygous dominant I^B 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). <u>Table 2</u> shows how the different combinations of the blood group alleles can produce the four blood groups, A, AB, B, and O.

Table 2: Bloodtype as Determined by Multiple Alleles				
	$\mathbf{I}^{\mathbf{A}}$	I^{B}	i	
I ^A	$I^A I^A$ "A"	I ^A I ^B "AB"	I ^A i "A"	
I^{B}	I ^B I ^A "AB"	I ^B I ^B "B"	I ^B i "B"	
i	iI ^A "A"	iI ^B "B"	ii "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. 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.

Comprehension Questions:

- 1. Which blood types are not possible for children of 2 parents with type AB blood?
 - A. Type A
 - B. Type O
 - C. Type B
 - D. Type AB
- 2. Why is color-blindness much less common in females?
 - A. because color blindness is a co-dominant trait in males
 - B. because color blindness is a recessive trait carried on the X chromosome
 - C. because color blindness is a dominant trait carried on the X chromosome
 - D. because the eye structure of males is more susceptible to color blindness

- *3.* What is the probability of a colorblind father (XcY) and normal visioned non-carrier mother (XX) producing a colorblind son?
 - A.~0%
 - B. 25%
 - C. 50%
 - D. 100%
 - Fill in the blank.
- 4. In blood types, both alleles are fully expressed and show no dominance over each other. This is an example of _____.
 - A. incomplete dominance
 - B. sex-linkage
 - C. a mutation
 - D. co-dominance
- 5. In human blood types, the A and B alleles are co-dominant. Predict the blood type if a person inherits the A allele from one parent and B allele from another.
 - A. A
 B. B
 C. O
 D. AB
- 4. The skin color of a human may have a genotype of WwDDRr. What kind of trait is this?
 - A. a mutated trait
 - B. a polygenic trait
 - C. a polyploid trait
 - D. a double allele trait

Sources:

Text: ck12 Biology FlexBook (August 2010)

- Figure 1: http://jan.ucc.nau.edu/~lrm22/lessons/ mitosis_notes/ mitosis_and_meiosis.html, License: Creative Commons
- Figure 2: <u>http://commons.wikimedia.org/wiki/Image:Doperwt_rijserwt_bloemen_Pisum_</u> sativum.jpg, *License*:GFDL;
- Figure 4: http://en.wikipedia.org/wiki/Image:PedigreechartB.png, *Diagram by*: Rozzychan, *License:* CC-BYSA

Figure 5: Created by: Niamh Gray Wilson, License: CC-BY-SA

Vocabulary

Allele: Different versions of a gene. Anther: The male reproductive part of a flower. Autosome: Any chromosome other than a sex chromosome. Blending inheritance hypothesis: Hypothesis that stated that offspring were a "mix" of their parents. Carrier: A person who is heterozygous for a recessive allele of a trait. Characteristic: A heritable feature, such as flower color. Classical Inheritance: Same as Mendelian inheritance/trait. **Codominance:** Occurs when both traits appear in a heterozygous individual. **Cross-pollination**: When pollen from one flower pollinates a flower on a different plant. Dihybrid cross: A cross in which the inheritance of two characteristics are tracked. **Dominant allele**: The allele that is expressed when two separate alleles are inherited. **F1 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. **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. Law of Independent Assortment: The inheritance of one trait will not affect the inheritance of another. Law of Segregation: A pair of alleles is separated, or segregated, during the formation of gametes Mendelian trait: A trait that is controlled by a single gene that has two alleles. Monohybrid Cross: Only one characteristic is examined in a genetic cross. Multiple alleles: When three or more alleles determine a trait; ex: human ABO blood group. Monohybrid cross: A hybridization in which only one characteristic is examined. **P** generation: Parental generation; first cross in an inheritance study. Pedigree: A chart which shows the inheritance of a trait over several generations. Phenotype: An organism's physical traits. **Polygenic traits**: Traits that are affected by more than one gene. **Probability**: The likelihood that a certain event will occur. **Punnett square**: A diagramthat helps predict the probable inheritance of alleles in different crosses. **Recessive:** The allele that is expressed only in the absence of a dominant allele. 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. 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 characteristic of an organism, such as flower color.

True-breeding: Homozygous dominant individuals; when cross involves classical inheritance, offspring will always be dominant.