Mendelian Inheritance
Extended Background

Individuals within populations of the same species vary greatly. In fact, within sexually reproducing populations, no two individuals have exactly the same genetic make-up. When sexually reproducing organisms mate, the offspring receives half of its genes from one parent, and half from the other parent, creating a new being with a unique combination of genes from both parents. This combination of genetic information is what leads to variation among individuals.

The question of how traits or characteristics are passed from one generation to the next has been pondered for hundreds of years. Although many questions on inheritance have been answered thus far, the connection between genes and traits, and the passage of these genes from parents to children still amazes scientists and lay people alike. And even if we don't understand the many intricate interactions genes have inside cells, several laws of inheritance have been observed and documented. These basic principles are understood to be some of the underlying rules that genes follow as they are passed from parent to offspring.

Amazingly enough, a few of these laws were documented in the 1800's before DNA was identified as the hereditary molecule of the cell. Gregor Mendel (1822-1884) made some of the very first groundbreaking scientific observations of inheritance. Sadly, when Mendel presented his findings to the rest of the world, he was met with rejection and disbelief. Finally, in the early 1900's, Mendel received proper recognition when scientists began making some of the same observations.

Gregor Mendel was a monk who lived at a monastery in an area that is now the Czech Republic. He was interested in mathematics and science, but had a passion for his garden. Through his observation of pea plants, Mendel discovered that certain traits, such as seed color and shape, flower color and plant height only came in two forms and were inherited in specific patterns. For example, seeds were either green or yellow, flowers were colored or white, and plants were either tall or short. Mendel set up an experiment to test what the result would be from a cross between two plants of opposing traits. He transferred pollen from the stamen of one plant to the stigma of another that had its own stamen removed, to prevent self-pollination. When Mendel crossed pure forms of each type of plant, much to his surprise, only one of the traits appeared in the offspring.

In one experiment, Mendel crossed a pure tall plant with a pure short plant, collected the seeds and planted them. After several experiments, the results of the cross were a surprise: all of the offspring were tall. It seemed as if the short trait had disappeared. The offspring of a cross between pure parents with different traits are called **hybrids**. Mendel continued his work by allowing the hybrid offspring to self-pollinate. He collected and planted the seeds, and observed that most of the offspring were tall, but some were short. The short trait reappeared in the second generation. Mendel concluded that the short factor (he used the word factor instead of gene) didn't disappear, but was somewhat hidden. He described that the tall trait was **dominant** over the short **recessive** trait, which is why it was not expressed in the first generation. The **law of dominance** resulted which states that if a dominant gene is present, the dominant trait will express itself. A recessive trait will only be expressed if there are no dominant factors present to mask over it.

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Next, Mendel attempted to explain why the recessive trait disappeared in the first generation, and reappeared in the second. He hypothesized that every trait in an organism is controlled by two factors, one from each parent. For every trait there is a pair of genes that control that trait. A parent can only pass one of the two factors to an offspring for each trait, and the factors from both parents recombine to create a new being. This is called the **law of segregation**. An offspring can inherit two dominant factors, one from each parent, and then express that trait. If an offspring inherits two different factors for a trait, the dominant trait will express itself. Although the recessive trait is not expressed, the factor is still present, and can be passed on to further generations. If both recessive factors are inherited, the recessive trait will express itself.

These laws are in fact true for humans as well as all other sexually reproducing organisms. In all human cells (except egg and sperm) there are 23 pairs of chromosomes. The first 22 pairs are called autosomes and the 23rd pair is the sex chromosomes. Within each pair, one set is inherited maternally and the other paternally. In autosomes, this means that each gene has a pair on the opposing chromosome. The sex chromosomes are a special case. For males, the last pair of chromosomes isn't a matching set. Males inherit an X chromosome maternally, but paternally inherit a Y chromosome. Females on the other hand, inherit an X chromosome from both parents. This means that at fertilization, the sperm cell's genetic information determines gender. A Punnett square, developed by Reginald Punnett (1875-1967), can be used to demonstrate the statistics involved in gender determination.

	X	X
X		
Y		

(mother)

(father)

	X	X
X	XX	XX
Y	XY	XY

Punnett squares are good tools to study how principles of probability can be applied to Mendelian inheritance. Use this tool like a multiplication table. Simply drag the gene or chromosome represented on the outside edge of the square into each of the center squares to see the possible number of outcomes in each cross. For gender, there are 2 possible outcomes: XX or XY. Statistically, there is a 50% chance of having a boy or a girl each time a woman conceives.

The same procedure can be used for individual genes, such as the genes for plant height or seed color in Mendel's peas, or for a simple human trait such as a widow's peak. As a rule, letters are used to represent each allele, or gene form in a pair. For plant height, tall is the dominant trait and short is the recessive. The possible genotypes (or inherited alleles) for this trait are: TT (pure bred tall), tt (pure bred short) and Tt (hybrid). A capital letter is always used to represent the dominant allele, and a lower case letter is always used to represent the recessive. Use a Punnett square to investigate the statistical inheritance of each trait.

For example, if you cross a pure tall plant(TT) with a pure short plant(tt), the resulting offspring will all be tall hybrids(Tt), inheriting the tall (T) allele from one parent and the short (t) allele from the other. A Punnett square can be used to show that every time two pure plants of opposing traits are crossed, there is a 100% chance the dominant trait will express itself. Two hybrids(Tt) can be crossed to study the probabilities of the next generation. Using a Punnet square, you will observe a 75% tall and 25% short ratio for the cross between two hybrid tall plants. 50% of the offspring will be hybrids