Lesson 3: Genetics

The Beginning of Genetics—Mendel

Gregor Mendel, the father of genetics, playing with his peas.

Gregor Mendel (1822-1884), was an Austrian (now part of Czechoslovakia) is best known for his experiments with peas. His experiments and careful record-keeping allowed him to be the first to develop the **laws of heredity**.

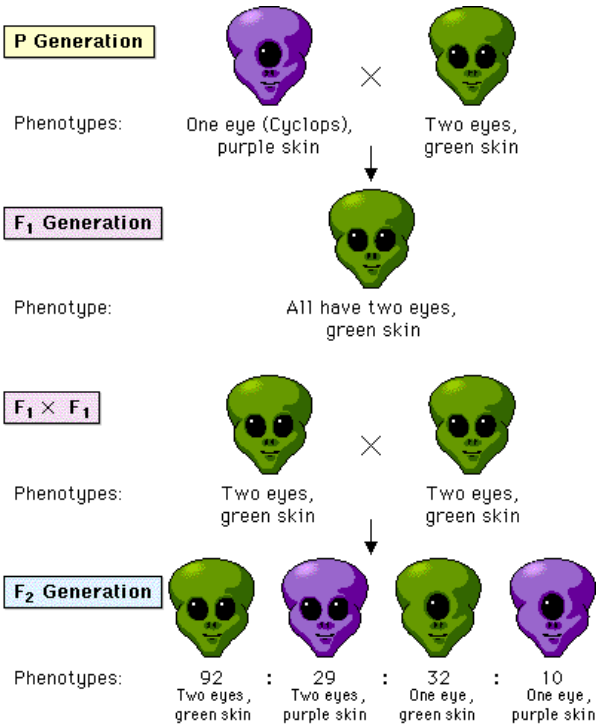
Mendel experimented with peas that did not carry all the same features. Some were tall, some were short, some had white flowers, some had purple flowers, some had wrinkled seeds while others had round ones… the peas carried many different features (which we will call **traits**[4] from now on).

Mendel followed the traits over generations. Peas were well suited to the experiment because peas have such short lives. Can you imagine trying to study generations of dogs? It would take years to see traits passed from one generation to the next! Mendel kept good records, and always noted when a trait was passed to the next generation, if it skipped a generation, or if it never appeared again. Another advantage was the way the pea flowers were pollinated; they could either self pollinate, or be cross-pollinated by Mendel. This way, Mendel meticulously constructed and observed the passage of traits over many generations of many varieties of plants.

P, F1, and F2

Mendel used a short-hand to discuss his results. Mendel called the parental generation of his peas the **P** generation. He called the offspring of the P generation the **F1** generation (short for *Filius*, meaning sons and daughters in latin). The offspring of the F1 generation he then called the **F2** generation.

[4] Trait is just another word for "feature". However it's the word used by biologists, so it's the one we'll use. The official definition of a trait is "a distinguishing characteristic or quality" (dictionary.com)



Mendelian nomenclature. Mendel used P to refer to the parent generation, F1 to refer to the offspring of P, and F2 to refer to the offspring of F1. In this case we're looking at alien inheritance of Mend-alien traits. An experiment Mendel never managed to get around to…

Mendel's strange results

At the time, people expected that traits were mixed and averaged each generation. If a tall woman and a short man had a child, that child was expected to be of average height. Likewise, when Mendel crossed a tall plant and a short plant the expected result in the F1 generations was a short plant. But it wasn't!

All of the F1 plants were tall—a completely unexpected result. But, even more unexpectedly, in the F2 generation 75% of the plants were tall, and 25% were short.

The law of segregation

This lead Mendel to develop **the law of segregation**—a law that still serves as a fundamental law of modern genetics. The law states that each organism gets two copies of the same gene[5], which separate (the "segregation" in the name of the law) when gametes[6] are produced.

Mendel used this law to resolve the curious results found in the short pea plant/tall pea plant experiment. He guessed that each plant has two copies of the same trait, but could only pass on one copy through reproduction. He then guessed that some traits were more dominant—more likely to show itself in the offspring—than others. For example, the *tall* trait was more dominant than the *short* trait. He called these traits that were more likely to be chosen (like the tall trait) **dominant**, and the

[5] At the time Mendel did not know that genes existed, so he simply referred to them as "hereditary factors".

[6] Remember: gametes are the reproductive cells produced via meiosis. Gametes only have one copy of DNA—half the amount found in the organism.

traits less likely to be chosen (like the short trait) **recessive**.

His experiments showed that if a plant with a dominant trait and a plant with a recessive trait reproduced the F1 generation would be 100% dominant trait. For example, all of the offspring of the tall and short plant were tall. But, the F2 would always be 75% dominant and 25% recessive—and F2 generation that was 75% tall and 25% short, in our example.

The dominant traits are designated by a capital letter, and the recessive traits are designated by a lowercase letter. For example, the dominant trait "tall" is designated the letter *T*, while short is given the lowercase letter *t*. Since the plants have two copies of each trait the combinations can be either TT, Tt, tT, or tt.

If both copies are dominant, than the dominant trait is seen (TT= tall plant). If there's a mix, than the dominant trait is seen (Tt/tT = tall plant). If both traits are recessive, than the recessive trait is seen (tt = short plant). Since three of the four options result in tall plants, and one of the four results in short plants, it makes sense that Mendel observed the results he did.

That's because after the first generation of TT X tt all of the plants were Tt and tT. But, when the tT/Tt generation was crossed, the plants could have all four combinations. Three out of the four combinations (TT, tT, and Tt) yield dominant traits, while the fourth combination (tt) yields short plants. Thus ¾ = 75%, and ¼ = 25%.

A good way of visualizing these results is with **Punnett Squares**. Punnett squares are simply tables we can use to show the possible combinations of traits. In our Tall/short example we can draw this Punnett Square:

Parental (P) Generation: TT crossed with tt

	The Recessive Plant		
The Dominant Plant		**t**	**T**
	T	Tt	Tt
	T	Tt	Tt

Result: 100% Tt. 100% tall.

F1 Generation: Tt crossed with Tt

	T	**T**
T	TT	Tt
t	tT	Tt

Result: 25% TT, 50% Tt, 25% tt. 75% tall, 25% short.

Modern Genetics: Additions, Further Explanations, and Exceptions to Mendel's Laws

It's really rare to get everything completely right the first time. It's only natural that Mendel's laws—while essential to the science of genetics—didn't fully explain the mechanisms of inheritance and overlooked some exceptions to the rules. After Mendel's contributions to genetics, there were still some questions to be answered.

First and foremost: What *are* these "inheritance factors"? What is the "tall" trait? What does it look like? What are these mysterious things? Could finding out cause a revolution in the field of biology[7]?

Second: What about the exceptions? Sometimes offspring are neither fully dominant, nor fully recessive. Sometimes a white flower crosses with a red flower and produce a *pink-flowered* F1 generation! How can we explain exceptions like this and others?

[7] Yes.

DNA—the "inheritance factors"

DNA (short for deoxyribonucleic acid), the double-helix shaped molecule found in all cells, answered the question of what these "inheritance factors" were. DNA is often thought of as the cell's "recipe book." DNA holds the instructions for building proteins the same way recipe books hold the instructions for making dishes. The individual codes for making proteins are called **genes**, and are like the recipes of the cookbook. These genes are the inheritance traits.

The genes are passed on from generation to generation and instruct the cell how to make proteins. Here's the tricky part: the proteins made by the cell—according to the recipe from the gene—is the trait exhibited by the organism.

For example: In Mendel's peas there were some peas that had a "wrinkled" appearance, and others that hand a smoother "round" appearance. As it turns out, "wrinkled" and "round" are just two versions of the same recipe. Some plants used the "wrinkled" recipe; others used the "round" recipe. Although we're calling it a recipe right now, we can just as easily call it by its scientific name—a gene.

These genes are what is inherited. These genes are the "inheritance factors" Mendel discussed.

DNA is simply two strands of genes. That is to say, the way traits are inherited as genes. To be exact, two copies of each gene in inherited. Which version of the gene is expressed (shows in the offspring) depends on dominance as well as other factors.

So, to answer the question "what are the 'inheritance factors' discussed by Mendel?": genes.

Different versions of the same gene: Alleles

In every cell there is DNA which is composed of two copies of every gene. That means, each gene has a corresponding copy. For example, if there's a tall pea plant that means there's a *T* gene as well as either another *T* or a *t*. These corresponding copies are called **alleles**. The alleles can either be the same or different[8].

Genotype and Phenotype

Genotype and **phenotype** are the words we use to describe what genes an organism has, and which traits are expressed, respectively.

They are extremely useful because organisms do not express all of their genes—mostly the dominant ones. We use the word "genotype" to describe the genetic composition of a cell. Is the cell a TT? Is it a Tt? Is it a tt? To talk about which genes an organism has we use the word "genotype".

Phenotype, on the other hand, is just used to describe the appearance of the organism. Is it round? Wrinkled? Tall? Short? The phenotype is the physical trait expressed.

Exceptions to Mendel's Laws

Incomplete dominance and codominance

Mendel's laws do not explain when traits are averaged. When both a dominant and a recessive trait are present in the DNA (a heterozygous genotype), but an average of the two traits is expressed. For example, when a red Snapdragon Flower is crossed with a white Snapdragon Flower, and the F1 offspring are… Pink! There is no explanation for that in Mendel's laws. According to his laws, the dominant must be expressed.

This exception is called **incomplete dominance**. The

[8] If they are the same they are called "homozygous" and if they are different they are called "heterozygous".

dominant trait is expressed, but not fully.

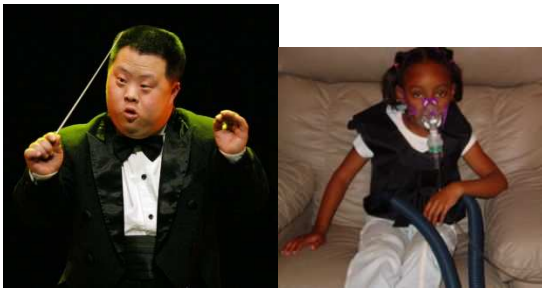
Pink Snapdragon Flower. Incomplete dominance in action!

Another exception is when there are two different types of dominant alleles which are both expressed. This is called **codominance**. For example the AB bloodtype carries traits of both the A and B blood type despite both of them being different dominant alleles.

Polygenic traits

Polygenic traits are traits controlled by multiple genes. For example, human height and skin color traits are controlled by multiple genes.

Human Genetics



Genetic disorders. *Left to right: a man with Down Syndrome, and a girl with cystic fibrosis.*

Peas are great, but what does genetics have to do with me? What does genetics have to say about human beings? A lot, actually.

Mendel's laws help us study the various **genetic disorders** (such as Down Syndrome, and Cystic Fibrosis) that affect humans. A genetic disorder is simply a disease or disorder that is inherited genetically. Genetic disorders can be caused by single defective gene, or by entire defective chromosomes. These disorders caused by defective chromosomes are called **chromosomal disorders**. Downs Syndrome is an example of a chromosomal disorder.

Using simple Punnett Squares, and Mendel's laws, we can figure out how genetic disorders are inherited.

Sex-linked Inheritance

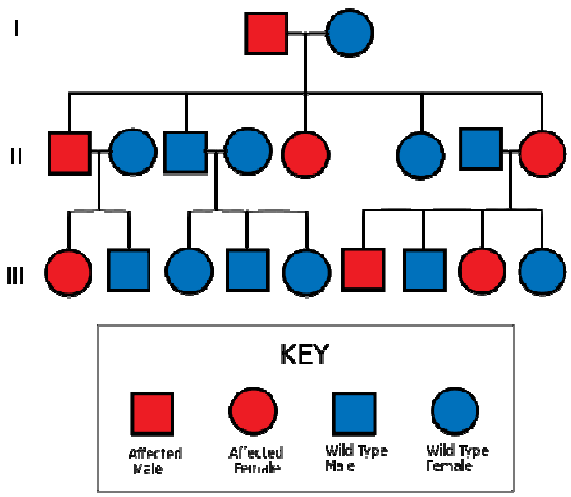
What determines if a baby is a boy or a girl? Recall that you have 23 pairs of chromosomes, one pair of which are the sex chromosomes. Everyone has two sex chromosomes, X or Y, that determine if we're male or female.

Females have two X chromosomes, while males have one Y chromosome and one X chromosome. So if a baby inherits an X from the father and an X from the mother, it will be a girl. If the baby inherits the Y chromosome, it will be a boy.

Notice that a mother can only pass on an X chromosome, so the sex of the baby is determined by the father. The father has a 50 percent chance of passing on the Y or X chromosome; hence it is a 50 percent chance whether a child will be a boy or a girl.

One special pattern of inheritance that doesn't fit Mendel's rules is **sex-linked inheritance,** referring to the inheritance of traits which are due to genes located on the sex chromosomes. The X chromosome and Y chromosome carry many genes and some of them code for traits that have nothing to do with determining sex. Since males and females do not have the same sex chromosomes, there will be differences between the sexes in how these sex-linked traits are expressed.

Pedigree Analysis



What is this chart? It's a **pedigree analysis**. These charts, usually used for families, allow us to visualize the inheritance of genotypes and phenotypes (traits). In this chart, the P, F1, and F2 generation are represented by the numerals I, II, and III respectively. Notice that those carrying the trait are colored red, and those not carrying the trait (the normal-looking ones) are in blue. The normal, non-trait carrying organisms on the chart are called the **wild-type**.

The term wild-type is used in genetics often to refer to organisms not carrying the trait

being studied. For example, if we were studying a gene that turns house-flies orange, we would call the normal-looking ones the wild-type.

Modern Genetics

Genes are the stuff of life. Naturally, current genetic research is extremely relevant and exciting. Advances in genetics include better treatments of diseases, more productive agriculture, and improved crime-fighting. However, as we learn to control the basic units of life we begin to walk into ethical grey-zones. Should humans be able to clone themselves? Should we be able to choose what traits we want our children to have? These are questions that still need to be answered. As we reap the benefits of our control, we must still ask ourselves—at each step—whether what we are doing is ethically correct.

How to cut and paste genes

We can manipulate DNA to get proteins we want, and even entire organisms we want.

This is done by creating **recombinant DNA**. Recombinant DNA is DNA from multiple sources. For example, we can take genes (that is, a piece of DNA) from one bacterium and put them into the DNA of another. Because we have to separate the genes we want from the first bacterium and then *re-combine* them into the second, we call the DNA we create recombinant.

To cut genes out of a strand of DNA, geneticists use an extremely useful tool called **restriction enzymes**. Restriction enzymes cut DNA at specific points (specific sequences of DNA). There are more than 3,000 known restriction enzymes. Because we know where theses restriction enzymes cut the DNA, we can use them to cut the specific section of DNA we want to re-combine. Restriction enzymes' ability to cut DNA at specific sequences makes them an important part of the process of creating recombinant DNA.

The Human Genome Project

A genome is all the genetic information of an organism. The human genome is all of the genetic information that humans have—all that makes us human.

The **Human Genome Project**, completed in 2003, was an international effort to identify and

locate[9] the over 20,000 human genes. Although the project was completed, the results (the identified and mapped genes) are still being analyzed. The results will make help us understand how we work, and who we are. That knowledge will help us fight diseases and genetic disorders. It can lead to advances in **gene therapy,** the manipulation of human genetic information to cure disorders. Additionally, since it contains the information of our ancestors, it will also help us understand where we come from. The Human Genome Project sequenced and mapped all human genetic information.

[9] On the chromosome.