Study Guide - Classical Genetics (by Mendel)

The science we call genetics was developed by a monk named Gregor Mendel in experiments he performed in his garden in Austria. Though the concepts he studied were not widely accepted at first, the principles and ”laws“ that he established are recognized today as classical genetics and are often called Mendelian genetics.

Genetics (genes, alleles, genotype, phenotype)

Genetics is the study of how genes bring about certain traits expressed in organisms. Well remember that genes are segments of DNA that define what certain traits or characteristics and organism may have. For example, genes are responsible for the color of eyes that you have.

Since most complex organisms are **diploid cells**, they have a double set of chromosomes. We learned in the last lesson about **meiosis** that diploid cells are the result of two haploid cells called gametes uniting through the process of sexual reproduction to for a new individual. Remember that in humans, these **haploid** gametes are in the form of eggs and sperm (each from a different parent and each containing 23 chromosomes) when they unite, they have 46 chromosomes. The offspring of such a union then has **genes** from each of these parents. Each of the parents offers a different possibility for each gene. Lets think of the example of eye color we talked about earlier. Your parents have a certain color of eyes, with the combination of the gametes from them, you now have the possibility to have the same color of eyes as one (or both) of your parents. The different forms of the same gene (like eye color) are called **alleles**. The type of eye color that you have is ultimately determined by the alleles that you inherited from your parents.

We have genes that determine virtually everything about us. The genes that make up an organism are known as the **genotype** (the type of genes that exist). The expression of these genes is called the **phenotype** - or what we actually see. For example, if you have blue eyes, we could say that you have the phenotype for blue eyes, even though there may have been a genotype present for brown eyes.

Homozygous vs. Heterozygous

In genetics we have some terms that allow us to talk about the pairs of alleles that are inherited. When there are two identical alleles for a particular trait, they are said to be **homozygous** (hom - meaning the same). In homozygous individuals the alleles express themselves. On the other hand, when there are two different alleles for a particular characteristic they are said to be **heterozygous** (hetero - meaning different). In the case of a heterozygous individual, the alleles may interact with each other, or only one allele may be expressed. Well talk more about this later in the lesson.

Dominant vs. Recessive

The word **dominant** is used when talking about one allele expressing itself and another one not. For convenience, we generally label a dominant allele with a capital letter like **A**. The word **recessive** is used to talk about those traits that are overshadowed by the dominant allele. Also for convenience, we label the recessive allele with a small letter like **a**.

Now we can combine the terms weve just learned (**homozygous, heterozygous, dominant, recessive**) to help us along with talking about genetics. We will talk about them here and then after a few examples it will become clearer to you.

We use the term **homozygous dominant** when we talk about a trait that has two dominant alleles for that trait, like blue eyes. If we were to label this type we would use two capital letters **AA**. Another combination is described as being **heterozygous dominant** simply means that there is a dominant allele and a recessive allele present and is labeled **Aa**. In this case the dominant allele is generally expressed. There are also cases where we have a condition described as being **homozygous recessive**, which simply means that there are two alleles for the recessive trait and is labeled **aa**.

Mendels Laws

We have Gregor Mendel to thank for reaching this type of description of genes, alleles, and how their respective traits are all expressed. Mendel worked in his garden with pea plants. He studied different traits that the plants expressed like: height, pod color, pod shape, seed shape, seed color, flower color, and flower location. The nice thing about the pea plants that he studied is that they are self-pollinating (they pollinate themselves). This means that after many different generations of plants the peas develop individuals that are homozygous (either dominant or recessive AA or aa) for certain traits. These types of individuals are called pure-line.

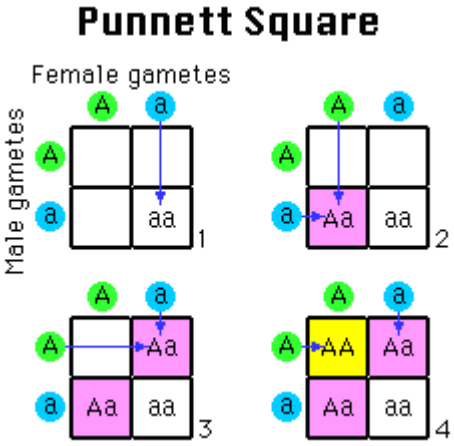
Mendel started his experiments with just such a line and he called them the **parent generation** (**P**). He crossed homozygous plants that were tall with homozygous plants that were short. He discovered that all the plants that came from this crossing were tall. He called this the **F1 generation** (**first generation**). Continuing the experiment, he crossed the F1 generation among themselves and the new offspring were called the **F2 generation** (**second generation**). He observed that ¾ of the plants were tall and that ¼ of the plants were short. Over many years of experimenting with the other traits of pea plants that were mentioned earlier, he came up with several conclusions that are commonly called Mendels laws of genetics. Here are three of them:

1. **Law of segregation** - diploid cells have pairs of genes, and during meiosis the two genes of each pair separate and end up in different gametes.

2. **Law of dominance** - when organism has two different alleles for the same trait, one allele dominates.

3. **Law of independent assortment** - gene pairs of homologous chromosomes are sorted into one gamete or another independently of how gene pairs on other chromosomes are sorted.

One of the nice things about genetics is that it allows us to predict the probability of the traits that are inherited from one generation to the next. Remember the labels we learned for the homozygous/heterozygous dominant and the homozygous recessive traits (AA, Aa, aa)? These labels help us do just that. Lets say we wanted to know the probability of attached earlobes in the first generation. We know that the parents are both heterozygous for the dominant trait (which is attached earlobes - so they will each be represented by the label Aa). Now, since meiosis offers the possibility that either one of these alleles (the dominant A or the recessive a) could be passed along in the gametes to the next generation. To help us figure out the possible traits that will be expressed, we will keep track of the gamete using a box called the **Punnett square**. (See diagram)



Notice that the female gamete possibilities (A and a) are placed along the top of the square, and the male gamete possibilities (A and a) are placed along the side. We know follow the arrows and place each gamete in its respective row and column.

To express the results of such a cross, we use some ratios - one is the **genotypic ratio** which will tell us the genes present in all the possibilities and the other is the **phenotypic ratio** which will tell us exactly what will be seen. From the Punnett square we see that the genotypic is 1 homozygous dominant (AA) for attached earlobes, 2 heterozygous dominant alleles (Aa) for attached earlobes, and 1 homozygous allele (aa) for the recessive trait - unattached earlobes. Another way to write the genotypic ratio is 1:2:1. Now for the phenotypic ratio. From the genotype we see that there will be the possibility of 3 to 1 that the dominant trait will be expressed (remember that in heterozygous situations the dominant allele overshadows the recessive one). The ratio is then written 3:1 for the dominant trait. We got the 3 because of the 2 heterozygous alleles and the single homozygous allele.

Other ways that genes are expressed

There are other principles that govern genetics that are not completely explained by Mendel. So here are some of them.

Incomplete Dominance

Incomplete dominance simply means that in the combinations of some alleles, no one trait dominates another. For example, in some flowers, there may be two alleles for color. That means if two alleles for white are present - then the flower would be white, and if two alleles for red were present - then the flower would be red, but when there is one allele for white flowers and one allele for red flowers - the flower is pink (the combination of the two colors).

Multiple Alleles

In some cases there are times when there are more than two possible alleles that define a certain trait or characteristic - that is what is meant by **multiple alleles**. An example of multiple alleles is **human blood type**. Our blood type is determined by a single gene that has three possible alleles A,B, and O. The alleles for both types A and B are codominant, but the allele for type O is recessive to both A and B. So lets see the possibilities since a person can have only two of the three alleles. If a person were to have an A allele and a B allele, they would have blood type AB (remember they are codominant). If a person has two A alleles, or an A and an O allele, they would have blood type O (remember that O is recessive to types A and B). Likewise, if a person had two B alleles or a B and an O allele, they would have blood type B. The only way a person could have blood type O is if both alleles were type O.

Polygenic Inheritance

We have talked about the fact that certain characteristics are determined by alleles found at a single place along the chromosome (called the gene locus). There are characteristics though that are determined by the interaction of many genes on several different chromosomes or many genes on the same chromosome. This type of occurrence is known as **polygenic inheritance** and a good example of that is **skin color**. The genes for skin color are located on many different genes.

Gene Linkage

A chromosome has many thousands of genes. It is, therefore, very common for a large number of genes to be inherited together if they are all on the same chromosome. This concept is called **gene linkage**. The groups of genes that are inherited together are called **linkage groups**. We must add that the closer the genes are to each other on the chromosome, the higher the probability that they will be inherited together.

Sex Linkage

In humans there are 23 pairs of chromosomes, 22 of those are called **autosomes**, and there is one pair of **sex chromosomes**. There are two types of sex chromosomes: **X** (females have two X chromosomes) and **Y** (males have an X and a Y chromosome). The Y chromosome is much shorter than the X chromosome and, for that reason, lacks a certain number of genes. This may cause some **sex-linked** conditions to occur. A sex-linked condition occurs because when a gene is found on the X chromosome the other allele probably is found on the other X chromosome (in females). This may not be the case for males. Sex-linked traits are those genes found on the sex chromosomes. An example of a sex-linked trait is **colorblindness**. The gene for color blindness is found on the X chromosome. A female is hardly ever colorblind because the gene for normal vision is dominant and found on the other X chromosome and therefore overshadows the colorblind gene. However with males, because of the shortened Y chromosome, the dominant allele for normal vision may not be found and the recessive allele for colorblindness is expressed.

Conclusion

As we have seen there are many factors that influence the traits that organisms have. Those traits are studied by looking at genetics. In this lesson we have established a base for us to understand these concepts. In the next lesson we will look at the molecular genetics and DNA.