Gregor Mendel and the Principles of Inheritance

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Gregor Mendel's principles of inheritance form the cornerstone of modern genetics. So just what are they?

Ever wonder why you are the only one in your family with your grandfather's nose? The way in which traits are passed from one generation to the next-and sometimes skip generations-was first explained by Gregor Mendel. By experimenting with pea plant breeding, Mendel developed three principles of inheritance that described the transmission of genetic traits, before anyone knew genes existed. Mendel's insight greatly expanded the understanding of genetic inheritance, and led to the development of new experimental methods.

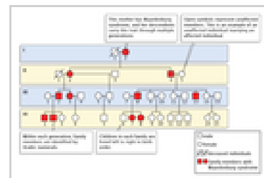

Figure 1

Figure Detail

Traits are passed down in families in different patterns. Pedigrees can illustrate these patterns by following the history of specific characteristics, or phenotypes, as they appear in a family. For example, the pedigree in Figure 1 shows a family in which a grandmother (generation I) has passed down a characteristic (shown in solid red) through the family tree. The inheritance pattern of this characteristic is considered dominant, because it is observable in every generation. Thus, every individual who carries the genetic code for this characteristic will show evidence of the characteristic. In contrast, Figure 2 shows a different pattern of inheritance, in which a characteristic disappears in one generation, only to reappear in a subsequent one. This pattern of inheritance, in which the parents do not show the phenotype but some of the children do, is considered recessive. But where did our knowledge of dominance and recessivity first come from?

Gregor Mendel's Courage and Persistence

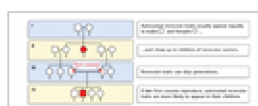

Figure 2

Figure 3

Our modern understanding of how traits may be inherited through generations comes from the principles proposed by Gregor Mendel in 1865. However, Mendel didn't discover these foundational principles of inheritance by studying human beings, but rather by studying *Pisum sativum*, or the common pea plant. Indeed, after eight years of tedious experiments with these plants, and—by his own admission—"some courage" to persist with them, Mendel proposed three foundational principles of inheritance. These principles eventually assisted clinicians in human disease research; for example, within just a couple of years of the rediscovery of Mendel's work, Archibald Garrod applied Mendel's principles to his study of alkaptonuria. Today, whether you are talking about pea plants or human beings, genetic traits that follow the rules of inheritance that Mendel proposed are called Mendelian.

Mendel was curious about how traits were transferred from one generation to the next, so he set out to understand the principles of heredity in the mid-1860s. Peas were a good model system, because he could easily control their fertilization by transferring pollen with a small paintbrush. This pollen could come from the same flower (self-fertilization), or it could come from another plant's flowers (cross-fertilization). First, Mendel observed plant forms and their offspring for two years as they self-fertilized, or "selfed," and ensured that their outward, measurable characteristics remained constant in each generation. During this time, Mendel observed seven different characteristics in the pea plants, and each of these characteristics had two forms (Figure 3). The characteristics included height (tall or short), pod shape (inflated or constricted), seed shape (smooth or winkled), pea color (green or yellow), and so on. In the years Mendel spent letting the plants self, he verified the purity of his plants by confirming, for example, that tall plants had only tall children and grandchildren and so forth. Because the seven pea plant characteristics tracked by Mendel were consistent in generation after generation of self-fertilization, these parental lines of peas could be considered pure-breeders (or, in modern terminology, homozygous for the traits of interest). Mendel and his assistants eventually developed 22 varieties of pea plants with combinations of these consistent characteristics.

Mendel not only crossed pure-breeding parents, but he also crossed hybrid generations and crossed the hybrid progeny back to both parental lines. These crosses (which, in modern terminology, are referred to as F_1, F_1 reciprocal, F_2, B_1, and B_2) are the classic crosses to generate genetically hybrid generations.

Understanding Dominant Traits

Before Mendel's experiments, most people believed that traits in offspring resulted from a blending of the traits of each parent. However, when Mendel cross-pollinated one variety of purebred plant with another, these crosses would yield offspring that looked like either one of the parent plants, not a blend of the two. For example, when Mendel cross-fertilized plants with wrinkled seeds to those with smooth seeds, he did not get progeny with semi-wrinkly seeds. Instead, the progeny from this cross had only smooth seeds. In general, if the progeny of crosses between purebred plants looked like only one of the parents with regard to a specific trait, Mendel called the expressed parental trait the dominant trait. From this simple observation, Mendel proposed his first principle, the principle of uniformity; this principle states that all the progeny of a cross like this (where the parents differ by only one trait) will appear identical. Exceptions to the principle of uniformity include the phenomena of penetrance, expressivity, and sex-linkage, which were discovered after Mendel's time.

Understanding Recessive Traits

When conducting his experiments, Mendel designated the two pure-breeding parental generations involved in a particular cross as P_1 and P_2, and he then denoted the progeny resulting from the crossing as the filial, or F_1, generation. Although the plants of the F_1 generation looked like one parent of the P generation, they were actually hybrids of two different parent plants. Upon observing the uniformity of the F_1 generation, Mendel wondered whether the F_1 generation could still possess the nondominant traits of the other parent in some hidden way.

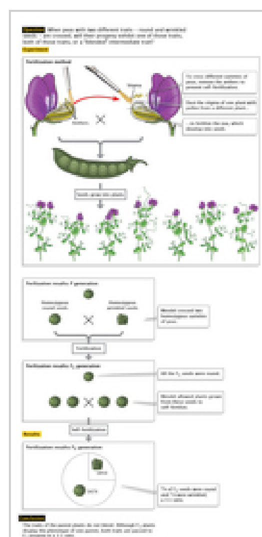

Figure 4

Figure Detail

To understand whether traits were hidden in the F_1 generation, Mendel returned to the method of self-fertilization. Here, he created an F_2 generation by letting an F_1 pea plant self-fertilize (F_1 x F_1). This way, he knew he was crossing two plants of the exact same genotype. This technique, which involves looking at a single trait, is today called a monohybrid cross. The resulting F_2 generation had seeds that were either round or wrinkled. Figure 4 shows an example of Mendel's data.

When looking at the figure, notice that for each F_1 plant, the self-fertilization resulted in more round than wrinkled seeds among the F_2 progeny. These results illustrate several important aspects of scientific data:

1. Multiple trials are necessary to see patterns in experimental data.
2. There is a lot of variation in the measurements of one experiment.
3. A large sample size, or "N," is required to make any quantitative comparisons or conclusions.

In Figure 4, the result of Experiment 1 shows that the single characteristic of seed shape was expressed in two different forms in the F_2 generation: either round or wrinkled. Also, when Mendel averaged the relative proportion of round and wrinkled seeds across all F_2 progeny sets, he found that round was consistently three times more frequent than wrinkled. This 3:1 proportion resulting from F_1 x F_1 crosses suggested there was a hidden recessive form of the trait. Mendel recognized that this recessive trait was carried down to the F_2 generation from the earlier P generation.

Mendel and Alleles

As mentioned, Mendel's data did not support the ideas about trait blending that were popular among the biologists of his time. As there were never any semi-wrinkled seeds or greenish-yellow seeds, for example, in the F_2 generation, Mendel concluded that blending should not be the expected outcome of parental trait combinations. Mendel instead hypothesized that each parent contributes some particulate matter to the offspring. He called this heritable substance "elementen." (Remember, in 1865, Mendel did not know about DNA or genes.) Indeed, for each of the traits he examined, Mendel focused on how the elementen that determined that trait was distributed among progeny. We now know that a single gene controls seed form, while another controls color, and so on, and that elementen is actually the assembly of physical genes located on chromosomes. Multiple forms of those genes, known as alleles, represent the different traits. For example, one allele results in round seeds, and another allele specifies wrinkled seeds.

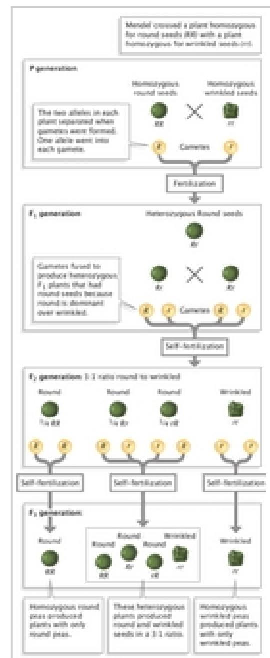

Figure 5

One of the most impressive things about Mendel's thinking lies in the notation that he used to represent his data. Mendel's notation of a capital and a lowercase letter (*Aa*) for the hybrid genotype actually represented what we now know as the two alleles of one gene: *A* and *a*. Moreover, as previously mentioned, in all cases, Mendel saw approximately a 3:1 ratio of one phenotype to another. When one parent carried all the dominant traits (*AA*), the F_1 hybrids were "indistinguishable" from that parent. However, even though these F_1 plants had the same phenotype as the dominant P_1 parents, they possessed a hybrid genotype (*Aa*) that carried the potential to look like the recessive P_1 parent (*aa*). After observing this potential to express a trait without showing the phenotype, Mendel put forth his second principle of inheritance: the principle of segregation. According to this principle, the "particles" (or alleles as we now know them) that determine traits are separated into gametes during meiosis, and meiosis produces equal numbers of egg or sperm cells that contain each allele (Figure 5).

Dihybrid Crosses

Mendel had thus determined what happens when two plants that are hybrid for one trait are crossed with each other, but he also wanted to determine what happens when two plants that are each hybrid for two traits are crossed. Mendel therefore decided to examine the inheritance of two characteristics at once. Based on the concept of segregation, he predicted that traits must sort into gametes separately. By extrapolating from his earlier data, Mendel also predicted that the inheritance of one characteristic did not affect the inheritance of a different characteristic.

Mendel tested this idea of trait independence with more complex crosses. First, he generated plants that were purebred for two characteristics, such as seed color (yellow and green) and seed shape (round and wrinkled). These plants would serve as the P_1 generation for the experiment. In this case, Mendel crossed the plants with wrinkled and yellow seeds (*rrYY*) with plants with round, green seeds (*RRyy*). From his earlier monohybrid crosses, Mendel knew which traits were dominant: round and yellow. So, in the F_1 generation, he expected all round, yellow seeds from crossing these purebred varieties, and that is exactly what he observed. Mendel knew that each of the F_1 progeny were dihybrids; in other words, they contained both alleles for each characteristic (*RrYy*). He then crossed individual F_1 plants (with genotypes *RrYy*) with one another. This is called a dihybrid cross. Mendel's results from this cross were as follows:

- 315 plants with round, yellow seeds
- 108 plants with round, green seeds
- 101 plants with wrinkled, yellow seeds
- 32 plants with wrinkled, green seeds

Thus, the various phenotypes were present in a 9:3:3:1 ratio (Figure 6).

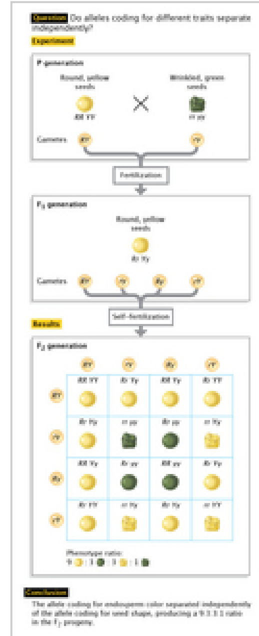

Figure 6

Figure Detail

Next, Mendel went through his data and examined each characteristic separately. He compared the total numbers of round versus wrinkled and yellow versus green peas, as shown in Tables 1 and 2.

Table 1: Data Regarding Seed Shape

	Round	**Wrinkled**
Number of plants	315 + 108 = 423	101 + 32 = 133
Proportion of total	3.2	1

Table 2: Data Regarding Pea Color

	Yellow	**Green**
Number of plants	315 + 101 = 416	108 + 32 = 140
Proportion of total	2.97	1

The proportion of each trait was still approximately 3:1 for both seed shape and seed color. In other words, the resulting seed shape and seed color looked as if they had come from two parallel monohybrid crosses; even though two characteristics were involved in one cross, these traits behaved as though they had segregated independently. From these data, Mendel developed the third principle of inheritance: the principle of independent assortment. According to this principle, alleles at one locus segregate into gametes independently of alleles at other loci. Such gametes are formed in equal frequencies.

Mendel’s Legacy

More lasting than the pea data Mendel presented in 1862 has been his methodical hypothesis testing and careful application of mathematical models to the study of biological inheritance. From his first experiments with monohybrid crosses, Mendel formed statistical predictions about trait inheritance that he could test with more complex experiments of dihybrid and even trihybrid crosses. This method of developing statistical expectations about inheritance data is one of the most significant contributions Mendel made to biology.

But do all organisms pass their on genes in the same way as the garden pea plant? The answer to that question is no, but many organisms do indeed show inheritance patterns similar to the seminal ones described by Mendel in the pea. In fact, the three principles of inheritance that Mendel laid out have had far greater impact than his original data from pea plant manipulations. To this day, scientists use Mendel's principles to explain the most basic phenomena of inheritance.

References and Recommended Reading

Mendel, G. Versuche über Plflanzen-hybriden.Verhandlungen des naturforschenden Ver-eines in Brünn, Bd. IV für das Jahr 1865, Abhand-lungen, 3-47 (1866) (Bateson translation) (link to article)

Strachan, T., & Read, A.P. Mendelian pedigree patterns. *Human Molecular Genetics 2* (Garland Science, 1999)