The law of segregation

Mendel's law of segregation. Genotype, phenotype, and alleles. Heterozygous/homozygous. 2 x 2 Punnet squares.

Key points:

Gregor Mendel studied inheritance of traits in pea plants. He proposed a model where pairs of "heritable elements, " or genes, specified traits. Genes come in different versions, or alleles. A Dominant allele hides a recessive allele and determines the organism's appearance.

When an organism makes gametes, each gamete receives just one gene copy, which is selected randomly. This is known as the law of segregation.

A Punnet square can be used to predict genotypes (allele combinations) and phenotypes (observable traits) of offspring from genetic crosses.

A test cross can be used to determine whether an organism with a dominant phenotype is Homozygous or heterozygous.

Introduction

Today, we know that many of people's characteristics, from hair colour to height to risk of diabetes, are influenced by genes. We also know that genes are the way parents pass characteristics on to their children (including things like dimples, or—in the case of me and my father—a terrible singing voice). In the last hundred years, we've come to understand that genes are actually pieces of DNA that are found on chromosomes and specify proteins.

But did we always know those things? Not by a long shot! About 150 years ago, a monk named Gregor Mendel published a paper that first proposed the existence of genes and presented a model for how they were inherited. Mendel's work was the first step on a long road, involving many hard-working scientists, that's led to our present understanding of genes and what they do.

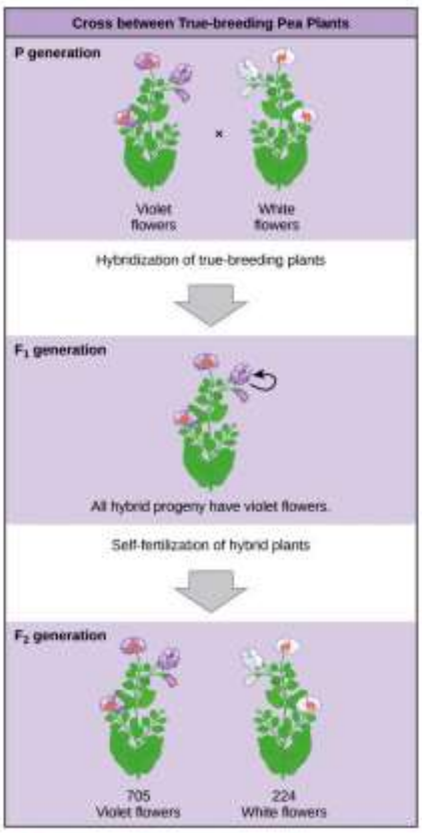
In this article, we'll trace the experiments and reasoning that led Mendel to formulate his model for the inheritance of single genes.

Mendel's model: It started with a 3 : 1 Ratio

Mendel studied the genetics of pea plants, and he traced the inheritance of a variety of characteristics, including flower colour, flower position, seed colour , and seed shape. To do so, he started by crossing pure breeding parent plants with different forms of a characteristic, such as violet and white flowers. Pure breeding just means that the plant will always make more offspring like itself, when self-fertilized over many generations. What is self-fertilization?

What results did Mendel find in his crosses for flower colour? In the parental, or P generation, Mendel crossed a pure-breeding violet-flowered plant to a pure-breeding white-flowered plant. When he gathered and planted the seeds produced in this cross, Mendel found that 100 percent of the plants in the next generation, or F generation, had violet flowers.

Conventional wisdom at that time would have predicted that the hybrid flowers should be pale violet —that is, that the parents' traits should blend in the offspring. Instead, Mendel’s results showed that the white flower trait had completely disappeared. He called the trait that was visible in the F generation (violet flowers) the dominant trait, and the trait that was hidden or lost (white flowers) the recessive trait.



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Importantly, Mendel did not stop his experimentation there. Instead, he let the F plants self-fertilize. Among their offspring, called the F generation, he found that 705 plants had violet flowers and 224 had white flowers. This was a ratio of 3.15 violet flowers to one white flower, or approximately 3:1.

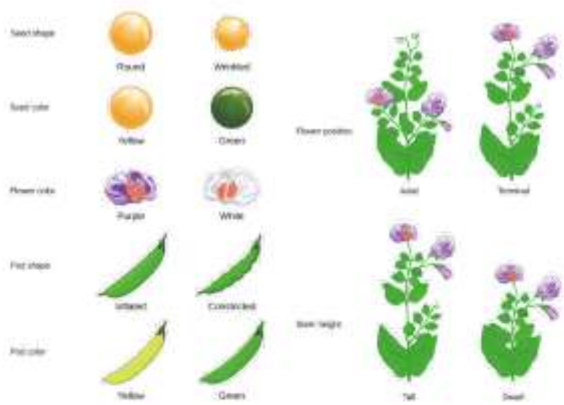
This 3:1ratio was no fluke. For the other six characteristics that Mendel examined, both the F and F generations behaved in the same way they did for flower colour. One of the two traits would disappear completely from the F generation, only to reappear in the F generation in a ratio of roughly 3:1

As it turned out, the 3:1ratio was a crucial clue that let Mendel crack the puzzle of inheritance. Let's take a closer look at what Mendel figured out.

Mendel's model of inheritance

Based on his results (including that magic 3:1ratio), Mendel came up with a model for the inheritance of individual characteristics, such as flower colour.

In Mendel's model, parents pass along “heritable factors, "which we now call genes that determine the traits of the offspring. Each individual has two copies of a given gene, such as the gene for seed colour (Y gene) shown below. If these copies represent different versions, or alleles, of the gene, one allele—the dominant one—may hide the other allele—the recessive one. For seed colour, the dominant yellow allele Y hides the recessive green allele y.



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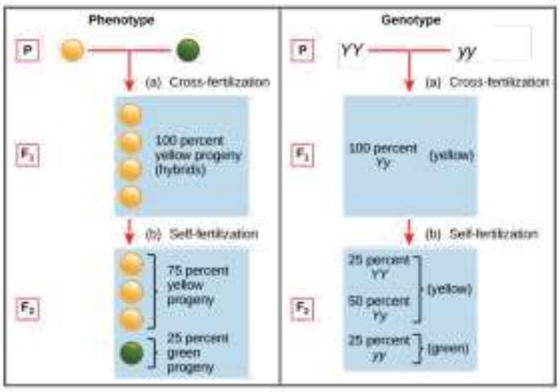

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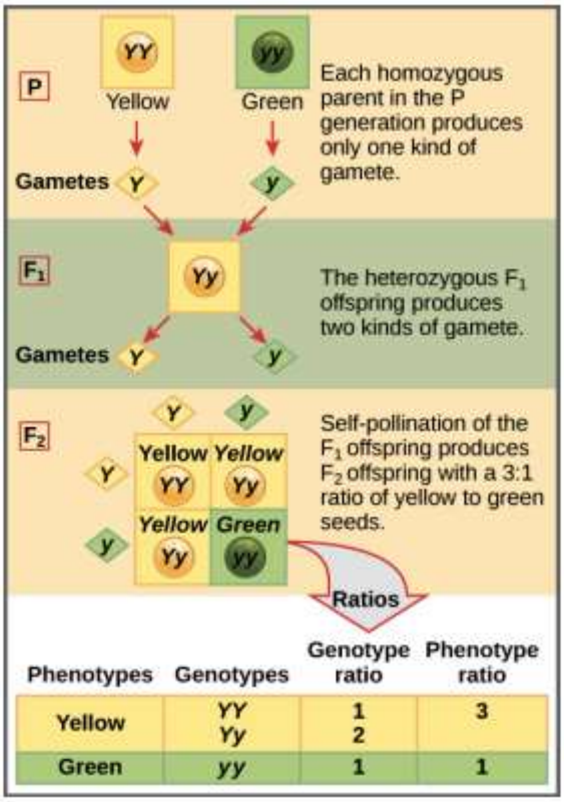
The set of alleles carried by an organism is known as its genotype. Genotype determines phenotype, an organism's observable features. When an organism has two copies of the same allele (say, YY or yy), it is said to be homozygous for that gene. If, instead, it has two different copies (like Y y), we can say it is heterozygous. Phenotype can also be affected by the environment in many real-life cases, though this did not have an impact on Mendel's work.

Mendel's model: The law of segregation

So far, so good. But this model alone doesn't explain why Mendel saw the exact patterns of inheritance he did. In particular , it doesn't account for the 3:1ratio.

For that, we need Mendel's law of segregation.

According to the law of segregation, only one of the two gene copies present in an organism is distributed to each gamete (egg or sperm cell) that it makes, and the allocation of the gene copies is random. When an egg and a sperm join in fertilization, they form a new organism, whose genotype consists of the alleles contained in the gametes. The diagram below illustrates this idea:



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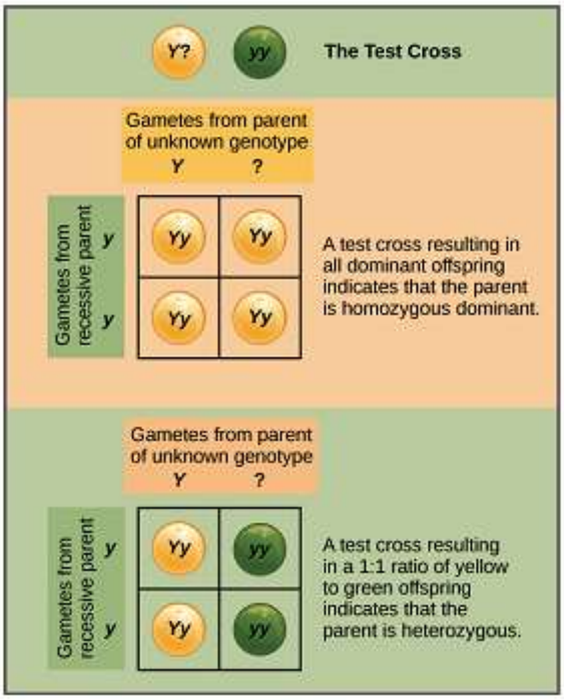
The four-squared box shown for the F generation is known as a punnet square. To prepare a punnet square, all possible gametes made by the parents are written along the top (for the father) and side (for the mother) of a grid. Here, since it is self-fertilization, the same plant is both mother and father.

The combinations of egg and sperm are then made in the boxes in the table, representing fertilization to make new individuals. Because each square represents an equally likely event, we can determine genotype and phenotype ratios by counting the squares.

The test cross

Mendel also came up with a way to figure out whether an organism with a dominant phenotype (such as a yellow-seeded pea plant) was a heterozygote (Y y) or a homozygote (YY). This technique is called a test cross and is still used by plant and animal breeders today.

In a test cross, the organism with the dominant phenotype is crossed with an organism that is Homozygous recessive (e.g., green-seeded):



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If the organism with the dominant phenotype is homozygous, then all of the F offspring will get a dominant allele from that parent, be heterozygous, and show the dominant phenotype. If the organism with the dominant phenotype organism is instead a heterozygote, the F offspring will be half heterozygotes (dominant phenotype) and half recessive homozygotes (recessive phenotype). The fact that we get a 1:1ratio in this second case is another confirmation of Mendel's law of segregation.

Is that Mendel's complete model of inheritance?

Not quite! We've seen all of Mendel's model for the inheritance of single genes. However, Mendel's complete model also addressed whether genes for different characteristics (such as flower colour and seed shape) influence each other's inheritance. You can learn more about Mendel's model for the inheritance of multiple genes in the law of independent assortment article.

One thing I find pretty amazing is that Mendel was able to figure out his entire model of inheritance simply from his observations of pea plants. This wasn't because he was some kind of crazy super genius, but rather, because he was very careful, persistent, and curious, and also because he thought about his results mathematically (for instance, the 3:1ratio). These are some of the qualities of a great scientist—ones that anyone, anywhere, can develop!