

THE BIRTH OF GENETICS AND GREGOR MENDEL'S LAW OF SEGREGATION

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Mendel's Abbey, 2005



Mendel's Abbey, a side view, 2005

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Historical Background

Science begins with simple questions. "Why is the sky blue?" Why do non-freckled parents have only non-freckled children?" "Why do pea plants only give rise to peas, not to cows or roses?" Some scientific discoveries are made, routinely, by all of us. For instance, sometime during your early childhood you might have looked at a mirror and figured out, all by yourself, that you were actually facing your own reflection. Similarly, discoveries, even great discoveries made by professional scientists, do not seem to require much more than a healthy dose of curiosity, open-mindedness, and imagination (and a very small dose of TV, materialism, and propaganda and memorization masquerading as schooling). [Antony van Leeuwenhoek](#) discovered a whole world in a drop of pond water. With his improved telescope, Galileo could see Jupiter's moons. Though we admire the technical ingenuity, courage to challenge dogmas, or curiosity about the world which such discoveries require, we understand how the likes of Leeuwenhoek and Galileo could come around to making them. With a bit more time, brains, and imagination, we too, we feel, could have discovered the existence of spermatozoa or Jupiter's moons. It took genius to discover them, but only "ordinary" genius. There is, however, another kind of genius which has an almost magical quality to it. We might, or might not, understand such work, but we are astounded by the

psychological process that gave rise to it. How in heaven's name, we wonder, did it occur to a "magical" genius to carry out his/her work? And how did this person manage to interpret it in such a radical new way?



One such magical genius was Gregor Mendel, perhaps one of the greatest scientists of the nineteenth century. His experiments not only revolutionized biology, but left historians of science wondering: What mental processes led to their conception? How did Mendel have the intellectual courage to traverse new territories for so many years, totally alone? What made it possible for him to come up with his beautiful deductions and with a radically new theory of heredity? How did he come to see, for the first time in the history of biology, that simple numerical ratios provided an important key to unlocking one of nature's secrets?

Mendel lived in central Europe, in a region which now belongs to the Czech Republic but that, during his lifetime, belonged to the Austrian-Hungarian Empire. He spent most of his adult life as a monk (and later, abbot) in an Augustinian monastery. As a farmer's son in class-conscious Europe, the monastic life, in his words, spared him the "perpetual anxiety about a means of livelihood." It also gave him free time to tinker with peas, tomatoes, hawkweed, bees, and other curiosities of nature.

He carried out his experiments with the common edible pea in his small garden plot in the monastery. These experiments were begun in 1856 and completed some eight years later. In 1865, he described his experiments in two lectures he gave at a regional scientific conference. In the first lecture he described his observations and experimental results. In the second, which was given one month later, he boldly explained them.

The forty or so scientists which made up his audience listened to him politely enough, but no one asked a single question. Most likely, they didn't understand what he was talking about. Later, he also carried a correspondence with one of the most eminent biologists of the time, who also failed to appreciate Mendel's work. In 1886, Mendel's results were published in an obscure scientific journal. He might as well have dropped a leaded spoon into the ocean; the paper was ignored by the scientific community. At times, he must have entertained doubt about his work, but not always: "My time will come," he reportedly told a friend.

His time came, though later than he might have expected. His emergence from obscurity began in 1900--sixteen years after his death. In the West his work was generally accepted by the scientific community by the 1920s.

Conceptually, Mendel's experiments were not all that difficult. His scientific contemporaries lacked, for one thing, open-mindedness. Moreover, they might have been unable to accept, deep down, the proposition that an obscure monk, a farmer's son, and a person who was discouraged from completing his university education because he "lacked insight," could have something important to

say. At any rate, in your case all this is not a problem. You probably accept history's judgment that he was an insightful person and that geniuses can spring from impoverished, plebian backgrounds. What you do have in common with Mendel's scientific contemporaries is, most likely, ignorance of elementary statistics. The next section will give you the necessary background information. To get the most out of it, try to actually answer each question as you go along.

Probability: Random Events

Q1. Let us first examine the results of single random events. Suppose you toss a coin once. What are the chances of getting heads (H)? ____ Tails (T)? ____

Q2. What are the chances that your next child will be a boy? ____ A girl? ____

Q3. Now toss a coin 10 times and record the number of heads (Hs) and tails (Ts) in the space below. (Make sure that the process is indeed random. Flip the coin a few times in your hand before each toss; examine it only after it lands on the floor).

H ____ T ____

For the class as a whole, the results were: H ____ T ____

Q4. Most likely, some of your classmates' observations are not in line with the expected 50/50 distribution? Why?

Q5. Are class totals more or less in line with the expected distribution? Why?

Mark Twain (citing Benjamin Disraeli)--a contemporary of Gregor Mendel--observed that there are "lies, damn lies, and statistics." In a sense, he was right. If you tossed a coin just once, and got H, then you didn't get the 1 in 2 distribution statistical theory seems to demand, but a 1 in 1 distribution. Similarly, although each participant flipped a coin 10 times, the data of some diverged considerably from the expected 1 in 2 distribution. Assuming a minimum of 10 students in your class, however, data for the entire class almost certainly approached the theoretical expectation of equal odds. These commonsense observations suggest two generalizations:

--Actual distributions of random events rarely match theoretical expectations. A coin that has been randomly tossed 400 times will almost never yield 200 Ts and 200 Hs. As a rule, something like 195 Ts and 205 Hs is all we can expect.

--The larger the number of trials, the closer the fit between observed and theoretical values. Thus, if you toss a coin just once, theory demands a 1 in 2 chance of H or T, in real life you would always observe just one event. In 30 trials, you very well might get a 20/10 distribution. But in 3000 trials, you will almost never get a 2000/1000 distribution.

Until artificial offspring sex selection becomes a reality, the distribution of boys and girls obeys similar rules. The coin flipper in this case is nature. To put it in a somewhat simplified form, as far as a child's sex is concerned, a woman's anatomy plays a fairly passive role. Semen contains two populations of sperm in roughly equal numbers. One type, when united with a woman's egg, gives rise to a girl (G). The other, when united with the same egg, gives rise to a boy (B). For any given

conception, chance determines which type of sperm will fertilize the egg.

Q6. What are a couple's chances of having a girl?

Q7. Assuming that either type of sperm is as likely as the other to fertilize an egg (reality is a bit more complicated), what is the expected distribution of boys and girls in a small village of 100 souls?
 ____ In a city of two million? ____

Probability: Two Independent Events

Imagine that you simultaneously tossed a nickel and a dime. Each coin alone, we know already, has an equal chance of yielding H or T, but what can we expect when the 2 are flipped at the same time? Obviously, the nickel can be H or T, and so can the dime. Our problem is putting the two together to determine the types of events and their respective probabilities.

Let us then construct a table, and carry out an experiment. Please divide into pairs, and construct the following table (a penny or a quarter will be just as good). Simultaneously, one person should toss a nickel and the other person should toss a quarter. For each toss of the two coins, 4 outcomes are possible. Please record each outcome with one vertical line in the table below, and then summarize the total of 10 simultaneous tossed for your pair. Next, summarize the totals for your entire row. We shall then summarize, on board, the results for the entire class.

Q8. Our prediction: ??? Our actual results for the class as a whole?

		Dime	
		H	T
Nickel	H		
	T		

How can we explain what we have just seen? For any given throw, the nickel has a 1 in 2 chance of yielding H. Assuming that the nickel yielded an H, the dime is equally likely to yield H or T. So the chances of [nickel: H; dime: H] is one-half of that one-half, or one-quarter. Similarly, there is a one-quarter chance of [nickel: H, dime T].

The nickel also has a one-half chance of yielding T, leading again to 2 equally probable events; [Nickel: T, dime H]; and [Nickel: T;, dime: T]. Looking then at the simultaneous toss of two coins, we have four equally probable events, as we have seen.

Similar probabilities prevail in a large population of two-child families:

		Dad	
		B	G
Mom	G		
	G		

In other words, in a population of 100,000 such families, one expects some 25,000 with 2 boys, 25,000 with an older girl and a younger boy, 25,000 with an older boy and a younger girl, and 25,000 with two girls.

These examples suggest the following generalization:

--The probability that two independent events will occur together equals the product of their individual probabilities. Thus, when you randomly toss two coins, the two events are independent: the outcome of one has no bearing on the outcome of the other. Since the probability of each is 1/2, their combined probability is $1/2 \times 1/2$ or 1/4.

You are ready now to work out a few more puzzles.

Q9. In this population of 100, 000, what are the chances of having a boy and a girl, irrespective of their order of birth? ____

Q10. What are the chances for two girls? ____

Q11. Let's divide these 100,000 two-child families into 2 categories. The first consists of families with 2 girls. The second consists of all other two-child families. How many families would belong to the first group (the one with 2 girls)? ____ How many to the second (all the other combinations)? If you divide the second number by the first, the expected ratio between them would be ____ to 1.

Q12. Imagine that a philanthropist came up with a \$1000 prize to any family in your community that meets the following conditions: it has 2, and only 2, natural born daughters. Imagine that all qualified families applied for the bonus, and that after being checked for accuracy, 100,000 received the prize. The philanthropist now comes up with an even more curious announcement: He will give a \$1,000,000 prize to the first person who would tell him, and then properly explain his deductions, the approximate number of two-child families in this community who do not have two girls. How would you go about getting this prize?

Dad	
B	G

Mom	G		
	G		

Imagine yourself a banker. To save money, you devised an ingenious scheme of monitoring the vigilance of your bank's night watchman. At the opposite ends of the building, there are 2 light switches. Every hour during his 10-hour shift, the guard must approach one switch and flip a coin. If it's Heads, he is to turn the switch on, if it's Tails, he is to turn it off. He then must approach the second switch and repeat the same procedure. If both switches are on, the light is on. If either one is off, or if both are off, the light is off. You have installed a special machine that tells you, for every hour, whether the light has been on or off. Four months and 1000 hours later, you examine the lighting record to determine whether the guard should receive a raise or be fired. You observe that during that time, the light has been on 570 hours and off 430 hours.

		Switch II	
		On	Off
Switch I	On		
	Off		

Q13. What are your theoretical expectations for the number of hours the lights should have been on and off? Why? Should the guard be promoted or sacked?

Now let us carry out this thought experiment. Imagine that I gave you 2 vials, each containing one red and one green toothpick. Each vial is covered with a lid. At the bottom of each lid there is a hole through which, at any given time, the head of one, and only one, toothpick can pass when the vial is shaken. So shaking a vial leads to the *segregation* of the two toothpicks--they separate from each other, one remains wholly inside, the other is protruding out of the vial.

Imagine that the class is now divided into pairs. One member of each pair is holding the first vial, the other, holds the second vial. In each case, by shaking both vials at the same time, either a red or a green toothpick pops through the hole: the two toothpicks *segregate* from each other. We expect in this case 4 equally probable 2-toothpicks permutations. From this we can calculate any given ratio. If a red toothpick is just as likely to penetrate the hole as the green one, what is your theoretical expectation for 100 trials for both events?

		Vial 2	
		Red	Green
Vial 1	Red		
	Green		

Q14. How many are expected to be either Red/Green or Red/Red? ____

Q15. What is the ratio between them [Red/Green + Red/Red] : [Green/Green]? ____ to 1.

Mendel's Observations

One wonders what might have happened had Mendel given three lectures on the subject of heredity, instead of two, with the first paper containing the elementary lesson in statistics you have just struggled through. But let's leave these idle speculations to historians, and see if we can make heads or tails of Mendel's observations.



Mendel's experimental plot, the way it appeared in 2005 (Mendel's statue is in the background, to the left)

In his day, gardeners could obtain all kinds of true-breeding pea varieties from commercial seed houses. For example, one variety was guaranteed to give only tall plants (6 ft or so); another, only short plants (about 1 ft in height). If you crossed one tall plant to itself or to another tall plant,

collected the resultant seeds some three months later, planted them, and observed the height of this second-generation of plants, all would be tall. Similarly, only short plants would result from a cross between true-breeding short peas. This would continue generation after generation. One stock would only give rise to tall plants; the other only to short plants. (Incidentally, in humans, freckle-lessness is more or less true-breeding--without knowing any of the freckle-less couples you are familiar with, a geneticist can be fairly certain that their children are freckle-less too).

Mendel then crossed the two varieties.

Q16. Before we continue, can you guess the height of the offspring? The cross Short X Short, we have seen, yields short plants. Tall X Tall yields only Tall. But how tall would the descendants of a Tall X Short cross be?

The surprising answer is that, in this case, *all* the first generation plants were tall.

Q17. Mendel then crossed these new tall plants (the offspring of the Tall X Short cross) to themselves or to each other. Can you guess the outcome?

The actual results from this cross were: 787 plants among the second generation ("grandchildren" of the original true-breeding tall/true-breeding short "couple") were tall, and 277 were short. Note that the short characteristic which disappeared from sight in the first generation, reappeared in the second. This means that although the factor which caused short stature was temporarily out of sight, it was still there. Note also that the ratio between tall and short plants was 787/277, or 2.84 to 1 (approximately 3 to 1).

Mendel obtained similar results for many other characteristics, suggesting that a general rule is at work here. For instance, in Mendel's time one could order dry pea seeds with either smooth or wrinkled surfaces. Again, Mendel established that neither type was true-breeding. When plants arising from smooth seeds were crossed to plants arising from wrinkled seeds, all their offspring produced smooth seeds. In the next generation, 5,474 of the plants produced smooth seeds and 1,850 wrinkled seeds, yielding a 2.96 to 1 ratio (an almost exact 3 to 1 ratio).

These are some of the observations Mendel recounted to his audience on that cold February evening of 1865, in the little Moravian (a region of the Czech Republic) town of Brno. A month later he reappeared before them and put forward an elegant explanation for his results.

Q18. Imagine you found yourself in Mendel's shoes. What would you tell your audience?

Mendel deductions are by no means confined to peas. Owing to the close affinity between them, living creatures often obey the same rules. When crossed to fruit flies of the same strain, red-eyed individuals produce only red-eyed offspring while brown-eyed individuals produce only brown-eyed offspring. In one experiment, all the progeny of a cross between a red-eyed male and a brown-eyed female had red eyes. When these sisters and brothers were mated to each other, they produced 308 red-eyed and 93 brown-eyed offspring.

Q19. Please use the insights you gained from this exercise to explain these observations.

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