

CLASSICAL GENETICS (KHAN ACADEMY)



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1.1: Introduction to heredity



Heredity and Classical Genetics. Dominant and recessive traits. Heterozygous and homozygous genotypes. Created by Sal Khan.

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1.2: Alleles and genes



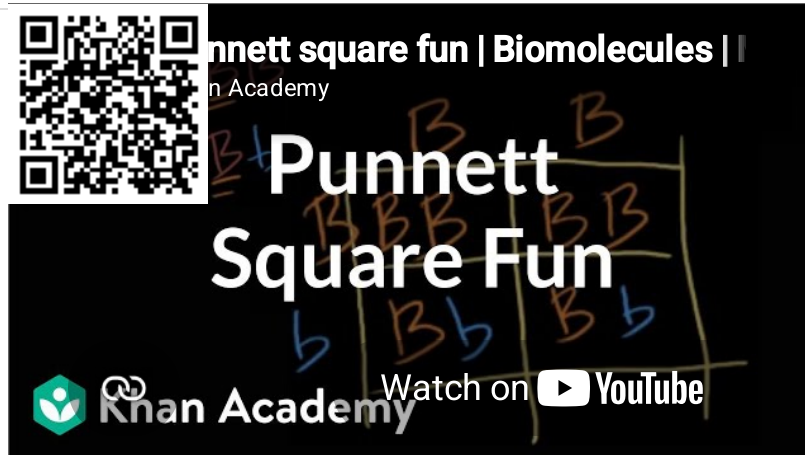
A gene as a stretch of DNA on a chromosome. Alleles as versions (sequence variants) of a gene.

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1.3: Worked example - Punnett squares



Learn how to use Punnett squares to calculate probabilities of different phenotypes. Includes worked examples of dihybrid crosses, independent assortment, incomplete dominance, codominance, and multiple alleles. Created by Sal Khan.

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1.4: Mendel and his peas

How can we study inheritance?

When spending time with your own family, friends, and neighbors, you may have noticed that many traits run in families. For instance, members of a family may share similar facial features, hair color (like the brother and sister below), or a predisposition to health problems such as diabetes. Characteristics that run in families often have a **genetic basis**, meaning that they depend on genetic information a person inherits from his or her parents.



Image credit "[Brother, sister, portrait, russet,](#)" by Adina Voicu (CC0, public domain).

What if you wanted to figure out how genetic information is transmitted between generations? For instance, you might be curious how traits can "skip" a generation, or why one child in a family may suffer from a genetic disease while another does not. How could you go about asking these kinds of questions scientifically?

An obvious first idea would be to study human inheritance patterns directly, but that turns out to be a tricky proposition (see the pop-up below for details). In this article, we'll see how a nineteenth-century monk named Gregor Mendel instead uncovered the key principles of inheritance using a simple, familiar system: the pea plant.

[Why didn't Mendel study humans?]

Many of us are curious about genetics because we want to understand human inheritance. If the burning questions of genetics are about humans, why would anyone study inheritance in something like the pea plant?

When it comes to basic principles of inheritance, humans may be the organisms we most want to learn about, but they aren't always the best organisms to study experimentally. For instance, it takes many years for a human being to grow, mature, and have children, making data collection slow. Also, when humans have children, they often have one or two (rather than, say, several thousand), making it harder to see mathematical patterns in the data. Finally, and perhaps most critically, it wouldn't be

possible (or ethical) to set up controlled experiments in human genetics – that is, you couldn't ask a pair of people to have children just because you were curious what those children would look like.

Peas, on the other hand, grow quickly, make many seeds, and can be bred to one another in a simple, controlled way. In other words, they're good organisms to study in many ways that humans aren't. Using pea plants, Mendel was able to uncover fundamental principles of inheritance that apply to many different kinds of organisms. Although these principles would have been nearly impossible to deduce from human family trees alone, they form the core of the modern field of human genetics. Mendel's laws, and later findings building on those laws, allow us to understand and predict the inheritance of many human traits, including genetic disorders.

The monk in the garden: Gregor Mendel

Johann Gregor Mendel (1822–1884), often called the “father of genetics,” was a teacher, lifelong learner, scientist, and man of faith. It would be fair to say that Mendel had a lot of grit: he persevered through difficult circumstances to make some of the most important discoveries in biology.



Image credit: "Mendel's experiments and the laws of probability: Figure 1," by OpenStax College, Biology (CC BY 3.0).

As a young man, Mendel had difficulty paying for his education due to his family's limited means, and he also suffered bouts of physical illness and depression; still, he persevered to graduate from high school and, later, university¹. After finishing university, he joined the Augustinian Abbey of St. Thomas in Brno, in what is now the Czech Republic. At the time, the monastery was the cultural and intellectual hub of the region, and Mendel was immediately exposed to new teachings and ideas¹.

His decision to join the order (against the wishes of his father, who expected him to carry on the family farm) appears to have been motivated in part by a desire to continue his education and pursue his scientific interests². Supported by the monastery, he taught physics, botany, and natural science courses at the secondary and university levels.

Research on heredity

In 1856, Mendel began a decade-long research project to investigate patterns of inheritance. Although he began his research using mice, he later switched to honeybees and plants, ultimately settling on garden peas as his primary model system². A **model system** is an organism that makes it easy for a researcher to investigate a particular scientific question, such as how traits are inherited. By studying a model system, researchers can learn general principles that apply to other, harder-to-study organisms or biological systems, such as humans.

Mendel studied the inheritance of seven different features in peas, including height, flower color, seed color, and seed shape. To do so, he first established pea lines with two different forms of a feature, such as tall vs. short height. He grew these lines for

generations until they were pure-breeding (always produced offspring identical to the parent), then bred them to each other and observed how the traits were inherited.

In addition to recording how the plants in each generation looked, Mendel counted the exact number of plants that showed each trait. Strikingly, he found very similar patterns of inheritance for all seven features he studied:

- One form of a feature, such as tall, always concealed the other form, such as short, in the first generation after the cross. Mendel called the visible form the **dominant trait** and the hidden form the **recessive trait**.
- In the second generation, after plants were allowed to self-fertilize (pollinate themselves), the hidden form of the trait reappeared in a minority of the plants. Specifically, there were always about 3 plants that showed the dominant trait (e.g., tall) for every 1 plant that showed the recessive trait (e.g., short), making a 3:1 ratio.
- Mendel also found that the features were inherited independently: one feature, such as plant height, did not influence inheritance of other features, such as flower color or seed shape.

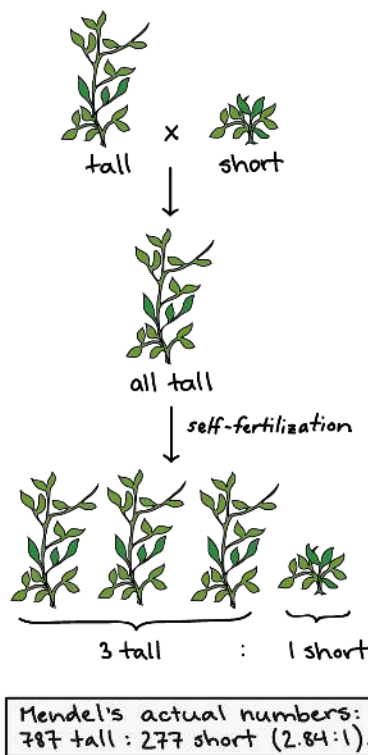


Image modified from "[Mendel seven characters](#)," by Mariana Ruiz Villareal (public domain).

In 1865, Mendel presented the results of his experiments with nearly 30,000 pea plants to the local Natural History Society. Based on the patterns he observed, the counting data he collected, and a mathematical analysis of his results, Mendel proposed a model of inheritance in which:

- Characteristics such as flower color, plant height, and seed shape were controlled by pairs of heritable factors that came in different versions.
- One version of a factor (the dominant form) could mask the presence of another version (the recessive form).
- The two paired factors separated during gamete production, such that each gamete (sperm or egg) randomly received just one factor.
- The factors controlling different characteristics were inherited independently of one another.

We'll take a closer look at how Mendel reached these conclusions in the articles on the [law of segregation](#) and the [law of independent assortment](#). In 1866, Mendel published his observations and his model of inheritance, under the title *Experiments in Plant Hybridization*^{3,4}, in the Proceedings of the Natural History Society of Brünn.

Scientific legacy

Mendel's work went largely unnoticed by the scientific community during his lifetime. How could this have been the case?

In part, Mendel's contemporaries failed to recognize the importance of his work because his findings went against prevailing (popular) ideas about inheritance. In addition, although we now see Mendel's mathematical approach to biology as innovative and pioneering, it was new, unfamiliar, and perhaps confusing or unintuitive to other biologists of the time⁵.

In the mid-1800s, when Mendel was doing his experiments, most biologists subscribed to the idea of **blending inheritance**. Blending inheritance wasn't a formal, scientific hypothesis, but rather, a general model in which inheritance involved the permanent blending of parents' characteristics in their offspring (producing offspring with an intermediate form of a characteristic)⁶. The blending model fit well with some observations of human inheritance: for instance, children often look a bit like both of their parents.

But the blending model could not explain why Mendel crossed a tall and a short pea plant and got only tall plants, or why self-fertilization of one of those tall plants would produce a 3:1 ratio of tall to short plants in the next generation. Instead, if the blending model were correct, a tall plant crossed with a short plant should produce a medium plant, which would go on to produce more medium plants (see below).

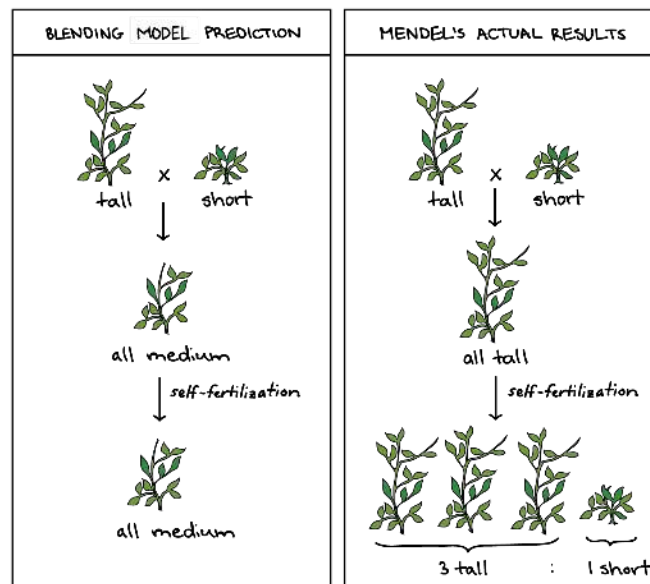


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As it turns out, both pea plant height and human height (along with many other characteristics in a wide range of organisms) are controlled by pairs of heritable factors that come in distinctive versions, just as Mendel proposed. In humans, however, there are many different factors (genes) that contribute fractionally to height and vary among individuals. This makes it difficult to see the contribution of any one factor and produces inheritance patterns that can resemble blending. In Mendel's experiments, in contrast, there was just one factor that differed between the tall and short pea plants, allowing Mendel to clearly see the underlying pattern of inheritance.

In 1868, Mendel became abbot of his monastery and largely set aside his scientific pursuits in favor of his pastoral duties. He was not recognized for his extraordinary scientific contributions during his lifetime. In fact, it was not until around 1900 that his work was rediscovered, reproduced, and revitalized. Its rediscoverers were biologists on the brink of discovering the chromosomal basis of heredity – that is, about to realize that Mendel's "heritable factors" were carried on chromosomes.

Mendel's model system: The pea plant

Mendel carried out his key experiments using the garden pea, *Pisum sativum*, as a model system. Pea plants make a convenient system for studies of inheritance, and they are still studied by some geneticists today.

Useful features of peas include their rapid life cycle and the production of lots and lots of seeds. Pea plants also typically self-fertilize, meaning that the same plant makes both the sperm and the egg that come together in fertilization. Mendel took advantage of this property to produce **true-breeding** pea lines: he self-fertilized and selected peas for many generations until he got lines that consistently made offspring identical to the parent (e.g., always short).

Pea plants are also easy to cross, or mate in a controlled way. This is done by transferring pollen from the anthers (male parts) of a pea plant of one variety to the carpel (female part) of a mature pea plant of a different variety. To prevent the receiving plant from self-fertilizing, Mendel painstakingly removed all of the immature anthers from the plant's flowers before the cross.

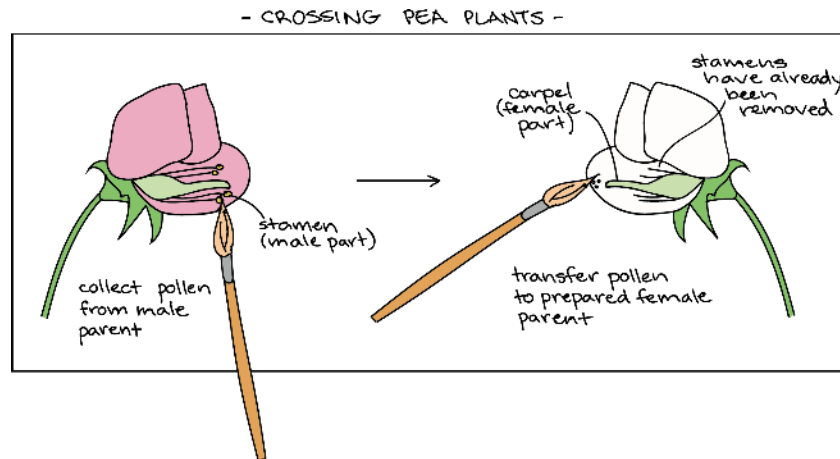


Image based on similar illustration from Reece et al.⁷

Because peas were so easy to work with and prolific in seed production, Mendel could perform many crosses and examine many individual plants, making sure that his results were consistent (not just a fluke) and accurate (based on many data points).

Mendel's experimental setup

Once Mendel had established true-breeding pea lines with different traits for one or more features of interest (such as tall vs. short height), he began to investigate how the traits were inherited by carrying out a series of crosses.

First, he crossed one true-breeding parent to another. The plants used in this initial cross are called the **P generation**, or parental generation.

Mendel collected the seeds from the P generation cross and grew them up. These offspring were called the **F₁ generation**, short for first filial generation. (*Filius* means “son” in Latin, so this name is slightly less weird than it seems!)

Once Mendel examined the F₁ plants and recorded their traits, he let them self-fertilize naturally, producing lots of seeds. He then collected and grew the seeds from the F₁ plants to produce an **F₂ generation**, or second filial generation. Again, he carefully examined the plants and recorded their traits.

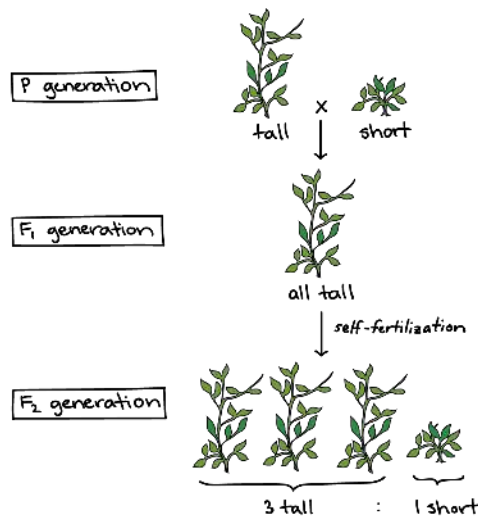


Image modified from "Mendel seven characters," by Mariana Ruiz Villareal (public domain).

Mendel's experiments extended beyond the F₂ generation to F₃, F₄, and later generations, but his model of inheritance was based mostly on the first three generations (P, F₁, and F₂).

Mendel didn't just record what his plants looked like in each generation (e.g., tall vs. short). Instead, he counted exactly how many plants with each trait were present. This may sound tedious, but by recording numbers and thinking mathematically, Mendel made discoveries that eluded famous scientists of his time (such as Charles Darwin, who carried out similar experiments but didn't grasp the significance of his results)⁵.

You can use the links below to learn more about Mendel's laws of inheritance:

- The [law of segregation](#), describing how individual traits are inherited.
- The [law of independent assortment](#), describing how two or more traits are inherited relative to one another.

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1.5: The law of segregation

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 **Punnett 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 color 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 color, flower position, seed color, 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?]

In self-fertilization, sperm and eggs from the same pea plant combine inside a closed flower, producing seeds with a single plant as both mother and father.

What results did Mendel find in his crosses for flower color? 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 F₁ 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 F₁ generation (violet flowers) the **dominant trait**, and the trait that was hidden or lost (white flowers) the **recessive trait**.

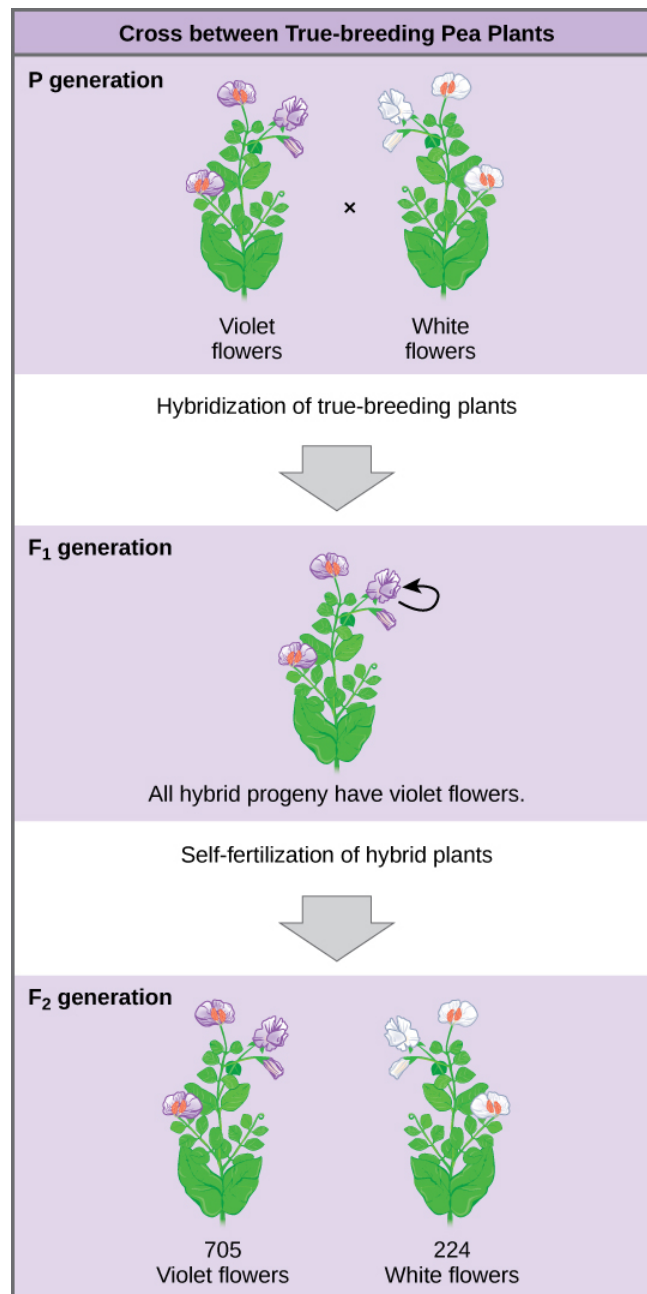


Image credit: "Mendel's experiments: Figure 2," by Robert Bear et al., OpenStax, [CC BY 4.0](https://creativecommons.org/licenses/by/4.0/)

Importantly, Mendel did not stop his experimentation there. Instead, he let the F₁ plants self-fertilize. Among their offspring, called the F₂ 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 3:1 ratio was no fluke. For the other six characteristics that Mendel examined, both the F₁ and F₂ generations behaved in the same way they did for flower color. One of the two traits would disappear completely from the F₁ generation, only to reappear in the F₂ generation in a ratio of roughly 3:1.

[See Mendel's data for all seven characteristics]

Character	Dominant trait	Recessive trait	F ₂ generation	F ₂ ratio
Seed color	Yellow	Green	6022 yellow, 2001 green	3.01:1
Seed shape	Round	Wrinkled	5474 round, 1850 wrinkled	2.96:1
Pod color	Green	Yellow	428 green, 152 yellow	2.82:1
Pod shape	Inflated	Constricted	882 inflated, 299 constricted	2.95:1
Plant height	Tall	Short (dwarf)	787 tall, 277 short	2.84:1
Flower color	Purple	White	705 purple, 224 white	3.15:1
Flower position	Axial (along branch)	Terminal (end of branch)	651 axial, 207 terminal	3.14:1

Adapted from "[The results of Mendel's garden pea hybridizations](#)," by OpenStax College, Biology (CC BY 3.0).

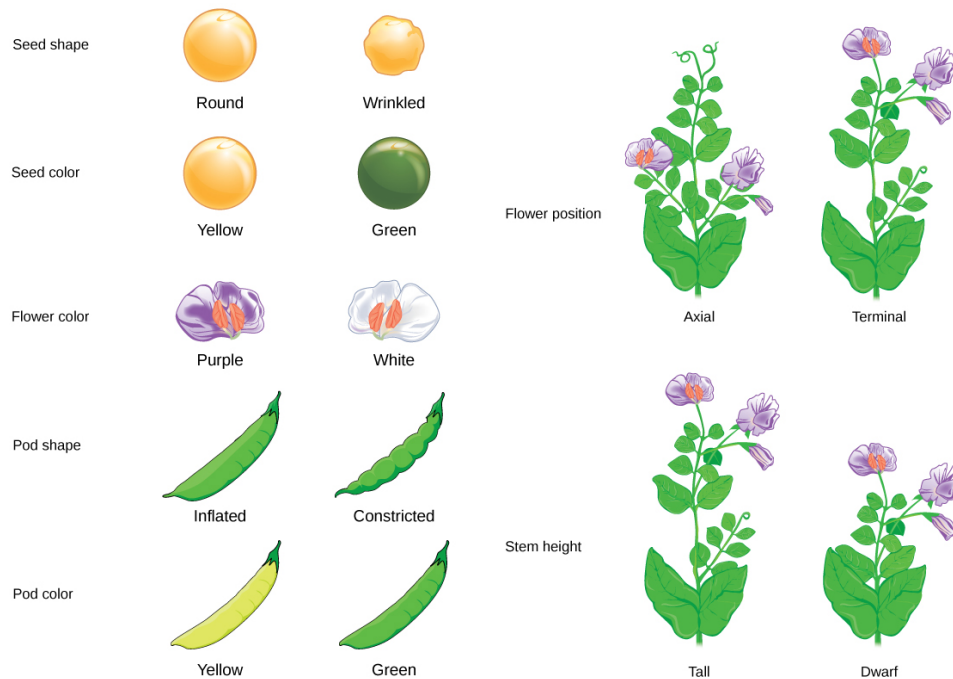


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As it turned out, the 3:1 ratio was a crucial clue that let Mendel crack the puzzle of inheritance. Let's take a closer look at what Mendel figured out.

Mendel's model of inheritance

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

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 color (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 color, the dominant yellow allele Y hides the recessive green allele y.

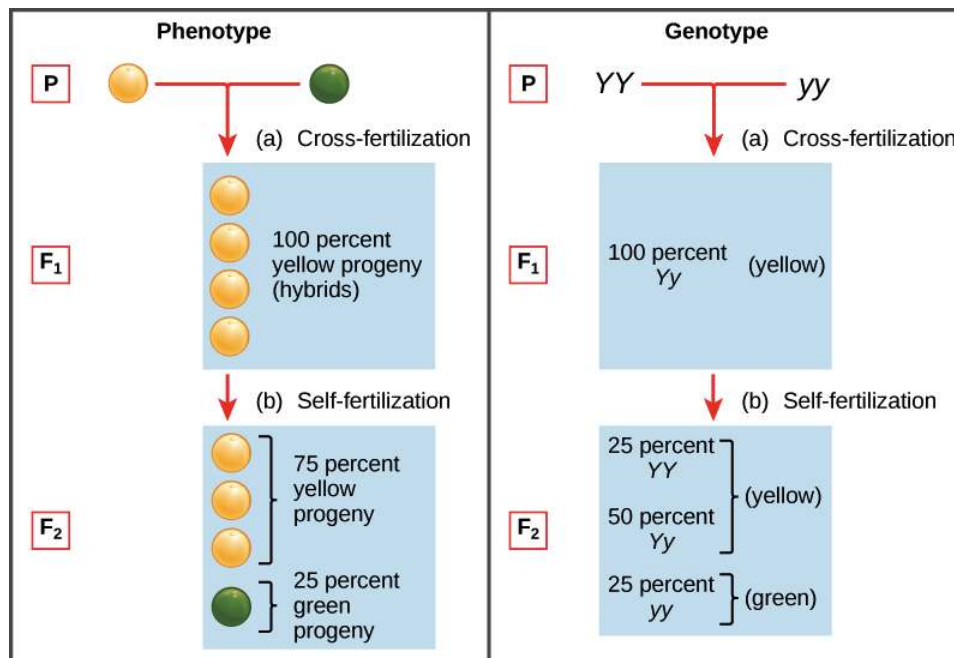


Image modified from "Laws of inheritance: Figure 1," by Robert Bear et al., OpenStax, CC BY 4.0

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 Yy), 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 3:1 ratio. 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:

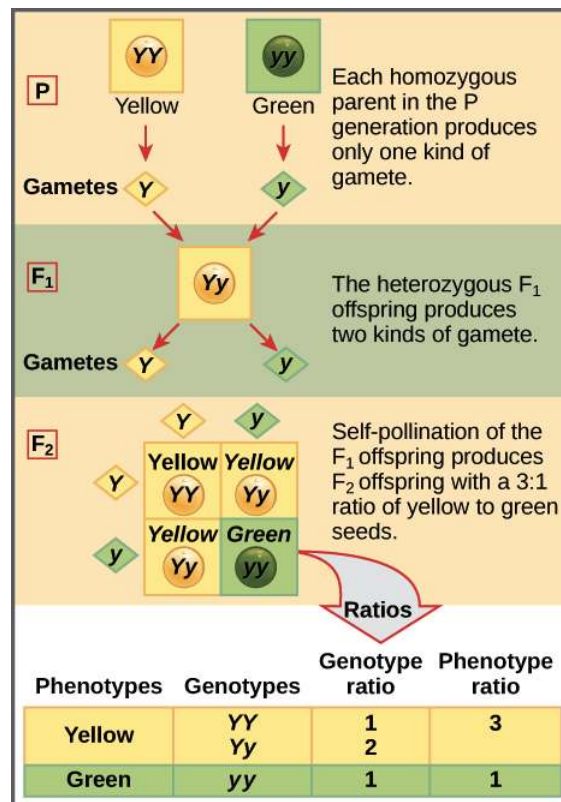


Image modified from "Laws of inheritance: Figure 5," by Robert Bear et al., OpenStax, CC BY 4.0

The four-squared box shown for the F_2 generation is known as a **Punnett square**. To prepare a Punnett 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.

[Why are the boxes all equally likely?]

A key point of the law of segregation is that a parent's two gene copies are randomly distributed to its gametes. Thus, for a Yy heterozygote, Y and y gametes are equally likely to be made: 50% of the sperm and eggs will have a Y allele, 50% will have a y allele, and the same will be true for eggs.

Since each type of gamete is equally common, each fertilization event (meeting of gametes, corresponding to a square of the table) also has an equal chance of happening. Thus, the boxes of the table represent four equal-probability events.

Since the table contains 1 box with a YY genotype, 2 boxes with a Yy genotype, and 1 box with a yy genotype, we'd expect to see YY , Yy , and yy plants in a ratio of 1:2:1 in the F_2 generation. Since both YY and Yy plants are yellow, this genotype ratio translates into a phenotypic ratio of 3:1 yellow-seeded to green-seeded plants, almost exactly what Mendel observed.

YY and yy plants each appear in just one square, while Yy plants are found in two squares. This is because there are two different fertilization events that lead to a Yy plant: the fusion of a Y egg and a y sperm, or the fusion of a y egg and a Y sperm. Either event is equally likely, and has the same likelihood as a YY or yy fertilization event.

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 (Yy) 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):

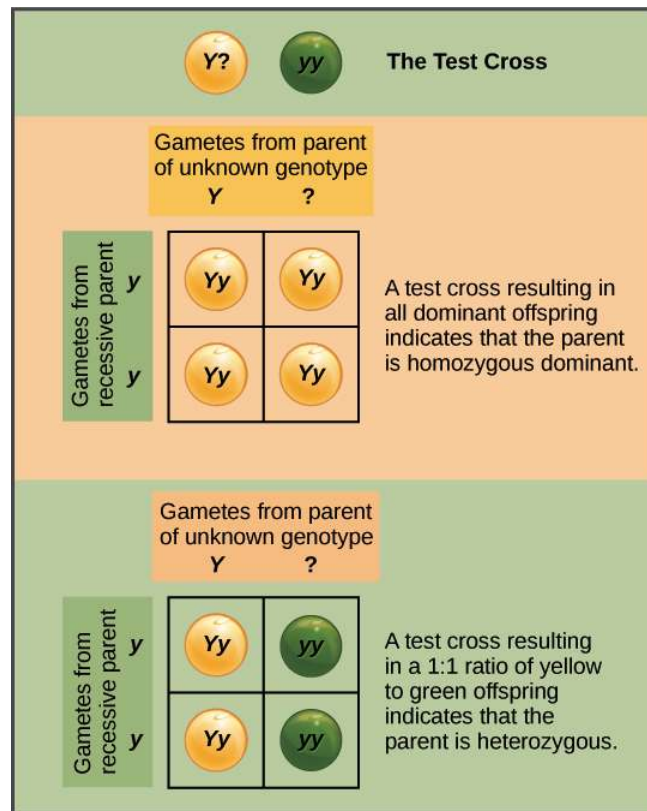


Image credit: "Laws of inheritance: Figure 4," by Robert Bear et al., OpenStax, CC BY 4.0

If the organism with the dominant phenotype is homozygous, then all of the F_1 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_1 offspring will be half heterozygotes (dominant phenotype) and half recessive homozygotes (recessive phenotype).

The fact that we get a 1:1 ratio 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 color 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 3:1 ratio). These are some of the qualities of a great scientist—ones that anyone, anywhere, can develop!

Check your understanding

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1.6: The law of independent assortment

Introduction

The [law of segregation](#) lets us predict how a single feature associated with a single gene is inherited. In some cases, though, we might want to predict the inheritance of two characteristics associated with two different genes. How can we do this?

[Refresher on the law of segregation]

The **law of segregation** states that each gamete (sperm or egg cell) made by an organism will get just one of the two gene copies present in a parent organism, and that the gene copies are randomly allocated to the gametes. For instance, if an organism has a genotype of Aa , half of its gametes will contain an A allele, and the other half will contain an a allele.

You can use the link at the start of the paragraph to learn more about the law of segregation.

To make an accurate prediction, we need to know whether the two genes are inherited independently or not. That is, we need to know whether they "ignore" one another when they're sorted into gametes, or whether they "stick together" and get inherited as a unit.

When [Gregor Mendel](#) asked this question, he found that different genes were inherited independently of one another, following what's called the **law of independent assortment**. In this article, we'll take a closer look at the law of independent assortment and how it is used to make predictions. We'll also see when and why the law of independent assortment does (or doesn't!) hold true.

Note: If you are not yet familiar with how individual genes are inherited, you may want to check out the article on the [law of segregation](#) or the [introduction to heredity](#) video before you dive into this article.

What is the law of independent assortment?

Mendel's **law of independent assortment** states that the alleles of two (or more) different genes get sorted into gametes independently of one another. In other words, the allele a gamete receives for one gene does not influence the allele received for another gene.

Example: Pea color and pea shape genes

Let's look at a concrete example of the law of independent assortment. Imagine that we cross two pure-breeding pea plants: one with yellow, round seeds ($YYRR$) and one with green, wrinkled seeds ($yyrr$). Because each parent is homozygous, the law of segregation tells us that the gametes made by the wrinkled, green plant all are ry , and the gametes made by the round, yellow plant are all RY . That gives us F_1 offspring that are all $RrYy$.

The allele specifying yellow seed color is dominant to the allele specifying green seed color, and the allele specifying round shape is dominant to the allele specifying wrinkled shape, as shown by the capital and lower-case letters. This means that the F_1 plants are all yellow and round. Because they are heterozygous for two genes, the F_1 plants are called **dihybrids** (*di-* = two, *-hybrid* = heterozygous).

A cross between two dihybrids (or, equivalently, self-fertilization of a dihybrid) is known as a **dihybrid cross**. When Mendel did this cross and looked at the offspring, he found that there were four different categories of pea seeds: yellow and round, yellow and wrinkled, green and round, and green and wrinkled. These **phenotypic** categories (categories defined by observable traits) appeared in a ratio of approximately 9:3:3:1.

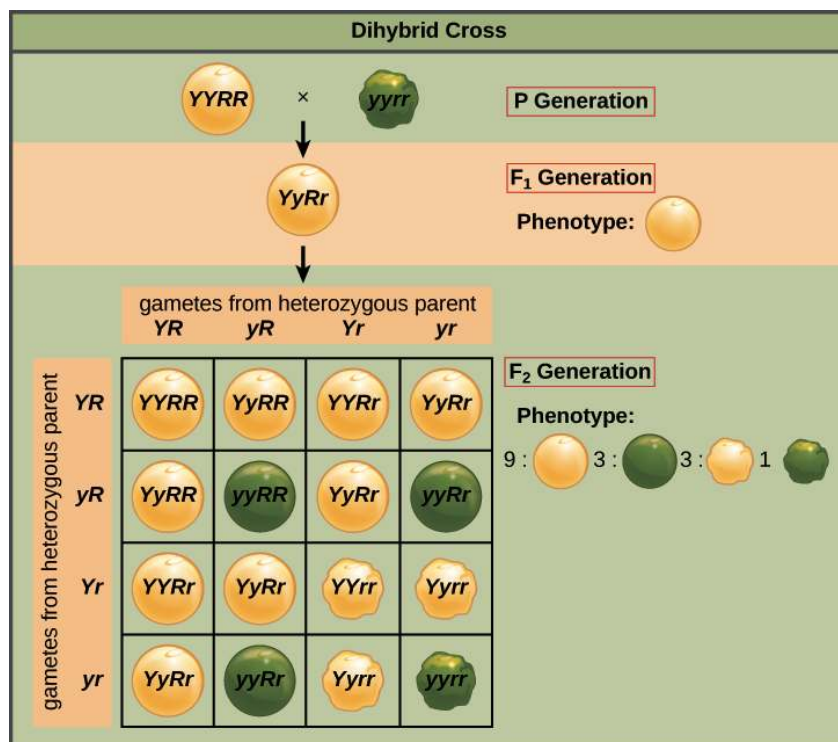


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This ratio was the key clue that led Mendel to the law of independent assortment. That's because a 9:3:3:1 ratio is exactly what we'd expect to see if the F₁ plant made four types of gametes (sperm and eggs) with equal frequency: YR, Yr, yR, and yr. In other words, this is the result we'd predict if each gamete randomly got a Y or y allele, and, in a separate process, also randomly got an R or r allele (making four equally probable combinations).

We can confirm the link between the four types of gametes and the 9:3:3:1 ratio using the Punnett square above. To make the square, we first put the four equally probable gamete types along each axis. Then, we join gametes on the axes in the boxes of the chart, representing fertilization events. The 16 equal-probability fertilization events that can occur among the gametes are shown in the 16 boxes. The offspring genotypes in the boxes correspond to a 9:3:3:1 ratio of phenotypes, just as Mendel observed.

[More about two-gene Punnett squares]

We can draw a Punnett square for a two-gene scenario by following the same basic rules as for a monohybrid cross, placing the gametes along the axes and combining them in the squares to represent fertilization events. However, since there are now more gamete types, there must also be more squares in the table: 4 possible types of maternal gametes x 4 possible types of paternal gametes = 16 squares total.

As with a [single-gene Punnett square](#), we place all the possible types of gametes along the axes, then combine them in the squares where the columns and rows intersect to represent fertilization events (the formation of zygotes, or offspring).

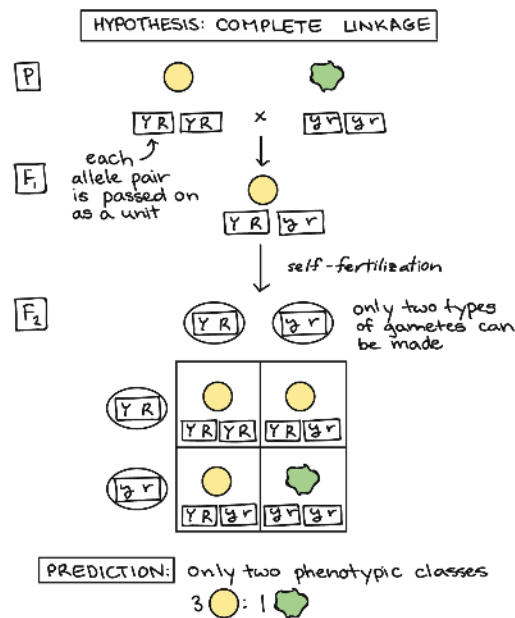
To learn how you can use the rules of probability to predict the outcome of a dihybrid cross, see the [probabilities in genetics](#) article.

Independent assortment vs. linkage

The section above gives us Mendel's law of independent assortment in a nutshell, and lets us see how the law of independent assortment leads to a 9:3:3:1 ratio. But what was the alternative possibility? That is, what would happen if two genes *didn't* follow independent assortment?

In the extreme case, the genes for seed color and seed shape might have always been inherited as a pair. That is, the yellow and round alleles might always have stayed together, and so might the green and wrinkled alleles.

To see how this could work, imagine that the color and shape genes are physically stuck together and cannot be separated, as represented by the boxes around the alleles in the diagram below. For instance, this could happen if the two genes were located very, very close together on a chromosome (an idea we'll explore further at the end of the article).



Rather than giving a color allele and, separately, giving a shape allele to each gamete, the F₁ dihybrid plant would simply give one “combo unit” to each gamete: a YR allele pair or a yr allele pair.

We can use a Punnett square to predict the results of self-fertilization in this case, as shown above. If the seed color and seed shape genes were in fact always inherited as a unit, or **completely linked**, a dihybrid cross should produce just two types of offspring, yellow/round and green/wrinkled, in a 3:1 ratio. Mendel's actual results were quite different from this (the 9:3:3:1 ratio we saw earlier), telling him that the genes assorted independently.

The reason for independent assortment

To see why independent assortment happens, we need to fast-forward half a century and discover that genes are physically located on chromosomes. To be exact, the two copies of a gene carried by an organism (such as a Y and a y allele) are located at the same spot on the two chromosomes of a **homologous pair**. Homologous chromosomes are similar but non-identical, and an organism gets one member of the pair from each of its two parents.

The physical basis for the law of independent assortment lies in meiosis I of gamete formation, when homologous pairs line up in random orientations at the middle of the cell as they prepare to separate. We can get gametes with different combos of “mom” and “dad” homologues (and thus, the alleles on those homologues) because the orientation of each pair is random.

To see what this means, compare chromosome arrangement 1 (top) and chromosome arrangement 2 (bottom) at the stage of metaphase I in the diagram below. In one case, the red “mom” chromosomes go together, while in the other, they split up and mix with the blue “dad” chromosomes. If meiosis happens many times, as it does in a pea plant, we will get both arrangements—and thus RY, Ry, rY, and ry classes of gametes—with equal frequency.

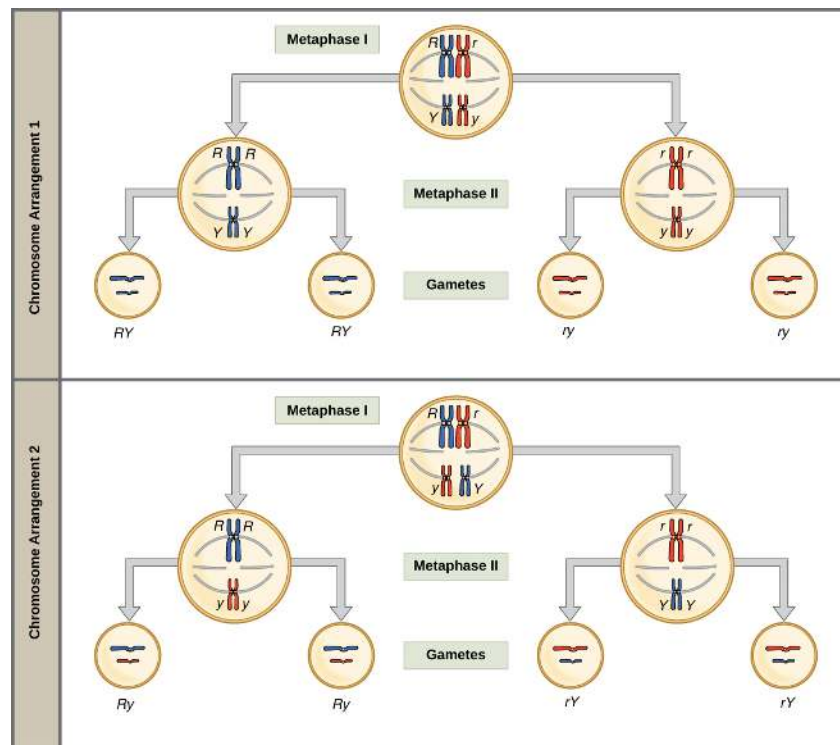
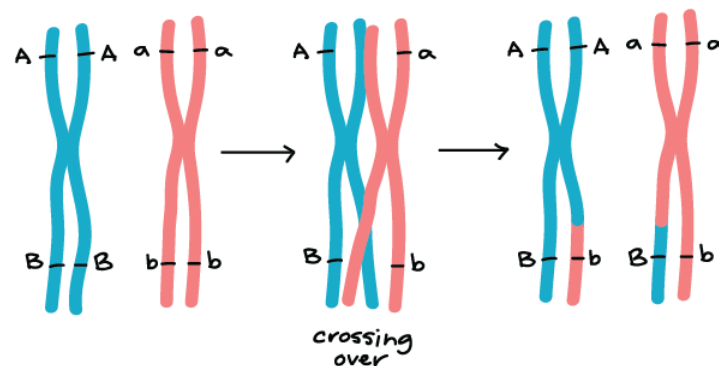


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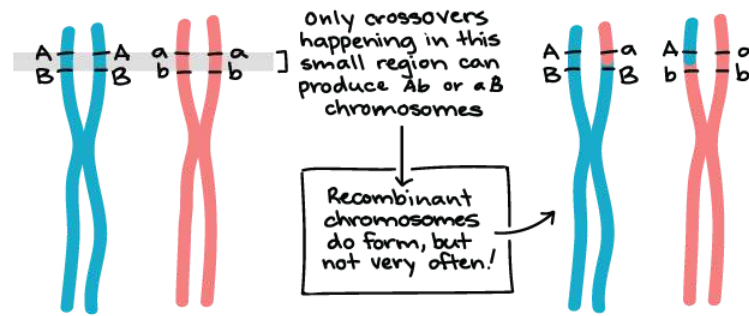
Genes that are on different chromosomes (like the Y and R genes) assort independently. The seed color and seed shape genes are on chromosomes 1 and 7 of the pea genome, respectively, in real life¹. Genes that are far apart on the same chromosome also assort independently thanks to the crossing over, or exchange of homologous chromosome bits, that occurs early in meiosis I.

[See a picture]



There are, however, gene pairs that do not assort independently. When genes are close together on a chromosome, the alleles on the same chromosome tend to be inherited as a unit more frequently than not. Such genes do not display independent assortment and are said to be **linked**. We'll take a closer look at genetic linkage in other articles and videos.

[See a picture]



Check your understanding

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1.7: Probabilities in genetics

Introduction

The [Punnett square](#) is a valuable tool, but it's not ideal for every genetics problem. For instance, suppose you were asked to calculate the frequency of the recessive class not for an $Aa \times Aa$ cross, not for an $AaBb \times AaBb$ cross, but for an $AaBbCcDdEe \times AaBbCcDdEe$ cross. If you wanted to solve that question using a Punnett square, you could do it – but you'd need to complete a Punnett square with 1024 boxes. Probably not what you want to draw during an exam, or any other time, if you can help it!

The five-gene problem above becomes less intimidating once you realize that a Punnett square is just a visual way of representing probability calculations. Although it's a great tool when you're working with one or two genes, it can become slow and cumbersome as the number goes up. At some point, it becomes quicker (and less error-prone) to simply do the probability calculations by themselves, without the visual representation of a clunky Punnett square. In all cases, the calculations and the square provide the same information, but by having both tools in your belt, you can be prepared to handle a wider range of problems in a more efficient way.

In this article, we'll review some probability basics, including how to calculate the probability of two independent events both occurring (event X and event Y) or the probability of either of two mutually exclusive events occurring (event X or event Y). We'll then see how these calculations can be applied to genetics problems, and, in particular, how they can help you solve problems involving relatively large numbers of genes.

[Solution to the five-gene cross problem]

In this problem, we're supposed to find the frequency of the recessive class among the offspring of an $AaBbCcDdEe \times AaBbCcDdEe$ cross – that is, the frequency of $aabbccdde$ individuals. How do we get an $aabbccdde$ individual? There's only one way for that to happen: both parents must contribute an $abcde$ gamete.

What, then, is the probability that one of the parents will make an $abcde$ gamete? Both parents are heterozygous for all five genes, so there's a $1/2$ chance of getting the recessive (lowercase) allele for any one gene. To get our desired gamete, we need all five genes in recessive form (a and b and c and d and e). This is a case where we can apply the **product rule**, which states that the probability of event X and event Y happening is the product of their individual probabilities (probability of X times probability of Y), assuming that X and Y are independent events. Thus, the overall probability of one parent producing an $abcde$ gamete is:

Probability of $abcde$ gamete = (probability of a) $\times$ (probability of b) $\times$ (probability of c) $\times$ (probability of d) $\times$ (probability of e)

$$P(abcde) = P(a) \cdot P(b) \cdot P(c) \cdot P(d) \cdot P(e)$$

$$P(abcde) = (1/2) \cdot (1/2) \cdot (1/2) \cdot (1/2) \cdot (1/2) = (1/2)^5 = 1/32$$

If that's the probability of one parent making an $abcde$ gamete, what's the likelihood of both parents doing so? Again, we can apply the "and" rule (product rule), since we need both parent 1 and parent 2 to make an $abcde$ gamete in order to get our target recessive homozygote. Thus, the overall probability is:

Probability of $aabbccdde$ individual = (probability of parent 1 making an $abcde$ gamete) $\times$ (probability of parent 2 making an $abcde$ gamete)

$$P(aabbccdde) = P(abcde_{\text{parent A}}) \cdot P(abcde_{\text{parent B}})$$

$$P(aabbccdde) = (1/32) \cdot (1/32) = 1/1024$$

That's our overall probability for a recessive homozygote for all five genes.

The $1/1024$ probability corresponds to 1 box out of the 1024 boxes of the Punnett square you'd have to draw to represent this cross. The probability calculation is the same calculation we'd implicitly do by drawing the Punnett square, just faster and with fewer chances for mistakes.

Probability basics

Probabilities are mathematical measures of likelihood. In other words, they're a way of quantifying (giving a specific, numerical value to) how likely something is to happen. A probability of 1 for an event means that it is guaranteed to happen, while a probability of 0 for an event means that it is guaranteed not to happen. A simple example of probability is having a $1/2$ chance of getting heads when you flip a coin, as Sal explains in this [intro to probability](#) video.

Probabilities can be either empirical, meaning that they are calculated from real-life observations, or theoretical, meaning that they are predicted using a set of rules or assumptions.

- The **empirical probability** of an event is calculated by counting the number of times that event occurs and dividing it by the total number of times that event could have occurred. For instance, if the event you were looking for was a wrinkled pea seed, and you saw it 1,850 times out of the 7,324 total seeds you examined, the empirical probability of getting a wrinkled seed would be $1,850/7,324 = 0.253$, or very close to 1 in 4 seeds.
- The **theoretical probability** of an event is calculated based on information about the rules and circumstances that produce the event. It reflects the number of times an event is *expected* to occur relative to the number of times it could possibly occur. For instance, if you had a pea plant heterozygous for a seed shape gene (Rr) and let it self-fertilize, you could use the rules of probability and your knowledge of genetics to predict that 1 out of every 4 offspring would get two recessive alleles (rr) and appear wrinkled, corresponding to a 0.25 ($1/4$) probability. We'll talk more below about how to apply the rules of probability in this case.

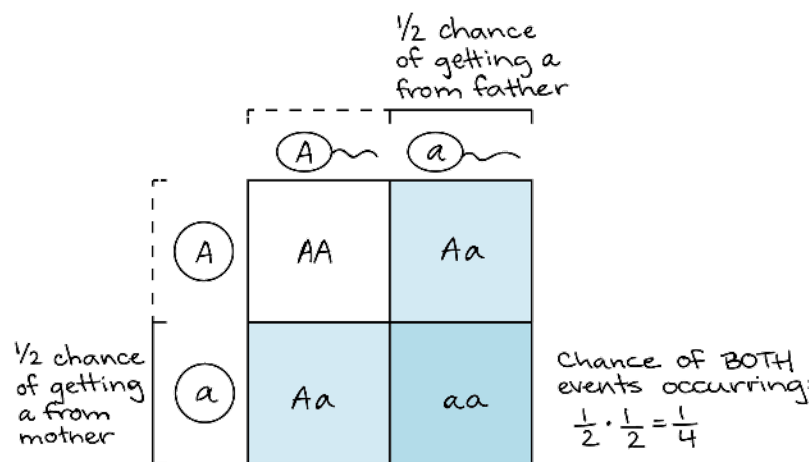
In general, the larger the number of data points that are used to calculate an empirical probability, such as shapes of individual pea seeds, the more closely it will approach the theoretical probability.

The product rule

One probability rule that's very useful in genetics is the **product rule**, which states that the probability of two (or more) independent events occurring together can be calculated by multiplying the individual probabilities of the events. For example, if you roll a six-sided die once, you have a $1/6$ chance of getting a six. If you roll two dice at once, your chance of getting two sixes is: (probability of a six on die 1) $\times$ (probability of a six on die 2) = $(1/6) \cdot (1/6) = 1/36$.

In general, you can think of the product rule as the "and" rule: if both event X *and* event Y must happen in order for a certain outcome to occur, and if X and Y are independent of each other (don't affect each other's likelihood), then you can use the product rule to calculate the probability of the outcome by multiplying the probabilities of X and Y.

We can use the product rule to predict frequencies of fertilization events. For instance, consider a cross between two heterozygous (Aa) individuals. What are the odds of getting an aa individual in the next generation? The only way to get an aa individual is if the mother contributes an a gamete and the father contributes an a gamete. Each parent has a $1/2$ chance of making an a gamete. Thus, the chance of an aa offspring is: (probability of mother contributing a) $\times$ (probability of father contributing a) = $(1/2) \cdot (1/2) = 1/4$.



This is the same result you'd get with a Punnett square, and actually the same logical process as well—something that took me years to realize! The only difference is that, in the Punnett square, we'd do the calculation visually: we'd represent the $1/2$ probability of an a gamete from each parent as one out of two columns (for the father) and one out of two rows (for the mother). The 1-square intersect of the column and row (out of the 4 total squares of the table) represents the $1/4$ chance of getting an a from both parents.

The sum rule of probability

In some genetics problems, you may need to calculate the probability that any one of several events will occur. In this case, you'll need to apply another rule of probability, the sum rule. According to the **sum rule**, the probability that any of several mutually exclusive events will occur is equal to the sum of the events' individual probabilities.

For example, if you roll a six-sided die, you have a $1/6$ chance of getting any given number, but you can only get one number per roll. You could never get both a one and a six at the same time; these outcomes are mutually exclusive. Thus, the chances of getting either a one or a six are: (probability of getting a 1) + (probability of getting a 6) = $(1/6) + (1/6) = 1/3$.

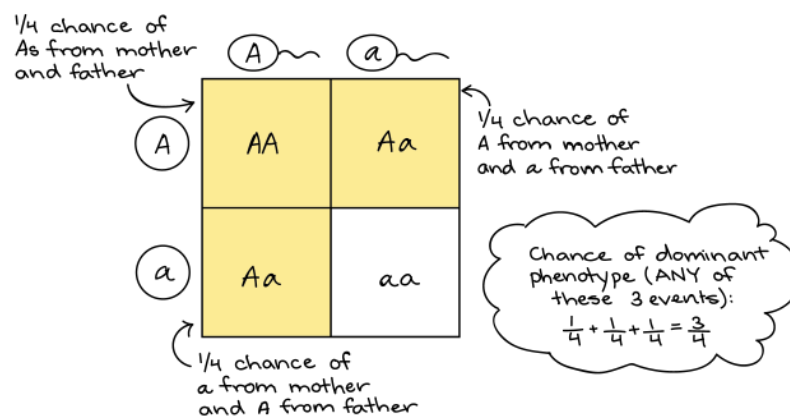
You can think of the sum rule as the “or” rule: if an outcome requires that either event X or event Y occur, and if X and Y are mutually exclusive (if only one or the other can occur in a given case), then the probability of the outcome can be calculated by adding the probabilities of X and Y.

As an example, let's use the sum rule to predict the fraction of offspring from an $Aa \times Aa$ cross that will have the dominant phenotype (AA or Aa genotype). In this cross, there are three events that can lead to a dominant phenotype:

- Two A gametes meet (giving AA genotype), or
- A gamete from Mom meets a gamete from Dad (giving Aa genotype), or
- a gamete from Mom meets A gamete from Dad (giving Aa genotype)

In any one fertilization event, only one of these three possibilities can occur (they are mutually exclusive).

Since this is an “or” situation where the events are mutually exclusive, we can apply the sum rule. Using the product rule as we did above, we can find that each individual event has a probability of $1/4$. So, the probability of offspring with a dominant phenotype is: (probability of A from Mom and A from Dad) + (probability of A from Mom and a from Dad) + (probability of a from Mom and A from Dad) = $(1/4) + (1/4) + (1/4) = 3/4$.



Once again, this is the same result we'd get with a Punnett square. One out of the four boxes of the Punnett square holds the dominant homozygote, AA. Two more boxes represent heterozygotes, one with a maternal A and a paternal a , the other with the opposite combination. Each box is 1 out of the 4 boxes in the whole Punnett square, and since the boxes don't overlap (they're mutually exclusive), we can add them up ($1/4 + 1/4 + 1/4 = 3/4$) to get the probability of offspring with the dominant phenotype.

The product rule and the sum rule

Product rule	Sum rule
For independent events X and Y, the probability (P) of them both occurring (X and Y) is $P(X) \cdot P(Y)$.	For mutually exclusive events X and Y, the probability (P) that one will occur (X or Y) is $P(X) + P(Y)$.

Applying probability rules to dihybrid crosses

Direct calculation of probabilities doesn't have much advantage over Punnett squares for single-gene inheritance scenarios. (In fact, if you prefer to learn visually, you may find direct calculation trickier rather than easier.) Where probabilities shine, though, is when you're looking at the behavior of two, or even more, genes.

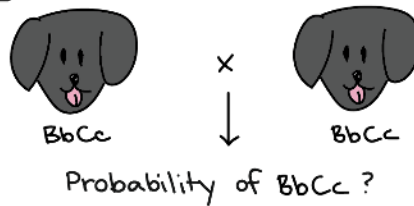
For instance, let's imagine that we breed two dogs with the genotype $BbCc$, where dominant allele B specifies black coat color (versus b , yellow coat color) and dominant allele C specifies straight fur (versus c , curly fur). Assuming that the two genes assort independently and are not sex-linked, how can we predict the number of $BbCc$ puppies among the offspring?

One approach is to draw a 16-square Punnett square. For a cross involving two genes, a Punnett square is still a good strategy. Alternatively, we can use a shortcut technique involving four-square Punnett squares and a little application of the product rule. In this technique, we break the overall question down into two smaller questions, each relating to a different genetic event:

1. What's the probability of getting a Bb genotype?
2. What's the probability of getting an Cc genotype?

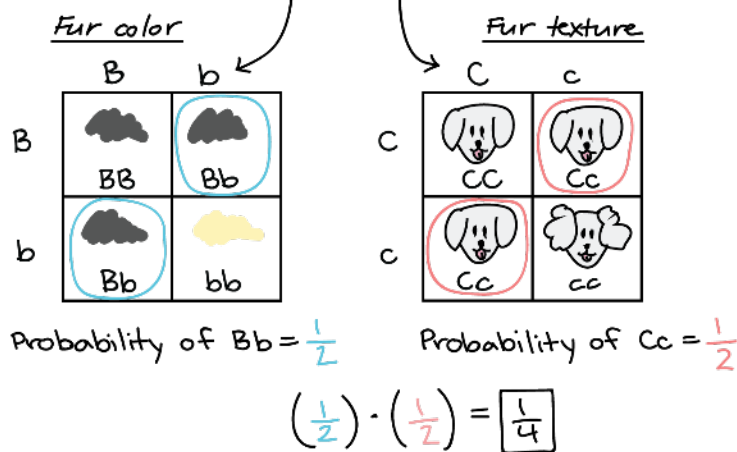
In order for a puppy to have a $BbCc$ genotype, both of these events must take place: the puppy must receive Bb alleles, *and* it must receive Cc alleles. The two events are independent because the genes assort independently (don't affect one another's inheritance). So, once we calculate the probability of each genetic event, we can multiply these probabilities using the product rule to get the probability of the genotype of interest ($BbCc$).

QUESTION:



SOLUTION:

$$\text{Probability of } BbCc = (\text{probability of } Bb) \cdot (\text{probability of } Cc)$$



To calculate the probability of getting a Bb genotype, we can draw a 4-square Punnett square using the parents' alleles for the coat color gene only, as shown above. Using the Punnett square, you can see that the probability of the Bb genotype is $\frac{1}{2}$. (Alternatively, we could have calculated the probability of Bb using the product rule for gamete contributions from the two parents and the sum rule for the two gamete combinations that give Bb .) Using a similar Punnett square for the parents' fur texture alleles, the probability of getting an Cc genotype is also $\frac{1}{2}$. To get the overall probability of the $BbCc$ genotype, we can simply multiply the two probabilities, giving an overall probability of $\frac{1}{4}$.

[Let's check that with a Punnett square]

QUESTION:

$BbCc$

$\times$
 $\downarrow$

$BbCc$

Probability of $BbCc$?

SOLUTION:

$BbCc$

		BC	Bc	bC	bc
	$BbCc$	BC	Bc	bC	bc
		$BBCC$	$BBCc$	$BbCC$	$BbCc$
		$BBCc$	$BBcc$	$BbCc$	$Bbcc$
		$BbCC$	$BbCc$	$bbCC$	$bbCc$
		$BbCc$	$Bbcc$	$bbCc$	$bbcc$

$\frac{4}{16} = \frac{1}{4} BbCc$

You can also use this technique to predict phenotype frequencies. Give it a try in the practice question below!

[Check your understanding](#)

Beyond dihybrid crosses

The probability method is most powerful (and helpful) in cases involving a large number of genes.

For instance, imagine a cross between two individuals with various alleles of four unlinked genes: $AaBbCCdd \times AabbCcDd$. Suppose you wanted to figure out the probability of getting offspring with the dominant phenotype for all four traits. Fortunately, you can apply the exact same logic as in the case of the dihybrid crosses above. To have the dominant phenotype for all four traits, and organism must have: one or more copies of the dominant allele A and one or more copies of dominant allele B and one or more copies of the dominant allele C and one or more copies of the dominant allele D .

Since the genes are unlinked, these are four independent events, so we can calculate a probability for each and then multiply the probabilities to get the probability of the overall outcome.

- The probability of getting one or more copies of the dominant A allele is $3/4$. (Draw a Punnett square for $Aa \times Aa$ to confirm for yourself that 3 out of the 4 squares are either AA or Aa .)
- The probability of getting one or more copies of the dominant B allele is $1/2$. (Draw a Punnett square for $Bb \times bb$: you'll find that half the offspring are Bb , and the other half bb .)
- The probability of getting one or more copies of the dominant C allele is 1. (If one of the parents is homozygous CC , there's no way to get offspring without a C allele!)

- The probability of getting one or more copies of the dominant D allele is $1/2$, as for B . (Half the offspring will be Dd , and the other half will be dd .)

To get the overall probability of offspring with the dominant phenotype for all four genes, we can multiply the probabilities of the four independent events: $(3/4) \cdot (1/2) \cdot (1) \cdot (1/2) = 3/16$.

[Check your understanding](#)

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- “[Mendel’s experiments and the laws of probability](#),” by OpenStax College, Biology (CC BY 3.0). Download the original article for free at <http://cnx.org/contents/185cbf87-c72e-48f5-b51e-f14f21b5eabd@9.85>.
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1.8: Introduction to heredity review

Key terms

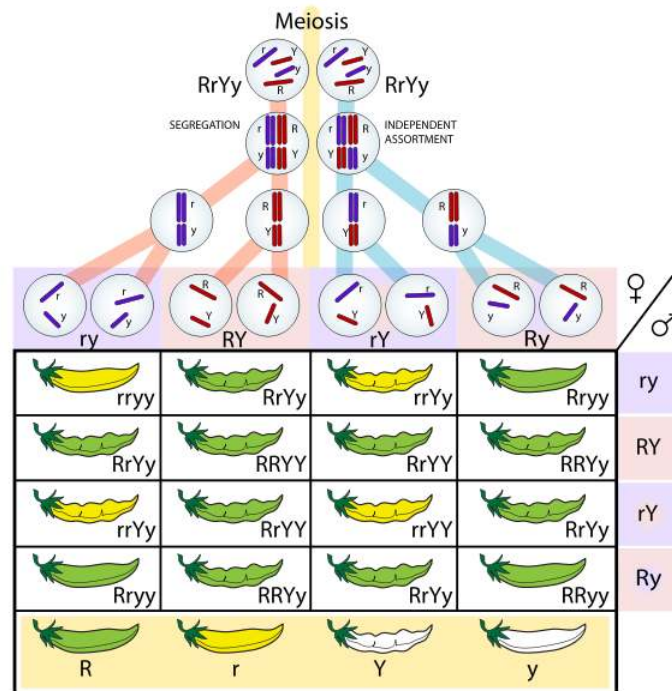
Term	Meaning
Genetics	The study of biological inheritance
Trait	A specific characteristic of an individual
Gene	A unit of heredity that is passed from parent to offspring
Allele	One of different forms of a gene
Genotype	The genetic makeup of an organism (ex: TT)
Phenotype	The physical characteristics of an organism (ex: tall)
Dominant allele	Allele that is phenotypically expressed over another allele
Recessive allele	Allele that is only expressed in absence of a dominant allele
Homozygous	Having two identical alleles for a particular gene
Heterozygous	Having two different alleles for a particular gene
Punnett square	Diagram that can be used to predict the genotypes and phenotypes resulting from a genetic cross

Mendelian inheritance

Gregor Mendel's principles of heredity, observed through patterns of inheritance in pea plants, form the basis of modern **genetics**.

Mendel proposed that **traits** were specified by "heritable elements" called **genes**. Genes come in different versions, or **alleles**, with **dominant alleles** being expressed over **recessive alleles**. Recessive alleles are only expressed when no dominant allele is present.

In most sexually reproducing organisms, each individual has two alleles for each gene (one from each parent). This pair of alleles is called a **genotype** and determines the organism's appearance, or **phenotype**.



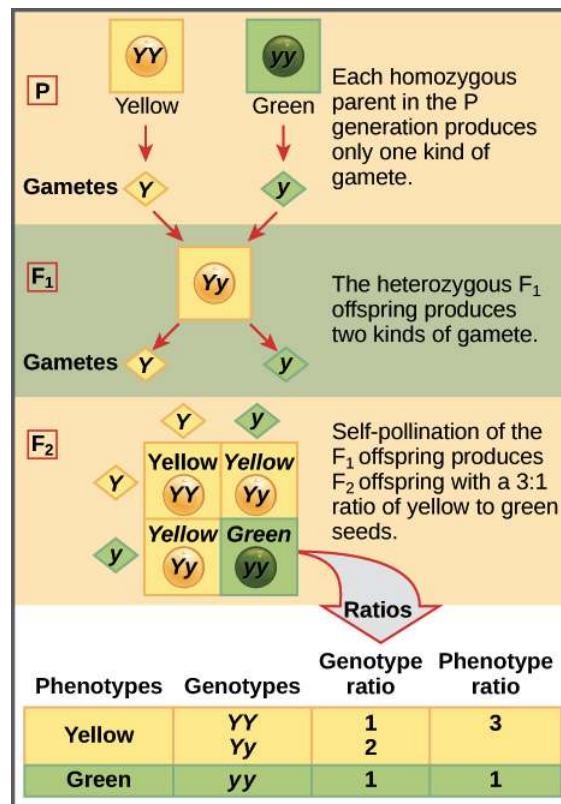
Laws of segregation and independent assortment. Image modified from [Wikimedia](#), [Public domain](#)

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

Mendel's second law is the *law of independent assortment*, which states that the alleles for one gene sort into gametes independently of the alleles of another gene.

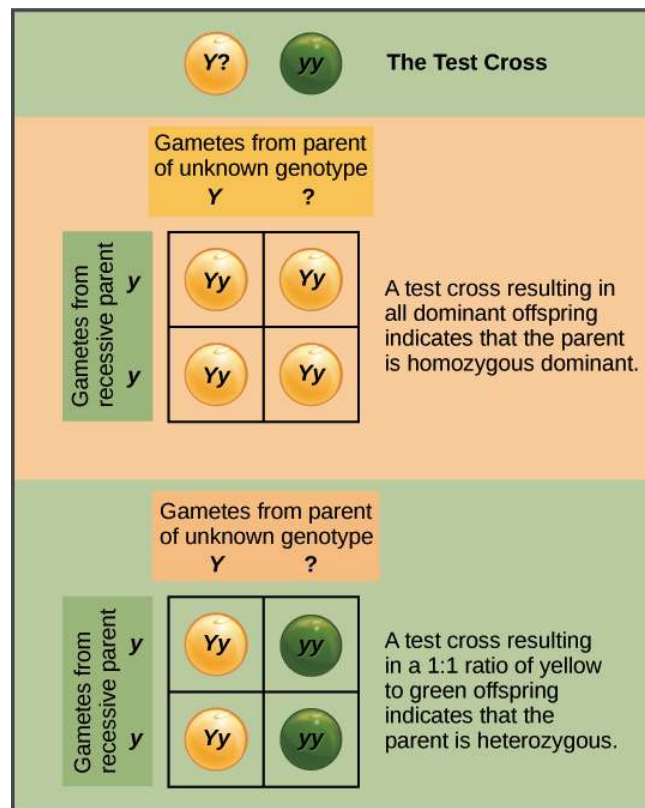
Punnett squares and probability

A **Punnett square** can be used to predict genotype and phenotypes of offspring from genetic crosses. A single-gene, or *monohybrid* cross is pictured below.



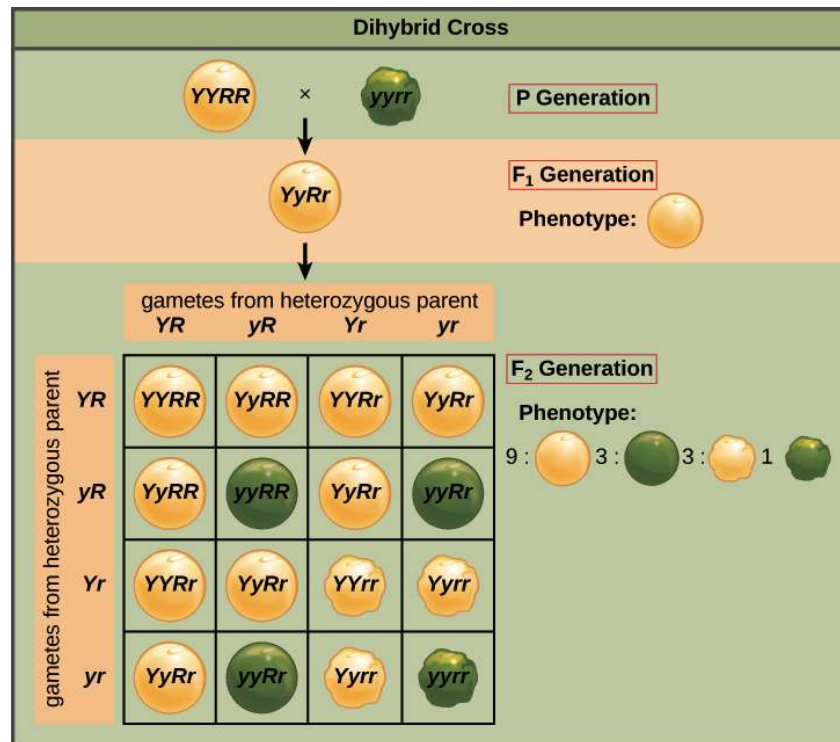
Monohybrid Punnett square. Image modified from [OpenStax](#), CC BY 4.0

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



Example test cross. Image credit: [OpenStax](#), CC BY 4.0

Punnett squares can be used for two-gene crosses, or *dihybrid crosses* by following the same basic rules as for a monohybrid cross. However, since there are now more gamete types, there must also be more squares in the table.

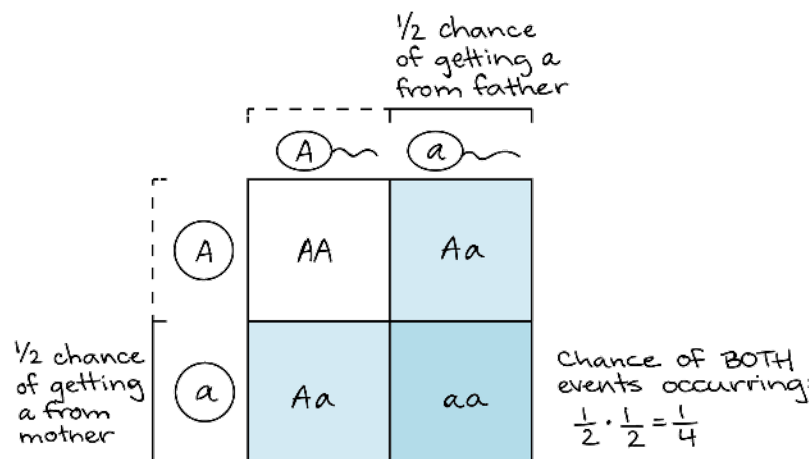


Dihybrid cross. Image credit: "OpenStax," CC BY 4.0.

Probabilities in genetics

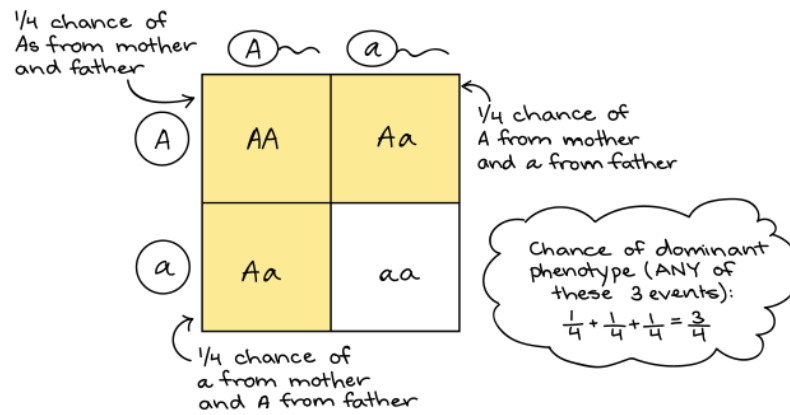
The two probability rules that are most relevant to Punnett squares are the *product rule* and the *sum rule*.

The product rule states that the probability of two (or more) independent events occurring together can be calculated by multiplying the individual probabilities of the events.



Example of the product rule using a Punnett square.

In some genetics problems, you may need to calculate the probability that any one of several events will occur. In this case, you'll need to apply another rule of probability, the *sum rule*. According to the sum rule, the probability that any of several mutually exclusive events will occur is equal to the sum of the events' individual probabilities.



Example of the sum rule using a Punnett square.

Common mistakes and misconceptions

- **Dominant traits are not always the most common.** Some people may think that dominant trait is the most likely to be found in the population, but the term "dominant" only refers to the fact that the allele is expressed over another allele. An example of this is Huntington's disease. Even though Huntington's is caused by a dominant allele, it only affects about 30,000 people in the United States¹.
- **Traits are not always the product of a single gene.** For example, there are at least 3 different genes that are associated with eye color in humans. In addition, there are sometimes more than two alleles for each gene. For example, there are 3 different alleles of one gene that determine coat color of cats.

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1.9: Practice - Introduction to heredity

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CHAPTER OVERVIEW

2: Non-Mendelian inheritance

- 2.1: Co-dominance and Incomplete Dominance
- 2.2: Multiple alleles, incomplete dominance, and codominance
- 2.3: Pleiotropy and lethal alleles
- 2.4: Polygenic inheritance and environmental effects
- 2.5: Non-Mendelian inheritance review
- 2.6: Practice - Non-Mendelian inheritance

Thumbnail: Incomplete dominance in snapdragons. (CC BY-NC-SA / cropped from original; [Khan Academy](#)).

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2.1: Co-dominance and Incomplete Dominance



In complete dominance, only one allele in the genotype is seen in the phenotype. In codominance, both alleles in the genotype are seen in the phenotype. In incomplete dominance, a mixture of the alleles in the genotype is seen in the phenotype. Created by Ross Firestone.

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2.2: Multiple alleles, incomplete dominance, and codominance

Introduction

Gregor Mendel knew how to keep things simple. In Mendel's work on pea plants, each gene came in just two different versions, or **alleles**, and these alleles had a nice, clear-cut dominance relationship (with the dominant allele fully overriding the recessive allele to determine the plant's appearance).

Today, we know that not all alleles behave quite as straightforwardly as in Mendel's experiments. For example, in real life:

- Allele pairs may have a variety of dominance relationships (that is, one allele of the pair may not completely “hide” the other in the heterozygote).
- There are often many different alleles of a gene in a population.

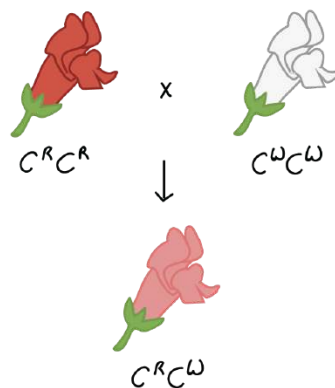
In these cases, an organism's **genotype**, or set of alleles, still determines its **phenotype**, or observable features. However, a variety of alleles may interact with one another in different ways to specify phenotype.

As a side note, we're probably lucky that Mendel's pea genes didn't show these complexities. If they had, it's possible that Mendel would not have understood his results, and wouldn't have figured out the core principles of inheritance—which are key in helping us understand the special cases!

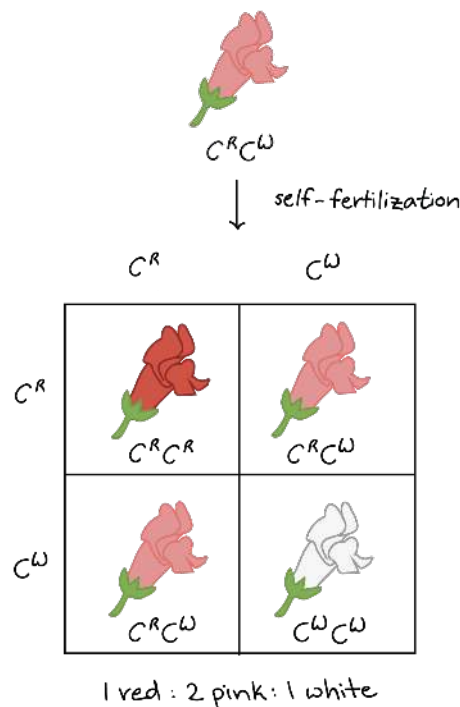
Incomplete dominance

Mendel's results were groundbreaking partly because they contradicted the (then-popular) idea that parents' traits were permanently blended in their offspring. In some cases, however, the phenotype of a heterozygous organism *can* actually be a blend between the phenotypes of its homozygous parents.

For example, in the snapdragon, *Antirrhinum majus*, a cross between a homozygous white-flowered plant ($C^W C^W$) and a homozygous red-flowered plant ($C^R C^R$) will produce offspring with pink flowers ($C^R C^W$). This type of relationship between alleles, with a heterozygote phenotype intermediate between the two homozygote phenotypes, is called **incomplete dominance**.



We can still use Mendel's model to predict the results of crosses for alleles that show incomplete dominance. For example, self-fertilization of a pink plant would produce a genotype ratio of $1 C^R C^R : 2 C^R C^W : 1 C^W C^W$ and a phenotype ratio of $1 : 2 : 1$ red: pink: white. Alleles are still inherited according to Mendel's basic rules, even when they show incomplete dominance.



Codominance

Closely related to incomplete dominance is **codominance**, in which both alleles are simultaneously expressed in the heterozygote.

We can see an example of codominance in the MN blood groups of humans (less famous than the ABO blood groups, but still important!). A person's MN blood type is determined by his or her alleles of a certain gene. An L^M allele specifies production of an M marker displayed on the surface of red blood cells, while an L^N allele specifies production of a slightly different N marker.

Homozygotes ($L^M L^M$ and $L^N L^N$) have only M or N markers, respectively, on the surface of their red blood cells. However, heterozygotes ($L^M L^N$) have both types of markers in equal numbers on the cell surface.

As for incomplete dominance, we can still use Mendel's rules to predict inheritance of codominant alleles. For example, if two people with $L^M L^N$ genotypes had children, we would expect to see M, MN, and N blood types and $L^M L^M$, $L^M L^N$, and $L^N L^N$ genotypes in their children in a 1 : 2 : 1 ratio (if they had enough children for us to determine ratios accurately!)

Multiple alleles

Mendel's work suggested that just two alleles existed for each gene. Today, we know that's not always, or even usually, the case! Although individual humans (and all diploid organisms) can only have two alleles for a given gene, multiple alleles may exist in a population level, and different individuals in the population may have different pairs of these alleles.

As an example, let's consider a gene that specifies coat color in rabbits, called the C gene. The C gene comes in four common alleles: C , c^{ch} , c^h , and c :

- A CC rabbit has black or brown fur.
- A $c^{ch}c^{ch}$ rabbit has chinchilla coloration (grayish fur).
- A $c^h c^h$ rabbit has Himalayan (color-point) patterning, with a white body and dark ears, face, feet, and tail.
- A cc rabbit is albino, with a pure white coat.

Genotype			
CC	$c^{ch}c^{ch}$	$c^h c^h$	cc
Phenotype			
BLACK	CHINCHILLA	HIMALAYAN	ALBINO
			

Image credit: "Characteristics and traits: Figure 5," by OpenStax College, Biology (CC BY 3.0).

Multiple alleles makes for many possible dominance relationships. In this case, the black C allele is completely dominant to all the others; the chinchilla c^{ch} allele is incompletely dominant to the Himalayan c^h and albino c alleles; and the Himalayan c^h allele is completely dominant to the albino c allele.

Rabbit breeders figured out these relationships by crossing different rabbits of different genotypes and observing the phenotypes of the heterozygous kits (baby bunnies).

[How do these alleles change the rabbit's color?]

The C gene in rabbits encodes an enzyme that's needed to make a type of pigment called melanin in hairs^{1,2}.

- The C allele of this gene encodes a fully functional enzyme that makes lots of pigment and results in black fur.
- The c^{ch} allele encodes an enzyme that is less effective at making pigment, resulting in lighter, more grayish fur.
- The c^h allele encodes a defective enzyme, where the defect makes the enzyme very sensitive to temperature: it works fine at low temperatures, but doesn't work at all at higher temperatures. The rabbit's extremities (paws, ears, etc.) are cooler, so the enzyme functions there and makes pigment. The rabbit's main body is warmer, so the enzyme does not function and no pigment is made.
- The c allele encodes a completely nonfunctional enzyme, leading to an albino rabbit (one that does not produce any pigment in its hairs).

Note to rabbit fanciers: Rabbit coat color is determined by a number of genes, not just by the C gene. The allelic series described here for C assumes a certain genetic background for the other genes, one in which a CC genotype results in a black rabbit.

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2.3: Pleiotropy and lethal alleles

Introduction

From Mendel's experiments, you might imagine that all genes control a single characteristic and affect some harmless aspect of an organism's appearance (such as color, height, or shape). Those predictions are true for some genes, but definitely not all of them! For example:

- A human genetic disorder called Marfan syndrome is caused by a mutation in one gene, yet it affects many aspects of growth and development, including height, vision, and heart function. This is an example of **pleiotropy**, or one gene affecting multiple characteristics.
- A cross between two heterozygous yellow mice produces yellow and brown mice in a ratio of 2:1, not 3:1. This is an example of **lethality**, in which a particular genotype makes an organism unable to survive.

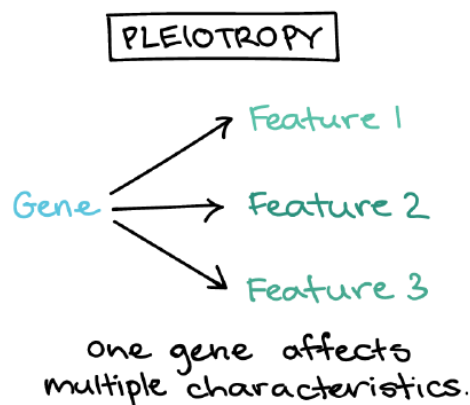
In this article, we'll take a closer look at pleiotropic genes and lethal alleles, seeing how these variations on Mendel's rules fit into our modern understanding of inheritance.

Pleiotropy

When we mentioned Mendel's [experiments](#) with purple-flowered and white-flowered plants, we didn't discuss any other phenotypes associated with the two flower colors. However, Mendel noticed that the flower colors were always correlated with two other features: the color of the seed coat (covering of the seed) and the color of the axils (junctions where the leaves met the stem)^{1,2}.

In plants with white flowers, the seed coats and axils were colorless. In plants with purple flowers, on the other hand, the seed coats were brown-gray and the axils were reddish. Thus, rather than affecting just one characteristic, the flower color gene actually affected three.

Genes like this, which control multiple, seemingly unrelated features, are said to be **pleiotropic** (*pleio-* = many, *-tropic* = effects)¹. We now know that Mendel's flower color gene specifies a protein that causes colored particles, or pigments, to be made². This protein works in several different parts of the pea plant (flowers, seed coat, and leaf axils). In this way, the seemingly unrelated phenotypes can be traced back to a defect in one gene with several jobs.



Based on similar diagram by Ingrid Lobo¹.

Importantly, alleles of pleiotropic genes are transmitted in the same way as alleles of genes that affect single traits. Although the phenotype has multiple elements, these elements are specified as a package, and the dominant and recessive versions of the package would appear in the offspring of two heterozygotes in a ratio of 3:1.

Pleiotropy in human genetic disorders

Genes affected in human genetic disorders are often pleiotropic. For example, people with a hereditary disorder called Marfan syndrome may have a set of seemingly unrelated symptoms, including the following^{1,3}:

- Unusually tall height
- Thin fingers and toes
- Dislocation of the lens of the eye
- Heart problems (in which the aorta, the large blood vessel carrying blood away from the heart, bulges or ruptures).

These symptoms don't seem directly related, but as it turns out, they can all be traced back to the mutation of a single gene. This gene encodes a protein that assembles into chains, making elastic fibrils that give strength and flexibility to the body's connective tissues⁴. Mutations that cause Marfan syndrome reduce the amount of functional protein made by the body, resulting in fewer fibrils.

How does the identity of this gene explain the range of symptoms? Our eyes and the aortas normally contain many fibrils that help maintain structure, which is why these two organs are affected in Marfan syndrome⁵. In addition, the fibrils serve as “storage shelves” for growth factors. When there are fewer of them in Marfan syndrome, the growth factors cannot be shelved and thus cause excess growth (leading to the characteristic tall, thin Marfan build)⁴.

Lethality

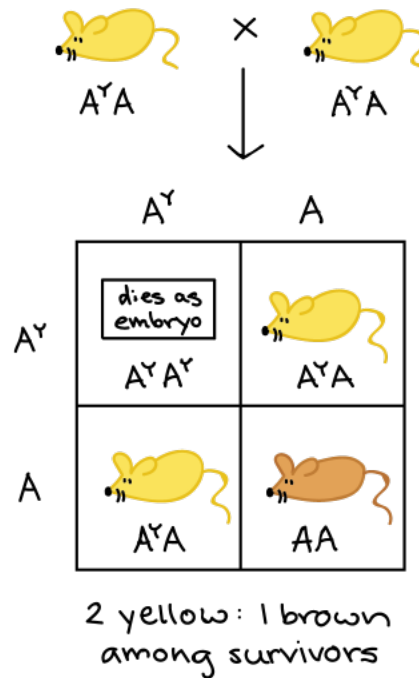
For the alleles that Mendel studied, it was equally possible to get homozygous dominant, homozygous recessive, and heterozygous genotypes. That is, none of these genotypes affected the survival of the pea plants. However, this is not the case for all genes and all alleles.

Many genes in an organism's genome are needed for survival. If an allele makes one of these genes nonfunctional, or causes it to take on an abnormal, harmful activity, it may be impossible to get a living organism with a homozygous (or, in some cases, even a heterozygous) genotype.

Example: The yellow mouse

A classic example of an allele that affects survival is the lethal yellow allele, a spontaneous mutation in mice that makes their coats yellow. This allele was discovered around the turn of the 20th century by the French geneticist Lucien Cuenót, who noticed that it was inherited in an unusual pattern^{6,7}.

When yellow mice were crossed with normal agouti (brown) mice, they produced half yellow and half brown offspring. This suggested that the yellow mice were heterozygous, and that the yellow allele, A^Y , was dominant to the agouti allele, A . But when two yellow mice were crossed with each other, they produced yellow and brown offspring in a ratio of 2:1, and the yellow offspring did not breed true (were heterozygous). Why was this the case?



As it turned out, this unusual ratio reflected that some of the mouse embryos (homozygous $A^Y A^Y$ genotype) died very early in development, long before birth. In other words, at the level of eggs, sperm, and fertilization, the color gene segregated normally, resulting in embryos with a 1:2:1 ratio of $A^Y A^Y$, $A^Y A$, and $A A$ genotypes. However, the $A^Y A^Y$ mice died as tiny embryos, leaving a 2:1 genotype and phenotype ratio among the surviving mice^{7,8}.

Alleles like A^Y , which are lethal when they're homozygous but not when they're heterozygous, are called **recessive lethal alleles**.

[Wait, isn't yellow color dominant?]

The lethal recessive A^Y allele also happens to give a dominant, non-lethal phenotype in the heterozygote (yellow coloration, along with health problems such as obesity, diabetes, and tumor formation). The trick is that the *lethality* is recessive, even though the color phenotype is dominant.

The color of the yellow mouse heterozygote is helpful because it lets us keep track of genotypes, but in many other cases, recessive lethal genes don't have a dominant phenotype in the heterozygote. Instead, they are purely recessive to the normal allele, resulting in a normal phenotype for the heterozygote and an embryonic lethal phenotype for the homozygote.

Lethal alleles and human genetic disorders

Some alleles associated with human genetic disorders are recessive lethal. For example, this is true of the allele that causes achondroplasia, a form of dwarfism. A person heterozygous for this allele will have shortened limbs and short stature (achondroplasia), a condition that is not lethal. However, homozygosity for the same allele causes death during embryonic development or the first months of life, an example of recessive lethality^{7,9}.

Some human disorders are also caused by **dominant lethal alleles**. These are alleles that cause death when they are present in just a single copy. If an allele leads to death of heterozygotes before birth, we'll never see that allele in the living human population (but rather, as an implantation failure or miscarriage). However, if a dominant lethal allele allows heterozygotes to survive past birth, it can be seen in the population as a genetic disorder.

In fact, if a dominant lethal allele lets a person survive to reproductive age, it may even be passed on to children. This is the case in Huntington's disease, a fatal genetic disorder affecting the nervous system. People with a Huntington allele inevitably develop the disease, but they may not show any symptoms until age 40 and can unknowingly pass the allele on to their children.

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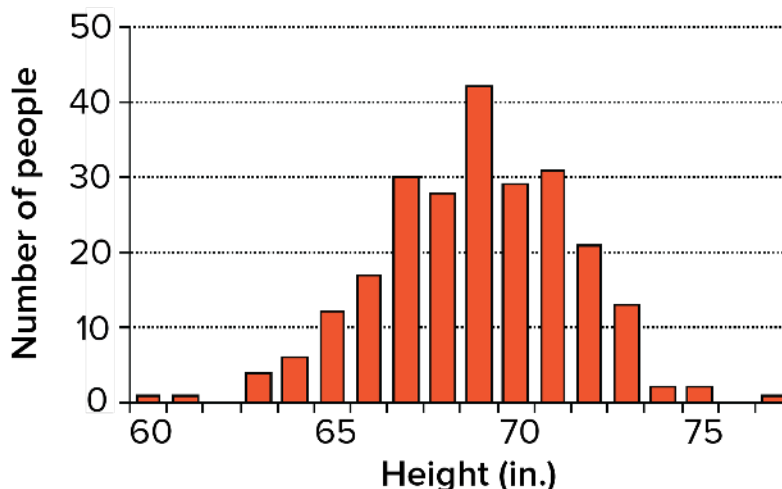
2.4: Polygenic inheritance and environmental effects

How is height inherited?

If what you're really interested in is human genetics, learning about Mendelian genetics can sometimes be frustrating. You'll often hear a teacher use a human trait as an example in a genetics problem, but then say, "that's an oversimplification" or "it's much more complicated than that." So, what's actually going on with those interesting human traits, such as eye color, hair and skin color, height, and disease risk?

As an example, let's consider human height. Unlike a simple Mendelian characteristic, human height displays:

- **Continuous variation.** Unlike Mendel's [pea plants](#), humans don't come in two clear-cut "tall" and "short" varieties. In fact, they don't even come in four heights, or eight, or sixteen. Instead, it's possible to get humans of many different heights, and height can vary in increments of inches or fractions of inches¹.



The heights of a group of male high school seniors. Image modified from "[Continuous variation: Quantitative traits](#)," by J. W. Kimball ([CC BY 3.0](#))

- **A complex inheritance pattern.** You may have noticed that tall parents can have a short child, short parents can have a tall child, and two parents of different heights may or may not have a child in the middle. Also, siblings with the same two parents may have a range of heights, ones that don't fall into distinct categories.

Simple models involving one or two genes can't accurately predict all of these inheritance patterns. How, then, is height inherited?

Height and other similar features are controlled not just by one gene, but rather, by multiple (often many) genes that each make a small contribution to the overall outcome. This inheritance pattern is sometimes called **polygenic inheritance** (*poly-* = many). For instance, a recent study found over 400 genes linked to variation in height².

When there are large numbers of genes involved, it becomes hard to distinguish the effect of each individual gene, and even harder to see that gene variants (alleles) are inherited according to Mendelian rules. In an additional complication, height doesn't just depend on genetics: it also depends on environmental factors, such as a child's overall health and the type of nutrition he or she gets while growing up.

In this article, we'll examine how complex traits such as height are inherited. We'll also see how factors like genetic background and environment can affect the **phenotype** (observable features) produced by a particular **genotype** (set of gene variants, or alleles).

Polygenic inheritance

Human features like height, eye color, and hair color come in lots of slightly different forms because they are controlled by many genes, each of which contributes some amount to the overall phenotype. For example, there are two major eye color genes, but at least 14 other genes that play roles in determining a person's exact eye color³.

Looking at a real example of a human polygenic trait would get complicated, largely because we'd have to keep track of tens, or even hundreds, of different allele pairs (like the 400 involved in height!). However, we can use an example involving wheat kernels to see how several genes whose alleles "add up" to influence the same trait can produce a spectrum of phenotypes^{1,4}.

In this example, there are three genes that make reddish pigment in wheat kernels, which we'll call *A*, *B*, and *C*. Each comes in two alleles, one of which makes pigment (the capital-letter allele) and one of which does not (the lowercase allele). These alleles have additive effects: the *aa* genotype would contribute no pigment, the *Aa* genotype would contribute some amount of pigment, and the *AA* genotype would contribute more pigment (twice as much as *Aa*). The same would hold true for the *B* and *C* genes^{1,4}.

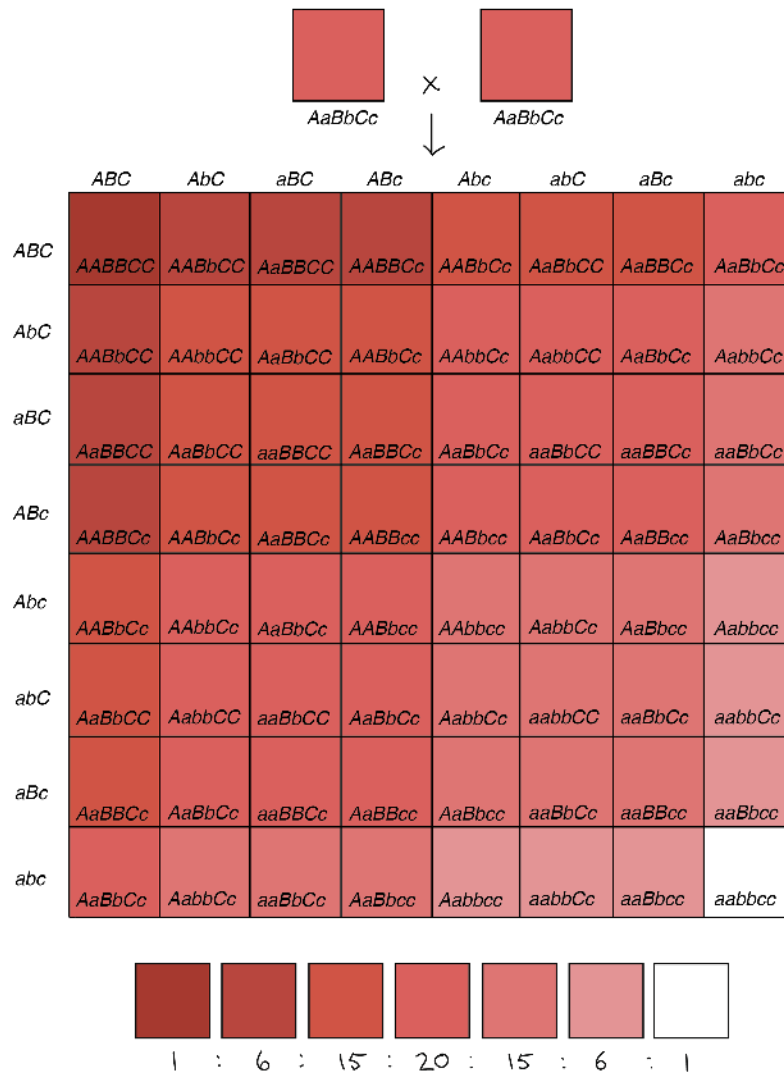


Diagram based on similar diagram by W. P. Armstrong⁵.

Now, let's imagine that two plants heterozygous for all three genes (*AaBbCc*) were crossed to one another. Each of the parent plants would have three alleles that made pigment, leading to pinkish kernels. Their offspring, however, would fall into seven color groups, ranging from no pigment whatsoever (*aabbcc*) and white kernels to lots of pigment (*AABBCC*) and dark red kernels. This is in fact what researchers have seen when crossing certain varieties of wheat^{1,4}.

This example shows how we can get a spectrum of slightly different phenotypes (something close to continuous variation) with just three genes. It's not hard to imagine that, as we increased the number of genes involved, we'd be able to get even finer variations in color, or in another trait such as height.

Environmental effects

Human phenotypes—and phenotypes of other organisms—also vary because they are affected by the environment. For instance, a person may have a genetic tendency to be underweight or obese, but his or her actual weight will depend on diet and exercise (with these factors often playing a greater role than genes). In another example, your hair color may depend on your genes—until you dye your hair purple!

One striking example of how environment can affect phenotype comes from the hereditary disorder **phenylketonuria (PKU)**⁶. People who are homozygous for disease alleles of the PKU gene lack activity of an enzyme that breaks down the amino acid phenylalanine. Because people with this disorder cannot get rid of excess phenylalanine, it rapidly builds up to toxic levels in their bodies⁷.

If PKU is not treated, the extra phenylalanine can keep the brain from developing normally, leading to intellectual disability, seizures, and mood disorders. However, because PKU is caused by the buildup of too much phenylalanine, it can also be treated in a very simple way: by giving affected babies and children a diet low in phenylalanine⁸.

If people with phenylketonuria follow this diet strictly from a very young age, they can have few, or even no, symptoms of the disorder. In many countries, all newborns are screened for PKU and similar genetic diseases shortly after birth through a simple blood test, as shown in the image below.



Image credit: "Phenylketonuria testing," by Eric T. Sheler, USAF Photographic Archives (public domain).

Variable expressivity, incomplete penetrance

Even for characteristics that are controlled by a single gene, it's possible for individuals with the same genotype to have different phenotypes. For example, in the case of a genetic disorder, people with the same disease genotype may have stronger or weaker forms of the disorder, and some may never develop the disorder at all.

In **variable expressivity**, a phenotype may be stronger or weaker in different people with the same genotype. For instance, in a group of people with a disease-causing genotype, some might develop a severe form of the disorder, while others might have a milder form. The idea of expressivity is illustrated in the diagram below, with the shade of green representing the strength of the phenotype.

[Example]

The genetic disorder retinoblastoma causes cancerous tumors of the eyes, but the disease varies in severity and speed of onset. Children with retinoblastoma may develop tumors in just one eye or in both eyes, and the tumors may appear more quickly or slowly after birth⁹.



Illustration modeled after similar image by Steven M. Carr¹⁰.

In **incomplete penetrance**, individuals with a certain genotype may or may not develop a phenotype associated with the genotype. For example, among people with the same disease-causing genotype for a hereditary disorder, some might never actually develop the disorder. The idea of penetrance is illustrated in the diagram below, with green or white color representing the presence or absence of a phenotype.

[Example]

Some mutations in the retinoblastoma gene have high penetrance, but others have lower (incomplete) penetrance. For mutations in this latter category, some family members with the mutation are normal, while others develop retinoblastoma tumors^{6,11}.

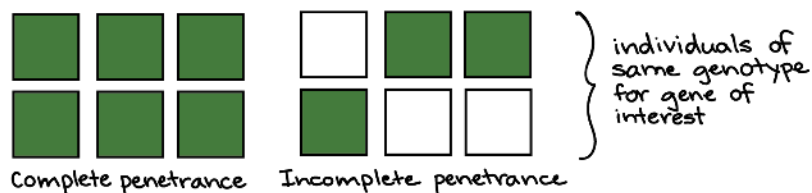


Illustration modeled after similar image by Steven M. Carr¹⁰.

What causes variable expressivity and incomplete penetrance? Other genes and environmental effects are often part of the explanation. For example, disease-causing alleles of one gene may be suppressed by alleles of another gene elsewhere in the genome, or a person's overall health may influence the strength of a disease phenotype¹¹.

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2.5: Non-Mendelian inheritance review

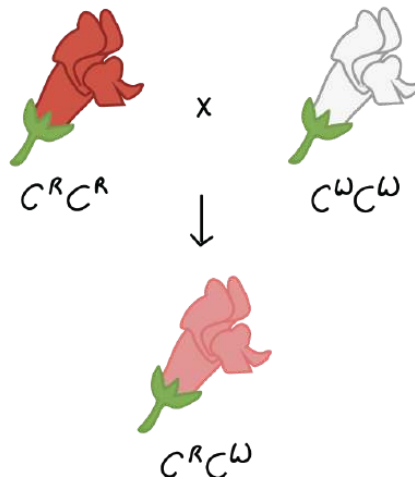
Key terms

Term	Meaning
Incomplete dominance	Pattern of heredity in which one allele is not completely dominant over another
Codominance	Pattern of heredity in which both alleles are simultaneously expressed in the heterozygote
Multiple alleles	A gene that is controlled by more than two alleles
Pleiotropy	When one gene affects multiple characteristics
Lethal allele	Allele that results in the death of an individual
Polygenic trait	Traits that are controlled by multiple genes

Variations involving single genes

Some of the variations on Mendel's rules involve single genes.

- **Incomplete dominance.** Two alleles may produce an intermediate phenotype when both are present, rather than one fully determining the phenotype.



An example of this is the snapdragon plant. A cross between a homozygous white-flowered plant ($C^W C^W$) and a homozygous red-flowered plant ($C^R C^R$) will produce offspring with pink flowers ($C^R C^W$).

- **Codominance.** Two alleles may be simultaneously expressed when both are present, rather than one fully determining the phenotype.



Codominance in erminette chicken. Image from [Wikimedia](#), CC BY 2.0.

In some varieties of chickens, the allele for black feathers is codominant with the allele for white feathers. A cross between a black chicken and a white chicken will result in a chicken with both black and white feathers.

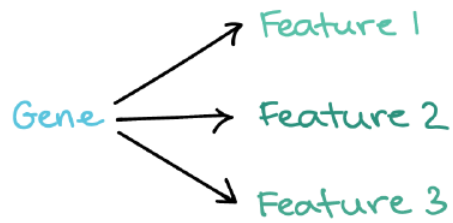
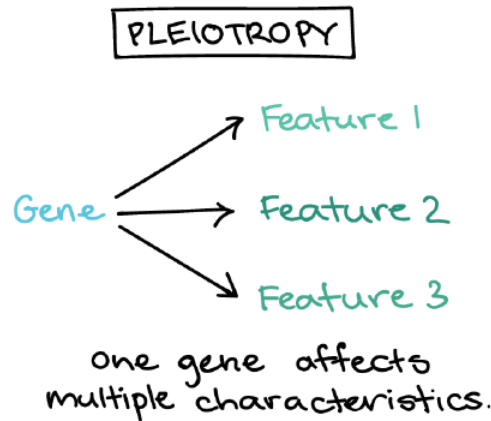
- **Multiple alleles.** Mendel studied just two alleles of his pea genes, but real populations often have multiple alleles of a given gene.

Genotype			
CC	$c^{ch}c^{ch}$	$c^h c^h$	cc
Phenotype			
BLACK	CHINCHILLA	HIMALAYAN	ALBINO
			

Image from [OpenStax](#), CC BY 3.0

An example of this is the gene for coat color in rabbits (the C gene) which comes in four common alleles: C , c^{ch} , c^h , and c .

- **Pleiotropy.** Some genes affect many different characteristics, not just a single characteristic.

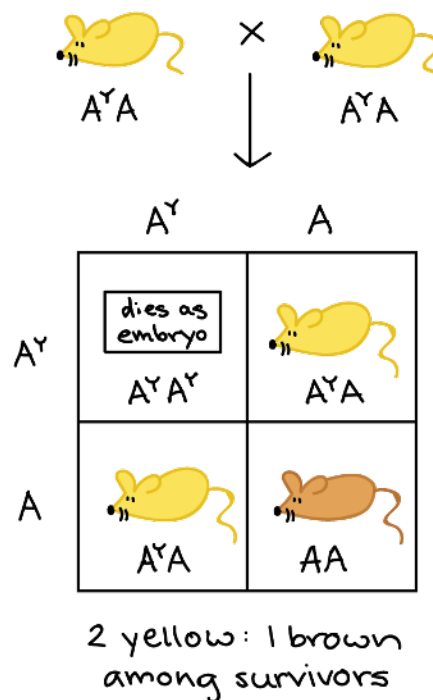


one gene affects multiple characteristics.

Based on similar diagram by Ingrid Lobo¹.

An example of this is Marfan syndrome, which results in several symptoms (unusually tall height, thin fingers and toes, lens dislocation, and heart problems). These symptoms don't seem directly related, but as it turns out, they can all be traced back to the mutation of a single gene.

- **Lethal alleles.** Some genes have alleles that prevent survival when homozygous or heterozygous.

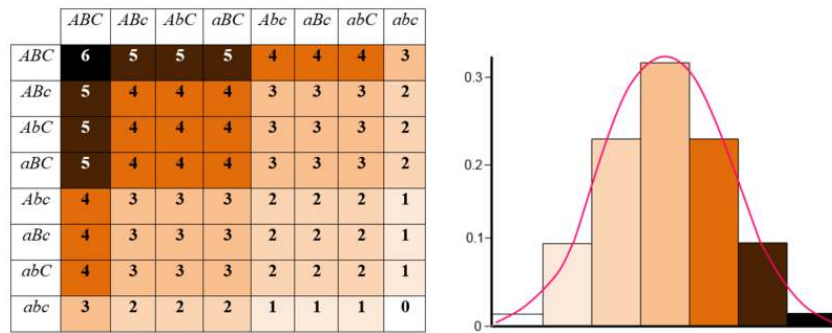


A classic example of an allele that affects survival is the lethal yellow allele, a spontaneous mutation in mice that makes their coats yellow. Mice that are homozygous ($A^Y A^Y$) genotype die early in development. Although this particular allele is dominant, lethal alleles can be dominant or recessive, and can be expressed in homozygous or heterozygous conditions.

Polygenic inheritance and environmental effects

Many characteristics, such as height, skin color, eye color, and risk of diseases, are controlled by many factors. These factors may be genetic, environmental, or both.

- **Polygenic inheritance.** Some characteristics are **polygenic**, meaning that they're controlled by a number of different genes. In polygenic inheritance, traits often form a phenotypic spectrum rather than falling into clear-cut categories.



Human skin color chart. Image from [Wikimedia](#), CC BY-SA 4.0.

An example of this is skin pigmentation in humans, which is controlled by several different genes.

- **Environmental effects.** Most real-world characteristics are determined not just by genotype, but also by environmental factors that influence how genotype is translated into phenotype.



Blue and pink hydrangea due to variance in soil pH. Image by [Lynn Greyling](#), Public domain

An example of this is the hydrangea flower. Hydrangea of the same genetic variety may vary in color from blue to pink depending on the pH of the soil they are in.

Common mistakes and misconceptions

- **Some people confuse pleiotropy and polygenic inheritance.** The major difference between the two is that pleiotropy is when *one gene* affects multiple characteristics (e.g. Marfan syndrome) and polygenic inheritance is when *one trait* is controlled by multiple genes (e.g. skin pigmentation).
- **Codominance and incomplete dominance are not the same.** In codominance, neither allele is dominant over the other, so both will be expressed equally in the heterozygote. In incomplete dominance, there is an intermediate heterozygote (such as a pink flower when the parents' phenotypes are red and white).

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2.6: Practice - Non-Mendelian inheritance

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CHAPTER OVERVIEW

3: Sex linkage

3.1: Example punnet square for sex-linked recessive trait

3.2: X-linked inheritance

3.3: X-inactivation

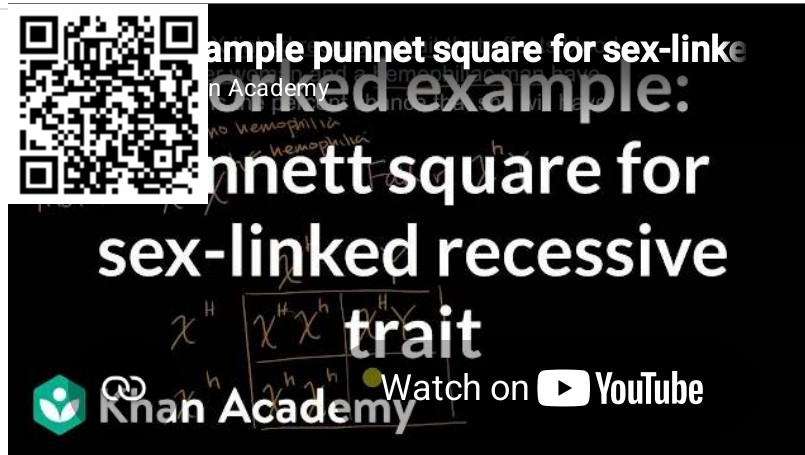
3.4: Sex linkage review

3.5: Practice - Sex linkage

Thumbnail: Tortoiseshell cat. (Public domain; Michael Bodega via [Wikimedia Commons](#)).

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3.1: Example punnet square for sex-linked recessive trait



Example punnet square for sex-linked recessive trait.

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3.2: X-linked inheritance

Key points:

- In humans and other mammals, biological sex is determined by a pair of **sex chromosomes**: XY in males and XX in females.
- Genes on the X chromosome are said to be **X-linked**. X-linked genes have distinctive inheritance patterns because they are present in different numbers in females (XX) and males (XY).
- X-linked human genetic disorders are much more common in males than in females due to the X-linked inheritance pattern.

Introduction

If you're a human being (which seems like a good bet!), most of your chromosomes come in **homologous pairs**. The two chromosomes of a homologous pair contain the same basic information – that is, the same genes in the same order – but may carry different versions of those genes.

Are all of your chromosomes organized in homologous pairs? The answer depends on whether you're (chromosomally) male.

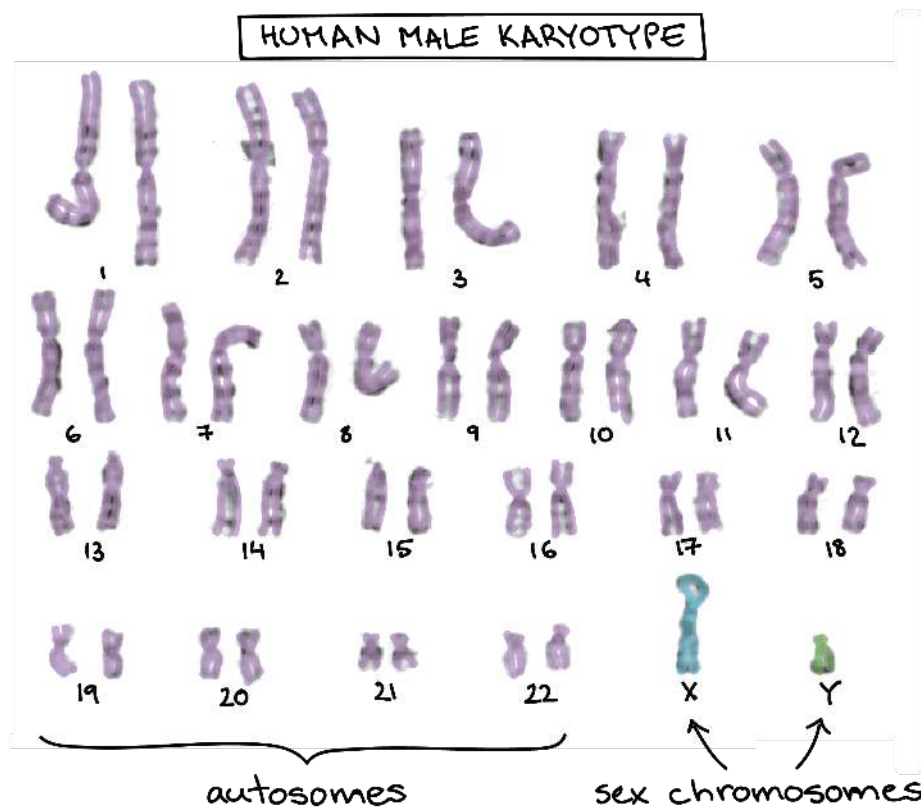


Image modified from "Karyotype," by Can H. (CC BY 2.0).

- A human male has two **sex chromosomes**, the X and the Y. Unlike the 44 **autosomes** (non-sex chromosomes), the X and Y don't carry the same genes and aren't considered homologous.
- Instead of an X and a Y, a human female has two X chromosomes. These X chromosomes do form a *bona fide* homologous pair.

Because sex chromosomes don't always come in homologous pairs, the genes they carry show unique, distinctive patterns of inheritance.

Sex chromosomes in humans

Human X and Y chromosomes determine the biological sex of a person, with XX specifying female and XY specifying male. Although the Y chromosome contains a small region of similarity to the X chromosome so that they can pair during meiosis, the Y chromosome is much shorter and contains many fewer genes.

To put some numbers to it, the X chromosome has about 800-900 protein-coding genes with a wide variety of functions, while the Y chromosome has just 60-70 protein-coding genes, about half of which are active only in the testes (sperm-producing organs)^{1,2,3,4}.

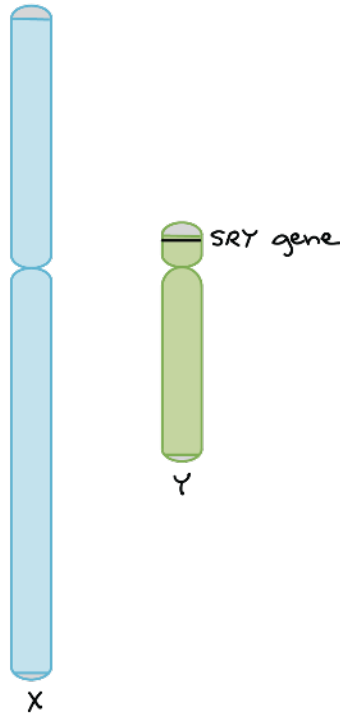


Image based on ideograms from the [Genome Decoration Page](#), maintained by the U.S. NCBI.

The human Y chromosome plays a key role in determining the sex of a developing embryo. This is mostly due to a gene called *SRY* (“sex-determining region of Y”). *SRY* is found on the Y chromosome and encodes a protein that turns on other genes required for male development^{5,6}.

- XX embryos don't have *SRY*, so they develop as female.
- XY embryos do have *SRY*, so they develop as male.

In rare cases, errors during meiosis may transfer *SRY* from the Y chromosome to the X chromosome. If an *SRY*-bearing X chromosome fertilizes a normal egg, it will produce a chromosomally female (XX) embryo that develops as a male⁷. If an *SRY*-deficient Y chromosome fertilizes a normal egg, it will produce a chromosomally male embryo (XY) that develops as a female⁸.

[Do all species with X and Y chromosomes have the *SRY* gene?]

No, not all of them. The molecular mechanisms of sex determination vary greatly among species, even those that use an X-Y sex determination system. In most placental mammals, *SRY* is found on the Y chromosome and is used for sex determination. However, the *SRY* gene is not present in other species with X-Y sex determination systems, such as fruit flies and other insects⁹.

X and Y chromosomes have evolved independently many times

To understand how this is possible, it's useful to keep in mind that “X” and “Y” are just generic labels applied to the dimorphic (*di-* = two, *-morphe* = form), or dissimilar, chromosomes found in species with X-Y sex determination systems¹⁰. X is whatever chromosome the female has two of, while Y is the chromosome paired with a single X in the male. The nature of the chromosomes — what genes are on them, and how they determine maleness or femaleness — may be quite different between species.

Sex chromosomes have evolved independently many times¹¹. Thus, although fruit flies and humans both have X and Y chromosomes that match the above definition, and although XY and XX chromosome combinations correspond to maleness and femaleness, respectively, in both species, the sex determination mechanism in flies is very different from that of humans.

Example: Humans vs. fruit flies

In humans, as described above, the Y chromosome specifies maleness through the action of the SRY gene, a master regulator of male development.

In flies, the presence of a single X chromosome specifies maleness, while the presence of two Xs specifies femaleness, regardless of the presence or absence of a Y chromosome. (More specifically, it appears that sex is determined by the ratio of X chromosomes to autosomes, with a 1:2 ratio specifying maleness and a 2:2 ratio specifying femaleness. These ratios are "measured" through the levels of proteins produced by specific genes on the X chromosome and on autosomes.)¹²

Because of these differences in sex determination mechanisms, an XXY fly will develop as a female (see article on [Thomas Hunt Morgan's experiments](#)), while an XXY human will develop as a male (in a condition known as Klinefelter syndrome; see article on [large-scale chromosomal changes](#)).

X-linked genes

When a gene is present on the X chromosome, but not on the Y chromosome, it is said to be **X-linked**. X-linked genes have different inheritance patterns than genes on non-sex chromosomes (autosomes). That's because these genes are present in different copy numbers in males and females.

[What about genes on the Y?]

Genes on either the X or the Y have unusual inheritance patterns and are called **sex-linked**, but X-linkage is much more common than Y-linkage (since there are many more genes on the X than the Y).

Since a female has two X chromosomes, she will have two copies of each X-linked gene. For instance, in the fruit fly *Drosophila* (which, like humans, has XX females and XY males), there is an eye color gene called *white* that's found on the X chromosome, and a female fly will have two copies of this gene. If the gene comes in two different alleles, such as X^W (dominant, normal red eyes) and X^w (recessive, white eyes), the female fly may have any of three genotypes: $X^W X^W$ (red eyes), $X^W X^w$ (red eyes), and $X^w X^w$ (white eyes).

A male has different genotype possibilities than a female. Since he has only one X chromosome (paired with a Y), he will have only one copy of any X-linked genes. For instance, in the fly eye color example, the two genotypes a male can have are $X^W Y$ (red eyes) and $X^w Y$ (white eyes). Whatever allele the male fly inherits for an X-linked gene will determine his appearance, because he has no other gene copy—even if the allele is recessive in females. Rather than homozygous or heterozygous, males are said to be **hemizygous** for X-linked genes.

We can see how sex linkage affects inheritance patterns by considering a cross between two flies, a white-eyed female ($X^w X^w$) and a red-eyed male ($X^W Y$). If this gene were on a non-sex chromosome, or autosome, we would expect all of the offspring to be red-eyed, because the red allele is dominant to the white allele. What we actually see is the following:

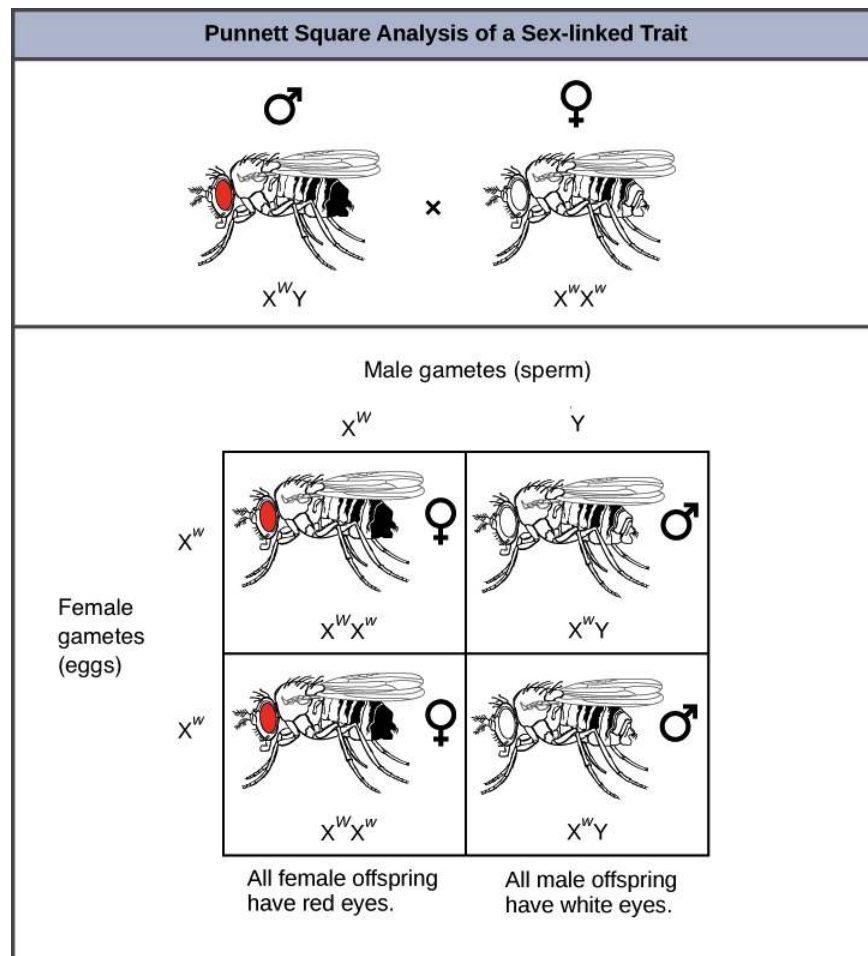


Image credit: "Characteristics and traits: Figure 10," by OpenStax College, Biology, CC BY 4.0

However, because the gene is X-linked, and because it was the female parent who had the recessive phenotype (white eyes), all the male offspring—who get their only X from their mother—have white eyes ($X^w Y$). All the female offspring have red eyes because they received two Xs, with the X^W from the father concealing the recessive X^w from the mother.

X-linked genetic disorders

The same principles we see at work in fruit flies can be applied to human genetics. In humans, the alleles for certain conditions (including some forms of color blindness, hemophilia, and muscular dystrophy) are X-linked. These diseases are much more common in men than they are in women due to their X-linked inheritance pattern.

Why is this the case? Let's explore this using an example in which a mother is heterozygous for a disease-causing allele. Women who are heterozygous for disease alleles are said to be **carriers**, and they usually don't display any symptoms themselves. Sons of these women have a 50% chance of getting the disorder, but daughters have little chance of getting the disorder (unless the father also has it), and will instead have a 50% chance of being carriers.

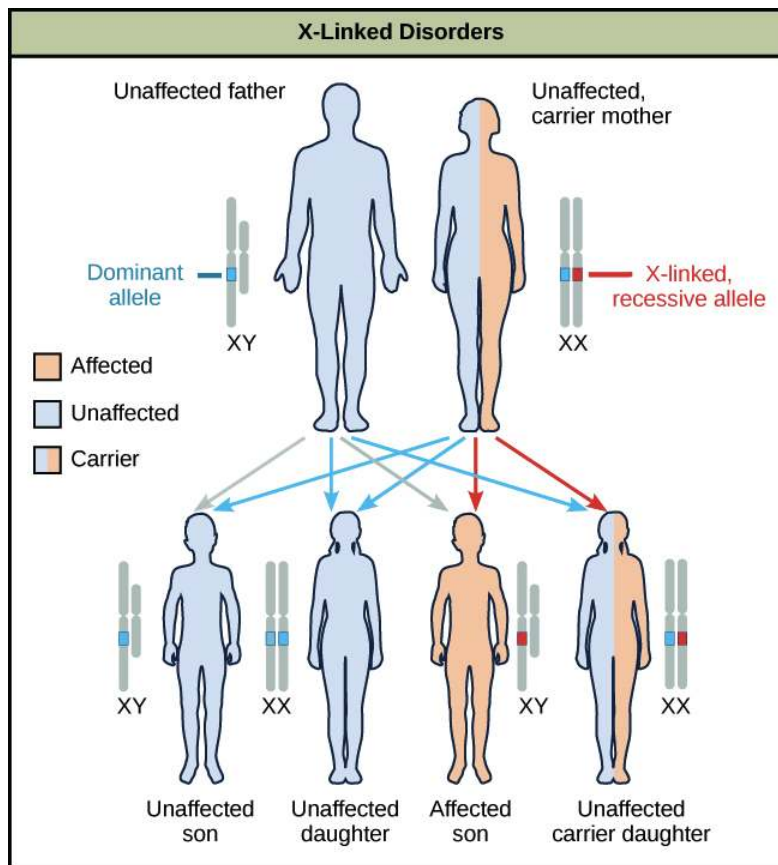


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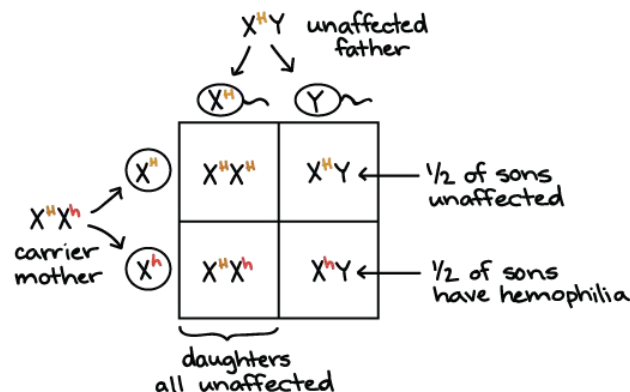
Why is this the case? Recessive X-linked traits appear more often in males than females because, if a male receives a "bad" allele from his mother, he has no chance of getting a "good" allele from his father (who provides a Y) to hide the bad one. Females, on the other hand, will often receive a normal allele from their fathers, preventing the disease allele from being expressed.

Case study: Hemophilia

Let's look at a Punnett square example using an X-linked human disorder: hemophilia, a recessive condition in which a person's blood does not clot properly¹³. A person with hemophilia may have severe, even life-threatening, bleeding from just a small cut.

Hemophilia is caused by a mutation in either of two genes, both of which are located on the X chromosome. Both genes encode proteins that help blood clot¹⁴. Let's focus on just one of these genes, calling the functional allele X^H and the disease allele X^h .

In our example, a woman who is heterozygous for normal and hemophilia alleles ($X^H X^h$) has children with a man who is hemizygous for the normal form ($X^H Y$). Both parents have normal blood clotting, but the mother is a carrier. What is the chance of their sons and daughters having hemophilia?



Since the mother is a carrier, she will pass on the hemophilia allele (X^h) on to half of her children, both boys and girls.

- None of the daughters will have hemophilia (zero chance of the disorder). That's because, in order to have the disorder, they must get a X^h allele from both their mother and their father. There is 0 chance of the daughters getting an X^h allele from their father, so their overall chance of having hemophilia is zero.
- The sons get a Y from their father instead of an X, so their only copy of the blood clotting gene comes from their mother. The mother is heterozygous, so half of the sons, on average, will get an X^h allele and have hemophilia (1/2 chance of the disorder).

[Can a woman ever have hemophilia?]

Yes, it's possible for a woman to have a recessive, X-linked condition such as hemophilia. However, she must get two recessive copies of the X-linked gene (one from each parent) in order to have the condition. The odds of this are much lower than the odds of a man getting just one recessive disease allele from his mother.

Check your understanding

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3.3: X-inactivation

Introduction

Having [extra or missing chromosomes](#) is not usually a good thing. In fact, for most chromosomes, having an extra or missing copy is lethal to humans (causing an embryo to die early in development).

Yet, human females have two X chromosomes (XX), while human males have just one (XY). Why doesn't it cause problems for men to have just one copy of the X chromosome, while women have two?

X-inactivation

As it turns out, the level of gene activity produced by a single X chromosome is the normal "dosage" for a human. Men have this dosage because, well, they only have one X chromosome! Women have the same dosage for a different reason: they shut down one of their two X chromosomes in a process called X-inactivation.

In **X-inactivation**, an X chromosome is compacted (or, as my intro bio professor liked to say, "crumpled up into a ball"), to make a small, dense structure called a **Barr body**. Most of the genes on the Barr body are inactive, meaning that they are not transcribed. The process of X-inactivation was discovered by the British geneticist Mary F. Lyon and is sometimes called **lyonization** in her honor¹.



Image modified from "Photo of an English geneticist, Mary Frances Lyon," by Jane Gitschier (CC BY 2.5).

A woman has two X chromosomes, one from each parent. Which one will she inactivate? X-inactivation is a random process that happens separately in individual cells during embryonic development. One cell might shut down the paternal X, while its next-door neighbor might shut down the maternal X instead. All the cells descended from each of these original cells will maintain the same pattern of X-inactivation.

Interesting note: if you were a kangaroo, what I just said would not be true! In kangaroos and other marsupials, it is always the paternal X chromosome that undergoes X-inactivation².

X-inactivation example: Calico cat

A classic example of X-inactivation is seen in cats. If a female cat is heterozygous for black and tan alleles of a coat color gene found on the X, she will inactivate her two Xs (and thus, the two alleles of the coat color gene) at random in different cells during development.

The result is a tortoiseshell coat pattern, made up of alternating patches of black and tan fur. The black patches come from groups of cells in which the X with the black allele is active, while the tan patches come from cells in which the X with the tan allele is

active.

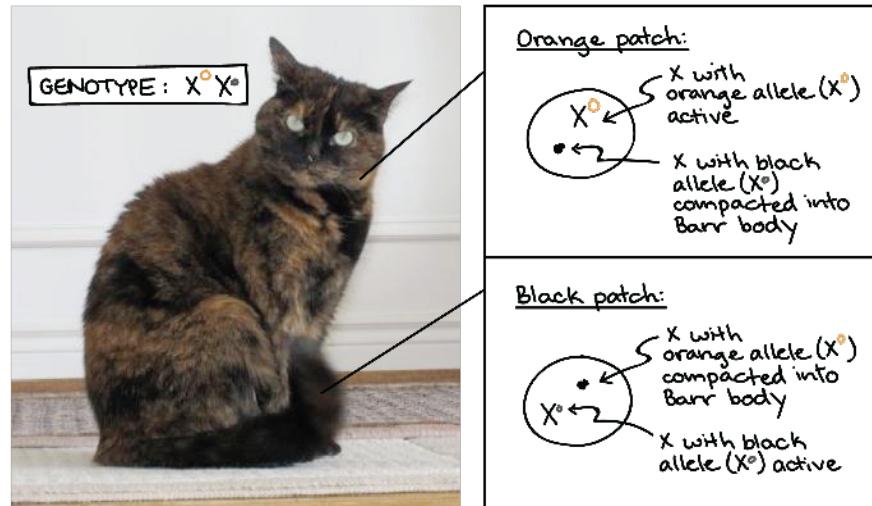


Image modified from "6-year old tortoiseshell cat," by Michael Bodega (public domain).

Although it's rarely as easy to see as in the case of the tortoiseshell cat, human females are also "mosaic" for any genes that are present in different alleles on their two X chromosomes.

[If that's true, why don't female carriers show X-linked disorders?]

At this point, you may be wondering why women heterozygous for a recessive, X-linked allele don't display the associated condition. After all, roughly half of their cells will inactivate the normal allele, leaving only the disorder-causing allele active.

In many cases, it appears that having 50% of cells with a normal copy of a gene is sufficient to produce a normal, or close to normal, phenotype.

A good example of this comes from the X-linked condition of red-green color vision deficiency (colorblindness). Women who are carriers for red-green color vision deficiency alleles have a mixture of working and non-working photoreceptor cells in their eyes. However, the working cells allow them to perceive red and green well enough to function normally in daily life. In more sensitive tests in a laboratory setting, these women actually do show subtle color vision deficiencies relative to women who are not carriers³.

Sex chromosome aneuploidies

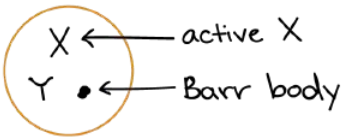
When an organism has an extra or missing copy of a chromosome, it is said to be **aneuploid**. Aneuploidies involving autosomes (non-sex chromosomes), especially large ones, are usually so harmful to development that an aneuploid embryo can't survive to birth.

Aneuploidies of X chromosomes, however, tend to be much less harmful, despite the fact that the X is a large chromosome. This is mostly thanks to X inactivation. Although the purpose of the X-inactivation system is to shut down the second X of an XX female, it can also do a pretty good job of shutting down more X chromosomes if they are present.

Examples of X chromosome aneuploidies include:

- **Triple X syndrome**, in which a woman has an XXX genotype, which occurs in about 1 out of every 1,000 female newborns⁴. Women with an XXX genotype have female sex characteristics and are fertile (able to have children). In some cases, triple X syndrome may be associated with learning difficulties, late development of motor skills in infants, and problems with muscle tone⁴.
- **Klinefelter syndrome**, in which males have an extra X chromosome, leading to a genotype of XXY. (In rarer cases, Klinefelter syndrome can involve several extra Xs, leading to an XXXY or XXXXY genotype.) Affected men may be infertile or develop less dense body and facial hair than other men. Klinefelter syndrome is thought to affect 1 out of every 500 to 1,000 male newborns⁵.

Like females, XXY males with Klinefelter syndrome will convert one X to a Barr body in each cell. Triple X females (as well as Klinefelter males with more than two X chromosomes) neutralize their extra Xs by forming additional Barr bodies. For example, there would be two Barr bodies in a cell from an XXX female or XXXY male.

XX female	
XY male	
XXY male (Klinefelter)	
XXX female (triple X)	

In **Turner syndrome**, a woman lacks part or all of one of her X chromosomes (leaving her with just one functional X). People with this disorder develop as females, but often have short stature and may exhibit symptoms like infertility and learning difficulties. Turner syndrome is thought to occur in about 1 out of every 2,500 female births⁶. It has relatively mild effects because humans normally have only one X active in the cells of their body anyway.

Check your understanding

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3.4: Sex linkage review

Key terms

Term	Meaning
Sex chromosome	One of two chromosomes that determines an organism's biological sex
Autosome	Chromosome that is not a sex chromosome
Sex-linked gene	Gene that is located on one of the two sex chromosomes
Carrier	Heterozygous individual that inherited a recessive allele for a genetic disorder but does not display symptoms of that disorder
Barr body	A condensed region in the nucleus of a cell, consisting of an inactivated X chromosome
Aneuploidy	Condition of having too many or too few chromosomes

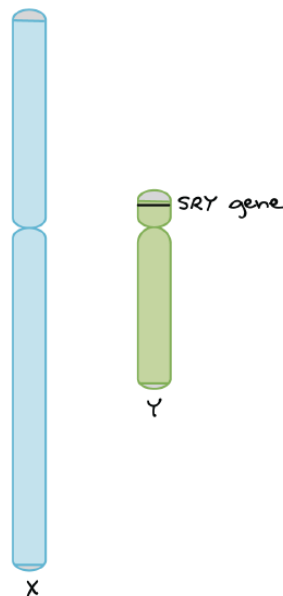
Sex linkage

In humans, biological sex is determined by a pair of **sex chromosomes**: *XX* in females and *XY* in males. The other 44 chromosomes are **autosomes**.

Genes on either the X or Y chromosome are **sex-linked traits**. Genes found on the X chromosome can be found in either males or females, while genes found on the Y chromosome can only be found in males.

X-linked inheritance

There are many more X-linked traits than Y-linked traits because the Y chromosome is much shorter and fewer genes than the X chromosome.



X-linked genes have distinctive inheritance patterns because they are present in different numbers in females (*XX*) and males (*XY*).

Females have two X chromosomes, so she will have two copies of each X-linked gene, giving her the opportunity to be either homozygous or heterozygous for each sex-linked gene.

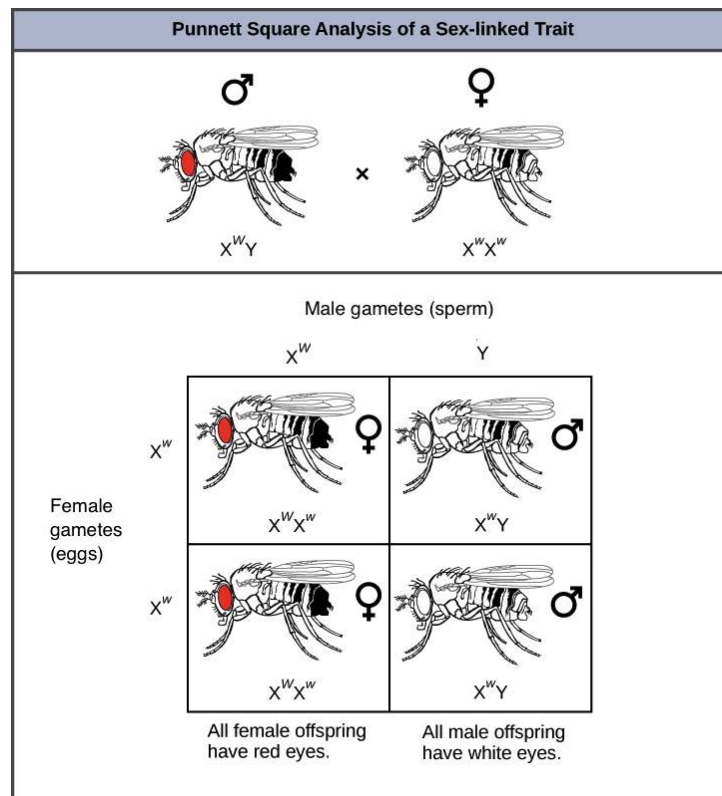


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X-linked disorders

X-linked human genetic disorders are much more common in males than in females. Since males only have one X chromosome, and therefore one copy of any X-linked genes, whatever allele the male inherits for an X-linked gene will be expressed.

An example of this is the blood-clotting disorder, hemophilia. Women who are heterozygous for hemophilia are **carriers**, and they usually don't display any symptoms themselves.

Sons of these women have a 50% chance of having hemophilia. Daughters have little chance of having hemophilia (unless the father also has it), and will instead have a 50% chance of being carriers.

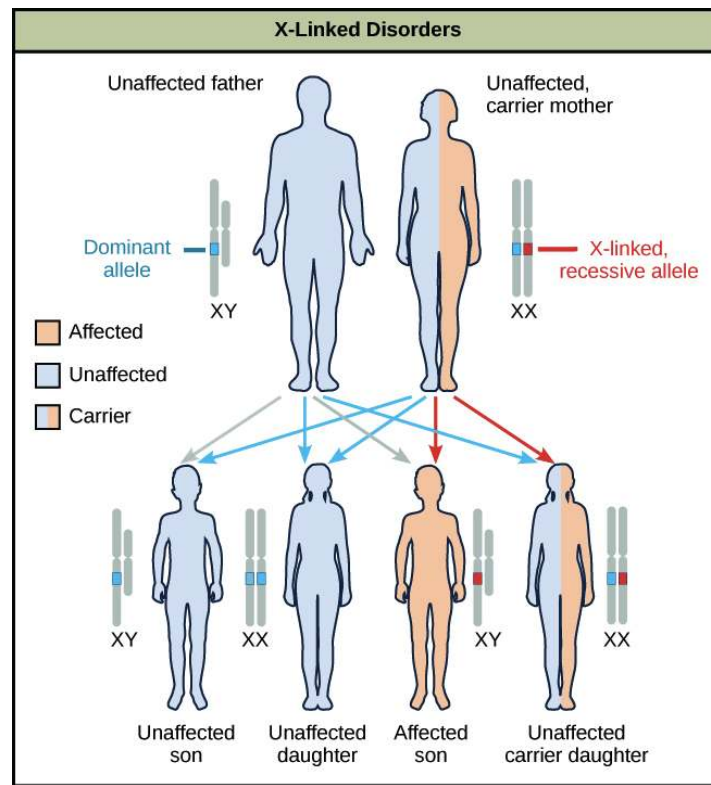


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X-inactivation

If males can survive with only one X chromosome, why doesn't it cause problems for women who have two X chromosomes?

As it turns out, for females, most of the genes in one of the X chromosomes is inactivated, forming a **Barr body**. This inactivation happens randomly during embryonic development.

Example:

A common example of X-inactivation is seen in cats. If a female cat is heterozygous for black and tan alleles of a coat color gene found on the X, two Xs (and thus, the two alleles of the coat color gene) will be inactivated at random in different cells during development.

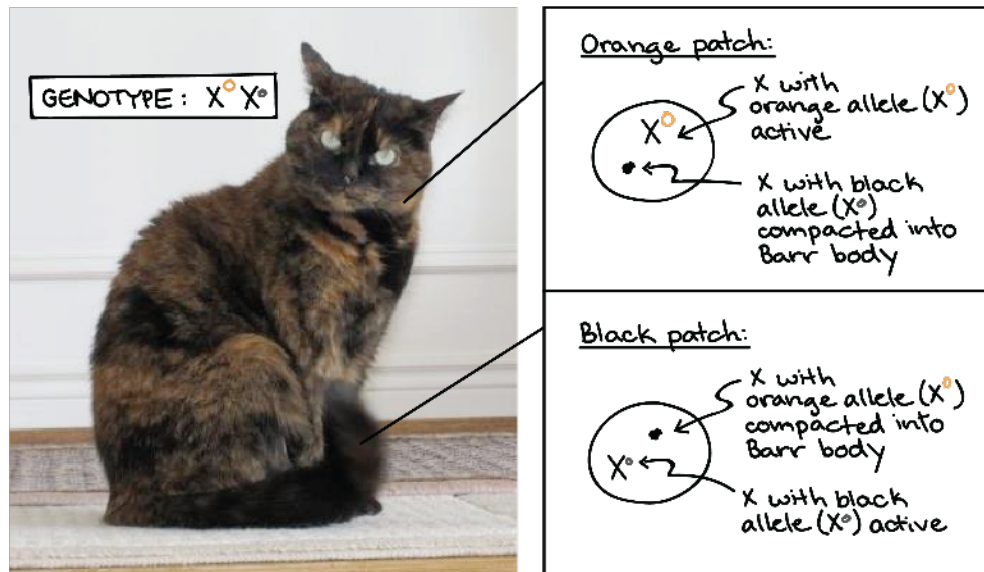
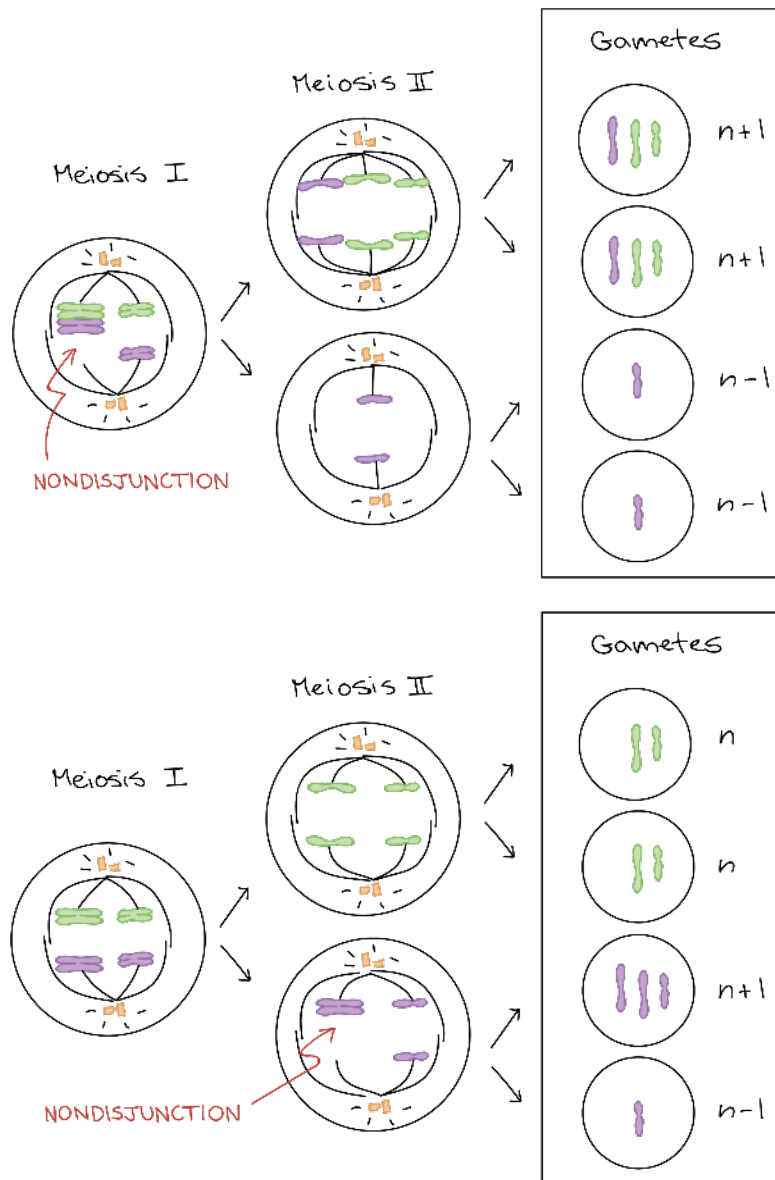


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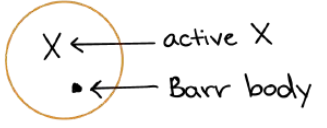
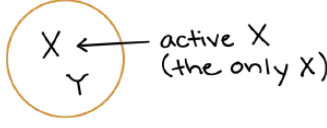
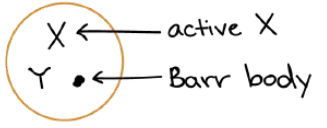
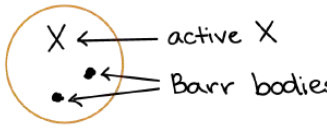
The result of this is a tortoiseshell coat pattern, made up of alternating patches of black and tan fur.

Sex chromosome aneuploidy

Aneuploidy, or disorders of chromosome number, are generally caused by *nondisjunction*. This occurs when pairs of homologous chromosomes or sister chromatids fail to separate during cell division.



Individuals that have autosomal **aneuploidy** rarely survive to birth. However, due to the size of the X chromosome and because of X-inactivation, X chromosome aneuploidies tend to be much less harmful.

XX female	
XY male	
XXY male (Klinefelter)	
XXX female (triple X)	

In *Klinefelter syndrome*, males have one or more extra X chromosomes, leading to a genotype of XXY. (Or in rare cases, XXXY or XXXXY!) Affected men may be infertile or develop less dense body and facial hair than other men.

Women affected with *Triple X syndrome* have an XXX genotype. Women with Triple X syndrome have female sex characteristics and are fertile (able to have children).

Women with *Turner syndrome* lack part or all of one of their X chromosomes (leaving her with just one functional X). People with this disorder develop as females, but often have short stature and may experience infertility and learning difficulties.

Common mistakes and misconceptions

- **Some people think that a recessive X-linked trait will show up more often in women because they have two X chromosomes.** However, women are less likely to express recessive X-linked traits because there is potential for a "good" allele to mask a "bad" allele. On the other hand, if a male receives a "bad" allele from his mother, he has no chance of getting a "good" allele from his father (who provides a Y) to hide the bad one.
- **Codominance and X-inactivation are not the same.** Although these two concepts may result in similar looking organisms, a heterozygous individual expressing a codominant trait will express both alleles fully and separately. In X-inactivation, females express only one X chromosome in each cell, meaning that genes on the X chromosome are expressed singly instead of in a pair. Because the inactivated X chromosome is not the same in every cell, neighboring cells may express different proteins if different X chromosomes carry different alleles.

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CHAPTER OVERVIEW

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[4.2: Pedigree for determining probability of exhibiting sex linked recessive trait](#)

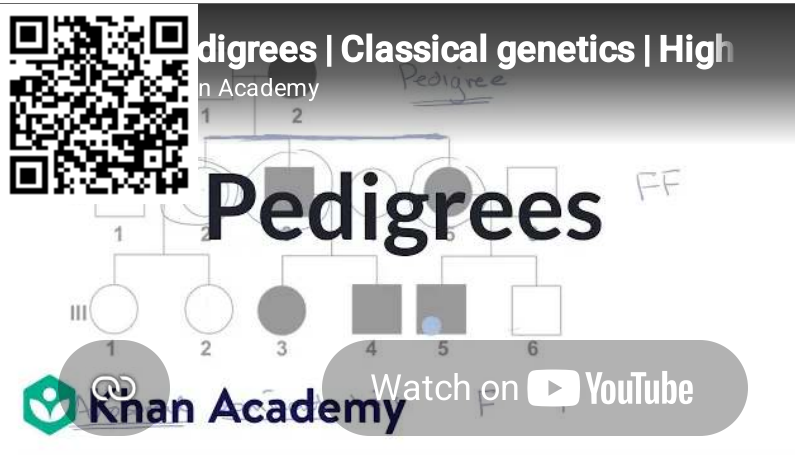
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Thumbnail: Pedigree for an autosomal dominant trait. (CC BY-NC-SA; [Khan Academy](#)).

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4.1: Pedigrees



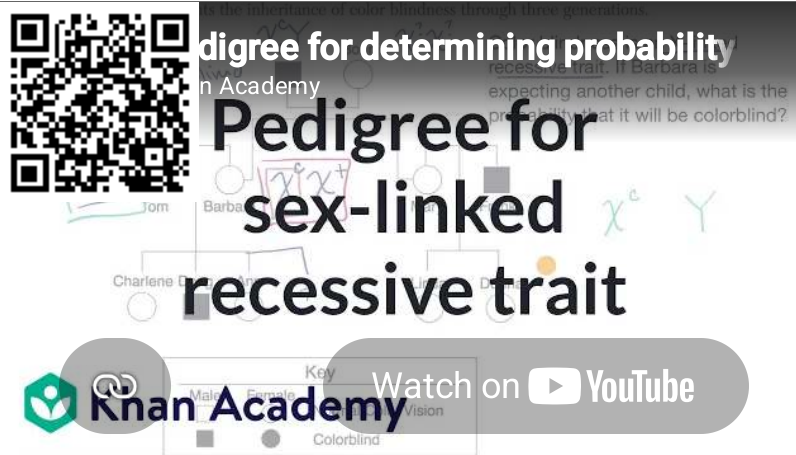
An introduction to reading and analyzing pedigrees.

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Pedigree for determining probability of exhibiting sex linked recessive trait.

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4.3: Pedigrees review

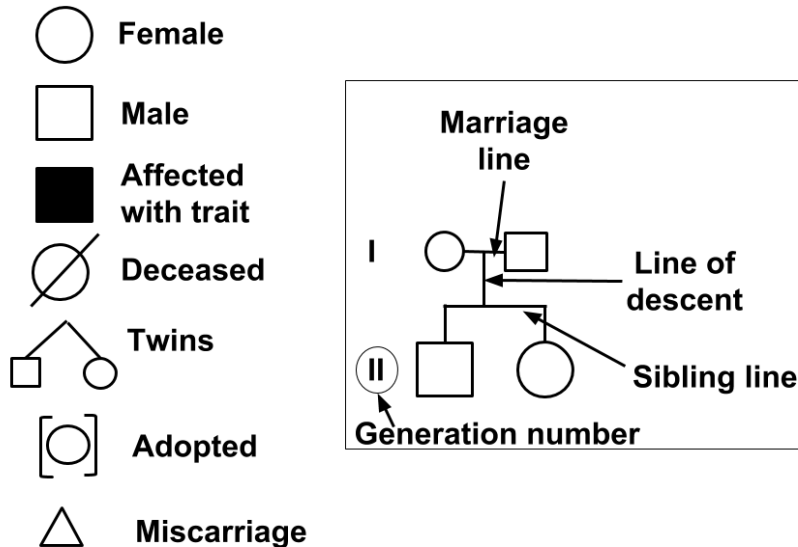
Key terms

Term	Meaning
Pedigree	Chart that shows the presence or absence of a trait within a family across generations
Genotype	The genetic makeup of an organism (ex: TT)
Phenotype	The physical characteristics of an organism (ex: tall)
Dominant allele	Allele that is phenotypically expressed over another allele
Recessive allele	Allele that is only expressed in absence of a dominant allele
Autosomal trait	Trait that is located on an autosome (non-sex chromosome)
Sex-linked trait	Trait that is located on one of the two sex chromosomes
Homozygous	Having two identical alleles for a particular gene
Heterozygous	Having two different alleles for a particular gene

Pedigrees

Pedigrees are used to analyze the pattern of inheritance of a particular trait throughout a family. Pedigrees show the presence or absence of a trait as it relates to the relationship among parents, offspring, and siblings.

Reading a pedigree



Common pedigree symbols.

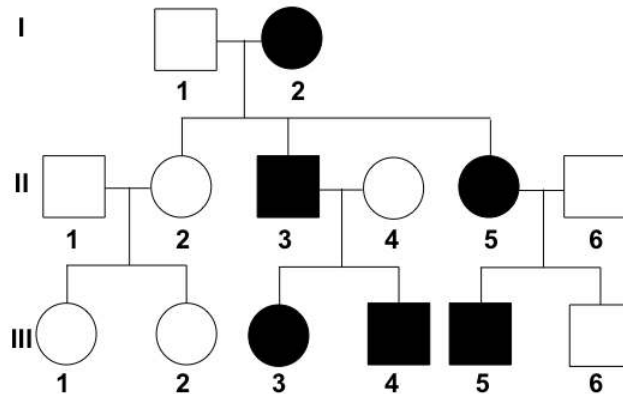
Pedigrees represent family members and relationships using standardized symbols.

By analyzing a pedigree, we can determine **genotypes**, identify **phenotypes**, and predict how a trait will be passed on in the future. The information from a pedigree makes it possible to determine how certain alleles are inherited: whether they are **dominant**, **recessive**, **autosomal**, or **sex-linked**.

To start reading a pedigree:

1. **Determine whether the trait is dominant or recessive.** If the trait is dominant, one of the parents *must* have the trait. Dominant traits will not skip a generation. If the trait is recessive, neither parent is required to have the trait since they can be heterozygous.
2. **Determine if the chart shows an autosomal or sex-linked (usually X-linked) trait.** For example, in X-linked recessive traits, males are much more commonly affected than females. In autosomal traits, both males and females are equally likely to be affected (usually in equal proportions).

Example: Autosomal dominant trait



The diagram shows the inheritance of freckles in a family. The allele for freckles (**F**) is dominant to the allele for no freckles (**f**).

At the top of the pedigree is a grandmother (individual I-2) who has freckles. Two of her three children have the trait (individuals II-3 and II-5) and three of her grandchildren have the trait (individuals III-3, III-4, and III-5).

[What is the genotype of individual I-2?]

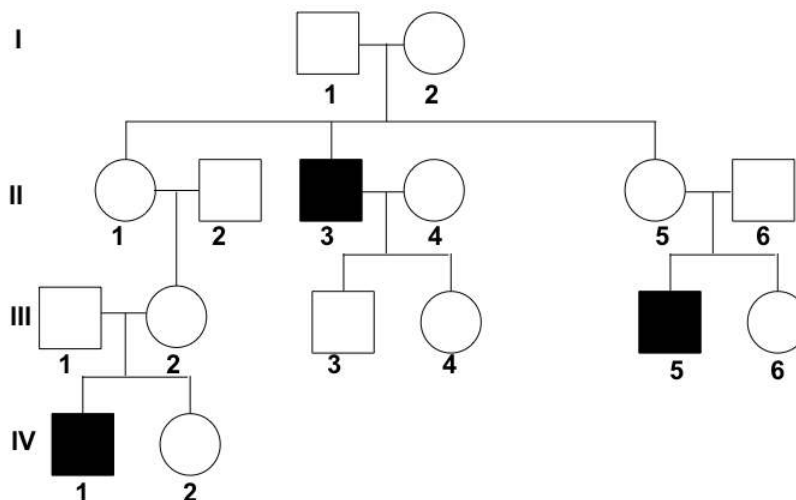
Since freckles are dominant to no freckles, an affected individual such as I-2 must at least have one **F** allele.

The trait shows up in all generations and affects both males and females equally. This suggests that it is an autosomal dominant trait.

Unaffected individuals must have two recessive alleles (**ff**) in order to not have freckles. If we notice, I-2 has some children who do *not* have freckles. In order to produce children with a genotype of **ff**, I-2 must be able to donate a **f** allele.

We can therefore conclude that her genotype is **Ff**.

Example: X-linked recessive trait



The diagram shows the inheritance of colorblindness in a family. Colorblindness is a recessive and X-linked trait (X^b). The allele for normal vision is dominant and is represented by X^B .

In generation I, neither parent has the trait, but one of their children (II-3) is colorblind. Because there are unaffected parents that have affected offspring, it can be assumed that the trait is recessive. In addition, the trait appears to affect males more than females (in this case, exclusively males are affected), suggesting that the trait may be X-linked.

[What is the genotype of individual III-2?]

We can determine the genotype of III-2 by looking at her children. Since she is an unaffected female, she must have at least one normal vision allele (X^B). Her two genotype options are then $X^B X^B$ or $X^B X^b$.

However, her son (IV-1) is colorblind, meaning that he has a genotype of $X^b Y$. Because males always get their X chromosome from their mothers (and their Y from their fathers), his colorblind allele must come from III-2.

We can then determine that III-2's genotype is $X^B X^b$, so she can pass the X^b on to her son.

Common mistakes and misconceptions

- **The presence of many affected individuals in a family does not always mean that the trait is dominant.** The terms dominant and recessive refer to the way that a trait is expressed, not by how often it shows up in a family. In fact, although it is uncommon, a trait may be recessive but still show up in all generations of a pedigree.
- **You may not always be able to determine the genotype of an individual based on a pedigree.** Sometimes an individual can either be homozygous dominant or heterozygous for a trait. Often, we can use the relationships between an individual and their parents, siblings, and offspring to determine genotypes. However, not all carriers are always explicitly indicated in a pedigree, and it may not be possible to determine based on the information provided.

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Autosomal recessive trait

X-linked recessive trait

Autosomal dominant trait

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