

CHAPTER 12

Mendel's Experiments and Heredity



Figure 12.1 Experimenting with thousands of garden peas, Mendel uncovered the fundamentals of genetics. (credit: modification of work by Jerry Kirkhart)

INTRODUCTION Genetics is the study of heredity. Johann Gregor Mendel set the framework for genetics long before chromosomes or genes had been identified, at a time when meiosis was not well understood. Mendel selected a simple biological system and conducted methodical, quantitative analyses using large sample sizes. Because of Mendel's work, the fundamental principles of heredity were revealed. We now know that genes, carried on chromosomes, are the basic functional units of heredity with the capability to be replicated, expressed, or mutated. Today, the postulates put forth by Mendel form the basis of classical, or Mendelian, genetics. Not all genes are transmitted from parents to offspring according to Mendelian genetics, but Mendel's experiments serve as an excellent starting point for thinking about inheritance.

Chapter Outline

- 12.1 Mendel's Experiments and the Laws of Probability
- 12.2 Characteristics and Traits
- 12.3 Laws of Inheritance

12.1 Mendel's Experiments and the Laws of Probability

By the end of this section, you will be able to do the following:

- Describe the scientific reasons for the success of Mendel's experimental work
- Describe the expected outcomes of monohybrid crosses involving dominant and recessive alleles
- Apply the sum and product rules to calculate probabilities



Figure 12.2 Johann Gregor Mendel is considered the father of genetics.

Johann Gregor Mendel (1822–1884) (Figure 12.2) was a lifelong learner, teacher, scientist, and man of faith. As a young adult, he joined the Augustinian Abbey of St. Thomas in Brno in what is now the Czech Republic. Supported by the monastery, he taught physics, botany, and natural science courses at the secondary and university levels. In 1856, he began a decade-long research pursuit involving inheritance patterns in honeybees and plants, ultimately settling on pea plants as his primary **model system** (a system with convenient characteristics used to study a specific biological phenomenon to be applied to other systems). In 1865, Mendel presented the results of his experiments with nearly 30,000 pea plants to the local Natural History Society. He demonstrated that traits are transmitted from parents to offspring independently of other traits and in dominant and recessive patterns. In 1866, he published his work, *Experiments in Plant Hybridization*,¹ in the proceedings of the Natural History Society of Brünn.

Mendel's work went virtually unnoticed by the scientific community, which believed, incorrectly, that the process of inheritance involved a blending of parental traits that produced an intermediate physical appearance in offspring. The **blending theory of inheritance** asserted that the original parental traits were lost or absorbed by the blending in the offspring, but we now know that this is not the case. This hypothetical process appeared to be correct because of what we know now as continuous variation. **Continuous variation** results from the action of many genes to determine a characteristic like human height. Offspring appear to be a “blend” of their parents' traits.

Instead of continuous characteristics, Mendel worked with traits that were inherited in distinct classes (specifically, violet versus white flowers); this is referred to as **discontinuous variation**. Mendel's choice of these kinds of traits allowed him to see experimentally that the traits were not blended in the offspring, nor

¹Johann Gregor Mendel, *Versuche über Pflanzenhybriden Verhandlungen des naturforschenden Vereines in Brünn, Bd. IV für das Jahr, 1865 Abhandlungen, 3–47.* [For English translation see <http://www.mendelweb.org/Mendel.plain.html> (http://openstax.org/l/mendel_experiments)]

were they absorbed, but rather that they kept their distinctness and could be passed on. In 1868, Mendel became abbot of the monastery and exchanged his scientific pursuits for his pastoral duties. He was not recognized for his extraordinary scientific contributions during his lifetime. In fact, it was not until 1900 that his work was rediscovered, reproduced, and revitalized by scientists on the brink of discovering the chromosomal basis of heredity.

Mendel's Model System

Mendel's seminal work was accomplished using the garden pea, *Pisum sativum*, to study inheritance. This species naturally self-fertilizes, such that pollen encounters ova within individual flowers. The flower petals remain sealed tightly until after pollination, preventing pollination from other plants. The result is highly inbred, or "true-breeding," pea plants. These are plants that always produce offspring that look like the parent. By experimenting with true-breeding pea plants, Mendel avoided the appearance of unexpected traits in offspring that might occur if the plants were not true breeding. The garden pea also grows to maturity within one season, meaning that several generations could be evaluated over a relatively short time. Finally, large quantities of garden peas could be cultivated simultaneously, allowing Mendel to conclude that his results did not come about simply by chance.

Mendelian Crosses

Mendel performed **hybridizations**, which involve mating two true-breeding individuals that have different traits. In the pea, which is naturally self-pollinating, this is done by manually transferring pollen from the anther of a mature pea plant of one variety to the stigma of a separate mature pea plant of the second variety. In plants, pollen carries the male gametes (sperm) to the stigma, a sticky organ that traps pollen and allows the sperm to move down the pistil to the female gametes (ova) below. To prevent the pea plant that was receiving pollen from self-fertilizing and confounding his results, Mendel painstakingly removed all of the anthers from the plant's flowers before they had a chance to mature.

Plants used in first-generation crosses were called **P₀**, or parental generation one ([Figure 12.3](#)). After each cross, Mendel collected the seeds belonging to the P₀ plants and grew them the following season. These offspring were called the **F₁**, or the first filial (*filial* = offspring, daughter or son) generation. Once Mendel examined the characteristics in the F₁ generation of plants, he allowed them to self-fertilize naturally. He then collected and grew the seeds from the F₁ plants to produce the **F₂**, or second filial, generation. Mendel's experiments extended beyond the F₂ generation to the F₃ and F₄ generations, and so on, but it was the ratio of characteristics in the P₀–F₁–F₂ generations that were the most intriguing and became the basis for Mendel's postulates.

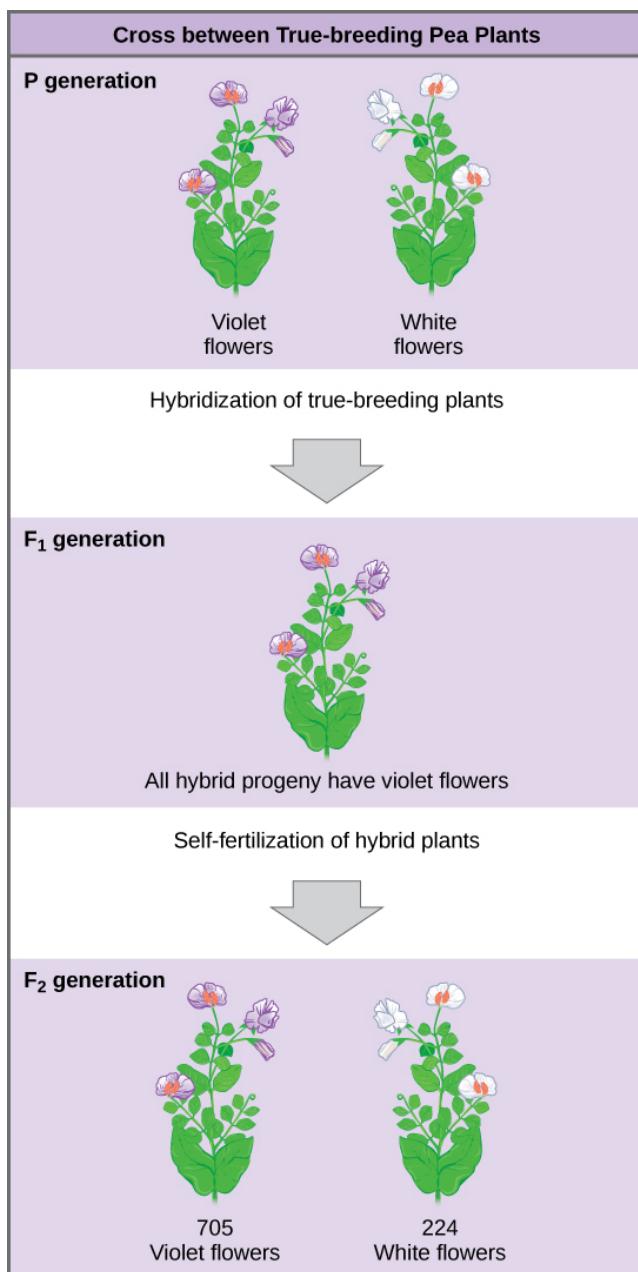


Figure 12.3 In one of his experiments on inheritance patterns, Mendel crossed plants that were true-breeding for violet flower color with plants true-breeding for white flower color (the P generation). The resulting hybrids in the F₁ generation all had violet flowers. In the F₂ generation, approximately three quarters of the plants had violet flowers, and one quarter had white flowers.

Garden Pea Characteristics Revealed the Basics of Heredity

In his 1865 publication, Mendel reported the results of his crosses involving seven different characteristics, each with two contrasting traits. A **trait** is defined as a variation in the physical appearance of a heritable characteristic. The characteristics included plant height, seed texture, seed color, flower color, pea pod size, pea pod color, and flower position. For the characteristic of flower color, for example, the two contrasting traits were white versus violet. To fully examine each characteristic, Mendel generated large numbers of F₁ and F₂ plants, reporting results from 19,959 F₂ plants alone. His findings were consistent.

What results did Mendel find in his crosses for flower color? First, Mendel confirmed that he had plants that bred true for white or violet flower color. Regardless of how many generations Mendel examined, all self-crossed offspring of parents with white flowers had white flowers, and all self-crossed offspring of parents with violet flowers had violet flowers. In addition, Mendel

confirmed that, other than flower color, the pea plants were physically identical.

Once these validations were complete, Mendel applied the pollen from a plant with violet flowers to the stigma of a plant with white flowers. After gathering and sowing the seeds that resulted from this cross, *Mendel found that 100 percent of the F₁ hybrid generation had violet flowers*. Conventional wisdom at that time (the blending theory) would have predicted the hybrid flowers to be pale violet or for hybrid plants to have equal numbers of white and violet flowers. In other words, the contrasting parental traits were expected to blend in the offspring. Instead, Mendel's results demonstrated that the white flower trait in the F₁ generation had completely disappeared.

Importantly, Mendel did not stop his experimentation there. He allowed the F₁ plants to self-fertilize and found that, of F₂-generation plants, 705 had violet flowers and 224 had white flowers. This was a ratio of 3.15 violet flowers per one white flower, or approximately 3:1. When Mendel transferred pollen from a plant with violet flowers to the stigma of a plant with white flowers and vice versa, he obtained about the same ratio regardless of which parent, male or female, contributed which trait. This is called a **reciprocal cross**—a paired cross in which the respective traits of the male and female in one cross become the respective traits of the female and male in the other cross. For the other six characteristics Mendel examined, the F₁ and F₂ generations behaved in the same way as they had for flower color. One of the two traits would disappear completely from the F₁ generation only to reappear in the F₂ generation at a ratio of approximately 3:1 ([Table 12.1](#)).

The Results of Mendel's Garden Pea Hybridizations

Characteristic	Contrasting P ₀ Traits	F ₁ Offspring Traits	F ₂ Offspring Traits	F ₂ Trait Ratios
Flower color	Violet vs. white	100 percent violet	705 violet 224 white	3.15:1
Flower position	Axial vs. terminal	100 percent axial	651 axial 207 terminal	3.14:1
Plant height	Tall vs. dwarf	100 percent tall	787 tall 277 dwarf	2.84:1
Seed texture	Round vs. wrinkled	100 percent round	5,474 round 1,850 wrinkled	2.96:1
Seed color	Yellow vs. green	100 percent yellow	6,022 yellow 2,001 green	3.01:1
Pea pod texture	Inflated vs. constricted	100 percent inflated	882 inflated 299 constricted	2.95:1
Pea pod color	Green vs. yellow	100 percent green	428 green 152 yellow	2.82:1

Table 12.1

Upon compiling his results for many thousands of plants, Mendel concluded that the characteristics could be divided into expressed and latent traits. He called these, respectively, dominant and recessive traits. **Dominant traits** are those that are inherited unchanged in a hybridization. **Recessive traits** become latent, or disappear, in the offspring of a hybridization. The recessive trait does, however, reappear in the progeny of the hybrid offspring. An example of a dominant trait is the violet-flower trait. For this same characteristic (flower color), white-colored flowers are a recessive trait. The fact that the recessive trait

reappeared in the F_2 generation meant that the traits remained separate (not blended) in the plants of the F_1 generation. Mendel also proposed that plants possessed two copies of the trait for the flower-color characteristic, and that each parent transmitted one of its two copies to its offspring, where they came together. Moreover, the physical observation of a dominant trait could mean that the genetic composition of the organism included two dominant versions of the characteristic or that it included one dominant and one recessive version. Conversely, the observation of a recessive trait meant that the organism lacked any dominant versions of this characteristic.

So why did Mendel repeatedly obtain 3:1 ratios in his crosses? To understand how Mendel deduced the basic mechanisms of inheritance that lead to such ratios, we must first review the laws of probability.

Probability Basics

Probabilities are mathematical measures of likelihood. The empirical probability of an event is calculated by dividing the number of times the event occurs by the total number of opportunities for the event to occur. It is also possible to calculate theoretical probabilities by dividing the number of times that an event is *expected* to occur by the number of times that it could occur. Empirical probabilities come from observations, like those of Mendel. Theoretical probabilities, on the other hand, come from knowing how the events are produced and assuming that the probabilities of individual outcomes are equal. A probability of one for some event indicates that it is guaranteed to occur, whereas a probability of zero indicates that it is guaranteed not to occur. An example of a genetic event is a round seed produced by a pea plant.

In one experiment, Mendel demonstrated that the probability of the event “round seed” occurring was one in the F_1 offspring of true-breeding parents, one of which has round seeds and one of which has wrinkled seeds. When the F_1 plants were subsequently self-crossed, the probability of any given F_2 offspring having round seeds was now three out of four. In other words, in a large population of F_2 offspring chosen at random, 75 percent were expected to have round seeds, whereas 25 percent were expected to have wrinkled seeds. Using large numbers of crosses, Mendel was able to calculate probabilities and use these to predict the outcomes of other crosses.

The Product Rule and Sum Rule

Mendel demonstrated that pea plants transmit characteristics as discrete units from parent to offspring. As will be discussed, Mendel also determined that different characteristics, like seed color and seed texture, were transmitted independently of one another and could be considered in separate probability analyses. For instance, performing a cross between a plant with green, wrinkled seeds and a plant with yellow, round seeds still produced offspring that had a 3:1 ratio of yellow:green seeds (ignoring seed texture) and a 3:1 ratio of wrinkled:round seeds (ignoring seed color). The characteristics of color and texture did not influence each other.

The **product rule** of probability can be applied to this phenomenon of the independent transmission of characteristics. The product rule states that the probability of two independent events occurring together can be calculated by multiplying the individual probabilities of each event occurring alone. To demonstrate the product rule, imagine that you are rolling a six-sided die (D) and flipping a penny (P) at the same time. The die may roll any number from 1–6 (D_6), whereas the penny may turn up heads (P_H) or tails (P_T). The outcome of rolling the die has no effect on the outcome of flipping the penny and vice versa. There are 12 possible outcomes of this action (Table 12.2), and each event is expected to occur with equal probability.

Twelve Equally Likely Outcomes of Rolling a Die and Flipping a Penny

Rolling Die	Flipping Penny
D_1	P_H
D_1	P_T
D_2	P_H
D_2	P_T
D_3	P_H
D_3	P_T
D_4	P_H
D_4	P_T
D_5	P_H
D_5	P_T
D_6	P_H
D_6	P_T

Table 12.2

Rolling Die	Flipping Penny
D_3	P_H
D_3	P_T
D_4	P_H
D_4	P_T
D_5	P_H
D_5	P_T
D_6	P_H
D_6	P_T

Table 12.2

Of the 12 possible outcomes, the die has a 2/12 (or 1/6) probability of rolling a two, and the penny has a 6/12 (or 1/2) probability of coming up heads. By the product rule, the probability that you will obtain the combined outcome 2 and heads is: $(D_2) \times (P_H) = (1/6) \times (1/2)$ or 1/12 ([Table 12.3](#)). Notice the word “and” in the description of the probability. The “and” is a signal to apply the product rule. For example, consider how the product rule is applied to the dihybrid cross: the probability of having both dominant traits (for example, yellow and round) in the F_2 progeny is the product of the probabilities of having the dominant trait for each characteristic, as shown here:

$$\frac{3}{4} \times \frac{3}{4} = \frac{9}{16}$$

On the other hand, the **sum rule** of probability is applied when considering two mutually exclusive outcomes that can come about by more than one pathway. The sum rule states that the probability of the occurrence of one event or the other event, of two mutually exclusive events, is the sum of their individual probabilities. Notice the word “or” in the description of the probability. The “or” indicates that you should apply the sum rule. In this case, let’s imagine you are flipping a penny (P) and a quarter (Q). What is the probability of one coin coming up heads and one coin coming up tails? This outcome can be achieved by two cases: the penny may be heads (P_H) and the quarter may be tails (Q_T), or the quarter may be heads (Q_H) and the penny may be tails (P_T). Either case fulfills the outcome. By the sum rule, we calculate the probability of obtaining one head and one tail as $[(P_H) \times (Q_T)] + [(Q_H) \times (P_T)] = [(1/2) \times (1/2)] + [(1/2) \times (1/2)] = 1/2$ ([Table 12.3](#)). You should also notice that we used the product rule to calculate the probability of P_H and Q_T , and also the probability of P_T and Q_H , before we summed them. Again, the sum rule can be applied to show the probability of having at least one dominant trait in the F_2 generation of a dihybrid cross:

$$(\frac{1}{4} \times \frac{3}{4}) + (\frac{3}{4} \times \frac{1}{4}) = \frac{3}{16} + \frac{3}{16} = \frac{6}{16} = \frac{3}{8}$$

The Product Rule and Sum Rule

Product Rule	Sum Rule
For independent events A and B, the probability (P) of them both occurring (A <i>and</i> B) is $(P_A \times P_B)$	For mutually exclusive events A and B, the probability (P) that at least one occurs (A <i>or</i> B) is $(P_A + P_B)$

Table 12.3

To use probability laws in practice, we must work with large sample sizes because small sample sizes are prone to deviations

caused by chance. The large quantities of pea plants that Mendel examined allowed him calculate the probabilities of the traits appearing in his F_2 generation. As you will learn, this discovery meant that when parental traits were known, the offspring's traits could be predicted accurately even before fertilization.

12.2 Characteristics and Traits

By the end of this section, you will be able to do the following:

- Explain the relationship between genotypes and phenotypes in dominant and recessive gene systems
- Develop a Punnett square to calculate the expected proportions of genotypes and phenotypes in a monohybrid cross
- Explain the purpose and methods of a test cross
- Identify non-Mendelian inheritance patterns such as incomplete dominance, codominance, recessive lethals, multiple alleles, and sex linkage

Physical characteristics are expressed through genes carried on chromosomes. The genetic makeup of peas consists of two similar, or homologous, copies of each chromosome, one from each parent. Each pair of homologous chromosomes has the same linear order of genes. In other words, peas are diploid organisms in that they have two copies of each chromosome. The same is true for many other plants and for virtually all animals. Diploid organisms produce haploid gametes, which contain one copy of each homologous chromosome that unite at fertilization to create a diploid zygote.

For cases in which a single gene controls a single characteristic, a diploid organism has two genetic copies that may or may not encode the same version of that characteristic. Gene variants that arise by mutation and exist at the same relative locations on homologous chromosomes are called **alleles**. Mendel examined the inheritance of genes with just two allele forms, but it is common to encounter more than two alleles for any given gene in a natural population.

Phenotypes and Genotypes

Two alleles for a given gene in a diploid organism are expressed and interact to produce physical characteristics. The observable traits expressed by an organism are referred to as its **phenotype**. An organism's underlying genetic makeup, consisting of both physically visible and non-expressed alleles, is called its **genotype**. Mendel's hybridization experiments demonstrate the difference between phenotype and genotype. When true-breeding plants in which one parent had yellow pods and one had green pods were cross-fertilized, all of the F_1 hybrid offspring had yellow pods. That is, the hybrid offspring were phenotypically identical to the true-breeding parent with yellow pods. However, we know that the allele donated by the parent with green pods was not simply lost because it reappeared in some of the F_2 offspring. Therefore, the F_1 plants must have been genotypically different from the parent with yellow pods.

The P_1 plants that Mendel used in his experiments were each homozygous for the trait he was studying. Diploid organisms that are **homozygous** at a given gene, or locus, have two identical alleles for that gene on their homologous chromosomes. Mendel's parental pea plants always bred true because both of the gametes produced carried the same trait. When P_1 plants with contrasting traits were cross-fertilized, all of the offspring were **heterozygous** for the contrasting trait, meaning that their genotype reflected that they had different alleles for the gene being examined.

Dominant and Recessive Alleles

Our discussion of homozygous and heterozygous organisms brings us to why the F_1 heterozygous offspring were identical to one of the parents, rather than expressing both alleles. In all seven pea-plant characteristics, one of the two contrasting alleles was dominant, and the other was recessive. Mendel called the dominant allele the expressed unit factor; the recessive allele was referred to as the latent unit factor. We now know that these so-called unit factors are actually genes on homologous chromosome pairs. For a gene that is expressed in a dominant and recessive pattern, homozygous dominant and heterozygous organisms will look identical (that is, they will have different genotypes but the same phenotype). The recessive allele will only be observed in homozygous recessive individuals ([Table 12.4](#)).

Human Inheritance in Dominant and Recessive Patterns

Dominant Traits	Recessive Traits
Achondroplasia	Albinism
Brachydactyly	Cystic fibrosis
Huntington's disease	Duchenne muscular dystrophy
Marfan syndrome	Galactosemia
Neurofibromatosis	Phenylketonuria
Widow's peak	Sickle-cell anemia
Wooly hair	Tay-Sachs disease

Table 12.4

Several conventions exist for referring to genes and alleles. For the purposes of this chapter, we will abbreviate genes using the first letter of the gene's corresponding dominant trait. For example, violet is the dominant trait for a pea plant's flower color, so the flower-color gene would be abbreviated as *V* (note that it is customary to italicize gene designations). Furthermore, we will use uppercase and lowercase letters to represent dominant and recessive alleles, respectively. Therefore, we would refer to the genotype of a homozygous dominant pea plant with violet flowers as *VV*, a homozygous recessive pea plant with white flowers as *vv*, and a heterozygous pea plant with violet flowers as *Vv*.

The Punnett Square Approach for a Monohybrid Cross

When fertilization occurs between two true-breeding parents that differ in only one characteristic, the process is called a **monohybrid** cross, and the resulting offspring are monohybrids. Mendel performed seven monohybrid crosses involving contrasting traits for each characteristic. On the basis of his results in *F*₁ and *F*₂ generations, Mendel postulated that each parent in the monohybrid cross contributed one of two paired unit factors to each offspring, and every possible combination of unit factors was equally likely.

To demonstrate a monohybrid cross, consider the case of true-breeding pea plants with yellow versus green pea seeds. The dominant seed color is yellow; therefore, the parental genotypes were *YY* for the plants with yellow seeds and *yy* for the plants with green seeds, respectively. A **Punnett square**, devised by the British geneticist Reginald Punnett, can be drawn that applies the rules of probability to predict the possible outcomes of a genetic cross or mating and their expected frequencies. To prepare a Punnett square, all possible combinations of the parental alleles are listed along the top (for one parent) and side (for the other parent) of a grid, representing their meiotic segregation into haploid gametes. Then the combinations of egg and sperm are made in the boxes in the table to show which alleles are combining. Each box then represents the diploid genotype of a zygote, or fertilized egg, that could result from this mating. Because each possibility is equally likely, genotypic ratios can be determined from a Punnett square. If the pattern of inheritance (dominant or recessive) is known, the phenotypic ratios can be inferred as well. For a monohybrid cross of two true-breeding parents, each parent contributes one type of allele. In this case, only one genotype is possible. All offspring are *Yy* and have yellow seeds ([Figure 12.4](#)).

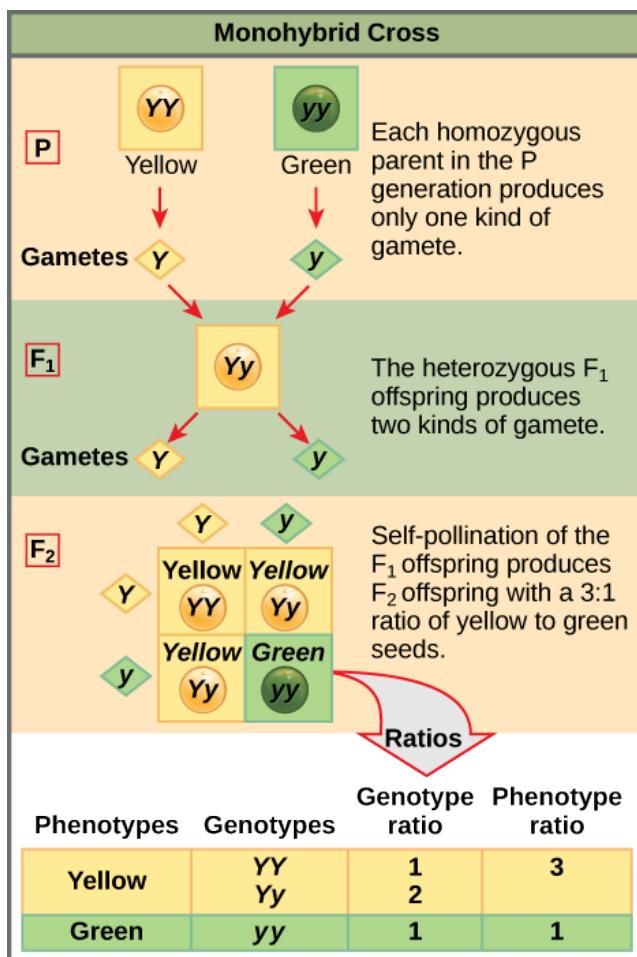


Figure 12.4 In the P generation, pea plants that are true-breeding for the dominant yellow phenotype are crossed with plants with the recessive green phenotype. This cross produces F₁ heterozygotes with a yellow phenotype. Punnett square analysis can be used to predict the genotypes of the F₂ generation.

A self-cross of one of the Yy heterozygous offspring can be represented in a 2×2 Punnett square because each parent can donate one of two different alleles. Therefore, the offspring can potentially have one of four allele combinations: YY, Yy, yY, or yy (Figure 12.4). Notice that there are two ways to obtain the Yy genotype: a Y from the egg and a y from the sperm, or a y from the egg and a Y from the sperm. Both of these possibilities must be counted. Recall that Mendel's pea-plant characteristics behaved in the same way in reciprocal crosses. Therefore, the two possible heterozygous combinations produce offspring that are genotypically and phenotypically identical despite their dominant and recessive alleles deriving from different parents. They are grouped together. Because fertilization is a random event, we expect each combination to be equally likely and for the offspring to exhibit a ratio of YY:Yy:yy genotypes of 1:2:1 (Figure 12.4). Furthermore, because the YY and Yy offspring have yellow seeds and are phenotypically identical, applying the sum rule of probability, we expect the offspring to exhibit a phenotypic ratio of 3 yellow:1 green. Indeed, working with large sample sizes, Mendel observed approximately this ratio in every F₂ generation resulting from crosses for individual traits.

Mendel validated these results by performing an F₃ cross in which he self-crossed the dominant- and recessive-expressing F₂ plants. When he self-crossed the plants expressing green seeds, all of the offspring had green seeds, confirming that all green seeds had homozygous genotypes of yy. When he self-crossed the F₂ plants expressing yellow seeds, he found that one-third of the plants bred true, and two-thirds of the plants segregated at a 3:1 ratio of yellow:green seeds. In this case, the true-breeding plants had homozygous (YY) genotypes, whereas the segregating plants corresponded to the heterozygous (Yy) genotype. When these plants self-fertilized, the outcome was just like the F₁ self-fertilizing cross.

The Test Cross Distinguishes the Dominant Phenotype

Beyond predicting the offspring of a cross between known homozygous or heterozygous parents, Mendel also developed a way to determine whether an organism that expressed a dominant trait was a heterozygote or a homozygote. Called the **test cross**,

this technique is still used by plant and animal breeders. In a test cross, the dominant-expressing organism is crossed with an organism that is homozygous recessive for the same characteristic. If the dominant-expressing organism is a homozygote, then all F_1 offspring will be heterozygotes expressing the dominant trait (Figure 12.5). Alternatively, if the dominant-expressing organism is a heterozygote, the F_1 offspring will exhibit a 1:1 ratio of heterozygotes and recessive homozygotes (Figure 12.5). The test cross further validates Mendel's postulate that pairs of unit factors segregate equally.



VISUAL CONNECTION

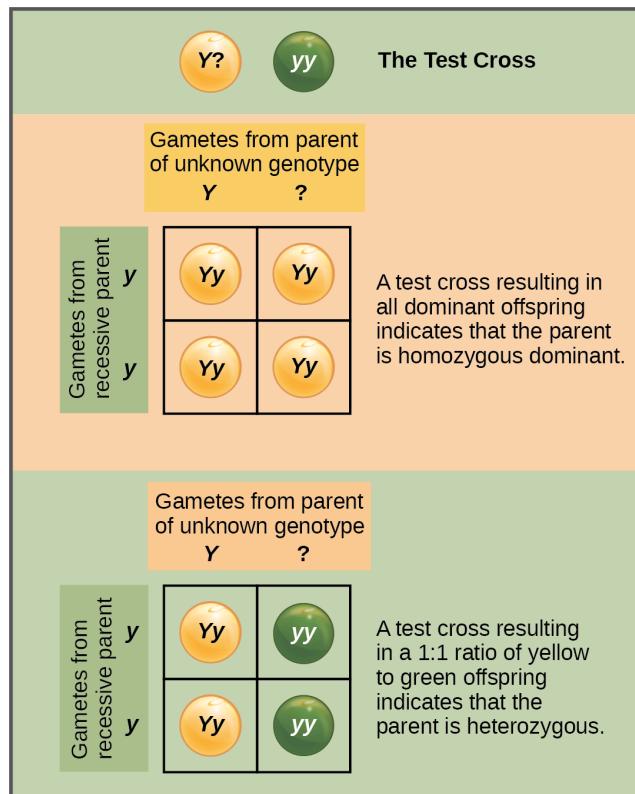


Figure 12.5 A test cross can be performed to determine whether an organism expressing a dominant trait is a homozygote or a heterozygote.

In pea plants, round peas (R) are dominant to wrinkled peas (r). You do a test cross between a pea plant with wrinkled peas (genotype rr) and a plant of unknown genotype that has round peas. You end up with three plants, all which have round peas. From this data, can you tell if the round pea parent plant is homozygous dominant or heterozygous? If the round pea parent plant is heterozygous, what is the probability that a random sample of 3 progeny peas will all be round?

Many human diseases are genetically inherited. A healthy person in a family in which some members suffer from a recessive genetic disorder may want to know if he or she has the disease-causing gene and what risk exists of passing the disorder on to his or her offspring. Of course, doing a test cross in humans is unethical and impractical. Instead, geneticists use **pedigree analysis** to study the inheritance pattern of human genetic diseases (Figure 12.6).



VISUAL CONNECTION

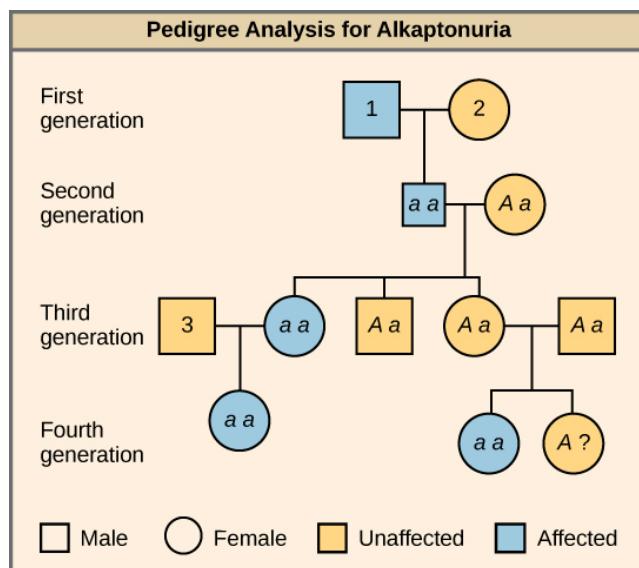


Figure 12.6 Alkaptonuria is a recessive genetic disorder in which two amino acids, phenylalanine and tyrosine, are not properly metabolized. Affected individuals may have darkened skin and brown urine, and may suffer joint damage and other complications. In this pedigree, individuals with the disorder are indicated in blue and have the genotype aa. Unaffected individuals are indicated in yellow and have the genotype AA or Aa. Note that it is often possible to determine a person's genotype from the genotype of their offspring. For example, if neither parent has the disorder but their child does, they must be heterozygous. Two individuals on the pedigree have an unaffected phenotype but unknown genotype. Because they do not have the disorder, they must have at least one normal allele, so their genotype gets the "A?" designation.

What are the genotypes of the individuals labeled 1, 2, and 3?

Alternatives to Dominance and Recessiveness

Mendel's experiments with pea plants suggested that: (1) two "units" or alleles exist for every gene; (2) alleles maintain their integrity in each generation (no blending); and (3) in the presence of the dominant allele, the recessive allele is hidden and makes no contribution to the phenotype. Therefore, recessive alleles can be "carried" and not expressed by individuals. Such heterozygous individuals are sometimes referred to as "carriers." Further genetic studies in other plants and animals have shown that much more complexity exists, but that the fundamental principles of Mendelian genetics still hold true. In the sections to follow, we consider some of the extensions of Mendelism. If Mendel had chosen an experimental system that exhibited these genetic complexities, it's possible that he would not have understood what his results meant.

Incomplete Dominance

Mendel's results, that traits are inherited as dominant and recessive pairs, contradicted the view at that time that offspring exhibited a blend of their parents' traits. However, the heterozygote phenotype occasionally does appear to be intermediate between the two parents. For example, in the snapdragon, *Antirrhinum majus* (Figure 12.7), a cross between a homozygous parent with white flowers ($C^W C^W$) and a homozygous parent with red flowers ($C^R C^R$) will produce offspring with pink flowers ($C^R C^W$). (Note that different genotypic abbreviations are used for Mendelian extensions to distinguish these patterns from simple dominance and recessiveness.) This pattern of inheritance is described as **incomplete dominance**, denoting the expression of two contrasting alleles such that the individual displays an intermediate phenotype. The allele for red flowers is incompletely dominant over the allele for white flowers. However, the results of a heterozygote self-cross can still be predicted, just as with Mendelian dominant and recessive crosses. In this case, the genotypic ratio would be $1 C^R C^R : 2 C^R C^W : 1 C^W C^W$, and the phenotypic ratio would be 1:2:1 for red:pink:white.



Figure 12.7 These pink flowers of a heterozygote snapdragon result from incomplete dominance. (credit: "storebukkebruse"/Flickr)

Codominance

A variation on incomplete dominance is **codominance**, in which both alleles for the same characteristic are simultaneously expressed in the heterozygote. An example of codominance is the MN blood groups of humans. The M and N alleles are expressed in the form of an M or N antigen present on the surface of red blood cells. Homozygotes ($L^M L^M$ and $L^N L^N$) express either the M or the N allele, and heterozygotes ($L^M L^N$) express both alleles equally. In a self-cross between heterozygotes expressing a codominant trait, the three possible offspring genotypes are phenotypically distinct. However, the 1:2:1 genotypic ratio characteristic of a Mendelian monohybrid cross still applies.

Multiple Alleles

Mendel implied that only two alleles, one dominant and one recessive, could exist for a given gene. We now know that this is an oversimplification. Although individual humans (and all diploid organisms) can only have two alleles for a given gene, multiple alleles may exist at the population level such that many combinations of two alleles are observed. Note that when many alleles exist for the same gene, the convention is to denote the most common phenotype or genotype among wild animals as the **wild type** (often abbreviated “+”); this is considered the standard or norm. All other phenotypes or genotypes are considered **variants** of this standard, meaning that they deviate from the wild type. The variant may be recessive or dominant to the wild-type allele.

An example of multiple alleles is coat color in rabbits (Figure 12.8). Here, four alleles exist for the *c* gene. The wild-type version, $C^+ C^+$, is expressed as brown fur. The chinchilla phenotype, $c^{ch} c^{ch}$, is expressed as black-tipped white fur. The Himalayan phenotype, $c^h c^h$, has black fur on the extremities and white fur elsewhere. Finally, the albino, or “colorless” phenotype, cc , is expressed as white fur. In cases of multiple alleles, dominance hierarchies can exist. In this case, the wild-type allele is dominant over all the others, chinchilla is incompletely dominant over Himalayan and albino, and Himalayan is dominant over albino. This hierarchy, or allelic series, was revealed by observing the phenotypes of each possible heterozygote offspring.

Allele			
C	c^{ch}	c^h	c
Genotype			
CC	$c^{ch}c^{ch}$	$c^h c^h$	cc
Phenotype			
WILD TYPE: Brown fur	CHINCHILLA: Black-tipped white fur	HIMALAYAN: White fur with black paws, nose, ears, tail	ALBINO: White fur
			

Figure 12.8 Four different alleles exist for the rabbit coat color (C) gene.

The complete dominance of a wild-type phenotype over all other mutants often occurs as an effect of “dosage” of a specific gene product, such that the wild-type allele supplies the correct amount of gene product whereas the mutant alleles cannot. For the allelic series in rabbits, the wild-type allele may supply a given dosage of fur pigment, whereas the mutants supply a lesser dosage or none at all. Interestingly, the Himalayan phenotype is the result of an allele that produces a temperature-sensitive gene product that only produces pigment in the cooler extremities of the rabbit’s body.

Alternatively, one mutant allele can be dominant over all other phenotypes, including the wild type. This may occur when the mutant allele somehow interferes with the genetic message so that even a heterozygote with one wild-type allele copy expresses the mutant phenotype. One way in which the mutant allele can interfere is by enhancing the function of the wild-type gene product or changing its distribution in the body. One example of this is the *Antennapedia* mutation in *Drosophila* (Figure 12.9). In this case, the mutant allele expands the distribution of the gene product, and as a result, the *Antennapedia* heterozygote develops legs on its head where its antennae should be.



Figure 12.9 As seen in comparing the wild-type *Drosophila* (left) and the *Antennapedia* mutant (right), the *Antennapedia* mutant has legs on its head in place of antennae.



EVOLUTION CONNECTION

Multiple Alleles Confer Drug Resistance in the Malaria Parasite

Malaria is a parasitic disease in humans that is transmitted by infected female mosquitoes, including *Anopheles gambiae* (Figure 12.10a), and is characterized by cyclic high fevers, chills, flu-like symptoms, and severe anemia. *Plasmodium falciparum* and *P. vivax* are the most common causative agents of malaria, and *P. falciparum* is the most deadly (Figure 12.10b). When promptly and correctly treated, *P. falciparum* malaria has a mortality rate of 0.1 percent. However, in some parts of the world, the parasite has evolved resistance to commonly used malaria treatments, so the most effective malarial treatments can vary by geographic region.

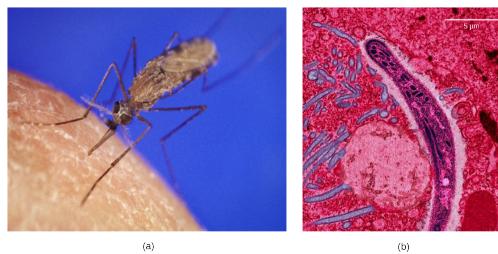


Figure 12.10 The (a) *Anopheles gambiae*, or African malaria mosquito, acts as a vector in the transmission to humans of the malaria-causing parasite (b) *Plasmodium falciparum*, here visualized using false-color transmission electron microscopy. (credit a: James D. Gathany; credit b: Ute Frevert; false color by Margaret Shear; scale-bar data from Matt Russell)

In Southeast Asia, Africa, and South America, *P. falciparum* has developed resistance to the anti-malarial drugs chloroquine, mefloquine, and sulfadoxine-pyrimethamine. *P. falciparum*, which is haploid during the life stage in which it is infectious to humans, has evolved multiple drug-resistant mutant alleles of the *dhps* gene. Varying degrees of sulfadoxine resistance are associated with each of these alleles. Being haploid, *P. falciparum* needs only one drug-resistant allele to express this trait.

In Southeast Asia, different sulfadoxine-resistant alleles of the *dhps* gene are localized to different geographic regions. This is a common evolutionary phenomenon that occurs because drug-resistant mutants arise in a population and interbreed with other *P. falciparum* isolates in close proximity. Sulfadoxine-resistant parasites cause considerable human hardship in regions where this drug is widely used as an over-the-counter malaria remedy. As is common with pathogens that multiply to large numbers within an infection cycle, *P. falciparum* evolves relatively rapidly (over a decade or so) in response to the selective pressure of commonly used anti-malarial drugs. For this reason, scientists must constantly work to develop new drugs or drug combinations to combat the worldwide malaria burden.²

X-Linked Traits

In humans, as well as in many other animals and some plants, the sex of the individual is determined by sex chromosomes. The sex chromosomes are one pair of non-homologous chromosomes. Until now, we have only considered inheritance patterns among non-sex chromosomes, or **autosomes**. In addition to 22 homologous pairs of autosomes, human females have a homologous pair of X chromosomes, whereas human males have an XY chromosome pair. 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. When a gene being examined is present on the X chromosome, but not on the Y chromosome, it is said to be **X-linked**.

Eye color in *Drosophila* was one of the first X-linked traits to be identified. Thomas Hunt Morgan mapped this trait to the X chromosome in 1910. Like humans, *Drosophila* males have an XY chromosome pair, and females are XX. In flies, the wild-type eye color is red (X^W) and it is dominant to white eye color (X^w) (Figure 12.11). Because of the location of the eye-color gene, reciprocal crosses do not produce the same offspring ratios. Males are said to be **hemizygous**, because they have only one allele for any X-linked characteristic. Hemizygosity makes the descriptions of dominance and recessiveness irrelevant for XY males. *Drosophila* males lack a second allele copy on the Y chromosome; that is, their genotype can only be X^WY or X^wY . In contrast, females have two allele copies of this gene and can be X^WX^W , X^WX^w , or X^wX^w .

²Sumiti Vinayak, et al., "Origin and Evolution of Sulfadoxine Resistant *Plasmodium falciparum*," *Public Library of Science Pathogens* 6, no. 3 (2010): e1000830, doi:10.1371/journal.ppat.1000830.



Figure 12.11 In *Drosophila*, several genes determine eye color. The genes for white and vermilion eye colors are located on the X chromosome. Others are located on the autosomes. Clockwise from top left are brown, cinnabar, sepia, vermilion, white, and red. Red eye color is wild-type and is dominant to white eye color.

In an X-linked cross, the genotypes of F_1 and F_2 offspring depend on whether the recessive trait was expressed by the male or the female in the P_1 generation. With regard to *Drosophila* eye color, when the P_1 male expresses the white-eye phenotype and the female is homozygous red-eyed, all members of the F_1 generation exhibit red eyes (Figure 12.12). The F_1 females are heterozygous ($X^W X^w$), and the males are all $X^W Y$, having received their X chromosome from the homozygous dominant P_1 female and their Y chromosome from the P_1 male. A subsequent cross between the $X^W X^w$ female and the $X^W Y$ male would produce only red-eyed females (with $X^W X^W$ or $X^W X^w$ genotypes) and both red- and white-eyed males (with $X^W Y$ or $X^w Y$ genotypes). Now, consider a cross between a homozygous white-eyed female and a male with red eyes. The F_1 generation would exhibit only heterozygous red-eyed females ($X^W X^w$) and only white-eyed males ($X^w Y$). Half of the F_2 females would be red-eyed ($X^W X^w$) and half would be white-eyed ($X^w X^w$). Similarly, half of the F_2 males would be red-eyed ($X^W Y$) and half would be white-eyed ($X^w Y$).



VISUAL CONNECTION

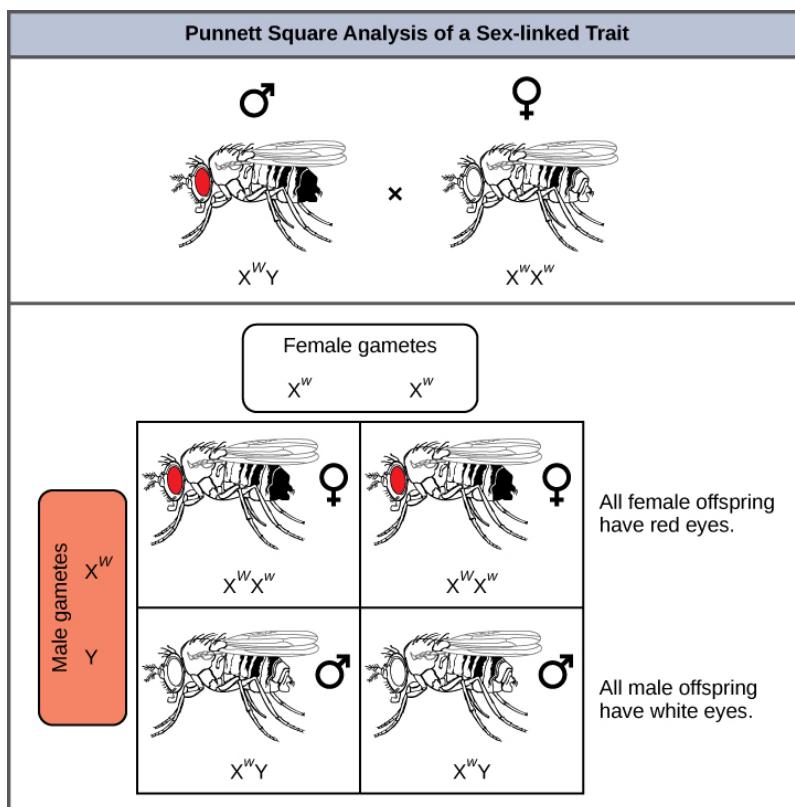


Figure 12.12 Punnett square analysis is used to determine the ratio of offspring from a cross between a red-eyed male fruit fly and a white-eyed female fruit fly.

What ratio of offspring would result from a cross between a white-eyed male and a female that is heterozygous for red eye color?

Discoveries in fruit fly genetics can be applied to human genetics. When a female parent is homozygous for a recessive X-linked trait, she will pass the trait on to 100 percent of her offspring. Her male offspring are, therefore, destined to express the trait, as they will inherit their father's Y chromosome. In humans, the alleles for certain conditions (some forms of color blindness, hemophilia, and muscular dystrophy) are X-linked. Females who are heterozygous for these diseases are said to be carriers and may not exhibit any phenotypic effects. These females will pass the disease to half of their sons and will pass carrier status to half of their daughters; therefore, recessive X-linked traits appear more frequently in males than females.

In some groups of organisms with sex chromosomes, the sex with the non-homologous sex chromosomes is the female rather than the male. This is the case for all birds. In this case, sex-linked traits will be more likely to appear in the female, in which they are hemizygous.

Human Sex-linked Disorders

Sex-linkage studies in Morgan's laboratory provided the fundamentals for understanding X-linked recessive disorders in humans, which include red-green color blindness, and Types A and B hemophilia. Because human males need to inherit only one recessive mutant X allele to be affected, X-linked disorders are disproportionately observed in males. Females must inherit recessive X-linked alleles from both of their parents in order to express the trait. When they inherit one recessive X-linked mutant allele and one dominant X-linked wild-type allele, they are carriers of the trait and are typically unaffected. Carrier females can manifest mild forms of the trait due to the inactivation of the dominant allele located on one of the X chromosomes. However, female carriers can contribute the trait to their sons, resulting in the son exhibiting the trait, or they can contribute the recessive allele to their daughters, resulting in the daughters being carriers of the trait (Figure 12.13). Although some Y-linked recessive disorders exist, typically they are associated with infertility in males and are therefore not transmitted to subsequent generations.

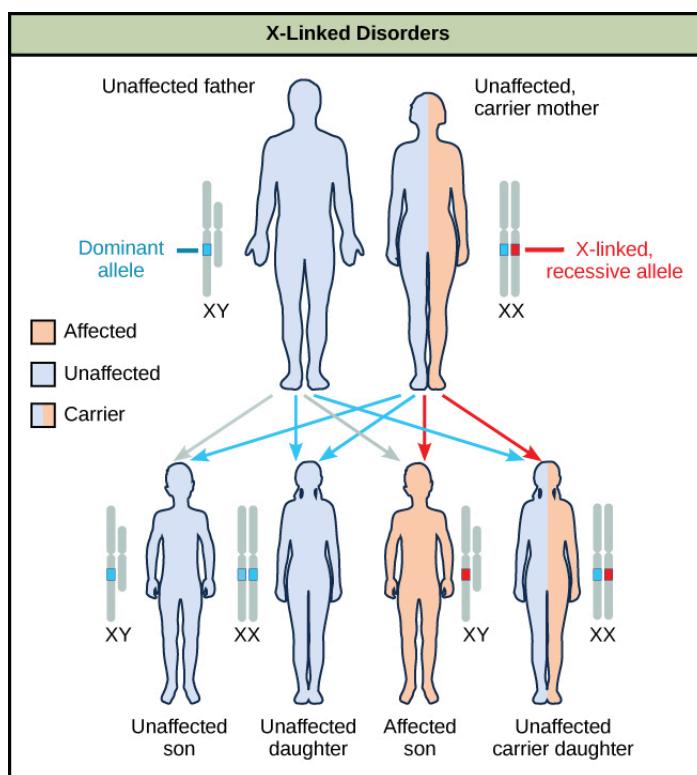


Figure 12.13 The son of a woman who is a carrier of a recessive X-linked disorder will have a 50 percent chance of being affected. A daughter will not be affected, but she will have a 50 percent chance of being a carrier like her mother.

LINK TO LEARNING

Watch this video to learn more about sex-linked traits.

[Click to view content \(\[https://www.openstax.org/l/sex-linked_trts\]\(https://www.openstax.org/l/sex-linked_trts\)\)](https://www.openstax.org/l/sex-linked_trts)

Lethality

A large proportion of genes in an individual's genome are essential for survival. Occasionally, a nonfunctional allele for an essential gene can arise by mutation and be transmitted in a population as long as individuals with this allele also have a wild-type, functional copy. The wild-type allele functions at a capacity sufficient to sustain life and is therefore considered to be dominant over the nonfunctional allele. However, consider two heterozygous parents that have a genotype of wild-type/nonfunctional mutant for a hypothetical essential gene. In one quarter of their offspring, we would expect to observe individuals that are homozygous recessive for the nonfunctional allele. Because the gene is essential, these individuals might fail to develop past fertilization, die *in utero*, or die later in life, depending on what life stage requires this gene. An inheritance pattern in which an allele is only lethal in the homozygous form and in which the heterozygote may be normal or have some altered nonlethal phenotype is referred to as **recessive lethal**.

For crosses between heterozygous individuals with a recessive lethal allele that causes death before birth when homozygous, only wild-type homozygotes and heterozygotes would be observed. The genotypic ratio would therefore be 2:1. In other instances, the recessive lethal allele might also exhibit a dominant (but not lethal) phenotype in the heterozygote. For instance, the recessive lethal *Curly* allele in *Drosophila* affects wing shape in the heterozygote form but is lethal in the homozygote.

A single copy of the wild-type allele is not always sufficient for normal functioning or even survival. The **dominant lethal** inheritance pattern is one in which an allele is lethal both in the homozygote and the heterozygote; this allele can only be transmitted if the lethality phenotype occurs after reproductive age. Individuals with mutations that result in dominant lethal alleles fail to survive even in the heterozygote form. Dominant lethal alleles are very rare because, as you might expect, the allele only lasts one generation and is not transmitted. However, just as the recessive lethal allele might not immediately manifest the phenotype of death, dominant lethal alleles also might not be expressed until adulthood. Once the individual reaches reproductive age, the allele may be unknowingly passed on, resulting in a delayed death in both generations. An example of this

in humans is Huntington's disease, in which the nervous system gradually wastes away (Figure 12.14). People who are heterozygous for the dominant Huntington allele (Hh) will inevitably develop the fatal disease. However, the onset of Huntington's disease may not occur until age 40, at which point the afflicted persons may have already passed the allele to 50 percent of their offspring.

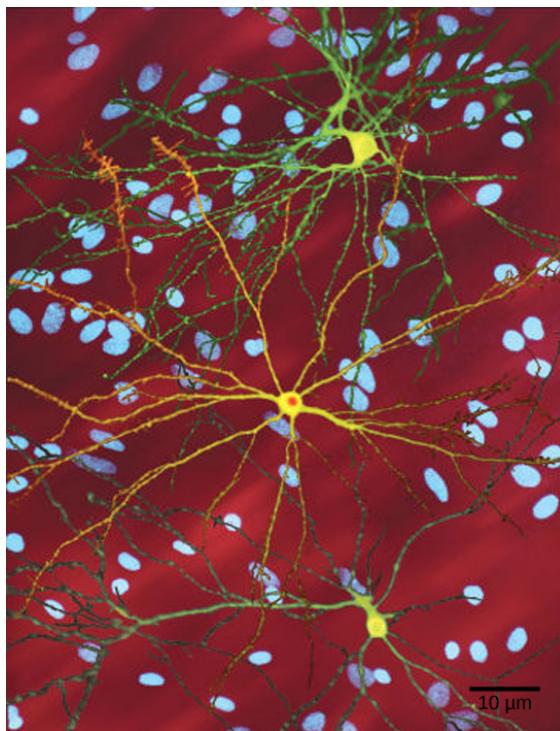


Figure 12.14 The neuron in the center of this micrograph (yellow) has nuclear inclusions characteristic of Huntington's disease (orange area in the center of the neuron). Huntington's disease occurs when an abnormal dominant allele for the Huntington gene is present. (credit: Dr. Steven Finkbeiner, Gladstone Institute of Neurological Disease, The Taube-Koret Center for Huntington's Disease Research, and the University of California San Francisco/Wikimedia)

12.3 Laws of Inheritance

By the end of this section, you will be able to do the following:

- Explain Mendel's law of segregation and independent assortment in terms of genetics and the events of meiosis
- Use the forked-line method and the probability rules to calculate the probability of genotypes and phenotypes from multiple gene crosses
- Explain the effect of linkage and recombination on gamete genotypes
- Explain the phenotypic outcomes of epistatic effects between genes

Mendel generalized the results of his pea-plant experiments into four postulates, some of which are sometimes called “laws,” that describe the basis of dominant and recessive inheritance in diploid organisms. As you have learned, more complex extensions of Mendelism exist that do not exhibit the same F_2 phenotypic ratios (3:1). Nevertheless, these laws summarize the basics of classical genetics.

Pairs of Unit Factors, or Genes

Mendel proposed first that paired unit factors of heredity were transmitted faithfully from generation to generation by the dissociation and reassociation of paired factors during gametogenesis and fertilization, respectively. After he crossed peas with contrasting traits and found that the recessive trait resurfaced in the F_2 generation, Mendel deduced that hereditary factors must be inherited as discrete units. This finding contradicted the belief at that time that parental traits were blended in the offspring.

Alleles Can Be Dominant or Recessive

Mendel's **law of dominance** states that in a heterozygote, one trait will conceal the presence of another trait for the same characteristic. Rather than both alleles contributing to a phenotype, the dominant allele will be expressed exclusively. The recessive allele will remain "latent" but will be transmitted to offspring by the same manner in which the dominant allele is transmitted. The recessive trait will only be expressed by offspring that have two copies of this allele ([Figure 12.15](#)), and these offspring will breed true when self-crossed.

Since Mendel's experiments with pea plants, researchers have found that the law of dominance does not always hold true. Instead, several different patterns of inheritance have been found to exist.



Figure 12.15 The child in the photo expresses albinism, a recessive trait.

Equal Segregation of Alleles

Observing that true-breeding pea plants with contrasting traits gave rise to F_1 generations that all expressed the dominant trait and F_2 generations that expressed the dominant and recessive traits in a 3:1 ratio, Mendel proposed the **law of segregation**. This law states that paired unit factors (genes) must segregate equally into gametes such that offspring have an equal likelihood of inheriting either factor. For the F_2 generation of a monohybrid cross, the following three possible combinations of genotypes could result: homozygous dominant, heterozygous, or homozygous recessive. Because heterozygotes could arise from two different pathways (receiving one dominant and one recessive allele from either parent), and because heterozygotes and homozygous dominant individuals are phenotypically identical, the law supports Mendel's observed 3:1 phenotypic ratio. The equal segregation of alleles is the reason we can apply the Punnett square to accurately predict the offspring of parents with known genotypes. The physical basis of Mendel's law of segregation is the first division of meiosis, in which the homologous chromosomes with their different versions of each gene are segregated into daughter nuclei. The role of the meiotic segregation of chromosomes in sexual reproduction was not understood by the scientific community during Mendel's lifetime.

Independent Assortment

Mendel's **law of independent assortment** states that genes do not influence each other with regard to the sorting of alleles into gametes, and every possible combination of alleles for every gene is equally likely to occur. The independent assortment of genes can be illustrated by the **dihybrid** cross, a cross between two true-breeding parents that express different traits for two characteristics. Consider the characteristics of seed color and seed texture for two pea plants, one that has green, wrinkled seeds ($yyrr$) and another that has yellow, round seeds ($YYRR$). Because each parent is homozygous, the law of segregation indicates that the gametes for the green/wrinkled plant all are yr , and the gametes for the yellow/round plant are all YR . Therefore, the F_1 generation of offspring all are $YyRr$ ([Figure 12.16](#)).



VISUAL CONNECTION

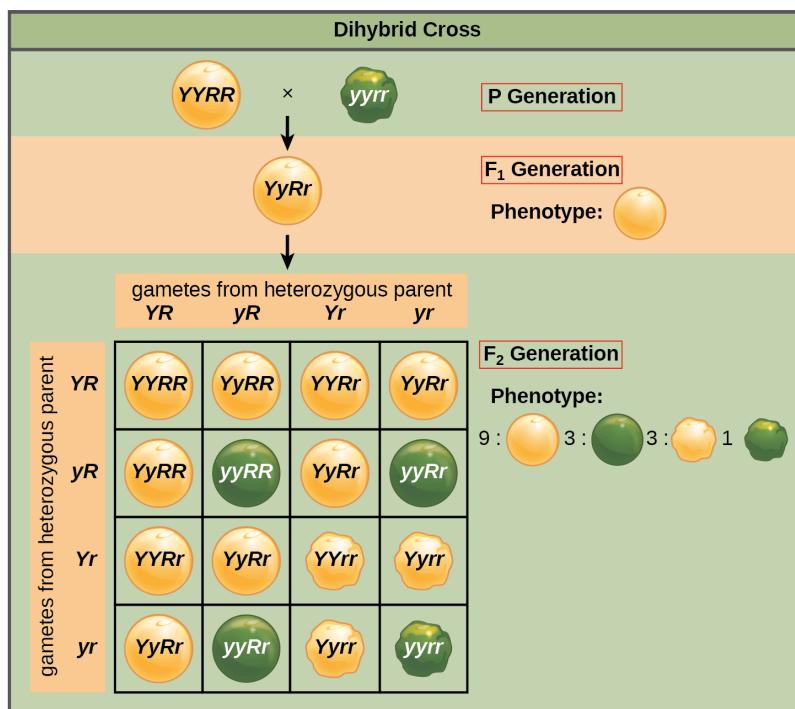


Figure 12.16 This dihybrid cross of pea plants involves the genes for seed color and texture.

In pea plants, purple flowers (P) are dominant to white flowers (p) and yellow peas (Y) are dominant to green peas (y). What are the possible genotypes and phenotypes for a cross between PpYY and ppYy pea plants? How many squares do you need to do a Punnett square analysis of this cross?

For the F₂ generation, the law of segregation requires that each gamete receive either an *R* allele or an *r* allele along with either a *Y* allele or a *y* allele. The law of independent assortment states that a gamete into which an *r* allele sorted would be equally likely to contain either a *Y* allele or a *y* allele. Thus, there are four equally likely gametes that can be formed when the *YyRr* heterozygote is self-crossed, as follows: *YR*, *Yr*, *yR*, and *yr*. Arranging these gametes along the top and left of a 4 × 4 Punnett square ([Figure 12.16](#)) gives us 16 equally likely genotypic combinations. From these genotypes, we infer a phenotypic ratio of 9 round/yellow:3 round/green:3 wrinkled/yellow:1 wrinkled/green ([Figure 12.16](#)). These are the offspring ratios we would expect, assuming we performed the crosses with a large enough sample size.

Because of independent assortment and dominance, the 9:3:3:1 dihybrid phenotypic ratio can be collapsed into two 3:1 ratios, characteristic of any monohybrid cross that follows a dominant and recessive pattern. Ignoring seed color and considering only seed texture in the above dihybrid cross, we would expect that three quarters of the F₂ generation offspring would be round, and one quarter would be wrinkled. Similarly, isolating only seed color, we would assume that three quarters of the F₂ offspring would be yellow and one quarter would be green. The sorting of alleles for texture and color are independent events, so we can apply the product rule. Therefore, the proportion of round and yellow F₂ offspring is expected to be $(3/4) \times (3/4) = 9/16$, and the proportion of wrinkled and green offspring is expected to be $(1/4) \times (1/4) = 1/16$. These proportions are identical to those obtained using a Punnett square. Round, green and wrinkled, yellow offspring can also be calculated using the product rule, as each of these genotypes includes one dominant and one recessive phenotype. Therefore, the proportion of each is calculated as $(3/4) \times (1/4) = 3/16$.

The law of independent assortment also indicates that a cross between yellow, wrinkled (YYrr) and green, round (yyRR) parents would yield the same F₁ and F₂ offspring as in the YYRR × yyrr cross.

The physical basis for the law of independent assortment also lies in meiosis I, in which the different homologous pairs line up in random orientations. Each gamete can contain any combination of paternal and maternal chromosomes (and therefore the genes on them) because the orientation of tetrads on the metaphase plane is random.

Forked-Line Method

When more than two genes are being considered, the Punnett-square method becomes unwieldy. For instance, examining a cross involving four genes would require a 16×16 grid containing 256 boxes. It would be extremely cumbersome to manually enter each genotype. For more complex crosses, the forked-line and probability methods are preferred.

To prepare a forked-line diagram for a cross between F_1 heterozygotes resulting from a cross between $AABBCC$ and $aabbcc$ parents, we first create rows equal to the number of genes being considered, and then segregate the alleles in each row on forked lines according to the probabilities for individual monohybrid crosses (Figure 12.17). We then multiply the values along each forked path to obtain the F_2 offspring probabilities. Note that this process is a diagrammatic version of the product rule. The values along each forked pathway can be multiplied because each gene assorts independently. For a trihybrid cross, the F_2 phenotypic ratio is 27:9:9:3:3:1.

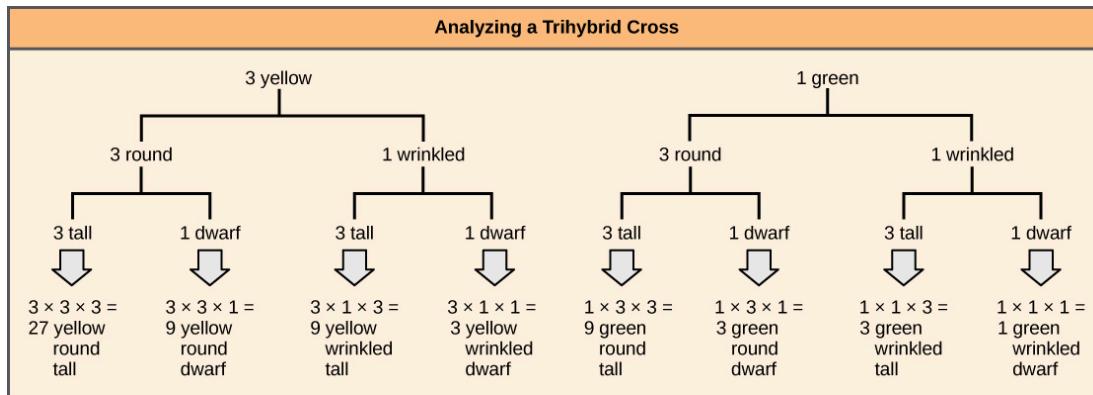


Figure 12.17 The forked-line method can be used to analyze a trihybrid cross. Here, the probability for color in the F_2 generation occupies the top row (3 yellow:1 green). The probability for shape occupies the second row (3 round:1 wrinkled), and the probability for height occupies the third row (3 tall:1 dwarf). The probability for each possible combination of traits is calculated by multiplying the probability for each individual trait. Thus, the probability of F_2 offspring having yellow, round, and tall traits is $3 \times 3 \times 3$, or 27.

Probability Method

While the forked-line method is a diagrammatic approach to keeping track of probabilities in a cross, the probability method gives the proportions of offspring expected to exhibit each phenotype (or genotype) without the added visual assistance. Both methods make use of the product rule and consider the alleles for each gene separately. Earlier, we examined the phenotypic proportions for a trihybrid cross using the forked-line method; now we will use the probability method to examine the genotypic proportions for a cross with even more genes.

For a trihybrid cross, writing out the forked-line method is tedious, albeit not as tedious as using the Punnett-square method. To fully demonstrate the power of the probability method, however, we can consider specific genetic calculations. For instance, for a tetrahybrid cross between individuals that are heterozygotes for all four genes, and in which all four genes are sorting independently and in a dominant and recessive pattern, what proportion of the offspring will be expected to be homozygous recessive for all four alleles? Rather than writing out every possible genotype, we can use the probability method. We know that for each gene, the fraction of homozygous recessive offspring will be $1/4$. Therefore, multiplying this fraction for each of the four genes, $(1/4) \times (1/4) \times (1/4) \times (1/4)$, we determine that $1/256$ of the offspring will be quadruply homozygous recessive.

For the same tetrahybrid cross, what is the expected proportion of offspring that have the dominant phenotype at all four loci? We can answer this question using phenotypic proportions, but let's do it the hard way—using genotypic proportions. The question asks for the proportion of offspring that are 1) homozygous dominant at A or heterozygous at A , and 2) homozygous at B or heterozygous at B , and so on. Noting the “or” and “and” in each circumstance makes clear where to apply the sum and product rules. The probability of a homozygous dominant at A is $1/4$ and the probability of a heterozygote at A is $1/2$. The probability of the homozygote or the heterozygote is $1/4 + 1/2 = 3/4$ using the sum rule. The same probability can be obtained in the same way for each of the other genes, so that the probability of a dominant phenotype at A and B and C and D is, using the product rule, equal to $3/4 \times 3/4 \times 3/4 \times 3/4$, or $81/256$. If you are ever unsure about how to combine probabilities, returning to the forked-line method should make it clear.

Rules for Multihybrid Fertilization

Predicting the genotypes and phenotypes of offspring from given crosses is the best way to test your knowledge of Mendelian

genetics. Given a multihybrid cross that obeys independent assortment and follows a dominant and recessive pattern, several generalized rules exist; you can use these rules to check your results as you work through genetics calculations ([Table 12.5](#)). To apply these rules, first you must determine n , the number of heterozygous gene pairs (the number of genes segregating two alleles each). For example, a cross between $AaBb$ and $AaBb$ heterozygotes has an n of 2. In contrast, a cross between $AABb$ and $AABb$ has an n of 1 because A is not heterozygous.

General Rules for Multihybrid Crosses

General Rule	Number of Heterozygous Gene Pairs
Number of different F_1 gametes	2^n
Number of different F_2 genotypes	3^n
Given dominant and recessive inheritance, the number of different F_2 phenotypes	2^n

Table 12.5

Linked Genes Violate the Law of Independent Assortment

Although all of Mendel's pea characteristics behaved according to the law of independent assortment, we now know that some allele combinations are not inherited independently of each other. Genes that are located on separate non-homologous chromosomes will always sort independently. However, each chromosome contains hundreds or thousands of genes, organized linearly on chromosomes like beads on a string. The segregation of alleles into gametes can be influenced by **linkage**, in which genes that are located physically close to each other on the same chromosome are more likely to be inherited as a pair. However, because of the process of recombination, or "crossover," it is possible for two genes on the same chromosome to behave independently, or as if they are not linked. To understand this, let's consider the biological basis of gene linkage and recombination.

Homologous chromosomes possess the same genes in the same linear order. The alleles may differ on homologous chromosome pairs, but the genes to which they correspond do not. In preparation for the first division of meiosis, homologous chromosomes replicate and synapse. Like genes on the homologs align with each other. At this stage, segments of homologous chromosomes exchange linear segments of genetic material ([Figure 12.18](#)). This process is called *recombination*, or crossover, and it is a common genetic process. Because the genes are aligned during recombination, the gene order is not altered. Instead, the result of recombination is that maternal and paternal alleles are combined onto the same chromosome. Across a given chromosome, several recombination events may occur, causing extensive shuffling of alleles.

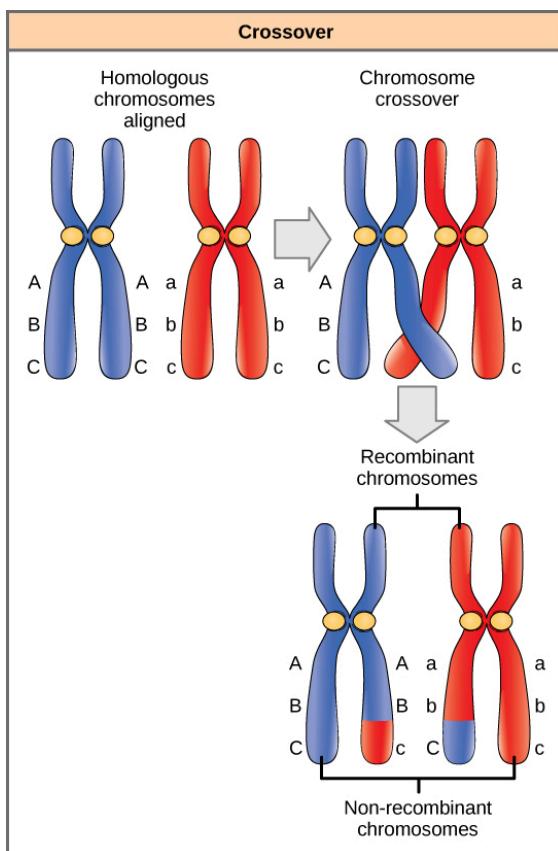


Figure 12.18 The process of crossover, or recombination, occurs when two homologous chromosomes align during meiosis and exchange a segment of genetic material. Here, the alleles for gene C were exchanged. The result is two recombinant and two non-recombinant chromosomes.

When two genes are located in close proximity on the same chromosome, they are considered linked, and their alleles tend to be transmitted through meiosis together. To exemplify this, imagine a dihybrid cross involving flower color and plant height in which the genes are next to each other on the chromosome. If one homologous chromosome has alleles for tall plants and red flowers, and the other chromosome has genes for short plants and yellow flowers, then when the gametes are formed, the tall and red alleles will go together into a gamete and the short and yellow alleles will go into other gametes. These are called the parental genotypes because they have been inherited intact from the parents of the individual producing gametes. But unlike if the genes were on different chromosomes, there will be no gametes with tall and yellow alleles and no gametes with short and red alleles. If you create the Punnett square with these gametes, you will see that the classical Mendelian prediction of a 9:3:3:1 outcome of a dihybrid cross would not apply. As the distance between two genes increases, the probability of one or more crossovers between them increases, and the genes behave more like they are on separate chromosomes. Geneticists have used the proportion of recombinant gametes (the ones not like the parents) as a measure of how far apart genes are on a chromosome. Using this information, they have constructed elaborate maps of genes on chromosomes for well-studied organisms, including humans.

Mendel's seminal publication makes no mention of linkage, and many researchers have questioned whether he encountered linkage but chose not to publish those crosses out of concern that they would invalidate his independent assortment postulate. The garden pea has seven chromosomes, and some have suggested that his choice of seven characteristics was not a coincidence. However, even if the genes he examined were not located on separate chromosomes, it is possible that he simply did not observe linkage because of the extensive shuffling effects of recombination.



SCIENTIFIC METHOD CONNECTION

Testing the Hypothesis of Independent Assortment

To better appreciate the amount of labor and ingenuity that went into Mendel's experiments, proceed through one of Mendel's

dihybrid crosses.

Question: What will be the offspring of a dihybrid cross?

Background: Consider that pea plants mature in one growing season, and you have access to a large garden in which you can cultivate thousands of pea plants. There are several true-breeding plants with the following pairs of traits: tall plants with inflated pods, and dwarf plants with constricted pods. Before the plants have matured, you remove the pollen-producing organs from the tall/inflated plants in your crosses to prevent self-fertilization. Upon plant maturation, the plants are manually crossed by transferring pollen from the dwarf/constricted plants to the stigmata of the tall/inflated plants.

Hypothesis: Both trait pairs will sort independently according to Mendelian laws. When the true-breeding parents are crossed, all of the F_1 offspring are tall and have inflated pods, which indicates that the tall and inflated traits are dominant over the dwarf and constricted traits, respectively. A self-cross of the F_1 heterozygotes results in 2,000 F_2 progeny.

Test the hypothesis: Because each trait pair sorts independently, the ratios of tall:dwarf and inflated:constricted are each expected to be 3:1. The tall/dwarf trait pair is called T/t , and the inflated/constricted trait pair is designated I/i . Each member of the F_1 generation therefore has a genotype of $TtIi$. Construct a grid analogous to [Figure 12.16](#), in which you cross two $TtIi$ individuals. Each individual can donate four combinations of two traits: TI , Ti , tI , or ti , meaning that there are 16 possibilities of offspring genotypes. Because the T and I alleles are dominant, any individual having one or two of those alleles will express the tall or inflated phenotypes, respectively, regardless if they also have a t or i allele. Only individuals that are tt or ii will express the dwarf and constricted alleles, respectively. As shown in [Figure 12.19](#), you predict that you will observe the following offspring proportions: tall/inflated:tall/constricted:dwarf/inflated:dwarf/constricted in a 9:3:3:1 ratio. Notice from the grid that when considering the tall/dwarf and inflated/constricted trait pairs in isolation, they are each inherited in 3:1 ratios.

		$TtIi$			
		TI	Ti	tI	ti
$TtIi$	TI	$TTII$	$TTIi$	$TtII$	$TtIi$
	Ti	$TTIi$	$TTii$	$TtIi$	$Ttii$
	tI	$TtII$	$TtIi$	$ttII$	$ttIi$
	ti	$TtIi$	$Ttii$	$ttIi$	$ttii$

Figure 12.19 This figure shows all possible combinations of offspring resulting from a dihybrid cross of pea plants that are heterozygous for the tall/dwarf and inflated/constricted alleles.

Test the hypothesis: You cross the dwarf and tall plants and then self-cross the offspring. For best results, this is repeated with hundreds or even thousands of pea plants. What special precautions should be taken in the crosses and in growing the plants?

Analyze your data: You observe the following plant phenotypes in the F_2 generation: 2706 tall/inflated, 930 tall/constricted, 888 dwarf/inflated, and 300 dwarf/constricted. Reduce these findings to a ratio and determine if they are consistent with Mendelian laws.

Form a conclusion: Were the results close to the expected 9:3:3:1 phenotypic ratio? Do the results support the prediction? What might be observed if far fewer plants were used, given that alleles segregate randomly into gametes? Try to imagine growing that many pea plants, and consider the potential for experimental error. For instance, what would happen if it was extremely windy one day?

Epistasis

Mendel's studies in pea plants implied that the sum of an individual's phenotype was controlled by genes (or as he called them, unit factors), such that every characteristic was distinctly and completely controlled by a single gene. In fact, single observable characteristics are almost always under the influence of multiple genes (each with two or more alleles) acting in unison. For example, at least eight genes contribute to eye color in humans.

LINK TO LEARNING

Eye color in humans is determined by multiple genes. Use the [Eye Color Calculator](http://openstax.org/l/eye_color_calc) (http://openstax.org/l/eye_color_calc) to predict the eye color of children from parental eye color.

In some cases, several genes can contribute to aspects of a common phenotype without their gene products ever directly interacting. In the case of organ development, for instance, genes may be expressed sequentially, with each gene adding to the complexity and specificity of the organ. Genes may function in complementary or synergistic fashions, such that two or more genes need to be expressed simultaneously to affect a phenotype. Genes may also oppose each other, with one gene modifying the expression of another.

In **epistasis**, the interaction between genes is antagonistic, such that one gene masks or interferes with the expression of another. "Epistasis" is a word composed of Greek roots that mean "standing upon." The alleles that are being masked or silenced are said to be hypostatic to the epistatic alleles that are doing the masking. Often the biochemical basis of epistasis is a gene pathway in which the expression of one gene is dependent on the function of a gene that precedes or follows it in the pathway.

An example of epistasis is pigmentation in mice. The wild-type coat color, agouti (*AA*), is dominant to solid-colored fur (*aa*). However, a separate gene (*C*) is necessary for pigment production. A mouse with a recessive *c* allele at this locus is unable to produce pigment and is albino regardless of the allele present at locus *A* (Figure 12.20). Therefore, the genotypes *AAcc*, *Aacc*, and *aacc* all produce the same albino phenotype. A cross between heterozygotes for both genes (*AaCc* x *AaCc*) would generate offspring with a phenotypic ratio of 9 agouti:3 solid color:4 albino (Figure 12.20). In this case, the *C* gene is epistatic to the *A* gene.

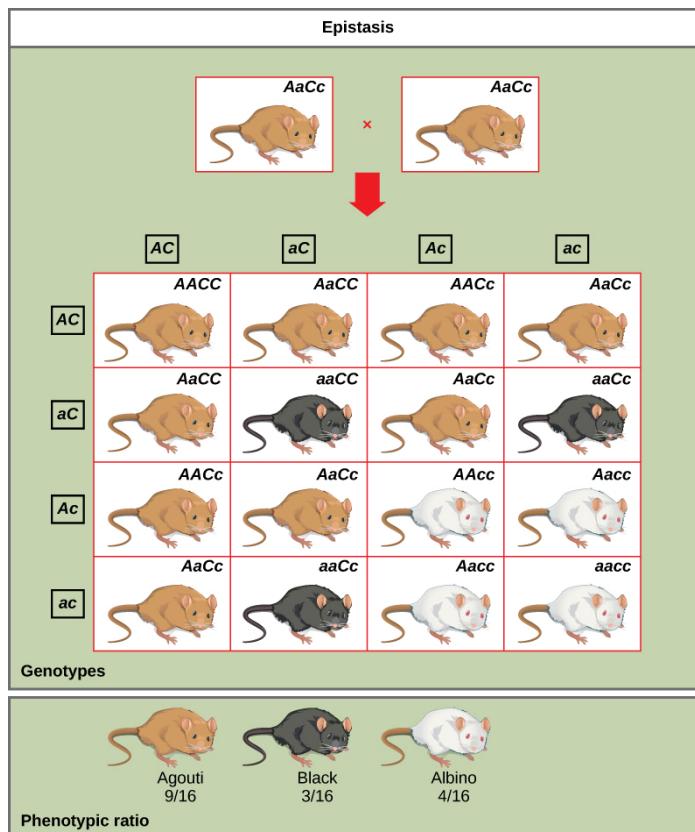


Figure 12.20 In mice, the mottled agouti coat color (*A*) is dominant to a solid coloration, such as black or gray. A gene at a separate locus (*C*) is responsible for pigment production. The recessive *c* allele does not produce pigment, and a mouse with the homozygous recessive *cc* genotype is albino regardless of the allele present at the *A* locus. Thus, the *C* gene is epistatic to the *A* gene.

Epistasis can also occur when a dominant allele masks expression at a separate gene. Fruit color in summer squash is expressed in this way. Homozygous recessive expression of the *W* gene (*ww*) coupled with homozygous dominant or heterozygous expression of the *Y* gene (*YY* or *Yy*) generates yellow fruit, and the *wwyy* genotype produces green fruit. However, if a dominant copy of the *W* gene is present in the homozygous or heterozygous form, the summer squash will produce white fruit regardless of the *Y* alleles. A cross between white heterozygotes for both genes (*WwYy* × *WwYy*) would produce offspring with a phenotypic ratio of 12 white:3 yellow:1 green.

Finally, epistasis can be reciprocal such that either gene, when present in the dominant (or recessive) form, expresses the same phenotype. In the shepherd's purse plant (*Capsella bursa-pastoris*), the characteristic of seed shape is controlled by two genes in a dominant epistatic relationship. When the genes *A* and *B* are both homozygous recessive (*aabb*), the seeds are ovoid. If the dominant allele for either of these genes is present, the result is triangular seeds. That is, every possible genotype other than *aabb* results in triangular seeds, and a cross between heterozygotes for both genes (*AaBb* × *AaBb*) would yield offspring with a phenotypic ratio of 15 triangular:1 ovoid.

As you work through genetics problems, keep in mind that any single characteristic that results in a phenotypic ratio that totals 16 is typical of a two-gene interaction. Recall the phenotypic inheritance pattern for Mendel's dihybrid cross, which considered two noninteracting genes—9:3:3:1. Similarly, we would expect interacting gene pairs to also exhibit ratios expressed as 16 parts. Note that we are assuming the interacting genes are not linked; they are still assorting independently into gametes.

LINK TO LEARNING

For an excellent review of Mendel's experiments and to perform your own crosses and identify patterns of inheritance, visit the [Mendel's Peas](http://openstax.org/l/mendels_peas) (http://openstax.org/l/mendels_peas) web lab.

KEY TERMS

- allele** gene variations that arise by mutation and exist at the same relative locations on homologous chromosomes
- autosomes** any of the non-sex chromosomes
- blending theory of inheritance** hypothetical inheritance pattern in which parental traits are blended together in the offspring to produce an intermediate physical appearance
- codominance** in a heterozygote, complete and simultaneous expression of both alleles for the same characteristic
- continuous variation** inheritance pattern in which a character shows a range of trait values with small gradations rather than large gaps between them
- dihybrid** result of a cross between two true-breeding parents that express different traits for two characteristics
- discontinuous variation** inheritance pattern in which traits are distinct and are transmitted independently of one another
- dominant** trait which confers the same physical appearance whether an individual has two copies of the trait or one copy of the dominant trait and one copy of the recessive trait
- dominant lethal** inheritance pattern in which an allele is lethal both in the homozygote and the heterozygote; this allele can only be transmitted if the lethality phenotype occurs after reproductive age
- epistasis** antagonistic interaction between genes such that one gene masks or interferes with the expression of another
- F₁** first filial generation in a cross; the offspring of the parental generation
- F₂** second filial generation produced when F₁ individuals are self-crossed or fertilized with each other
- genotype** underlying genetic makeup, consisting of both physically visible and non-expressed alleles, of an organism
- hemizygous** presence of only one allele for a characteristic, as in X-linkage; hemizygosity makes descriptions of dominance and recessiveness irrelevant
- heterozygous** having two different alleles for a given gene on the homologous chromosome
- homozygous** having two identical alleles for a given gene on the homologous chromosome
- hybridization** process of mating two individuals that differ with the goal of achieving a certain characteristic in their offspring
- incomplete dominance** in a heterozygote, expression of two contrasting alleles such that the individual displays an intermediate phenotype
- law of dominance** in a heterozygote, one trait will conceal the presence of another trait for the same characteristic
- law of independent assortment** genes do not influence each other with regard to sorting of alleles into gametes; every possible combination of alleles is equally likely to occur
- law of segregation** paired unit factors (i.e., genes) segregate equally into gametes such that offspring have an equal likelihood of inheriting any combination of factors
- linkage** phenomenon in which alleles that are located in close proximity to each other on the same chromosome are more likely to be inherited together
- model system** species or biological system used to study a specific biological phenomenon to be applied to other different species
- monohybrid** result of a cross between two true-breeding parents that express different traits for only one characteristic
- P₀** parental generation in a cross
- phenotype** observable traits expressed by an organism
- product rule** probability of two independent events occurring simultaneously can be calculated by multiplying the individual probabilities of each event occurring alone
- Punnett square** visual representation of a cross between two individuals in which the gametes of each individual are denoted along the top and side of a grid, respectively, and the possible zygotic genotypes are recombined at each box in the grid
- recessive** trait that appears “latent” or non-expressed when the individual also carries a dominant trait for that same characteristic; when present as two identical copies, the recessive trait is expressed
- recessive lethal** inheritance pattern in which an allele is only lethal in the homozygous form; the heterozygote may be normal or have some altered, nonlethal phenotype
- reciprocal cross** paired cross in which the respective traits of the male and female in one cross become the respective traits of the female and male in the other cross
- sex-linked** any gene on a sex chromosome
- sum rule** probability of the occurrence of at least one of two mutually exclusive events is the sum of their individual probabilities
- test cross** cross between a dominant expressing individual with an unknown genotype and a homozygous recessive individual; the offspring phenotypes indicate whether the unknown parent is heterozygous or homozygous for the dominant trait
- trait** variation in the physical appearance of a heritable characteristic
- X-linked** gene present on the X, but not the Y chromosome

CHAPTER SUMMARY

12.1 Mendel's Experiments and the Laws of Probability

Working with garden pea plants, Mendel found that crosses between parents that differed by one trait produced F_1 offspring that all expressed the traits of one parent. Observable traits are referred to as dominant, and non-expressed traits are described as recessive. When the offspring in Mendel's experiment were self-crossed, the F_2 offspring exhibited the dominant trait or the recessive trait in a 3:1 ratio, confirming that the recessive trait had been transmitted faithfully from the original P_0 parent. Reciprocal crosses generated identical F_1 and F_2 offspring ratios. By examining sample sizes, Mendel showed that his crosses behaved reproducibly according to the laws of probability, and that the traits were inherited as independent events.

Two rules in probability can be used to find the expected proportions of offspring of different traits from different crosses. To find the probability of two or more independent events occurring together, apply the product rule and multiply the probabilities of the individual events. The use of the word "and" suggests the appropriate application of the product rule. To find the probability of two or more events occurring in combination, apply the sum rule and add their individual probabilities together. The use of the word "or" suggests the appropriate application of the sum rule.

12.2 Characteristics and Traits

When true-breeding or homozygous individuals that differ for a certain trait are crossed, all of the offspring will be heterozygotes for that trait. If the traits are inherited as dominant and recessive, the F_1 offspring will all exhibit the same phenotype as the parent homozygous for the dominant trait. If these heterozygous offspring are self-crossed, the resulting F_2 offspring will be equally likely to inherit gametes carrying the dominant or recessive trait, giving rise to offspring of which one quarter are homozygous dominant, half are heterozygous, and one quarter are homozygous recessive. Because homozygous dominant and heterozygous individuals are phenotypically identical, the observed traits in the F_2 offspring will exhibit a ratio of three dominant to one recessive.

Alleles do not always behave in dominant and recessive patterns. Incomplete dominance describes situations in which the heterozygote exhibits a phenotype that is intermediate between the homozygous phenotypes. Codominance describes the simultaneous expression of both of the alleles in the heterozygote. Although diploid

organisms can only have two alleles for any given gene, it is common for more than two alleles of a gene to exist in a population. In humans, as in many animals and some plants, females have two X chromosomes and males have one X and one Y chromosome. Genes that are present on the X but not the Y chromosome are said to be X-linked, such that males only inherit one allele for the gene, and females inherit two. Finally, some alleles can be lethal. Recessive lethal alleles are only lethal in homozygotes, but dominant lethal alleles are fatal in heterozygotes as well.

12.3 Laws of Inheritance

Mendel postulated that genes (characteristics) are inherited as pairs of alleles (traits) that behave in a dominant and recessive pattern. Alleles segregate into gametes such that each gamete is equally likely to receive either one of the two alleles present in a diploid individual. In addition, genes are assorted into gametes independently of one another. That is, alleles are generally not more likely to segregate into a gamete with a particular allele of another gene. A dihybrid cross demonstrates independent assortment when the genes in question are on different chromosomes or distant from each other on the same chromosome. For crosses involving more than two genes, use the forked line or probability methods to predict offspring genotypes and phenotypes rather than a Punnett square.

Although chromosomes sort independently into gametes during meiosis, Mendel's law of independent assortment refers to genes, not chromosomes, and a single chromosome may carry more than 1,000 genes. When genes are located in close proximity on the same chromosome, their alleles tend to be inherited together. This results in offspring ratios that violate Mendel's law of independent assortment. However, recombination serves to exchange genetic material on homologous chromosomes such that maternal and paternal alleles may be recombined on the same chromosome. This is why alleles on a given chromosome are not always inherited together. Recombination is a random event occurring anywhere on a chromosome. Therefore, genes that are far apart on the same chromosome are likely to still assort independently because of recombination events that occurred in the intervening chromosomal space.

Whether or not they are sorting independently, genes may interact at the level of gene products such that the expression of an allele for one gene masks or modifies the expression of an allele for a different gene. This is called epistasis.

VISUAL CONNECTION QUESTIONS

- Figure 12.5** In pea plants, round peas (*R*) are dominant to wrinkled peas (*r*). You do a test cross between a pea plant with wrinkled peas (genotype *rr*) and a plant of unknown genotype that has round peas. You end up with three plants, all which have round peas. From this data, can you tell if the round pea parent plant is homozygous dominant or heterozygous? If the round pea parent plant is heterozygous, what is the probability that a random sample of 3 progeny peas will all be round?
- Figure 12.6** What are the genotypes of the individuals labeled 1, 2, and 3?
- Figure 12.12** What ratio of offspring would result from a cross between a white-eyed male and a female that is heterozygous for red eye color?
- Figure 12.16** In pea plants, purple flowers (*P*) are dominant to white flowers (*p*) and yellow peas (*Y*) are dominant to green peas (*y*). What are the possible genotypes and phenotypes for a cross between *PpYY* and *ppYy* pea plants? How many squares do you need to do a Punnett square analysis of this cross?

REVIEW QUESTIONS

- Mendel performed hybridizations by transferring pollen from the _____ of the male plant to the female ova.
 - anther
 - pistil
 - stigma
 - seed
- Which is one of the seven characteristics that Mendel observed in pea plants?
 - flower size
 - seed texture
 - leaf shape
 - stem color
- Imagine you are performing a cross involving seed color in garden pea plants. What *F*₁ offspring would you expect if you cross true-breeding parents with green seeds and yellow seeds? Yellow seed color is dominant over green.
 - 100 percent yellow-green seeds
 - 100 percent yellow seeds
 - 50 percent yellow, 50 percent green seeds
 - 25 percent green, 75 percent yellow seeds
- Consider a cross to investigate the pea pod texture trait, involving constricted or inflated pods. Mendel found that the traits behave according to a dominant/recessive pattern in which inflated pods were dominant. If you performed this cross and obtained 650 inflated-pod plants in the *F*₂ generation, approximately how many constricted-pod plants would you expect to have?
 - 600
 - 165
 - 217
 - 468
- A scientist pollinates a true-breeding pea plant with violet, terminal flowers with pollen from a true-breeding pea plant with white, axial flowers. Which of the following observations would most accurately describe the *F*₂ generation?
 - 75% violet flowers; 75% terminal flowers
 - 75% white flowers in a terminal position
 - 75% violet flowers; 75% axial flowers
 - 75% violet flowers in an axial position
- The observable traits expressed by an organism are described as its _____.
 - phenotype
 - genotype
 - alleles
 - zygote
- A recessive trait will be observed in individuals that are _____ for that trait.
 - heterozygous
 - homozygous or heterozygous
 - homozygous
 - diploid
- If black and white true-breeding mice are mated and the result is all gray offspring, what inheritance pattern would this be indicative of?
 - dominance
 - codominance
 - multiple alleles
 - incomplete dominance

- 13.** The ABO blood groups in humans are expressed as the I^A , I^B , and i alleles. The I^A allele encodes the A blood group antigen, I^B encodes B, and i encodes O. Both A and B are dominant to O. If a heterozygous blood type A parent ($I^A i$) and a heterozygous blood type B parent ($I^B i$) mate, one quarter of their offspring will have AB blood type ($I^A I^B$) in which both antigens are expressed equally. Therefore, ABO blood groups are an example of:
- multiple alleles and incomplete dominance
 - codominance and incomplete dominance
 - incomplete dominance only
 - multiple alleles and codominance
- 14.** In a mating between two individuals that are heterozygous for a recessive lethal allele that is expressed *in utero*, what genotypic ratio (homozygous dominant:heterozygous:homozygous recessive) would you expect to observe in the offspring?
- 1:2:1
 - 3:1:1
 - 1:2:0
 - 0:2:1
- 15.** If the allele encoding polydactyly (six fingers) is dominant why do most people have five fingers?
- Genetic elements suppress the polydactyl gene.
 - Polydactyly is embryonic lethal.
 - The sixth finger is removed at birth.
 - The polydactyl allele is very rare in the human population.
- 16.** A farmer raises black and white chickens. To his surprise, when the first generation of eggs hatch all the chickens are black with white speckles throughout their feathers. What should the farmer expect when the eggs laid after interbreeding the speckled chickens hatch?
- All the offspring will be speckled.
 - 75% of the offspring will be speckled, and 25% will be black.
 - 50% of the offspring will be speckled, 25% will be black, and 25% will be white.
 - 50% of the offspring will be black and 50% of the offspring will be white.
- 17.** Assuming no gene linkage, in a dihybrid cross of $AABB \times aabb$ with $AaBb$ F_1 heterozygotes, what is the ratio of the F_1 gametes (AB , aB , Ab , ab) that will give rise to the F_2 offspring?
- 1:1:1:1
 - 1:3:3:1
 - 1:2:2:1
 - 4:3:2:1
- 18.** The forked line and probability methods make use of what probability rule?
- test cross
 - product rule
 - monohybrid rule
 - sum rule
- 19.** How many different offspring genotypes are expected in a trihybrid cross between parents heterozygous for all three traits when the traits behave in a dominant and recessive pattern? How many phenotypes?
- 64 genotypes; 16 phenotypes
 - 16 genotypes; 64 phenotypes
 - 8 genotypes; 27 phenotypes
 - 27 genotypes; 8 phenotypes
- 20.** Labrador retrievers' fur color is controlled by two alleles, E and B. Any dog with the ee__ genotype develops into a yellow lab, while B_E__ dogs become black labs and bbE__ dogs become chocolate labs. This is an example of ____.
- epistasis
 - codominance
 - incomplete dominance
 - linkage
- 21.** Which of the following situations does **not** follow the Law of Independent Assortment?
- A blond man and a brunette woman produce three offspring over time, all of who have blond hair.
 - A white cow crossed with a brown bull produces roan cattle.
 - Mating a hog with a sow produces six female piglets.
 - Men are more likely to experience hemophilia than women.

CRITICAL THINKING QUESTIONS

- 22.** Describe one of the reasons why the garden pea was an excellent choice of model system for studying inheritance.
- 23.** How would you perform a reciprocal cross for the characteristic of stem height in the garden pea?
- 24.** Mendel performs a cross using a true-breeding pea plant with round, yellow seeds and a true-breeding pea plant with green, wrinkled seeds. What is the probability that offspring will have green, round seeds? Calculate the probability for the F_1 and F_2 generations.
- 25.** Calculate the probability of selecting a heart or a face card from a standard deck of cards. Is this outcome more or less likely than selecting a heart suit face card?

26. The gene for flower position in pea plants exists as axial or terminal alleles. Given that axial is dominant to terminal, list all of the possible F_1 and F_2 genotypes and phenotypes from a cross involving parents that are homozygous for each trait. Express genotypes with conventional genetic abbreviations.
27. Use a Punnett square to predict the offspring in a cross between a dwarf pea plant (homozygous recessive) and a tall pea plant (heterozygous). What is the phenotypic ratio of the offspring?
28. Can a human male be a carrier of red-green color blindness?
29. Why is it more efficient to perform a test cross with a homozygous recessive donor than a homozygous dominant donor? How could the same information still be found with a homozygous dominant donor?
30. Use the probability method to calculate the genotypes and genotypic proportions of a cross between $AABBCC$ and $Aabbcc$ parents.
31. Explain epistasis in terms of its Greek-language roots “standing upon.”
32. In Section 12.3, “Laws of Inheritance,” an example of epistasis was given for the summer squash. Cross white $WwYy$ heterozygotes to prove the phenotypic ratio of 12 white:3 yellow:1 green that was given in the text.
33. People with trisomy 21 develop Down’s syndrome. What law of Mendelian inheritance is violated in this disease? What is the most likely way this occurs?
34. A heterozygous pea plant produces violet flowers and yellow, round seeds. Describe the expected genotypes of the gametes produced by Mendelian inheritance. If all three genes are found on the same arm of one chromosome should a scientist predict that inheritance patterns will follow Mendelian genetics?