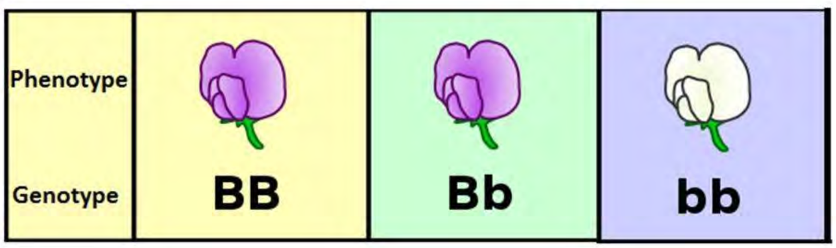
Mendel's hypotheses were based on the physical characteristic that he could observe. An organism's observable physical traits are referred to as its **phenotype**; for example, violet or white flowers (Figure 9.6). Mendel could not examine an organism's genetic makeup. He made inferences on whether an organism was homozygous or heterozygous for a particular gene but could not provide genomic data that supported this. An organism's underlying genetic makeup is called its **genotype** (Figure 9.6). A genotype is usually denoted by using two of the same letters (Figure 9.6). The letter that is used is often the first letter of the dominant trait, but geneticists prefer to use letters that have distinct upper- and lower-case forms (P and p may be mistaken for each other, while B and b are more distinct). The genotype may be two upper case letters, two lower case letters, or an upper and a lower-case letter (BB, bb, or Bb).



9.6 Phenotype shows an organism's physical observable traits, whereas genotype is an organism's genetic makeup. (credit: Modified by Elizabeth O'Grady original work of Madeleine Price Ball)

What do the genotype letters represent? The P generation plants that Mendel used in his experiments were each homozygous for the trait he was studying, meaning that for a given gene, it had two identical alleles for that gene. Genotypes of individuals that have two identical alleles are represented by either two identical upper-case letters (BB) which represent homozygous dominant individuals, or two identical lower-case letters (bb) which represent homozygous recessive individuals. The dominant allele is capitalized, and the recessive allele is lower case. When P plants with contrasting traits, for example, violet flowers (BB) vs. white flowers (bb), were cross-fertilized, all offspring were heterozygous. Heterozygous plants have two different alleles, one violet and one white, from each corresponding parent. The heterozygous genotype is denoted by one upper-case letter and one lower-case letter (Bb). It is the phenotype that is observed in a heterozygous individual that determines which trait is dominant and which trait is recessive.

Monohybrid cross and Punnett Square

Mendel's cross-fertilization experiments demonstrate the difference between phenotype and genotype. When fertilization occurs between two true-breeding parents that differ in only one characteristic, the process is called a **monohybrid cross**.

To demonstrate a monohybrid cross, consider the case of true-breeding pea plants with yellow seeds versus green seeds. The dominant seed color is yellow; therefore, the parental genotypes were *YY* for the homozygous dominant plants with yellow seeds and *yy* for the homozygous recessive plants with green seeds. A **Punnett square**, devised by the British geneticist Reginald Punnett, can be drawn that applies the rules of probability to predict the possible genotype outcomes of a genetic cross and their expected frequencies. To prepare a Punnett square, a table is drawn where all possible combinations of the parental alleles are listed along the top for one parent, and all possible combinations of the second parental alleles are listed on the left side of the table (Figure 9.7). This allows the alleles to be separated into separate boxes, which represent their meiotic separation into haploid gametes. The different 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 fertilization event. Because each possibility is equally likely, genotypic ratios can be determined from a Punnett square. If the pattern of inheritance 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 F_1 offspring are heterozygous, *Yy,* and have yellow seeds because they have a dominant allele (Figure 9.7).

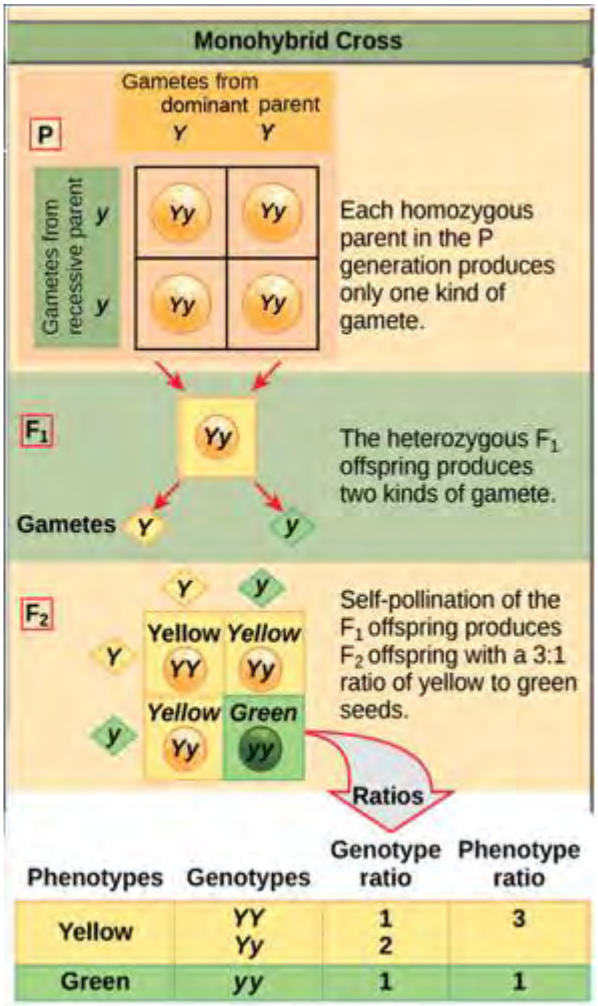


Figure 9.7 This Punnett square shows the cross between plants with yellow seeds and green seeds. The cross between the true-breeding P plants produces F_1 heterozygotes that can be self-fertilized. The self-fertilization of the F_1 generation can be analyzed with a Punnett square to predict the genotypes of the F_2 generation. Given an inheritance pattern of dominant–recessive, the genotypic and phenotypic ratios can then be determined. (credit: Modified by Elizabeth O'Grady original work of Clark et al. / Biology 2E OpenStax)

A self-cross of one of the *Yy* heterozygous F_1 offspring can also be represented in a Punnett square. 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 possibilities must be counted. 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 9.7). Furthermore, the *YY* and *Yy* offspring all have yellow seeds and are phenotypically identical. Therefore, we expect the offspring to exhibit a phenotypic ratio of 3 yellow:1 green. In all the characteristics that Mendel observed, he found this ratio in every F_2 generation.

Mendel's law of independent assortment and Dihybrid cross

Mendel's **law of independent assortment** states that genes do not influence each other with regard to the sorting of alleles into gametes. It also states that 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 pea plant has yellow, round seeds (*YYRR*), and is crossed with a different pea plant that has green, wrinkled 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 9.8).

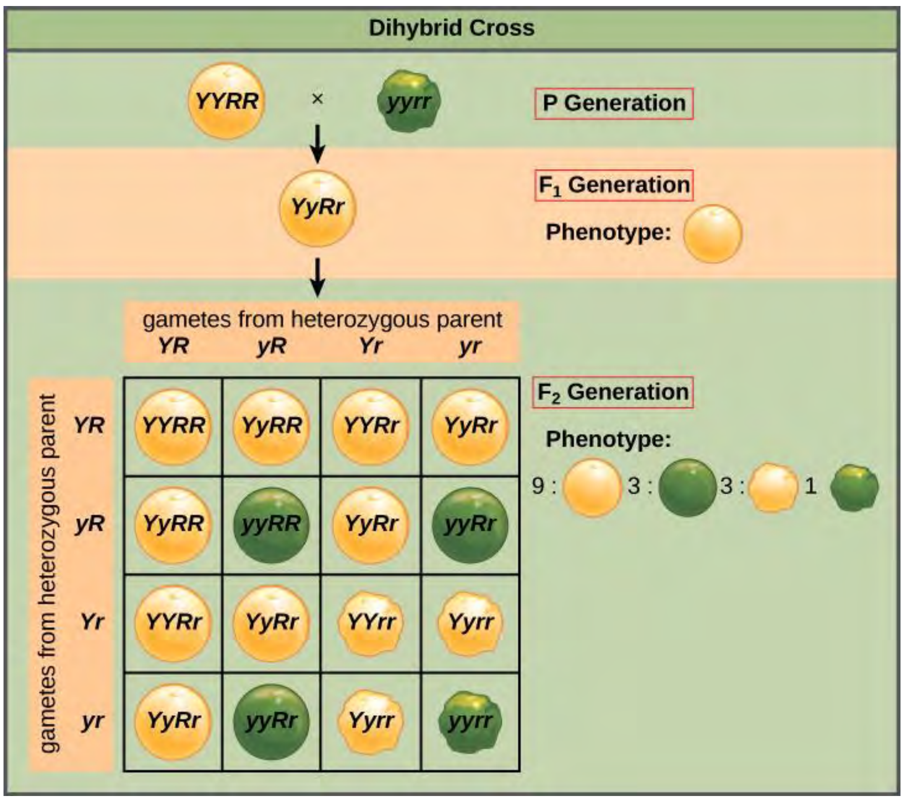


Figure 9.8 A dihybrid cross in pea plants involves the genes for seed color and texture. The P cross produces F_1 offspring that are all heterozygous for both characteristics. The resulting 9:3:3:1 F_2 phenotypic ratio is obtained using a Punnett square. (credit: Fowler et al. / Concepts of Biology OpenStax)

For the F_2 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 possible gametes that can be formed when the *YyRr* heterozygote is self-crossed: *YR*, *Yr*, *yR*, and *yr*. Arranging these gametes along the top and left of a four × four Punnett square (Figure 9.8) gives us 16 equally likely genotypic