CONCEPTS OF BIOLOGY

Chapter 8 PATTERNS OF INHERITANCE

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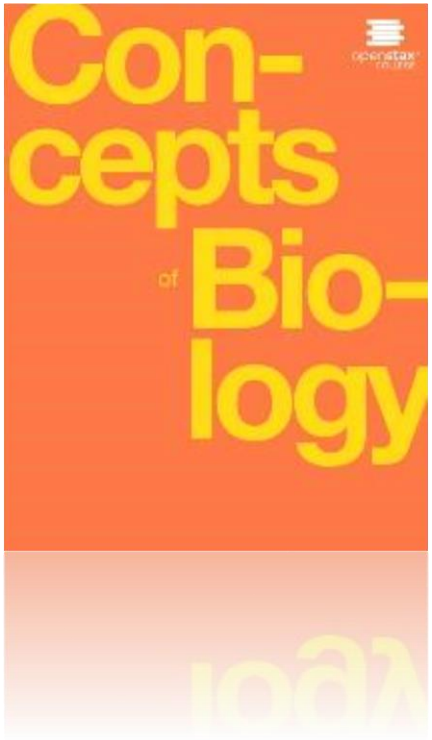

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Introduction

- **Genetics** - the study of heredity
- **Gregor Mendel** set the framework for genetics long before chromosomes or genes had been identified
- Mendel conducted methodical, quantitative analyses using large sample sizes and revealed the fundamental principles of heredity
- We now know that genes, carried on chromosomes, are the basic functional units of heredity with the ability to be replicated, expressed, or mutated

FIGURE 8.1

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

FIGURE 8.2

Johann Gregor Mendel set the framework for the study of genetics.

8.1 MENDEL'S EXPERIMENTS (1 of 4)

- Gregor Mendel, (1822–1884) supported by the monastery, 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 pea plants
- 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 faithfully from parents to offspring in specific patterns

8.1 Mendel's Experiments (2 of 4)

- Mendel's work went virtually unnoticed by the scientific community, which incorrectly believed that the process of inheritance involved a blending of parental traits that produced an intermediate physical appearance in offspring
- This hypothetical process appeared to be correct because of what we know now as continuous variation

8.1 Mendel's Experiments (3 of 4)

- Mendel worked instead with traits that show discontinuous variation
- **Discontinuous variation** - the variation seen among individuals when each individual shows one of two—or a very few—easily distinguishable traits, such as violet or white flowers
- Mendel's choice of these kinds of traits allowed him to see experimentally that the traits were not blended in the offspring, but that they were inherited as distinct traits

8.1 Mendel's Experiments (4 of 4)

- 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 Crosses (1 of 4)

- Mendel's work was accomplished using the garden pea, *Pisum sativum*, to study inheritance
- This species naturally self-fertilizes
- 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

Mendel's Crosses (2 of 4)

- 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
- Plants used in first-generation crosses were called **P**, or **parental generation**, plants (Figure 8.3)

Mendel's Crosses (3 of 4)

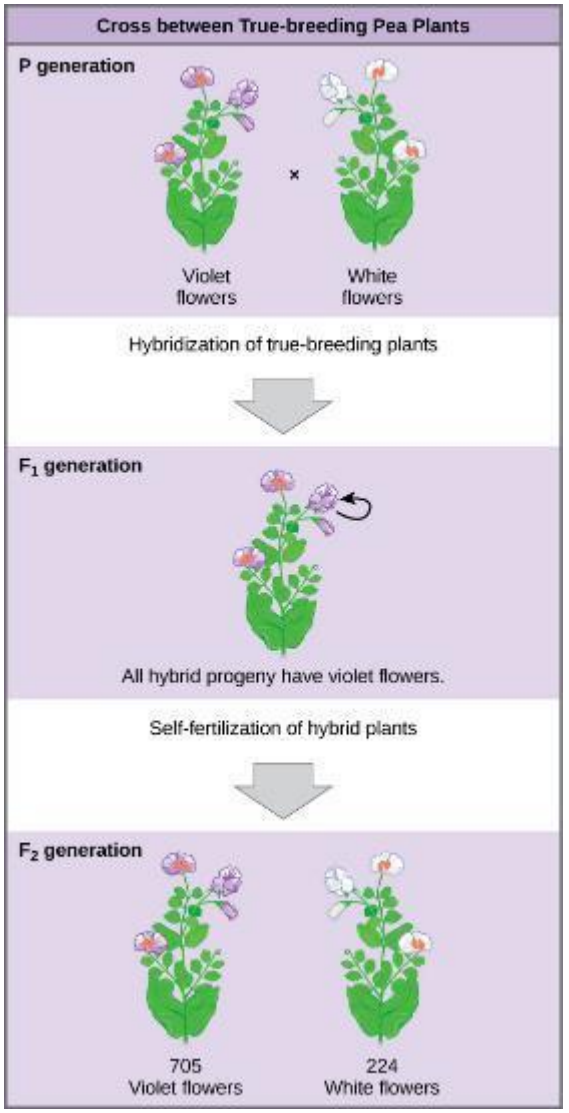
- Mendel collected the seeds produced by the P plants that resulted from each cross and grew them the following season
- These offspring were called the **F_1**, or the **first filial generation** (filial = daughter or son)
- Once Mendel examined the characteristics in the F_1 generation of plants, he allowed them to self-fertilize naturally
- He then collected and grew the seeds from the F_1 plants to produce the **F_2**, or **second filial generation**

Mendel's Crosses (4 of 4)

- Mendel's experiments extended beyond the F_2 generation to the F_3 generation, F_4 generation, and so on, but it was the ratio of characteristics in the P, F_1, and F_2 generations that were the most intriguing and became the basis of Mendel's postulates

FIGURE 8.3

Mendel's process for performing crosses included examining flower color.



Garden Pea Characteristics Revealed the Basics of Heredity (1 of 11)

- Mendel reported the results of his crosses involving seven different characteristics, each with two contrasting traits
- **Trait** - 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

Garden Pea Characteristics Revealed the Basics of Heredity (2 of 11)

- Mendel generated large numbers of F_1 and F_2 plants and reported results from thousands of F_2 plants
- What results did Mendel find in his crosses for flower color?

Garden Pea Characteristics Revealed the Basics of Heredity (3 of 11)

- First, Mendel confirmed that he was using plants that bred true for white or violet flower color, meaning 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

Garden Pea Characteristics Revealed the Basics of Heredity (4 of 11)

- In addition, Mendel confirmed that, other than flower color, the pea plants were physically identical
- This was an important check to make sure that the two varieties of pea plants only differed with respect to one trait, flower color
- Mendel then applied the pollen from a plant with violet flowers to the stigma of a plant with white flowers (cross-pollination)
- Mendel found that 100 percent of the F_1 hybrid generation had violet flowers

Garden Pea Characteristics Revealed the Basics of Heredity (5 of 11)

- At that time, it would have been 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 had completely disappeared in the F_1 generation

Garden Pea Characteristics Revealed the Basics of Heredity (6 of 11)

- Importantly, Mendel did not stop his experimentation there
- He allowed the F_1 plants to self-fertilize and found that 705 plants in the F_2 generation had violet flowers and 224 had white flowers
- This was a ratio of 3:1 violet flowers to one white flower

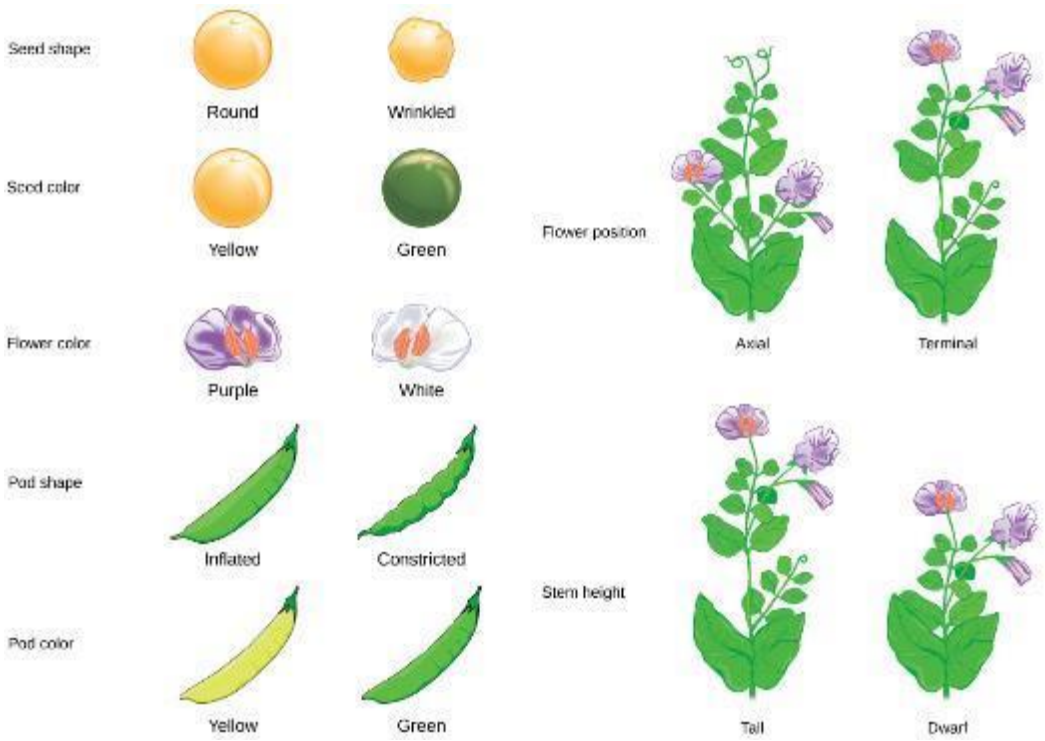
Garden Pea Characteristics Revealed the Basics of Heredity (7 of 11)

- When Mendel transferred pollen from a plant with violet flowers to the stigma of a plant with white flowers and vice versa, he obtained approximately the same ratio irrespective 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

Garden Pea Characteristics Revealed the Basics of Heredity (8 of 11)

- For the other six characteristics that Mendel examined, the F_1 and F_2 generations behaved in the same way that they behaved for flower color
- One of the two traits would disappear completely from the F_1 generation, only to reappear in the F_2 generation at a ratio of roughly 3:1 (Figure 8.4)

FIGURE 8.4



Mendel identified seven pea plant characteristics.

Garden Pea Characteristics Revealed the Basics of Heredity (9 of 11)

- 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 dominant and recessive traits, respectively
- **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

Garden Pea Characteristics Revealed the Basics of Heredity (10 of 11)

- An example of a dominant trait is the violet colored 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 (and were not blended) in the plants of the F_1 generation
- Mendel proposed that the plants possessed two copies of the trait for the flower color characteristic, and each parent transmitted one of their two copies to their offspring, where they came together

Garden Pea Characteristics Revealed the Basics of Heredity (11 of 11)

- 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

8.2 LAWS OF INHERITANCE (1 of 4)

- The seven characteristics that Mendel evaluated in his pea plants were each expressed as one of two traits
- Mendel deduced from his results that each individual had two discrete copies of the characteristic that are passed individually to offspring
- We now call those two copies **genes**, which are carried on chromosomes

8.2 LAWS OF INHERITANCE (2 of 4)

- The reason we have two copies of each gene is that we inherit one from each parent
- In fact, it is the chromosomes we inherit and the two copies of each gene are located on paired chromosomes
- Recall that in meiosis these chromosomes are separated out into haploid gametes
- This separation, or **segregation**, of the homologous chromosomes also means that only one of the copies of the gene gets moved into a gamete

8.2 LAWS OF INHERITANCE (3 of 4)

- The offspring are formed when one gamete unites with one from the other parent and the two copies of each gene (and chromosome) are restored
- 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
- For example, one individual may carry a gene that determines white flower color and a gene that determines violet flower color

8.2 LAWS OF INHERITANCE (4 of 4)

- 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 (1 of 5)

- 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 the physically visible and the non-expressed alleles, is called its **genotype**

Phenotypes and Genotypes (2 of 5)

- Mendel's hybridization experiments demonstrate the difference between phenotype and genotype
- For example, seed color is governed by a single gene with two alleles
- The yellow-seed allele is dominant and the green-seed allele is recessive

Phenotypes and Genotypes (3 of 5)

- When true-breeding plants were cross-fertilized, in which one parent had yellow seeds and one had green seeds, all of the F_1 hybrid offspring had yellow seeds
- That is, the hybrid offspring were phenotypically identical to the true-breeding parent with yellow seeds even though we know that the allele donated by the parent with green seeds 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 seeds

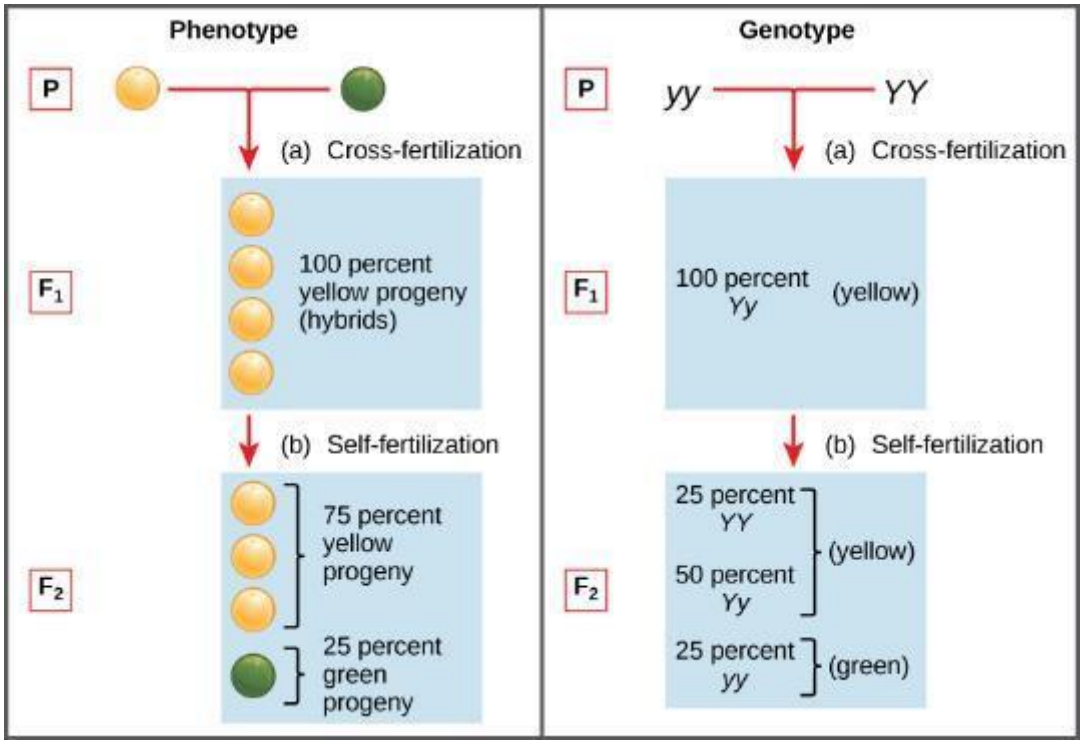
Phenotypes and Genotypes (4 of 5)

- Diploid organisms that are **homozygous** for a gene have two identical alleles, one on each of their homologous chromosomes
- The genotype is often written as YY or yy, for which each letter represents one of the two alleles in the genotype
- The dominant allele is capitalized and the recessive allele is lower case
- The letter used for the gene (seed color in this case) is usually related to the dominant trait (yellow allele, in this case, or “Y”)

Phenotypes and Genotypes (5 of 5)

- Mendel's parental pea plants always bred true because both produced gametes carried the same allele
- When P generation plants with contrasting traits were cross-fertilized, all of the offspring were heterozygous for the contrasting trait, meaning their genotype had different alleles for the gene being examined
- For example, the F_1 yellow plants that received a Y allele from their yellow parent and a y allele from their green parent had the genotype Yy

FIGURE 8.5



Phenotypes are physical expressions of traits that are transmitted by alleles. Capital letters represent dominant alleles and lowercase letters represent recessive alleles. The phenotypic ratios are the ratios of visible characteristics. The genotypic ratios are the ratios of gene combinations in the offspring, and these are not always distinguishable in the phenotypes.

Law of Dominance (1 of 3)

- 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

Law of Dominance (2 of 3)

- We now know that these so-called unit factors are actually genes on homologous chromosomes
- 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 the same phenotype but different genotypes)
- The recessive allele will only be observed in homozygous recessive individuals (Table 8.1)

Correspondence of Genotype and Phenotype

Correspondence between Genotype and Phenotype for a Dominant-Recessive Characteristic.

	Homozygous	Heterozygous	Homozygous
Genotype	*YY*	*Yy*	*yy*
Phenotype	yellow	yellow	green

Table 8.1

Law of Dominance (3 of 3)

- Mendel's **law of dominance** states that in a heterozygote, one trait will conceal the presence of another trait for the same characteristic
- For example, when crossing true-breeding violet-flowered plants with true-breeding white-flowered plants, all of the offspring were violet-flowered, even though they all had one allele for violet and one allele for white

FIGURE 8.6

The allele for albinism, expressed here in humans, is recessive. Both of this child's parents carried the recessive allele.

Monohybrid Cross and the Punnett Square (1 of 6)

- When fertilization occurs between two true-breeding parents that differ by only the characteristic being studied, the process is called a **monohybrid cross**, and the resulting offspring are called **monohybrids**
- Mendel performed seven types of monohybrid crosses, each involving contrasting traits for different characteristics

Monohybrid Cross and the Punnett Square (2 of 6)

- Out of these crosses, all of the F_1 offspring had the phenotype of one parent, and the F_2 offspring had a 3:1 phenotypic ratio
- The results of Mendel's research can be explained mathematically in terms of probabilities

Monohybrid Cross and the Punnett Square (3 of 6)

- To demonstrate this with a monohybrid cross, consider the case of true-breeding pea plants with yellow versus green 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
- A **Punnett square** (devised by the British geneticist Reginald Punnett) is useful for determining probabilities because it is drawn to predict all possible outcomes of all possible random fertilization events and their expected frequencies

Monohybrid Cross and the Punnett Square (4 of 6)

- Figure 8.9 shows a Punnett square for a cross between a plant with yellow peas and one with green peas
- To prepare a Punnett square, all possible combinations of the parental alleles (the genotypes of the gametes) are listed along the top (for one parent) and side (for the other parent) of a grid
- The combinations of egg and sperm gametes are then made in the boxes in the table on the basis of which alleles are combining
- Each box then represents the diploid genotype of a zygote, or fertilized egg

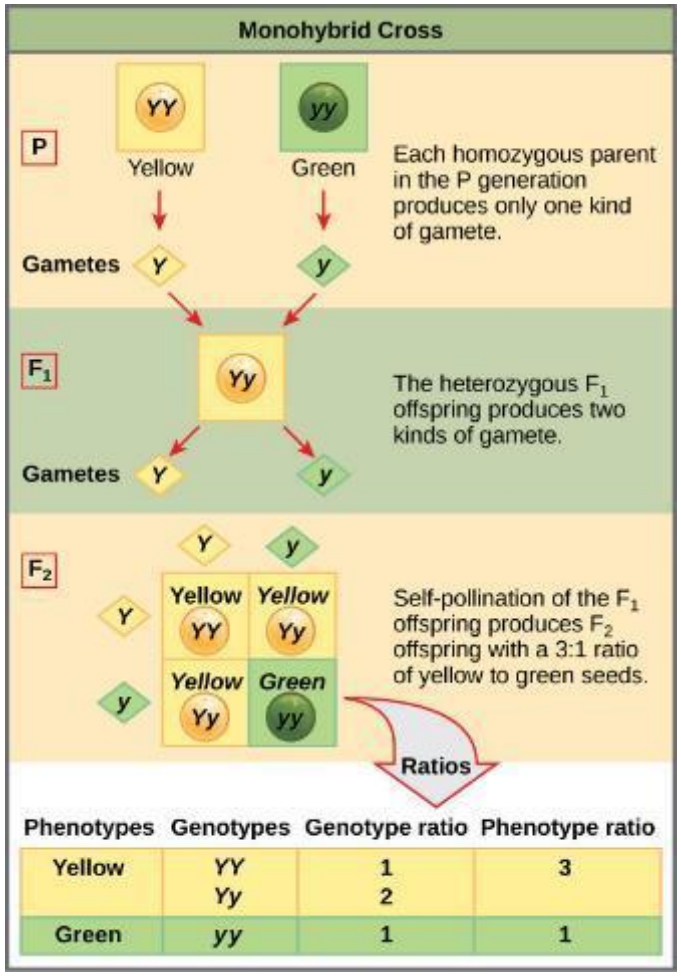
Monohybrid Cross and the Punnett Square (5 of 6)

- Because each possibility is equally likely, genotypic ratios can be determined from a Punnett square
- If the pattern of inheritance (dominant and recessive) is known, the phenotypic ratios can be inferred as well
- In this case, only one genotype is possible in the F_1 offspring: All offspring are Yy and have yellow seeds

Monohybrid Cross and the Punnett Square (6 of 6)

- When the F_1 offspring are crossed with each other, the result is a 25% (1 in 4) probability of the offspring having YY genotype; a 50% (2 in 4) probability of the offspring having Yy; and a 25% (1 in 4) probability of the offspring having yy genotype
- When counting all four possible outcomes, there is a 75% (3 in 4) probability of offspring having the yellow phenotype and a 25% (1 in 4) probability of offspring having the green phenotype
- This explains why the results of Mendel's F_2 generation occurred in a 3:1 phenotypic ratio

FIGURE 8.9



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-cross 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.

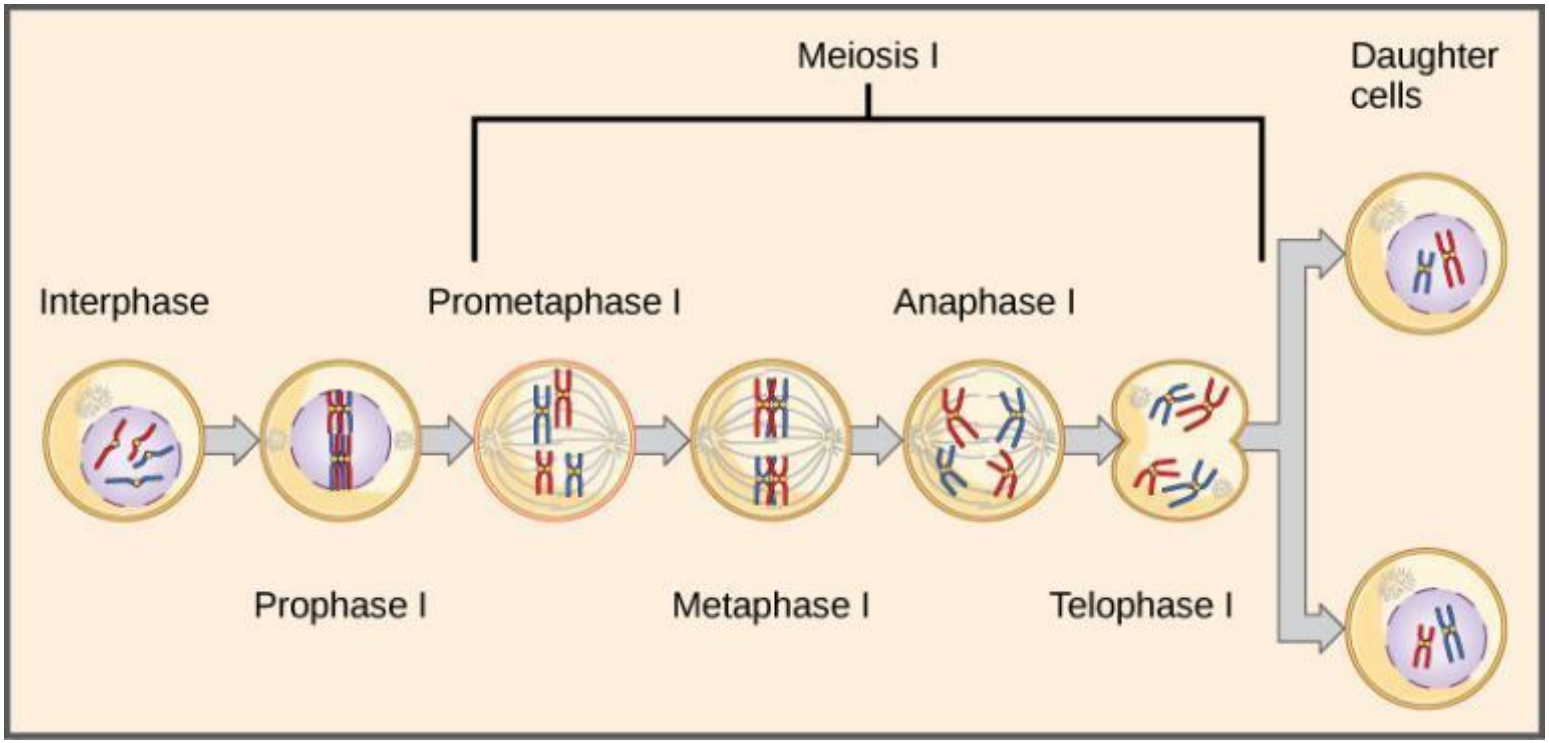
Law of Segregation (1 of 2)

- 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

Law of Segregation (2 of 2)

- 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 (but remember, this process was not understood by the scientific community during Mendel's lifetime)

FIGURE 8.7



The first division in meiosis is shown.

Test Cross (1 of 2)

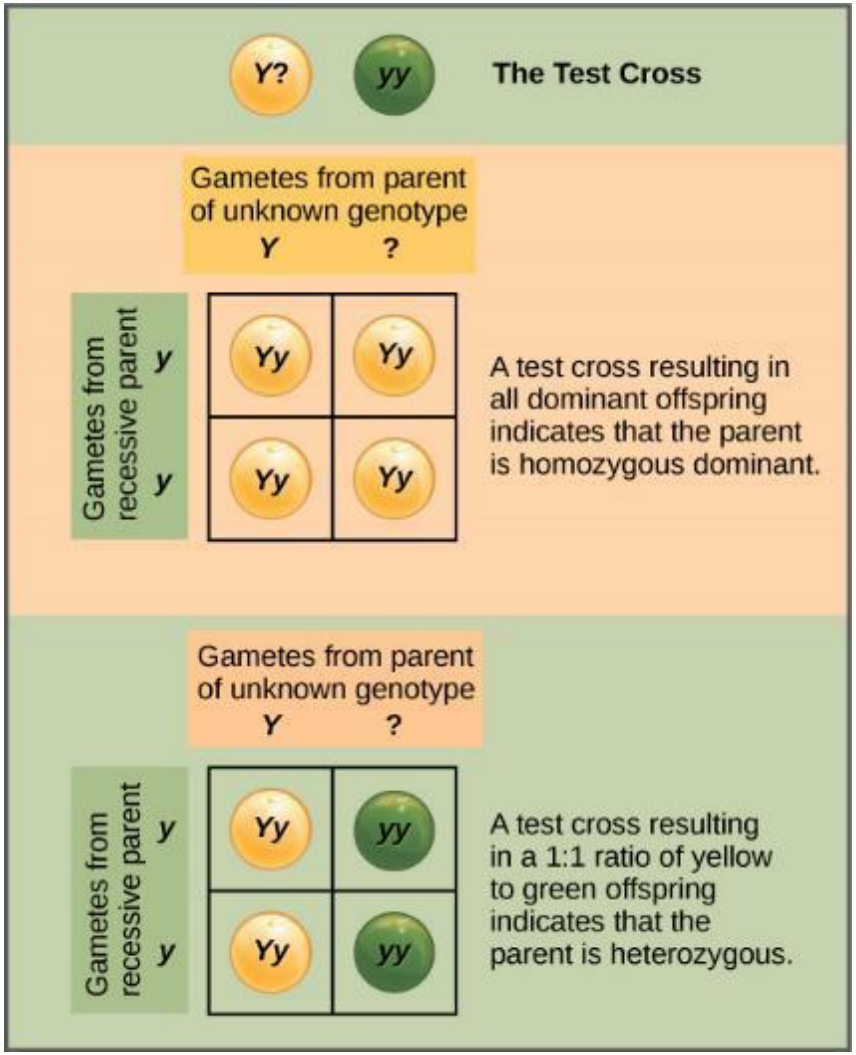
- Beyond predicting the offspring of a cross between known homozygous or heterozygous parents, Mendel also developed a way to determine whether an organism that expressed a dominant trait was a heterozygote or a homozygote
- This technique, called the test cross, is still used by plant and animal breeders

Test Cross (2 of 2)

- 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
- 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 8.8

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



Law of Independent Assortment (1 of 6)

- 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
- 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

Law of Independent Assortment (2 of 6)

- Consider the characteristics of seed color and seed texture for two pea plants, one that has wrinkled, green seeds (rryy) and another that has round, yellow seeds (RRYY)
- Because each parent is homozygous, the law of segregation indicates that the gametes for the wrinkled–green plant all are ry, and the gametes for the round–yellow plant are all RY. Therefore, the F_1 generation of offspring all are RrYy (Figure 8.10)

Law of Independent Assortment (3 of 6)

- The gametes produced by the F_1 individuals must have one allele from each of the two genes
- For example, a gamete could get the R allele or the r allele for the seed shape gene and either the Y or the y allele for the seed color gene

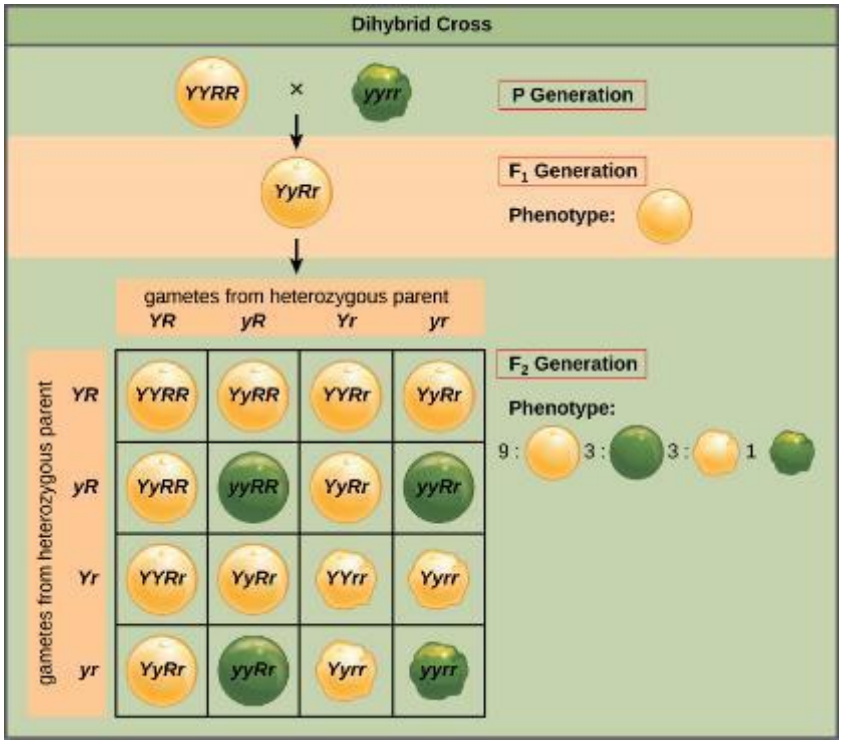
Law of Independent Assortment (4 of 6)

- The **law of independent assortment** states that a gamete into which an r allele is sorted would be equally likely to contain either a Y or a y allele
- Thus, there are four equally likely gametes that can be formed when the RrYy heterozygote is self-crossed, as follows: RY, rY, Ry, and ry

Law of Independent Assortment (5 of 6)

- Arranging these gametes along the top and left of a 4 × 4 Punnett square (Figure 8.10) gives us 16 equally likely genotypic combinations
- From these genotypes, we find a phenotypic ratio of 9 round–yellow:3 round–green:3 wrinkled–yellow:1 wrinkled–green (Figure 8.10)
- These are the offspring ratios we would expect, assuming we performed the crosses with a large enough sample size

FIGURE 8.10

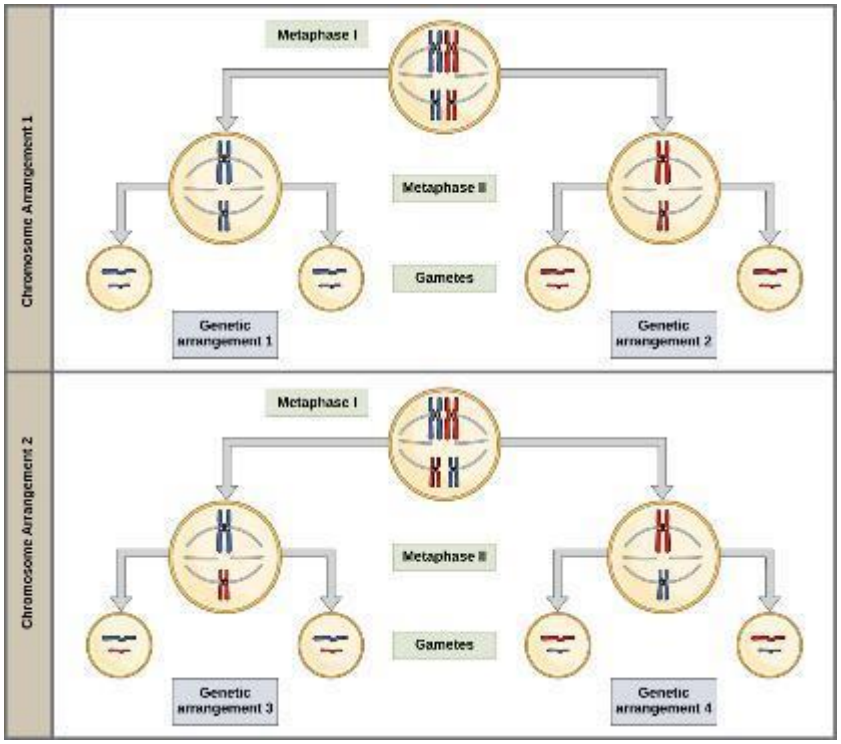


A dihybrid cross in pea plants involves the genes for seed color and texture. The P cross produces F_1 offspring that are all heterozygous for both characteristics. The resulting 9:3:3:1 F_2 phenotypic ratio is obtained using a Punnett square.

Law of Independent Assortment (6 of 6)

- 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 (Figure 8.11)

FIGURE 8.11



The random segregation into daughter nuclei that happens during the first division in meiosis can lead to a variety of possible genetic arrangements.

8.3 EXTENSIONS OF THE LAWS OF INHERITANCE

- Mendel studied traits with only one mode of inheritance in pea plants, the relatively simple pattern of dominant and recessive alleles for a single characteristic
- There are several important modes of inheritance (discovered after Mendel's work) that do not follow the dominant and recessive, single-gene model

Alternatives to Dominance and Recessiveness (1 of 2)

- Mendel's experiments with pea plants suggested that:
  1) two types of "units" or alleles exist for every gene
  2) alleles maintain their integrity in each generation (no blending)
  3) in the presence of the dominant allele, the recessive allele is hidden, with 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**"

Alternatives to Dominance and Recessiveness (2 of 2)

- Since then, genetic studies in other organisms have shown that much more complexity exists, but that the fundamental principles of Mendelian genetics still hold true
- Other types of inheritance patterns are considered **extensions of Mendelism**

Incomplete Dominance (1 of 3)

- Mendel's results, demonstrating 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

Incomplete Dominance (2 of 2)

- For example, in the snapdragon, *Antirrhinum majus* (Figure 8.12), a cross between a homozygous parent with white flowers (C^WC^W) and a homozygous parent with red flowers (C^RC^R) will produce offspring with pink flowers (C^RC^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**

Incomplete Dominance (3 of 3)

- 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^RC^R : 2 C^RC^W : 1 C^WC^W, and the phenotypic ratio would be 1:2:1 for red:pink:white

FIGURE 8.12

These pink flowers of a heterozygote snapdragon result from incomplete dominance. (credit: “storebukkebruse”/Flickr)

Codominance (1 of 3)

- 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 occurs in the ABO blood groups of humans

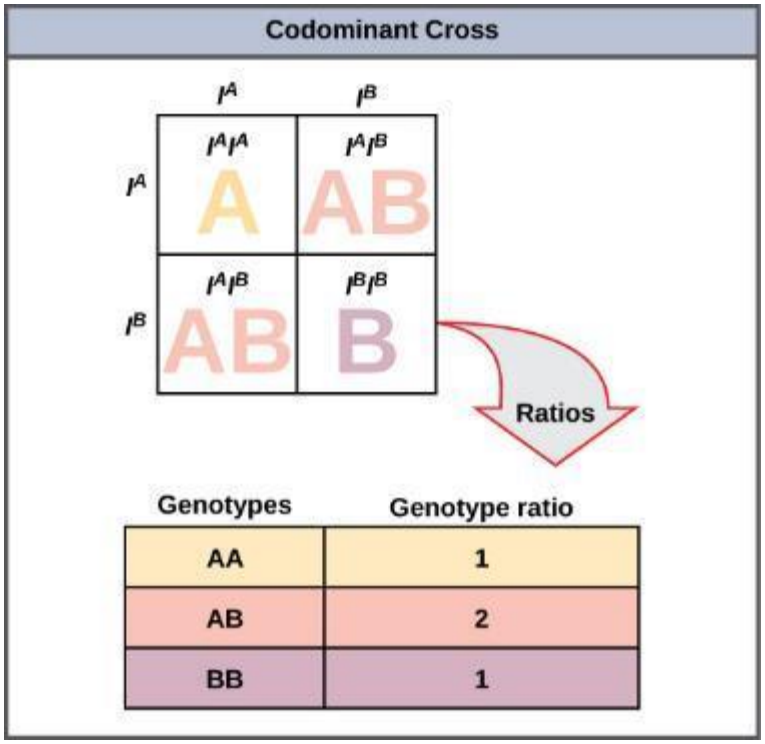
Codominance (2 of 3)

- The A and B alleles are expressed in the form of A or B molecules present on the surface of red blood cells
- Homozygotes (I^AI^A and I^BI^B) express either the A or the B phenotype, and heterozygotes (I^AI^B) express both phenotypes equally
- The I^AI^B individual has blood type AB

Codominance (3 of 3)

- 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 (Figure 8.13)

FIGURE 8.13



This Punnett square shows an AB/AB blood type cross

Multiple Alleles (1 of 4)

- 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

Multiple Alleles (2 of 4)

- Note that when many alleles exist for the same gene, the convention is to denote the most common phenotype or genotype in the natural population as the wild type (often abbreviated “+”)
- All other phenotypes or genotypes are considered variants (mutants) of this typical form, meaning they deviate from the wild type
- The variant may be recessive or dominant to the wild-type allele

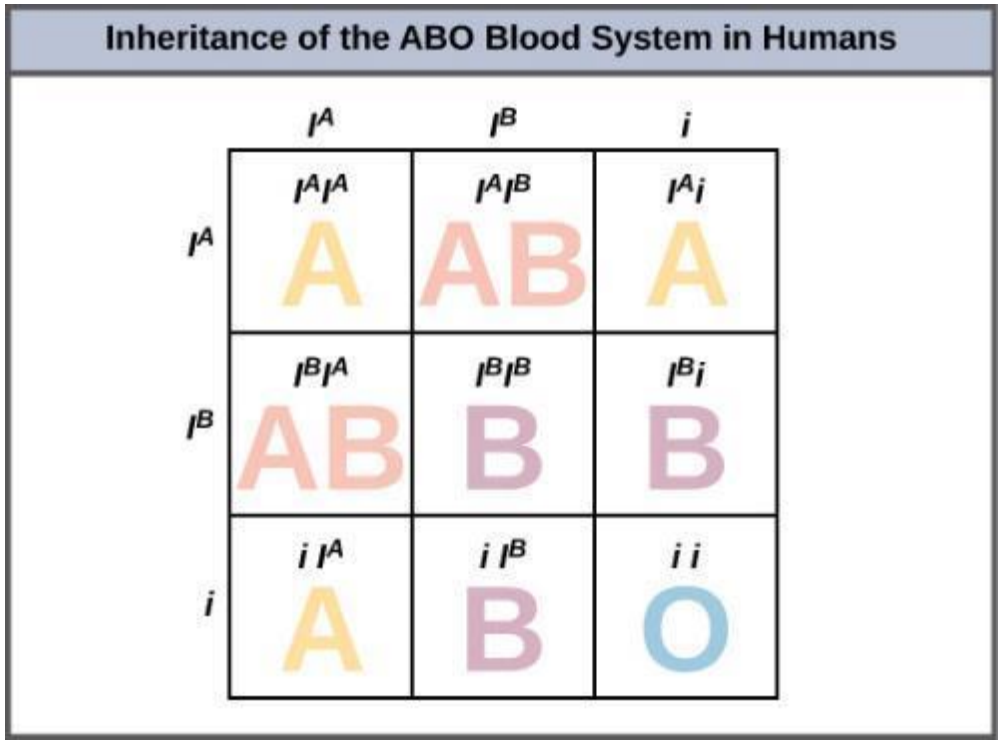
Multiple Alleles (3 of 4)

- An example of multiple alleles is the ABO blood-type system in humans
- In this case, there are three alleles circulating in the population: the I^A allele codes for A molecules on the red blood cells, the I^B allele codes for B molecules on the surface of red blood cells, and the i allele codes for no molecules on the red blood cells
- In this case, the I^A and I^B alleles are codominant with each other and are both dominant over the recessive i allele

Multiple Alleles (4 of 4)

- Although there are three alleles present in a population, each individual only gets two of the alleles, one from each of their parents
- This produces the genotypes and phenotypes shown in Figure 8.14
- Notice that instead of three genotypes, there are six different genotypes when there are three alleles
- The number of possible phenotypes depends on the dominance relationships between the three alleles

FIGURE 8.14



Inheritance of the ABO blood system in humans is shown.

Sex-Linked Traits (1 of 7)

- In humans, as well as in many other animals and some plants, the sex of the individual is determined by sex chromosomes—one pair of non-homologous chromosomes
- 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

Sex-Linked Traits (2 of 7)

- 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 fewer genes
- When a gene being examined is present on the X, but not the Y chromosome, it is called **X-linked**

Sex-Linked Traits (3 of 7)

- Eye color in Drosophila, the common fruit fly, was the first X-linked trait 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 is dominant to white eye color (X^w) (Figure 8.15)

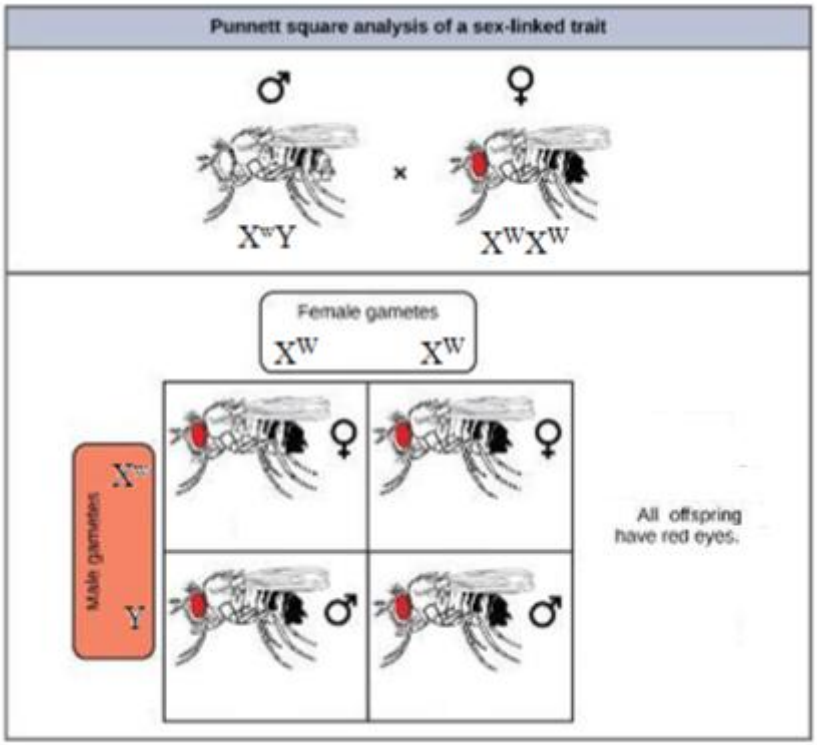
FIGURE 8.15

In *Drosophila*, the gene for eye color is located on the X chromosome. Red eye color is wild-type and is dominant to white eye color.

Sex-Linked Traits (4 of 7)

- 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 generation
- With respect to Drosophila eye color, when the P male expresses the white-eye phenotype and the female is homozygously red-eyed, all members of the F_1 generation exhibit red eyes (Figure 8.16)

FIGURE 8.16-A

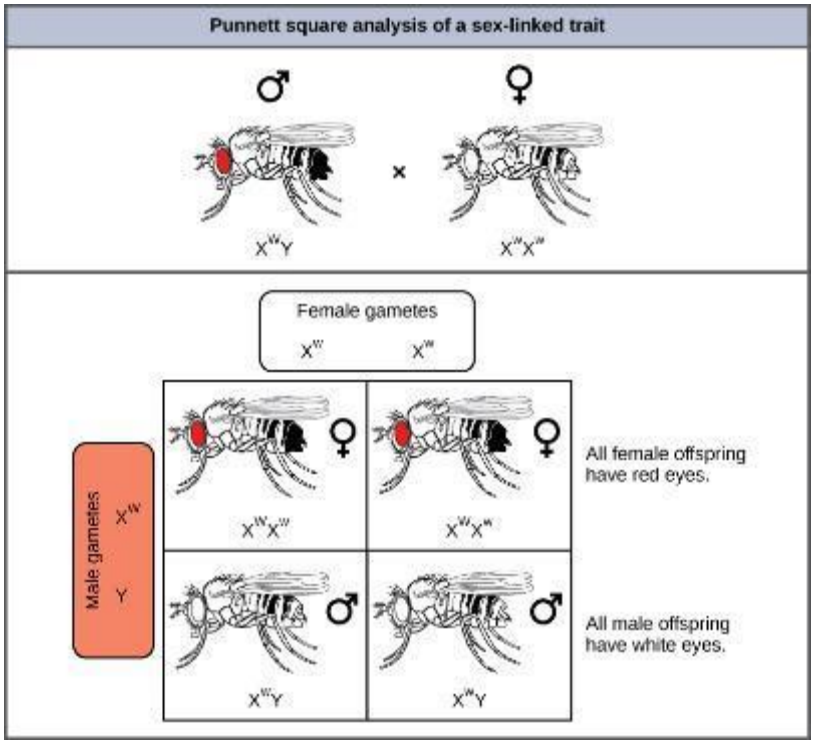


as is the case for this cross involving red and white eye color in *Drosophila*. In the diagram, *w* is the white-eye mutant allele and *W* is the wild-type, red-eye allele. The female is homozygous for the wild-type, red eyes; the male is hemizygous for the recessive white eyes. All offspring will inherit the dominant red eye allele from the female and will therefore exhibit red eyes. (original illustration modified to show the example cross described)

Sex-Linked Traits (5 of 7)

- 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^WX^w) and only white-eyed males (X^wY)

FIGURE 8.16-B



Crosses involving sex-linked traits often give rise to different phenotypes for the different sexes of offspring, as is the case for this cross involving red and white eye color in *Drosophila*. In the diagram, *w* is the white-eye mutant allele and *W* is the wild-type, red-eye allele.

Sex-Linked Traits (6 of 7)

- In humans, the alleles for certain conditions (some 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, X-linked traits appear more frequently in males than females

Sex-Linked Traits (7 of 7)

- In some groups of organisms with sex chromosomes, the sex with the non-homologous sex chromosomes is the female rather than the male (for example, birds)
- In this case, sex-linked traits will be more likely to appear in the female, because they are **hemizygous**

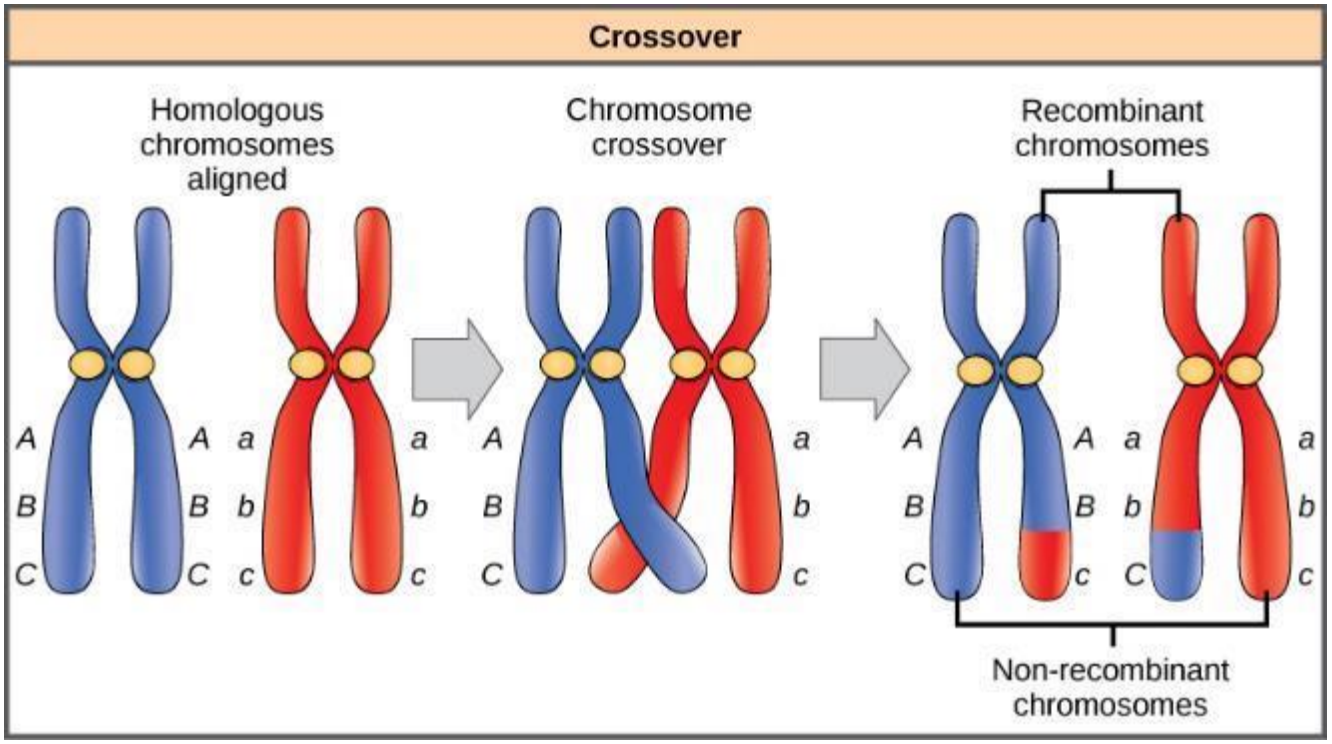
Linked Genes Violate the Law of Independent Assortment (1 of 2)

- Genes that are located on separate, nonhomologous 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

Linked Genes Violate the Law of Independent Assortment (2 of 2)

- 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
- Recall that during interphase and prophase I of meiosis, homologous chromosomes first replicate and then synapse, and exchange linear segments of genetic material (Figure 8.17)
- This process is called **recombination**, or **crossover**, and it is a common genetic process

FIGURE 8.17



The process of crossover, or recombination, occurs when two homologous chromosomes align and exchange a segment of genetic material.

Epistasis (1 of 2)

- Genes may also oppose each other, with one gene suppressing 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** - a gene pathway in which expression of one gene is dependent on the function of a gene that precedes or follows it in the pathway

Epistasis (2 of 2)

- 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, when present as the recessive homozygote (cc), negates any expression of pigment from the A gene and results in an albino mouse (Figure 8.18)
- Therefore, the genotypes AAcc, Aacc, and aacc all produce the same albino phenotype
- The C gene is epistatic to the A gene

FIGURE 8.18

In this example of epistasis, one gene (*C*) masks the expression of another (*A*) for coat color. When the *C* allele is present, coat color is expressed; when it is absent (*cc*), no coat color is expressed. Coat color depends on the *A* gene, which shows dominance, with the recessive homozygote showing a different phenotype than the heterozygote or dominant homozygote.