

Genetics

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OVERVIEW

In this chapter, the topic of genetics is introduced with the story of Barbara McClintock's discovery of transposable elements ("jumping genes") in corn. The chapter progresses from genetics at the molecular level to the cellular level (cytogenetics) to the organismal level (Mendelian genetics and related topics) and then finishes with population genetics (the Hardy-Weinberg law).

Some Learning Goals

1. Identify components of a DNA molecule and know how they are arranged in the molecule.
2. Describe the functions of DNA.
3. Describe how a DNA molecule replicates.
4. Know the function of transcription and outline its steps.
5. Know the function of translation and outline its steps.
6. Distinguish between somatic and germ-line mutations.
7. Describe the significance of translocations and inversions.
8. Distinguish between aneuploids and polyploids.
9. Understand the significance of Mendel's experiments with peas.
10. Give the ratios of the offspring in the first two generations from a monohybrid and a dihybrid cross. Describe the genotypes involved.
11. Distinguish between genotype and phenotype; heterozygous and homozygous.
12. Be able to solve simple genetics problems involving dominance and incomplete dominance.
13. Show how genes may interact with each other to influence phenotype.
14. Explain how genotype influences phenotype.
15. Describe features of a quantitative trait.
16. Describe where extranuclear DNA is located and how it differs from nuclear DNA.
17. Explain how linkage can produce ratios that deviate from Mendelian ratios.
18. Describe the Hardy-Weinberg law.

very so often, someone comes along who is able to push beyond our limits of understanding in science and change the way we view the natural world. Barbara McClintock was one of the most significant scientists in 20th-century biology because she caused a major shift in the way we view gene organization. She was a geneticist who carried out most of her work at the Cold Spring Harbor laboratory in New York in the 1940s and 1950s. While looking at corn seedlings for one of her genetic studies, she noticed that streaks of color often appeared where they did not seem to belong. There were patches of yellow in green leaves or patches of green in white leaves. She also noticed spots of color in corn kernels that should have been colorless, based on her genetic studies (Fig. 13.1). Instead of ignoring these plants as genetic oddities, McClintock undertook years of research to identify the cause of these patterns.

In the early 1950s, McClintock published work introducing the concept of **transposition**, or the movement of a piece of a chromosome to another chromosomal location. A **transposable genetic element** (or *jumping gene* in popular literature) is a

gene or a small DNA fragment that can move to a new location on the same chromosome or even to another chromosome. If it moves into an existing gene, then the function of that gene is disrupted. A transposable element can also move out of a gene and restore its original function. So, the patches of yellow in green leaves were regions where transposable elements were inserted into a gene involved in chlorophyll synthesis. Green areas on white leaves resulted when a transposable element moved out of a chlorophyll synthesis gene and allowed it to function again.

McClintock was a highly respected figure in the field of genetics and was able to describe in detail the actions of several transposable elements in corn. Accordingly, even though the concept of movable genes was difficult to fit into the framework of conventional genetics, McClintock's ideas were generally accepted by her peers. Before long, transposable elements were found in other organisms as well.

The discovery of the structure of DNA by James Watson and Francis Crick in 1953 ushered in the new field of **molecular genetics**. At that time, geneticists thought of McClintock's work as a remnant of a previous era and not relevant to modern genetics. However, studies of DNA and gene structure transformed the concept of a gene. Genes were no longer abstract entities but were actual sequences of nucleic acids that could be isolated and characterized. The concept of mutation within a gene became believable. In addition, numerous studies found transposable elements in other organisms.

McClintock's work was formally recognized in 1983 when she was awarded a Nobel Prize. We now know that transposable elements are widespread and have probably been in organisms for a long time. We do not know, however, if they represent genetic parasites causing mutations as they move about an organism's genetic material or if they perform valuable functions. One theory is that they allow nature to tinker with chromosomes much as human genetic engineers do. It may be evolutionarily beneficial to copy, move, and rearrange pieces of chromosomes, creating new and occasionally better combinations of genes within an organism.

MOLECULAR GENETICS

In Chapter 3, we mentioned that chromosomes in the nucleus of each cell carry genetic information that is passed on from one generation to the next. Chromosomes are composed of two types of large molecules—DNA and protein. Until the early 1950s, most scientists believed that proteins carried the genetic information because they thought that the 20 amino acids in proteins could provide more diversity than the four bases in DNA. We now know, of course, that DNA is responsible for carrying genetic information.

Structure of DNA

Chromosomes are composed of chromatin, which is DNA and associated proteins. A DNA molecule is a simple, elegant chain of building blocks called **nucleotides**. Each nucleotide consists of three parts: (1) a nitrogen-containing compound, called a **base**; (2) a 5-carbon sugar, named **deoxyribose**; and (3) a phosphate group. Both the nitrogenous base and the phosphate group are bonded to the sugar (Fig. 13.2). Four types of nucleotides occur in DNA. Each has a unique nitrogenous base, but all have the same phosphate group and deoxyribose sugar. Two of the bases—**adenine** (A) and **guanine** (G)—are called **purines**. Each has a molecular structure that resembles two linked rings. The other two bases—**cytosine** (C) and **thymine** (T)—are called **pyrimidines**. They each have a molecular structure consisting of a single ring.

The nucleotides in a DNA molecule are bonded to each other in such a way that they form a chain that looks like a ladder twisted into a spiral, or a helix. Each of the two sides of the "ladder" is composed of alternating sugar and phosphate groups (Fig. 13.3). Each sugar is also bound to a base. Hydrogen bonds hold each base on one side of the helix to another base on the other side, making the "rungs" of the ladder. Although the structure of a DNA molecule is very simple—alternating sugar and phosphate groups on the outside and pairs of bases in the middle—it provides all the genetic diversity on the planet. The variation comes in the form of base pairs. A single chromosome contains a DNA molecule that is often hundreds of millions of base pairs in length. The sequence of base pairs on a DNA molecule determines whether, for example, that DNA will direct the synthesis of a chlorophyll molecule or a hemoglobin molecule.

As mentioned earlier, in the early 1950s, James Watson and Francis Crick began to unravel the mystery of DNA structure. Much of their work involved piecing together existing information to develop a model. They knew Linus Pauling had postulated that the structure of DNA might be similar to the structure of protein. Pauling had shown part of the structure of some proteins to be helical and maintained by hydrogen bonds between the amino acids. Watson and Crick also knew from X-ray work by Rosalind Franklin and Maurice Wilkins that DNA molecules are composed of regularly repeating units in a helical arrangement. Finally, studies by Erwin Chargaff indicated that the amount of pyrimidine nucleotides (cytosine + thymine) in DNA equals the amount of purine nucleotides (adenine + guanine). This suggested to Watson and Crick that purines pair with pyrimidines.

When they had put together all the facts, Watson and Crick concluded that a DNA molecule consists of a double helix whose two strands appear to be wrapped around an invisible pole—one strand spiraling in one direction and the other strand spiraling at the same angle in the opposite direction. They also concluded that nucleotides are linked in ladder-like fashion between the two strands. Furthermore, in order for the nucleotides to fit precisely in the DNA molecule, two linked purines or

two linked pyrimidines would either be too wide or too narrow. However, a purine and a pyrimidine linked together would fit perfectly. Accordingly, they concluded that the ladder rungs had to consist exclusively of purine-pyrimidine pairs. In order for hydrogen bonds to form correctly between base pairs, guanine must pair with cytosine, and thymine must pair with adenine. Watson and Crick's DNA molecule is now universally accepted as an authentic representation.

DNA Functions

The DNA in a cell must perform four major tasks. It must (1) store genetic information; (2) copy that information for future generations of cells; (3) express that information; and (4) occasionally change its message, or **mutate**.

Storage of Genetic Information

Most DNA in plants resides in the nucleus. The nucleus is a compartment in which proteins that interact with DNA are concentrated. This compartmentalization is important because it creates an environment in which biochemical reactions are optimized by high concentrations of substrates and the enzymes that act on them.

The genetic information in a DNA molecule resides in the sequence of nucleotides along the rungs of the ladder. The sequence GAATCC, for example, codes for a different set of amino acids than CTTAGA. Early scientists were fooled by the simplicity of a DNA molecule and did not believe it could provide information for the survival of all organisms on earth. However, the DNA in a typical plant cell contains millions or billions of base pairs. A barley plant with 5 billion base pairs of DNA in its nucleus could produce $4^{5,000,000,000}$ different DNA sequence combinations! This provides more than enough genetic variability for the plant.

At this point, we need to introduce some important concepts, beginning with that of the gene. We are all familiar with the idea that genes control physical traits, such as eye color in people and height in plants. How do they do that? From a molecular perspective, a **gene** is a segment of DNA (several thousand base pairs long) that directs the synthesis of a protein. That protein is then used by the cell as structural or storage material, or it influences the activities of the cell by acting as an **enzyme**. As you recall from Chapter 2, enzymes are organic catalysts. Any activity in the cell, from photosynthesis to cell-wall construction, absolutely depends on enzymes to facilitate chemical reactions. Therefore, all activities of the cell are controlled by enzymes, which require genes for their synthesis. Each kind of organism has many different genes in its **genome**, which is a term for *the sum total of DNA in an organism's chromosomes*. A relatively simple organism such as a bacterium typically has several thousand genes in its genome. On the other hand, more complex eukaryotic organisms usually have between 10,000 and 50,000 genes per genome. The differences between organisms lie in the differences in the composition of their genomes. Organisms that are very dissimilar—for example, bacteria and higher plants—will have many differences in their DNA sequences. The genomes of similar organisms, such as domestic plants and their wild relatives, are very similar, yet different enough to account for dramatic differences in appearance.

Replication (Duplication) of Information

DNA must duplicate itself precisely in order to pass along its information from generation to generation. DNA replication occurs during the S phase of the cell cycle (see Chapter 3). Differences between the DNA of one organism and that of another lie in the sequence of the four possible types of ladder rungs and their total number. If the four nucleotides are synthesized in living cells, then what tells the cell exactly how to put them together to form DNA molecules with proper nucleotide sequences?

Watson and Crick predicted that if the two strands of DNA are “unzipped” by an enzyme breaking the hydrogen bonds between the nucleotides down the middle of the ladder, then each separated chain provides all the information needed to put together a new ladder (Fig. 13.4). In other words, since guanine can pair only with cytosine, and vice versa, a guanine nucleotide synthesized by the cell can lock on, or bond, only to a thymine nucleotide. If the two strands of a DNA molecule “unzip,” then individual nucleotide “building blocks” can line up next to each of the single chains in precise sequence and be bonded together to form a new chain; each single chain acts as a mold or template for the creation of a new double-stranded DNA molecule. When replication is complete, two double helices have been created from a single one. Each new DNA molecule consists of one strand from the original molecule and another built using that parental strand as a template. This is called **semiconservative replication**.

Replication is a remarkably rapid and precise process. During DNA replication, nucleotides are added by the enzyme **DNA polymerase** at an amazing rate of approximately 833 per second in prokaryotes and 33 per second in eukaryotes. However, even at this rate, a plant chromosome that is, for example, 200 million base pairs long, would take *70 days* to replicate. Because a cell must replicate its DNA before it undergoes mitosis, this replication rate is far too slow for normal plant growth. How is this problem resolved for the cells? In two ways: (1) replication proceeds simultaneously in both directions on the chromosome, cutting the replication time in half; and (2) replication begins at several to many points on a chromosome nearly simultaneously.

The nucleus contains enzymes that are capable of “proofreading” during replication. They detect and correct mismatched base pairs, such as guanine paired with adenine or thymine paired with cytosine. Consequently, about only one in every million

nucleotides added during replication is incorrectly paired. However, each nucleus of a corn plant contains 4 billion base pairs of DNA. Therefore, about 4,000 mutations (changes in DNA sequence) occur with each cell division.

Expression of Information

Since the nature and structure of DNA is of fundamental importance to its ability to control the form and function of organisms, there appears to be a paradox. If every cell in an organism contains the same genetic information, then why do cells within a plant differ from each other? That is, why is a bark cell so different from a leaf cell?

The answers to these questions stem from the fact that each cell in a multicellular plant contains the same DNA information, but different subsets of this master plan are read in each cell type. For example, a plant has epidermal cells that secrete a protective waxy cuticle, and it also has mesophyll cells that are highly specialized for photosynthesis. The epidermal cells express genes (read DNA sequences) that encode enzymes necessary to build a waxy cuticle. Mesophyll cells, on the other hand, will not express cuticle genes but do express a set of genes that encode enzymes for chlorophyll synthesis. One of the most active areas of biology research today is directed toward understanding how organisms control gene expression.

A cell's environment can influence the set of genes expressed by that cell. Many cellular changes occur as a result of the expression of new sets of genes induced by hormones (see Chapter 11). We know, for example, that an epidermal cell expresses genes for cell elongation if the hormone auxin is present; when mesophyll cells are moved from dark into light, many dramatic changes in gene expression occur as genes for the photosynthetic machinery are turned on.

The expression of genetic information requires two processes. First, the information coded by DNA is *transcribed*. During **transcription**, a copy of the gene message is made using RNA (ribonucleic acid) building blocks. RNA is similar to DNA with a few important differences. First, RNA nucleotides contain **ribose** sugars instead of **deoxyribose** sugars. Ribose contains one more oxygen atom than deoxyribose. Second, thymine is replaced by **uracil**, another nitrogenous base. Finally, RNA molecules are typically single-stranded. After transcription has been completed in the nucleus, the RNA is modified and then travels to the cytoplasm, where *translation* occurs. During **translation**, the RNA message provides the information necessary to construct proteins. It might help to remember the following when you are trying to distinguish between transcription and translation. During transcription, a nucleic acid, RNA, is made using another nucleic acid, DNA, as a template. This is analogous to a stenographer transcribing shorthand to longhand. During translation, a protein is made using a nucleic acid (RNA) as a template. This is analogous to a court reporter who transcribes notes based on combinations of letters and words into the text record of the court's proceedings.

Transcription Three different types of RNA are made by transcription. Chromosomes contain genes for the production of *messenger RNA (mRNA)*, *transfer RNA (tRNA)*, and *ribosomal RNA (rRNA)*. Messenger RNA will be translated to produce proteins, while tRNA and rRNA will be used as part of the machinery for translation.

The mechanism of RNA synthesis is relatively simple. It involves the addition of RNA nucleotides to single-stranded DNA molecules, using complementary base pairing, similar to DNA replication. **RNA polymerase** is responsible for assembling the RNA strand. However, while all regions of the genome are replicated, only a very small portion of the genome is transcribed. In most eukaryotes, less than 10% of the DNA in the genome contains genes. The remainder is **noncoding DNA**. The transcription machinery must therefore search through millions of base pairs to identify small regions that contain genes. How does it do that? We have unraveled some of this mystery, but have much to learn. At the beginning of every gene, a DNA sequence called a **promoter region** acts as a flag to signal the transcription machinery scanning the DNA that a gene is ahead. This causes the transcription enzymes to associate with one strand of the DNA molecule and begin to make a copy of the gene. The complementary DNA strand is not used for transcription. A *terminator* DNA sequence at the end of each gene signals the transcription enzymes to fall off the DNA molecule. At this point, a single-stranded RNA molecule, called a **transcript**, has been made using one strand of a gene as a template. The transcript will undergo several modifications and move to the cytoplasm before translation begins.

As we mentioned before, not every gene is expressed in every cell. In addition to identifying genes, the transcription machinery must determine which genes to transcribe, or express. This is a very complex process and involves interactions among genes and signals from the environment. In addition, cells must control the amount of each type of protein they produce. Until recently, scientists thought that the large amount of nonprotein-coding DNA in the genome represents evolutionary "junk." That is, this DNA is what is left over from failed experimentation by organisms during evolution. On the contrary, it now appears that this nonprotein-coding DNA is fundamental to the control of gene expression. Because nonprotein-coding DNA is not translated, scientists never see protein products. However, RNA transcription products from these sequences appear to carry out an amazing array of activities that control the amount of protein cells produce from conventional "coding" genes.

Nonprotein-coding genes are also responsible for making some of the machinery needed for translation. Chromosomes contain genes for building tRNA. When transcribed, these genes produce molecules of tRNA approximately 80 nucleotides long. The single-stranded RNA produced by transcription spontaneously forms base pairs in some regions, creating a three-dimensional molecule of a characteristic shape. Transfer RNA will act as the "translator" during translation. One end of the tRNA molecule binds to the mRNA, while the other end carries a specific amino acid. There is at least one form of tRNA for each of the 20 amino acids. Each form of tRNA has a specific *anticodon* loop. The **anticodon** is a sequence of three nucleotides that recognizes a codon on mRNA and base pairs with it. An amino acid binds to the other end of the tRNA with the aid

of an enzyme that specifically identifies both a single tRNA strand and its proper amino acid.

Genes for rRNA are also transcribed in the nucleus. This RNA is used to construct **ribosomes**, which act as workbenches during protein synthesis. A ribosome is composed of a large and a small subunit, each containing a specific combination of rRNA molecules and proteins. The rRNA molecules create the three-dimensional structure of a ribosome and help to orient the proteins, most of which appear to assist with the assembly of new proteins during translation. It is believed that mRNA slides between the large and small subunits during translation. All ribosomes in a cell are identical and capable of reading any mRNA.

Translation RNA transcripts that can be decoded into proteins are called *mRNAs*. It seems puzzling that a nucleic acid with only four possible nucleotides (adenine, uracil, cytosine, and guanine) can be used to make a protein polymer that may have 20 different amino acids. One nucleotide is incapable of encoding one amino acid. Even two nucleotides together could be combined in a maximum of only 16 different ways. It turns out that the genetic code is based on trios of nucleotides we earlier referred to as *codons*. Like words and sentences, codons and genes can be read from one direction only, thereby preventing the possibility of expressing the wrong amino acid. There are 64 possible ways to combine nucleotides, taking three at a time. This is more than enough to encode all 20 amino acids. The order of nucleotides on an mRNA molecule (determined by the sequence of nucleotides in a gene during transcription) determines the sequence of amino acids added during translation. For example, the codon AUG (adenine-uracil-guanine) on an mRNA encodes the amino acid methionine, whereas the codon AGU on an mRNA encodes the amino acid serine. The genetic code (Fig. 13.5) shows the matching of each three-nucleotide codon with a specific amino acid. With few exceptions, the genetic code is universal. In other words, the same code is used in bacteria, plants, and animals.

As mentioned before, tRNAs act as translators, or decoders, during translation. The anticodon part of the tRNA binds to the mRNA codon by base pairing with it, while the other end of the tRNA molecule has, according to the genetic code, the correct amino acid bound to it. In a sense, then, we can think of tRNAs as the dictionary used to decode an mRNA during translation.

The start of translation is usually signaled by a ribosome in the cytoplasm binding to the mRNA and scanning it for the first AUG codon. This sets the reading frame because each set of three nucleotides is translated in succession after the first AUG. A start tRNA with the amino acid methionine and an anticodon that base-pairs with the AUG on the mRNA joins the mRNA and the ribosome, forming a three-part complex (Fig. 13.6). Another tRNA that base-pairs to the next codon and has its correct amino acid attached inserts in the complex next to the start tRNA. An enzyme in the ribosome then links the two adjacent amino acids together to begin to form a protein. As translation continues, new tRNAs base-pair to each mRNA codon in order, and new amino acid linkages are made. For example, an mRNA with the sequence AUG-AAG-UGU will be translated into a protein with the sequence methionine-lysine-cysteine until a stop codon (UGA, UAA, or UAG) is reached. At that point, a newly made protein is released from the complex.

To summarize, the flow of information in a cell is from the DNA genome in the nucleus, which is transcribed into mRNA, to the cytoplasm, where the mRNA is translated into protein. Because these processes occur in all living organisms, it has come to be known as the *Central Dogma of Molecular Genetics* (Fig. 13.7). Truly, a cell's identity is determined by the genes it expresses, which, in turn, produce the myriad of proteins required for its survival and function.

Mutation

When my six-year-old son recently told a friend that his Mom was a scientist, the boy asked, "Can she make mutants?" While the term *mutation* may conjure up images of *Teenage Mutant Ninja Turtles*, most mutations are not so dramatic. In fact, as mentioned earlier, **mutations**, or changes in DNA sequence, happen every time DNA replicates. In addition to DNA replication errors and other spontaneous types of changes, agents called **mutagens** can alter DNA sequences. Ultraviolet light, ionizing radiation, and certain chemicals act as mutagens. DNA repair enzymes can often find and correct damaged DNA. If left uncorrected, though, a permanent change in DNA sequence, or mutation, occurs. Most mutations are *silent*, with no visible consequence of the DNA alteration. Remember that most DNA in a cell is noncoding. Even altered coding sequences may produce functional proteins.

Mutations are either *somatic* or *germ-line*. A **somatic mutation** occurs in a body cell and will exist in all cells produced by mitosis of the mutant cell. A somatic mutation may show up as a *sport*, or a branch that looks different from the others on a tree. These types of mutations are sometimes sources of new types of horticultural plants. Navel oranges and Red Delicious apples originated from sports and have been clonally propagated by grafting (see Chapter 14).

A **germ-line** mutation occurs in tissue that will produce gametes, or sex cells. Unlike somatic mutations, germ-line mutations will be passed on to future generations through seeds. A germ-line mutation is generally not apparent until it is passed on to offspring, which will carry the altered DNA in all cells. This mutation has now become a permanent feature of that plant's lineage. Germ-line mutations are important for the genetic improvement of horticultural and agronomic plants. They are responsible for variability in all traits, including flower color and fragrance, fruit taste and texture, grain yield and quality, and disease and stress tolerance.

While we tend to think of mutations as "bad," they are an essential feature of DNA. All the genetic variability in nature has arisen as a result of mutation. The accumulation of mutations is the mechanism that drives evolution of species (see

Chapters 15 and 16). That variability was essential for the first plants, which lived in the ocean, to become established on land; it continues to be necessary for plants to survive and compete in an ever-changing environment.

CYTOGENETICS

During the middle of the 20th century, corn cytogenetics dominated the biological sciences, due in large part to the efforts of Barbara McClintock and other corn geneticists, including Roland Emerson, George Beadle, Charles Burnam, and Marcus Rhoades. **Cytogenetics** is the study of chromosome behavior and structure from a genetic point of view. This includes chromosome movement during meiosis and mitosis, chromosome pairing during meiosis, and chromosomal variation due to changes in chromosome structure and number. Chapters 3 and 12 discuss chromosome behavior during mitosis and meiosis, respectively. Therefore, this section will emphasize changes in chromosome structure and number. These changes occur at low frequency, but since their effects are often large, they can have a dramatic influence on the evolutionary success of populations or species.

Changes in Chromosome Structure

Occasionally, a chromosome will break into two or more pieces. Enzymes may repair the damage, leaving the chromosome unaltered. However, sometimes a broken chromosome may become reorganized before the pieces are attached together again. The most significant events from an evolutionary perspective include *inversions* and *translocations*.

An **inversion** results when a piece of a chromosome is broken and reinserted in the opposite orientation. For example, if the chromosome contains genes ABCDE in that order, then an inversion might create a chromosome with the genes in this order: **ABDCE**. The inverted chromosome can pair up with the normal homologous one at meiosis, but crossing-over within the inverted region will create gametes that do not survive because they contain some duplicated chromosomal regions and are missing other segments of chromosomes. Plants carrying an inverted chromosome therefore are less fertile than normal ones. The only gametes that are passed on from one generation to another are those in which crossovers occurred only outside the inverted region. Therefore, allele combinations within an inverted region are not rearranged by meiosis and are inherited together as blocks.

A **translocation** results when a piece of a chromosome breaks off and becomes attached to another one. A plant carrying a translocation, like one carrying an inversion, will not be as fertile as a normal one. This is because some gametes from translocation plants carry unusual combinations of chromosomes.

Translocations and inversions can be important factors in **speciation** (the formation of new species) during evolution. Plants carrying chromosomes from these events may become reproductively isolated from normal plants and will eventually develop into new species. Crosses between the new species and the old one will produce offspring with low fertility and, therefore, a poor reproductive capacity. These types of events have been important in the evolution of many plants. For example, cultivated rye (*Secale cereale*) and four wild species (*S. vavilovii*, *S. africanum*, *S. montanum*, *S. silvestre*) each contain 14 chromosomes. However, translocations have resulted in the wild species being reproductively isolated from the cultivated species. Genetic exchange among these species is limited by translocations.

Changes in Chromosome Number

Compared to animals, plants are remarkably tolerant of changes in chromosome number. Mistakes made during the pairing and separation of chromosomes during meiosis can result in gametes carrying extra or missing chromosomes. If one of these gametes is involved in fertilization, then the resulting zygote will be *aneuploid*. An **aneuploid** plant carries an extra one or more chromosomes or, less commonly, is missing one or more chromosomes. For example, the cells in a carrot plant typically contain nine pairs of homologous chromosomes, for a total of 18 chromosomes. An aneuploid carrot plant would have 19 chromosomes if it had an extra copy of one chromosome or 17 chromosomes if it was missing one of a pair. As you would expect, aneuploid plants generally differ in appearance from their normal counterparts due to the effects of extra or missing doses of many genes. Geneticists use aneuploids to determine the chromosomal locations of genes.

Occasionally, meiosis may completely fail to halve the chromosome number, producing gametes with the somatic (body cell) chromosome number. These are called $2n$ gametes. If these gametes are involved in fertilization, then the resulting offspring are *polyploid*. A **polyploid** plant has at least one complete extra set of chromosomes. Using the carrot example, a polyploid carrot could have 27 chromosomes (three sets of nine chromosomes). Polyploid plants are often larger and higher-yielding than their diploid counterparts. For this reason, many of our cultivated plants are polyploid. These include potato, cotton, peanut, wheat, oats, strawberry, and sugar cane (Fig. 13.8). The larger and longer-lasting flowers of polyploids make them attractive as ornamental plants; marigold, snapdragon, lily, and hyacinth are among the numerous examples of ornamental polyploids. In addition, triploid plants (polyploids containing three copies of each chromosome) are sterile and do not produce seeds. Seedless fruits, such as bananas and watermelons, are triploid. In Chapter 15, polyploidy is discussed as an important mechanism for the production of interspecific hybrids in nature.

MENDELIAN GENETICS

Genetics as a science originated about a generation before its significance became appreciated in the scientific community as a whole. An Austrian monk, Gregor Mendel (Fig. 13.9) taught between 1853 and 1868 in what became the Czechoslovakian city of Brno. In the monastery there, he carried out a wide range of studies in physics, mathematics, and natural history. He also became an authority on bees and meteorology and kept notes on experiments involving two dozen different kinds of plants. Today, he is best known for the studies he conducted with peas.

Mendel published the results of his studies on pea plants in a biological journal in 1866 and sent copies of his paper to leading European and American libraries. He also sent copies to at least two eminent botanists of the time. His work, nevertheless, was completely ignored or overlooked until 1900, when three botanists (Eric von Tschermak of Austria, Carl Correns of Germany, and Hugo de Vries of Holland), working independently in their own countries, reached the same conclusions as Mendel. Each, as a result of library research, came across Mendel's original paper.

Mendel's Studies

Peas, unlike many flowering plants, are *self-fertile*. In other words, a pollen grain of a pea flower can germinate on its own stigma, allowing a sperm cell to fertilize an egg and the ovule to develop into a viable pea seed. Although Mendel did not know about mitosis and meiosis, he had noticed that if a pea plant grew to a certain height, its offspring, if grown under the same conditions, would always reach approximately the same height, generation after generation. He wondered what would happen if he crossed a tall plant with a short one. Would the offspring be intermediate in height?

To make such a cross, Mendel needed to prevent self-pollination. He did so by reaching into a flower of one plant and removing the stamens before the pollen had matured. Then, he took pollen from another plant and applied it by hand to the stigma of the first flower. He also covered the experimental flowers with small bags to prevent insects from bringing pollen from other flowers after the cross had been made.

When Mendel made such crosses, the results were astonishing. All of the offspring were tall. There were no short or intermediate plants. He found, however, that if he allowed the offspring plants to pollinate themselves, they produced offspring in a ratio of approximately three tall plants to one short plant (Fig. 13.10).

Mendel then tried crosses between peas with smooth seeds and those with wrinkled seeds. He also crossed green-seeded plants with yellow-seeded ones and several others with pairs of contrasting characteristics.

Mendel referred to the original plants involved in making the crosses as the **parental generation (P)** and their offspring were the **first filial (F₁)** generation. Filial means "of or relating to a son or daughter." The offspring of F₁ plants were called the **second filial (F₂)** generation.

There are several reasons why Mendel was successful in developing a model for inheritance when countless others before him failed. He performed carefully planned and executed crosses between pure-breeding parents. Each parent was genetically stable for the trait under study—a green-seeded parent produced only green-seeded offspring. Mendel also counted the number of offspring in each cross, something none of his predecessors had done. His use of his math background to apply statistical analyses to his data was unique, allowing him to develop and test inheritance models. Finally, he chose traits under simple genetic control. His predecessors usually studied traits such as weight, which is controlled by many genes and strongly influenced by environment. A summary of some of Mendel's data is shown in Table 13.1.

As Mendel began to collect data in his experiments, he realized that there must be elements in the pea plant responsible for controlling traits such as seed color and stem height. He referred to this unknown agent as a *factor*. He also deduced that each plant must have two such factors for each characteristic, since even though all the offspring of an F₁ generation appeared identical, the F₂ generation revealed some plants with one characteristic and others with the contrasting form.

These discoveries and deductions came to be known as the **law of unit characters**. Stated simply, this law says that *factors, which always occur in pairs, control the inheritance of various characteristics*. Today, we know that the paired factors are **alleles** of genes. Alleles are alternative forms of a gene. For example, the seed color gene has an allele for yellow color and an allele for green color. Genes are always at the same position (**locus**) on homologous chromosomes (see Chapter 12). We also now know that there are typically thousands of genes on every chromosome.

From his data and analyses, Mendel also deduced that one allele may conceal the expression of the other. For example, in the F₁ generation following a cross between green-podded and yellow-podded parents, all the plants had green pods. However, both parents obviously had to contribute something to the cross, since some of the F₂ plants had green pods while others had yellow pods. This deduction led to another principle, known as the **law of dominance**. This principle says that *for any given pair of alleles, one may mask the expression of the other*. The expressed allele is referred to as **dominant**, and the one that is not expressed (masked) is **recessive**. In the green-podded by yellow--podded cross, the green-pod allele is dominant, and the yellow-pod allele is recessive.

Mendel's crosses also made it clear that a plant's having green pods did not indicate whether or not both members of the pair of alleles controlling pod color were present, because the dominant one could mask the expression of the recessive one. To distinguish between the physical appearance of an organism and the genetic information it contains, we use the terms *phenotype* and *genotype*. **Phenotype** refers to the physical appearance of the organism, while **genotype** refers to the genetic

information responsible for contributing to that phenotype. We customarily use words to describe phenotypes and letters to designate genotypes.

In Mendel's crosses, for example, seven pairs of phenotypes are shown as parents; these phenotypes include yellow seeds and green seeds, smooth seeds and wrinkled seeds, green pods and yellow pods, and so on. In designating the genotypes of each of these plants, a capital letter is used to indicate the dominant member of a pair of genes; *the same letter* in lowercase is used to indicate the corresponding recessive allele. In the green-podded \times yellow-podded pea cross, for example, green is dominant over yellow. If a plant has a green phenotype, then its genotype could be either GG or Gg . The genotype of yellow-podded plants would always be gg . The plant is said to be **homozygous** if both alleles of a pair are identical (e.g., GG or gg) and **heterozygous** if the pair is composed of contrasting alleles (e.g., Gg). Mendel's true-breeding parents were homozygous for the traits he studied.

It is amazing that, nearly 150 years ago, Mendel was able to develop hereditary laws that are widely recognized today. Remember that he did his work before anyone knew about chromosomes, meiosis, or genes.

The Monohybrid Cross

Mendel was successful largely because he carried out well-planned crosses with true-breeding parents. Geneticists today perform similar experiments when trying to understand the genetic basis of uncharacterized traits. A common strategy is to generate a **monohybrid cross**. In this scenario, a single trait is studied. A cross is made between two true-breeding parents differing for that trait, producing an F_1 generation. Then, these F_1 plants are intercrossed to produce an F_2 generation. As an example, we will examine a cross between a homozygous green-podded pea plant and a homozygous yellow-podded one (Fig. 13.11). The genotype of the homozygous dominant parent (green-podded) is GG ; the genotype of the homozygous recessive parent (yellow-podded) is gg . You will recall from Chapter 12 that gametes have only one member of each pair of homologous chromosomes. The gametes of the green-podded parent (GG), therefore, will be G ; the gametes of the yellow-podded parent (gg) will be g . No matter which egg unites with which sperm of the other parent, all the zygotes of this cross will be heterozygous, with the genotype Gg . This means that all individuals in the F_1 generation will have the green-podded phenotype, and all will be heterozygous.

The next step is the actual monohybrid cross. Members of the F_1 generation are crossed with each other (or self-pollinated) to produce an F_2 generation. All of the F_1 plants have the same phenotype (green-podded) and genotype (Gg). Each F_1 plant produces two kinds of gametes (G and g) in roughly equal numbers. When the gametes are produced, half will be G , and half will be g . A G egg may unite with a G sperm, producing a GG zygote. Alternatively, purely at random, the G egg may unite with a g sperm producing a Gg zygote. The same type of random combinations occur with g eggs so that either Gg or gg zygotes are produced. When all the offspring of a large number of such crosses are counted, a genotypic ratio of 1 GG : 2 Gg : 1 gg is produced. The phenotypic ratio will be approximately three green-podded plants (GG and Gg) to one yellow-podded plant (gg). These four equally possible F_2 offspring are shown at the bottom of Figure 13.11.

The Dihybrid Cross

So far, our examples have involved single genes. However, it is often desirable to study a pair of genes using a **dihybrid cross**. This is especially valuable in gene mapping studies. That topic is discussed later in this chapter. The **law of independent assortment** states that *the factors (genes) controlling two or more traits segregate independently of each other*. This is one law that, while true for many traits, often breaks down. That is because the two genes under study may be physically close to each other on a chromosome. If this is the case, then the genes are basically attached together and will follow each other through meiosis. They do not segregate independently. In this situation, the genes are said to be **linked**. In the dihybrid example that follows, however, we will assume that the pair of genes is **unlinked**. That is, they are on different chromosomes or are far apart on a single chromosome.

Let's consider the genes for plant height and pod color. Recall that tallness in peas is dominant and dwarfness is recessive, while green pod is dominant over yellow pod. The homozygous dominant ($GGTT$) parent will have a green-podded, tall phenotype, while the recessive parent ($ggtt$) will be yellow-podded and dwarf. All the gametes from the dominant parent will be GT , and those of the recessive parent will be gt . The F_1 generation will be composed of dihybrid ($GgTt$) plants that are green-podded and tall (Fig. 13.12).

When intercrossed to produce the F_2 generation, these dihybrid members of the F_1 generation will produce four types of gametes in equal proportions. The gamete genotypes are GT , Gt , gT , and gt . Remember that each gamete will carry only one allele of each gene. Since any one of the four kinds of gametes can unite randomly with any of the four kinds of gametes of the other parent, 16 combinations are possible.

In order to avoid confusion when trying to make all possible combinations of gametes, a diagram called a **Punnett square** is used to determine the genotypes of the zygotes. The Punnett square diagram looks somewhat like a checkerboard, with the gametes of one parent across the top and the gametes of the other parent on the left side. To fill in each square on the checkerboard, look at the gamete designation above the square and to the side of it. Multiply the gamete proportions (e.g. $1/4 \times 1/4$) and add together the gamete letters (e.g., $GT + GT$). In Figure 13.11, the first square would therefore be $1/16 GGTT$.

Nine different zygote genotypes are possible in the $16 \square$ Punnett square. They are produced in a ratio of 1 $GGTT$: 2 $GGTt$:

1 *GGtt*: 2 *GgTT*: 4 *GgTt*: 2 *Ggtt*: 1 *ggTT*: 2 *ggTt*: 1 *ggtt*. Four phenotypes are possible in a ratio of 9 green-podded, tall: 3 green-podded, dwarf: 3 yellow-podded, tall: 1 yellow--podded, dwarf. A geneticist observing a 9:3:3:1 ratio from a dihybrid cross can deduce that the genes controlling the two traits are unlinked and are exhibiting dominance. Genetic ratios such as the ones previously outlined are expected when large numbers of individuals are studied. Note in Mendel's data (Table 13.1) that he looked at over 8,000 F₂ offspring following a monohybrid cross involving seed color. With small numbers, chance alone may not produce the expected ratios. Human populations, for example, are divided relatively equally into males and females (a genetically controlled trait), but in smaller groups, such as families, the general population ratio of one male to one female may not be evident.

The Backcross

If a scientific theory is valid, then one should be able to test it experimentally. Mendel himself tested his predictions by means of backcrosses. A **backcross** is a cross between a hybrid and one of its parents. Mendel had found in pea flowers that red color is dominant and that white is recessive. Therefore, all F₁ offspring of a cross between a homozygous red-flowered parent (*RR*) and a homozygous white--flowered parent (*rr*) would be red (*Rr*). The gametes of these F₁ plants would be either *R* or *r*, with each type being produced in equal numbers. If the F₁ hybrid (*Rr*) is crossed to a homozygous recessive plant (*rr*), then half of the offspring would be *Rr*. These plants received the dominant allele from the heterozygous parent and a recessive allele from the homozygous parent. The remaining offspring from the backcross received a recessive allele from the heterozygous parent, so they would be *rr*. Following backcrosses of F₁ hybrids to recessive parents, Mendel observed the expected ratio of 1 red-flowered plant (*Rr*) to 1 white-flowered plant (*rr*). This provided further confirmation for his inheritance theory.

The Testcross

In the F₂ generation of a monohybrid cross, there are two genetically different types of plants that look identical. Homozygous dominant plants look just like heterozygotes. Suppose you are interested in determining which plants are homozygous dominant for the tall allele. How would you identify them? One of the easiest ways is to cross the tall plants with short plants. A cross between a plant with the dominant phenotype and a homozygous recessive plant is called a **testcross**. This cross will determine whether the plant with the dominant phenotype is homozygous or heterozygous. The short plants are homozygous recessive, so you know their genotype. If all the offspring from a cross between a tall plant and a short plant are tall, then the tall plant must be homozygous dominant. If some (approximately 50%) of the offspring are short, then the tall plant must be heterozygous.

Incomplete Dominance

Although the principle of dominance was amply demonstrated by Mendel's crosses, we know now that in many other instances, neither member of a pair of genes completely dominates the other. In other words, some genes exhibit **incomplete dominance** (or absence of dominance), in which a heterozygote is intermediate in phenotype to the two homozygotes. In snapdragons, for example, the F₁ plants of a cross between a red-flowered parent and a white--flowered parent are all pink-flowered. When an F₂ generation is produced from such a cross, the flowers of the offspring appear in a ratio of 1 red: 2 pink: 1 white (Fig. 13.13).

Interactions Among Genes

So far, our discussion of inheritance has focused on single genes or pairs of genes acting independently of each other. This type of autonomous behavior, however, is the exception rather than the rule. Genes are generally responsible for the production of proteins that are components of complex biochemical pathways. It is often necessary to consider the genotype of an individual at more than one locus before a prediction of phenotype can be made. For example, in a plant called blue-eyed Mary (*Collinsia parviflora*), two genes control flower color in a biochemical pathway as follows:



If a plant contains at least one dominant *W* allele, then it can proceed through the first step of the pathway and produce either magenta or blue flowers. The flowers will be magenta if the plant is homozygous recessive for the *M* gene (*mm*) or blue if it has at least one copy of the dominant *M* allele (*MM* or *Mm*), allowing it to complete the second step in the pathway. The enzyme produced by the dominant *W* allele catalyzes a pathway that synthesizes a compound used by the gene *M* enzyme. Therefore, a homozygous recessive (*ww*) plant cannot produce colored flowers even if it contains a dominant *M* allele. Of course, a plant that is homozygous recessive for both genes (*wwmm*) will also produce colorless (white) flowers.

How Genotype Controls Phenotype

Mendel had no way of knowing how his factors (genes) could produce plants that varied in phenotype. However, as our understanding of the molecular basis of genetics increases, so does our ability to explain Mendelian inheritance patterns.

Mendel discovered that the allele for smooth seeds is dominant over the wrinkled seed allele. How does one allele mask the expression of another? Often, the dominant allele codes for a protein that can effectively catalyze a reaction in a cell and produce a phenotype we recognize. The recessive allele, on the other hand, represents a mutant form that has an altered DNA sequence. The protein product that results from transcription and translation of that recessive (mutant) allele is defective. It cannot catalyze the reaction, and, therefore, does not produce a functional product. One copy of the normal allele is usually enough to allow the cell to produce ample quantities of normal protein. Therefore, a heterozygous plant looks normal. A homozygous recessive plant, on the other hand, cannot make any normal protein from its two mutant alleles.

What is the difference between the smooth seed allele and the wrinkled seed one? The smooth pea phenotype (RR or Rr) produces an enzyme allowing the seeds to accumulate high levels of starch. Seeds with the rr genotype do not produce a functional form of that enzyme and so have high levels of sucrose rather than starch. The sucrose in the rr seeds causes them to absorb water during development. When they dry, they lose that water and shrivel. The RR and Rr seeds have lower levels of sucrose and do not absorb as much water during development; consequently, they do not shrivel when they dry. Recent molecular genetic studies have revealed that the r allele produces a defective enzyme because it has had an 800-nucleotide piece of DNA inserted into the gene. As you might expect, compared with the normal allele, transcription and translation of the allele carrying this insert produce a much different (and, in this case, nonfunctional) protein. How did the insert get there? The best guess is that a transposable element inserted into the pea chromosome at this location some time during the pea plant's evolutionary history.

Molecular genetics can also explain how other Mendelian traits are expressed. For example, as you recall, flower color in snapdragons is inherited as an incompletely dominant trait. In this case, one copy of the functional allele produces some red pigment, but two copies produce noticeably more pigment. Red-flowered plants therefore have two copies of an allele that is critical in the red pigment synthesis pathway and are RR . Pink-flowered plants have one copy of that allele, producing light red (pink) pigmentation, and are RR' . (We use superscripts instead of upper- and lower-case letters here because we are not looking at complete dominance.) Finally, white plants have two copies of the defective allele ($R'R'$) and cannot make the enzyme necessary for red pigment production.

QUANTITATIVE TRAITS

While simply inherited traits such as snapdragon flower color effectively demonstrate Mendel's principles, many traits are under much more complex genetic control. **Quantitative traits**, such as yield and days to flowering, exhibit a range of phenotypes, rather than the discrete phenotypes studied by Mendel. Typically, they are dramatically influenced by environment. Consider tomato fruit yield as an example of a quantitative trait. Under identical environments, some tomato plants produce more fruit than others due to genetic differences. However, genetically identical plants will also produce different amounts of fruit when grown under different environments. They will produce large yields under optimal growing conditions and lower yields under stressed conditions. Therefore, phenotype (yield in this example) is determined by both genotype and environment. This is the "nature versus nurture" concept. Because quantitative traits are strongly influenced by environment and are generally controlled by many genes, they do not produce typical Mendelian ratios. Instead, statistical tools such as distributions, means, and variances are used to study quantitative traits. Molecular geneticists are able to identify chromosomal fragments, called **quantitative trait loci**, or QTLs, associated with quantitative traits. Presumably, these fragments contain genes that influence the trait and behave like Mendelian genes. Using this approach, quantitative geneticists are beginning to understand the nature of complex traits such as growth rate, fruit quality, and flowering time.

EXTRANUCLEAR DNA

You may recall a statement earlier in this chapter indicating that *most* DNA in plants resides in the nucleus. Where is the remaining DNA found? In plants, **extranuclear DNA** is found in both mitochondria and chloroplasts. According to the **endosymbiont hypothesis**, mitochondria and chloroplasts were free-living bacteria at some time in their evolutionary history. They became incorporated into cells of organisms that evolved into plants and established a symbiotic association. As you might expect, then, the DNA in mitochondria and chloroplasts is similar to bacterial DNA.

Some genes involved in photosynthesis and respiration are located in the chloroplast and mitochondrial genomes, respectively. Because many copies of each organelle exist in a plant cell and multiple copies of each gene may be found in each organelle, extranuclear genes do not exhibit Mendelian segregation ratios. In addition, sperm cells rarely carry mitochondria and chloroplasts, so that extranuclear genes are typically passed on to the next generation by only the female parent. This is called **maternal inheritance**.

Leaf variegation in plants such as four-o'clocks exhibits maternal inheritance because it results from mutations in chloroplast synthesis genes. Some herbicides are effective because they inhibit enzymes encoded by chloroplast genes. Genetic engineers are currently trying to manipulate chloroplast genomes to create herbicide-resistant crop plants. See Chapter 14 for a more detailed discussion of genetically engineered plants.

LINKAGE AND MAPPING

There are typically thousands of genes on each chromosome, and the closer the genes are to one another on a chromosome, the more likely they are to be inherited together. Genes that are together on a chromosome are said to be **linked**.

In 1906, just a few years after the basic details of meiosis became known, W. Bateson, R. C. Punnett (after whom the Punnett square is named), and E. B. Saunders became the first to report on linkage in sweet peas. They knew from earlier work with sweet peas that purple flower color was dominant and red was recessive. They also knew that pollen grains with an oblong shape ("long pollen") were dominant and that spherical pollen grains ("round pollen") were recessive.

When Bateson and Punnett crossed a homozygous purple long plant ($PPLL$) with a homozygous red round plant ($ppll$), all the F_1 offspring were purple and long as expected. When they crossed the F_1 plants with one another to obtain an F_2 generation, however, the F_2 offspring phenotypes were produced in numbers that differed markedly from the expected 9:3:3:1 ratio. Puzzled by these results, they tried a backcross, crossing F_1 plants ($PpLl$) with the recessive parent ($ppll$). Again, the expected 1:1:1:1 ratio was not produced. Instead, they obtained a ratio of 7 purple long: 1 purple round: 1 red long: 7 red round. They could not adequately explain what they had observed. In 1910, however, T. H. Morgan, who had observed similarly puzzling ratios in his experiments with fruit flies, correctly theorized that linkage and crossing-over were responsible.

If the genes for purple and red and those for long and round were on separate pairs of chromosomes, then the genotypes of the F_1 would be all $PpLl$, and the homozygous recessive would be $ppll$. A backcross to the F_1 ($PpLl \times ppll$) should have produced 1 $PpLl$: 1 $Ppll$: 1 $pPll$: 1 $ppll$ instead of the 7:1:1:7 ratio actually obtained. Apparently, the F_1 parent produced many more PL and pl gametes than Pl and pL gametes. If we assume the genes are linked, then we can use a slash (/) to separate the alleles on one chromosome from those on the homologous chromosome. The parents were PL/PL and pl/pl . The first parent would produce gametes containing the PL chromosome and the other would produce pl gametes. The F_1 plant, then, would be PL/pl because it received a PL chromosome from one parent and a homologous pl chromosome from the other. The F_1 plant would produce just two kinds of gametes, namely PL and pl , and the backcross ($PL/pl \times pl/pl$) would have produced only two kinds of phenotypes, namely purple long (PL/pl) and red round (pl/pl) in equal numbers. But some purple round and red long phenotypes were also produced. The only plausible explanation for this seems to be that some crossing-over (see prophase I of meiosis in Chapter 12) occurs between genes P and L .

Crossing-over is a regular, seemingly random event during meiosis. Therefore, it may occur by chance between any pair of linked genes. Each gene has a specific location (**locus**) on a chromosome and, presumably, crossing-over is more likely to happen between two genes that are far apart on a chromosome than between two genes that are close together. We can use the frequency of crossing-over between genes to create a genetic map of each chromosome. A genetic map is similar to a map of a straight road, with genes corresponding to cities and map distances corresponding to the distances between the cities. A **map unit** equals 1% crossing-over between a pair of genes. Suppose we make a backcross ($PL/pl \times pl/pl$), and 90% of the offspring are either PL/pl or pl/pl . These are **parental type** offspring because the gametes from the heterozygous parent (PL and pl) do not carry chromosomes for which a crossover between P and L occurred. The remaining 10% of the offspring are **recombinant types** (Pl/pl and pL/pl) because a crossover occurred between genes P and L to produce Pl and pL gametes. Remember that the heterozygous parent contains P and L on one chromosome and p and l on the homologous chromosome. The only way, then, it can produce Pl and pL gametes is through crossing-over. Because 10% of the offspring (and, therefore, gametes) were recombinant, the map distance between genes P and L is 10 map units.

In Bateson and Punnett's backcross, two of every sixteen (12.5%) of the offspring were produced through recombination due to crossing-over. We can deduce from this that the genes controlling flower color and pollen shape in sweet peas are 12.5 map units apart on a chromosome. It is interesting that Mendel did not observe linkage in his studies. The traits he analyzed happened to be on different chromosomes in peas. Imagine the difficulty he would have encountered if he had chosen to work on genes that were linked.

Linkage maps are essential tools for geneticists because they give us a physical framework in which to organize genes. Massive mapping efforts are currently underway in all major crop plants, with new genes and DNA sequences being added to maps every day. A simplified example of a genetic map of the pea is shown in Figure 13.14.

THE HARDY-WEINBERG LAW

Before Mendel's work became known, most biologists studied quantitative traits and believed that inherited characteristics resulted from a blending of those furnished by the parents. It was difficult to understand why unusual characteristics did not

eventually become so diluted that they essentially disappeared. After Mendel, R. C. Punnett and other biologists asked why dominant genes did not eventually completely eliminate recessive ones in breeding populations. After all, a cross between two heterozygotes produces only 25% homozygous recessive individuals. G. H. Hardy, a mathematician, and W. Weinberg, a physician, pointed out the reason in 1908, and their observation became known as the Hardy-Weinberg law. The **Hardy-Weinberg law**, which essentially specifies the criteria for genetic equilibrium in large populations, states that the proportions of dominant alleles to recessive ones in a large, random mating population will remain the same from generation to generation unless there are forces that change those proportions. In small populations, for example, random losses of alleles can occur if, by chance, the individuals carrying those alleles do not mate as often as other individuals. In populations of any size, selection is the most significant cause of exceptions to the Hardy-Weinberg law and can cause dramatic changes in the proportions of dominant to recessive alleles. Discussions of artificial selection and natural selection are found in Chapters 14 and 15, respectively.

Summary

1. McClintock's work with corn changed beliefs about genes and chromosomes.
2. A DNA molecule consists of nucleotides. A typically single-stranded RNA molecule has a different sugar, and uracil instead of thymine.
3. Watson, Crick, and Wilkins developed a model of a DNA molecule, now considered authentic. It is a -double-stranded helix resembling a spirally twisted ladder.
4. DNA stores genetic information, replicates itself, expresses its information, and can mutate.
5. A genome is the sum total of the information in an organism's DNA.
6. A DNA molecule replicates by "unzipping"; the resulting single strands serve as templates for two double-stranded molecules.
7. With DNA as a template, mRNA, tRNA, and rRNA are made during transcription.
8. During translation mRNA from transcription is used to make proteins; tRNA decodes mRNA information. Ribosomes are made of rRNA and proteins.
9. Spontaneous or induced mutations are changes in DNA sequence.
10. Inversions and translocations may be important speciation mechanisms.
11. An aneuploid has a normal chromosome number plus or minus one to several chromosomes. A polyploid has extra complete sets of chromosomes.
12. Gregor Mendel originated the science of genetics while experimenting with peas. The pea varieties he crossed had pairs of contrasting characteristics on homologous chromosomes. Parent plant offspring were called the F_1 generation; the F_2 generation resulted from F_1 plant crosses. F_1 plants, called hybrids, all resembled their parents. F_2 offspring were produced in a 3:1 ratio of plants resembling one or the other parent.
13. The agents controlling the plants' characteristics were called "factors"; Mendel deduced that each plant had two factors (later known as alleles) for each characteristic. His law of unit characters states that "factors, which always occur in pairs, control the inheritance of various characteristics." The suppressing factor was called dominant and its counterpart recessive.
14. Phenotypes (described with words) denote appearance; genotypes (shown with letters) designate genetic makeup. Capital letters designate dominant alleles; lower case letters designate recessive alleles.
15. Paired homozygous plant alleles are identical. Paired heterozygous plant alleles are contrasting.
16. Monohybrid cross offspring are produced in a ratio of 3 dominant phenotypes to 1 recessive phenotype. Dihybrid crosses produce a 9:3:3:1 phenotypic ratio. A backcross between an F_1 hybrid and its recessive parent produces a phenotypic ratio of 1:1 (monohybrid) or 1:1:1:1 (dihybrid). A testcross is used to determine if a plant with the dominant phenotype is homozygous or heterozygous.
17. Compared with homozygotes the heterozygotes are intermediate when dominance is incomplete.
18. Enzymes control biochemical reactions. Genotypes control phenotypes.
19. Quantitative trait genotypes are strongly influenced by environment.
20. Linked genes are inherited together. Bateson, Punnett, and Saunders observed a 7:1:1:7 ratio in a dihybrid cross, and postulated it resulted from linkage and crossing-over.
21. Chromosomal mapping requires calculating crossover percentages to determine the positions of genes on chromosomes.
22. The Hardy-Weinberg law explains why recessives in a population do not disappear. Selection is the most significant cause for seven exceptions to the Hardy-Weinberg law.

Review Questions

1. Using S, P, and B as abbreviations for sugar, phosphate, and base, respectively, draw a DNA double helix in the arrangement you would see if you untwisted the helix so you were looking at a ladder structure.
2. If a DNA molecule had a string of cytosine-thymine base pairs, how would it differ in width from a normal molecule?
3. Explain how DNA replicates.
4. Using the genetic code, determine the amino acid sequence produced by the following gene (DNA) sequence. Remember that during transcription, RNA forms complementary base pairs with DNA. For example, if the DNA sequence is TAG, then transcription would produce an RNA fragment with the sequence AUC. TACACAGCAACT

Discussion Questions

1. Why do you suppose gene expression requires an intermediate RNA molecule? That is, why does the translation machinery read mRNA instead of the gene itself?
2. If proofreading enzymes became more accurate and efficient, then what do you suppose would happen to the speed at which organisms evolve?
3. Mendel's peas were self-pollinating. Describe advantages and disadvantages of self-pollination.

Additional Reading

- Comfort, N. C. 2001. *The tangled field: Barbara McClintock's search for the patterns of genetic control*. Cambridge, MA: Harvard University Press.
- Frank-Kamenetskii, M. D. 1997. *Unraveling DNA: The most important molecule of life*. Reading, MA: Addison-Wesley.
- Griffiths, A. J., W. M. Gelbert, and S. R. Wessler. 2004. *Introduction to genetic analysis*. New York: W. H. Freeman.
- Hartwell, L. H., A. E. Reynolds, L. M. Silver, R. C. Veres, and L. Hood. 2003. *Genetics: From genes to genomes*. 2d ed. Boston: McGraw-Hill.
- Henig, R. M. 2000. *The monk in the garden: The lost and found genius of Gregor Mendel, the father of genetics*. Boston: Houghton Mifflin.
- King, R. C., and W. D. Stansfield. 1997. *A dictionary of genetics*, 5th ed. New York: Oxford University Press.
- Lewin, B. 2003. *Genes VIII*. New York: Oxford University Press.
- Pierce, B. A. 2004. *Genetics: A conceptual approach*. New York: W. H. Freeman.
- Watson, J. D. 2001. *Double helix: A personal account of the discovery of the structure of DNA*. New York: Simon & Schuster.

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Narrow-tubed monkey flowers (*Mimulus angustatus*), a vernal pool species of northern California. (Courtesy Robert A. Schlising)

Figure 13.1 Part of an ear of corn showing the effects of transposable elements. The speckles in the center kernel are clusters of cells in which a transposable element has removed itself from a purple pigment gene so purple pigment is produced. Yellow areas in that kernel do not express purple pigment because the transposable element has interrupted that pigment gene.

Figure 13.2 Structure of a DNA molecule. In this enlargement of a small portion of a DNA molecule, the rungs of the twisted ladder formed by the two entwined spiraling strands consist of nitrogen-containing bases supported by alternating units of sugar (S) and phosphate (P) molecules. The purines adenine or guanine (A or G) occur opposite the pyrimidines thymine or cytosine (T or C). The purines and pyrimidines opposite each other are held together by hydrogen bonds linking the nitrogenous bases of the paired molecules. The double helix is 2 nanometers wide.

Figure 13.3 The pairing of nucleotides in a tiny portion of a molecule of DNA. The variations in sequences of pairs are virtually unlimited.

Figure 13.4 Replication (duplication) of DNA. A molecule replicates by “unzipping” along the hydrogen bonds, which link the pairs of nucleotides in the middle, and then each half serves as a template for nucleotides available in the cell. As the new nucleotides fit into the appropriate places and are linked together, a double helix is re-formed for each original half.

The Polymerase Chain Reaction (PCR)

How many times have you said to yourself, “Why didn’t I think of that?” Many scientists must have had that thought when Kary Mullis described the polymerase chain reaction (PCR) in 1984. While Mullis was driving to his cabin in northern California one evening, he was thinking about a method for determining nucleotide sequences in DNA molecules. The technique used DNA replication enzymes to synthesize short strands of DNA in a test tube. After running through several scenarios in his head, he realized that he could create a chain reaction in a test tube. Every reaction would double the amount of DNA synthesized, causing an exponential growth in the number of DNA molecules created. Twenty cycles of DNA replication would create from a single molecule a million identical DNA molecules. For years scientists had worked with replication enzymes in DNA synthesis experiments, but none of them had conceived the power of a chain reaction in a test tube.

The polymerase chain reaction allows scientists to replicate (duplicate) specific DNA sequences in small test tubes. The reaction requires DNA polymerase, nucleotides (actually nucleoside triphosphates, the high-energy precursors to nucleotides), short (approximately 20 nucleotide) single-stranded DNA molecules called *primers*, and DNA from an organism to serve as a template. The reaction follows these steps:

1. The primer is made with the aid of a DNA synthesizer. The primer DNA sequence is critical because it determines what piece of an organism’s DNA will be replicated during the PCR reaction.
2. DNA polymerase, nucleotide building blocks, primers, and DNA template are mixed together in a small test tube.
3. The test tube is placed in a *thermal cycler*. This machine is capable of rapidly heating and cooling samples.
4. The tube is heated to approximately 94°C to separate the template DNA into single-stranded molecules. In a living cell, enzymes carry out this process. In an experimental setting, however, heat provides a simple way to make the DNA single-stranded.
5. The tube is then cooled to 35°C to 60°C. This allows the primers to locate and bind to complementary base sequences on the template DNA. Remember that DNA molecules are typically double helices with adenine--thymine pairs and guanine-cytosine pairs.
6. The tube is heated to 72°C, which is the optimal temperature for the DNA polymerase used in PCR reactions. Because the enzyme for PCR is derived from thermophilic (heat-loving) bacteria, it is not destroyed by heating (see Chapter 17). DNA polymerase locates the short, double-stranded regions where primers have bound to template DNA; it then uses nucleotide building blocks to make double-stranded DNA near the primer.
7. Steps 4 through 6 are repeated 20 to 30 times.

After completion of the PCR reaction, millions of copies of a double-stranded DNA molecule are present in the test tube. The real power of the reaction, though, comes from its ability to amplify only the DNA sequence adjacent to the primer binding site. This allows a researcher to make an endless supply of a specific segment of DNA. The illustration (Box Figure 13.1) shows how a target DNA sequence is amplified through cycles of PCR.

The polymerase chain reaction has become a standard tool in many research laboratories. Countless applications for this technique have been developed and more are on the way. PCR is an important tool because (1) large quantities of DNA can be synthesized from very small samples and (2) only the target DNA sequence is amplified.

How is the PCR reaction used today? Here is a short list of examples:

1. **Forensics.** Samples of hair, blood, and skin contain enough DNA to be amplified by PCR. A comparison of amplified DNA samples from a crime scene with DNA from a suspect can provide compelling evidence.
2. **Disease detection.** Primers have been developed specifically to amplify DNA from pathogens. For example, a person’s blood could provide template DNA for the PCR reaction with a primer that binds only to a sequence from the human immunodeficiency virus (HIV). Then, even low levels of DNA produced by the virus would be amplified by PCR. This makes early disease detection possible. PCR is also being used to identify pathogens in plants.
3. **Food safety.** Pathogens in food can be detected quickly and accurately using PCR. Primers

specific for pathogenic strains of *Escherichia coli* can detect very low levels of the pathogen in hamburger. Similar tests are available for detection of the organisms that cause *Salmonella* poisoning and botulism, among others.

4. **Fetal testing.** A small sample of fetal tissue can be amplified using primers specific to DNA regions that, in mutant form, cause human diseases. Primers are available to detect sickle cell anemia, phenylketonuria, cystic fibrosis, hemophilia, and Huntington's disease, to name a few. Of course, a moral dilemma is created if the fetus tests positive.
5. **Evolutionary relationships.** DNA from extinct animals can be amplified and compared with the DNA of modern relatives. With the use of PCR, DNA from a 120-million-year-old weevil and from a 250-million-year-old bacterium have already been extracted and amplified.
6. **Conservation efforts.** DNA from tissue samples collected at poaching sites can be amplified and compared to the DNA of pelts from suspected poachers. PCR also allows scientists to determine whether commercial products containing ivory, meat, or feathers were taken from endangered species.
7. **Genetics of dung.** Even DNA from dung can be amplified with PCR! Scientists trying to understand why the giant ground sloth became extinct have located 20,000-year-old dung in caves. PCR amplification of the dung is being used for comparison with modern relatives. In addition, careful choice of primers allows researchers to amplify DNA from the animal's food and its parasites.

Only a few examples have been presented here. Application of PCR seems to be limited only by our imagination. We are capable of performing this amazingly simple, yet powerful reaction because we understand the elements responsible for DNA replication. The next time, then, when you are asked to memorize the steps in a biological process, don't grumble about it. Instead, see if you can look at it in a unique way and make your friends wonder, "Why didn't I think of that?"

Box Figure 13.1 Polymerase chain reaction (PCR) technology.

Figure 13.5 The genetic code. There are 64 different codons, each of which consists of 3 bases and nucleotides, and specifies 1 of the 20 amino acids plus *stop* and *start* information.

Figure 13.6 Translation. *A.* During initiation, the components necessary for translation bind together. These include the large and small ribosomal subunits, messenger RNA, and the initiator transfer RNA carrying the amino acid methionine. *B.* During elongation, the ribosome moves along the messenger RNA. Transfer RNA molecules bring amino acids to the ribosome, where a polypeptide chain is constructed. *C.* When the ribosome reaches a stop codon, translation terminates. The large and small ribosomal subunits, messenger RNA, transfer RNA, and polypeptide then separate from each other.

Figure 13.7 The Central Dogma of Molecular Genetics. The flow of information in a cell proceeds from the DNA in the nucleus via mRNA to the cytoplasm, where information from the mRNA is translated into proteins.

Figure 13.8 Leaves of potato plants. *Left:* A tetraploid. *Right:* A diploid.

Figure 13.9 Gregor Mendel. Photo ca. 1870s, Brünn, Moravia. (Courtesy Brooklyn Botanic Gardens, New York. Print at Hunt Institute, Pittsburgh, PA.)

Figure 13.10 Two generations of offspring in peas. The F₁ generation is composed of tall plants and was created by crossing a true-breeding tall plant with a true-breeding dwarf (short) plant. The F₂ generation is composed of 75% tall plants and 25% dwarf plants, and was created by intercrossing F₁ plants.

TABLE 13.1

Summary of Mendel's Data

PARENTS	F ₁	F ₂
Yellow × green seeds	All yellow	6,022 yellow: 2,001
Smooth × wrinkled seeds	All smooth	5,474 smooth: 1,850

Green pod × yellow pod	All green	428 green: 152 yellow
Long stem × short stem	All long	787 long: 277 short
Axial flowers × terminal flowers	All axial	651 axial: 207
Inflated pods × constricted pods	All inflated	882 inflated: 299
Purple flowers × white flowers	All purple	705 purple: 224 white

Figure 13.11 A monohybrid cross between a green-podded pea plant and a -yellow-podded pea plant. Green (*G*) is dominant; yellow (*g*) is recessive.

Figure 13.12 A dihybrid cross between a green-podded, tall pea plant and a yellow-podded, dwarf pea plant. Green (*G*) and tall (*T*) are dominant; yellow (*g*) and dwarf (*t*) are recessive.

Figure 13.13 Incomplete dominance. A cross between a red-flowered and a white-flowered variety of snapdragons produces F_1 offspring with an intermediate phenotype.

Figure 13.14 A partial genetic map of the pea. Chromosomes 1 through 7 are listed from left to right. Numbers along the right side of each chromosome are map distances. Letters along the left side are gene abbreviations. Drawings illustrate mutant phenotypes.