CHAPTER 10

Genes and Chromosomes





White tigers have inherited an altered gene for coat color. Because their cells cannot make orange pigment, white tigers are white where normal tigers are orange. Studies of the fruit fly Drosophila *(inset) have given geneticists insight into how normal genes and altered genes regulate the inheritance of traits.*

In the preceding chapters we set the stage for a detailed discussion of genes and their activities. We have seen that DNA is the genetic material. We know how the genetic code is written into DNA and how that code is translated into protein. We understand many of the things that proteins can do, and we appreciate their importance. We have recently learned how the science of genetics was developed, how the location of genes can be determined, how the chromosome is put together, and the rules that govern the passage of genetic information from one generation to the next. With all of these things behind us, we are like pilots with all of our ground training and study completed. Now we will begin to fly. We will see how the gene actually works!

CHAPTER OUTLINE

10–1 The Chromosome Theory of Heredity
- Genes and Chromosomes
- Gene Linkage
- Crossing-Over
- Gene Mapping
- Sex Linkage

10–2 Mutations
- Chromosomal Mutations
- Mutations in Genes

10–3 Regulation of Gene Expression
- Gene Interactions
- Gene Expression in Prokaryotes
- Gene Expression in Eukaryotes

10–1 The Chromosome Theory of Heredity

Section Objectives

- Relate genes to chromosomes.
- Explain how gene linkage and crossing-over affect heredity.
- Describe the patterns of inheritance for sex-linked traits.

History records Gregor Mendel as the founder of the science of genetics. Yet Mendel's work was incomplete because he never asked an important question that was the logical outcome of his work: Where in the cell are the factors that control heredity? In other words, where are the genes?

We can hardly blame Mendel for this shortcoming. Even if he had been inclined to ask, he would not have been able to search for genes among the bewildering variety of structures and components within cells. Shortly after completing his genetic experiments, Mendel was promoted to abbot, or head of the monastery. His administrative duties kept him too busy to continue his experiments. Fortunately, other scientists who had the time had begun to study cell structures and the processes of cell division. Their observations would prove to have enormous impact on the study of heredity.

Genes and Chromosomes

By the time Mendel's work was rediscovered in 1900, cell biologists had discovered most of the major structures within cells. They had also recorded the sequences of events that

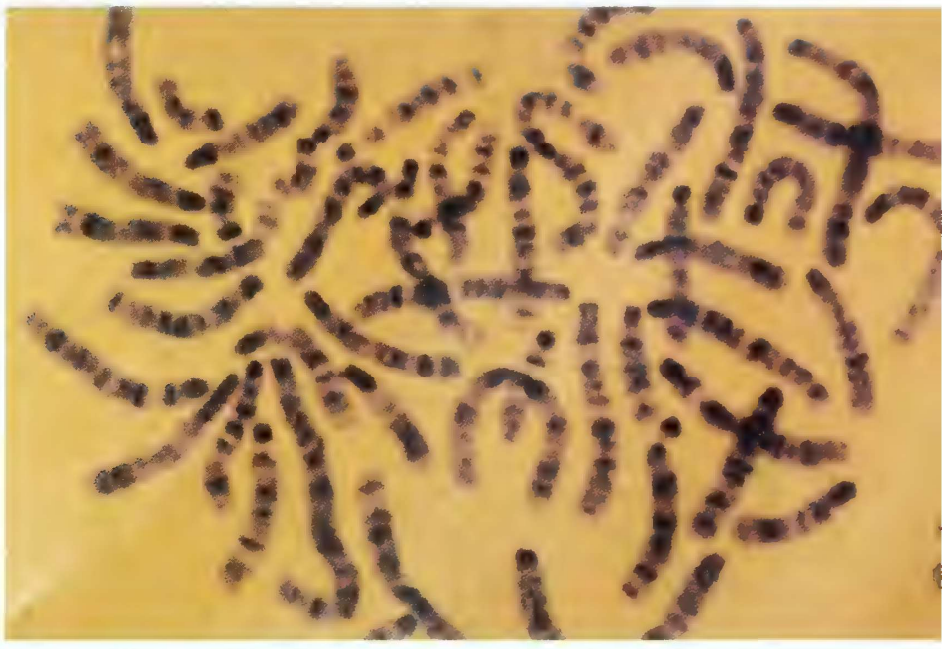


Figure 10–1 *These chromosomes are from a mouse cell. Chromosomes, which are located in the nucleus of a cell, are precisely separated during cell division—an indication that they contain something extremely critical to the cell.*

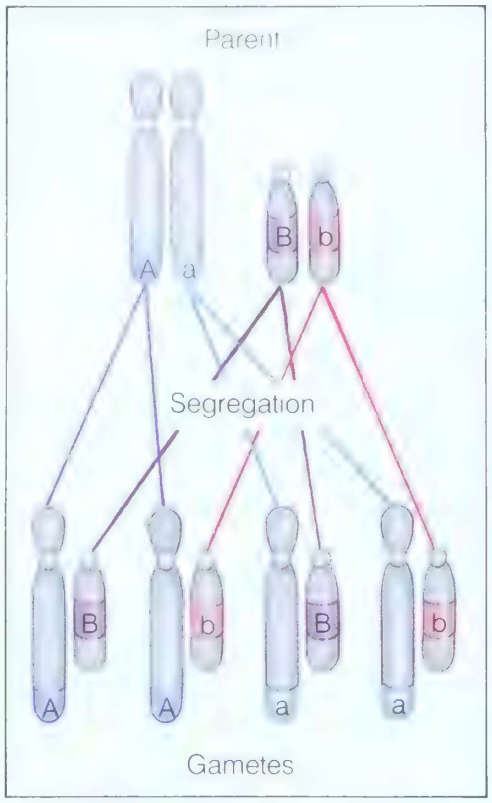


Figure 10–2 *According to Mendel's hypothesis, the two factors for each trait are segregated during gamete formation. Thus gametes have only one factor for each trait. This hypothesis is supported by the observations that homologous chromosomes are separated during meiosis and that gametes contain only one of the chromosomes of each homologous pair.*

occur during mitosis and meiosis. With this information, it seemed logical that the nucleus was the place for genes to be located. In addition to its large size and central location (which make it look important), the nucleus contains chromosomes—structures that behave in complex and interesting ways during cell divisions. The careful and precise mechanism by which chromosomes are separated during mitosis clearly suggests that they are structures vital to the cell. Could this be because they carry the cell's genes?

Further evidence that genes are located on chromosomes is the behavior of chromosomes during meiosis, the process by which reproductive cells are formed. In meiosis, diploid cells, which have 2 copies of each chromosome, divide to form haploid cells, which have 1 copy of each chromosome. This fits in with Mendel's hypothesis that an organism has 2 factors for each trait and that reproductive cells have only 1 factor for each trait. During the first part of meiosis, homologous chromosomes are separated from each other. This fits in with Mendel's hypothesis that the 2 factors for each trait are segregated, or separated, during the formation of gametes.

Walter Sutton, a young graduate student at Columbia University, arrived at the answer to the question of gene location in 1902. **The factors (genes) described by Mendel are located on chromosomes.** The tools with which Sutton made this discovery were a microscope—which he used to observe chromosomes during sperm formation in grasshoppers—and his own imagination, in which he compared the chromosomes to Mendel's factors. When the numbers and movements of chromosomes were analyzed, it was clear to Sutton that chromosomes behaved exactly as one would expect of the carriers of genetic information.

Sutton's **chromosome theory of heredity** states that genes are located on the chromosomes and each gene occupies a specific place on a chromosome. A gene may exist in several forms, or alleles, and each chromosome contains one allele for each gene.

Gene Linkage

The fact that genes are located on chromosomes is important. Genes on a chromosome are linked together. This means that they are inherited together. In other words, **linked genes** do not undergo independent assortment. Recall Mendel's experiment in which the genes for seed color and seed shape were shown to assort independently. Unlike linked genes, genes that are located on separate chromosomes do, in fact, assort independently during meiosis.

One of the earliest examples of linked genes was discovered in the first part of this century by the American geneticist Thomas Hunt Morgan. Morgan studied the tiny fruit fly, *Drosophila melanogaster*, which can produce a new generation every

four weeks. This short generation time makes *Drosophila* an ideal organism to study because traits in succeeding generations can be observed relatively quickly.

THE EFFECTS OF GENE LINKAGE Morgan crossed pure-bred flies that had gray bodies and normal wings with pure-bred flies that had black bodies and small wings. Because gray (*G*) is dominant over black (*g*), and normal wings (*W*) are dominant over small wings (*w*), all of the F_1 flies should have been gray with normal wings (genotype *GgWw*). That is exactly what Morgan observed.

However, when the F_1 flies (*GgWw*) were crossed with black small-winged flies (*ggww*), Morgan did not observe the expected results. If the principle of independent assortment were true for the *GgWw* X *ggww* cross, Morgan would have observed 25 percent (1/4) gray normal-winged, 25 percent (1/4) black small-winged, 25 percent (1/4) gray small-winged, and 25 percent (1/4) black normal-winged. Instead, Morgan obtained very different results for the cross.

As you can see in Figure 10–4, Morgan's actual results differed significantly from those predicted. Most gray-bodied flies had normal wings, and most black-bodied flies had small wings. These results indicated that the gene for body color and the gene for wing size were somehow connected, or linked. Morgan concluded that the two genes were linked by a physical bond in such a way that they could not assort independently.

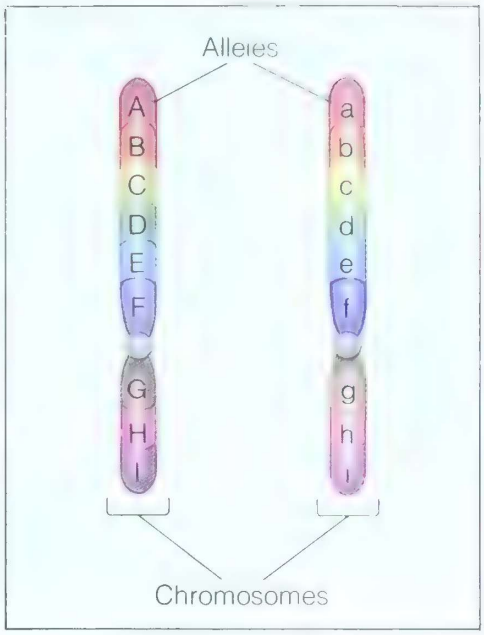
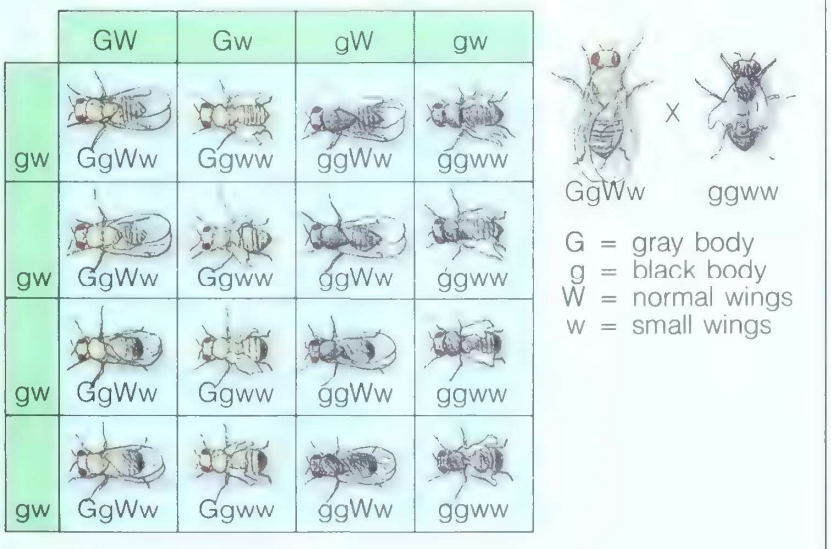


Figure 10–3 *According to Sutton's chromosome theory of heredity, genes are located on the chromosomes. Homologous chromosomes have alleles for the same traits. These alleles may be the same on both homologous chromosomes or they may be different.*



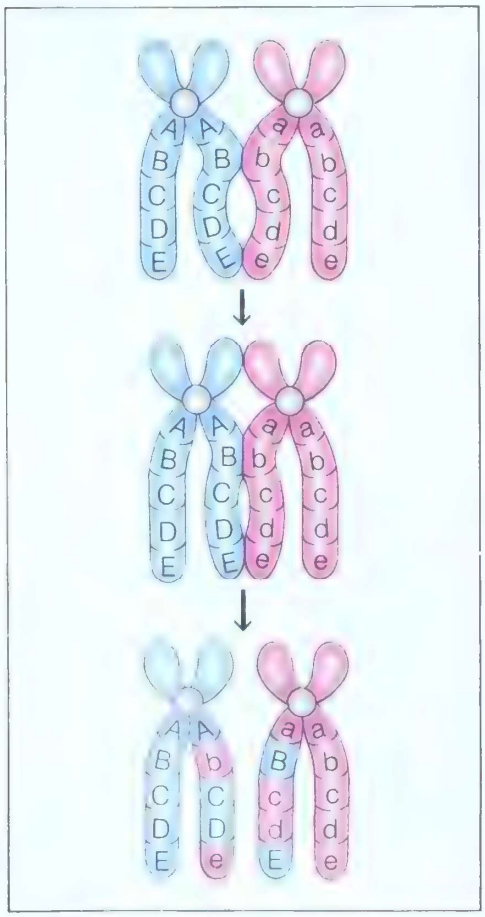
Expected results:	Actual results:
25% (¼) gray normal	41.5% gray normal
25% black small	41.5% black small
25% black normal	8.5% black normal
25% gray small	8.5% gray small

Figure 10–4 *If the principle of independent assortment were true for the genes for body color and wing size in the fruit fly* Drosophila, *the offspring of the cross* GgWw × ggww *would have a 1:1:1:1 phenotypic ratio. Because the actual results did not resemble the expected results, Morgan concluded that the genes for body color and wing size were linked.*



Figure 10–5 *A cobra has 38 chromosomes. How many linkage groups would this make?*

Figure 10–6 *During prophase I of meiosis, homologous chromosomes may exchange sections of their chromatids in a process called crossing-over.*



LINKAGE GROUPS As Morgan and his associates studied more and more genes, they found that the genes fell into distinct **linkage groups**, or "packages" of genes that always tended to be inherited together. The linkage groups themselves assorted independently, but all the genes on one group were inherited together. The linkage groups, of course, were chromosomes. Because homologous chromosomes contain the same genes, there is one linkage group for every homologous pair of chromosomes. *Drosophila* has four linkage groups. It also has four pairs of chromosomes. Corn has ten linkage groups and ten pairs of chromosomes.

Crossing-Over

Although the idea of linkage groups explains some of the results of the fruit fly crosses, it does not provide a complete explanation. Look at the results of the test cross between the *GgWw* and *ggww* flies again. Although 83 percent (41.5% + 41.5% = 83%) of the flies have gene combinations like their parents, 17 percent have new combinations. The 17 percent are, in the language of geneticists, **recombinants**—individuals with new combinations of genes.

If the genes for body color and wing size are linked, why aren't they linked all the time? Morgan and his associate, Alfred Sturtevant, proposed that the linkages could be broken some of the time. If two homologous chromosomes were positioned side by side, sections of the two chromosomes might cross, break, and reattach. As you can see in Figure 10–6, this process would rearrange the genes on the chromosome and produce new linkage groups. Morgan and Sturtevant called this hypothetical process crossing-over and suggested that it might be the reason for the recombinants in the offspring of the *Drosophila* test cross.

Crossing-over does indeed take place. It occurs during the first meiotic division, when homologous chromosomes are paired. Crossing-over produces new combinations of alleles on each chromosome and thus increases genetic variety.

Gene Mapping

Sturtevant further reasoned that crossing-over occurs at random along the linkage groups, and the distance between two genes determines how often crossing-over occurs between them. If two genes are close together, then crossing-over between them is rare. However, if two genes are far apart, then crossing-over between them is more common. Knowing the frequency with which crossing-over between two genes occurs makes it possible to map the positions of genes on a chromosome. This is precisely what Sturtevant tried to do with the genes in *Drosophila*'s linkage groups. And by 1915, just two years after he had begun, Sturtevant had mapped 85 genes in *Drosophila*.

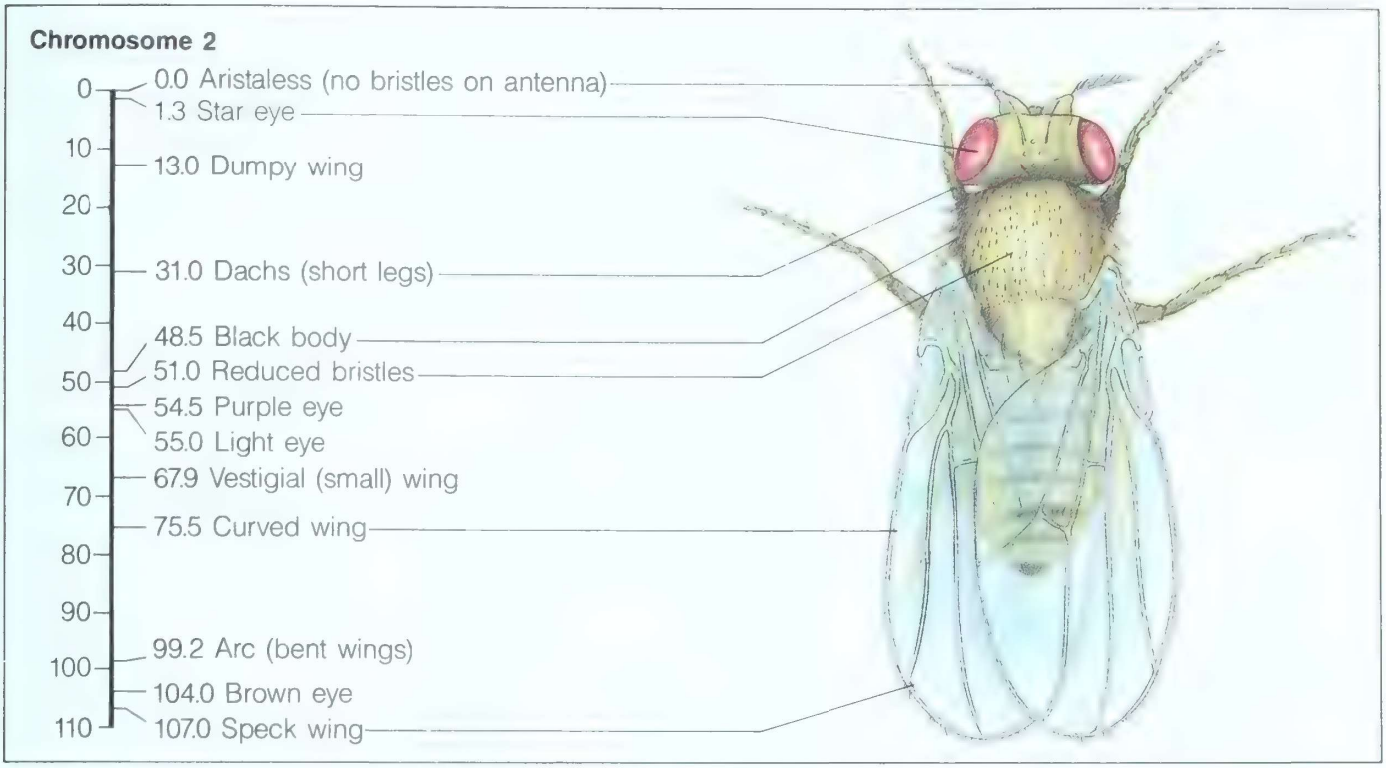


Figure 10–7 *Knowing the frequency with which crossing-over occurs between genes makes it possible to map the positions of genes on chromosomes.*

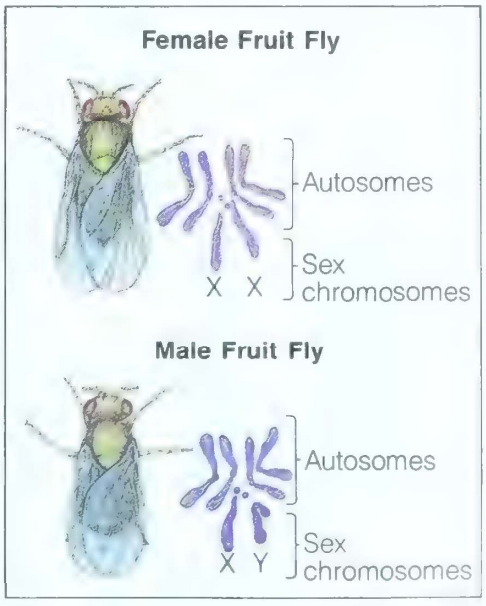
Morgan and Sturtevant were correct in their ideas about linkage groups, crossing-over, and the formation of gene maps. Today we have detailed maps of *Drosophila* that pinpoint the locations of more than 1500 different genes.

Sex Linkage

There is an exception to the rule that every chromosome has a corresponding homologous chromosome. This exception was discovered in 1905 by American biologist Nettie Stevens. She noticed that the cells of the female mealworm contain "20 large chromosomes, while those of the male contain 19 large ones and 1 small one." One of the pairs of chromosomes in the male mealworm is not, in the strictest sense of the term, homologous. The 2 chromosomes in the pair have very different shapes. Stevens found the same situation in *Drosophila*. The cells of the female *Drosophila* contain 8 chromosomes that can be arranged in 4 pairs of 2. The cells of the male, however, contain 3 sets of chromosomes that pair up nicely and 2 remaining chromosomes that do not match. One chromosome looks like a member of a pair of chromosomes found in the female, but the other looks completely different. It is small, shaped like a hook, and in no way similar to any of the other 7 chromosomes.

These seemingly mismatched chromosomes are the **sex chromosomes**. The other chromosomes, which are the same in both males and females, are called **autosomes**. The female *Drosophila* has two matching sex chromosomes—**X chromosomes**. The male has two dissimilar sex chromosomes—one X

Figure 10–8 *A female fruit fly has 2 matching sex chromosomes—X chromosomes. A male fruit fly has sex chromosomes that do not match—1 X chromosome and 1 Y chromosome.*



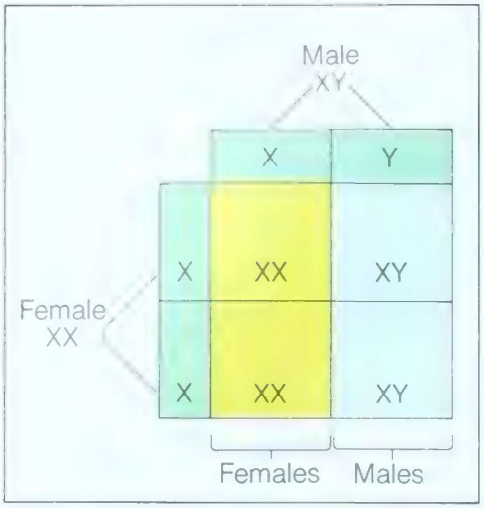


Figure 10–9 *In animals such as fruit flies and humans, the sex chromosomes in the male's gametes usually determine the sex of the offspring.*

chromosome and one **Y chromosome**. It is the Y chromosome that is small and hook-shaped. If we consider just sex chromosomes, we can refer to the male as XY and the female as XX.

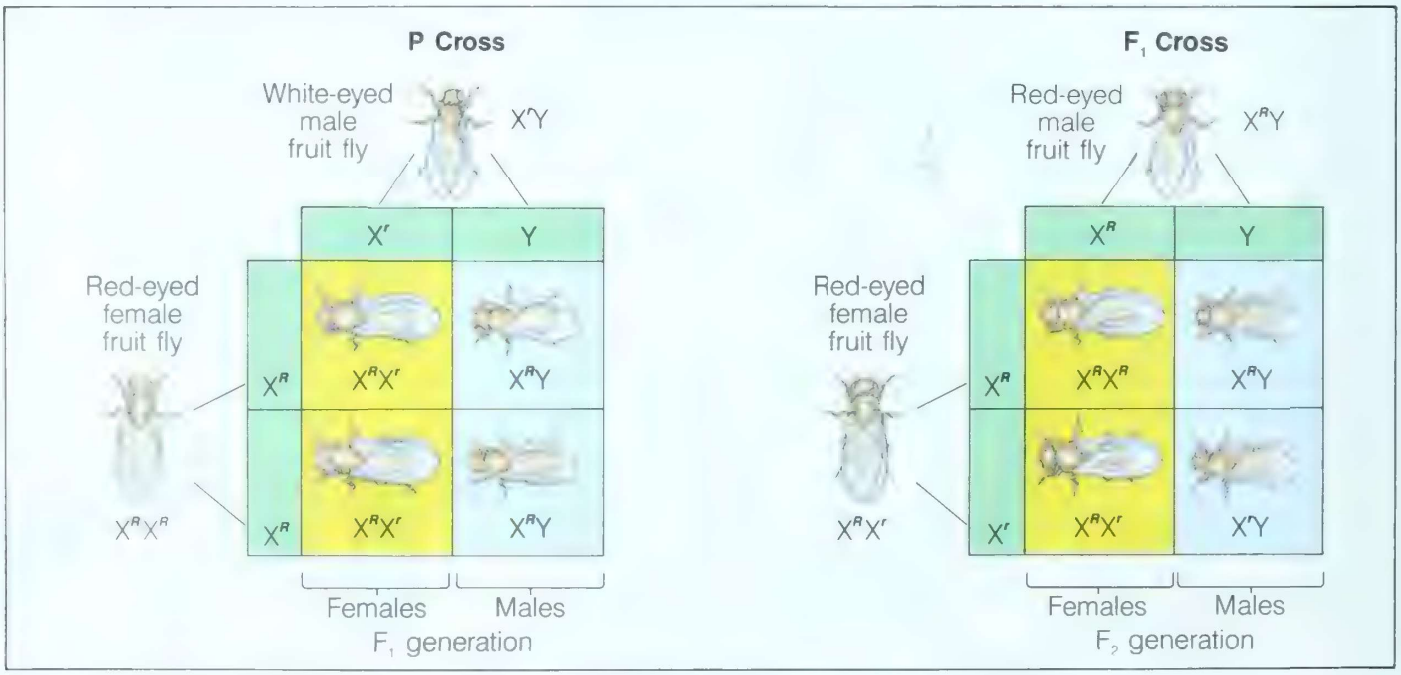
SEX DETERMINATION When female gametes are produced, meiosis separates one of the X chromosomes into each egg cell. In the male, meiosis separates the X and Y chromosomes so that 50 percent of the sperm cells carry a Y chromosome and 50 percent carry an X chromosome. When a Y sperm fertilizes an egg, a male fly (XY) is produced. When an X sperm fertilizes an egg, a female fly (XX) is produced. In a sense, the male is responsible for the sex of its offspring.

Many animals, including humans, follow the same system. We have 22 pairs of autosomes and 1 pair of sex chromosomes: XX in females and XY in males. Because about 50 percent of male sperm have the X chromosome and about 50 percent have the Y, the ratio of males to females is about 50:50, or 1:1.

GENES ON SEX CHROMOSOMES In addition to determining the sex of an individual, the sex chromosomes carry genes that affect other traits. A gene located on one of the sex chromosomes is said to be **sex-linked**. Morgan discovered the first sex-linked gene in his work with *Drosophila*.

Morgan crossed a group of white-eyed male flies with a group of red-eyed female flies. The allele for red eyes (R) is dominant over the allele for white eyes (r). Just as you might have expected, the hybrid flies of the F_1 generation all had red eyes. See Figure 10–10. Morgan allowed the F_1 flies to interbreed and produce a new generation of flies (the F_2 generation). Mendelian genetics predicts a 3 to 1 ratio of red-eyed flies to white-eyed flies, and that was exactly the ratio Morgan

Figure 10–10 *Morgan discovered the first sex-linked gene in experiments with the fruit fly* Drosophila. *The recessive allele, symbolized as* X^r, *causes white eyes in males—genotype* X^rY.



obtained. But Morgan noticed something strange about the F_2 flies. All of the white-eyed flies were male! There was not a single white-eyed female in the bunch.

Why should half of the males but none of the females have white eyes? Could it be that the gene for eye color was on one of the sex chromosomes?

Morgan realized that he could account for his results if the gene for eye color was on the X chromosome. Refer to the Punnett square in Figure 10–10 as you take a closer look at how the F_2 generation is formed.

The female flies produce two kinds of egg cells. One kind of egg cell has the *R* allele on its X chromosome (we can call those egg cells X^R); the other kind of egg cell has the *r* allele on its X chromosome (X^r). The male flies make two kinds of sperm cells: One kind of sperm cell has the X chromosome and the other kind of sperm cell has the Y chromosome. The X chromosome has the *R* allele (X^R) on it. The Y chromosome, however, has neither the *r* nor the *R* allele on it.

As you can see from the Punnett square, only one combination of chromosomes will produce a white-eyed fly—$X^r Y$. And every fly with that combination of chromosomes is male.

Morgan tested his hypothesis that the gene for eye color was sex-linked in a famous experiment. He crossed the F_2 white-eyed males ($X^r Y$) with F_1 ($X^R X^r$) females. The Punnett square in Figure 10–11 shows the probable results of the cross if, and only if, the gene for eye color is indeed on the X chromosome. According to the Punnett square, 1/2 of the offspring should have red eyes and 1/2 should have white eyes. In addition, the male offspring should be 1/2 red-eyed and 1/2 white-eyed. And the female offspring should also be 1/2 red-eyed and 1/2 white-eyed. Such results were exactly what Morgan obtained. He had confirmed his idea that the gene for eye color was sex-linked.

Sex-linked genes are not only found in *Drosophila*. They are important in humans as well. As you will see in Chapter 11, several important human genes are located on the X chromosome. Such genes include the genes for color vision and blood clotting. As with the eye color gene in *Drosophila*, the effects of recessive alleles tend to show up more frequently in males than in females.

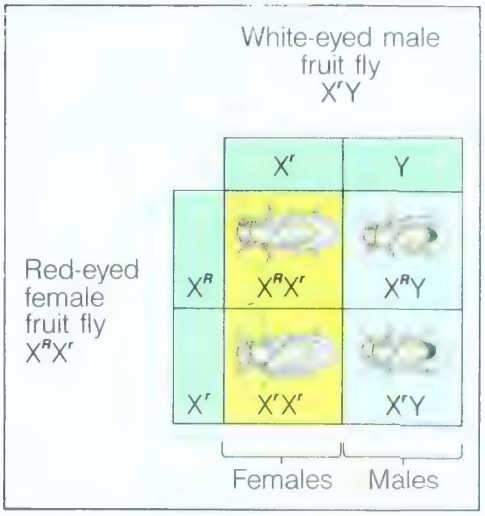


Figure 10–11 *The sex-linked* X^r *allele causes white eyes in females that have a "double dose" of the allele—genotype* X^rX^r. *White-eyed females inherit one* X^r *allele from their male parent (who must be* X^rY*) and one from their female parent (who must be either* X^RX^r *or* X^rX^r*).*

10–1 SECTION REVIEW

1. How are genes related to chromosomes?
2. How does crossing-over make genetic mapping possible?
3. What are sex chromosomes? Autosomes?
4. Why are the effects of recessive sex-linked alleles seen more often in males than in females?

Section Objectives

- Describe different kinds of mutations.
- Explain how mutations can affect heredity.

10–2 Mutations

As you will recall from Chapter 7, the processes involved in duplicating genetic information and transmitting it to the next generation are complex and precise. Although mistakes are rare, they do occur. These mistakes are called **mutations**, from a Latin word that means change. Not all mutations are harmful. Many mutations either have no effect or cause slight, harmless changes. And once in a while a mutation may be beneficial to an organism.

Mutations may occur in any cell. Mutations that affect the reproductive cells, or germ cells, are called germ mutations. The variant characters studied in genetics (such as white eyes in *Drosophila* or wrinkled seeds in pea plants) are the result of germ mutations. Mutations that affect the other cells of the body are called somatic (from the Greek word *soma*, which means body) mutations. Because they do not affect the reproductive cells, somatic mutations are not inheritable. Many cancers are caused by somatic mutations. Both somatic and germ mutations can occur at two levels—the level of chromosomes and the level of genes. **Chromosomal mutations involve segments of chromosomes, whole chromosomes, and even entire sets of chromosomes. Gene mutations involve individual genes.**



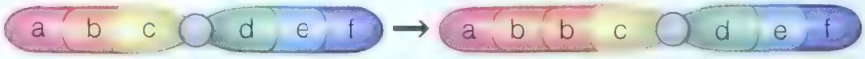
Figure 10–12 *Mutations in genes that regulate development resulted in extra hind legs on this frog.*

Chromosomal Mutations

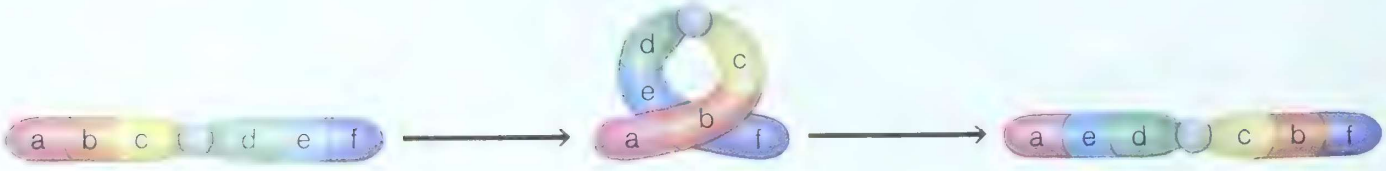
Whenever a **chromosomal mutation** occurs, there is a change in the number or structure of chromosomes. There are four types of chromosomal mutations that involve a change in the structure of a chromosome: deletions, duplications, inversions, and translocations. A deletion involves the loss of part of a chromosome.



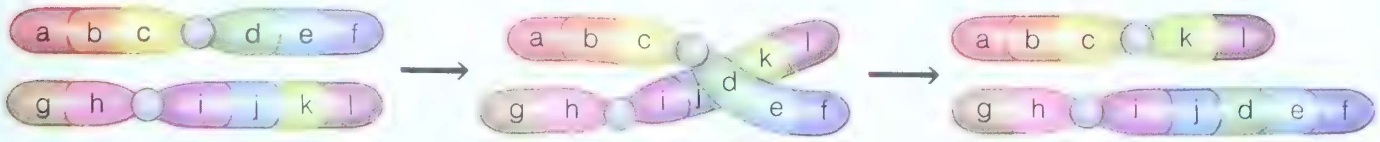
The opposite of a deletion is a duplication, in which a segment of a chromosome is repeated.



When part of a chromosome becomes oriented in the reverse of its usual direction, the result is an inversion.



A translocation occurs when part of one chromosome breaks off and attaches to another, nonhomologous chromosome. In most cases, nonhomologous chromosomes exchange segments, so that two translocations occur at the same time.



Chromosomal mutations that involve whole chromosomes or complete sets of chromosomes result from a process known as **nondisjunction**. Nondisjunction is the failure of homologous chromosomes to separate normally during meiosis. Nondisjunction literally means not coming apart.

When one chromosome is involved, nondisjunction results in an extra copy of a chromosome in one cell and a loss of that chromosome from the other. A number of human disorders that result from nondisjunction will be described in Chapter 11.

Nondisjunction can involve more than one chromosome. When all the homologous chromosomes fail to separate, the result may be a dramatic increase in chromosome number, producing triploid (3N) or tetraploid (4N) organisms. The condition in which an organism has extra sets of chromosomes is called **polyploidy**. Polyploidy is almost always fatal in animals. However, polyploid plants are often larger and hardier than normal plants.

Mutations in Genes

Mutations can occur in individual genes and can seriously affect gene function. Any chemical change that affects the DNA molecule has the potential to produce **gene mutations**. Some gene mutations result from a change involving many nucleotides within a gene; some involve only one nucleotide. The smallest changes, known as **point mutations**, affect no more than a single nucleotide.

Remembering how a DNA base sequence determines the amino acid sequence of a polypeptide, can you predict the possible effects of a point mutation in which one base is substituted for another? Because base substitutions usually affect only a single codon, only one amino acid is affected. In some cases, the sequence of amino acids that the gene codes for may not be changed at all, since different codons may code for the same amino acid. When the point mutation involves the insertion or deletion of a nucleotide, however, the situation may be much more serious. Remember that mRNA is read in groups of three bases. If a single base is inserted or deleted, the groupings are shifted for every codon following the point mutation. Such **frameshift mutations** can completely change the polypeptide product produced by a gene.

Figure 10–13 *A frameshift mutation, such as that caused by the addition of a single nucleotide, can greatly change the polypeptide product of a gene (top). This can make it as useless and nonsensical as a sentence in which the deletion of a letter and a frameshift has occurred (bottom).*

DNA:	TACGCATGGAATACC
mRNA:	AUG CGU ACC UUA UGG
amino acids:	met – arg – thr – leu – try
DNA:	TATCGCATGGAATAC
mRNA:	AUA GCG UAC CUU AUG
amino acids:	iso – ala – tyr – leu – met

THE	FAT	CAT	ATE	THE	RAT
THE	F AT	C AT	A TE	T HE	R AT
TEF	ATC	ATA	TET	HER	AT

Manipulating Chromosome Numbers

From Mendel's work we have come to expect that organisms will be diploid (2N). In other words, they will have two sets of chromosomes—one set inherited from each parent. However, plants with the "wrong" number of sets of chromosomes do exist. And, interestingly enough, these plants are extremely useful in agricultural research.

For example, a plant with only one set of chromosomes shows the effects of all its genes, including potentially desirable recessive genes. A plant with only one set of chromosomes is obtained by taking cells formed in meiosis and growing them under special conditions. However, plants with only one set of chromosomes cannot reproduce sexually because meiosis cannot occur normally in their cells. If these plants show a desirable characteristic, they can be made into diploid plants by treating them with the drug colchicine. Colchicine prevents spindle formation during mitosis and thus causes the number of chromosomes in a cell to double. The diploid plants produced by treatment with colchicine are purebred for the desired traits and can reproduce normally.



By manipulating chromosome numbers with colchicine, it was possible to produce high-protein Triticale.

Colchicine can also be used to change the number of sets of chromosomes in a naturally occurring plant. For example, colchicine has been used to create strawberry and blueberry plants that are polyploid, or have more than two sets of chromosomes. Polyploid plants produce larger fruit than plants with the normal number of sets of chromosomes. The ability to change the number of sets of chromosomes makes it possible to cross plants that have different diploid numbers. The high-protein grain *Triticale* was created by crossing wheat (2N=42) and rye (2N=14). The plants grown from the hybrid seeds were sterile; they could not produce offspring. However, when the hybrid plants were treated with colchicine, the chromosome number was doubled and meiosis could occur normally. Thus the second-generation plants were fertile.

Manipulating chromosome numbers is useful in studying plant genetics. It is also a technique that allows researchers to create new and potentially better crops.

10-2 SECTION REVIEW

1. What is a somatic mutation? How does it differ from a germ mutation?
2. Compare a chromosomal mutation and a gene mutation.
3. How does nondisjunction result in chromosomal mutations?

10–3 Regulation of Gene Expression

Section Objectives

- Discuss gene interactions that influence gene expression.
- Explain how an operon is turned on and off.
- Describe introns and exons.

Like the individual cells of a large organism, individual genes do not function in isolation. The expression and activities of genes, like those of cells, are regulated and controlled, thereby enabling a complex genetic system to function smoothly. **As biologists have intensified their studies of gene activity, it has become clear that interactions between different genes and between genes and their environment are critically important.**

This section focuses on some of the ways in which genes are regulated by conditions within cells. However, genes are also influenced by environmental conditions such as temperature and light.

Gene Interactions

Dominance is the simplest example of how genes interact with each other. In Chapter 9, you learned that the effects of the dominant allele are seen even when the recessive allele is present. But what causes dominance?

Remember that a gene is a section of DNA, and DNA codes for a polypeptide, or string of amino acids. In many cases, the dominant allele codes for a polypeptide that works, whereas the recessive allele codes for a polypeptide that does not work. For example, suppose that the allele *B* codes for an enzyme that makes a black pigment in a mouse's fur and allele *b* codes for a defective enzyme that cannot make the pigment. A mouse that has the genotype *bb* will have white fur because it lacks the enzyme that makes the black pigment. But a mouse that has the genotype *BB* or *Bb* will have black fur because it possesses the enzyme that makes the black pigment. Although each cell in the *Bb* animal has just one copy of the functioning allele, that single copy can code for thousands of mRNA molecules. And each mRNA molecule can code for thousands of enzymes. This is the reason the *B* allele is dominant over the *b* allele.

INCOMPLETE DOMINANCE In 1760 the German scientist Josef Kölreuter reported on experiments in which he crossed white carnations (*rr*) with red carnations (*RR*). Kölreuter found that all of the offspring from his crosses had pink flowers (*Rr*). In other words, the hybrids had a phenotype that was intermediate between those of the parents. At first glance, it might appear as if the parents' genes had blended together. But when Kölreuter crossed his pink F_1 hybrids with each other to form an F_2 generation, the parents' phenotypes reappeared. In the F_2 generation, 1/4 of the plants had red flowers, 1/2 had pink flowers, and 1/4 had white flowers. (This 1:2:1 ratio should be familiar to you from Mendel's crosses.)

Figure 10–14 *For some organisms, such as these caterpillars, gene expression is regulated by chemicals in the food they eat. Each caterpillar of this species has the genetic ability to develop either the twiglike or the flowerlike form. However, caterpillars that eat gray-green oak twigs look like twigs (top). And caterpillars that eat fuzzy gold oak flowers look like flowers (bottom).*



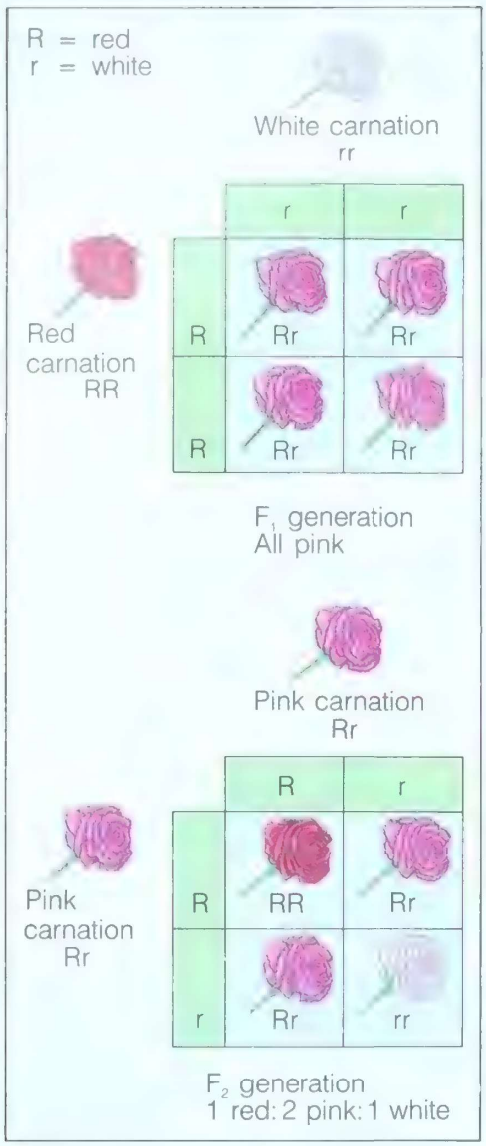


Figure 10–15 *Flower color in carnations is an example of incomplete dominance. The heterozygous phenotype, pink, is somewhere in between the homozygous phenotypes, red and white.*

In carnations, the *R* allele, which codes for an enzyme that makes red pigment, is incompletely dominant over the *r* allele, which codes for a defective enzyme that cannot make pigment. In **incomplete dominance** the active allele does not compensate for the inactive allele, and the heterozygous phenotype is somewhere in between the homozygous phenotypes.

CODOMINANCE Many genes display **codominance**, a condition in which both alleles of a gene are expressed. In other words, both alleles are active. (Remember that in incomplete dominance only one of the alleles is active.) Codominant alleles are written as capital letters with subscripts (for example, B_1 and B_2) or superscripts (for example, R and R').

Codominance is seen in many organisms. For example, red hair (H^R) is codominant with white hair (H^W) in cattle. Cattle that have the genotype H^RH^W are roan, or pinkish brown, in color because their coats are a mixture of red and white hairs. Much the same thing happens in certain varieties of chickens. Black feathers (F^B) are codominant with white feathers (F^W). Erminette chickens (F^BF^W) are speckled black and white.

POLYGENIC INHERITANCE It would be a great mistake to assume that all traits are produced by single genes. Many traits are produced by the interaction of many genes. Traits controlled by two or more genes are said to be **polygenic** (*poly-* means many). For example, at least three enzymes—each of which is produced by a different gene—are involved in making the reddish-brown pigment in the eyes of fruit flies. Different combinations of enzymes (which may be present in the normal form or altered or absent due to a mutation) produce different eye colors. More complicated traits, such as the shape of your nose or the color and markings on an animal's coat, are the result of interactions between large numbers of genes.

Gene Expression in Prokaryotes

In Chapter 7 you learned that individual genes on a chromosome serve as a template, or pattern, for the production of mRNA. In turn, mRNA serves as the instructions for the production of a protein (polypeptide). The genes of a single organism cannot all be activated at the same time. A cell that activated all of its genes at once would make a great many molecules that it did not need and would waste energy and raw materials in doing so. However, when the cell does need the product of a gene, it must be able to produce that product quickly and in adequate amounts.

When the product of a gene (a specific protein) is being actively produced by a cell, we say that the gene is being expressed. Within a single organism, some genes are rarely expressed, some are constantly expressed, and some are expressed for a time and then turned off. But how does a cell

"know" when to make a protein and when not to make it? In other words, how does a cell "know" which genes to turn on and which to turn off?



Figure 10–16 *In certain varieties of chickens, black and white feather colors are caused by codominant alleles. Thus the heterozygous phenotype, speckled black and white, is a result of the expression of both alleles.*

THE OPERON The mechanisms by which genes are turned on and off are quite complex. In this section we will examine one way genes are regulated in prokaryotes (organisms without a nucleus). This particular mechanism was discovered by the French scientists François Jacob and Jacques Monod in 1961. They noticed that genes that work together are often clustered together on a small area of a prokaryote's chromosome. Jacob and Monod also determined that there are regions on a chromosome that lie near these gene clusters but that do not code for the production of proteins. These regions are, however, involved in the regulation and expression of nearby gene clusters. These regions and the gene cluster they regulate are called an **operon** because they operate together.

An operon consists of the following parts: a cluster of genes that work together; a region of the chromosome near the cluster of genes called the **operator**; and a region of the chromosome next to the operator called the **promoter**. As you can see in Figure 10–17, the operator and promoter regions overlap slightly.

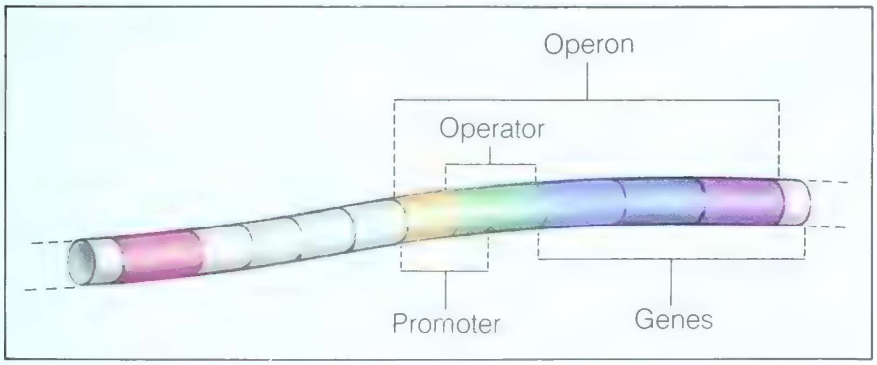


Figure 10–17 *Some genes in prokaryotes are regulated by an operon. An operon consists of several sections of DNA that are operated together. The operon studied by Jacob and Monod, which breaks down the sugar lactose, consists of a promoter, an operator, and a cluster of genes that code for lactose-metabolizing enzymes.*

The gene cluster in the operon that Jacob and Monod studied produces enzymes that break down lactose, a sugar that bacteria can use as a food source. Bacteria do not produce these lactose-breaking enzymes in large amounts unless lactose is present. In other words, the very presence of lactose induces the production of the enzymes necessary to break down lactose for use as food. In this operon system, lactose is called the **inducer** because it induces the production of enzymes.

In order to make the enzymes, RNA polymerase must move along the genes on the chromosome, producing mRNA in the process. Before the RNA polymerase can get to the desired genes, it must first attach to the promoter region near the genes. We might think of the promoter as a "Start Here!" instruction to RNA polymerase. Once the RNA polymerase attaches to the promoter, it can move along the chromosome, past

the operator region, to the genes. When the RNA polymerase reaches the genes, it can produce mRNA, which "instructs" the ribosomes to make the enzymes. When this process is taking place, we say the genes are activated, or being expressed. The enzymes coded for by the genes can then perform their task in the cell—in this case, breaking down the inducer lactose.

Remember that Jacob and Monod observed that the enzymes coded for by the genes in the operon were not produced in the absence of the inducer (lactose). Does this mean that the cell has a way to turn the operon off? Indeed it does.

THE REPRESSOR The cell produces a special protein called a **repressor**. When the repressor nears the operator region of an operon, it attaches itself to the operator so that it sits between the promoter and the genes. The repressor's position blocks the access of RNA polymerase to the genes. Like the guard outside a locked factory, the repressor prevents the workers (RNA polymerase) from getting to their jobs (making mRNA). In other words, the repressor turns the genes of the operon off.

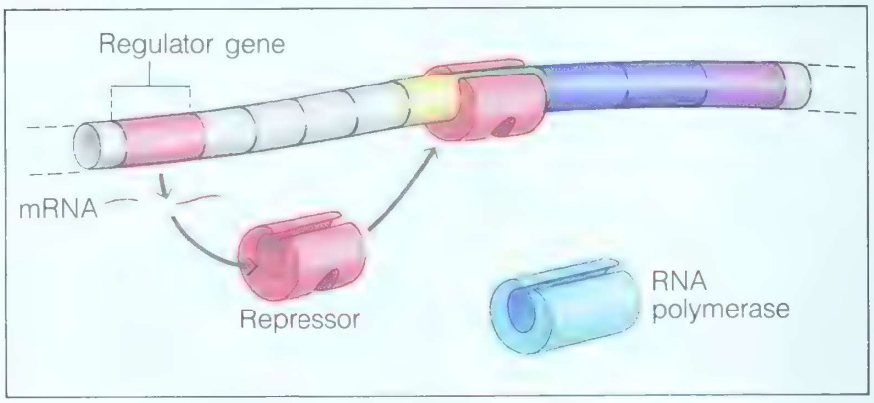


Figure 10–18 *The genes in this operon are turned off when a protein called the repressor binds to the operator and thus prevents the transcription of mRNA.*

You might wonder how the repressor "knows" which operon to attach to and thereby turn off. The actual mechanism is quite elegant and specific. Each repressor is shaped to fit a specific region of DNA on the chromosome. It can attach only to the specific operator on the operon it regulates. Thus each repressor turns off a specific operon.

GENE ACTIVATION You now know that a gene cluster is part of an operon that includes an operator and a promoter. And you also know that the genes are turned off when the repressor binds to the operator. But there is more to the story. How is the operon turned back on when it is needed?

When the inducer enters the cell, it binds to the repressor. And something quite remarkable happens: The repressor changes shape and can no longer bind to the operator. The repressor actually falls off the operator.

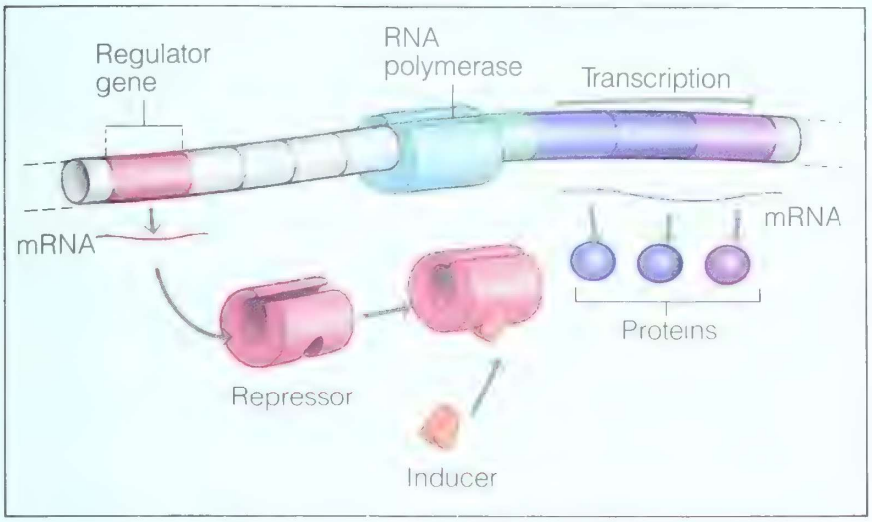


Figure 10–19 *The genes in this operon are turned on when lactose (the inducer) binds to the repressor and causes it to fall off the operator. This allows RNA polymerase to bind to the promoter, move across the operator, and transcribe mRNA from the cluster of genes. The operon system ensures that genes are activated only when their products are needed. In this case, the lactose-metabolizing enzymes are produced only when lactose is present.*

When the repressor falls off the operator, the RNA polymerase can bind to the promoter, move across to the genes, and produce messenger RNA. The mRNA codes for the enzymes that are used to break down the inducer. When the cell runs out of the inducer, the repressor can bind to the operator again, and the operon is turned off. The complete system is automatic and self-regulating. The presence of the inducer causes the cell to make the enzymes needed to use it. And when the inducer disappears, the enzymes are no longer made.

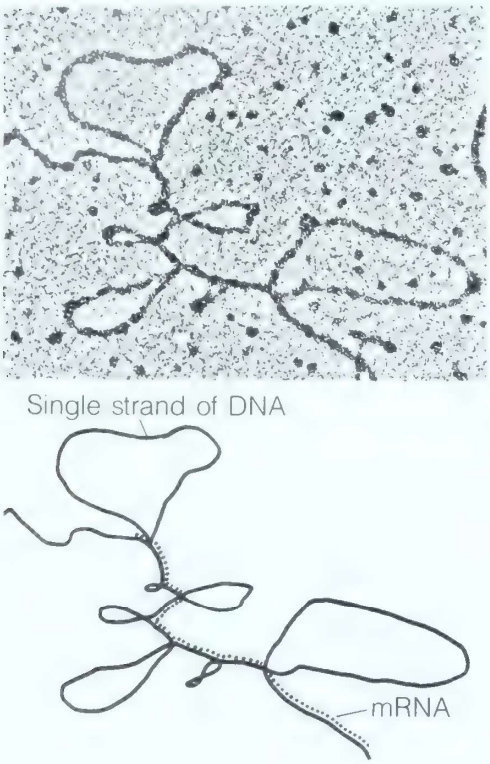
The repressor-operon system provides us with one model of how a group of genes can be regulated so that they are expressed only when they are needed. A number of other gene-regulation systems in prokaryotes have been identified. Some of these gene systems use repressors in a manner similar to that just described. Others use proteins that enhance the rate of transcription. In other systems, regulation occurs at the level of protein synthesis. Regardless of the actual system involved, the result is the same: Cells are able to turn their genes on and off, using the genes only when needed.

Gene Expression in Eukaryotes

The cells of eukaryotes contain a nucleus and membrane-enclosed organelles. Gene regulation in eukaryotes is more complex than in prokaryotes. Increased understanding of control systems in eukaryotes indicates that there are several systems of gene regulation. One system involves a substance called an inducer. Like the inducer in the operon system, inducers in eukaryotes induce, or cause, the activation of genes. In eukaryotes, inducers bind directly to DNA and either start or increase transcription of particular genes.

In 1976 Philip Sharp and Susan Berget discovered that mRNA produced during transcription may be altered before it is used to make protein (polypeptide) during translation. You may recall that during transcription, the base sequence in a

Figure 10–20 *The sequence of bases on the mRNA for this egg-white protein does not exactly match that of its corresponding gene. The DNA sequences that are not complementary to the mRNA appear as loops called introns.*



gene is transcribed into the complementary base sequence on mRNA. Most scientists assumed that a sequence of bases on mRNA was exactly complementary to a DNA sequence for a gene.

Sharp and Berget carefully compared the mRNA of a protein with a piece of DNA containing the gene for that protein. They expected to find an exact pairing between the bases on the gene and the bases on the mRNA that helps produce the protein. Instead, they discovered that the DNA fragment contained large sequences that were not complementary to the base sequence on the mRNA. What was going on?

Sharp and Berget quickly realized that the presence of DNA sequences that are not complementary to mRNA sequences implies that the gene is in "pieces." In other words, DNA sequences that code for protein (the gene pieces) are separated by DNA sequences that do not code for protein. Today we know that many eukaryotic genes contain sequences that are complementary to mRNA and sequences that are not. The sequences that are complementary code for protein. These "expressed" sequences are called **exons**. The segments that are not complementary do not code for protein. These "intervening" sequences are called **introns**.

When RNA polymerase moves along a gene, it transcribes the entire gene. This means that the RNA produced by transcription, or pre-mRNA, contains introns. Before the cell can produce protein, the pre-mRNA must be processed into functional mRNA. During this processing, the introns on the pre-mRNA are removed and the exons are spliced back together. In addition, a chemical "cap" and "tail" are attached to the RNA. At this point, the pre-mRNA can be called mRNA.

The conversion of pre-mRNA into mRNA has further significance because of the fact that the processing takes place entirely within the nucleus. This means that mRNA must complete its processing before it is allowed to leave the nucleus. There may be some kind of a molecular watchman at the gate of the nucleus that allows only processed molecules to leave. Such a system would regulate gene expression by preventing the protein products of a gene from being produced.

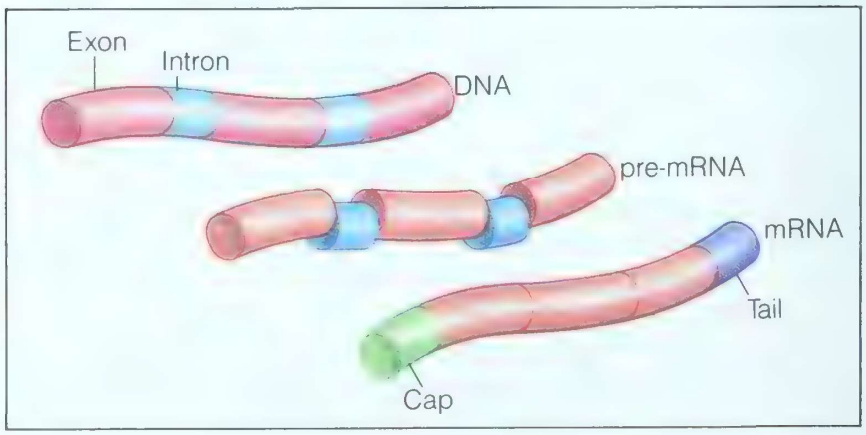
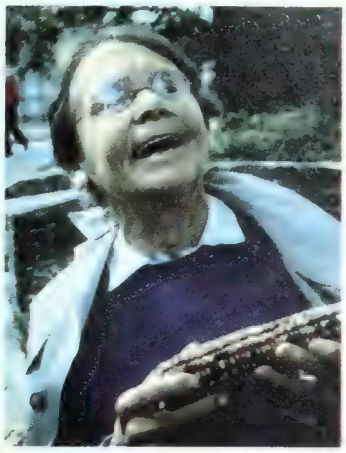


Figure 10–21 *RNA polymerase transcribes an entire gene, introns and exons alike. Before the transcribed RNA, or pre-mRNA, can leave the nucleus and code for protein, the introns must be removed and a chemical "cap" and "tail" must be attached.*

Jumping Genes



In the 1940s a young scientist named Barbara McClintock began to study certain genes in corn. She was particularly interested in why certain kernels of corn developed a spotty, or speckled, appearance. She knew that the color of each seed was genetically determined and that all of the cells in the coating of the seed had the same genetic makeup (genotype). Yet the speckled colors indicated that genes were being suddenly and unpredictably switched on and off in one cell or another. She could find only one reasonable explanation for this observation: There are special controlling elements that can be transposed or move from one place to another around the chromosome. These transposable elements, which are sometimes called jumping genes, can insert themselves into different parts of the chromosome and turn the genes near them on or off.

McClintock's theory of transposable elements was far ahead of its time, and found little support. But McClintock patiently continued her studies, providing experimental proof and documentation that she was right about these moving genes. Finally, in 1975, the techniques of molecular biology had advanced to the point where scientists could actually look for jumping genes. And scientists found jumping genes in bacteria, insects, mammals (including humans), and corn.

Jumping genes may play a role in gene regulation in some organisms, such as corn. They also are involved in the formation of certain proteins that show a great number of variations, such as antibodies—proteins that defend the body against disease-causing organisms. More jumping genes are discovered every day, and it will be a long time before scientists are able to explain all of their different functions.

McClintock has lived to see the importance of her discovery appreciated and is now the recipient of many honors, including the highest prize that can be awarded to a scientist, the Nobel prize (in 1983).

10-3 SECTION REVIEW

1. Compare incomplete dominance and codominance.
2. What is a polygenic trait?
3. Suppose the gene that codes for a repressor was deleted. What effect would this have on the operon?
4. "Mutations in introns are less likely to affect phenotype than mutations in exons." Defend or refute this statement.

LABORATORY INVESTIGATION

MAPPING CHROMOSOMES

PROBLEM

How can rates of crossing-over be used to map chromosomes?

MATERIALS *(per group)*

wooden stick from a frozen-dessert bar
metric ruler
pen
sheet of unlined paper

PROCEDURE

1. Using a pen and a ruler, draw a vertical line 15 cm long in the center of a clean sheet of unlined paper. Make a small horizontal mark at the bottom of the line. Measuring from the bottom of the line, add horizontal marks at 1 cm, 3 cm, 6 cm, 10 cm, and 15 cm.
2. Label each horizontal mark alphabetically (A through F), starting from the bottom.
3. The vertical line on the paper represents a chromosome that has six genes on it—A, B, C, D, E, and F. This chromosome's homologous chromosome is represented by the colored edge of the wooden stick.
4. Adjust the sheet of paper so that its bottom edge is about 15 cm from the edge of your lab table or desk. Move your chair back so that the front edge of your seat is about 30 cm from the edge of your lab table or desk.
5. Toss the wooden stick, underhand, toward the vertical line until the stick lands across the line. The landing of the stick across the line represents crossing-over.
6. When crossing-over occurs, look at the colored edge of the stick to determine which genes have been separated. Make a tally mark in the appropriate place in the data table for each gene that has become separated from gene A. For example, if the colored edge of the stick lands between D and E, make tally marks for genes E and F because they have been separated from gene A as a result of crossing-over.
7. Toss the stick and tally the results until crossing-over has occurred 100 times.
8. Count up the number of tally marks for each of the five genes. In the appropriate place in your data table, record the number of times each gene was separated from gene A.
9. Calculate the frequency of crossing-over by dividing the number of times each gene was separated from gene A by 100. Record the results of your calculations.
10. Calculate the location of each gene by multiplying the frequency of its crossing-over by 15 and rounding off to the nearest integer. Record the calculated gene locations.

OBSERVATIONS

Genes Separated from Gene A	Times Separated from Gene A		Frequency of Crossing-over	Gene Locations	
	Tally	Number		Calculated	Actual
B					1
C					3
D					6
E					10
F					15

1. Did every toss result in crossing-over?
2. Which genes became separated from gene A most frequently? Most infrequently?

ANALYSIS AND CONCLUSIONS

1. What is the relationship between the frequency of gene separation due to crossing-over and the distance between genes?
2. Are your calculated gene locations exactly the same as the actual gene locations? If so, discuss why the experiment went as expected. If not, discuss possible sources of error.
3. How can the frequencies of crossing-over be used to map chromosomes?

STUDENT STUDY GUIDE

SUMMARIZING THE CONCEPTS

The key concepts in each section of this chapter are listed below to help you review the chapter content. Make sure you understand each concept and its relationship to other concepts and to the theme of this chapter.

10–1 The Chromosome Theory of Heredity

- Genes are located on the chromosomes.
- Genes that are located on the same chromosome tend to be inherited together.
- Gene linkages may be broken by crossing-over. The farther apart genes are located on a chromosome, the more likely that they will be separated by a cross-over event.
- Sex-linked genes are located on the sex chromosomes.

10–2 Mutations

- Mutations are changes in the genetic material. There are two kinds of mutations. Chromosomal mutations involve segments of chromosomes, whole chromosomes, or entire sets of chromosomes. Gene mutations involve individual genes.

10–3 Regulation of Gene Expression

- Interactions between genes affect gene expression.
- Genes can be turned on when they are needed and turned off when they are not.
- Many genes contain sequences that are not expressed. These sequences are removed when the RNA transcribed from DNA is made into mRNA.

REVIEWING KEY TERMS

Vocabulary terms are important to your understanding of biology. The key terms listed below are those you should be especially familiar with. Review these terms and their meanings. Then use each term in a complete sentence. If you are not sure of a term's meaning, return to the appropriate section and review its definition.

10–1 The Chromosome Theory of Heredity

chromosome theory of heredity
linked genes
linkage group
recombinant
sex chromosome
autosome
X chromosome
Y chromosome
sex-linked

10–2 Mutations

mutation
chromosomal mutation
nondisjunction
polyploidy
gene mutation
point mutation
frameshift mutation

10–3 Regulation of Gene Expression

incomplete dominance
codominance
polygenic
operon
operator
promoter
inducer
repressor
exon
intron

CHAPTER REVIEW

CONTENT REVIEW

Multiple Choice

Choose the letter of the answer that best completes each statement.

1. An organism has an extra copy of a chromosome in all its cells. This is an example of
   a. polyploidy.
   b. a chromosomal mutation.
   c. a gene mutation.
   d. a somatic mutation.
2. Both alleles are expressed in
   a. codominance.
   b. nondisjunction.
   c. polyploidy.
   d. dominance.
3. The expressed allele does not make up for the inactive allele in
   a. polyploidy.
   b. translocation.
   c. codominance.
   d. incomplete dominance.
4. Which kind of mutation would result if an extra nucleotide were inserted into a gene?
   a. germ
   b. frameshift
   c. chromosomal
   d. deletion
5. Traits that are caused by the interaction of many genes are said to be
   a. polyploid.
   b. linked.
   c. polygenic.
   d. autosomal.
6. The gene product is not produced when the repressor binds to
   a. the operator.
   b. RNA polymerase.
   c. lactose.
   d. the enzyme gene.
7. A point mutation can best be classified as a
   a. chromosomal mutation.
   b. germ mutation.
   c. deletion mutation.
   d. gene mutation.
8. Gene linkages may be broken by
   a. nondisjunction.
   b. duplication.
   c. crossing-over.
   d. polyploidy.

True or False

Determine whether each statement is true or false. If it is true, write "true." If it is false, change the underlined word or words to make the statement true.

1. Crossing-over is more likely to occur between genes that are close together.
2. A somatic mutation affects sperm cells or egg cells and can be inherited.
3. The effects of recessive sex-linked alleles are seen more often in males.
4. Changes in DNA are called mutations.
5. Segments of nonhomologous chromosomes are exchanged in crossing-over.
6. Sex-linked genes are located on the autosomes.
7. Polyploidy is the condition of having extra sets of chromosomes.
8. A point mutation affects a single nucleotide.

Word Relationships

A. *In each of the following sets of terms, three of the terms are related. One term does not belong. Determine the characteristic common to three of the terms and then identify the term that does not belong.*

1. inversion, translocation, duplication, nondisjunction
2. operator, promoter, repressor, inducer
3. dominance, codominance, incomplete dominance, polygenic inheritance
4. gene mutation, point mutation, frameshift mutation, chromosomal mutation

B. *Give the vocabulary word whose meaning is opposite that of the following words.*

5. sex chromosome
6. crossing-over
7. somatic mutation
8. intron

CONCEPT MASTERY

Use your understanding of the concepts developed in the chapter to answer each of the following in a brief paragraph.

1. List and describe the four types of chromosomal mutations that result in a change in the structure of a chromosome.
2. Explain why the operon is turned on when the inducer is present and turned off in the absence of the inducer.
3. Describe two ways genes are regulated in eukaryotes.
4. What is a linkage group? What kind of evidence suggests that linkage groups are the same as chromosomes?

CRITICAL AND CREATIVE THINKING

Discuss each of the following in a brief paragraph.

1. **Making predictions** Would a mutation in an intron affect gene expression? Explain.
2. **Relating concepts** How are crossing-over and linkage related to gene mapping?
3. **Problem solving** Examine the data for the crosses in cats shown below. How are the black and yellow coat colors inherited? (*Hint:* Male cats are XY and female cats are XX.) Using a Punnett square, predict the results of a cross involving a black male and a tortoise-shell female.
4. **Applying concepts** Two of the genes that Mendel studied are located on the same chromosome. Despite this, they show independent assortment in test crosses. What can you conclude about their relative positions on the chromosome?
5. **Using the writing process** Pretend you are the sex-linked gene for colorblindness. Describe the effects you have on each generation as you are passed down through two generations.

Parents	Offspring
black male X yellow female	½ yellow males ½ tortoise-shell (black and yellow) females
yellow male X black female	½ black males ½ tortoise-shell females
yellow male X tortoise-shell female	¼ black males ¼ yellow males ¼ yellow females ¼ tortoise-shell females