Chapter 8

Heredity

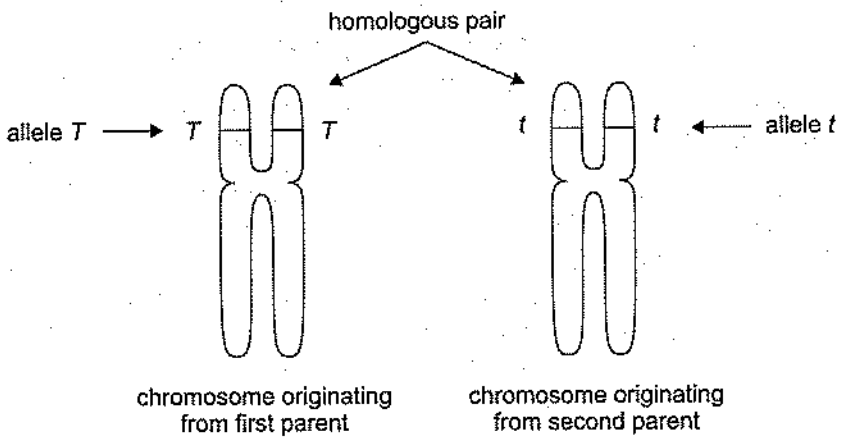
Review

Much of the material in this chapter prepares you for solving genetics problems. A genetics problem is an analysis of the characteristics (traits) of parents and offspring (progeny). Given the traits of one of these generations, you are required to determine the traits of the other generation.

Genetics problems require the application of probability rules. If a coin is tossed, there is a ½ (50%) chance, or probability, that it will be heads. If a coin is tossed again, there is, again, a ½ chance that it will be heads. The first toss does not affect the second toss; that is, the two tosses are independent. To determine the probability of two or more independent events occurring together, you merely multiply the probabilities of each event happening separately. This is the **multiplication rule** of probability. For two consecutive tosses of a coin, the probability of getting two heads is ½ × ½ = ¼. For three tosses, the probability of three heads would be ½ × ½ × ½ = ⅛.

The following terms are used in genetics:

1. A **gene** represents the genetic material on a chromosome that contains the instructions for creating a particular trait. Since the formula for carrying out these instructions is described by a genetic code, a gene is often said to code for a trait. In pea plants, for example, there is a gene that codes for stem length.
2. An **allele** is one of several varieties of a gene. In pea plants, there are two alleles of the gene for stem length—the tall allele, which codes for tall plants, and the dwarf allele, which codes for dwarf plants.
3. A **locus** refers to the location on a chromosome where a gene is located. Every gene has a unique locus on a particular chromosome.
4. **Homologous chromosomes** refer to a pair of chromosomes (a **homologous pair**) that contains the same genetic information, gene for gene. Each parent contributed one of the chromosomes in the pair (Figure 8-1). At any one particular locus, the two genes on a pair of homologous chromosomes (a **gene pair**) might represent two different alleles for that gene because they originated from different parents. For example, the allele for stem length on one pea plant chromosome (inherited from one parent) might code for tall plants, whereas the allele on the homologue of that chromosome (inherited from the second parent) might code for dwarf plants.



Homologous Pair of Chromosomes
Figure 8-1

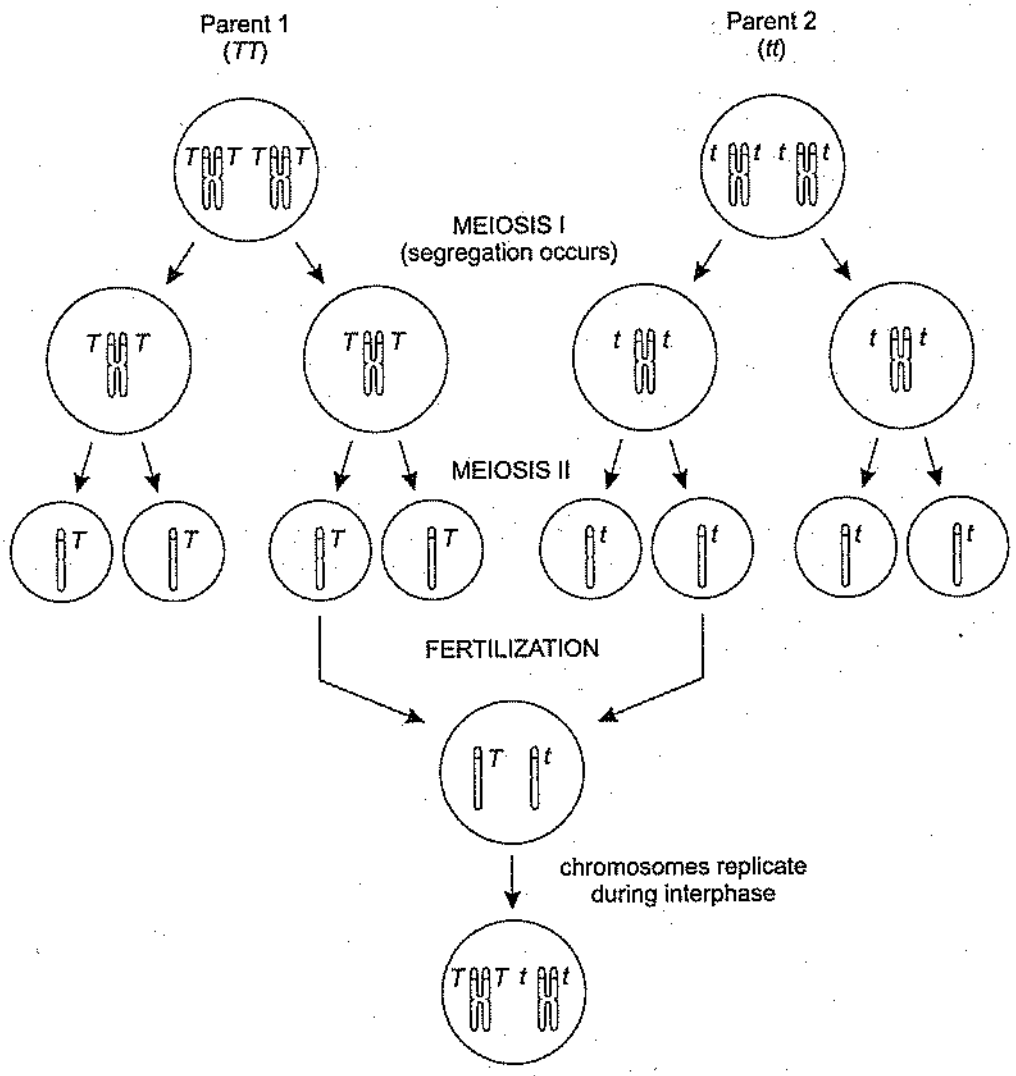
5. If the two alleles inherited for a gene (each on one of the two homologous chromosomes) are different, one allele may be **dominant,** while the other is **recessive.** The trait encoded by the dominant allele is the actual trait expressed. In pea plants, the tall stem allele is dominant and the dwarf allele is recessive. Therefore, if a pea plant inherits one of each of these alleles, only the dominant allele is expressed, producing tall plants. In genetics problems, a dominant allele is often represented by a capital letter, while the recessive allele is represented by the lowercase form of the same letter. In addition, the first letter of the dominant allele is often used to represent the gene. Thus, *T* and *t* represent the dominant (tall) and recessive (dwarf) alleles, respectively, of the same gene. (It is easiest to read these two alleles as "big" *T* and "small" *t*.)
6. **Homozygous dominant** refers to the inheritance of two dominant alleles (*TT*). In this condition, the dominant trait is expressed. In the **homozygous recessive** condition, two recessive alleles are inherited (*tt*) and the recessive trait is expressed. **Heterozygous** refers to the condition where the two inherited alleles are different (*Tt*—it is normal convention to write a pair of alleles with the dominant allele first). In this condition, only the dominant allele is expressed.
7. The **phenotype** is the expression of an allele. Tall stems, blue eyes, and brown hair each represent the phenotype of their respective alleles. On the other hand, the **genotype** represents the actual alleles. For example, *TT* describes the genotype for the homozygous dominant condition. If *T* represents the allele for tall stems and *t* is the allele for dwarf stems, the *genotype Tt* would express the *phenotype* of tall stems. To help you remember their meanings, think *genes* for genotype and *ph*ysical trait for phenotype (although phenotypes might describe physiological or behavioral traits, as well). The term "genotype" is also used to describe just those alleles under discussion (or even all of an individual's alleles), and its phenotype is used to describe the expression of those alleles.

Gregor Mendel, a nineteenth-century monk, is credited with the discovery of the laws of segregation and independent assortment. The following laws describe the separation of chromosomes during meiosis (Figure 8-2). Because the distribution of alleles among gametes is random, the rules of probability can be used to describe how the different chromosomes (and their alleles) in parents assemble in offspring.

1. The **law of segregation** refers to the segregation (separation) of alleles (and their chromosomes) to individual gametes. In other words, one member of each chromosome pair migrates to an opposite pole so that each gamete contains only one copy of each chromosome (and each allele).
2. The **law of independent assortment** refers to the independent assortment of alleles (and their chromosomes). The process is independent because the migration of homologues within one pair of homologous chromosomes to opposite poles does not influence the migration of homologues of other homologous pairs.

Mendel mated, or **crossed,** two varieties of pea plants to form offspring, or **hybrids.** In these kinds of experiments, the **P generation** represents the parents, the **F_1 generation** represents the offspring from the crossing of the parents, and the **F_2 generation** represents the offspring produced from crosses among the F_1. (The letter F stands for *filial,* which refers to sons and daughters.) Two kinds of genetic experiments are common:

1. A **monohybrid cross** is an experiment in which only one trait is being investigated. For example, a cross between a tall pea plant and a dwarf pea plant is a monohybrid cross because it is investigating a gene for only one trait, the gene for stem length.
2. A **dihybrid cross** occurs when two traits are involved. A cross investigating the traits of stem length (tall or dwarf) and flower color (purple or white) is a dihybrid cross. Another example is a cross investigating seed color (yellow, green) and seed texture (round, wrinkled).



Meiosis and Fertilization

Figure 8-2

Complete Dominance, Monohybrid Cross

When traits are expressed as if one allele is dominant to a second allele, the inheritance pattern is called **complete** (or **full) dominance.** Take, for example, a **monohybrid cross,** in which a single trait (originating from a single gene) is examined. For pea plants, let *T,* a dominant allele, represent the allele for tall stems and *t,* the recessive allele, represent the allele for dwarf stems. For inheritance by complete dominance, *TT* and *Tt* produce tall plants, and *tt* produces dwarf plants. Suppose that a plant heterozygous for tall stems (*Tt*) is crossed with a dwarf-stemmed plant (*tt*). The cross can be represented as $Tt \times tt$. Now, what predictions can be made for the genotypes of the offspring?

The first step in analyzing this genetics problem is to determine the genotypes for all possible gametes produced by both parents. Thus, the tall-stemmed parent (genotype *Tt*) produces a gamete with the *T* allele or a gamete with the *t* allele. This is because the genotype *Tt* is represented by a pair of homologous chromosomes, one with the *T* allele and the other with the *t* allele. Each chromosome (with its respective allele) migrates to an opposite pole and ends up in a separate gamete (the law of segregation). Similarly, the dwarf-stemmed parent produces a gamete with the *t* allele and another gamete with the *t* allele.

The next step in the genetic analysis is to determine all the possible ways in which the gametes can combine. This is most easily accomplished by creating a Punnett square (Figure 8-3). (Sometimes, you might see a Punnett square rotated so that it appears in the shape of a diamond; either method can be used.)

	t	*t*
T	*Tt*	*Tt*
t	*tt*	*tt*

Tt × *tt* — P generation

↓

½ *Tt* + ½ *tt* — F_1 generation

Monohybrid Cross (F_1 Generation)
Figure 8-3

In a Punnett square for a monohybrid cross, the gametes from one parent are represented in two spaces at the top of the diagram (*t* and *t* in Figure 8-3). The gametes of the second parent are represented at the left side (*T* and *t* in Figure 8-3). In the middle are four boxes, each box combining the allele found at the top with the allele found to the left. The four boxes represent all of the possibilities of combining the two gametes from one parent with the two gametes from the second parent. In Figure 8-3, the results of the cross *Tt* × *tt* are ½ *Tt* and ½ *tt*. These results represent the genotypic frequencies of the offspring. The phenotypic results are ½ tall and ½ dwarf plants. Results can be given as frequencies (fractions), as percents (½ = 50%), or as ratios. The ratio of dwarf to tall plants is 1:1.

Suppose that you were asked to find the frequencies of the F_2 generation for the cross *TT* × *tt*. Following the procedures above, all of the F_1 progeny would have the genotype *Tt*.

To find the F_2 generation, cross the F_1 offspring with themselves, *Tt* × *Tt* (Figure 8-4). The genotypic results of this cross would produce ¼ *TT*, ½ *Tt*, and ¼ *tt*, while the phenotypic frequencies would be ¾ tall and ¼ dwarf because both *TT* and *Tt* genotypes produce tall plants.

	T	*t*
T	*TT*	*Tt*
t	*Tt*	*tt*

Tt × *tt* — P generation

↓

Tt × *Tt* — F_1 generation

↓

¼ *TT* + ½ *Tt* + ¼ *tt* — F_2 generation

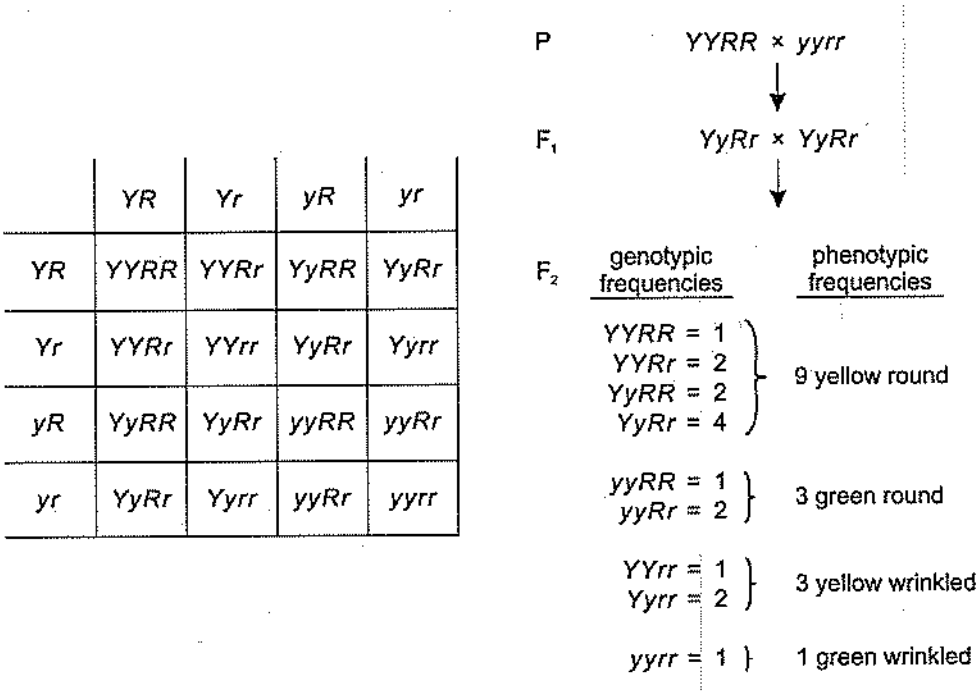
Monohybrid Cross (F_2 Generation)
Figure 8-4

Complete Dominance, Dihybrid Cross

For a **dihybrid cross,** genes for two different traits are observed at the same time. In peas, seed color can be yellow (*Y*) or green (*y*), and seed texture can be round (*R*) or wrinkled (*r*). Thus, *Y* and *y* are used to represent the two alleles (yellow and green) for the gene for seed color, and *R* and *r* (round and wrinkled) are used for the alleles for seed texture. A cross between one pea plant homozygous dominant for both traits and a second plant homozygous recessive for both traits would be given as *YYRR* × *yyrr*.

The first step in analyzing this cross is to determine the alleles of all possible gametes. The *YYRR* plant can produce only one kind of gamete, *YR*. This is determined by the law of segregation; that is, one allele of each allele pair migrates to opposite poles and ends up in separate gametes. Thus, one *Y* and one *R* end up in a single gamete. All segregation possibilities produce gametes that are *YR*. For the second plant, there is also only one kind of gamete produced, *yr*. The next step is to determine the different ways that the gametes from one parent can combine with the gametes from the second parent. Usually you would make a Punnett square for this, but since each parent has only one kind of gamete, you can quickly conclude, without a Punnett square, that all the F_1 offspring will result from the union of a *YR* gamete and a *yr* gamete. The result of that union produces individuals that have the genotype *YyRr*, which bears the phenotype yellow and round.

Now, analyze the F_2 generation produced by *YyRr* × *YyRr*. Each parent can produce four kinds of gametes: *YR*, *Yr*, *yR*, and *yr*. The Punnett square, illustrated in Figure 8-5, shows the gametes of one parent in spaces on the top line and the gametes of the second parent in spaces along the left margin. In the 16 boxes of the



Dihybrid Cross

Figure 8-5

square, alleles from gametes at the top and left are combined. By convention, both alleles from the color gene are arranged together (for example, *Y* and *y*), and both alleles of the texture gene are arranged together (for example, *R* and *r*). The next step is to list each kind of genotype and count the number of times each genotype appears.

This information is shown at the right side of Figure 8-5. The final step is to identify the phenotype of each genotype and to count how many times each phenotype appears. You will see that some phenotypes have more than one genotype. For example, *YYRR, YYRr, YyRR,* and *YyRr* all code for a yellow and round seed. The conclusion is that the F_2 progeny consist of nine plants with yellow and round seeds, three plants with green and round seeds, three plants with yellow and wrinkled seeds, and one plant with green and wrinkled seeds. This ratio, 9:3:3:1, is the same ratio Mendel observed in his experiments for this dihybrid cross.

Test Crosses

Suppose that you wanted to know the genotype of a dwarf pea plant. That would easily be identified as *tt* because dwarf-stemmed plants must have two copies of the recessive allele. Suppose, however, you wanted to know the genotype of a plant with the dominant trait, tall stems. Would it be *TT* or *Tt*? To determine which genotype is correct, you would perform a test cross. A **test cross** is a mating of an individual whose genotype you are trying to determine with an individual whose genotype is known. You will always know the genotype of the individual that expresses the recessive trait (dwarf stems). So, the cross is *T*____ × *tt*. Since you do not know the second allele for the first individual, represent it with an underscore, leaving a blank space for the unknown allele. The next step is to perform both possible crosses, *TT* × *tt* and *Tt* × *tt*. For the first cross, all F_1 will be tall (*Tt*). For the second cross, ½ will be tall (*Tt*) and ½ will be dwarf (*tt*). A farmer would perform a test cross if he or she wanted to know if the tall plant in question was *TT* or *Tt*. If there were any dwarf-stemmed offspring, the farmer could be sure that the tall parent was *Tt*, since you cannot obtain dwarf-stemmed offspring unless both parents contribute the dwarf allele (*t*). In contrast, if all offspring were tall, the farmer could reasonably conclude that the tall parent was *TT*. It is possible, though not likely, that the tall-stemmed parent could be *Tt*, and that, due to chance, no short-stemmed offspring were produced.

A coin toss presents an analogous situation. If you toss a coin six times, the prediction is that half the time it will be tails. However, there is a small chance, 1/64 (½ × ½ × ½ × ½ × ½ × ½), that all six tosses will be tails. As the number of coin tosses increases, though, the probability of obtaining all tails gets smaller and smaller. For the farmer who finds all tall offspring in the test cross, there is a small possibility that the tall parent will be *Tt*. However, the identification of the genotype as *TT* will become more conclusive as the number of offspring observed increases.

Incomplete Dominance

Sometimes, the alleles for a gene do not exhibit the dominant and recessive behaviors discussed above. Instead, the combined expression of two different alleles in the heterozygous condition produces a blending of the individual expressions of the two alleles called **incomplete dominance.** In snapdragons, for example, the heterozygous condition consisting of one allele for red flowers (*R*) and one allele for white flowers (*r*) results in a pink phenotype (*Rr*). Sometimes, both alleles are written with the same uppercase or lowercase letter but with a prime or a superscript or subscript number or letter to differentiate the two. As an example, *R* and *R'* might represent red and white alleles for snapdragons. As another example, H_1 and H_2 might represent straight-hair and curly-hair alleles in humans. The H_1 H_2 phenotype is expressed as an intermediate trait, wavy hair. In still other cases, there may be no apparent rationale, except perhaps historical, for the notation used to indicate different alleles.

Codominance

Another kind of inheritance pattern is termed **codominance.** In this pattern, both inherited alleles are completely expressed. For example, the M and N blood types produce two molecules that appear on the surface of human red blood cells. The *M* (sometimes written as L^M) allele produces a certain blood-cell molecule. The *N* (sometimes written as L^N) allele produces another molecule. Individuals who are *MM* (L^ML^M) produce one kind of molecule; those who are *NN* (L^NL^N) produce a second kind of molecule; and those who are *MN* (L^ML^N) produce both kinds of molecules.

To help you distinguish the three kinds of inheritance, imagine a continuum. At one extreme, there is complete dominance by a dominant allele over a recessive allele. At the other extreme, both alleles are expressed (codominance). Between the two extremes, a blending of two different alleles produces an intermediate phenotype (incomplete dominance).

Multiple Alleles

In the blood group that produces A, B, and O blood types, there are three possible alleles, represented by I^A, I^B, or *i*. Superscripts are used because the two alleles, *A* and *B*, are codominant. A lowercase *i* is used for the third allele because it is recessive when expressed with I^A or I^B. There are six possible genotypes representing all possible combinations of two alleles: I^AI^A and I^Ai (A blood type), I^BI^B and I^Bi (B blood type), I^AI^B (AB blood type), and *ii* (O blood type). The four phenotypes (A, B, AB, and O types) correspond to the presence or absence of an A or B sugar component attached to proteins of the plasma membrane of red blood cells. Thus, the I^AI^A and I^Ai genotypes have proteins with the A sugar attached, the I^BI^B and I^Bi genotypes have proteins with the B sugar attached, and the I^AI^B genotype has both kinds of proteins (half with the A sugar attached and half with the B sugar attached). For the *ii* genotype, neither sugar is attached.

You should be aware of why blood transfusions must be made between individuals of like phenotypes. If an individual with I^BI^B, I^Bi, or *ii* blood is given type A blood, then the immune system of the recipient will identify the A sugar on the introduced red blood cells as a foreign substance. The immune system responds to foreign substances **(antigens)** by producing **antibodies** that attack the antigens. The result is clumping, or **agglutination,** of the blood and possibly death. Individuals with AB type blood can accept any blood type because both A and B sugars are recognized as "self." Also, anyone can accept O type blood because it contains neither A nor B sugars. Thus, a person with O type blood is a universal donor for the ABO blood group (other blood-group types, such as Rh, also need to match).

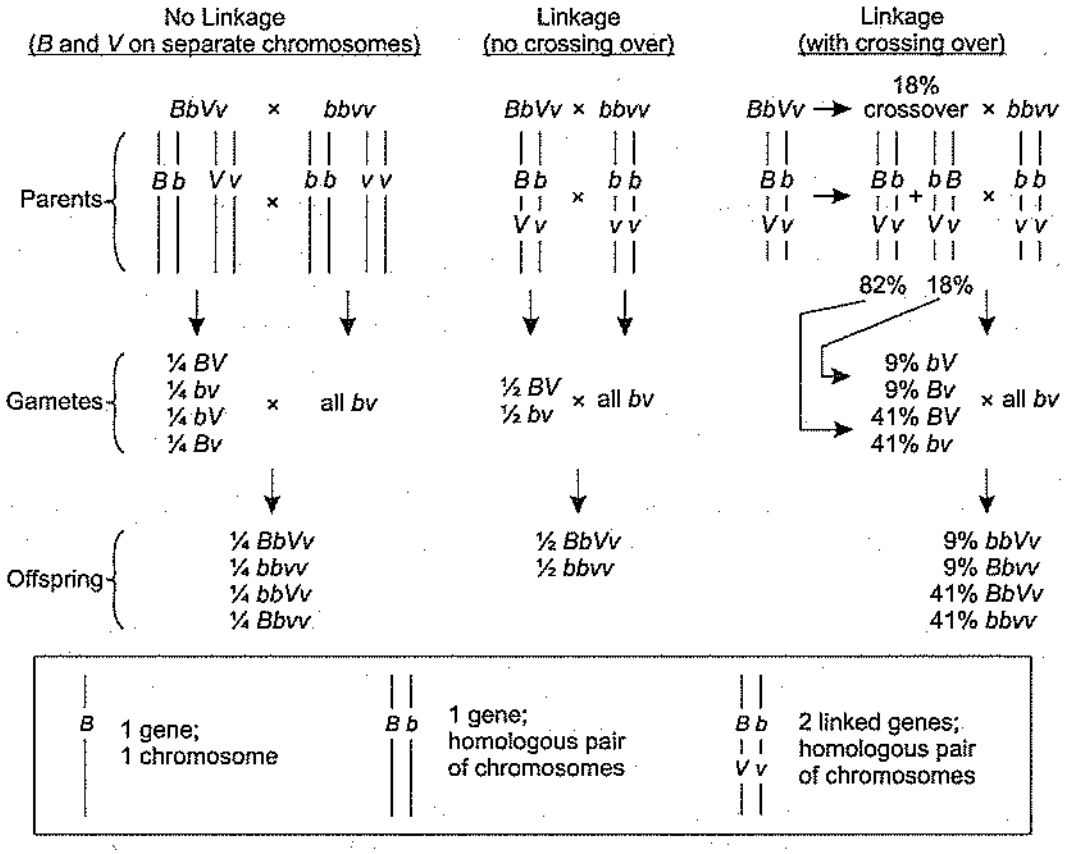
Polygenic Inheritance

Many traits are not expressed in just two or three varieties, such as yellow and green pea seeds or A, B, and O blood types, but as a range of varieties. The heights of humans, for example, are not just short or tall but are displayed as a **continuous variation** from very short to very tall. Continuous variation usually results from **polygenic inheritance,** the interaction of many genes to shape a single phenotype.

Linked Genes and Crossing Over

If two genes are on different chromosomes, such as the seed color and seed texture genes of pea plants, the genes segregate independently of one another (law of independent assortment). **Linked genes** are genes that reside on the *same* chromosome and, thus, cannot segregate independently because they are physically connected. Genes that are linked do not obey the law of independent assortment and are usually inherited together.

In the fruit fly *Drosophila melanogaster,* flies reared in the laboratory occasionally exhibit mutations in their genes. Two such mutations, affecting body color and wing structure, are linked. The normal, or wild, body color is gray (*B*), while the mutant allele is expressed as black (*b*). The second mutation, for wing structure, transforms normal wings (*V*) into vestigial wings (*v*) (small, underdeveloped, and nonfunctional). (Note that for *Drosophila* mutations, the gene notation uses letters that denote the name for the mutation.) Since these two genes are linked, a fly heterozygous for a gray body and normal wings (called gray-normal), indicated by *BbVv,* would have the *BV* on one chromosome and the *bv* on the homologous chromosome. If the linkage between these genes were not known, then the expected results from a cross between this gray-normal fly (*BbVv*) and a black fly with vestigial wings (called black-vestigial, *bbvv*) would be ¼ *BbVv*, ¼ *bbvv*, ¼ *Bbvv*, and ¼ *bbVv* (Figure 8-6). However, since



Dihybrid Cross with Linked Genes and Crossing Over

Figure 8-6

the two genes are on the same chromosome and cannot assort independently, the gray-normal fly produces only two kinds of gametes, *BV* and *bv*. *Bv* and *bV* gametes are not produced. Taking linkage into consideration, the expected offspring would be ½ *BbVv* and ½ *bbvv* (Figure 8-6). If this cross were actually carried out, however, the results would produce a ratio among the four offspring *BbVv*:*bbvv*:*Bbvv*:*bbVv* closer to 41:41:9:9. This is because linked genes cross over (recombine) during prophase I—in this case, about 18% of the time. Instead of 50% of the gametes being *BV* and 50% *bv*, an 18% crossover rate would produce 41% *BV* and 41% *bv* (which sum to 82%) and 9% *Bv* and 9% *bV* (which sum to 18%) (Figure 8-6).

The greater the distance between two genes on a chromosome, the more places between the genes that the chromosome can break and, thus, the more likely the two genes are to cross over during synapsis. As a result, recombination frequencies are used to give a picture of the arrangement of genes on a chromosome. Suppose you knew that for a fly with genotype *BBVVAA* (where *A* is the apterous, or wingless, mutant) the crossover frequency between *B* and *V* was 18%, between *A* and *V* was 12%, and between *B* and *A* was 6%. Since greater recombination frequencies indicate greater distances between genes, *B* and *V* are separated by the greatest distance. Using the frequencies as a direct measure of distance (so that a 1% crossover frequency represents 1 map unit), *B* and *V* are 18 map units apart, *A* and *V* are 12 map units apart, and *B* and *A* are 6 map units apart (Figure 8-7). This suggests that the three genes are arranged in the order *B*-*A*-*V* (or alternatively, *V*-*A*-*B*), with *B* and *A* separated by 6 units and *A* and *V* separated by 12 units. The sum of these two distances, 18 (or 6 + 12), is the map distance between *B* and *V* (other positions for *A* do not preserve the frequencies of *AV* and *AB*). A chromosome map created in this fashion is a **linkage map** and is a portrayal of the sequence of genes on a chromosome. A map portraying the true relative positions of the genes, a **cytological map,** requires additional experimental analyses.

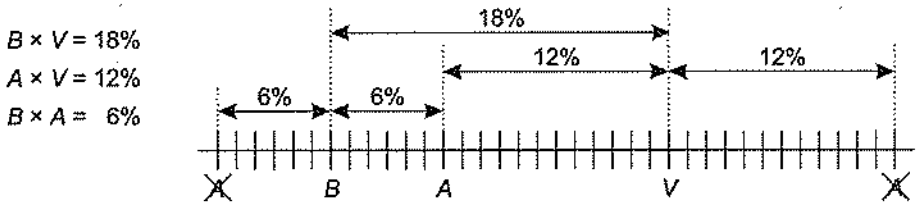


Figure 8-7

Sex Chromosomes and Sex-Linked Inheritance

There is one pair of homologous chromosomes in animals that does not have exactly the same genes. These two chromosomes, the X and Y chromosomes, are called the **sex chromosomes.** All other chromosomes are called **autosomes.** In mammals and fruit flies, inheritance of two X chromosomes (XX) produces a female and inheritance of one X and one Y (XY) produces a male.

Although inheritance of a Y chromosome makes a male, the Y chromosome is small (compared to the X) and has relatively few genes. Some of its genes code for testes and sperm development. One gene in particular, the *SRY* (sex-determining region of Y), regulates gene activities on other chromosomes, which, in turn, stimulate development of male characteristics. As a result, many traits that distinguish the sexes (milk production, male pattern baldness) are **sex limited,** that is, they can only develop in one or the other sex, even though the genes for those traits are on autosomes.

Sex-linked (or **X-linked**) genes are genes that reside on the X, or sex, chromosome. **Y-linked** genes are also possible, but because so few genes reside on the Y chromosome, Y-linkage is rarely encountered. Note the different uses of the word "linkage": Used alone, the word "linkage" refers to two or more genes that reside on the same chromosome; "sex linkage" refers to a single gene residing specifically on a sex chromosome.

You have additional considerations when working with sex-linked genes. When females (XX) inherit a sex-linked gene, they receive two copies of the gene, one on each X chromosome. This situation is similar to that for autosomal inheritance. In contrast, however, a male (XY) will inherit only one copy of the gene because only the X chromosome delivers the gene. There is no similar gene delivered by the Y chromosome. As a result, the allele on the X chromosome of a male is the allele whose trait is expressed, regardless of whether it is dominant or recessive.

Color blindness is caused by a sex-linked, recessive gene (n) in humans. Females and males who inherit the normal allele (N) are X^NX^N and X^NY, respectively, and both are normal. In order for a female to be color blind, she must have two copies of the defective allele (X^nX^n). A male, however, has to inherit only one copy of the defective allele (X^nY) to be color blind. (Note how the Y chromosome is written with no allele superscript because it has none of the genes that are on the X chromosome.) As a result, color blindness, as well as other sex-linked genetic defects, are much more common in males. Heterozygous individuals, who possess a recessive allele for a genetic disorder, do not express the disorder, but they are said to be **carriers** since they can pass the defective allele to their offspring. Thus, heterozygous females, females who are X^NX^n, have normal vision but are carriers.

X-Inactivation

During embryonic development in female mammals, one of the two X chromosomes in each cell does not uncoil into chromatin. Instead, **X-inactivation** occurs, and one chromosome remains coiled as a dark, compact body, called a **Barr body.** Barr bodies are mostly inactive X chromosomes—most of the genes are not expressed, nor do they interact (in a dominant/recessive or codominant manner) with their respective alleles on the X chromosome that is expressed. Thus, only the alleles of the genes on the one active X chromosome are expressed by that cell. When X-inactivation begins, one of the two chromosomes in each embryonic cell randomly and independently becomes inactive. Subsequent daughter cells will have the same X chromosome inactivated as did the embryonic parent cell from which they originated. In the fully developed fetus, then, some groups of cells will have one X chromosome inactivated, while other groups will have the other X chromosome inactivated. Thus, all of the cells in a female mammal are not functionally identical.

A very visible example of X-inactivation can be seen in the different groups of cells producing different patches of color in an individual calico cat. Calico cats have orange, black, and white hair, randomly arranged in patches over their bodies. The orange and black colors are determined by a gene on the X chromosome (the white color is controlled by a different gene). When the X chromosome with the orange allele is inactivated, the black color allele on the active chromosome is expressed, and the hair is black. In other patches, the chromosome with the black allele may be inactivated, and those patches will be orange.

What does this mean for sex-linked genetic disorders in humans, such as color blindness? A carrier female (X^NX^n) should usually be normal with respect to this trait, because all or at least some cells will have X^N activated and produce normal functioning color vision. However, in the very unlikely case that all of the relevant cells have X^N inactivated, the carrier female should express the same symptoms of color blindness as a male.

Nondisjunction

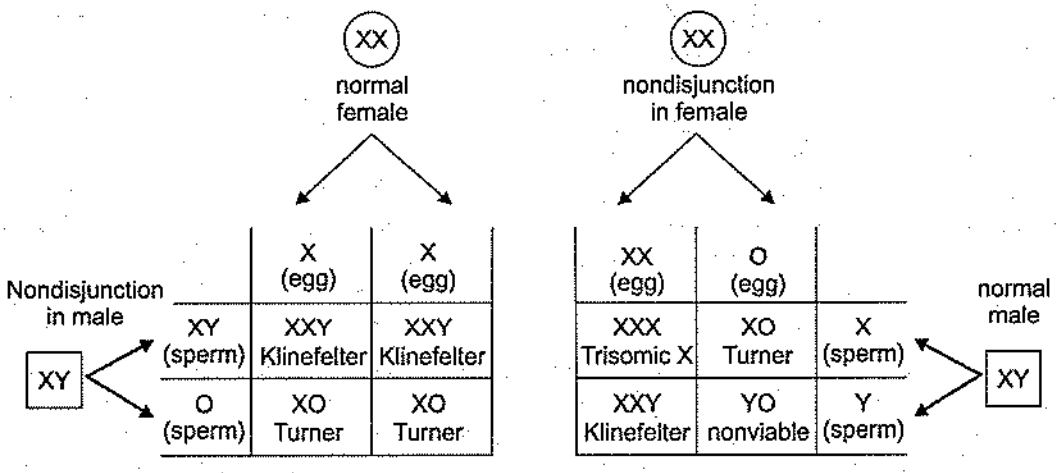
Nondisjunction is the failure of one or more chromosome pairs or chromatids of a single chromosome to properly move to opposite poles during meiosis or mitosis. Some examples of nondisjunction follow:

1. During meiosis, the failure of two homologous chromosomes (during anaphase I) or two chromatids of a single chromosome (during anaphase II) to separate produces gametes with extra or missing chromosomes.
2. During mitosis, the failure of two chromatids of a single chromosome (during anaphase) to separate produces daughter cells with extra or missing chromosomes. This happens most often during embryonic development and results in **mosaicism,** in which a fraction of the body cells, those descendent of a cell where nondisjunction occurs, have an extra or missing chromosome.
3. **Polyploidy** occurs if all of the chromosomes undergo meiotic nondisjunction and produce gametes with twice the number of chromosomes. If a polyploid gamete is fertilized with a similar gamete, then a polyploidy zygote and individual can form. Polyploidy is common in plants.

Human Genetic Disorders

Some of the causes of genetic disorders follow:

1. **Point mutations** occur when a single nucleotide in the DNA of a gene is incorrect. This can occur if a different nucleotide is substituted for the correct one **(substitution),** if a nucleotide base-pair is omitted **(deletion),** or if an extra base-pair is inserted **(insertion).** Most point mutations have deleterious effects on gene function. Two examples follow:
   - **Sickle-cell disease,** caused by a nucleotide substitution, results in the production of defective hemoglobin, the oxygen-carrying protein in red blood cells. The defective hemoglobin molecule causes the red blood cell, usually circular, to become sickle shaped when low-oxygen conditions occur (high altitudes, strenuous exercise). In response, red blood cells do not flow through capillaries freely and oxygen is not adequately delivered throughout the body (anemia). For individuals who are homozygous for the defective allele (sickle-cell *disease*), inadequate oxygen supplies can lead to organ damage, bone abnormalities, and impaired mental functioning. Heterozygous individuals (sickle-cell *trait*) are generally without symptoms, as the normal allele is sufficient to produce adequate amounts of normal hemoglobin.
   - **Tay-Sachs disease,** usually caused by a nucleotide insertion, results when lysosomes lack the functional enzyme to break down certain fats (glycolipids). When these fats accumulate in the nerve cells of the brain, brain cells die and death usually follows early in childhood.
2. **Aneuploidy** is a genome with extra or missing chromosomes. It is most often caused by nondisjunction. Although most aneuploid gametes do not produce viable offspring, some zygotes, with certain chromosome imbalances, survive. These almost always lead to genetic disorders. Three examples follow:
   - **Down syndrome** occurs when an egg or sperm with an extra number 21 chromosome fuses with a normal gamete. The result is a zygote with three copies of chromosome 21 **(trisomy 21).** Down syndrome individuals bear various abnormalities, including mental retardation, heart defects, respiratory problems, and deformities in external features.
   - **Turner syndrome** results when there is nondisjunction of the sex chromosomes. Sperm will have either both sex chromosomes (XY) or no sex chromosomes (O, used to indicate the absence of a chromosome). Similarly, eggs will be either XX or O. When a normal X egg or sperm combines with an O sperm or egg, a Turner syndrome zygote, **XO,** results. XO individuals are sterile females with physical abnormalities. Although the absence of a single chromosome is usually fatal, missing the Y chromosome, with its few, male-necessary genes, is not nearly so deleterious (Figure 8-8).
   - **Klinefelter syndrome** occurs when an XY or XX gamete, produced as a result of nondisjunction (as in Turner syndrome), combines with a normal X gamete to produce an **XXY** individual. Because of the presence of a Y chromosome, these individuals are male but may be sterile. In addition, individuals may mildly express a variety of female secondary sex characteristics (for example, reduced facial and chest hair and breast development). Although the extra X chromosome is mostly inactive (forming a Barr body), some activity apparently remains. An XXX individual can result when an XX egg combines with a normal X sperm, but these trisomic X females are usually without serious disorders, though they are often tall and may have some learning disabilities (Figure 8-8).



Aneuploidy of Sex Chromosomes
Figure 8-8

3. **Chromosomal aberrations** are caused when chromosome segments are changed.
   - **Duplications** occur when a chromosome segment is repeated on the same chromosome. For example, **Huntington's disease** is caused by the insertion of multiple repeats of three nucleotides. The mutant gene codes for a defective enzyme, which results in the death of nerve cells in the brain.
   - **Inversions** occur when chromosome segments are rearranged in reverse orientation on the same chromosome. Individuals with inversions usually do not express any abnormalities as long as the inversion does not introduce any duplications or deletions.
   - **Translocations** occur when a segment of a chromosome is moved to another chromosome. For example, Down syndrome ordinarily occurs when an individual inherits an extra chromosome 21 (as a result of nondisjunction). However, Down syndrome can also occur after a translocation of a chromosome segment from chromosome 21 to chromosome 14. An individual inheriting a chromosome with a 14/21 translocation has a normal number of chromosomes but inherits three copies of a segment from chromosome 21 (two chromosomes 21 and a chromosome 14 with a segment of chromosome 21). The result has the same phenotypic effect as trisomy 21.

Environmental Influences on Phenotypic Expression

If you have ever compared "identical" twins, you know that, though very similar, they are not identical. Although their DNA may be identical, the expression of their genes is influenced by environmental factors. Before identical twins are born, they may experience slightly difference environments in the uterus, and these differences lead to different phenotypic expressions of height, weight, and fingerprints. Some examples of environmental factors that influence gene expression follow:

- Nutrition can heavily influence physical development in both animals and plants. Insufficient calcium or other dietary deficiencies may result in short stature, while plants growing in soils that lack adequate amounts of nitrogen may not flower or may flower and produce smaller-than-normal fruits.
- Nutrition may also influence the expression of genetic disorders. For example, individuals with phenylketonuria cannot metabolize the amino acid phenylalanine. As a result, phenylalanine accumulates, brain cells die, and death follows. However, if dietary phenylalanine is reduced to minimal levels, quantities of phenylalanine in the body remain safe. Similarly, individuals who are lactose intolerant and unable to break down lactose can avoid symptoms of nausea, gas, and diarrhea by omitting dairy products containing lactose from their diet.

- Temperature influences sex determination in various reptiles. Eggs incubated at lower temperatures become males; those at higher temperatures become females.
- Temperature often influences the color of animal fur. The genes for fur color in Siamese cats and Himalayan rabbits produce a dark fur pigment in cold areas of the animal (feet, ears, face, and tail) and a light fur pigment in the remaining warm areas.
- Seasonal changes in daylight length influence the expression of hair color from brown (in summer) to white (in winter, for camouflage) in the snowshoe hare. Similarly, an increased exposure to UV radiation in humans stimulates a corresponding increase in melanin, the skin-darkening pigment.
- Soil pH influences flower color in certain species of *Hydrangea*. Flower color is blue when the soil pH is acidic, pink when the soil pH is basic.
- Expression of one individual's genes is sometimes dependent upon chemicals in the environment that are produced by other individuals. This kind of chemical signaling between organisms is often required to elicit mating. For example, when nutrient availability is scarce, slime bacteria secrete signaling molecules that stimulate nearby bacteria to aggregate, form a multicellular collective, grow into a fruiting body, and produce spores. Similarly, certain yeast cells only mate with yeast cells of an opposite mating type. To signal its presence, a yeast cell secretes a pheromone (a signaling molecule), to which only yeasts of the opposite mating type respond.

Non-Nuclear Inheritance

Mitochondria and chloroplasts carry within their own DNA genes that are responsible for some of their metabolic processes. Mitochondria assort randomly during cell division and, in most animals, are inherited only from the mother, as the male gamete (sperm) delivers very little cytoplasm. Similarly, in many plants, both mitochondria and chloroplasts are inherited through the female gamete (ovules), as the male gamete (pollen) does not usually carry these organelles. Where it occurs, this **maternal inheritance** can be used to trace a specific genome from progeny back through multiple generations to its original mother. One example of maternal inheritance in humans is a genetic mutation that impairs oxidative phosphorylation, reduces ATP output, and results in nerve deterioration. Because the mutation occurs on a mitochondrial gene, the trait can only be inherited from mothers who possess the defective mitochondria.

Review Questions

Multiple-Choice Questions

The questions that follow provide a review of the material presented in this chapter. Use them to evaluate how well you understand the terms, concepts, and processes presented. Actual AP multiple-choice questions are often more general, covering a broad range of concepts, and often more lengthy. For multiple-choice questions typical of the exam, take the two practice exams in this book.

Directions: Each of the following questions or statements is followed by four possible answers or sentence completions. Choose the one best answer or sentence completion.

1. If you roll a pair of dice, what is the probability that they will both turn up a three?

A. $\frac{1}{2}$
B. $\frac{1}{8}$
C. $\frac{1}{16}$
D. $\frac{1}{36}$

2. Which of the following best expresses the concept of the word *allele*?

A. genes for wrinkled and yellow
B. genes for wrinkled and round
C. the expression of a gene
D. phenotypes

3. The ability to taste a chemical called PTC is inherited as an autosomal dominant allele. What is the probability that children can taste PTC if they are descended from parents who are both heterozygous for this trait?

A. 0
B. ¼
C. ½
D. ¾

4. In fruit flies, dumpy wings are shorter and broader than normal wings. The allele for normal wings (*D*) is dominant to the allele for dumpy wings (*d*). Two normal-winged flies mated and produced 300 normal-winged and 100 dumpy-winged flies. The parents were probably

A. *DD* and *DD*
B. *DD* and *Dd*
C. *Dd* and *Dd*
D. *Dd* and *dd*

5. Which of the following is true of the gametes produced by an individual with genotype *Dd*?

A. ¼ *D* + ½ *Dd* + ¼ *D*
B. ½ *D* and ½ *d*
C. ½ *Dd* and ½ *dD*
D. all *Dd*

6. Suppose that in sheep, a dominant allele (*B*) produces black hair and a recessive allele (*b*) produces white hair. If you saw a black sheep, you would be able to identify

A. its phenotype for hair color
B. its genotype for hair color
C. the genotypes for only one of its parents
D. the phenotypes for both of its parents

Questions 7–9 refer to the following key and description of fruit fly traits. Each answer in the key may be used once, more than once, or not at all.

A. 0
B. 1/16
C. 9/16
D. 1

In fruit flies, the gene for curved wings (*c*) and the gene for spineless bristles (*s*) are on different chromosomes. The respective wild-type alleles for each of these genes produce normal wings and normal bristles.

7. From the cross *CCSS* × *ccss*, what is the probability of having an offspring that is *CcSs*?

8. From the cross *CcSs* × *CcSs*, what is the probability of having an offspring that is *ccss*?

9. From the cross *CcSs* × *CcSs*, what is the probability of having an offspring that is normal for both traits?

Questions 10–11 refer to the following.

In snapdragons, the allele for tall plants (T) is dominant to the allele for dwarf plants (t), and the allele for red flowers (R) is codominant with the allele for white flowers (R'). The heterozygous condition for flower color is pink (RR').

10. If a dwarf red snapdragon is crossed with a white snapdragon homozygous for tall, what are the probable genotypes and phenotypes of the F1 generation?

A. all $TtRR'$ (tall and pink)
B. all $TtRR$ (tall and red)
C. all $TtR'R'$ (tall and white)
D. all $ttRR$ (dwarf and red)

11. If $ttRR'$ is crossed with $TtRR$, what would be the probable frequency for offspring that are dwarf and white?

A. 0
B. ¼
C. ½
D. ¾

12. For the cross $AABBCCDd \times AAbbCcDd$, what is the probability that an offspring will be $AABbCcDd$?

A. 1/16
B. ⅛
C. ¼
D. ½

13. The inheritance of skin color in humans is an example of which of the following?

A. X-linked inheritance
B. codominance
C. polygenic inheritance
D. gene linkage

14. Red-headed people frequently have freckles. This is best explained by which of the following?

A. The genes for these two traits are linked on the same chromosome.
B. The genes for these two traits are sex-linked.
C. Alleles for these two traits are codominant.
D. Both parents have red hair and freckles.

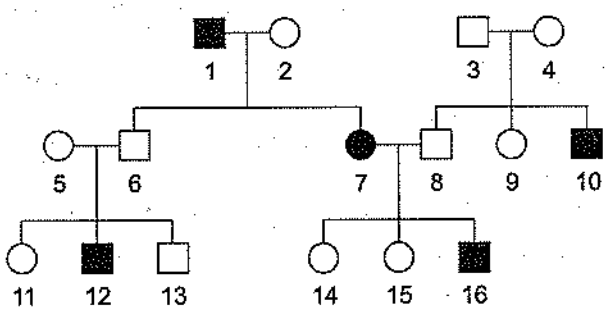
15. Let A and a represent two alleles for one gene and B and b represent two alleles for a second gene. If for a particular individual, A and B were on one chromosome and a and b were on a second chromosome, then all of the following are true EXCEPT:

A. The two genes are linked.
B. The two chromosomes are homologous.
C. All gametes would be either AB or ab.
D. The genotype of this individual is $AaBb$.

16. Four genes, *J*, *K*, *L*, and *M*, reside on the same chromosome. Given that the crossover frequency between *K* and *J* is 3, between *K* and *L* is 8, between *J* and *M* is 12, and between *L* and *M* is 7, what is the order of the genes on the chromosome?

A. *J K L M*
B. *J K M L*
C. *K J L M*
D. *K J M L*

Questions 17–19 refer to the following pedigree. Circles indicate females, and squares indicate males. A horizontal line connecting a male and a female indicates that these two individuals produced offspring. Offspring are indicated by a descending vertical line that branches to the offspring. A filled circle or filled square indicates that the individual has a particular trait, in this case, red-green color blindness. Color blindness is inherited as a sex-linked, recessive allele.



Use the following key for the next three questions. Each answer in the key may be used once, more than once, or not at all.

A. X^NX^N
B. X^NX^n
C. X^NY
D. X^nY

17. Identify the genotype for individual 10.

18. Identify the genotype for individual 5.

19. Identify the genotype for individual 14.

20. In domestic cats, two alleles of a sex-linked (X-linked) gene code for hair color. One allele codes for orange hair, and the other allele codes for black hair. Cats can be all orange or all black, or they can be calico, a coat characterized by randomly arranged patches of orange and black hair. With respect to this gene, all of the following are true EXCEPT:

A. A black female and an orange male can produce a black male cat.
B. A black female and an orange male can produce a female calico cat.
C. A calico female and an orange male can produce a female calico cat.
D. A calico female and an orange male can produce a male calico cat.

21. From which parent(s) did a male with red-green color blindness inherit the defective allele?

A. *only* the mother
B. *only* the father
C. the mother or the father, but not both
D. *both* the mother and the father

22. A human genetic disorder that is caused by nondisjunction of the sex chromosomes is

A. sickle-cell disease
B. hemophilia
C. Down syndrome
D. Turner syndrome

23. Two genes, *A* and *B*, are linked. An individual who is *AaBb* produces equal numbers of four gametes: *AB*, *Ab*, *aB*, and *ab*. The best explanation for this would be that

A. nondisjunction occurred
B. the genes are on nonhomologous chromosomes
C. the two genes are close together on the same chromosome
D. the two genes are separated by a large distance on the same chromosome

24. In peas, the gene for seed color (yellow or green) and flower color (colored or white) are located on the same chromosome. Find the crossover frequency if a cross between a plant heterozygous for both traits and a plant homozygous recessive for both traits produces the following progeny.

yellow and colored	green and white	yellow and white	green and colored
652	683	77	88

A. 5.5%
B. 11%
C. 15%
D. 22%

25. For the cross *AaBbCCddEE* × *AaBbCcDdEE*, what is the probability that an offspring will be *aaBBCCddEE*?

A. $1/16$
B. $1/32$
C. $1/64$
D. $1/128$

Free-Response Questions

The AP exam has long and short free-response questions. The long questions have considerable descriptive information that may include tables, graphs, or figures. The short questions are brief but may also include figures. Both kinds of questions have four parts and generally require that you bring together concepts from multiple areas of biology.

The questions that follow are designed to further your understanding of the concepts presented in this chapter. Unlike the free-response questions on the exam, they are narrowly focused on the material in this chapter. For free-response questions typical of the exam, take the two practice exams in this book.

Directions: The best way to prepare for the AP exam is to write out your answers as if you were taking the exam. Use complete sentences for all your answers. But do *not* use outline form or bullets. You may use diagrams to supplement your answers, but be sure to describe the importance or relevance of your diagrams.

1. Color blindness is inherited as an X-linked recessive allele. Explain why males have a greater chance of expressing the color-blindness trait.

2. A form of vitamin D resistance is inherited as an X-linked dominant allele. Explain why females have a greater chance of inheriting and expressing the vitamin D–resistant trait.

3. The height of an individual can be any value over a wide range, from short to tall. Describe a mechanism that can produce such continuous variation.

4. Describe Mendel's laws of segregation and independent assortment with respect to
   a. genes that are not linked
   b. genes that are linked
   c. crossing over
   d. sex-linkage
   e. Down syndrome
   f. Turner syndrome

Answers and Explanations

Multiple-Choice Questions

1. **D.** The chance that one die will turn up a three is 1 in 6, or $\frac{1}{6}$. For both dice to turn up a three, the probability is determined by multiplying the probability of each event happening independently, or $\frac{1}{6} \times \frac{1}{6} = \frac{1}{36}$.

2. **B.** Alleles refer to the various forms of a gene, or the various forms in which a particular gene can be expressed. Wrinkled and round are alleles that refer to two forms of a single gene (at a particular locus) that code for seed texture. Answer choice A is incorrect because wrinkled and yellow refer to two different genes (at different loci), one for seed texture and the other for seed color.

3. **D.** If you let *T* represent the dominant allele for the ability to taste PTC, then the cross would be *Tt* × *Tt*. The Punnett square that follows shows that ¾ of the offspring have ability to taste PTC (¼ *TT* + ½ *Tt*).

	T	*t*
T	*TT*	*Tt*
t	*Tt*	*tt*

4. **C.** If both parents have normal wings (*DD* or *Dd*), there are three possible parent crosses: *DD* × *DD*, *DD* × *Dd*, or *Dd* × *Dd*. All of the progeny of *DD* × *DD* and of *DD* × *Dd* would have normal wings. Only the progeny of *Dd* × *Dd* would consist of ¾ normal-winged flies.

DD × *DD*

	D	*D*
D	*DD*	*DD*
D	*DD*	*DD*

↓

All *DD*

All normal wings

DD × *Dd*

	D	*D*
D	*DD*	*DD*
d	*Dd*	*Dd*

↓

½ *DD* + ½ *Dd*

All normal wings

Dd × *Dd*

	D	*d*
D	*DD*	*Dd*
d	*Dd*	*dd*

↓

¼ *DD* + ½ *Dd* + ¼*dd*

¾ normal wings + ¼ vestigial wings

5. **B.** At the end of meiosis I, the two homologous chromosomes, one with *D* and one with *d*, would separate and migrate to opposite poles, which will form separate cells. During meiosis II, each chromosome separates into two chromatids (both of which will have exactly the same allele, assuming no crossing over), which migrate to opposite poles and become separate gametes. Thus, the cell containing the *D* chromosome will produce two gametes, each with a *D* chromosome (previously a chromatid). Similarly, the cell containing the *d* chromosome will produce two gametes, each with a *d* chromosome. At the end of meiosis II, then, there will be two gametes with a *D* chromosome and two gametes with a *d* chromosome.

6. **A.** Black is the phenotype of the sheep. That is given to you in the question. Without further information, you cannot identify the genotype of a black sheep because it could be either *BB* or *Bb*. The possible genotypes of the parents of a black sheep could be *BB* × *BB*, *BB* × *Bb*, *BB* × *bb*, or *Bb* × *Bb*. Thus, there is no one single genotype for either parent. Answer choice D is incorrect because although one parent would always be black, you cannot be certain whether the second parent is black or white.

7. **D.** In the *CCSS* × *ccss* cross, *CCSS* produces only *CS* gametes, and *ccss* produces only *cs* gametes. Thus, all offspring are *CcSs*.

8. **B.** The cross of *CcSs* × *CcSs* is the same kind of cross illustrated in Figure 8-5. Among the 16 genotypes in the Punnett square, only one would be *ccss*.

9. **C.** The cross of *CcSs* × *CcSs* is the same kind of cross illustrated in Figure 8-5. Among the 16 genotypes in the Punnett square, 9 would be normal for both traits (1 of *CCSS*, 2 of *CCSs*, 2 of *CcSS*, and 4 of *CcSs*).

10. **A.** The question involves the progeny of the cross *ttRR* × *TTR′R′*. Since *ttRR* produces only *tR* gametes, and *TTR′R′* produces only *TR′* gametes, all progeny will be *TtRR′*. The *Tt* genotype codes for tall, and *RR′* codes for pink.

11. **A.** This is a trick question because there's a long way and a very short way to solve this problem. The long way would be to construct a Punnett square and sort and count all the offspring. The short, easy way is to recognize that a white flower has the genotype *R′R′*. Looking at only the color gene for each parent, the cross is *RR′* × *RR*. In order for a cross to produce a white-flowered offspring (*R′R′*), both parents must contribute *R′*. Since this is not the case, no offspring will be white flowered.

12. **C.** It is not usually practical to make a Punnett square for genotypes involving more than two genes. In this problem, you are asked about the frequency of one specific offspring, *AABbCcDd*. To solve this problem, look at each gene separately. Looking at the first gene, the parents are *AA* × *AA* and all offspring will be *AA* (frequency of 1). For the second gene, *BB* × *bb*, all offspring will be *Bb* (1). For the third gene, the parents are *CC* × *Cc*, which produces ½ *CC* and ½ *Cc* (do a Punnett square to confirm this). Finally, a cross of the fourth gene, *Dd* × *Dd*, produces ¼ *DD*, ½ *Dd*, and ¼ *dd*. To find the probability of *AABbCcDd*, first find the frequency of each gene separately. The probability of *AA* is 1, of *Bb* is 1, of *Cc* is ½, and of *Dd* is ½. Then find the product of these frequencies. For *AABbCcDd*, the product is 1 × 1 × ½ × ½ = ¼.

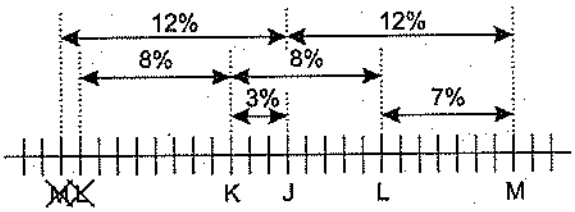
13. **C.** Since the range of skin colors in humans shows continuous variation from very pale to very dark, it is most likely coded by many genes (polygenic inheritance).

14. **A.** When two traits frequently occur together, then they are probably linked. Sometimes, a red-headed person may not have freckles, or a freckled person may not have red hair. In these cases, there was probably a crossover event, exchanging one of the two genes with an allele that did not code for freckles or red hair.

15. **C.** Answer choice C is the false statement. Since *A* and *B* are on one chromosome, then, by definition, they are linked and answer choice A is true. Answer choice B is also true because the homologous chromosome would have the same genes but with possibly different alleles. In this case, both chromosomes carry the same genes, but the alleles are different (*A* and *B* on one chromosome and *a* and *b* on its homologue). Taking

both chromosomes together, you get the genotype *AaBb,* and, thus, answer choice D is true. Answer choice C is false because even though the chromosomes separate during meiosis to produce gametes that are *AB* and *ab,* crossing over can take place, producing some gametes that are *Ab* and some that are *aB.*

16. C. Begin by drawing a horizontal line with about 30 tick marks. Since *K* and *J* have a crossover frequency of 3, write the letters *K* and *J* on two marks 3 ticks apart, near the middle. Next, add the letter *L* in two positions, 8 units to the right of *K* and 8 units to the left of *K.* At this point of the solution, both positions are possible. Next, add the letter *M* 12 units to the right of *J* and, again, 12 units to the left of *J.* Both are possible. Last, use the *L-M* frequency to determine which configuration of *M* and *L* is correct. Since the *L-M* frequency is 7, only the *M* and *L* positions at the right are correct. That leaves only one possible sequence, *K-J-L-M.*



17. D. In any pedigree problem, you should begin by first identifying genotypes for which there is only one possibility. For a sex-linked recessive pattern of inheritance, you can identify the genotypes of all males and of all females that express the trait that the pedigree is describing. In this case, color-blind males are X^nY (filled boxes, 1, 10, 12, 16), normal males are X^NY (open boxes, 3, 6, 8, 13), and color-blind females are X^nX^n (filled circle, 7).

18. B. The next step in this pedigree problem is to identify the normal females (open circles). Are they X^NX^N or X^NX^n? Note that box 12 is a color-blind son (X^nY). Since a son can inherit only his Y chromosome from his father (box 6), box 12 must have inherited his X^n gene from his mother (circle 5). Thus, you can conclude that the mother, circle 5, is X^NX^n.

19. B. There are two possibilities for female 14, X^NX^N or X^NX^n. The color-blind mother, 7, is X^nX^n. The normal father, 8, is X^NY. A cross between these two individuals can produce only one kind of daughter, X^NX^n (confirm this with a Punnett square). For practice, you should try to identify every female. Female 7 is color blind (X^nX^n), females 2, 4, 5, 14, and 15 are carriers (X^NX^n), and females 9 and 11 are either normal (X^NX^N) or carriers (X^NX^n). (There is not enough information to determine which genotype is correct.)

20. D. To be a calico, a cat must have two X chromosomes, one with the orange allele and one with the black allele. Since a male cat has only one X chromosome, it normally can be only orange or black.

21. A. Since red-green color blindness is inherited as a sex-linked recessive allele, a color-blind male must be X^nY. Because he is a male, he received the *Y* from his father. Therefore, he inherited the X^n from his mother.

22. D. Turner syndrome is caused by the nondisjunction of the sex chromosomes. The result is a sperm or egg that is missing a sex chromosome. The formation of a zygote from the union of one of these sperm or eggs with a normal egg or sperm (with an X chromosome) results in Turner syndrome (XO). It is also possible to form an OY zygote, but because this zygote is missing an X chromosome, a chromosome with many essential genes, the zygote is nonviable. Answer choice C, Down syndrome, is also caused by nondisjunction, but of chromosome 21, not the sex chromosomes. Sickle-cell disease is inherited as an autosomal recessive allele, while hemophilia is inherited as a sex-linked recessive allele.

23. D. Since the genes *A* and *B* are on the same chromosome (linked), there are two possible allele arrangements for an *AaBb* individual. The first is that *AB* is on one chromosome and *ab* is on the homologous chromosome. The second possibility is that *Ab* is on one chromosome and *aB* is on the homologous

chromosome. Using the first possibility as an example, one would expect only two kinds of gametes in the absence of crossing over—*AB* and *ab* in equal quantities. Crossing over would produce gametes that are *Ab* and *aB*. If the two genes are very close together, there would be very few crossovers because there are few places between the genes where the chromosomes can break and cross over. If the genes are far apart, there would be many crossovers because there are many places for chromosome breaks. When the genes are very far apart, they cross over so frequently that by observing the allele frequencies of the gametes the genes seem to assort independently, as if they were on different chromosomes (not linked). That is exactly what has happened in this question. The observed frequencies are those that would have been expected had the genes been on different chromosomes. Since the question states that the genes are linked, they must be far apart to allow so large a number of crossovers.

24. **B.** Without crossing over, the homozygous recessive parent can only produce one gamete genotype and the heterozygous parent can only produce two gamete genotypes (see Figure 8-6). Without crossing over, then, only two kinds of progeny phenotypes are produced, and they would be produced in equal numbers. These would be the progeny in the table with the largest numbers, 652 and 683. The smaller numbers, then, represent the progeny that are produced as a result of crossing over. The total number of crossover progeny produced is 77 + 88, or 165. Since the total number of progeny is 77 + 88 + 652 + 683, or 1,500, the crossover frequency is 165 ÷ 1,500, or 0.11 (11%).

25. **C.** First, find the probabilities for each gene:

$Aa \times Aa \rightarrow \frac{1}{4}\ AA + \frac{1}{2}\ Aa + \frac{1}{4}\ aa$

$Bb \times Bb \rightarrow \frac{1}{4}\ BB + \frac{1}{2}\ Bb + \frac{1}{4}\ bb$

$CC \times Cc \rightarrow \frac{1}{2}\ CC + \frac{1}{2}\ Cc$

$dd \times Dd \rightarrow \frac{1}{2}\ Dd + \frac{1}{2}\ dd$

$EE \times EE \rightarrow 1\ EE$

Then multiply the probabilities for each gene in the offspring: $aa \times BB \times CC \times dd \times EE = \frac{1}{4} \times \frac{1}{4} \times \frac{1}{2} \times \frac{1}{2} \times 1 = \frac{1}{64}$.

Free-Response Questions

1. Because males inherit only one X chromosome, they express all the genes on that chromosome, regardless of whether the alleles are dominant or recessive. In contrast, females inherit two X chromosomes and both alleles for a gene must be recessive in order for a female to express a recessive trait.

2. Because females have two X chromosomes, they have two chances of inheriting a trait on that chromosome and they need only one copy of a dominant allele to express the trait.

3. A trait that is produced by the expression of many interacting genes (polygenic inheritance) generates continuous variation in a population. Also, multiple alleles of a single gene can recombine to produce many genotypic and phenotypic combinations.

4. **a.** When chromosomes align (on the metaphase plate) during meiosis I, homologous chromosomes are paired. Each homologue migrates to a separate pole and becomes a member of a separate gamete. The migration to separate poles is random—that is, either chromosome of a homologous pair can migrate to either pole (Mendel's law of segregation).

Different homologous pairs of chromosomes act independently of other homologous chromosome pairs. Thus, genes that are on different chromosomes (unlinked) migrate independently of genes on other chromosomes (Mendel's law of independent assortment).

b. When two genes are linked, they are on the same chromosome. If they are on the same chromosome, they migrate together to either pole (unless crossing over occurs). Thus, they violate Mendel's law of independent assortment and are inherited together as if they were a single gene in a monohybrid cross. If the dominant alleles *A* and *B* are on one chromosome and the recessive alleles *a* and *b* are on the homologous chromosome, they produce only two kinds of gametes, *AB* and *ab*. Then, the dihybrid cross *AaBb* × *AaBb* would produce a 1:2:1 genotypic ratio for *AABB, AaBb,* and *aabb* with a phenotypic ratio of 3:1, not the typical 9:3:3:1 phenotypic ratio that Mendel found when using unlinked genes. The 1:2:1 and 3:1 genotypic and phenotypic ratios are those expected from a typical monohybrid cross.

c. Crossing over occurs between linked genes. Instead of producing only two kinds of gametes—say, *AB* and *ab*—exchanges occur between homologous chromosomes, producing some *Ab* and *aB* gametes, quantities of which depend on the frequency of crossing over. The frequency of crossing over increases as the distance between the gene loci increases.

d. Sex-linkage occurs when a gene is located on one of the sex chromosomes, usually the X chromosome. For example, in humans, hemophilia is inherited as a recessive allele on the X chromosome. Females receive two copies of the gene, one on each of their X chromosomes. If they receive two recessive alleles, they are hemophiliacs. If they receive two normal alleles, they are normal, but if they inherit one normal and one hemophilia allele, they will have normal clotting abilities (because the normal allele is dominant) but will be carriers of the disease. Males, on the other hand, inherit only one X chromosome and, thus, only one copy of the allele. If they receive the normal allele, they will have normal clotting; if they receive the hemophilia allele, they will be hemophiliacs. Because they need only one copy of the allele to express the trait, sex-linked diseases are more common in males than in females.

e. Down syndrome occurs as a result of the nondisjunction of the two number 21 chromosomes. As a result, the homologous pair does not separate and move to opposite poles (as the law of segregation implies), but rather both chromosomes end up at the same pole and in the same gamete. Two kinds of gametes are formed, one with two copies of chromosome 21 and one with no chromosome 21. Only the gamete with two copies of the chromosome is viable. The zygote formed between this gamete and a normal gamete will have three copies of chromosome 21, and the infant will express the Down syndrome phenotype, which consists of physical abnormalities and mental retardation.

f. Turner syndrome results from a nondisjunction of the sex chromosomes. This results in a gamete that has either two sex chromosomes or no sex chromosomes. If a gamete with no sex chromosomes (O) fuses with a normal gamete bearing the X chromosome, the resulting zygote will have only a single X chromosome (XO) and express the Turner syndrome phenotype. Turner syndrome individuals are female and exhibit physical abnormalities, including sterility.