CHAPTER 9

Patterns of Inheritance

Objectives

Introduction Explain how studies of dog breeding can provide insight into principles of genetics.

Mendel's Principles

9.1 Describe the pangenesis theory and blending hypothesis. Explain why both ideas are now rejected.

9.2 Explain why Mendel's decision to work with peas was a good decision. Define and distinguish between true-breeding organisms, hybrids, the P generation, the F_1 generation, and the F_2 generation.

9.3 Define and distinguish between the following pairs of terms: genotype vs. phenotype, dominant allele vs. recessive allele, and heterozygous vs. homozygous. Also define a monohybrid cross and a Punnett square.

9.3 Explain how Mendel's principle of segregation describes the inheritance of a single characteristic.

9.4 Describe the relationship between alleles for the same gene on separate homologous chromosomes.

9.5 Explain how Mendel's principle of independent assortment applies to a dihybrid cross. Illustrate this principle with examples of Mendel's work with peas and recent research on Labrador retrievers.

9.6 Explain how a testcross is performed to determine the genotype of an organism.

9.7 Explain when the rule of addition and the rule of multiplication should be used to determine the probability of an event. Explain why Mendel was wise to use large sample sizes in his studies.

9.8 Explain how family pedigrees can help determine the inheritance of many human traits.

9.9 Explain how recessive and dominant disorders are inherited. Provide examples of each.

9.10 Compare the health risks, advantages, and disadvantages of the following forms of fetal testing: amniocentesis, chorionic villus sampling, and ultrasound imaging.

Variations on Mendel's Principles

9.11–9.14 Describe the inheritance patterns of incomplete dominance, multiple alleles, and pleiotropy.

9.15 Define and distinguish between carrier testing, diagnostic testing, prenatal testing, newborn screening, and predictive testing.

9.16 Explain how a single characteristic can be influenced by many genes.

The Chromosomal Basis of Inheritance

9.17 Define the chromosome theory of inheritance. Explain the chromosomal basis of the principles of segregation and independent assortment.

9.18 Explain how linked genes are inherited differently from other, nonlinked genes.

9.19–9.20 Describe T. H. Morgan's studies of crossing over and explain how Sturtevant created linkage maps.

Sex Chromosomes and Sex-Linked Genes

9.21 Explain how sex is genetically determined in humans and the significance of the *SRY* gene. Explain how sex is determined differently in other organisms.

9.22–9.23 Describe the patterns of sex-linked inheritance, noting examples in fruit flies and humans.

Key Terms

genetics
self-fertilize
cross-fertilize
hybrid
cross
P generation
F_1 generation
F_2 generation
monohybrid cross
allele
dominant allele
recessive allele
homozygous
heterozygous
Punnett square
phenotype
genotype
principle of segregation
dihybrid cross
principle of independent assortment
testcross
rule of multiplication
rule of addition
carrier
cystic fibrosis
achondroplasia
Huntington's disease
amniocentesis
chorionic villus sampling (CVS)
ultrasound imaging
incomplete dominance
ABO blood groups
codominance
pleiotropy
polygenic inheritance
chromosome theory of inheritance
linked genes
recombination frequency
sex chromosome
monoecious
hermaphroditic
sex-linked gene
red-green color blindness
hemophilia
Duchenne muscular dystrophy

Word Roots

-centesis = a puncture (*amniocentesis:* a technique for determining genetic abnormalities in a fetus by the presence of certain chemicals or defective fetal cells in the amniotic fluid, obtained by aspiration from a needle inserted into the uterus)

co- = together (*codominance:* phenotype in which both dominant alleles are expressed in the heterozygote)

di- = two (*dihybrid cross:* a breeding experiment in which parental varieties differing in two traits are mated)

pleio- = more (*pleiotropy:* when a single gene impacts more than one characteristic)

poly- = many; **gen-** = produce (*polygenic:* an additive effect of two or more gene loci on a single phenotypic character)

Lecture Outline

Introduction *Purebreds and Mutts—A Difference of Heredity*

A. Close observations of breeding organisms and their offspring (Labrador puppies) show patterns in the inheritance of characteristics that can be predicted (chapter-opening

photo; Figure 9.0A). However, inheritance patterns can be rather unpredictable, as indicated in the photo of the mongrel pups (Figure 9.0B).

B. Behavior can be partially explained by genetic inheritance, but the environment in which an organism lives also influences behavior.

C. We will see that patterns of inheritance can be explained by the behavior of chromosomes during meiosis and fertilization.

I. Mendel's Principles

Module 9.1 The science of **genetics** has ancient roots.

A. The ancient Greeks believed in pangenesis, the idea that particles governing the inheritance of each characteristic collect in eggs and sperm and are passed on to the next generation.

B. But many, including Aristotle, realized there were problems with this idea: The potential to produce characteristics is inherited, not pieces of the characteristics themselves. Reproductive cells are not changed by the development or activity of other cells.

C. Based on artificial breeding, nineteenth-century observers believed in the "blending" hypothesis, in which characteristics from both parents blend in the offspring.

D. *Preview:* Not only did plant and animal breeders provide data for hypotheses concerning inheritance, but this information greatly influenced the ideas of Charles Darwin and Alfred Wallace, at about the same time (Chapter 13).

Module 9.2 Experimental genetics began in an abbey garden.

A. Mendel was university trained in precise experimental technique (Figure 9.2A). He studied peas because they offered advantages over other organisms. Peas grow easily, have relatively short life spans (one year), and have numerous and distinct characteristics (Figure 9.2D), and the mating of individuals can be controlled so that the parentage of offspring can be known for certain.
NOTE: This is a place to talk about the fact that good biological experimentation often results from the choice of suitable study organisms that enable the experimenter to focus on particular questions.

B. Mendel's paper, published 1866, argued that there are discrete, heritable factors (what we call genes) that retain their individuality when transmitted from generation to generation.
NOTE: Like Barbara McClintock's work on transposons (Module 12.13), Mendel's work was not appreciated under much later.

C. Mendel could intentionally **self-fertilize** a flower by covering it with a bag, or **cross-fertilize** two different plants by dusting the carpels of one with the pollen of another (Figure 9.2C).
NOTE: The life history of flowering plants, for our purposes here, is similar to that of most animals, with male and female gamete-producing organs found in flowers (Figure 9.2B).

D. By continuous self-fertilization for many generations, Mendel developed breeds of plants that bred true (continued to show a characteristic when self-fertilized) for each of the characteristics he followed. He found seven characteristics, each of which came in two distinct forms (Figure 9.2D).

E. Mendel developed two principles based on two types of experiments. In one type (monohybrid crosses), he hybridized true-breeding plants for each of the two forms of a characteristic. In a second type (dihybrid and trihybrid crosses), he hybridized plants that combined two or more of the seven characteristics.

F. In these experiments, the true-breeding parents are the **P (parental)** generation, their hybrid offspring is the **F_1 (first filial) generation,** and the offspring of mating two F_1 individuals is the **F_2 (second filial) generation.**

Module 9.3 Mendel's principle of segregation describes the inheritance of a single characteristic.

Review: Point out that principle of segregation is a reflection of the events of meiosis (Module 8.14).

A. Principle of segregation: Pairs of genes segregate (separate) during gamete formation; the fusion of gametes at fertilization pairs genes once again.

B. Mendel conducted a **monohybrid cross** with flower color (Figure 9.3A). The results of this experiment were: out of 929 F_2 offspring, 705 were purple, and 224 were white.
NOTE: The proportions are not exactly ¾ and ¼ because mating involves probabilities. See below.

C. Mendel observed that each of the seven characteristics exhibited the same inheritance pattern.

D. Mendel developed four hypotheses:

1. There are alternative forms of genes, the units that determine heritable characteristics. These alternative forms are called **alleles.**
2. For each inherited characteristic, an organism has two genes, one from each parent. They may be the same allele or different alleles.
3. A sperm or egg carries only one allele for each characteristic because the allele pairs segregate from each other during gamete production.
4. When the two alleles are different, the one that is fully expressed is said to be **dominant** and the one that is not noticeably expressed is said to be **recessive.**

E. Conventions for alleles: *P,* the dominant (purple) allele, and *p,* the recessive (white) allele. *P* generation: *PP* x *pp;* their gametes: *P* and *p;* F_1 generation: *Pp.*

F. Homozygous dominant, homozygous recessive, and heterozygous refer to the **genotypes** (the nature of the genes as inferred from observations and knowledge of how the system works). The **phenotypes** are what we see.

G. The **Punnett square** is used to keep track of the gametes (two sides of the square) and offspring (cells within the square) (Figure 9.3B).
NOTE: This pattern, whereby each gamete contains a single copy of each gene, is stated by Mendel's **principle of segregation** and is based on the events of meiosis (anaphase I and anaphase II; Module 8.14).

Module 9.4 Homologous chromosomes bear the two alleles for each characteristic.

A. *Review:* Homologous pairs (Module 8.12).

B. Although Mendel knew nothing about chromosomes, our knowledge of chromosome arrangements (in homologous pairs) strongly supports the principle of segregation.

C. Alleles of a gene reside at the same locus on homologous chromosomes.
NOTE: One of the chromosomes illustrated was inherited from the female parent, the other from the male parent (Figure 9.4).

Module 9.5 The principle of independent assortment is revealed by tracking two characteristics at once.

Review: Point out that the **principle of independent assortment** is a reflection of the events of meiosis (Module 8.14).

A. Principle of independent assortment: Each pair of alleles segregates independently during gamete formation.

B. Experimental procedure: Breed two strains true, each exhibiting one of the two forms of two characteristics (in the example used, round yellow seeded plants *[RRYY]* and wrinkled green-seeded plants *[rryy]*). Hybridize these two strains as the P generation, resulting in hybrid offspring (F_1: *RrYy*). Then allow the F_1 to self-fertilize (*RrYy* x *RrYy*).
NOTE: Each of these individuals produces the same four gametes: *RY, Ry, rY,* and *ry.* Taking one gamete from each individual means that there are $4^2 = 16$ possible gametic combinations.

C. Two hypotheses: The characteristics are inherited either dependently or independently of each other (Figure 9.5A).

D. Results. The F_1 generation exhibits only the dominant phenotype (this is expected). The F_2 generation exhibits a phenotypic ratio of 9:3:3:1 (round yellow: round green: wrinkled yellow: wrinkled green).
NOTE: 9 + 3 + 3 + 1 = 16, the same as the number of possible gametic combinations. That the phenotypic ratio adds up to the number of possible gametic combinations serves as a check of the results of a cross.

E. Use a Punnett square to analyze these results, with the sides of the square representing the male and female gametes possible if alleles of two characteristics segregate independently. Notice that the genotypes that produce the same phenotype are not all the same (Figure 9.5A).

F. Fur color and vision defects (PRA) in Labradors follow this pattern of assortment if pure strains of black labs and chocolate labs are used as the P generation. *B* allele, black fur; *b* allele, brown fur; *N* allele, normal vision; *n* allele, blind. (See Figure 9.5B.) If two Labs of genotype *BbNn* are bred, the phenotypic ratio will follow the expected ratio from the example with peas, 9:3:3:1. Four dogs will be blind; one of which is a chocolate Lab *(bbnn).*

Module 9.6 Geneticists use the testcross to determine unknown genotypes.

A. A **testcross** involves crossing an unknown genotype expressing the dominant phenotype with the recessive phenotype (by necessity, homozygous).

B. Each of two possible genotypes (homozygous or heterozygous) gives a different phenotypic ratio in the F_1 generation. Homozygous dominant gives all dominant. Heterozygous gives half recessive, half dominant (Figure 9.6).
NOTE: This technique uses phenotypic results to determine genotypes.

Module 9.7 Mendel's principles reflect the rules of probability.

A. Events that follow probability rules are independent events; that is, one such event does not influence the outcome of a later such event. If you flip a coin four times and get four heads, the probability for tails on the next flip is still ½.

B. The probability of two events occurring together is the product of the probabilities of the two events occurring apart (the **rule of multiplication**).

C. Thus, when studying how the alleles of two (or more) genes that segregate independently behave, use the probabilities of how they behave individually.
NOTE: The probability of a recessive phenotype occurring in a monohybrid cross is 1 out of 4 (¼) (Figure 9.7). The probability of two recessives occurring together in a dihybrid cross is ¼ × ¼, or 1 out of 16 (recall 9 + 3 + 3 + 1 = 16). In a trihybrid cross, as mentioned, the probability of a triple recessive is 1 out of 64.

D. If there is more than one way an outcome can occur, these probabilities must be added, as in the case of determining the chances for heterozygous mixtures (the **rule of addition**).

Module 9.8 Connection: Genetic traits in humans can be tracked through family pedigrees.

A. Point out the commonly used, symbolic conventions on the pedigree chart, showing the appearance of congenital deafness in a Martha's Vineyard family (Figure 9.8).

B. By applying Mendel's principles, one can deduce the information on the chart from the pattern of phenotypes.

C. Assuming that Jonathan Lambert inherited his deafness from his parents, the only explanation is that his deafness is caused by a recessive allele because neither of his parents was deaf. Because some of his children were deaf, his wife, Elizabeth Eddy, must have been a **carrier.** From this it follows that all their hearing children were carriers.

D. This final deduction shows the power of applying Mendelian principles to pedigrees and how to make predictions.
NOTE: Since the pattern in the pedigree is not tied to gender, the gene for congenital deafness is not sex-linked.

Module 9.9 Connection: Many inherited disorders in humans are controlled by a single gene.

A. Over 1000 known genetic traits are attributable to a single gene locus and show simple Mendelian patterns of inheritance (Table 9.9).

B. Many human characteristics are thought to be determined by simple dominant-recessive inheritance, and sometimes the ratio of dominant-to-recessive phenotype exhibits a Mendelian ratio (Figure 9.9A).

C. Most disorders are caused by recessive alleles and vary in the severity of the expressed trait.
NOTE: The terms *dominant* and *recessive* refer only to whether or not a characteristic is expressed in the heterozygote, not to whether it is the most common.

D. The vast majority of people afflicted with recessive disorders are born to normal, heterozygous parents (Figure 9.9A).
NOTE: It is really only the distribution of phenotypes in the offspring of one couple of known phenotype or genotype that will follow Mendelian principles.

E. **Cystic fibrosis** is the most common lethal genetic disease in the U.S.

F. Most genetic diseases of this sort are not evenly distributed across all racial and cultural groups because of the prior and existing reproductive isolation of various populations.

G. Laws forbidding inbreeding may have arisen from observations that such marriages more often resulted in miscarriages, stillbirths, and birth defects. On the other hand, there is a debate over this issue because seriously detrimental alleles would likely be eliminated from populations when expressed in the homozygous embryo, and there are societies where inbreeding occurs without detrimental results.

H. Some disorders are caused by dominant alleles. These disorders vary in how deadly they are. Some are nonlethal handicaps, some are lethal in the homozygous condition, and some are intermediate in severity.

I. **Achondroplasia,** a type of dwarfism, is lethal in the homozygous condition; individuals who express the trait are heterozygous.

J. Other conditions attributable to dominant alleles are lethal only in older adults, so the allele can be passed to children before it is realized that the parent has the condition.

Preview: These diseases either are, or in the future may be, treatable by use of the techniques of genetic engineering (Module 12.19).

K. With practice, the principles and techniques outlined above can be used to determine many interesting things about the genotypes of individuals. This information, in turn, can be used to predict future characteristics in offspring.
Preview: In large populations, the prevalence of dominant and recessive characteristics may depend on whether one or the other allele confers advantages or disadvantages on those who have it. Population genetics will be discussed in Chapter 13.

Module 9.10 Connection: Fetal testing can spot many inherited disorders early in pregnancy.

A. **Amniocentesis** involves taking a sample of the amniotic fluid that bathes the fetus, at 14–16 weeks. This fluid contains living fetal cells (from the skin and the mouth cavity) and can be karyotyped. Some chemical tests can be performed on the fluid itself (Figure 9.10A).

B. **Chorionic villus sampling** involves removing tissue from the fetal side of the placenta nurturing a fetus, at 8–10 weeks. These cells are rapidly dividing and can be immediately karyotyped. Some biochemical tests can be performed (Figure 9.10B).

C. **Ultrasound imaging** of the fetus provides a noninvasive view inside the womb (Figure 9.10C, D).

D. Fetoscopy provides a more direct view of the fetus through a needle-thin viewing scope inserted into the uterus.

E. Amniocentesis, chorionic villus sampling, and fetoscopy carry a small risk and are reserved for situations with higher probabilities of disorders (for example, older parents or situations where genetic counseling has uncovered a higher risk).

F. Analysis of the mother's blood can detect abnormal levels of certain hormones (HCG and estriol) or proteins produced by the fetus (alpha-fetoprotein). Abnormal levels may indicate that the fetus has Down syndrome or a neural tube defect.

G. If fetal testing suggests that there is a problem that cannot be helped by routine surgery or other therapy, the difficult choice must be made between terminating a pregnancy by abortion or carrying a defective baby to term.

II. Variations on Mendel's Principles

Module 9.11 The relationship of genotype to phenotype is rarely simple.

A. The inheritance of many characteristics among all eukaryotes follows the principles that Mendel discovered.
NOTE: Discussing these principles first has allowed us to focus on the conventions and basic functioning of the system that underlies inheritance patterns.

B. However, most characteristics are inherited in ways that follow more complex patterns.

C. Before looking at the chromosomal explanation of Mendel's principle of independent assortment, we will look at four such complex patterns: incomplete dominance, multiple alleles at a gene locus, pleiotropy, and polygenic inheritance.

D. These patterns are extensions of Mendel's principles, not exceptions to them.

Module 9.12 Incomplete dominance results in intermediate phenotypes.

A. **Incomplete dominance** describes the situation where one allele is not completely dominant in the heterozygote; the heterozygote usually exhibits characteristics intermediate between both homozygous conditions.

B. Snapdragon color is a good example of how this works. Note that the possibilities of each genotype are the same as in a case of complete dominance, but the phenotypic ratios are different (Figure 9.12A).

C. Another example: the inheritance of alleles that relate to hypercholesterolemia. Normal individuals, *HH,* have normal amounts of LDL receptor proteins; *hh* individuals (rare in the population, about 1 in 1 million) have no receptors and five times the amount of blood cholesterol; *Hh* individuals (1 in 500) have half the number of receptors and twice the amount of blood cholesterol (Figure 9.12B).
Preview: Lifestyle can also lead to hypercholesterolemia (Module 21.20).

D. *Preview:* In this last example, the relative numbers of each phenotype in the population depend on the manner in which genes are inherited in populations, the subject of Chapter 13.

Module 9.13 Many genes have more than two alleles in the population.

A. The **ABO blood groups** in humans follow this pattern, in which individuals can have two alleles from a set of three possible alleles.

B. These blood-type alleles code for two carbohydrates (or the absence of any carbohydrate) on the surface of red blood cells (a total of three alleles). There are six possible genotypes and four possible phenotypes.

C. When blood is transfused, recipients develop antibodies (discussed further in Chapter 24) for the types of carbohydrate on the donor red blood cells that the recipients lack.

D. Type O (universal donor) has neither carbohydrate and can receive no other type. Type AB (universal recipient) has both carbohydrates and can receive any type. Type A has carbohydrate A and can receive A or O. Type B has carbohydrate B and can receive B or O (Figure 9.13). The AB blood type is an example of **codominance.**

E. Blood types can be used to disprove or suggest parentage in paternity suits; however, more sophisticated blood analyses are needed to prove parentage.

Module 9.14 A single gene may affect many phenotypic characteristics.

A. This common situation is known as **pleiotropy.**

B. An example is the inheritance of an allele that codes for abnormal hemoglobin and, in the homozygous condition, causes sickle-cell disease.
Preview: The allelic variant that is responsible for sickle-cell disease is discussed in Module 10.16.

C. The sickle shape of the red blood cells confers a whole suite of symptoms on homozygous individuals, attributable to three underlying difficulties resulting from the abnormal cell shape (Figure 9.14).

D. The normal and abnormal alleles are another good example of codominance, so heterozygous individuals (carriers) can exhibit some symptoms, although normally they are healthy.

E. The incidence of the allele is relatively high in individuals of African descent (one in 10 African Americans is heterozygous), because sickle-cell carriers are somewhat protected from malaria, a protozoan-caused disease prevalent in tropical regions.
Preview: This is an example of the action of natural selection (Chapter 13).
NOTE: Another good example of pleiotropy is Marfan's syndrome, which results in long, loose-jointed limbs, cardiovascular defects, and several other problems.

Module 9.15 Connection: Genetic testing can detect disease-causing alleles.

A. The field of genetic testing (genetic screening) has expanded dramatically in the last decade. There are four major categories of genetic testing.

B. Carrier testing: The purpose of this analysis is to determine if a potential parent will pass on a harmful trait to offspring.

C. Diagnostic testing: This type of genetic analysis is used to confirm or rule out the existence of a genetic disorder. This procedure can be used on the unborn (prenatal testing) as well as after birth (particularly adults).

D. Newborn screening: This type of genetic screening is designed to catch childhood disease that can be treated effectively when detected early.

E. Predictive testing: This type of testing is designed to identify a person predisposed to certain disorders such as colon cancer (FAP) or breast cancer (BRCA1 or BRCA2).

F. Ethical, moral, and medical issues are being raised by the increased use of genetic testing. Insurability of persons with detected genetic disorders is also an issue of concern.

Module 9.16 A single characteristic may be influenced by many genes.

A. This situation is known as **polygenic inheritance.**

B. Skin pigmentation is just such a phenotypic character whose underlying genetics has not been completely determined. Figure 9.16 is hypothetical, showing the phenotypic outcome of mixtures of three genes, each with two alleles coding for "additive units," which produce the overall characteristic.
NOTE: Point out to your students how much easier it is to solve genetic problems such as these using probabilities instead of Punnett squares.

III. The Chromosomal Basis of Inheritance

Module 9.17 Chromosome behavior accounts for Mendel's principles.

A. While the existence and behavior of chromosomes was not appreciated by Mendel himself, the significance of his work was understood later, in the late 1800s and early 1900s. Out of this understanding came the **chromosome theory of inheritance.**

B. We have already seen that the fact that there are homologous pairs of chromosomes accounts for the principle of segregation.

C. The fact that there are several sets of homologous pairs of chromosomes accounts for the principle of independent assortment (Figure 9.17).
NOTE: Mendel's seven garden pea characteristics all sorted independently of each other because the genes governing each characteristic are all on separate chromosomes.

Module 9.18 Genes on the same chromosome tend to be inherited together.

A. **Linked genes** are located close together on the same chromosome.

B. The inheritance of such genes does not follow the pattern described by the principle of independent assortment because the two genes are normally inherited together on adjoining portions of the same chromosome.

C. The phenotypic ratios of such dihybrid crosses approach that of a monohybrid cross (3:1), rather than the typical pattern of the dihybrid cross (9:3:3:1) (Figure 9.18).

Module 9.19 Crossing over produces new combinations of alleles.

A. In the case of many linked genes (for example, pollen grain length and flower color in sweet peas), there are some offspring of a dihybrid cross that do involve independent assortment of the genes.

B. These situations of recombination are accounted for by crossing over, which occurs between the genes on homologous pairs of the same chromosome during some meiotic divisions, but not all (Figure 9.19A, B).
Review: Crossing over and genetic recombination are discussed in Module 8.18.

C. Early examples of recombination were demonstrated in fruit flies (Figure 9.19B) by embryologist T. H. Morgan and colleagues in the early 1900s.

D. The percentage of recombinant offspring is called the **recombination frequency** (Figure 9.19C).

Module 9.20 Geneticists use crossover data to map genes.

A. The study of fruit-fly genetics resulted in considerable additional understanding of genetic principles.
NOTE: This is another example of the use of an experimental organism that lends itself to study. Fruit flies have many phenotypic characters, are easily raised and bred in captivity, and have a short life cycle. In addition, they have only four chromosomes (simplifying the situation), and these chromosomes can be easily visualized in nondividing cells in the salivary glands.

B. A. H. Sturtevant, one of Morgan's colleagues, developed a technique of using crossover data to map the locations of genes on chromosomes on which they were linked.

C. Sturtevant assumed that the rate of recombination is proportional to the distance between two genes on a chromosome (Figure 9.20B) and this information can be used to construct a genetic map (Figure 9.20C).

IV. Sex Chromosomes and Sex-Linked Genes

Module 9.21 Chromosomes determine sex in many species.

A. **Sex chromosomes** in humans are nonidentical members of a homologous pair.

B. In humans, *XX* individuals are female, and *XY* are male (Figure 9.21A).

C. A crucial role in the human sex determination is played by the *SRY* (sex-determining region of the *Y* chromosome) gene. This gene initiates the development of testes. An individual who does not have a functioning *SRY* gene develops ovaries.

D. In other species, other patterns of sex chromosomes exist (Figure 9.21B, C).

E. In some species, chromosome number rather than chromosome type determine sex (Figure 9.21D). In some invertebrates, diploid individuals are female and haploid are male.
NOTE: In sea turtles, for example, the temperature at which the fertilized eggs are incubated determines sea turtles' sex.

F. Plants that produce both eggs and sperm are said to be **monoecious.** Animals that produce both eggs and sperm are **hermaphroditic.**

Module 9.22 Sex-linked genes exhibit a unique pattern of inheritance.

A. Sex chromosomes contain genes specifying sex and other genes for characteristics unrelated to sex. These genes are said to be **sex-linked.**

B. Because of linkage and location, the inheritance of these characteristics follows peculiar patterns.

C. Examples are given using eye color in fruit flies (*X*-linked recessive for white eyes; Figure 9.22A). Depending on the genotypes of the parents, three patterns emerge (Figure 9.22B, C, D).

D. In humans, most sex-linked characteristics result from genes on the *X* chromosome.
Preview: Thus, mostly males are affected (Module 9.23).
NOTE: Other sex-related patterns of inheritance include sex-influenced genes, sex-limited genes, genome imprinting, and mitochondrial inheritance. Pattern baldness is an example of a sex-influenced trait; the allele for pattern baldness behaves as a recessive in females and a dominant in males (its expression requires sufficient testosterone). A sex-limited gene is one that can be expressed in only one sex or the other; for example, some testicular tumors are the result of inheriting a particular allelic variant (obviously, testicular tumors cannot be expressed in females). In genome imprinting the same DNA sequence is expressed differently based on whether it was inherited from the female or male parent. For example, if an individual is missing a particular segment of paternal chromosome 15, the result is Prader-Willi syndrome; if the same segment is missing from maternal chromosome 15, the result is Angelman syndrome. Mitochondrial genes are all inherited from the female parent.

Module 9.23 Connection: Sex-linked disorders affect mostly males.

A. Examples of such characteristics are red-green color blindness, a type of muscular dystrophy, and hemophilia.

B. Because the male has only one *X* chromosome, his recessive *X*-linked characteristic will always be exhibited.

C. Most known sex-linked traits are caused by genes (alleles) on the *X* chromosome.

D. When these traits are recessive (most are), males express them because they have only one *X*. Females who have the allele are normally carriers and will exhibit the condition only if they are homozygous.

E. Males cannot pass sex-linked traits to sons (who get a *Y* from their father).

F. **Red-green color blindness** is a complex of sex-linked disorders, each of which is caused by an allele on the *X* chromosome. The result is considerable variation in the changes in color perception (Figure 9.23A).

G. **Hemophilia** is a sex-linked trait with a particularly well-studied history because of its incidence among the intermarrying royal families of Europe (Figure 9.23B).
NOTE: Hemophilia contributed to the Russian revolution of 1917. Rasputin gained influence over Czar Nicholas II and Czarina Alexandra by his apparent ability to control hemophilic episodes experienced by their child, Alexis.

H. **Duchenne muscular dystrophy** (DMD) is a severe disease that causes progressive loss and weakening of muscle tissue and has been traced to a particular nucleotide sequence.
NOTE: A functional version of the protein dystrophin is missing in individuals with DMD. Dystrophin is found in the plasma membrane (sarcolemma) of muscle fibers (Modules 20.6, 30.7, and 30.8). It appears that the result of not having a functional version of dystrophin is an increase in Ca^{2+} levels in the sarcoplasm (cytoplasm of a muscle fiber). The excess Ca^{2+} appears ultimately to lead to degeneration of the muscle fiber.

Class Activities

1. Remind students that most of the details covered in this chapter can be deduced by observation of patterns of inheritance across generations. Make an accounting of the ratios of some simple, Mendelian characteristics among your class (earlobes, mid-digital hair, tongue curling, etc.). You may want to see if it is possible to determine the genotypes of students for these characteristics by comparing how they express the characteristic with how each of their parents expresses that characteristic. Examine the distribution of phenotypes such as those illustrated in Figure 9.8A in your class by asking for a show of hands. You may want to explain that the class represents a "freely interbreeding" population in which the 3:1 ratio of dominant-to-recessive phenotypes is maintained only if there is no relative advantage of one characteristic over the other. However, since any class is a small sample size and there is no control over the parents of those in the class, it is unlikely that the class ratio will be 3:1 for any of these traits. Point out to the class how the dominant character is not always the most common character. This demonstration ties in with the process of science as discussed in Chapter 1 and helps preview population genetics (Chapter 13).
2. Ask your students which events of meiosis allow for the application of the rule of multiplication and the rule of addition.
3. See if, based on their phenotype and the phenotypes of their parents and other relatives for a particular trait, students can use a family pedigree to determine their genotype and the genotypes of their relatives for that trait.

Transparency Acetates

Figure 9.2B	Anatomy of a pea flower
Figure 9.2C	Mendel's technique for cross-fertilization of pea plants
Figure 9.2D	The seven pea characteristics studied by Mendel
Figure 9.3A	Crosses tracking one characteristic (flower color)
Figure 9.3B	Explanation of the crosses in Figure 9.3A
Figure 9.4	Homologous chromosomes
Figure 9.5A	Two hypotheses for segregation in a dihybrid cross
Figure 9.5B	Independent assortment of two genes in the Labrador retriever
Figure 9.6	Using a testcross to determine genotype
Figure 9.7	Segregation and fertilization as chance events
Figure 9.8B	Pedigree showing inheritance of deafness in a family from Martha's Vineyard
Figure 9.9A	Offspring produced by parents who are both carriers for a recessive disorder
Table 9.9	Some autosomal disorders in humans
Figure 9.10A	Amniocentesis
Figure 9.10B	Chorionic villus sampling
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Media

See the beginning of this book for a complete description of all media available for instructors and students. Animations and videos are available in the Campbell Image Presentation Library. Media Activities and Thinking as a Scientist investigations are available on the student CD-ROM and web site.

Animations and Videos	File Name
Human Fetus Ultrasound Video #1	09-10D-Ultrasound1Video-B.mov
Human Fetus Ultrasound Video #1	09-10D-Ultrasound1Video-S.mov
Human Fetus Ultrasound Video #2	09-10D-Ultrasound2Video-S.mov

Activities and Thinking as a Scientist	Module Number
Web/CD Activity 9A: *Monohybrid Cross*	9.3
Web/CD Activity 9B: *Dihybrid Cross*	9.6
Web/CD Activity 9C: *Gregor's Garden*	9.7
Web/CD Activity 9D: *Incomplete Dominance*	9.12
Web/CD Activity 9E: *Linked Genes and Crossing Over*	9.20
Web/CD Activity 9F: *Sex-Linked Genes*	9.22
Web/CD Thinking as a Scientist: *How Is the Chi-Square Test Used in Genetic Analysis?*	9.22
Biology Labs On-Line: *FlyLab*	9.22