Chapter 9 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). However, inheritance patterns can be rather unpredictable, as indicated in the photo of the mongrel pups (chapter-opening photo).

B. Behavior can be partially explained by genetics (the science of heredity), 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 Laws

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. The development or activity of other cells does not change reproductive 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: Plant and animal breeders provide data not only for hypotheses concerning inheritance, but this information also greatly influenced the ideas of Charles Darwin and Alfred Wallace, at about the same time (Module 13.1).

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), have numerous and distinct characteristics (Figure 9.2D), and the mating of individuals can be controlled so 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 in 1866, argued that there are discrete, heritable factors (what we call genes) transmitted from generation to generation.

C. Mendel could intentionally have a flower self-fertilize by covering it with a bag, or he could allow cross-fertilization between 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 true-breeding varieties of plants (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 then performed an experiment that produced hybrids (offspring from two different varieties) by performing a cross (cross-fertilization) of plants with two different traits. For example, peas with purple flowers and those with white flowers were crossed.

F. In these experiments, the true-breeding parents are the P generation (P for parental), their hybrid offspring is the F1 generation (F for filial, Latin for son), and the offspring of mating two F1 individuals is the F2 generation (second filial).

Module 9.3 Mendel’s law of segregation describes the inheritance of a single characteristic.

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

A. Law 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 as follows: out of 929 F2 offspring, 705 were purple, and 224 were white, a ratio of 3:1.

NOTE: The proportions are not exactly 3⁄4 and 1⁄4 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 (homozygous) or different alleles (heterozygous).

3. When the two alleles are different, the one that is fully expressed is said to be the dominant allele, and the one that is not noticeably expressed is said to be the recessive allele.

4. A sperm or egg carries only one allele for each characteristic because the allele pairs segregate from each other during gamete production. This is called the law of segregation (see anaphase I and anaphase II; Module 8.14).

E. Conventions for alleles: P, the dominant (purple) allele, and p, the recessive (white) allele. P generation: PP 3 pp; their gametes: P and p; F1 generation: Pp (Figure 9.3B).

F. 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).

G. Homozygous dominant (PP), homozygous recessive (pp), and heterozygous (Pp) refer to the genotype (the nature of the genes as inferred from observations and knowledge of how the system works). The phenotype is what we see (expressed traits).

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 law of segregation.

C. Alleles of a gene reside at the same locus on homologous chromosomes (Figure 9.4).

NOTE: One of the chromosomes illustrated was inherited from the female parent, the other from the male parent.

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

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

A. Law of independent assortment: Each pair of alleles segregates independently from the other pairs during gamete formation.

B. Dihybrid cross: Breed two true-breeding strains, 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 (F1: RrYy). Then allow the F1 to self-fertilize (RrYy 3 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 42, or 16, possible gametic combinations.

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

D. Results: The F1 generation exhibits only the dominant phenotype (this is expected). The F2 generation exhibits a phenotypic ratio of 9:3:3:1 (round yellow: round green: wrinkled yellow: wrinkled green).

NOTE: 9 1 3 1 3 1 1 5 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 5 black fur; b allele 5 brown fur; N allele 5 normal vision; n allele 5 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 F1 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 laws 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 1⁄2.

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 (1⁄4). The probability of two recessives occurring together in a dihybrid cross is 1⁄4 3 1⁄4, or 1 out of 16 (recall 9 1 3 1 3 1 1 5 16). In a trihybrid cross, as mentioned, the probability of a triple recessive is 1 out of 64 (or 1⁄4 3 1⁄4 3 1⁄4 5 1⁄64).

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 (family tree), showing the appearance of congenital deafness in a Martha’s Vineyard family (Figure 9.8).

B. By applying Mendel’s laws, 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 (possess the heterozygous genotype and express the dominant phenotype). From this it follows that all their hearing children were carriers.

D. This final deduction shows the power of applying Mendelian laws 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. There are thousands of known genetic traits that 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. Recessive Disorders: 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 heterozygous state, 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 laws.

E. Cystic fibrosis is the most common lethal genetic disease in the United States.

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 (mating with close relatives) 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. Dominant Disorders: 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. A prime example of this type of dominant disorder is Huntington’s disease.

K. With practice, the laws 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: New technologies can provide insight into one’s genetic legacy.

A. Circumstances may lead a couple to seek medical advice and counseling prior to conception, during a pregnancy, or after birth of a child. There are several methods that can be used to assist in the process.

B. Identifying carriers: Tests can be performed that can identify if a person is a carrier of a particular recessive allele that can cause disease. Carriers of recessive alleles such as Tay-Sachs disease, sickle-cell disease, and cystic fibrosis can be identified.

C. Fetal testing: During the pregnancy, several techniques can be used.

1. Amniocentesis involves taking a sample of the amniotic fluid that bathes the fetus, at 14–20 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, left).

2. Chorionic villus sampling (CVS) 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 9.10A, right). CVS has the added advantage of speed over amniocentesis; however, both carry a slight risk of complications to the mother and/or the fetus (1% and 2%, respectively).

3. 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. All three tests are often used as a triple screening regimen.

D. Fetal imaging:

1. Ultrasound imaging of the fetus provides a noninvasive view inside the womb (Figure 9.10B).

2. Fetoscopy provides a more direct view of the fetus through a needle-thin viewing scope inserted into the uterus. Fetoscopy, like CVS and amniocentesis, carries a small risk and is reserved for situations with higher probabilities of disorders (for example, older parents or situations where genetic counseling has uncovered a higher risk).

E. Newborn screening: State laws mandate the routine screening of all infants for the genetic disorder called PKU (phenylketonuria). If left untreated, the baby will develop mental retardation. If caught early, the child will most likely develop normally.

F. Ethical considerations: Ethical issues and questions abound.

1. Will all parties maintain confidentiality during and after genetic counseling? If not, will insurance coverage be compromised, will couples be stigmatized, and will misinformed people mistake carrier for contagious disease?

2. 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 and carrying a defective baby to term.

II. Variations on Mendel’s Laws

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

A. The inheritance of many characteristics among all eukaryotes follows the laws that Mendel discovered.

NOTE: Discussing these laws 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 law 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 laws, not exceptions to them.

Module 9.12 Incomplete dominance results in intermediate phenotypes.

A. Complete dominance was illustrated in the work done by Mendel. The flowers were always purple or white. 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 is the inheritance of alleles that relate to hypercholesterolemia. Normal individuals (HH) have normal amounts of LDL receptor proteins; while diseased (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).

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 either 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, a misnomer) has neither carbohydrate and can receive no other type. Type AB (universal recipient, also a misnomer) has both carbohydrates and should receive no other 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; both alleles are expressed in the heterozygous individual. Remember that codominance is not the same as incomplete dominance (the expression of one intermediate trait).

Module 9.14 A single gene may affect many phenotypic characteristics.

A. Pleiotropy is the common situation in which a gene influences multiple characteristics.

B. An example is the inheritance of an allele that encodes for an 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).

Module 9.15 A single characteristic may be influenced by many genes.

A. Polygenic inheritance is the additive effect of two or more genes on a single phenotypic characteristic.

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 for skin color, each with two alleles coding for “additive units,” which produce the overall characteristic. AABBCC would be a very-dark-skinned person while aabbcc would be a very-light-skinned person. Because the effect of the genes is additive, a person with the genotype AABbcc would have the same skin color as a person with the genotype AaBbCc. In reality, the human skin has even more variations than illustrated here.

NOTE: Point out to your students how much easier it is to solve genetic problems such as these using probabilities instead of Punnett squares.

Module 9.16 The environment affects many characteristics.

A. The example of skin color in Module 9.15 is a gross underexaggeration of the range of skin color in humans. The bell-shaped curve in Figure 9.15 would more accurately represent the range of skin color. However, because of environmental influences, no degree of characterization of genes will fully explain the range of skin color in humans.

B. Many characteristics are a result of the combination of genetic and environmental influences. This statement holds true for organisms besides humans. An oak tree is genetically locked into being an oak, but the leaf size, shape, and shades of green are influenced by the exposure to the wind and sun.

C. The old and hotly contested debate of “nature versus nurture” still rages. There are some traits that are undeniably determined by the genes that we inherit (for example, ABO blood group). But are all phenotypes determined by the genotype? Consider red blood cell count and altitude.

D. A good example of environmental influence is illustrated by identical twins that are easily distinguished. It is important to remember that environmental influences are not passed on the next generation.

Module 9.17 Genetic testing can detect disease-causing alleles.

A. The field of genetic testing (also known as genetic screening) has expanded dramatically in the past decade.

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

C. 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 (see breast cancer researcher, Mary Claire King, Module 11.19).

III. The Chromosomal Basis of Inheritance

Module 9.18 Chromosome behavior accounts for Mendel’s laws.

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. The theory states that genes occupy specific loci on chromosomes, and it is the chromosomes that undergo segregation and independent assortment during meiosis.

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

C. The fact that there are several sets of homologous pairs of chromosomes accounts for the law of independent assortment. Figure 9.18 illustrates Mendel’s laws.

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 9.19 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 law 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 9.19).

Module 9.20 Crossing over produces new combinations of alleles.

A. Homologous chromosomes will undergo crossing over during meiosis and produce new combinations of alleles (review Module 8.18).

B. Figure 9.20A illustrates the process of crossing over between two linked genes and the production of four different gamete genotypes.

C. Early examples of recombination were demonstrated in fruit flies (Figure 9.20B) by embryologist T. H. Morgan and colleagues in the early 1900s.

D. The percentage of recombinant offspring is called the recombination frequency (Figure 9.20C).

Module 9.21 Geneticists use crossover data to map genes.

A. The study of fruit-fly genetics resulted in considerable additional understanding of genetic laws. 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.

NOTE: This is another example of the use of an experimental organism that lends itself to study.

B. A. H. Sturtevant, one of Morgan’s colleagues (both seen in Figure 9.21A), 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 was proportional to the distance between two genes on a chromosome (Figure 9.21B) and this information could be used to construct a genetic map (Figure 9.21C).

IV. Sex Chromosomes and Sex-Linked Genes

Module 9.22 Chromosomes determine sex in many species.

A. Sex chromosome are the genes (commonly designated X and Y) present in many animals, including fruit flies and humans, which determine the sex of the individual.

B. Sex chromosomes in humans are nonidentical members of a homologous pair. In humans, XX individuals are female, and XY are male (Figure 9.22A).

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 (Figures 9.22B and C). For example, fish, butterflies, and birds use the egg to determine the sex of the offspring, and the sex chromosomes are designated with W and Z. The eggs are either W or Z, and males have the genotype ZZ, while females are genotype ZW.

E. In some species, chromosome number rather than chromosome type determines sex (Figure 9.22D). 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. The chromosome complement for all individuals in such species will be the same.

Module 9.23 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 genes.

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

C. Examples are presented using eye color in fruit flies (X-linked recessive for white eyes; Figure 9.23A). Depending on the genotypes of the parents, three patterns emerge:

1. Female XRXR 3 Male XrY: All offspring with red eyes regardless of the sex of the individuals (Figure 9.23B).

2. Female XRXr 3 Male XRY: All females with red eyes, half the males with red eyes and the other half with white eyes (Figure 9.23C).

3. Female XRXr 3 Male XrY: An even number of each type regardless of the sex (Figure 9.23D).

D. In humans, most sex-linked characteristics result from genes on the X chromosome.

Preview: Thus, mostly males are affected (Module 9.24).

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 9.24 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 9.24A).

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 9.24B).

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 son, 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 calcium ion levels in the sarcoplasm (cytoplasm of a muscle fiber). The excess calcium ions appear ultimately to lead to degeneration of the muscle fiber.