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# **Chapter 2: Understanding Genetics**

DNA CODES FOR PROTEINS THE CELL IS THE BASIC UNIT OF LIFE GROWTH AND REPRODUCTION GENETIC INHERITANCE DETERMINING GENDER COMMON MISCONCEPTIONS ABOUT INHERITANCE

The study of genetics (the inheritance of traits in living organisms) is a basic concept in biology. The same processes that provide the mechanism for organisms to pass genetic information to their offspring lead to the gradual change of species over time, which in turn produces biodiversity (the variety of life and the genetic differences among living organisms) and the evolution of new species. An understanding of genetics is becoming increasingly important as genetics research and technology—and the controversies surrounding them—gain greater influence on social trends and individual lives.

Rapid and revolutionary advances in genetics, medicine, and biotechnology have created new opportunities as well as a complex and far-reaching range of legal, ethical, and social issues. Genetic testing to screen for disease susceptibility, moral questions about the cloning of human organs, and the debate over genetically modified foods affect many lives. To fully appreciate the benefits, risks, and ramifications of genetics research and to critically evaluate the related issues raised by ethicists, scientists, and other stakeholders, it is vital to understand the basics of genetics, a discipline that integrates biology, mathematics, sociology, medicine, and public health.

To understand genetics, the development of organisms, and the diversity of species, it is important to learn how deoxyribonucleic acid (DNA) functions as the information-bearing molecule of living organisms. This chapter contains definitions of basic genetics terms such as *DNA*, *chromosomes*, and *proteins*, as well as an introduction to genetic concepts and processes, including reproduction and inheritance.

# **DNA CODES FOR PROTEINS**

From the perspective of genetics, the DNA molecule has two major attributes. The first is that it is able to replicate—that is, to make an exact copy of itself that can be passed to another cell, thereby conveying its precise genetic characteristics. Figure 2.1 is a diagram that shows how DNA replication produces two completely new and identical daughter strands of DNA. The second critical attribute is that it stores detailed instructions to manufacture specific proteins—molecules that are essential to every aspect of life. DNA is a blueprint or template for making proteins, and much of the behavior and physiology (life processes and functions) of a living organism depends on the repertoire of proteins its DNA molecules know how to manufacture.

The function of DNA depends on its structure. The double strand of DNA is composed of individual building blocks called nucleotides that are paired and connected by chemical bonds. A nucleotide contains one of four nitrogenous bases: the purines (nitrogenous bases with two rings) adenine (A) and guanine (G), or the pyrimidines (nitrogenous bases with one ring; pyrimidines are smaller than purines) cytosine (C) and thymine (T). The two strands of DNA lie side by side to create a predictable sequence of nitrogenous base pairs. (See Figure 2.2.) A stable DNA structure is formed when the two strands are a constant distance apart, which can occur only when a purine (A or G) on one strand is paired with a pyrimidine (T or C) on the other strand. A generally pairs with T, and G generally pairs with C.

Proteins are molecules that perform all the chemical reactions necessary for life and provide structure and shape to cells. The properties of each protein depend primarily on its shape, which is determined by the sequence of its building blocks, known as amino acids. Proteins may be tough like collagen, the most abundant protein in the human body, or they may be stretchy like elastin, a protein that mixes with collagen to make softer, more flexible tissues such as skin. Figure 2.3 shows the shapes of four types of protein structures.

Proteins can act as structural components by building the tissues of the body. For example, some of the proteins in an egg include a bond that acts like an axle, allowing other parts of the molecule to spin around like wheels. However,

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FIGURE 2.1 DNA Replication DNA replication. Argosy Publishing, Cengage Gale.

when the egg is heated, these bonds break or denature, locking the "wheels" of the molecule in place. This is why an egg gets hard when you cook it.

Enzymes such as lactase, which helps in the digestion of lactose (milk sugar), and hormones such as insulin (a hormone that regulates carbohydrate metabolism by controlling blood sugar levels) are proteins that act to facilitate and direct chemical reactions. Defense proteins, which are able to combat invasion by bacteria or viruses, are embedded in the walls of cells and act as channels, determining which substances to let into the cell and which to block. Some bacteria know how to make proteins that protect them from antibiotics (substances such as penicillin and streptomycin that inhibit the growth of or destroy microorganisms), while the human immune system can make proteins that target

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FIGURE 2.2 Deoxyribonucleic acid (DNA) "Deoxyribonucleic Acid (DNA)," in Talking Glossary of Genetic Terms, U.S. Department of Health and Human Services, National Institutes of Health, National Human Genome Research Institute, Division of Intramural Research, http://www.genome.gov/Pages/Hyperion//DIR/VIP/Glossary/Illustration/dna.shtml (accessed September 3, 2008)

bacteria or other germs for destruction. Many essential biological processes depend on the highly specific functions of proteins.

### **Proteins and Amino Acids**

All proteins are composed of building blocks called amino acids. (See Figure 2.4.) There are 20 different kinds of amino acids, and each has a slightly different chemical composition. The structure and function of each protein depends on its amino acid sequence—in a protein containing a hundred amino acids, a change in a single one may dramatically affect the function of the protein. Amino acids



FIGURE 2.3 Four protein structures "Protein," in Talking Glossary of Genetic Terms, U.S. Department of Health and Human Services, National Institutes of Health, National Human Genome Research Institute, Division of Intramural Research, http://www.genome.gov/Pages/Hyperion//DIR/VIP/Glossary/Illustration/protein.shtml (accessed September 3, 2008)

are small molecular groups that act like jigsaw puzzle pieces, linking together in a chain to make up the protein. Each amino acid links to its neighbor with a special kind of covalent bond (covalent bonds hold atoms together) called a peptide bond. Many amino acids link together, side by side, to make a protein. Figure 2.5 is a diagram of a protein structure. Proteins range in length from 50 to 500 amino acids, linked head to tail.

Besides their ability to form peptide bonds to their neighbors, amino acids also contain molecular appendages called side groups. Depending on the particular atomic arrangement of the side group, neighboring amino acids experience different pushes and pulls as they attract or repel one another. The combination of the side-by-side peptide bond linking the amino acids into a chain, along with the extra influences of the side groups, twists a protein into a specific shape. This shape is called the protein's conformation, which determines how the protein interacts with other molecules.

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FIGURE 2.4 Amino acids "Amino Acids," in Talking Glossary of Genetic Terms, U.S. Department of Health and Human Services, National Institutes of Health, National Human Genome Research Institute, Division of Intramural Research, http://www.genome.gov/Pages/Hyperion//DIR/VIP/Glossary/Illustration/amino\_acid.shtml (accessed September 3, 2008)

Amino acids are linked head to tail, so that at one end there is a free amino group, and at the other a free carboxyl group. Proteins are typically 50–500 amino acids in length.



FIGURE 2.5 A primary protein structure Adapted from Richard Robinson, ed., "Schematic Diagram of a Primary Protein Structure," in Genetics, vol. 3, K—P, Macmillan Reference USA, 2002

### **Genes and Proteins**

Along the length of a DNA molecule there are regions that hold the instructions to manufacture specific proteins—a specific sequence of amino acids linked side by side. These regions are called protein-encoding genes and are an essential element of the modern understanding of genetics. Like a jukebox that holds a hundred songs but only plays the one that is selected at any given moment, a DNA molecule can contain anywhere from a dozen to several thousand of these protein-encoding genes. However, as with the jukebox, at any given time only some of these genes will be expressed—that is, switched on to actively produce the protein they know how to make.

The protein-synthesizing instructions present in DNA are interpreted and acted on by ribonucleic acid (RNA). RNA, as its name suggests, is similar to DNA, except that the sugar in RNA is ribose (instead of deoxyribose), the base uracil (U) replaces thymine (T), and RNA molecules are usually single stranded and shorter than DNA molecules. (See Figure 2.6.) RNA is used to transcribe and translate the genetic code contained in DNA. Transcription is the process by which a molecule of messenger RNA (mRNA) is made, and translation is the synthesis of a protein using mRNA code. Figure 2.7 shows the transcription of mRNA and how mRNA is involved in protein synthesis (translation).

# **Genetic Synthesis of Proteins**

How does a gene make a protein? The process of protein synthesis is quite complex. This overview describes the basic sequence of protein synthesis, which includes the following steps:

- 1. A gene is triggered for expression-to synthesize a protein.
- 2. Half of the gene is copied into a single strand of mRNA in a process called transcription.
- 3. The mRNA anchors to a ribosome, an organelle (or membrane-bound cell compartment) where protein synthesis occurs.
- 4. Each sequence of three bases on the mRNA, called a codon, uses the right type of transfer RNA (tRNA) to pick up a corresponding amino acid from the cell. (See Figure 2.8.)
- 5. A string of amino acids is assembled on the ribosome, side by side, in the same order as the codons of the mRNA, which, in turn, correspond to the sequence of bases from the original DNA molecule in a process called translation.
- 6. When the codons have all been read and the entire sequence of amino acids has been assembled, the protein is released to twist into its final form.

First, the DNA molecule receives a trigger telling it to express a particular gene. Many influences may trigger gene expression, including chemical signals from hormones and energetic signals from light or other electromagnetic energy. For example, the spiral backbone of the DNA molecule can actually carry pulsed electrical signals that participate in activating gene expression.

Once a gene has been triggered for expression, a special enzyme system causes the DNA's double spiral to spring apart between the beginning and end of the gene sequence. The process is similar to a zipper with teeth that remain connected above and below a certain area, but pop open to create a gap along part of the zipper's length. At this point, the DNA base pairs that make up the gene sequence are separated. DNA replication is based on the understanding that any exposed nucleotide thymine (T) will pick up an adenine (A) and vice versa, whereas an exposed cytosine (C) will connect to an available guanine (G) and vice versa. Here the same process takes place except that instead of unzipping

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FIGURE 2.6 **Comparison of ribonucleic acid (RNA) with deoxyribonucleic acid (DNA)**Adapted from "Ribonucleic Acid (RNA)," in Talking Glossary of Genetic Terms, U.S. Department of Health and Human Services, National Institutes of Health, National Human Genome Research Institute, Division of Intramural Research, http://www.genome.gov/Pages/Hyperion//DIR/VIP/Glossary/Illustration/rna.shtml (accessed September 3, 2008)

and copying the entire length of the DNA molecule, only the region between the beginning and end of the gene is copied. Instead of making a new double spiral, only one side of the gene is replicated, creating a special, single strand of bases called mRNA.

Once the mRNA has made a copy of one side of the gene, it separates from the DNA molecule. The DNA returns to its original state and the mRNA molecule breaks away, eventually connecting to an organelle in the cell called a ribosome. (Figure 2.9 is a drawing of a ribosome.) Here it anchors to another type of RNA called ribosomal RNA (rRNA). This is where the actual protein synthesis takes place. Using a special genetic coding system, each sequence of three bases on the mRNA copied from the gene is used to catch a corresponding amino acid floating inside the cell. These sequences of three bases are the codons, and they link to tRNA. A different form of tRNA is used to catch each different type of amino acid.



FIGURE 2.7 Messenger RNA (mRNA)"mRNA," in Talking Glossary of Genetic Terms, U.S. Department of Health and Human Services, National Institutes of Health, National Human Genome Research Institute, Division of Intramural Research, http://www.genome.gov/Pages/Hyperion//DIR/VIP/Glossary/Illustration/mrna.shtml (accessed September 3, 2008)

One after another, the mRNA's codons cause the corresponding tRNAs to be captured and linked, side by side, using the peptide bonds to connect them. When the last codon has been read and the entire peptide sequence is complete, the newly formed protein molecule is released from the ribosome. When this happens, all the side groups are able to interact, twisting the protein into its final shape. In this way, a protein-encoding gene is able to manufacture a protein from a series of DNA bases.

# THE CELL IS THE BASIC UNIT OF LIFE

Ever since Matthias Jakob Schleiden (1804–1881) and Theodor Schwann (1810–1882) put forth their theories—Schleiden in 1838 and Schwann in 1839—that all plants and animals are composed of cells, there has been continuous refinement of cell theory. The early view that cells were made up of protoplasm (a jellylike substance) has given way to the more sophisticated understanding that cells are highly complex organizations of even smaller molecules and substructures. Cytology (the study of the formation, structure, and function of cells) has benefited from ever-improving technology, including powerful microscopes that enable researchers to identify the organelles (component parts of the cell) and determine their roles in inheritance.

Cells are the basic units and building blocks of nearly every organism. (One exception is viruses, which are simple



FIGURE 2.8 CodonAdapted from "Codon," in Talking Glossary of Genetic Terms, U.S. Department of Health and Human Services, National Institutes of Health, National Human Genome Research Institute, Division of Intramural Research, http://www.genome.gov/Pages/Hyperion//DIR/VIP/Glossary/Illustration/codon.shtml (accessed September 3, 2008)

organisms that are not composed of cells.) Each cell of an organism contains the same genetic information, which is passed on faithfully when cells divide. Different types of cells arise because they use different parts of the information, as determined by the cell's history and the immediate environment. Different cell types may be organized into tissues and organs.

# **Cell Structure and Function**

Plants and animals, as well as other organisms such as fungi, are composed of eukaryotic cells, or eukaryotes, because they have nuclei and membrane-bound structures known as organelles. In eukaryotic cells the organelles within the cell sustain, support, and protect it, creating a barrier between the cell and its environment, acting to build and repair cell parts, storing and releasing energy, transporting material, disposing of waste, and increasing in number.

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A ribosome is a tiny particle of RNA and protein found in a cell's cytoplasm

FIGURE 2.9 Ribosome Adapted from "Ribosome," in Talking Glossary of Genetic Terms, U.S. Department of Health and Human Services, National Institutes of Health, National Human Genome Research Institute, Division of Intramural Research, http://www.genome.gov/Pages/Hyperion//DIR/VIP/Glossary/Illustration/ribosomes.shtml (accessed September 3, 2008)

Each organelle functions like an organ system for the cell. For example, the nucleus is the command center, masterminding protein synthesis within the cell. (See Figure 1.1 in chapter 1.) The ribosomes work as protein factories, the Golgi apparatus is a protein sorter, and the endoplasmic reticulum operates as a protein processor. Lysosomes and peroxisomes serve as the cell's digestive system, and mitochondria convert energy in the cell. The surface membrane of the cell acts like skin, selectively permitting molecules in and out of the cell.

The nuclei of eukaryotes contain the chromosomes, which are chains of genetic material coded in DNA. The threadlike chromosomes are contained in the nucleus of a typical animal cell. Genes are segments of DNA that carry a basic unit of hereditary information in coded form. (See Figure 2.10.) They contain instructions for making proteins.

The eukaryotic chromosome is composed of chromatin (a combination of nuclear DNA and protein) and contains a linear array of genes. It is visible just before and during cell division. (See Figure 2.11.) Human cells normally contain 23 pairs of chromosomes, or a total of 46 chromosomes, that may be examined using a process known as karyotyping, the organization of a standard picture of the chromosomes. Figure 2.12 is a karyotype—a photo of an individual's chromosomes.

Cells without nuclei, such as bacteria and blue-green algae, are called prokaryotic cells or prokaryotes. Prokaryotic



FIGURE 2.10 GeneAdapted from "Gene," in Talking Glossary of Genetic Terms, U.S. Department of Health and Human Services, National Institutes of Health, National Human Genome Research Institute, Division of Intramural Research, http://www.genome.gov/Pages/Hyperion//DIR/VIP/Glossary/Illustration/gene.shtml (accessed September 3, 2008)

cells are smaller than eukaryotic cells, contain less genetic information, and are able to grow and divide more quickly. They perform these functions without organelles. A prokaryotic cell's DNA is not contained in one location; instead, it floats in different regions of the cell. The DNA of prokaryotes also contains jumping genes that are able to bind to other genes and transfer gene sequences from one site of a chromosome to another.

# **GROWTH AND REPRODUCTION**

The growth of an organism occurs as a result of cell division in a process known as mitosis. Many cells are relatively short lived, and mitosis allows for regular renewal of these cells. It is also the process that generates the millions of cells needed to grow an organism, or in the case of a human being, the trillions of cells needed to grow from birth to adulthood.

Mitosis is a continuous process that occurs in several stages. Between cell divisions, the cells are in interphase, during which there is cell growth, and the genetic material (DNA) contained in the chromosomes is duplicated so that when the cell divides, each new cell has a full-scale version of the same genetic material. The process of mitosis involves exact duplication—gene by gene—of the cell's chromosomal material and a systematic method for evenly distributing this material. It concludes with the physical division known as cytokinesis, when the identical chromosomes pull apart and each heads for the nucleus of one of the new daughter cells.

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FIGURE 2.11 Chromosome "Chromosome," in Talking Glossary of Genetic Terms, U.S. Department of Health and Human Services, National Institutes of Health, National Human Genome Research Institute, Division of Intramural Research, http://www.genome.gov/Pages/Hyperion//DIR/VIP/Glossary/Illustration/chromosome.shtml (accessed September 3, 2008)

Mitosis occurs in all eukaryotic cells, except the gametes (sperm and egg), and always produces genetically identical daughter cells with a complete set of chromosomes.

If, however, mitosis occurred in the gametes, then when fertilization—the joining of sperm and egg—took place, the offspring would receive a double dose of hereditary information. To prevent this from occurring, the gametes undergo a process of reduction division known as meiosis. Meiosis reduces the number of chromosomes in the gametes by half, so that when fertilization occurs the normal number of chromosomes is restored. For example, in humans the gametes produced by meiosis are haploid—they have just one copy of each of the 23 chromosomes. Besides preventing the number of chromosomes from doubling with each successive generation, meiosis also provides genetic diversity in offspring.

During meiosis the chromosomal material replicates and concentrates itself into homologous chromosomes (doubled chromosomes), each of which is joined at a central spot called the centromere. Figure 2.11 shows chromosomes joined at the centromere, and Figure 2.13 shows the location of the centromere in the chromosome. Pairing up along their entire lengths, they are able to exchange genetic material in a process known as crossing over. Figure 2.14 shows homologous chromosomes crossing over during meiosis to create new gene combinations. Crossing over results in much of the genetic variation observed among parents and their offspring. The pairing of homologous chromosomes and crossing over occur only in meiosis.

The process of meiosis also creates another opportunity to generate genetic diversity. During one phase of meiosis, called metaphase, the arrangement of each pair of homologous chromosomes is random, and different combinations of maternal and paternal chromosomes line up with varying orientations to create new gene combinations on different chromosomes. This action is called independent assortment. Figure 2.15 shows the process of meiosis in an organism with six chromosomes. Because recombination and independent assortment of parental chromosomes takes place during meiosis, the daughter cells are not genetically identical to one another.

# **Gamete Formation**

In male animals gamete formation, known as spermatogenesis, begins at puberty and takes place in the testes. Spermatogenesis involves a sequence of events that begins with the mitosis of primary germ cells to produce primary spermatocytes. (See Figure 2.16.) Four primary spermatocytes undergo two meiotic divisions, and as they undergo spermatogenesis they lose much of their cytoplasm and develop the spermatozoa's characteristic tail, the motor apparatus that provides the propulsion necessary to reach the egg cell. Like oocytes (egg cells) produced by the female, the mature sperm cells will be haploid, possessing just one copy of each chromosome.

In female animals gamete formation, known as oogenesis, takes place in the ovaries. Primary oocytes are produced by mitosis in the fetus before birth. Unlike the male, which continues to produce sperm cells throughout life, in the female the total number of eggs ever to be produced is present at birth. In April 2009, however, the online edition of the journal *Nature Cell Biology* reported the results of experiments by Chinese researchers on female mice showing evidence that mammalian ovaries might actually hold primitive cells that could be manipulated into producing

new eggs ("Making New Eggs in Old Mice," April 11, 2009, http://www.nature.com/news/2009/090411/full/news.2009.362.html ). At birth, or shortly before, meiosis begins and primary oocytes remain in the prophase of meiosis until puberty. At puberty the first meiotic division is completed, and a diploid cell becomes two haploid daughter cells; one large cell becomes the secondary oocyte and the other the first polar body. The secondary oocyte undergoes meiosis a second time but the meiosis does not continue to completion without fertilization. Figure 2.17 shows the sequence of events leading to the production of a mature ovum.





FIGURE 2.12 KaryotypeAdapted from "Karyotype," in Talking Glossary of Genetic Terms, U.S. Department of Health and Human Services, National Institutes of Health, National Human Genome Research Institute, Division of Intramural Research, http://www.genome.gov/Pages/Hyperion//DIR/VIP/Glossary/Illustration/karyotype.shtml (accessed September 3, 2008)



FIGURE 2.13 Centromere"Centromere," in Talking Glossary of Genetic Terms, U.S. Department of Health and Human Services, National Institutes of Health, National Human Genome Research Institute, Division of Intramural Research, http://www.genome.gov/Pages/Hyperion//DIR/VIP/Glossary/Illustration/centromere.shtml (accessed September 3, 2008)

# Fertilization

Like gamete formation, fertilization is a process, as opposed to a single event. It begins when the sperm and egg first come into contact and fuse together and culminates with the intermingling of two sets of haploid genes to reconstitute a diploid cell with the potential to become a new organism.

Of the millions of sperm released during an ejaculation, less than 1% survives to reach the egg. Of the few hundred sperm that reach the egg, only one will successfully fertilize it. While the sperm are in the female reproductive tract, swimming toward the egg, they undergo a process known as capacitation, during which they acquire the capacity to fertilize the egg. As the sperm approach the egg they become hyperactivated, and in a frenzy of mechanical energy the sperm attempt to burrow their way through the outer shell of the egg called the zona pellucida.

The cap of the sperm, known as the acrosome, contains enzymes that are crucial for fertilization. These acrosomal enzymes dissolve the zona pellucida by making a tiny hole in it, so that one sperm can swim through and reach the surface of the egg. At this time, the egg transforms the zona pellucida by creating an impenetrable barrier, so that no other sperm may enter.

Sperm penetration triggers the second meiotic division of the egg. With this division the chromosomes of the sperm and egg form a single nucleus. The resulting cell—the first

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FIGURE 2.14 Gene crossoversAdapted from Richard Robinson, ed., "New Combinations of Alleles Result from the Crossing over between Homologous Chromosomes," in Genetics, vol. 3, K—P, Macmillan Reference USA, 2002

cell of an entirely new organism—is called a zygote. The zygote then divides into two cells, which, in turn, continue to divide rapidly, producing a ball of cells now called the blastocyst. The blastocyst is an early stage of embryogenesis, the process that describes the development of the fertilized egg as it becomes an embryo. In humans the developing baby is considered an embryo until the end of the eighth week of pregnancy.

# **GENETIC INHERITANCE**

For inheritance of simple genetic traits, the two inherited copies of a gene determine the phenotype (the observable characteristic) for that trait. When genes for a particular trait exist in two or more different forms that may differ among individuals and populations, they are called alleles. For example, brown and blue eye colors are different alleles for eye color. For every gene, the offspring receives two alleles, one from each parent. The combination of inherited alleles is the genotype of the organism, and its expression (the observable characteristic) is its phenotype. Figure 2.18 is a graphic example of phenotype.

For many traits the phenotype is a result of an interaction between the genotype and the environment. Some of the most readily apparent traits in humans, such as height, weight, and skin color, result from interactions between genetic and environmental factors. In addition, there are complex phenotypes that involve multiple gene-encoded proteins; the alleles of these particular genes are influenced by other factors, either genetic or environmental. So while the presence of certain genes indicates susceptibility or likelihood to develop a certain trait, it does not guarantee expression of the trait.

For a specific trait some alleles may be dominant whereas others are recessive. The phenotype of a dominant





FIGURE 2.15 Meiosis in an organism with six chromosomes" Meiosis in an Organism with Six Chromosomes: Replication Precedes Meiosis,"

allele is always expressed, whereas the phenotype of a recessive allele is expressed only when both alleles are recessive. Recessive genes continue to pass from generation to generation, but they are only expressed in individuals who do not inherit a copy of the dominant gene for the specific trait. Figure 2.19 shows the inheritance of a recessive trait, in this example a recessive mutation in mice that produces an albino offspring from black parents.

There are also some instances, known as incomplete dominance, when one allele is not completely dominant over the other, and the resulting phenotype is a blend of both traits. Skin color in humans is an example of a trait often governed by incomplete dominance, with offspring appearing to be a blend of the skin tones of each parent. Furthermore, some traits are determined by a combination of several genes (multigenic or polygenic), and the resulting phenotype is determined by the final combination of alleles of all the genes that govern the particular trait.

Some multigenic traits are governed by many genes, each contributing equally to the expression of the trait. In such instances a defect in a single gene pair may not have a significant impact on expression of the trait. Other multigenic traits are predominantly directed by one major gene pair and only mildly influenced by the effects of other gene pairs. For these traits the impact of a defective gene pair depends on whether it is the major pair governing expression of the trait or one of the minor pairs influencing its expression.

A range of other factors enters into whether a trait will be evidenced and the extent to which it is expressed. For example, different individuals may express a trait with different levels of severity. This phenomenon is known as variable expressivity.

#### **Determining Genetic Probabilities of Inheritance**

Conventionally, geneticists use uppercase letters to represent dominant alleles and lowercase letters to stand for recessive alleles. An organism with a pair of identical alleles for a trait is described as a homozygote, or homozygous for that particular trait. When organisms are homozygous for a dominant trait, all uppercase letters symbolize the trait, whereas those that are homozygous for a recessive trait are represented by all lowercase letters. A heterozygote is an organism with different alleles for a trait, one donated from each parent, and when one is dominant and the other is recessive the trait is shown using a combination of uppercase and lowercase letters.

Even though the combination of alleles is a random event, it is possible to predict the probability that an offspring will have the same or a different phenotype from its parents when the genotypes with respect to the specific trait of both parents and the phenotype associated with each possible combination of alleles are known. The formula used to determine genetic possibilities was developed by the British geneticist Reginald Crundall Punnett (1875–1967). The Punnett square is a grid configuration that depicts genotype and phenotype. In Punnett squares the genotypes of parents are represented with four letters. There are two alleles for each trait. Genotypes of haploid gametes are represented with two letters. Gametes will contain one allele for each trait in every

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FIGURE 2.16 Process of spermatogenesis Danton H. O'Day, "Spermatogenesis: The Sequence of Events," University of Toronto at Mississauga, Canada



FIGURE 2.17 Process of oogenesisDanton H. O'Day, "Oogenesis: The Sequence of Events," University of Toronto at Mississauga, Canada

possible combination, and all possible fertilizations are calculated. Punnett squares are used to compute the cross of a single gene or two genes and their alleles, but they become extremely complicated when used to predict the offspring of more than two alleles.

To construct a Punnett square, the alleles in the gametes of one parent are placed along the top, and the alleles in the gametes of the other parent are placed along the left side of the grid. The number of characteristics considered determines the size of the Punnett square. A monohybrid cross looks at one trait and has a two-by-two structure, with four possible genotypes resulting from the cross. A dihybrid cross looks at two traits and has a four-by-four structure, providing sixteen possible genotypes. Figure 2.20 is a simple Punnett square that shows one parent homozygous for the AA allele and another that is heterozygous (Aa), with the four possible offspring: 50% AA and 50% Aa. This is an example of offspring that shows just one phenotype even though two genotypes are present. Figure 2.21 is a Punnett square that shows the predicted patterns of recessive inheritance. When both parents are carriers of the trait and dominant inheritance, the offspring is likely to arise from an affected parent and a normal parent.

#### **Mitochondrial Inheritance**

Mitochondria are the organelles involved in cellular metabolism and energy production and conversion. Mitochondria are inherited exclusively from the mother and contain their own DNA, known as mtDNA, some of which multiplies during the organism's growth and development and as such is more susceptible to mutation (changes in

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**FIGURE 2.18 Phenotype** "Phenotype," in Talking Glossary of Genetic Terms, U.S. Department of Health and Human Services, National Institutes of Health, National Human Genome Research Institute, Division of Intramural Research, http://www.genome.gov/Pages/Hyperion//DIR/VIP/Glossary/Illustration/phenotype.shtml (accessed September 3, 2008)

DNA sequence). The fact that mitochondria contain their own DNA has prompted scientists to speculate that they originally existed as independent one-celled organisms that over time developed interdependent relationships with more complex, eukaryotic cells.

Mitochondrial inheritance looks much like Mendelian inheritance (or genetic inheritance as described by Gregor Mendel's [1822–1884] laws), with two important exceptions. First, all maternal offspring are usually affected, whereas even in autosomal dominant disorders (those not related to the sex genes) only 50% of offspring are expected to be affected. (See Figure 2.22.) Second, mitochondrially inherited traits are never passed through the male parent. Males are as likely to be affected as females, but their offspring are not at risk. In other words, when there is a mutation in a mitochondrial gene, it is passed from a mother to all of her children; sons will not pass it on, but daughters will pass it on to all their children.

Mutations in mtDNA have been linked to the development of disease in humans. Leber's hereditary optic neuropathy, a painless loss of vision that afflicts people between the ages of 12 and 30, was the first human disease to be associated with a mutation in mtDNA. Many diseases linked to mtDNA affect the nervous system, heart or skeletal muscles, liver, or kidneys—sites of energy usage.

# **DETERMINING GENDER**

From the moment of fertilization, the new organism has been assigned a gender, and its growth will proceed to develop either as a male or a female organism. The first clues that prompted scientists to consider that the determination of gender was influenced by genetics came from two key observations. The first was the fact that there is a general tendency toward a one-to-one ratio of males to females in all species. The second was the realization that the determination of gender, or sex, followed the principles of Mendelian genetics—gender was predictable as expected when individuals pure for a recessive trait were crossed with individuals that were hybrid.

The determination of gender occurs in all complex organisms, but the processes vary, even among animals. In humans 22 of the 23 pairs of chromosomes are as likely to be found in males as in females. These 22 chromosomes are known as the autosomes, and the 23rd pair is the sex chromosome. The sex chromosomes of females are identical and are called X chromosomes. In males the pair consists of an X chromosome and a smaller Y chromosome. (See Figure 2.23.)

### **Chromosome Theory of Sex Determination**

The genetic influence on gender is called the chromosome theory of sex determination, which states that:

• Gender is determined by the sex chromosome.

- In females the sex chromosomes are identical-both are X chromosomes.
- Because females have an XX genotype, all egg cells contain an X chromosome.

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FIGURE 2.19 Recessive inheritance"Recessive," in Talking Glossary of Genetic Terms, U.S. Department of Health and Human Services, National Institutes of Health, National Human Genome Research Institute, Division of Intramural Research, http://www.genome.gov/Pages/Hyperion//DIR/VIP/Glossary/Illustration/recessive.shtml (accessed September 3, 2008)

	A	A
A	AA	AA
a	Aa	Aa

FIGURE 2.20 Punnett squareCreated by Barbara Wexler for Gale, July 2003

- In males the sex chromosomes are not identical; one is X and one is Y.
- Because males have an XY genotype, half of all sperm cells contain an X chromosome and half contain a Y chromosome.
- After fertilization, the egg may receive either an X or a Y chromosome from the sperm. Because all egg cells contain an X chromosome, the determination of gender is wholly dependent on the chromosomal composition of the sperm. Sperm carrying the Y chromosome are known as androsperm; those containing the X chromosome are called gynosperm. If the sperm carries the Y chromosome, the offspring will be male (XY); if it carries an X chromosome, the offspring will be female (XX).

The determination of gender occurs at conception with the designation of chromosomal composition that is either XX or XY. However, a number of other genetic and environmental influences determine sex differentiation—the way in which the genetically predetermined gender becomes a reality. Differentiation translates the genetically coded message for gender into the physical traits, such as the hormones that influence the development of male and female genitalia, body functions, and behaviors associated with gender identity.

Interestingly, humans have an inherent tendency toward female development. Research conducted during the 1940s and 1950s confirmed that in many animals, individuals with just a single X chromosome developed as females, although in many instances they did not develop completely and were sterile (unable to reproduce). The absence of the Y chromosome results in female development, whereas the presence of the Y chromosome sets in motion the series of events that result in male development. These findings led to the premise that female development is the default option in the process of gender determination.

#### **Distribution of Males and Females in the Population**

Because human males produce equal numbers of sperm bearing either the X or the Y chromosome, and fertilization is a random event, then it stands to reason that in each generation equal numbers of males and females should be born. An examination of birth statistics in the United States and in other countries where reliable statistics have been compiled over time show that every year there are more births of males than females. For example, according to Joyce A. Martin et al., in "Births: Final Data for 2005" (*National Vital Statistics Reports*, vol. 56, no. 6, December 5, 2007), there were 2,118,982 live male births in 2005, compared to 2,019,367 live female births, a ratio of 1,049 males per 1,000 females for births to mothers of all races in the United States. Table 2.1 shows that births by sex varied by race, from a low of 1,024 males per 1,000 females among Native Americans or Alaskan Natives to a high of 1,066 males per 1,000 females among Asians or Pacific Islanders. Martin et al. report that the annual distribution of births by

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FIGURE 2.21 Punnett square with predicted patterns of recessive inheritance. Argosy Publishing, Cengage Gale.

sex has remained essentially unchanged over the past 60 years, varying by less than 1%.

For years this difference was attributed to the idea that males were inherently stronger than females and better able to survive pregnancy and birth. This theory was dispelled when researchers found that nearly three times as many male fetuses spontaneously abort (dying before birth). In fact, male life expectancy is less than female life expectancy at every age, from conception to adulthood. (See Table 2.2.) The explanation for the higher proportion of male births appears to be that more male offspring are conceived—possibly as many as 125 to every 100—because the prenatal death rate

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#### Fully shaded indicates dominant trait; half-shaded indicates recessive trait.

FIGURE 2.22 Inheritance patterns Richard Robinson, ed., "Mitochondrial Inheritance Passes Only through the Mother," in Genetics, vol. 2, E—I, Macmillan Reference USA, 2002

for males is so high; at birth the gap closes to about 105 to every 100.

One explanation for the higher number of males conceived is that the smaller and stronger Y sperm are better able to swim quickly and

successfully reach the egg cell. Along with androsperm size and mobility, environmental conditions influence gender determination and the chances of fetal survival. For example, the mother's age and general health are strongly linked to favorable outcomes of conception and pregnancy and are less strongly linked to, but are associated with, gender. Younger mothers are more likely to conceive male offspring, by a ratio as high as 120 to 100, and unfavorable prenatal conditions such as poor health or maternal illness are more likely to compromise the survival of the male fetus than the female fetus.

### **Sex-Linked Characteristics**

The two sex chromosomes also differ in terms of the genes they contain, which relate to many traits other than gender. The Y chromosome is quite small and carries few genes other than the one that determines male gender. One of the few confirmed traits linked to the Y chromosome is the hairy ear trait, a characteristic that is distinctive but largely unrelated to health. Because this trait is located exclusively on the Y chromosome, it only appears in males.



FIGURE 2.23 Sex chromosomes "Sex Chromosomes," in Talking Glossary of Genetic Terms, U.S. Department of Health and Human Services, National Institutes of Health, National Human Genome Research Institute, Division of Intramural Research, http://www.genome.gov/glossary.cfm?key=sex%20chromosome (accessed September 3, 2008)

The X chromosome is larger and holds many genes that are as necessary for males as they are for females. The genes on the X chromosome are called X-linked, and characteristics or conditions arising from these genes are called X-linked traits or conditions. Most people, male and female, likely have several so-called defective genes with the potential to produce harmful characteristics or conditions, but these genes are usually recessive and are not expressed in the phenotype unless they are combined with a similar recessive gene on the

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#### TABLE 2.1 Total births, by race of mother and selected demographic characteristics, 2005

[Birth rates are live births per 1,000 population. Fertility rates are computed by relating total births, regardless of age of mother, to women aged 15–44 years. Total fertility rates are sums of birth rates for 5-year age groups multiplied by 5. Populations estimated as of July 1. Mean age at first birth is the arithmetic average of the age of mothers at the time of the birth, computed directly from the frequency of first births by age of mother]

		ska			
Characteristic	All races	White	Black	Native	Asian or Pacific Islander
*Male live births per 1.000 female live birth	IS.				

Notes: Race and Hispanic origin are reported separately on birth certificates. Race categories are consistent with the 1977 Office of Management and Budget (OMB) standards. Nineteen states reported multiple-race data for 2005. The multiple-race data for these states were bridged to the single-race categories of the 1977 OMB standards for comparability with other states. In this table all women (including Hispanic women) are classified only according to their race.

SOURCE: Joyce A. Martin et al., "Table 14. Total Number of Births, Rates (Birth, Fertility, and Total Fertility), and Percentage of Births with Selected Demographic Characteristics, by Race of Mother: United States, 2005," in "Births: Preliminary Data for 2005," National Vital Statistics Reports, vol. 56, no. 6, December 5, 2007, http://www.cdc.gov/nchs/data/nvsr/nvsr56/nvsr56\_06.pdf (accessed September 3, 2008)

			Number		
Births	4,138,349	3,229,294	633,134	44,813	231,108
			Rate		
Birth rate	14.0	13.4	16.2	14.2	16.5
Fertility rate	66.7	66.3	69.0	59.9	66.6
Total fertility rate	2,053.5	2,056.0	2,070.5	1,750.0	1,889.0
Sex ratio*	1,049	1,052	1,030	1,024	1,066
All births			Percent		
Births to mothers under 20 years	10.2	9.3	16.9	17.7	3.3
4th- and higher-order births	11.1	10.5	15.1	19.6	6.5
Births to unmarried mothers	36.9	31.7	69.3	63.5	16.2

Mothers born in the 50 states and D.C.	75.4	77.6	83.3	95.1	18.1
			Mean		
Age of mother at first birth	25.2	25.4	22.8	21.7	28.5

[Birth rates are live births per 1,000 population. Fertility rates are computed by relating total births, regardless of age of mother, to women aged 15-44 years. Total fertility rates are sums of birth rates for 5-year age groups multiplied by 5. Populations estimated as of July 1. Mean age at first birth is the arthmetic average of the age of mothers at the time of the birth, computed directly from the frequency of first births by age of mother]

Characteristic	All races	White	Black	American Indian or Alaska Native	Asian or Pacitic Islander
			Number		
Births	4,138,349	3,229,294	633,134	44,813	231,108
			Rate		
Birth rate	14.0	13.4	16.2	14.2	16.5
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Total fertility rate	2,053.5	2,056.0	2,070.5	1,750.0	1,889.0
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Age of mother at first birth	25.2	25.4	22.8	21.7	28.5

\*Male live births per 1,000 female live births.

Notes: Race and Hispanic origin are reported separately on birth certificates. Race categories are consistent with the 1977 Office of Management and Budget (CMB) standards. Nineteen states reported multiple-race data for 2005. The multiple-race data for these states were bridged to the single-race categories of the 1977 OMB standards for comparability with other states. In this table all women (including Hispanic women) are classified only according to their race.

**TABLE 2.1 Total births, by race of mother and selected demographic characteristics, 2005** Joyce A. Martin et al., "Table 14. Total Number of Births, Rates (Birth, Fertility, and Total Fertility), and Percentage of Births with Selected Demographic Characteristics, by Race of Mother: United States, 2005," in "Births: Preliminary Data for 2005," National Vital Statistics Reports, vol. 56, no. 6, December 5, 2007, http://www.cdc.gov/nchs/data/nvsr/nvsr56/nvsr56\_06.pdf (accessed September 3, 2008)

corresponding chromosome. For this to occur, both parents would have to contribute the same defective gene. Species are also protected from the harmful effects of single defective genes by virtue of the fact that most traits are multigenic (controlled by more than one gene).

Although X-linked dominant alleles affect males and females, males are more strongly affected because they inherit just one X chromosome and do not have a counterbalancing normal allele. Huntington's disease, an inherited neuropsychiatric disease that affects the body and mind, is an example of a disorder caused by an X-linked dominant allele. Males are affected more frequently and more severely than females by X-linked recessive alleles. The male receives his X chromosome from his mother. Because males have just one X chromosome, all the alleles it contains are expressed, including those that cause serious and sometimes lethal medical disorders. Examples of disorders caused by X-linked recessive alleles range from relatively harmless conditions such as red-green color blindness to the always-fatal Duchenne muscular dystrophy (DMD), one of a group of muscular dystrophies characterized by the enlargement of muscles. DMD is one of the most prevalent types of muscular dystrophy and involves rapid progression of muscle degeneration early in life. It is X-linked, affects mainly males, and is the most common neuro-muscular disease of childhood, with an estimated overall prevalence (the number of cases of a disease that are present in a particular population at a given time) of 63 cases per 1 million births. The National Institute of Neurological Disorders and Stroke indicates in *Muscular Dystrophy: Hope through Research* (September 15, 2008, http://www.ninds.nih.gov/disorders/md/detail\_md.htm#110183171 ) that one out of every 3,500 to 5,000 boys worldwide are diagnosed with it. Another X-linked recessive allele causes Tay-Sachs disease, a disease that is most common among people of Jewish descent and results in neurological disorders and death in childhood.

All the male offspring of females who carry X-linked recessive alleles will be affected by the recessive allele. Female children are not as likely to express harmful recessive X-linked traits because they have two X chromosomes. Fifty percent of the female offspring will receive the recessive allele from a mother who carries the allele and an unaffected father. (See Figure 2.22.)

# COMMON MISCONCEPTIONS ABOUT INHERITANCE

There are many myths and misunderstandings about genetics and inheritance. For example, some people mistakenly believe that in any population dominant traits are inevitably more common than recessive traits. This is simply not true, as evidenced by the observation that, among humans, the allele that produces six fingers and six toes on each hand and foot, respectively, is dominant over the allele for five fingers and five toes, but the incidence of polydactyly (extra digits) is actually quite low.

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TABLE 2.2 Life expectancy at selected ages, by race and gender, selected years 1900-2005

[Data are based on death certificates]

All races				White		Black or African American <sup>a</sup>			
Specified age and year	Both sexes	Male	Female	Both sexes	Male	Female	Both sexes	Male	Female
<b>D</b>									

Data not available.

<sup>a</sup> Data shown for 1900–1960 are for the nonwhite population.

<sup>b</sup> Death registration area only. The death registration area increased from 10 states and the District of Columbia (DC) in 1900 to the coterminous United States in 1933.

<sup>c</sup> Includes deaths of persons who were not residents of the 50 states and DC.

Notes: Populations for computing life expectancy for 1991–1999 are 1990-based postcensal estimates of U.S. resident population. In 1997, life table methodology was revised to construct complete life tables by single years of age that extend to age 100 (Anderson RN. Method for constructing complete annual U.S. life tables. National Center for Health Statistics. Vital Health Stat 2(129). 1999). Previously, abridged life tables were constructed for 5-year age groups ending with 85 years and over. Life table values for 2000 and later years were computed using a slight modification of the new life table method due to a change in the age detail of populations received from the U.S. Census Bureau. In 2003, seven states reported multiple-race data. In 2004, 15 states reported multiple-race data. In 2005, 21 states and the District of Columbia reported multiple-race data. The multiple-race data for these states were bridged to the single-race categories of the 1977 Office of Management and

Budget Standards for comparability with other states. Some data have been revised and differ from previous editions of Health, United States. Data for additional years are available.

SOURCE: "Table 27. Life Expectancy at Birth, at 65 Years of Age, and at 75 Years of Age, by Race and Sex: United States, Selected Years 1900–2005," in Health, United States, 2007. With Chartbook on Trends in Health of Americans, U.S. Department of Health and Human Services, Centers for Disease Control and Prevention, National Center for Health Statistics, 2007, http://www.cdc.gov/nchs/data/hus/hus07.pdf#027 (accessed September 3, 2008)

At birth	Remaining life expectancy in years								
1900 <sup>b,c</sup>	47.3	46.3	48.3	47.6	46.6	48.7	33.0	32.5	33.5
1950c	68.2	65.6	71.1	69.1	66.5	72.2	60.8	59.1	62.9
1960c	69.7	66.6	73.1	70.6	67.4	74.1	63.6	61.1	66.3
1970	70.8	67.1	74.7	71.7	68.0	75.6	64.1	60.0	68.3
1980	73.7	70.0	77.4	74.4	70.7	78.1	68.1	63.8	72.5
1990	75.4	71.8	78.8	76.1	72.7	79.4	69.1	64.5	73.6
1995	75.8	72.5	78.9	76.5	73.4	79.6	69.6	65.2	73.9
1997	76.5	73.6	79.4	77.1	74.3	79.9	71.1	67.2	74.7
1998	76.7	73.8	79.5	77.3	74.5	80.0	71.3	67.6	74.8
1999	76.7	73.9	79.4	77.3	74.6	79.9	71.4	67.8	74.7
2000	77.0	74.3	79.7	77.6	74.9	80.1	71.9	68.3	75.2
2001	77.2	74.4	79.8	77.7	75.0	80.2	72.2	68.6	75.5
2002	77.3	74.5	79.9	77.7	75.1	80.3	72.3	68.8	75.6
2003	77.4	74.7	80.0	77.9	75.3	80.4	72.6	68.9	75.9
2004	77.8	75.2	80.4	78.3	75.7	80.8	73.1	69.5	76.3
2005	77.8	75.2	80.4	78.3	75.7	80.8	73.2	69.5	76.5
At 65 years									
1950c	13.9	12.8	15.0		12.8	15.1	13.9	12.9	14.9
1960c	14.3	12.8	15.8	14.4	12.9	15.9	13.9	12.7	15.1
1970	15.2	13.1	17.0	15.2	13.1	17.1	14.2	12.5	15.7
1980	16.4	14.1	18.3	16.5	14.2	18.4	15.1	13.0	16.8
1990	17.2	15.1	18.9	17.3	15.2	19.1	15.4	13.2	17.2
1995	17.4	15.6	18.9	17.6	15.7	19.1	15.6	13.6	17.1
1997	17.7	15.9	19.2	17.8	16.0	19.3	16.1	14.2	17.6
1998	17.8	16.0	19.2	17.8	16.1	19.3	16.1	14.3	17.4
1999	17.7	16.1	19.1	17.8	16.1	19.2	16.0	14.3	17.3
2000	18.0	16.2	19.3	18.0	16.3	19.4	16.2	14.2	17.7
2001	18.1	16.4	19.4	18.2	16.5	19.5	16.4	14.4	17.9
2002	18.2	16.6	19.5	18.2	16.6	19.5	16.6	14.6	18.0
2003	18.4	16.8	19.7	18.4	16.8	19.7	16.8	14.8	18.3
2004	18.7	17.1	20.0	18.7	17.2	20.0	17.1	15.2	18.6
2005	18.7	17.2	20.0	18.8	17.2	20.0	17.2	15.2	18.7
At 75 years									
1980	10.4	8.8	11.5	10.4	8.8	11.5	9.7	8.3	10.7
1990	10.9	9.4	12.0	11.0	9.4	12.0	10.2	8.6	11.2
1995	11.0	9.7	11.9	11.1	9.7	12.0	10.2	8.8	11.1
1997	11.2	9.9	12.1	11.2	9.9	12.1	10.7	9.3	11.5
1998	11.3	10.0	12.2	11.3	10.0	12.2	10.5	9.2	11.3
1999	11.2	10.0	12.1	11.2	10.0	12.1	10.4	9.2	11.1
2000	11.4	10.1	12.3	11.4	10.1	12.3	10.7	9.2	11.6
2001	11.5	10.2	12.4	11.5	10.2	12.3	10.8	9.3	11.7
2002	11.5	10.3	12.4	11.5	10.3	12.3	10.9	9.5	11.7
2003	11.7	10.5	12.5	11.6	10.4	12.5	11.1	9.6	12.1
2004	11.9	10.7	12.8	11.9	10.7	12.8	11.4	9.9	12.2
2005	12.0	10.8	12.8	11.9	10.7	12.8	11.4	10.0	12.3

[Data are based on death certificates]



**TABLE 2.2 Life expectancy at selected ages, by race and gender, selected years 1900–2005** "Table 27. Life Expectancy at Birth, at 65 Years of Age, and at 75 Years of Age, by Race and Sex: United States, Selected Years 1900–2005," in Health, United States, 2007. With Chartbook on Trends in Health of Americans, U.S. Department of Health and Human Services, Centers for Disease Control and Prevention, National Center for Health Statistics, 2007, http://www.cdc.gov/nchs/data/hus/hus07.pdf#027 (accessed September 3, 2008)

Another lingering misconception is that sex-linked diseases occur only in males. This is untrue but it is easy to understand the source of the misunderstanding. For years it was thought that hemophilia (a disease characterized by uncontrolled bleeding) did not occur in females. The observation seemed reasonable because there were no reported cases of the disease among females. Even though it was true that there were no females with the disease, the reasoning

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was incorrect. For a female to suffer from hemophilia, she would require a defective recessive gene on both of her X chromosomes, meaning her mother was carrying the gene and the disease affected her father. Because most people with hemophilia died young, few lived to produce offspring. In other words, female hemophiliacs were rare because the pairings that might produce them were infrequent. During the 1950s the first cases of hemophilia in females were documented, and the theory was discarded.

Finally, the idea that humans are entirely unique in their genetic makeup is false. In fact, human beings share much of their genetic composition with other organisms in the natural world. Furthermore, most human genetic variation is relatively insignificant. Even variations that alter the sequence of amino acids in a protein often produce no discernable influence on the action of the protein. Differences in some portions of DNA with as yet unknown functions appear to have no impact at all.

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