CHAPTER 3

Genetics and Heredity

Key Themes in Genetics and Heredity

- **Nature/Nurture** What roles do nature and nurture play in development?
- **Child's Active Role** How does the child play an active role in the effects of heredity on development?
- **Individual Differences** How prominent are individual differences in development?

"Make a wish before you blow out the candles!" But before Sheila could finish speaking, all twelve candles had been extinguished. It had been a great birthday party for the two five-year-olds and their guests. Fortunately, enough candles had been found to signal each year of growth and to continue the custom of adding "one to grow on."

When Sheila first learned that she would become a mother, she had not realized just how much work it would be to rear a child, even though her husband had been an enormous help. Then again, she had not planned on having identical twins! Thus, for this party (as for just about everything else) there were two *cakes to be decorated,* two *presents to be wrapped,* two *sets of friends to invite for the festivities. Jasmine and Alyssa seemed to bask in the attention they were receiving on their special day. Their excited voices sounded so much alike, indistinguishable by others, including their parents, unless the listener could see which of the twins was speaking (and could remember what each was wearing for the day). The twins enjoyed many of the same games and activities; they displayed the same impatience when others couldn't tell whether they were speaking with Jasmine or Alyssa. But there were differences, too. Jasmine was more impulsive, willing to try new things, likely to jump into the fray of things without a second thought, and less sensitive to the concerns of others. Alyssa was more cautious and careful and seemed to become upset easily, but she was also more willing to help her friends. And although they shared many friends, each twin had preferences for certain playmates. For their mother it was a puzzle: Why were they similar in so many ways but at the same time clearly different?*

Parents of more than one child are aware of similarities among them. However, they often take particular notice of the differences; for example, by pointing out how one child "takes after," perhaps, his mother and another after her father. In the case of identical twins, of course, the similarities are far more striking, although parents of twins, like Jasmine's and Alyssa's mother, are able to quickly identify differences in the two children. What are the mechanisms by which such resemblances and differences come about? Though we may grant the contribution of nature to eye color, gender, height, and many other physical traits, heredity's role in other characteristics, such as whether we are contented or quick-tempered, prone to alcoholism, likely to suffer depression, bright and quick-witted, active or more sedentary, is far less certain. Is Jasmine more impulsive and Alyssa more cautious because these qualities developed untouched by the common genetic pattern the twins share or because their parents and others encouraged them to develop individual interests and styles? Or did their individuality come about, perhaps, because the twins actively pursued different paths of responding to their daily experiences despite their common genetic circumstance?

In this chapter, we examine hereditary contributions to development. Major advances in our understanding of the basic biological units of inheritance and their effects on behavior help us to better appreciate the mutual, interactive relationship between nature and nurture. Experiences mold, modify, and enhance biological predispositions, and in a similar manner, genetic endowment influences, perhaps even

actively promotes, selection and preference for certain kinds of environments. Our goal is to understand just how such complex interactions evolve.

We begin with a brief overview of the principles of heredity. The blueprint for development is replicated in nearly every cell of our body. This blueprint includes genetic instructions that distinguish us from other species of plants and animals. Regardless of the language we speak, the work we do, the color of our skin, or how friendly we are, we share a genetic underpinning that makes each of us a human being. This biological inheritance also contributes to our individuality. With the exception of identical twins such as Jasmine and Alyssa, each of us begins with a different set of genetic instructions. But even for identical twins, in whom genetic makeup is the same, the influence of distinctive experiences ensures that each of us is a unique individual, different from everyone else.

In this chapter, we also examine several examples of hereditary variations that pose problems for development. As researchers learn more about the ways in which genetic influences occur, we can design environments to help minimize the restrictions imposed by certain hereditary conditions. We consider too how genetic counseling assists parents in deciding whether to have children or how to prepare for a child who is likely to experience developmental problems.

Most psychological development, of course, cannot be linked to simple genetic instructions. Intelligence, temperament, and personality, along with susceptibility to various diseases and conditions, are the outcome of complex interactions between genetic and environmental events. In the final section of this chapter, we consider research involving identical and fraternal twins, siblings, adopted children, and other family relationships to help us understand the complex tapestry genetic and environmental factors weave for cognitive, social-emotional, and personality development (Gottlieb, Wahlsten, & Lickliter, 1998; Rutter, 2002).

Principles of Hereditary Transmission

KEY THEME
Nature/Nurture

Whether we have freckles, blonde hair, or a certain type of personality can be influenced by genetic factors, but none of these characteristics is bestowed on us at conception any more directly than is our ultimate height. We must make a distinction, then, between what our genetic makeup consists of and the kind of individual we eventually become. Thus, we must distinguish between the **genotype,** a person's constant, inherited genetic endowment, and the **phenotype,** his or her observable, measurable features, characteristics, and behaviors. A given phenotype is the product of complex interactions involving the genotype and the many events that are part of an individual's *experience.*

Modern theories of the genotype can be traced to a series of experiments reported in 1866 by Gregor Mendel, an Austrian monk. From his observations of the characteristics of successive generations of peas, Mendel theorized that hereditary characteristics are determined by pairs of particles called *factors* (later termed **genes,** the specialized sequences of molecules that form the genotype). He also proposed that the information provided by the two members of a pair of genes is not always identical. These different forms of a gene are today known as **alleles.** The terms *gene* and *allele* are often used interchangeably, but an allele refers to the specific variation, and sometimes many possible alternative versions exist for a particular gene.

Mendel also outlined the basic principle by which genes are transferred from one generation to another. He concluded that offspring randomly receive one member of every pair of genes from the mother and one from the father. This is possible because the parents' **gametes,** or sex cells (egg and sperm), carry only one member of each pair of genes. Thus, when egg and sperm combine during fertilization, a new pair of genes, one member of the pair inherited from each parent, is reestablished in the offspring. That individual, in turn, may transmit either member of this new pair to subsequent children. Thus, genetic information is passed on from one generation to the next.

genotype Total genetic endowment inherited by an individual.

phenotype Observable and measurable characteristics and traits of an individual; a product of the interaction of the genotype with the environment.

gene Large segment of nucleotides within a chromosome that codes for the production of proteins and enzymes. These proteins and enzymes underlie traits and characteristics inherited from one generation to the next.

allele Alternate form of a specific gene; provides a genetic basis for many individual differences.

gametes Sperm cells in males, egg cells in females, normally containing only twenty-three chromosomes.

At about the same time Mendel's research was published, biologists discovered **chromosomes,** long, threadlike structures in the nucleus of nearly every cell in the body. In the early 1900s, several researchers independently hypothesized that genes are located on chromosomes. Yet another major breakthrough occurred in 1953 when James Watson and Francis Crick deciphered the structure of chromosomes and, in so doing, proposed a powerfully elegant way by which genes are duplicated during cell division. By 1956, researchers had documented the existence of forty-six chromosomes in normal human body cells. Today, the monumental effort to map the entire sequence of the **human genome,** that is, the nearly 3 billion chemical base pairs that make up every human's biological inheritance, is essentially complete (Celera Genomics Sequencing Team, 2001; International Human Genome Mapping Consortium, 2001).

The Building Blocks of Heredity

How could hereditary factors play a part in the similarities displayed by Jasmine and Alyssa or in a child's remarkable mathematical ability or in yet another's mental retardation? To understand the genotype and its effects on appearance, behavior, personality, or intellectual ability, we must consider genetic mechanisms at many different levels.

To begin with, every living organism is composed of cells—in the case of mature humans, trillions of cells. As Figure 3.1 indicates, within the nucleus of nearly all cells are the chromosomes that carry genetic information critical to the cells' functioning. Genes, regions within the strands of chromosomes, determine the production of specific proteins in the cell. The genes, in turn, are made up of various arrangements of four different chemical building blocks called **nucleotides** that contain one of four nitrogen-based molecules (*adenine, thymine, cytosine,* or *guanine*). The nucleotides pair together in one of only two ways to form the rungs of a remarkably long, spiral staircaselike structure called **DNA** or **deoxyribonucleic acid** (see Figure 3.1). An average of about one thousand nucleotide pairs make up each gene, although some genes have substantially more pairings (National Research Council, 1988). Genes differ from one another in number and sequence of nucleotide pairings and in their location on the chemical spiral staircases, or chains of DNA that we call the chromosomes.

Just as Mendel had theorized, hereditary attributes are, in most cases, influenced by pairs of genes or, more specifically, the two allelic forms of the pair. One member of the pair is located on a chromosome inherited from the mother, the other on a similar, or *homologous,* chromosome acquired from the father. Figure 3.2 shows a **karyotype** or photomicrograph of the forty-six chromosomes that males normally possess.

As can be seen in Figure 3.2, the homologous sets of chromosomes that are not genetically involved in the determination of sex, called **autosomes,** can be arranged in pairs and numbered from 1 to 22 on the basis of their size. However, the remaining two chromosomes specify the genetic sex of an individual and differ for males and females. In females this pair consists of **X chromosomes;** both are relatively large and similar in size. The normal male, however, has one X chromosome and a much smaller **Y chromosome.** The Y chromosome is believed to carry only a few dozen genes, in sharp contrast to the two to three thousand genes estimated to be on the X chromosome (Jegalian & Lahn, 2001). Thus, although most genes are present in pairs, males typically have only one instance of each gene found on the sex chromosomes. Nevertheless, the Y chromosome carries a specific gene that promotes the development of the male *gonads* (testes) and, as a consequence, provides an important first step in determining whether an individual is likely to be identified as male or female.

chromosomes Threadlike structures of DNA, located in the nucleus of cells, that form a collection of genes. A human body cell normally contains forty-six chromosomes.

human genome Entire inventory of nucleotide base pairs that compose the genes and chromosomes of humans.

nucleotide Repeating basic building block of DNA consisting of nitrogen-based molecules of adenine, thymine, cytosine, and guanine.

deoxyribonucleic acid (DNA) Long, spiral staircaselike sequence of molecules created by nucleotides identified with the blueprint for genetic inheritance.

karyotype Pictorial representation of an individual's chromosomes.

autosomes Twenty-two pairs of homologous chromosomes. The two members of each pair are similar in size, shape, and genetic function. The two sex chromosomes are excluded from this class.

X chromosome Larger of the two sex chromosomes associated with genetic determination of sex. Normally females have two X chromosomes and males only one.

Y chromosome Smaller of the two sex chromosomes associated with genetic determination of sex. Normally males have one Y chromosome and females none.

Cell Division and Chromosome Duplication

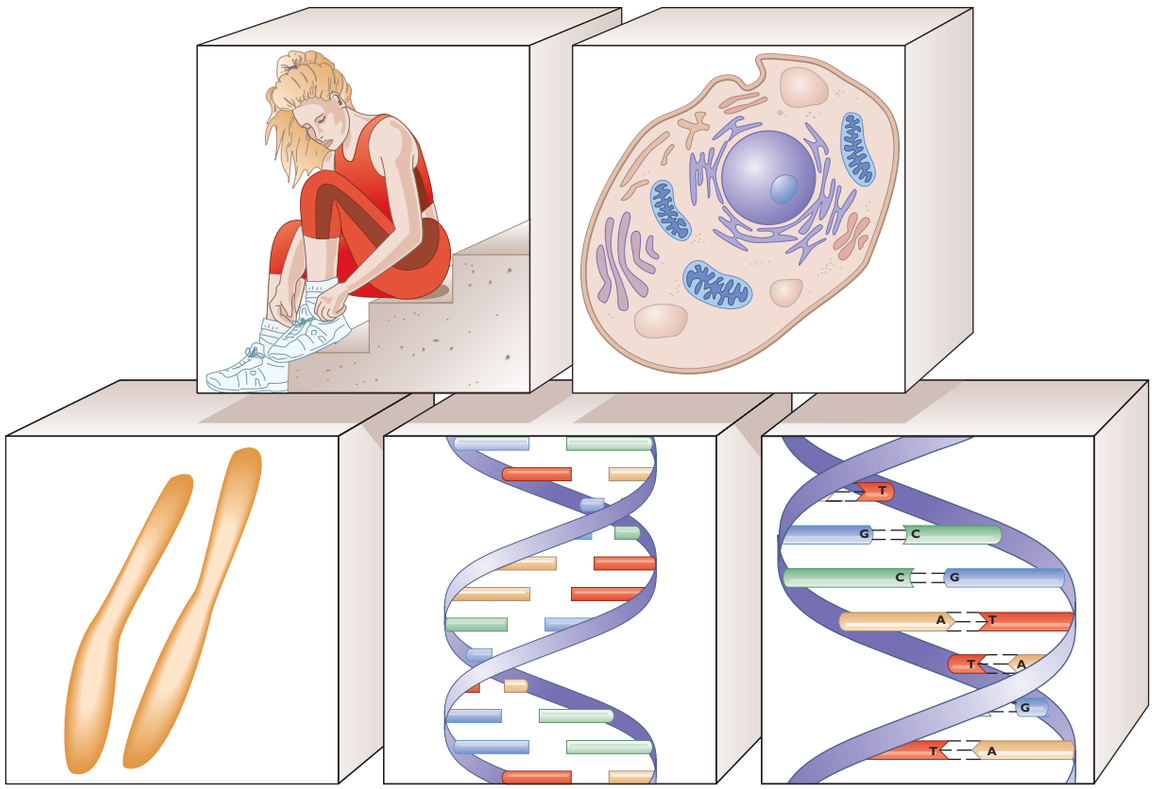
Each of us began life as a single cell created when a sperm cell, normally containing twenty-three chromosomes, from the father united with an ovum (egg), normally containing an additional twenty-three chromosomes, from the mother. The develop-

FIGURE 3.1 The Building Blocks of Heredity

Hereditary contributions to development can be observed at many levels. This figure depicts five major levels. Nearly every cell in the human body carries the genetic blueprint for development in the chromosomes. Specific regions on each chromosome, the genes, regulate protein production. Looked at in even more detail, the human genome consists of chemical molecules that are the building blocks for the genes. Each of these different levels of the individual's biological makeup can offer insights into the mechanisms by which the genotype affects the phenotype, the observable expression of traits and behaviors.

1. The **human body** has about 10 trillion cells. Proteins determine the structure and function of each cell.

2. Most **cells** contain a nucleus. Located within the nucleus are forty-six chromosomes that carry the instructions that signal the cell to manufacture various proteins.



3. A **chromosome** is a long thin strand of DNA organized as a coiled double helix. A full set of forty-six chromosomes in humans is believed to contain somewhere between 26,000 and 38,000 genes, far fewer than had been believed before the human genome was mapped.

4. A **gene** is made up of thousands of nucleotide pairs. Each gene typically has information designed to specify the production of one or more particular proteins.

5. **Nucleotides**, composed of four different kinds of chemical building blocks—adenine (A), thymine (T), cytosine (C), and guanine (G)—are the smallest genetic unit and are paired in specific combinations. Nearly 3 billion pairs of nucleotides make up the total complement of DNA in humans.

Source: Adapted from Isensee, 1986.

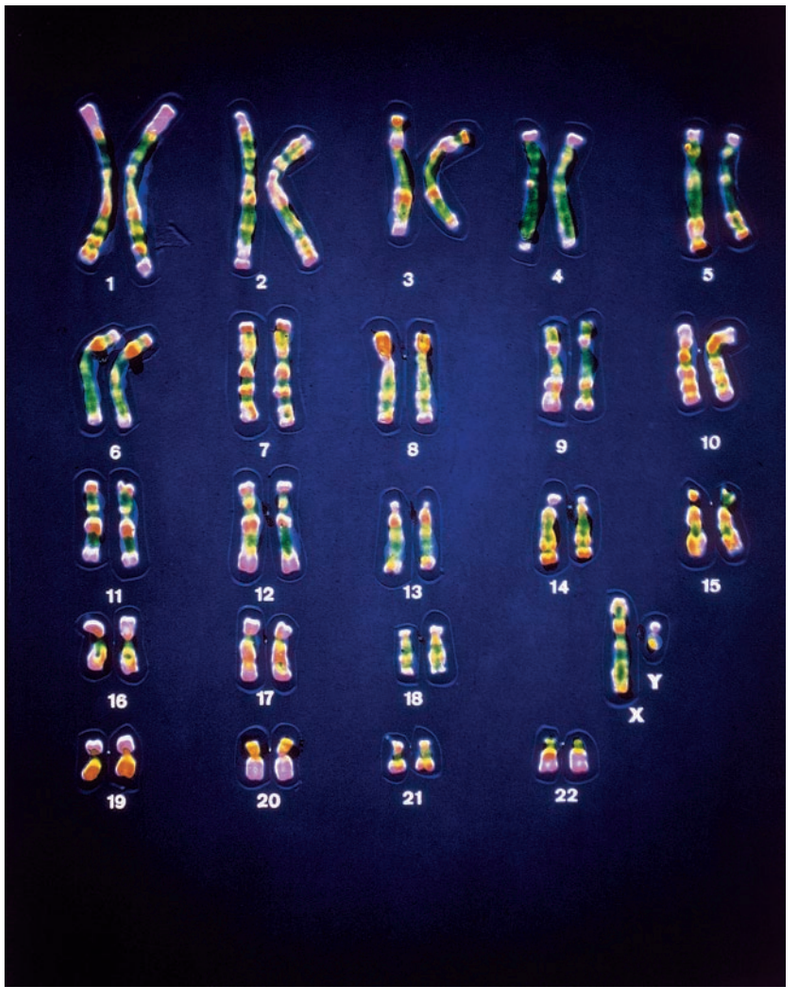
mental processes started by this fertilized egg cell, called a **zygote,** are more fully described in the chapter titled "The Prenatal Period and Birth." Remarkably, however, nearly every one of the millions of different cells in the newborn, whether specialized for bone or skin, heart or brain, or in some other way, contains the same genetic blueprint established in the initial zygote.

How does this extraordinary duplication of DNA from one cell to another and from one generation to the next take place? Most cells divide through the process

zygote Fertilized egg cell.

FIGURE 3.2
Chromosomes in the Normal Human Male

This karotype depicts the twenty-two homologous pairs of autosomes and the two sex chromosomes in the normal human male. In females, the twenty-third pair of chromosomes consists of an XX pair instead of an XY pair.



called **mitosis.** During mitosis, genetic material in the nucleus of the cell is reproduced such that a full complement of DNA becomes available to each new cell. Even before cell division occurs, the chemical bonds linking the nucleotides that form the rungs of the DNA ladder weaken. The pairs of nucleotides separate as though they were being unzipped from each other. At the same time, additional nucleotides are manufactured in the cell and attach to the separated nucleotides. Because each nucleotide can combine with only one other type, the two newly formed strands of DNA are rebuilt exactly as in their original sequence. The two newly formed copies of DNA eventually separate completely so that one becomes a member of each of the two new daughter cells, as depicted in Figure 3.3.

The process of cell division associated with the gametes (the sex cells) is called **meiosis.** Meiosis, which results in twenty-three chromosomes in the egg and sperm cells, actually involves *two* successive generations of cell divisions. In the first stage, each of the forty-six chromosomes begins to replicate in much the same way as mitosis begins. However, before the identical replicas split apart, the cell divides, so that each daughter cell receives only one chromosome from each of the twenty-three pairs, as pictured in Figure 3.4. In the second stage, the replicas of the twenty-three chromosomes completely separate, and the cell divides once more, each cell again receiving one of the replicas. Thus, from these two successive divisions, four cells are produced, each with twenty-three chromosomes.

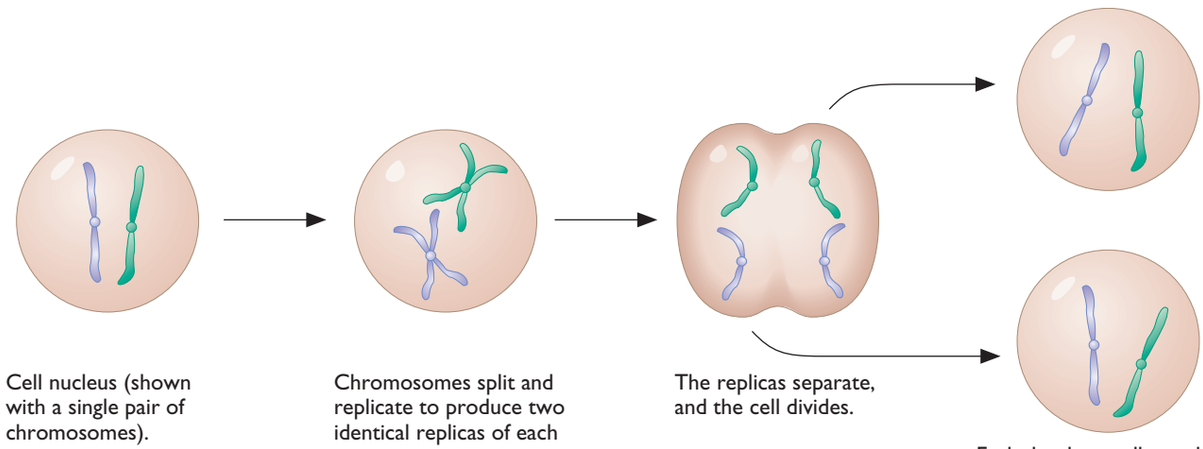
Random segregation of the twenty-three homologous chromosome pairs in the first stage of meiosis yields more than 8 million possible combinations of gametes with one or more different sets of chromosomes. Along with an equivalent number

mitosis Process of cell division that takes place in most cells of the human body and results in a full complement of identical material in the forty-six chromosomes in each cell.

meiosis Process of cell division that forms the gametes; normally results in twenty-three chromosomes in each human egg and sperm cell rather than the full complement of forty-six chromosomes.

FIGURE 3.3 The Process of Mitosis

The process of mitotic cell division generates nearly all the cells of the body except the gametes. During mitosis, each chromosome replicates to form two chromosomes with identical genetic blueprints. As the cell divides, one member of each identical pair becomes a member of each daughter cell. In this manner, complete genetic endowment is replicated in nearly every cell of the body.



Cell nucleus (shown with a single pair of chromosomes).

Chromosomes split and replicate to produce two identical replicas of each chromosome.

The replicas separate, and the cell divides.

Each daughter cell now has a pair of chromosomes that is identical to the original pair.

FIGURE 3.4 The Process of Meiosis for Sperm Cells

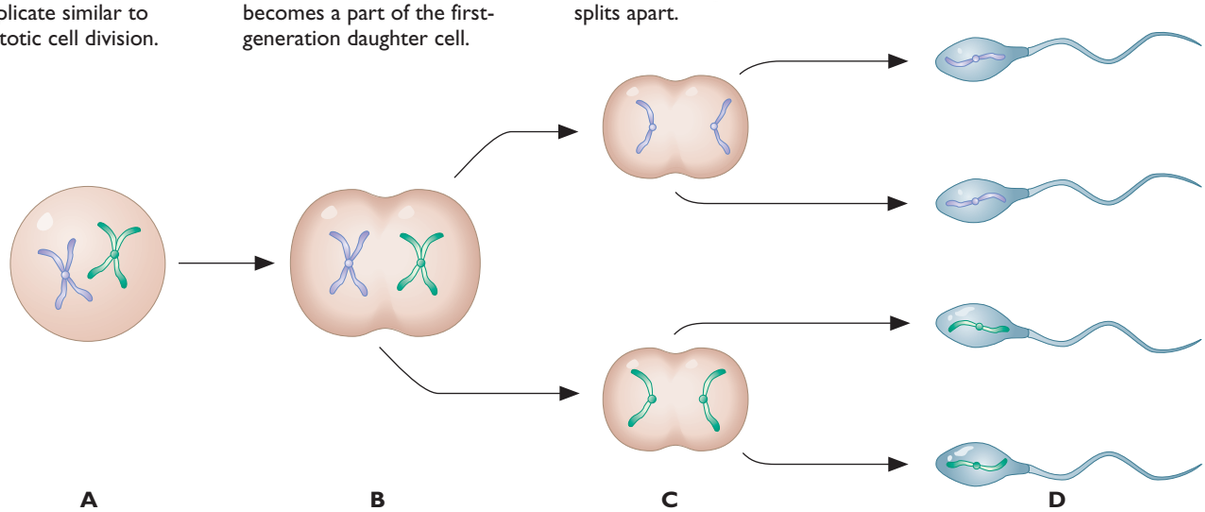
As meiosis begins (A), DNA replicates as in mitotic cell division. However, before the replicated arms split apart, one member of each pair of homologous chromosomes moves to become part of each first-generation daughter cell (B). Once the first generation of daughter cells is established, DNA replicas split as part of the second meiotic division (C). Thus one replica of one member of the pair of homologous chromosomes is contributed to each second-generation daughter cell (D). From these two successive divisions, four cells, each with twenty-three chromosomes, are produced.

Cell with forty-six chromosomes (only one pair of homologous chromosomes is shown here). Each member of the pair has begun to replicate similar to mitotic cell division.

First meiotic cell division begins but does not proceed as in mitosis. Instead of the replicated chromosome splitting apart, one member of each homologous pair becomes a part of the first-generation daughter cell.

The second meiotic division proceeds after the first is completed; now the replicated chromosome acquired in the first-generation daughter cell splits apart.

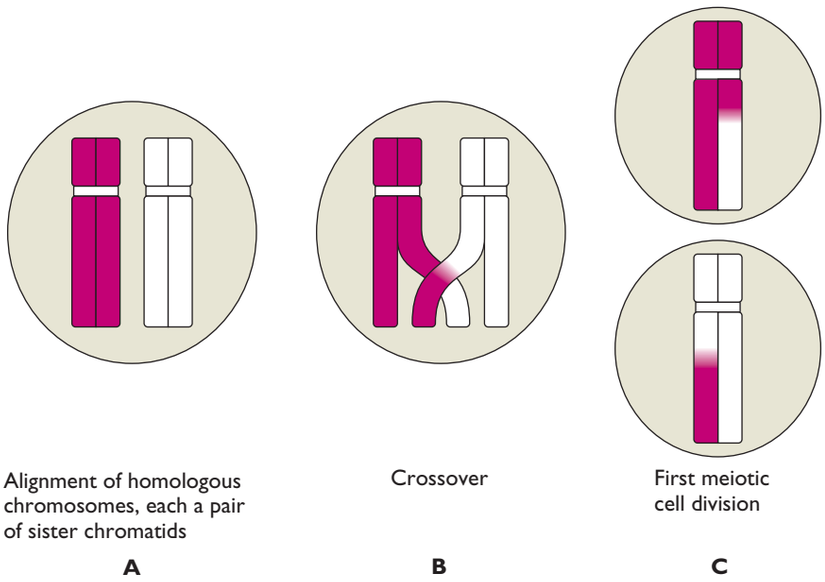
Each of the four gametes produced by the two-step process now has acquired one member of the pair of homologous chromosomes.



A B C D

FIGURE 3.5
Crossing Over: The Exchange of Genetic Material Between Chromosomes

In the process known as crossing over, genetic material is exchanged between homologous pairs of chromosomes during the first stage of meiotic cell division. (A) Initially, autosomes that have begun DNA replication align with each other. (B) Genetic material between homologous chromosomes is exchanged. (C) One member of each homologous pair of chromosomes randomly segregates or relocates to two different regions of the parent cell, and the first generation of cell division in meiosis takes place.



WHAT DO YOU THINK?

Should Research on Cloning Humans Be Permitted?

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of possible unique arrangements from a mate, mother and father together have a gene pool of about 64 trillion different combinations from which their offspring may derive. But the potential for genetic variability is actually far greater because of the phenomenon known as **crossing over,** a key part of the first stage of meiosis. Before homologous chromosome pairs separate in the first cell division, they mysteriously align, and segments of DNA transfer, or cross over, from one member to the other member of the pair, as shown in Figure 3.5. The genetic variability ensured by crossing over makes it virtually impossible for two individuals to have the same genetic makeup, even siblings, unless the two are identical twins.

Gene Expression

We have briefly described key structures of inheritance—nucleotides, genes, and chromosomes—and the way these are replicated in cells of the body, including gametes. But how does the genotype affect the phenotype? That is, how does the underlying genetic blueprint promote the appearance of blue eyes, baldness, and dark skin or such complex traits as shyness, schizophrenia, and intelligent problem solving? The answer begins with the alleles, the specific form a particular gene may take.

We have already learned that each of us typically inherits two genes that code for a particular protein in the cell, one from our mother and the other from our father. These may be identical—that is, have the same allelic form—or they may differ. When both have the same allelic form, a person's genotype is said to be **homozygous** for whatever characteristic that gene affects. For example, three different alleles exist for the gene that governs blood type: A, B, and O. When both inherited alleles are A, both B, or both O, a person has a homozygous genotype for blood type. But if an individual inherits two different alleles of the gene for blood type, let's say A and B, that person's genotype is **heterozygous;** he or she has Type AB blood.

The consequences of a homozygous genotype are usually straightforward: the child's phenotype will be influenced by whatever characteristics are specified by that particular allelic form. But the effects of a heterozygous genotype depend on how the alleles influence each other. When a child's phenotype shows the effects of only one of the two allelic forms, the one whose characteristics are observed is **dominant;** the allelic form whose influence is not evident in the phenotype is **recessive.** For example, a person who inherits both an A and an O allele for blood type will still be classified as having Type A; the allele for Type A is dominant and the allele for Type O recessive.

crossing over Process during the first stage of meiosis when genetic material is exchanged between autosomes.

homozygous Genotype in which two alleles of a gene are identical, thus having the same effects on a trait.

heterozygous Genotype in which two alleles of a gene are different. The effects on a trait will depend on how the two alleles interact.

dominant allele Allele whose characteristics are reflected in the phenotype even when part of a heterozygous genotype. Its genetic characteristics tend to mask the characteristics of other alleles.

recessive allele Allele whose characteristics do not tend to be expressed when part of a heterozygous genotype. Its genetic characteristics tend to be masked by other alleles.

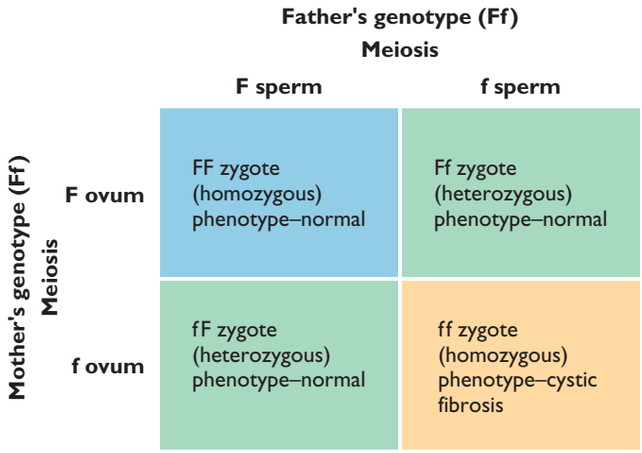

FIGURE 3.6
The Pattern of Inheritance for Cystic Fibrosis

The inheritance of cystic fibrosis is one of many traits and diseases that are influenced by a single pair of genes. In this figure, F symbolizes a normal allele and f represents the allele for cystic fibrosis. When parents with a heterozygous genotype for this disease have children, their offspring may inherit a homozygous genotype with normal alleles (FF), a heterozygous genotype with one normal and one abnormal allele (Ff or fF), or a homozygous genotype with two abnormal alleles (ff). Because the normal allele dominates, children with a heterozygous genotype will not exhibit cystic fibrosis. When both alleles carry genetic information for the disease, however, cystic fibrosis will occur.

Cystic fibrosis, the most common autosomal recessive disorder in Western Europe (Mueller & Cook, 1997) and a leading cause of childhood death among Caucasian children, provides another example of a dominant-recessive relationship between alleles. The shortened lifespan typically stems from a thickening of the mucus lining the respiratory tract that interferes with breathing. Most Caucasian children inherit a gene pair that does not include the allelic form that codes for cystic fibrosis; they have a homozygous genotype that contributes to normal development. About one in twenty-five people of Caucasian ancestry, however, has a heterozygous genotype in which one gene is normal and the other carries the genetic information that results in cystic fibrosis. The normal allele is *dominant.* Thus someone who is heterozygous for this condition can lead an ordinary, productive life. But a child of a mother and father, each of whom has a heterozygous genotype, may inherit either two normal alleles, both a normal and an abnormal allele, or two alleles coding for cystic fibrosis (see Figure 3.6). In the latter homozygous condition, the two recessive alleles are no longer masked by a normal gene; this child (about 1 in every 2,500 Caucasian children) will suffer from cystic fibrosis. Medical researchers today are actively investigating the potential for *gene therapy,* the replacement of the gene that codes for a disorder, to reduce and even eliminate the devastating consequences of cystic fibrosis and other inherited diseases (Friedmann, 1997).

KEY THEME
Individual Differences

For other genes, the child's phenotype will reflect the influence of both allelic forms if they differ. When the characteristics of both alleles are observed, they exhibit **codominance.** For example, a child with Type AB blood has inherited a gene for Type A blood from one parent and another gene for Type B blood from the other parent.

Table 3.1 summarizes a number of traits and characteristics of individuals who are affected by genes exhibiting dominant-recessive patterns. But we must be cautious when drawing inferences about these relationships. Many traits are **polygenic,** that is, determined by several genes, each located, perhaps, on different sets of chromosomes. For example, even eye color, although largely governed by the dominant-recessive relationship between allelic forms of a single gene, as suggested in Table 3.1, is affected by other genes as well.

Gene Functioning and Regulation of Development

How do genes influence the development of a phenotype? A major new field of study called *proteomics* has emerged precisely to attempt to answer that question (Ezzell, 2000). We can only give a brief glimpse into this rapidly evolving area. Although exceptions exist, genetic information is typically conveyed from the DNA in the cell's nucleus to the organic and inorganic substances in other parts of the cell. This

codominance Condition in which individual, unblended characteristics of two alleles are reflected in the phenotype.

polygenic Phenotypic characteristic influenced by two or more genes.

TABLE 3.1 Alleles of Genes That Display a Dominant and Recessive Pattern of Phenotypic Expression

Dominant Traits	Recessive Traits
Brown eyes	Gray, green, blue, hazel eyes
Curly hair	Straight hair
Normal hair growth	Baldness
Dark hair	Light or blond hair
Nonred hair (blond, brunette)	Red hair
Normal skin coloring	Albinism (lack of pigment)
Immunity to poison ivy	Susceptibility to poison ivy
Normal skin	Xeroderma pigmentosum (heavy freckling and skin cancers)
Thick lips	Thin lips
Roman nose	Straight nose
Earlobe free	Earlobe attached
Cheek dimples	No dimples
Extra, fused, or short digits	Normal digits
Second toe longer than big toe	Big toe longer than second toe
Double-jointedness	Normal joints
Normal color vision	Red-green colorblindness
Farsightedness	Normal vision
Normal vision	Congenital eye cataracts
Retinoblastoma (cancer of the eye)	Normal eye development
Normal hearing	Congenital deafness
Type A blood	Type O blood
Type B Blood	Type O blood
Rh-positive blood	Rh-negative blood
Normal blood clotting	Hemophilia
Normal metabolism	Phenylketonuria
Normal blood cells	Sickle cell anemia
Familial hypercholesterolemia (error of fat metabolism)	Normal cholesterol level at birth
Wilms tumor (cancer of the kidney)	Normal kidney
Huntington's disease	Normal brain and body maturation
Normal respiratory and gastrointestinal functioning	Cystic fibrosis
Normal neural and physical development	Tay-Sachs disease

process is performed by *messenger ribonucleic acid*, or *mRNA*, a molecule somewhat similar to DNA. mRNA replicates some segment of the DNA. This copy is transported outside the nucleus of the cell, where it can then initiate a series of events that produce proteins to give the cell its unique ability to function. Important to remember here is that the genes do not directly cause appearance, behavior, or any other phenotypic expression. Instead, our appearance and behavior are, in part, the end result of an extensive chain of biochemical processes started by the instructions provided by the DNA.

The instructions conveyed by different alleles of a gene may cause one or more biochemical events in the chain to be modified or disrupted. Such a disruption occurs, for example, in **phenylketonuria (PKU),** a genetic condition in which *phenylalanine,* an amino acid in milk and high-protein foods such as meat, cannot be metabolized normally by the liver. As a result, phenylalanine and other metabolic products accumulate in the blood, and the nervous system becomes deprived of needed nutrients. The eventual consequences are often convulsions, severe mental retardation, hyperactivity, and other behavioral problems. Remember, however, that a phenotype is the product of the interaction between genotype and environment. In the case of PKU, intervention in the form of reducing phenylalanine in the diet can help prevent severe mental retardation. Here, then, is an excellent example illustrating that genes do not have all the information built into them to cause particular developmental outcomes; environmental factors interact with the genotype to yield a specific phenotype.

KEY THEME
Nature/Nurture

Many mysteries remain concerning how genes influence development. For example, humans are believed to have between twenty-six and thirty-eight thousand *structural genes* that code for the production of proteins that govern the physiological functions of a cell (Pääbo, 2001). Yet structural genes account for only 1 to 2 percent of the nearly 3 billion base pairs estimated to make up the human genome (Pennisi, 2001). Some of the remaining DNA, for example, consists of other types of genes that start and stop or modify the functioning of structural genes. But large stretches of DNA are made up of repeat sequences of base pairs or of other patterns that seem to have simply replicated themselves and the functions of which, if any, remain unknown.

Other new discoveries are continuing to be made about genes and how they influence development. For example, sometimes it matters whether a particular gene has been inherited from the mother or the father. This phenomena, called **genomic imprinting,** is best illustrated by the *Prader-Willi* and *Angelman* syndromes, estimated to affect about one in ten thousand persons. Individuals with Prader-Willi syndrome display, among other physical and behavioral characteristics, short stature, obesity, and mild to moderate mental retardation. In contrast, individuals with Angelman syndrome display disturbances in gait suggestive of marionettelike movements, epilepsy, and more severe learning difficulties, including minimal or no speech. Prader-Willi syndrome stems from the absence of a particular gene or set of genes on chromosome 15 inherited from the father and the inability of the mother's genetic material on the homologous chromosome to compensate for this loss. In contrast, Angelman syndrome arises from the absence of that same gene or set of genes inherited from the mother and the inability of the father's genetic material to compensate for the loss (Everman & Cassidy, 2000; Lombroso, 2000). Susceptibility to certain cancers, growth disorders, and some types of diabetes are also known to occur as a result of genomic imprinting.

KEY THEME
Individual Differences

Substantial progress in understanding genetic influences has been made in recent years. However, important questions still exist concerning the effects of the human genome on a wide range of complex human behaviors of interest to psychologists. In the section that follows, we highlight additional examples of several specific gene mutations as well as chromosomal disturbances that can have profound repercussions for development. Keep in mind, however, that serious consequences associated with gene and chromosomal deviations affect a relatively small number of individuals. Nevertheless, the consequences often reverberate and extend well beyond those individuals and to their family and others within the community.

phenylketonuria (PKU) Recessive genetic disorder in which phenylalanine, an amino acid, fails to be metabolized. Unless dietary changes are made to reduce intake of phenylalanine, severe mental retardation occurs.

genomic imprinting Instances of genetic transmission in which the expression of a gene is determined by whether the particular allelic form has been inherited from the mother or the father.

FOR YOUR REVIEW

- What roles do genotype and the environment play in determining a phenotype?
- What is the human genome? How do the nucleus of the cell, chromosomes and DNA, genes, and nucleotides play a role in genetic influences on development?
- How many autosomes exist in the human karyotype? Of what importance are the X and Y chromosomes?

- What is the difference between mitosis and meiosis, and what is the impact of the phenomenon of crossing over?
- How do homozygous and heterozygous genes and the presence of dominant, recessive, and codominant allelic forms account for the inheritance patterns associated with various phenotypes? What are polygenic traits?
- How do genes regulate the development of the genotype? What is genomic imprinting?

KEY THEME
Individual Differences

Gene and Chromosomal Abnormalities

Changes in the structure of genes, or **mutations,** introduce genetic diversity among individuals. Mutations occur surprisingly often; perhaps as many as half of all human conceptions occur with some kind of genetic or chromosomal change (Plomin, DeFries, & McClearn, 1990). Most mutations are lethal, resulting in loss of the zygote through spontaneous abortion very soon after conception, often before a woman even knows she is pregnant. A small number of other mutations will have little impact on development. However, some can have enduring, often negative, consequences for an individual and his or her quality of life, consequences that may be passed on from one generation to the next. In fact, more than 5 percent of all diseases observed in individuals before the age of twenty-five are at least in part the result of some type of genetic or chromosomal anomaly (Rimoin, Connor, & Pyeritz, 1997). Moreover, birth defects and genetically based diseases contribute to a disproportionately high number of hospitalizations and medical expenses in the United States (Yoon et al., 1997). We consider here the consequences of just a few of the more than fourteen hundred gene and chromosome anomalies that have been identified as influencing physical and behavorial development (Peltonen & McKusick, 2001).

Gene Variations

An estimated 100,000 infants are born each year in the United States alone with some kind of problem caused by a single dominant or recessive gene. For about twenty thousand of these babies, the problem is serious (Knowles, 1985). Table 3.2 lists a few of the more serious of these. In many cases, the effects are evident at birth (*congenital*), but the consequences of some are not observed until childhood or even late adulthood. We will discuss several dominant and recessive disorders to illustrate their effects on development, the interventions and treatments available for them, and the insights they provide concerning the genotype's contribution to intellectual and behavorial capacities.

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Gene Locations for Various Disorders

mutation Sudden change in molecular structure of a gene; may occur spontaneously or be caused by an environmental event such as radiation.

Williams syndrome Dominant genetic disorder involving the deletion of a set of genes that results in affected individuals typically having a strong social orientation, good musical ability, and some unusual linguistic capabilities; accompanied by mental retardation and severe deficits in numerical and spatial ability.

● **Williams Syndrome: Discordances in Language, Cognition, and Social Behavior** About one in 10,000 children are born with **Williams syndrome,** caused by the deletion of a small number of genes on chromosome 7. The syndrome is autosomal dominant, although most occurrences are the result of a mutation. Children with Williams syndrome possess a distinctive set of facial features including a short, upturned nose and full lips. They also display knee and hip inflexions that produce an unusual postural appearance and gait, and they often have heart and kidney abnormalities.

Individuals with Williams syndrome are typically mildly to moderately retarded. Perhaps most puzzling, however, is their strikingly uneven profile of cognitive and social strengths and weaknesses. For example, as young children they seem especially preoccupied with the faces of adults and show relatively few social inhibitions, even among strangers, despite evidence of underlying anxiety in some contexts (Mervis, 2001). When young, they are also extremely sensitive to certain sounds, such as those made by a drill or vacuum cleaner, or the loud noises produced by fireworks or the bursting of a balloon. Their ability to acquire language is initially slow and may never reach a high

The smiling face of this girl with Williams syndrome epitomizes one of the behavorial characteristics common to such children, a strong orientation to initiating and maintaining social relationships. Their unusual pattern of strengths and weaknesses in various areas of cognition and language as well as their inquisitiveness about other people are of considerable interest to those who study child development. Such observations support the view that some aspects of development may be domain specific rather than more broadly determined by intelligence or personality.

TABLE 3.2 Some Inherited Gene Syndromes

Syndrome	Estimated Frequency (live births in U.S.)	Gene Located on Chromosome	Phenotype, Treatment, and Prognosis
Autosomal Dominant Syndromes			
Huntington's Disease	1 in 10,000–20,000	4	Personality changes, depression, gradual loss of motor control and memory caused by massive neuronal cell death that often begins in mid-adulthood. Thus affected individuals may transmit the disease to another generation of offspring before becoming aware of the disease. In some individuals, symptoms may begin earlier and be more severe if the dominant gene is transmitted by the father, another example of genomic imprinting.
Marfan Syndrome	1 in 10,000–20,000	15	Tall, lean, long limbed, with gaunt face (some believe Abraham Lincoln had syndrome). Frequent eye and heart defects. Cardiac failure in young adulthood common. Suicide second most common cause of death. Associated with increased paternal age.
Neurofibromatosis Type I	1 in 3,500	17	Symptoms range from a few pale brown spots on skin to severe tumors affecting peripheral nervous system and distorting appearance. Minimal intellectual deficits in about 40% of cases. Other forms of neurofibromatosis are associated with genes located on other chromosomes.
Williams Syndrome	1 in 10,000	7	See text.

(continued)

TABLE 3.2 Some Inherited Gene Syndromes *(continued)*

Syndrome	Estimated Frequency (live births in U.S.)	Gene Located on Chromosome	Phenotype, Treatment, and Prognosis
Autosomal Recessive Syndromes			
Albinism	1 in 10,000–20,000. Several forms. Most common occurs in about 1 in 15,000 African Americans, 1 in 40,000 Caucasians, but much more frequently among some Native American tribes (1 in 200 among Hopi and Navajo, 1 in 132 among San Blas Indians of Panama).	11 (also 15)	Affected individuals lack pigment *melanin*. Extreme sensitivity to sunlight and visual problems.
Cystic Fibrosis	Most common genetic disease in Caucasian populations in U.S., especially those of Northern European descent, affecting about 1 in 2,500. Less common among African American and Asian American populations.	7	Respiratory tract becomes clogged with mucus; lungs likely to become infected. Death often in young adulthood, but individuals may have children. Prognosis for females poorer than for males. Pulmonary therapy to remove mucus accumulation in lungs helps delay effects.
Galactosemia	1 in 60,000	9	Mental retardation, cataracts, cirrhosis of the liver caused by accumulation of galactose in body tissues because of absence of enzyme to convert this sugar to glucose. Heterozygous individuals have half the normal enzyme activity, enough for normal development. Galactose-free diet is only treatment, although many still display learning and behavioral problems.
Gaucher Disease	1 in 600 Ashkenazic Jews. Other, rarer forms found in all populations.	1	Enlarged spleen contributing to pain, cardiac failure, and failure to thrive. Frequent bone fractures, bruising, and bleeding. Limited treatment available.
Phenylketonuria	1 in 15,000. Somewhat higher rate of incidence in Caucasian than in Asian or African American populations.	12	See text.
Sickle Cell Disease	1 in 400 African Americans. Also frequently found in malaria-prone regions of world.	11	See text.
Tay-Sachs Disease	1 in 3,600 Ashkenazic Jews. Very rare in other populations.	15	Signs of mental retardation, blindness, deafness, and paralysis begin 1 to 6 months after birth. No treatment available. Death normally occurs by 3 or 4 years of age.
ß–Thalassemia (Cooley's anemia)	1 in 800–3,600 in populations of Greek and Italian descent. Much less frequent in other populations.	11	Severe anemia beginning within 2 to 3 months of birth, stunted growth, increased susceptibility to infections. Death usually occurs in 20s or 30s.

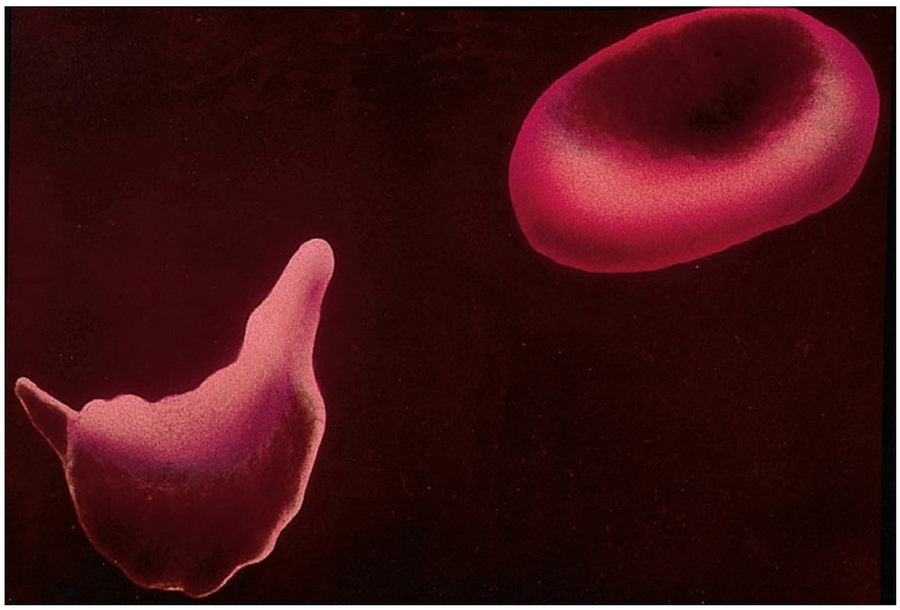
TABLE 3.2 Some Inherited Gene Syndromes *(continued)*

Syndrome	Estimated Frequency (live births in U.S.)	Gene Located on Chromosome	Phenotype, Treatment, and Prognosis
Sex-Linked Syndromes			
Colorblindness (red-green)	About 1 in 100 males of Caucasian descent see no red or green. About 1 in 15 Caucasian males experience some decrease in sensitivity to red or green colors.	X	If completely red-green colorblind, lack either green-sensitive or red-sensitive pigment for distinguishing these colors and see them as yellow. If decreased sensitivity to red or green, reds are perceived as reddish browns, bright greens as tan, and olive greens as brown.
Duchenne Muscular Dystrophy	1 in 3,500 males. Most common of many different forms of muscular dystrophy. Several forms, including Duchenne, are X linked.	X	Progressive muscle weakness and muscle fiber loss. Mental retardation in about ⅓ of cases. No cure, and few live long enough to reproduce. Responsible gene located on short arm of X chromosome; appears to be massive in number of nucleotide pairs.
Fragile X Syndrome	1 in 4000 males; 1 in 8,000 females.	X	See text.
Hemophilia A	1 in 10,000 Caucasian male births for the most common form.	X	Failure of blood to clot. Several different forms; not all are sex linked. Queen Victoria of England was carrier for the most common form. Potential for bleeding to death, but administration of clot-inducing drugs and blood transfusions reduces hazard.

Sources: Adapted from Beaudet et al., 1995; Committee on Genetics, 1996; Laskari, Smith, & Graham, 1999; McKusick & Amberger, 1997; Scriver et al., 1995.

level of grammatical complexity. However, children with Williams syndrome accumulate a surprisingly large vocabulary that permits them to engage in relatively sophisticated, although somewhat stereotyped, verbal interactions (Moldavsky, Lev, & Lerman-Sagie, 2001). Complementing these verbal abilities tend to be some rather uncommon abilities with respect to creating and imitating music (Levitin & Bellugi, 1998). For example, some, after hearing a selection only once and regardless of the language in which it may have been sung, are able to reproduce the piece with extraordinary skill. Despite these strengths, children with Williams syndrome show poor visual and spatial abilities, planning and problem solving, and little competence in acquiring numerical skills, although some can achieve relatively high levels of reading ability.

Individuals with Williams syndrome have become of special interest to developmental, cognitive, and social psychologists because of their unusual, and quite uneven, profile of intellectual and behavioral strengths and weaknesses. As is shown in the chapter titled "Cognition: Piaget and Vygotsky," many cognitive abilities may be modular; that is, they may undergo relatively specific patterns of development. Extensive debate occurs as well about whether to best view the nature of intelligence as a general capacity or as a set of more specific abilities (see the chapter titled "Intelligence"). The observations being carried out on children with Williams syndrome are helping to provide valuable new insights about the ways in which genes may have highly targeted consequences for intellectual, social, and other developmental capacities and about how best to conceptualize the concepts of cognition and intelligence.



Individuals who suffer from sickle cell anemia, a genetically inherited disorder, have a large proportion of crescent-shaped red blood cells like the one shown at the bottom left. A normal red blood cell (upper right) is round and doughnut-shaped. Sickle-shaped cells are ineffective in transporting oxygen and may cause damage to various organs and pain by blocking small blood vessels.

● **Sickle Cell Disease: A Problem Arising Out of Adaptive Circumstance** **Sickle cell disease** is a genetic disorder whose incidence is extremely high in many regions of West Africa and around the Mediterranean basin. Sickle cell disease is also found in about 1 out of every 400 African Americans (Ashley-Koch, Yang, & Olney, 2000) and in high numbers of Greek Americans and others whose ancestors came from regions in which malaria commonly occurs. The defect introduces a change in a single amino acid in hemoglobin, the molecule that permits the red blood cells to carry oxygen. As a result, red blood cells become crescent shaped rather than round.

Sickle-shaped cells are ineffective in transporting oxygen; they also survive for a much shorter duration than normal red blood cells, and the bone marrow has difficulty replacing them. The consequences are often anemia, jaundice, low resistance to infection, and susceptibility to stroke, severe pain, and damage to various organs when the distorted cells block small blood vessels. Blood transfusions can help to alleviate the more serious problems, and recent research in both Europe and the United States raises the possibility that bone marrow transplants can provide a cure (Davis & Roberts, 1996). Despite the physical limitations, elementary school children with sickle cell disease appear quite similar to unaffected peers in terms of emotional well-being and view themselves no differently in terms of social satisfaction, competencies, and feelings of depression. Still, children with sickle cell disease, especially girls, tend to be somewhat less popular in the classroom and boys somewhat less aggressive, perhaps because of their more limited energy and slower physical development (Noll et al., 1996).

About one in every twelve African Americans are carriers of the sickle cell gene. These individuals, who possess a heterozygous genotype, have the **sickle cell trait.** They manufacture a relatively small proportion of cells with abnormal hemoglobin. Few of these individuals show symptoms of sickle cell disease; most live normal lives. But insufficient oxygen, which may occur in high-altitude regions, when flying in unpressurized airplane cabins, or after strenuous exercise, can trigger sickling of red blood cells in those who have the trait. Nevertheless, carriers of the sickle cell gene are more resistant to malaria than are individuals who have normal hemoglobin—an adaptive feature that accounts for the high incidence and persistence of the trait in populations in which malaria is present.

sickle cell disease Genetic blood disorder common in regions of Africa and other areas where malaria is found and among descendents of the people of these regions. Abnormal blood cells carry insufficient oxygen.

sickle cell trait Symptoms shown by those possessing a heterozygous genotype for sickle cell anemia.

● **Phenylketonuria: A Genetic Problem Modifiable by Diet** Phenylketonuria (PKU), a recessive metabolic disorder, provides a good illustration of how changing

the child's environment, in this case diet, can reduce its more harmful consequences. An infant with PKU appears normal at birth. However, retardation sets in soon thereafter and becomes severe by four years of age if the condition is untreated. Fortunately, screening using blood samples collected a day or two after birth can detect elevated levels of phenylalanine. An infant identified as having PKU can then be placed on a diet low in phenylalanine to prevent its more serious effects. Experts agree the diet must be started relatively early, within the first few weeks after birth, and continued at least through adolescence to ensure nearly normal mental development (Phenylketonuria, 2000).

KEY THEME
Nature/Nurture

Even though the more serious consequences of phenylketonuria can be prevented, a completely normal prognosis for these children remains problematic. The diet is difficult to maintain; it requires a careful balance between excessive phenylalanine to prevent neural damage and sufficient nutrients. Weekly blood tests may be necessary to keep metabolite concentrations within an acceptable range, a regimen for which child, parents, and testing centers may be ill prepared. The bland and unappetizing diet can be a source of conflict between child and caregiver as well, creating management problems within households attempting to lead relatively normal lives (Phenylketonuria, 2000).

KEY THEME
Child's Active Role

Even under optimal conditions, children with PKU may show some growth and intellectual deficiencies. For example, these children seem to have difficulty in planning and problem-solving tasks in which working memory or sustained attention is required to inhibit well-learned, simpler reactions in order to master new, more complex responses (Diamond et al., 1997; Ris et al., 1994). Moreover, individuals with PKU who successfully reach adulthood still need to be concerned about their diets. For example, children born to mothers who continue to display elevated levels of phenylalanine during pregnancy show increased risk for heart defects and mental retardation (Platt et al., 2000; Rouse et al., 2000). If a mother returns to a low-phenylalanine diet before or early in her pregnancy, the risks can be reduced substantially. Although dietary modifications are helpful, it remains unclear whether this intervention completely eliminates some negative consequences of PKU.

● **Sex-Linked Syndromes** As already indicated, only a few dozen genes may be located on the Y chromosome, whereas the X chromosome carries many. This imbalance has substantial implications for a number of phenotypes said to be sex linked because the gene associated with them is carried only on the X chromosome. Hemophilia, red-green colorblindness, and Duchenne muscular dystrophy (see Table 3.2) have nothing to do with differentiation of sex but are sex linked because they are inherited via specific genes on the X chromosome. Thus, they occur much more frequently in males than in females.

As with genes for autosomes, those that are sex linked often have a dominant-recessive relationship. Females, who inherit two genes for sex-linked traits, one on each X chromosome, are much less likely to display the deleterious effects associated with an abnormal recessive gene than are males, who, if they inherit the damaging allele, have no second, normal allele to mask its effects. Hemophilia, a condition in which blood does not clot normally, is a good example because it is nearly always associated with a defective gene on the X chromosome. Because the allele for hemophilia is recessive, daughters who inherit it typically do not exhibit hemophilia; the condition is averted by an ordinary gene on the second X chromosome that promotes normal blood clotting. A female can, however, be a carrier. If she possesses a heterozygous genotype for hemophilia, the X chromosome with the abnormal allele has a fifty-fifty chance of being transmitted to either her son or her daughter. When a son inherits the abnormal allele, he will exhibit hemophilia because the Y chromosome does not contain genetic information to counter the allele's effects. If a daughter inherits the abnormal allele, she will be a carrier who may then transmit it to her sons and daughters, as has occurred in several interrelated royal families of Europe.

Fragile X Syndrome: A Sex-Linked Contributor to Mental Retardation

Geneticists have identified a structural anomaly that consists of a pinched or constricted site near the end of the long arm of the X chromosome in some individuals (see Figure 3.7). This anomaly, termed **fragile X syndrome,** may be the most frequently inherited source of mental retardation associated with a specific gene (Moldavsky et al., 2001). Males with fragile X syndrome commonly have a long, narrow face, large or prominent ears, and large testes. Cardiac defects and relaxed ligaments (permitting, e.g., hyperextension of finger joints) are also frequent components of the disorder. Behavioral attributes include poor eye contact and limited responsiveness to external stimulation, as well as hand flapping, hand biting, and other unusual mannerisms such as mimicry. Females who possess a heterozygous genotype often show, to a much lesser extent, some of the physical characteristics of the disorder. Many of these women display a normal or nearly normal level of intelligence, although, as with other sex-linked gene disorders, they are carriers for the syndrome.

An unusual feature of fragile X syndrome is that its severity seems to increase as the abnormal gene is passed on from one generation to the next, a phenomenon termed *anticipation.* This progression begins when one set of three nucleotides, which repeats between five and fifty times in the normal gene, for some reason expands to fifty to two hundred repetitions. Once this expansion begins, the gene seems to become unstable for subsequent offspring so that more copies of the three nucleotides are spewed out, as though the replication process has difficulty turning off (Eliez & Reiss, 2000). The inheritance of this unchecked expansion is accompanied by a spectrum of learning difficulties ranging from mild to severe mental retardation. Thus the size of the abnormal segment of the gene, along with the severity of the disorder, appears to increase as it is passed from a grandfather, in whom the initial amplification may occur (even if he shows no evidence of the disorder), to a daughter (who may be minimally affected because she has an additional X chromosome to compensate for the disorder), to a grandson (who now displays full-blown fragile X syndrome).

Chromosome Variations

Mutations in specific genes are only one of several sources of variation in the human genome. Occasionally whole sections of a chromosome are deleted, duplicated, or relocated to another chromosome, or an extra chromosome is transmitted to daughter cells during cell division. When this happens, normal development is often dis-

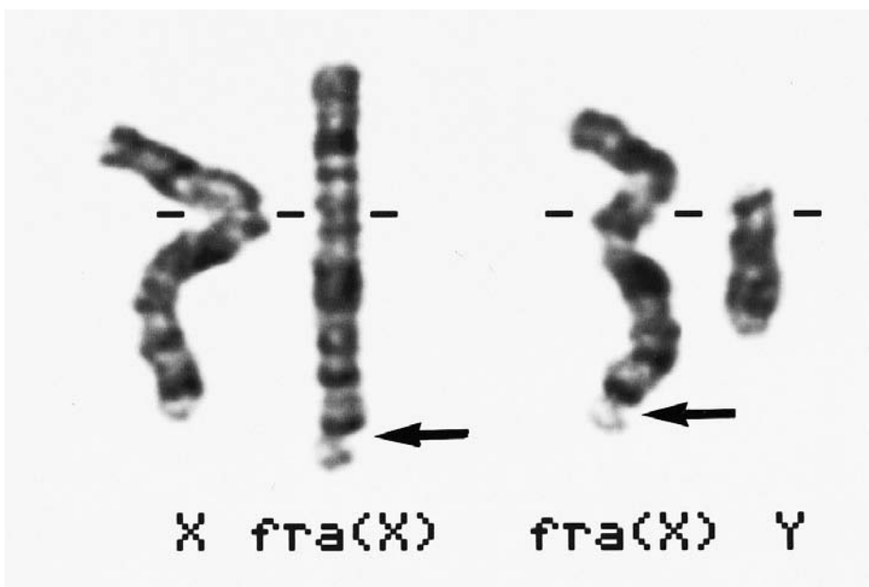


FIGURE 3.7
Chromosome Illustrating Fragile X Syndrome

Fragile X syndrome is one of the most frequently occurring genetic causes of mental retardation. This photomicrograph illustrates the pinched or constricted portion of one of the pair of X chromosomes in a heterozygous female and the X chromosome in an affected male.

fragile X syndrome Disorder associated with a pinched region of the X chromosome; a leading genetic cause of mental retardation in males.

rupted. Perhaps as many as half of all conceptions that result in spontaneous abortion include such chromosomal abnormalities (Garber, Schreck, & Carlson, 1997; Jacobs & Hassold, 1995). But this is not always the case. For example, the deletion of a small segment of the fifth chromosome is responsible for *cri du chat* or *cat-cry syndrome,* in which infants exhibit a cry similar to a cat's (hence its name), severe mental retardation, microcephaly (very small head size), short stature, and other congenital anomalies. Mental retardation and severe physical deformations often accompany structural aberrations observed in other chromosomes as well.

Human embryonic growth virtually never proceeds when a complete pair or even a member of one pair of autosomes is missing or when an extra pair of autosomes is inherited. **Trisomy,** the inheritance of an extra chromosome, also very often results in the loss of the zygote or miscarriage in early pregnancy (Jacobs & Hassold, 1995). However, three copies of chromosomes 13, 18, and 21 may be observed in surviving human newborns. Of these, trisomy 21, or Down syndrome, occurs most frequently. Even with this syndrome, however, fewer than 25 percent of conceptions survive to birth (Tolmie, 1997).

● **Trisomy 21 (Down Syndrome)** Trisomy 21, one of the most common genetic causes of mental retardation, arises in about one out of every eight hundred live births (Tolmie, 1997). Physically observable features include an epicanthal fold that gives an almond shape to the eye, flattened facial features, poor muscle tone, short stature, and short, broad hands, including an unusual crease of the palm. About 40 percent of infants with Down syndrome have congenital heart defects. Cataracts or other visual impairments, as well as deficiencies in the immune system, are also common. Physical development is slowed compared with normal children, as is intellectual development.

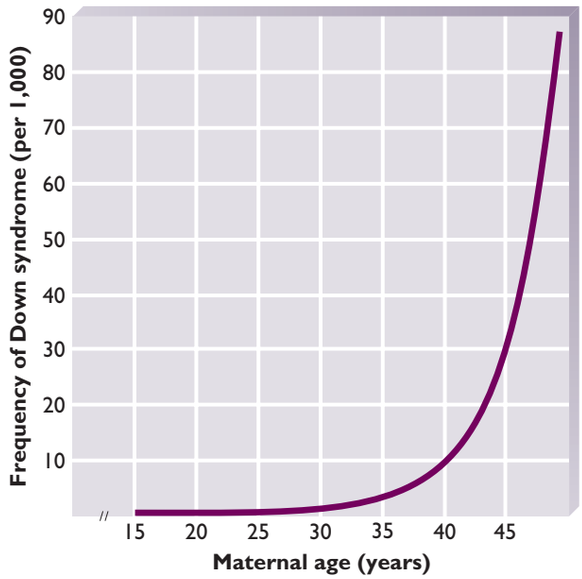
Approximately 95 percent of babies born with Down syndrome have an extra twenty-first chromosome. Nearly 90 percent of these errors originate in egg cells, and the remainder arise from errors during the production of sperm cells (Jacobs & Hassold, 1995). A small percentage of infants with Down syndrome have a segment of chromosome 21, perhaps as little as its bottom third, shifted to another chromosome (Moldavsky et al., 2001). Another small percentage display a mosaic genotype, that is, have chromosomal deviations in only a portion of their body cells. The severity of Down syndrome in these latter individuals seems to be related to the proportion of cells exhibiting trisomy.

This 13-year-old boy with trisomy 21, or Down syndrome, has learned to read. He has made considerable progress in academic subjects as is evident from his contribution to this geography project. By being provided with an enriching environment, many with Down syndrome are able to accomplish levels of skill that permit them to engage in meaningful work and be active members of their community.

trisomy Condition in which an extra chromosome is present.

FIGURE 3.8
Relationship Between Maternal Age and the Incidence of Down Syndrome

The incidence of Down syndrome increases dramatically as a function of the mother's age. One in every 1,500 babies born to a mother age twenty-one has Down syndrome. For forty-nine-year-old mothers, the incidence is much higher: one in every ten babies has Down syndrome. Several different explanations have been offered to account for these findings.



Source: Data from Epstein, 1989.

The probability of giving birth to an infant with trisomy 21 increases with the age of the mother, as is true for most other forms of trisomy (see Figure 3.8). Although mothers over thirty-five years of age give birth to only about 16 percent of all babies, they bear more than half of the infants with Down syndrome. The father's age shows virtually no relationship to the incidence of Down syndrome (Epstein, 1995). To explain these findings, experts have often proposed an "older egg" hypothesis. According to this view, the ova, which begin the first phases in meiosis even before the mother's own birth, change with age, either from the passage of time or perhaps because of increased exposure to potentially hazardous biological and environmental conditions. These older egg cells, released during ovulation in the later childbearing years, are then more susceptible to chromosomal errors while undergoing the final steps of meiosis. Other researchers have proposed a "relaxed selection" hypothesis to account for the increased frequency of Down syndrome in older mothers. According to this view, older mothers are less likely than younger mothers to spontaneously abort a zygote with trisomy 21. Still another view is that egg cells containing the extra chromosome are unlikely to be selected during ovulation. However, as they become a proportionally larger pool of the available ova over time, the possibility that one is released during ovulation increases (Tolmie, 1997).

KEY THEME
Individual Differences

Thanks to better medical and physical care, the majority of individuals born with Down syndrome may be expected to live more than fifty years (Tolmie, 1997). Many show considerable proficiency in reading and writing. But we still have much to discover about Down syndrome. For example, individuals with trisomy 21 who survive beyond age forty frequently develop the abnormal brain cells and show some of the same behavioral symptoms found in adults who acquire Alzheimer's disease (Janicki & Dalton, 2000). Alzheimer's disease is characterized by memory and speech disturbances, personality changes, and increasing loss of intellectual functioning, typically in individuals between fifty and seventy-five years of age, although the symptoms may begin much earlier. At least one form of Alzheimer's disease is thought to be inherited, and, not surprisingly, the responsible gene appears to be located on chromosome 21.

● **Sex Chromosome Syndromes** As we have already noted, males normally have an X and a Y sex chromosome and females have two X chromosomes. However, variations in the number of sex chromosomes as a result of errors during meiosis can occur in humans. For example, an individual may inherit only a single X, an extra chromosome (XXX, XXY, XYY), and, on rare occasions, even pairs of extra chromo-

somes (for example, XXXX, XXYY, XXXY). Table 3.3 describes several of these variations in more detail.

When it was first discovered that some humans possess an extra sex chromosome, these individuals were typically thought to display intellectual deficits, along with an assortment of abnormal and socially unacceptable behaviors. For example, in the 1960s some researchers claimed that an extra Y chromosome contributed to more aggressive and antisocial behavior. Males with an XYY chromosomal makeup tend to be taller than other males and display some learning difficulties but show a nearly normal range of intelligence. Furthermore, the majority of individuals who have this chromosomal pattern, along with those who possess other combinations of sex chromosome patterns, lead normal lives. In fact, variation in phenotypes associated with sex chromosome anomalies may be due as much, perhaps more, to experiential factors than to inheritance.

KEY THEME
Nature/Nurture

Bruce Bender and his colleagues at the University of Colorado School of Medicine studied forty-six children with variations in number of sex chromosomes such as those described in Table 3.3 (Bender, Linden, & Robinson, 1987). These children, born between 1964 and 1974, were identified by screening forty thousand consecutive births in the Denver area. Those with sex chromosome modifications were more likely to have

TABLE 3.3 Examples of Observed Sex Chromosome Syndromes

Disorder	Estimated Frequency (live births in U.S.)	Phenotype	Prognosis
45, XO (Turner syndrome)	1 in 2,500 females (more than 90% are spontaneously aborted); 80% of cases involve the absence of the paternal X chromosome.	Short stature, usually normal psychomotor development but limited development of secondary sexual characteristics. Failure to menstruate and sterility due to underdeveloped ovaries. About 50% have webbed, short neck. Near-average range of intelligence but serious deficiencies in spatial ability and directional sense.	Increased stature and sexual development, including menstruation, but not fertility, can be induced through administration of estrogen and other hormones. In vitro fertilization permits carrying of child when adult.
47, XXX (Triple-X syndrome)	1 in 1,200 females; 90% have received two copies of maternal X chromosome.	Not generally distinguishable. Some evidence of delay in speech and language development, lack of coordination, poor academic performance, and immature behavior. Sexual development usually normal. Tendency for tall stature.	Many are essentially normal, but substantial proportion have language, cognitive, and social-emotional problems.
47, XXY (Klinefelter syndrome)	1 in 600 males (increased risk among older mothers); 56% received two maternal chromosomes, 44% two paternal sex chromosomes.	Tend to be tall, beardless, with feminine body contour, high-pitched voice. Some evidence for poor auditory short-term memory and difficulty with reading. Testes underdeveloped, individuals usually sterile.	Many with normal IQ, but about 20% may have occasional mild to moderate retardation.
47, XYY (XXY syndrome)	1 in 1,000 males	Above-average height, some have learning disabilities, but near-average range of intelligence.	Most lead normal lives and have offspring with a normal number of chromosomes. Higher proportion than normal incarcerated, but crimes no more violent than those of XY men.

Sources: Adapted from Beaudet et al., 1995; Jacobs & Hassold, 1995; McGinniss & Kaback, 1997; McKusick & Amberger, 1997; Robinson & de la Chappelle, 1997.

neuromotor, psychosocial, language, and school problems than siblings who had a normal XX or XY complement. But this was true for school and psychosocial problems only if children were growing up in a family in which they experienced severe stress, such as exposure to drug abuse or considerable illness, lack of effective parenting by caregivers, or poverty. In the absence of such tensions, children with sex chromosome variations showed no greater evidence of school or psychosocial problems than their siblings, although they did continue to show more neuromotor and language impairment.

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Individual Differences

Bender and his colleagues followed the progress of thirty-nine of these children as they moved through adolescence (Bender et al., 1995). As Figure 3.9 shows, more of these students, compared with their siblings with a normal complement of sex chromosomes, needed special education assistance in high school and were less likely to graduate. This was especially true for girls with an extra X chromosome. These groups also exhibited somewhat more depression in psychiatric interviews and lower overall functioning and adaptation to adolescence. Even so, children with a mosaic pattern of abnormal sex chromosomes showed just as much progress in school as their siblings did. And, as in their earlier studies, the researchers note that the presence of a stable and supportive family environment seemed to promote more positive development in adolescents with sex chromosome variations, particularly those with XXY and X complements. These findings, then, further suggest that children and adolescents with different sex chromosome complements may be more vulnerable to disruptions in the caregiving environment than children with a normal complement of sex chromosomes.

FOR YOUR REVIEW

- What is the basis for each of the following genetically influenced disorders?

  Williams syndrome — Sickle cell disease and sickle cell trait
  Phenylketonuria — Fragile X syndrome

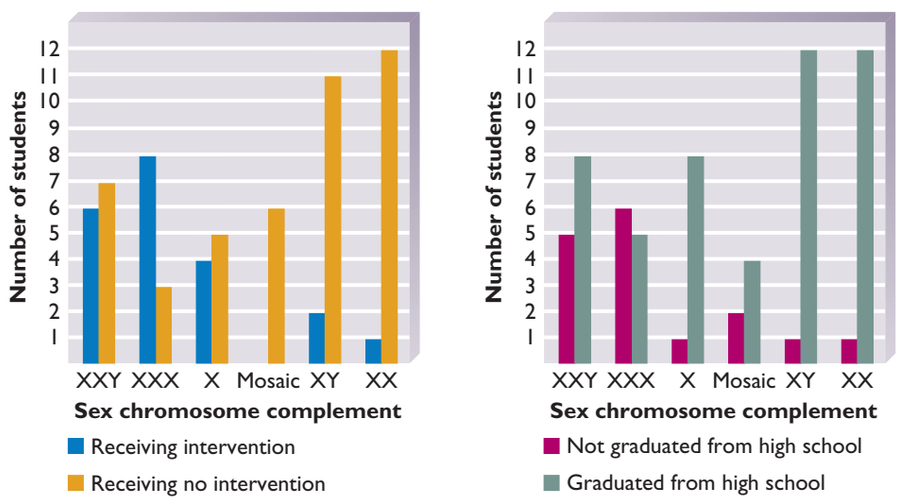
  What phenotypes are associated with each? Why is each of interest to developmental psychologists?

- What variations in the chromosomal makeup of an individual are possible? What is the most common example of chromosomal trisomy? What are examples of chromosomal variation associated with the sex chromosomes?

- What is the role of the environment in influencing behavioral outcomes for children with chromosomal variations?

FIGURE 3.9
Need for Educational Intervention Among Adolescents with Various Sex Chromosome Complements and Their Siblings

Adolescents who inherit an extra sex chromosome (X or Y) or only one X chromosome (Turner syndrome) often need more assistance in high school and may be somewhat less likely to graduate than their siblings who have a normal complement of sex chromosomes (XY or XX). Academic progress among high schoolers with a mosaic pattern of normal and abnormal sex chromosomes appears similar to that of their siblings. Closer inspection of the circumstances in which adolescents are reared also reveals that those with sex chromosome variations who are more successful academically live in families that provide more stable and supportive environments. Thus individuals with sex chromosome deviations may be more susceptible to disruptions or stress than their siblings.



Source: Adapted from Bender et al., 1995.

Genetic Counseling

Advances in detecting gene and chromosomal defects, as well as in understanding the biochemical and metabolic consequences of various inherited disorders, have led to a rapidly expanding medical and guidance specialty called **genetic counseling.** Obtaining a family history to summarize the occurrence of various diseases among ancestors and other relatives is usually the first step. If warranted, *parental* **genetic screening** may be carried out. For example, chromosomal abnormalities, as well as dominant and recessive genes associated with all of the disorders listed in Tables 3.2 and 3.3 (and many more not listed in these tables), can be detected through parental screening. Thus genetic counselors can provide prospective parents with estimates of the likelihood of bearing a child with a specific problem, although parental screening, of course, does not identify new mutations that may arise in offspring. Prospective parents can then use this information to decide whether, for example, adoption or other new reproductive technologies, such as those discussed in the chapter titled "The Prenatal Period and Birth," may be a better choice than bearing their own children.

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Genetic Counseling

Many reasons exist to carry out such tests. For example, prenatal testing is often recommended for women who are older than thirty-five because of the increased risk for Down syndrome; when a genetic disorder has been reported in the family history; if a previous child was born with a chromosomal variation or genetic disorder; if there has been difficulty in completing prior pregnancies; or if delayed or unusual development of the fetus occurs during pregnancy (Committee on Genetics, 1994). The finding of a serious inherited disorder may lead to a decision to terminate the pregnancy if religious and ethical values allow such a choice. However, prenatal screening tests, along with neonatal screening carried out shortly after birth, can serve another important purpose, that of suggesting therapy and treatment designed to prevent or minimize the more devastating consequences of some metabolic disorders (Erbe & Levy, 1997).

Prenatal Diagnosis

Procedures now exist that can be performed prenatally to detect hundreds of genetically and environmentally induced defects in fetal development. Some of these procedures provide unequivocal answers about the presence or absence of a problem. Other, often less invasive, procedures provide estimates of increased likelihood of the presence of some defect. If they are suggestive of a developmental disability, they are followed by more conclusive diagnostic tests. One of the best known definitive tests is **amniocentesis,** in which a small amount of amniotic fluid is withdrawn via a syringe inserted (under the guidance of an ultrasound image) in the woman's abdominal wall. Cells in the amniotic fluid are extracted and submitted to biochemical and chromosomal examination (see Figure 3.10). Amniocentesis is an especially effective procedure for detecting chromosomal variations, and it provides information about some metabolic problems as well. The test is usually performed between the thirteenth and eighteenth weeks of pregnancy. However, researchers are studying whether it can be safely carried out a few weeks sooner. Even when amniocentesis is performed later in pregnancy, the possibility of some increased risk exists from infections and spontaneous abortion (Elias & Simpson, 1997; Tongsong et al., 1998).

Another test that provides much the same information as amniocentesis but that can be carried out somewhat earlier in pregnancy (between ten and twelve weeks) is **chorionic villus sampling.** In this diagnostic procedure, a small sample of hairlike projections (*villi)* from the chorion, the outer wall of the membrane in which the fetus develops and that attaches to the woman's uterus, is removed by suction through a thin tube inserted through the vagina and cervix or, in some cases, through the abdominal wall. Information gained at this earlier time can reduce uncertainty and anxiety about the possibility of a developmental disability (Caccia et

genetic counseling Medical and counseling specialty concerned with determining and communicating the likelihood that prospective parents will give birth to a baby with a genetic disorder.

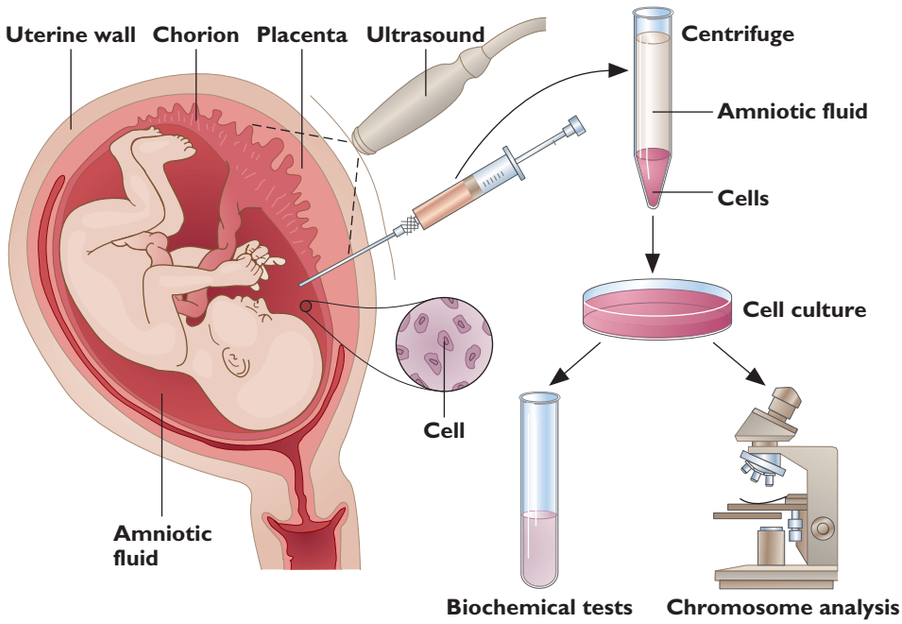
genetic screening Systematic search using a variety of tests to detect developmental risk due to genetic anomalies.

amniocentesis Method of sampling the fluid surrounding the developing fetus by insertion of a needle. Used to diagnose fetal genetic and developmental disorders.

chorionic villus sampling Method of sampling fetal chorionic cells. Used to diagnose embryonic genetic and developmental disorders.

FIGURE 3.10
The Process of Amniocentesis

In this prenatal screening procedure, a needle is inserted into the amniotic fluid surrounding the fetus. A small amount of fluid is withdrawn, and cells shed by the fetus are separated from the fluid by centrifuge. The cells are cultured and submitted to various biochemical and other tests to determine whether chromosomal, genetic, or other developmental defects exist.



Source: Adapted from Knowles, 1985.

al., 1991). However, chorionic villus sampling, in contrast to amniocentesis, does not provide information about possible neural tube defects, the procedure is somewhat more difficult to carry out unless performed by skilled technicians, and some researchers report a slightly increased risk of miscarriage and limb malformations (Elias & Simpson, 1997; Hsieh et al., 1995).

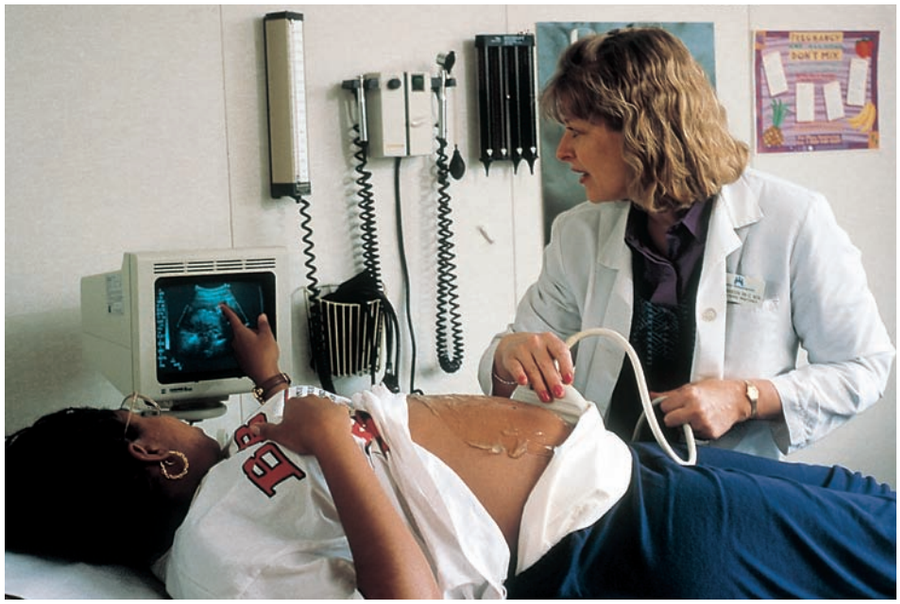
In **fetal blood sampling,** blood is directly withdrawn from the umbilical cord of the fetus for biochemical and chromosomal examination. This particular procedure, normally carried out from about the eighteenth week of pregnancy onward, permits the detection of various chromosomal and genetic errors and is especially useful in evaluating disorders associated with the blood. Although relatively safe, the procedure does appear to be slightly more risky than other invasive tests, although adequate evaluation of the risks remains to be carried out (Elias & Simpson, 1997).

Because of the possible increase in risk and their relatively high cost, amniocentesis, chorionic villus sampling, and fetal blood sampling are normally performed only when there is some reason to believe fetal abnormalities may occur. Testing can also be carried out to analyze fetal cells circulating in the pregnant woman's blood, but this and other new procedures remain experimental and of limited availability (Steele et al., 1996). Other, less invasive procedures are often performed to determine whether a fetal anomaly may exist. For example, several types of **maternal blood screening** tests such as the *alpha fetoprotein test* and the more extensive and accurate *triple screen test* can be carried out at around fifteen to twenty weeks of gestational age to provide evidence of increased risk for Down syndrome, neural tube defects, and certain kidney and other problems. Certainly the most widespread of the noninvasive diagnostic procedures, however, is **ultrasonography,** often called *ultrasound*. Ultrasound is now used routinely in many countries to help determine whether fetal growth is proceeding normally. Sound waves, reflecting at different rates from tissues of varying density, are represented on video monitors and even printed to form a picture of the fetus. The picture can reveal such problems as microcephaly (small head size), cardiac malformations, cleft lip and palate, and other physical disabilities. However, it is not possible to detect many other developmental disorders with great accuracy using this technique (Dooley, 1999; Grandjean et al., 1999).

fetal blood sampling Method of withdrawing blood from the umbilical cord of the fetus. Used to diagnose genetic disorders, especially those that affect the blood.

maternal blood screening Tests preformed on a woman's blood to determine if the fetus she is carrying has an increased risk for some types of chromosomal and metabolic disorders.

ultrasonography Method of using sound wave reflections to obtain a representation of the developing fetus. Used to estimate gestational age and detect fetal physical abnormalities.



The use of ultrasound to provide a visual image of the developing fetus has become a common practice in medical facilities around the world. Prospective parents are often thrilled by this opportunity to obtain a glimpse of the fetus. The use of ultrasound also can be important for obtaining a more accurate assessment of the age of the fetus. It provides assistance in carrying out other tests to determine if development is proceeding normally and, in some cases, permits surgical procedures designed to improve the likelihood of a healthy newborn.

Ultrasonography is widely used to assist in carrying out other prenatal diagnostic tests, to verify the age of the fetus (interpretation of maternal and fetal blood tests are often highly dependent on an accurate assessment of age), and to monitor life-saving operations that may on rare occasions be performed on the fetus within the womb (Harrison, 1996). Although not universally recommended in the United States because of its limits as a diagnostic tool, ultrasound has become a popular tool for informing specialists and parents about the course of prenatal development.

Ethical and Social Issues

The major prenatal diagnostic tests are summarized in Table 3.4. Note that many of these tests are carried out relatively late in pregnancy, and prospective parents may have to wait several weeks longer before they learn of the results. This waiting period, and even the somewhat shorter time frame required to obtain information from other procedures, can create enormous apprehension, especially among those who, on the basis of genetic counseling or less definitive tests such as maternal blood screening, have reason to believe that a problem may exist. Moreover, some expectant women almost feel coerced into using these technological advances to learn more about their pregnancies (Henifin, 1993; Wertz & Fletcher, 1993). Yet a physician who fails to at least offer prenatal diagnosis in circumstances in which it can be informative runs legal risks for incompetent obstetric practice (Wertz & Fletcher, 1998).

Prospective parents have choices regarding whether or not to have such tests performed. In many cases those about to become parents would like to know about possible problems, if for no other reason than to effectively prepare for and address them even before birth. In fact, a substantial number of expectant women who learn that the fetus carries some abnormality still elect to continue the pregnancy, especially if the problem is less severe and possibilities exist for prenatal or postnatal therapy (Pride et al., 1993). On the other hand, in some countries such tests may not be available. But even when they are, not everyone takes advantage of them. For example, women of African American and Hispanic ethnic identity, at least in some areas of the United States, are far less likely to undergo prenatal testing than Caucasian or Asian women (Kuppermann, Gates, & Washington, 1996).

TABLE 3.4 Prenatal Screening Tests

Prenatal Test	When Usually Administered (gestational age)	Typical Waiting Period for Results	Other Comments
Amniocentesis	13–18 weeks	About 2 weeks	Can be administered in weeks 11–14 but normally is not because the available supply of amniotic fluid is more limited.
Chorionic villus sampling	10–12 weeks	24–48 hours	Possibly a slightly greater risk than associated with amniocentesis, including limb deformities.
Fetal blood sampling	18 weeks or later	24–48 hours	Possibly somewhat greater risk than associated with amniocentesis.
Maternal blood screening	15–20 weeks	One week	Not definitive but provides information about increased risk for Down syndrome, and neural tube and some metabolic defects.
Ultrasonography	About 6 weeks and later	None	Provides picture of growing fetus. Not definitive for identifying many disorders. Little evidence of any risk. Often used to accompany other test procedures.

Another issue concerns access to the results of these tests. For example, might insurance companies or other health organizations drop coverage if they become aware of results that indicate expensive medical treatment in the future? What about employers who might choose to hire on the basis of fewer health risks, as, for example, when genetic information provides a hint of susceptibility to major diseases, such as cancer, diabetes, or heart disease, that could affect an individual's subsequent employment capability? There is also the issue of sex preselection.

Controversy: Thinking It Over

Should Sex Preselection Be Permitted?

At one time in medieval Germany couples placed a hammer under their bed if they wished to conceive a boy; in Denmark they chose a pair of scissors if they desired a girl (Golden, 1998). In China and India, where abortion is more widely practiced, evidence already exists that sex selection has been exercised; a disproportionate number of males have been born in these countries in recent years. As a consequence, China and India, as well as some other countries, have passed laws forbidding the use of prenatal tests solely to determine the sex of the fetus and to influence the course of pregnancy when the fetus is male or female (Wertz & Fletcher, 1998).

What Is the Controversy?

Although many adamantly oppose the use of a procedure that leads to the selective abortion of a healthy fetus on the basis of sex, *preselection*, the effort to tilt the probability toward having either a male or female conception, may be far less objectionable. In fact in 1996, 40 percent of individuals in the United States already supported the unrestricted availability of sex preselection if an effective procedure became available (Wertz & Fletcher, 1998).

We are, in fact, at the point at which this possibility exists. Previous efforts have focused on the timing or technique involved in procreation to increase conception of

a male or female. These attempts have generally failed to provide a reliable method of conceiving a boy or a girl. However, in 1998, a new, far more effective procedure was reported, at least for promoting the conception of females (Fackelmann, 1998). This new procedure is based on the difference in the amount of DNA found in the X and Y chromosomes. With the help of a dye that attaches to the DNA and glows when exposed to the light of a laser, sperm cells carrying a second X chromosome and containing about 2.8 percent more DNA than sperm cells carrying a Y chromosome shine more brightly. By introducing lopsided distributions of cells carrying the second X chromosome into the uterus during periods when a woman might become pregnant, these specialists claim to have attained markedly higher numbers of successful pregnancies resulting in girls, a procedure that could also be expected to be successful in skewing the odds toward boys. Other techniques associated with assisted reproduction (see the chapter titled "The Prenatal Period and Birth") can also be used to increase the likelihood of, or even virtually guarantee, a boy or a girl.

What Are the Opposing Arguments?

Proponents of sex preselection claim that parents in most Western countries do not show a strong preference for having a boy or a girl first; but family balancing, having one boy and one girl, is frequently seen as the ideal family complement (Silver, 1998). Having the opportunity to rear children of both sexes also is often viewed as a desirable experience for parents. For other couples, sex preselection may serve to prevent a sex-linked genetic disease from being passed on to their offspring.

Opponents of sex preselection are concerned that even among countries in which no strong preference exists for rearing either boys or girls, the practice supports the potential for sexual bias or increased sexism, a potential that may become more visible once a choice is available to prospective parents (Fackelmann, 1998). A second argument is that determining a conception on the basis of sex is but the first step in the emergence of a preference mentality that can ultimately lead to preselection on the basis of other desired traits and attributes intended to create "perfect" children. Thus the potential for discrimination against those who do not meet the ethnic, cultural, or community ideal may increase substantially.

What Answers Exist? What Questions Remain?

The American College of Obstetricians and Gynecologists (2002) currently supports the notion that sex preselection is ethical and justified in cases in which it will prevent sex-linked genetic disorders in offspring. In the United States, the public also seems to be in support of this position. Opinion is much more divided, however, with respect to sex preselection for other purposes. Recent advertisements have appeared in the United States that target some ethnic groups and emphasize, "Choose the sex of your next baby" and "Desire a son?" Perhaps to help address the controversy, psychologists need to conduct research on the extent to which such preferences exist and on what social and cultural factors promote them in order for the public to gauge whether regulations should be considered. Declarations in favor of sex preselection for the purpose of family balancing and even for selecting the sex of a child in a first pregnancy also have been made by individuals associated with committees and organizations—for example, the American Society for Reproductive Medicine—established to deal with these kinds of issues (Kolata, 2001; Sachs, 2001). Perhaps research on whether raising offspring of both sexes leads to different outcomes for children or results in more successful and effective parenting would provide informative data. Given advances in sex preselection, are we nearing a time when professionals will be recruited to help create "designer" children with respect to other traits? Could such efforts actually contribute to harmful effects because of the potential for lessening genetic diversity in humans? What other kinds of psychological research might be conducted to help answer whether a need exists for regulation of sex and other kinds of trait preselection?

FOR YOUR REVIEW

- What are the major diagnostic tests for prenatal development? What are their limitations and their advantages?
- What ethical and social issues emerge from the use of prenatal diagnostic tests?
- What is sex preselection, and what are arguments for and against the practice of it?

Developmental and Behavioral Genetics

KEY THEME
Nature/Nurture

As our previous discussion indicates, chromosomal errors and particular genes can have drastic, often devastating, effects on physical, intellectual, and social development. Yet similarities observed among relatives—the quick tempers of two brothers; the wry sense of humor in a mother and daughter; the musical talent of a grandfather and his grandchildren; and, as we saw at the beginning of the chapter, Jasmine's and Alyssa's impatience when others could not tell them apart—are not likely to be linked to a single, isolated gene. Might these attributes and behaviors reflect a contribution from many genes? Or are these phenotypic resemblances primarily the result of experiences shared by kin? Researchers engaged in **behavior genetics** attempt to learn to what extent the diversity of human traits, abilities, and behaviors stem from combinations of genes versus experience. Behavioral geneticists are helping to show that the entire realm of human behavior is influenced by nature and nurture in a variety of complex, sometimes surprising, ways (Plomin et al., 2001; Rutter et al., 1999a, 1999b).

The Methods of Behavioral Geneticists

When studying the fruit fly or mouse, behavior geneticists can use *selective breeding* experiments to learn whether certain phenotypic expressions can be increased or decreased in offspring. Members of a species that display a specific attribute are bred to each other, usually over many generations. If the attribute is inherited, subsequent generations of offspring can be expected to display it more and more frequently or strongly. For example, after thirty generations of selective breeding in which either highly active mice were bred only to each other or mice showing only a low level of activity were bred to each other, researchers observed no overlap in terms of the amount of activity displayed by members of the two groups (DeFries, Gervais, & Thomas, 1978). Those bred for high activity were thirty times more active; they would run the equivalent of a football field during two three-minute test periods compared to the other mice, which would not even run the equivalent of a first down (Plomin et al., 2001).

Selective breeding in various species of animals has revealed genetic contributions to many different attributes, including aggressiveness, emotionality, maze learning, and sex drive (Plomin et al., 2001). But selective breeding, of course, cannot be used to examine human behavior. Instead, behavior geneticists gain information about hereditary and environmental influences on human behavior by examining resemblances among family members. These studies investigate similarities among *identical* and *fraternal twins,* siblings, and other members of families who are genetically different from one another to varying degrees.

Identical, or *monozygotic,* **twins** come from the same zygote: a single egg fertilized by a single sperm. A cell division early in development creates two separate embryos from this zygote, and the twins are genetically identical. **Fraternal,** or *dizygotic,* **twins** come from two different zygotes, each created from a separate egg and separate sperm. Fraternal twins are no more genetically similar than siblings born at different times, each averaging about half of their genes in common. However, they do share some aspects of their prenatal environment and, by virtue of their being the same age, may share other experiences to a greater extent than siblings born at different times.

behavior genetics Study of how characteristics and behaviors of individuals, such as intelligence and personality, are influenced by the interaction between genotype and experience.

identical twins Two individuals who originate from a single zygote (one egg fertilized by one sperm), which early in cell division separates to form two separate cell masses. Also called *monozygotic twins*.

fraternal twins Siblings who share the same womb at the same time but originate from two different eggs fertilized by two different sperm cells. Also called *dizygotic twins*.

If identical twins resemble each other more than fraternal twins in intelligence or shyness, one *potential* explanation for this similarity is their common genotype. The degree of resemblance is usually estimated from one of two statistical measures: concordance rate or correlation coefficient. The **concordance rate** is the percentage of pairs of twins in which both members display a specific attribute when one twin is identified as having it. Concordance rate is used when measuring characteristics that are either present or absent, such as schizophrenia or depression. If both members of every twin pair have a particular trait, the concordance rate will be 100 percent. If only one member of every pair of twins has some particular trait and the other does not, the concordance rate will be 0 percent.

When attributes vary on a continuous scale such that they can be measured in terms of amount or degree, resemblances are estimated from a *correlation coefficient.* This statistic helps to determine whether variables such as intelligence or shyness, which have some quantitative value, are more similar for identical than for fraternal twins or more similar among siblings than among unrelated children.

Identical twins may resemble one another more than fraternal twins do because identical twins share the same genotype. However, another explanation for any greater resemblance may be that identical twins share more similar experiences. One way to potentially reduce the effects of similarity in experience is to study biologically related family members who have been adopted or reared apart from one another. If an attribute is influenced by genetic factors, children should still resemble their biological siblings, parents, or other family members more than their adoptive relatives. On the other hand, if the environment is the primary determinant of an attribute, separated children can be expected to resemble their adoptive parents or other adopted siblings more closely than their biological parents or siblings.

Adoption studies, just as in the case of twin studies, pose many challenges for evaluating hereditary and environmental influences (Rutter et al., 1999a). For example, in the past adopted children were often placed in homes similar to those of their biological parents. Under these circumstances, the relative contributions of family environment and heredity to an attribute are extremely difficult to distinguish. In addition, information on the biological family has not always been readily available in the case of adoption. Nevertheless, a number of large-scale investigations of genotype-environment effects involving twins, adopted children, siblings, half-siblings, and unrelated children in blended families are currently under way. For more than twenty-five years, the Colorado Adoption Project, for example, has been conducting longitudinal research on resemblances between (1) parents and their natural children, (2) adoptive parents and their adopted children, and (3) parents and their biological children who have been adopted into other homes (Plomin, DeFries, & Fulker, 1988). Another project, the Minnesota Study of Twins Reared Apart, has assessed a variety of psychological and physiological characteristics in identical and fraternal twins reared together or reared apart and having virtually no contact with each other prior to adulthood (Bouchard, 1997; Bouchard et al., 1990). The MacArthur Longitudinal Twin Study (MALTS), started in 1986, has been following 300 identical and fraternal twins, beginning at about one year of age, to investigate cognitive, social, emotional, and temperamental aspects of development (Emde & Hewitt, 2001). Yet another project, the Nonshared Environment in Adolescent Development (NEAD) study, has been observing 720 families, which include identical and fraternal twins, nontwin siblings in traditional families, and full, half, and unrelated siblings in stepfamilies, to explore the relationship between genetic and environmental influences in adolescence (Reiss et al., 2000). These and other studies are just beginning to illuminate the complex interactions and correlations that exist between heredity and experience, intricate relationships that we must consider more fully as well.

A major goal of many of these studies is to provide an estimate of the **heritability** of complex traits and various behaviors. Heritability refers to the extent to which the variability in a sample of individuals on some characteristic such as shyness or assertiveness is a result of genetic differences among those individuals. Of course, the variability that is not accounted for by the genotype must then be a result of the

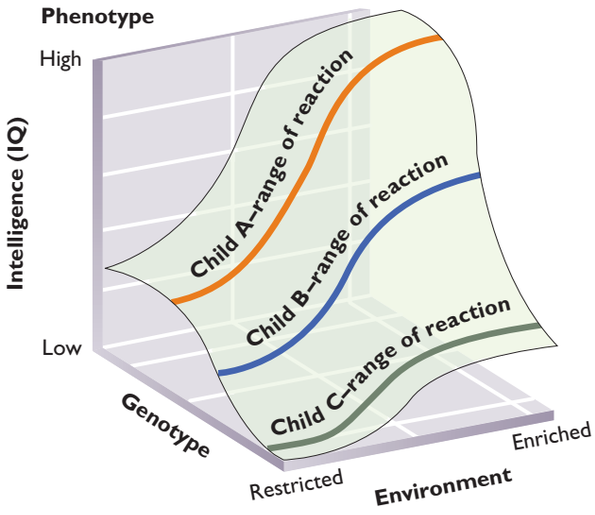
If you were given their names, would you be able to tell these twins apart once they moved and were no longer seated side by side? Virtually everyone would have a great deal of difficulty with such an assignment unless they could constantly keep an eye on at least one of them as he moved about. Because their genetic makeup is the same, identical or monozygotic twins typically look very much alike and display very similar traits and behaviors as can be seen here. Twin studies provide important information about the contributions of heredity and enviroment to development.

concordance rate Percentage of pairs of twins in which both members have a specific trait identified in one twin.

heritability Proportion of variability in the phenotype that is estimated to be accounted for by genetic influences within a known environmental range.

FIGURE 3.11
The Concept of Range of Reaction for Intellectual Performance

As the product of the interaction between genotype and the environment, the phenotype for any attribute shows a broad range of reaction. For example, the intelligence score of any child, even one who is born with the potential for high intellectual ability, is likely to be limited when the environment is severely restricted. An enriched environment will benefit all children, although perhaps those with greater intellectual potential (Child A or B) more than a child with less intellectual potential (Child C).



Source: Adapted from Turkheimer, Goldsmith & Gottesman, 1995.

environmental circumstances those individuals have had the opportunity to experience. Thus, although research on heritability was initially designed to provide answers about the contribution of the genotype, it also helps to shed light on the role of experience in development (Plomin et al., 2001). Unfortunately, estimates of heritability are not always easy to obtain because, as we discuss next, complex interactions and correlations exist between the genotype and experience.

Conceptualizing the Interaction Between Genotype and Environment

KEY THEME
Nature/Nurture

Neither genotype nor experience in isolation of one another explain development. Instead, how a genotype influences development depends to a great extent on the environment. Similarly, how an environment affects behavior often depends on the genotype. These conditional relationships are the basis for complex *interactions* between heredity and experience; the influence of one on the other is not constant across individuals or environmental circumstances or even during different periods of development (Collins et al., 2000; Rutter & Silberg, 2002).

● **Range of Reaction** The interactive relationship between genotype and environment can be conceptualized in terms of the concept of **range of reaction,** the notion that, depending on environmental conditions, a broad range of phenotypes may be expressed as a function of the genotype (Turkheimer, Goldsmith, & Gottesman, 1995). Figure 3.11 illustrates this concept for intellectual performance. Consider a child with Down syndrome (represented, e.g., by Child C in Figure 3.11). Transferring this child from an unstimulating institutional setting (a restricted environment) and engaging him in supportive learning activities (more enriched environments) very likely will help him to achieve a much higher level of cognitive functioning (Feuerstein, Rand, & Rynders, 1988). The performances of children with other genotypes (as represented by Child A and Child B in Figure 3.11) can be enormously affected, too, perhaps even more greatly, depending on whether they are reared in deprived or stimulating conditions.

range of reaction Range of phenotypic differences possible as a result of different environments interacting with a specific genotype.

We need to be cautious, however, in thinking about the concept of range of reaction. It reflects only what we presently know about the way genotypes are expressed in environments familiar to us (Gottlieb, 1995; Rutter et al., 1999a). For example, some day, when the ways in which trisomy 21 affects proteins essential to neural

events are more fully understood, an environment may be fashioned to promote far higher levels of intelligent behavior in children with Down syndrome. New advances in knowledge of biological processes and the other, much broader complex of multidirectional influences that we call environment may help to drastically modify the way a genotype is expressed in specific behavior (Bronfenbrenner & Ceci, 1994).

● **Canalization** The principle of **canalization,** a kind of "channeling" of development, suggests yet another way to think about genotype-environment relationships. This principle helps to shed light on the emergence of behaviors common to all members of a species, as well as individual differences. As originally proposed, a highly canalized attribute is one influenced primarily by the genotype; experiential factors can have an impact on the course of development, but only under extreme conditions (Waddington, 1971). Imagine an emerging capacity as something like water flowing through terrain in which channels of varying depth have been cut. The channels help to steer the flow in one of several possible directions. However, the channels are so deeply cut by the genotype that only extreme environmental pressures can change the course of some phenotypes.

KEY THEME
Individual Differences

Various aspects of early motor development tend to emerge on a fairly regular basis during infancy. Presumably the genotype has carved a relatively deep course for their appearance. However, as you will learn in the chapter titled "Physical Growth and Motor Skills," the emergence of early motor skills is not completely protected from disturbances in experience. Other aspects of early development, including the onset of babbling and smiling at interesting events, which are important components of social responsivity, may also be highly canalized, as may some aspects of language acquisition.

Gilbert Gottlieb (1991) has extended these ideas, proposing that the channeling process may come about not only through the genotype but also from early experiential influences. Thus exposure to certain types of stimulation during a critical period may steer development just as hereditary information can. As an example, Gottlieb cites research on mallard ducklings. If prevented from hearing their own vocalizations and instead exposed to a chicken's call early in development, the ducklings later show a preference for the chicken's call rather than for sounds produced by members of their own species.

Gottlieb (2000) has also argued that the genome itself is not "encapsulated" or isolated from the influences of the environment. A classic example is evident in some reptiles—the temperature during incubation determines the sex of the offspring. As we learn more about how genetic information is transcribed into proteins, we may discover other ways in which events both within and outside the organism change how the genetic transcription process takes place. In other words, we may begin to more fully appreciate that development is the outcome of bidirectional influences between the genotype and levels of experience that range from neural activity to behavior to social and cultural events.

Conceptualizing the Correlation Between Genotype and Environment

KEY THEME
Nature/Nurture

The task of determining what proportion of various traits such as activity level, sociability, or intelligence derives from the genes and what proportion comes from the environment is laden with further difficulties. Not only do genotype and environment interact, but they are also linked or *correlated* with each other in several complex ways (Plomin & Rutter, 1998; Rutter et al., 1999a; Scarr, 1992; Scarr & McCartney, 1983).

canalization Concept that the development of some attributes is governed primarily by the genotype and only extreme environmental conditions will alter the phenotypic pattern for these attributes.

● **Passive Links** One correlation between genotype and experience arises from the tendency for parents to establish a child-rearing environment in harmony with their own interests and preferences. Assume, for example, that sociability has some genetic basis. Sociable parents may transmit this orientation to their children either through

their genes, through the social environment created in their home, or through both mechanisms. This kind of correlation between genotype and environment is labeled as *passive,* because it has been created for the child by the parents.

In most families, the correlation between the genetic and environmental components of child rearing is likely to be positive; that is, the environment will contain features that support and complement the child's genetic potential. But a negative correlation is also possible, as in cases in which a highly active child is adopted into a sedentary family or in which parents elect to rear their children in ways that depart from their own backgrounds and genetic propensities. A parent who feels he or she was too shy during childhood and, as a consequence, missed out on many social opportunities may actively initiate play groups and other projects designed to promote sociability in a child.

● **Evocative Links** Another type of correlation between genotype and environment, termed *evocative* or *reactive,* occurs when aspects of the environment, particularly other people, support or encourage behaviors that may have a genetic component; that is, other people's behavior occurs in response to or is evoked by the child's genotype. For example, an active preschooler is likely to prompt teachers to provide large-muscle toys to dissipate some of her energy. A ten-year-old's eagerness to read may encourage a teacher to offer additional opportunities for learning. A sociable child is more likely to attract the attention of peers than a shy or passive child. Thus attributes that have a biological basis are likely to evoke patterns of behavior from others that complement the child's genetic tendencies.

Adoption studies are especially valuable for examining evocative genotype-environment correlations because the genetic makeup of the adopted child is independent of the genetic makeup of the adoptive parents. For example, Thomas O'Connor and his colleagues (O'Connor et al., 1998) wondered whether parental behaviors might be influenced by genetic factors that children bring to the caregiving situation. More specifically, could a child adopted shortly after birth whose biological parent had reported a history of antisocial behavior influence the way the adoptive parents treated him or her? In particular, would the adoptive parents of such children engage in more negative interactions than the adoptive parents of children whose biological parents did not report a history of antisocial behavior? Indeed, this was the finding, suggesting that something about the genotype inherited by the child was evoking more negative reactions from the adoptive parents. These children were, in fact, reported to engage in more antisocial behavior, thus perhaps drawing out the more negative reactions of their adoptive parents. However, the researchers also emphasize that other unknown environmental factors also seemed to play a role in increased negative parental reactions in some of these families.

KEY THEME
Child's Active Role

● **Active, Niche-Picking Links** In yet another type of correlation between genotype and environment, termed *active,* the child may be attracted by and eagerly seek out experiences more compatible with his or her genotype. Bright children could prefer to exercise their intellect and to play with peers who are also bright. The athletic child may find little pleasure in practicing the piano but spend countless hours skateboarding and playing basketball. Thus any genetic basis for various traits and activities influences the kind of environment a child attempts to create and experience. Sandra Scarr and Kathleen McCartney (1983) described this kind of linkage as **niche picking** to emphasize that children and adults selectively construct and engage environments responsive to their genetic orientations.

niche picking Tendency to actively select an environment compatible with a genotype.

Scarr and McCartney (1983; Scarr, 1992) believe that the strength of passive, evocative, and active correlations between genotype and environment changes with development. The experiences infants receive are often determined for them by their caregivers. Thus initial correlations between genotype and environment are more likely to be influenced by passive factors. As children gain greater independence and control of their environment, however, others around them are likely to notice and support their individual differences, and niche picking becomes an increasingly im-

Niche picking, the tendency for an individual to seek out and become attracted to activities that are compatible with his or her genotype, is an important aspect of the interaction between nature and nurture. Because of her talent at playing the piano, this young person may be drawn to a future career or avocation involving music. She is also likely to pursue additional training and opportunities to become even more skilled as a musican.

portant factor as children have the opportunity to choose their own interests and activities.

An important implication of these changing relationships is that children within the same family may become less similar to one another as they grow older and become freer of the common environment their parents provide. Older siblings can select niches befitting their individual genotypes more easily than can younger children. When Sandra Scarr and Richard Weinberg (1977) studied adopted children, they obtained support for this prediction. During early and middle childhood, adopted but biologically unrelated children in the same families showed similarities in intelligence, personality, and other traits. Perhaps these resemblances came about both as a result of adoption procedures that encouraged the placement of children in homes somewhat like their biological homes and from the common family environment the adopted children now shared. As adopted siblings neared the end of adolescence, however, they no longer exhibited similarities in intelligence, personality, or other traits; the passive influence of the common environment established by the adoptive parents had become supplanted by active niche picking.

The notion of niche picking provides us with an even more startling prediction. When identical twins are reared apart, they may, with increasing age, actually come to resemble each other more, and perhaps as much as, identical twins reared together! This greater correspondence would emerge as others react to their similar behaviors and as opportunities arise for the twins to make more choices. In the Minnesota Study of Twins Reared Apart, pairs of identical twins, separated as infants and having no interactions with each other until well into adulthood, revealed remarkable similarities not only in gait, posture, gestures, and habits such as straightening eyeglasses but also in storytelling skill, spontaneous giggling, phobic tendencies, hobbies, and interests, resemblances rarely observed between fraternal twins reared apart and usually not considered to have a strong genetic basis (Bouchard, 1984; Bouchard et al., 1990; Lykken et al., 1992). Furthermore, identical twins reared apart showed as high a correlation on many intellectual tasks and personality variables as those reared together. These results suggest that niche picking can be a powerful means of maintaining behaviors supported by the genotype.

Hereditary and Environmental Influences on Behavior

KEY THEME
Nature/Nurture

Research findings involving studies of family resemblances, adopted children, and identical and fraternal twins can be, and often are, interpreted in many different ways

precisely because of the complex interactions and relationships that genotype and environment share in shaping behavior. These interpretations sometimes have powerful implications for intervention and social policy (Baumrind, 1993; Jackson, 1993; Scarr, 1992, 1993). Should families or communities, for example, expend resources for educational and mental health efforts if a substantial biological basis for behavior exists? Or does this kind of question, concerned with *how much* heredity contributes to variations in the human phenotype, fail to recognize that educational and social opportunities are essential to maximize competencies, even where genetic contributions are considerable?

Consider the following: about 90 percent of the variability in height among individuals reared in a typical community is believed to be a consequence of genetic factors. However, even though height is strongly influenced by the genotype, its average increase among young adult males in Japan since the end of World War II has been about three-and-a-half inches (Angoff, 1988). Clearly changes in the environment have had a profound impact on a characteristic that receives a significant contribution from the genes. Environmental factors can surely be expected to have an impact on other inherited characteristics as well (Collins et al., 2000; Rutter, 2002).

Robert Plomin (1996) emphasizes one other interesting point about the work that often has revealed a substantial genetic contribution to various kinds of behaviors. Environmental influences frequently account for *more* of the variability in human behavior than does the genotype. Moreover, research in behavioral genetics has begun to provide intriguing insights into *how* environmental factors affect development. Let's examine some of the findings more closely.

● **Intelligence** Despite its many limitations (see the chapter titled "Intelligence"), most studies have relied on IQ to measure the contributions of genotype and environment to intelligence. Table 3.5 summarizes correlations on IQ test scores among individuals who share different genetic relationships with one another. Environmental contributions are revealed by findings that individuals reared together show somewhat higher correlations for intelligence scores than those with the same genetic relationship reared apart. Nevertheless, the impact of the genotype on intelligence is also evident. The correlations for IQ increase as the similarity in genotypes rises.

We can make sense of several additional findings by considering correlations between genotype and environment. For example, IQ scores for younger adopted children reared together are positively correlated (about .20), as indicated in Table 3.5. However, they are much closer to .00 for adolescents. Moreover, intelligence has been found to be highly correlated in infancy and early childhood for *both* identical and fraternal twins and, with increasing age, to become *even greater* for identical twins but decline to the level reported in Table 3.5 for fraternal twins (Fischbein, 1981; Wilson, 1986). These findings probably reflect the impact of passive links (the similar rearing environment created by the parents) on intelligence early in childhood and more opportunity for niche picking later in development.

KEY THEME
Child's Active Role

Identical twins, however, do not always become more similar as they grow older. As twins who have been reared together become older, fraternal, and to some extent identical, twins become more dissimilar on many aspects of intelligence tests (McCartney, Harris, & Bernieri, 1990). Perhaps they actively attempt to establish a *unique* niche in the family and community, efforts that may also be encouraged by parents of the twins (Schachter, 1982).

Despite the evidence that the heritability of intelligence is high, a classic investigation by Marie Skodak and Harold Skeels (1949) illustrates the substantial impact experience still has. One hundred children born to mentally retarded mothers, most of whom were from low socioeconomic backgrounds, were adopted before six months of age into homes that were economically and educationally well above average. These children displayed above-average intelligence throughout childhood and adolescence and substantially higher IQs than their biological parents, an outcome reflecting the contribution of environmental factors. Nevertheless, the children's IQs

TABLE 3.5 Correlation in IQ as a Function of Genetically Related and Unrelated Individuals Living Together and Apart

Relationship	Raised	Observed
Monozygotic twins	Together	0.85
Monozygotic twins	Apart	0.74
Dizygotic twins	Together	0.59
Siblings	Together	0.46
Siblings	Apart	0.24
Midparent/child	Together	0.50
Single parent/child	Together	0.41
Single parent/child	Apart	0.24
Adopting parent/child	Together	0.20

Note: The data are based on a meta-analysis of 212 IQ correlations reported in various studies. In the case of twins and siblings, the heritability estimates are increased by an unspecified amount because of the common environmental contribution from the prenatal environment shared simultaneously by twins and sequentially by siblings. "Midparent" refers to the mean of the IQ scores of both parents.

Source: From B. Devlin, S. E. Feinberg, D. P. Resnick, & K. Roeder (Eds.), *Intelligence, genes, and success: Scientists respond to* The Bell Curve, pp. 45–70. Copyright © 1997. Used by permission of Springer-Verlag and the author.

were still correlated with those of their biological mothers, indicating a hereditary contribution to these scores as well.

● **Temperament and Personality** Genetic influences typically account for 20 to 50 percent of the variability in personality differences within a population (Plomin et al., 2001; Saudino, 1997). The higher heritability estimates tend to be found when parental reports rather than direct observation of social behavior are recorded (Collins et al., 2000). **Temperament,** an early-appearing constellation of personality traits, has been of particular interest in terms of possible genetic influences. A number of broad qualities characterize temperament. One of these is *sociability,* the tendency to be shy or inhibited and somewhat fearful of new experiences versus outgoing and uninhibited, characteristics that are likely precursors to introversion and extroversion, respectively, in older children and adults. Another trait is *emotionality,* the ease with which an individual becomes distressed, upset, or angry and the intensity with which these emotions are expressed. A third trait is *activity,* as evidenced by the tempo and vigor with which behaviors are performed. Identical twins consistently show higher correlations on these characteristics (typically between .40 and .60) than fraternal twins (typically between .10 and .30), suggesting an inherited component (Emde et al., 1992; Goldsmith, Buss, & Lemery, 1997; Robinson et al., 1992). Perhaps inherited differences in physiological reactivity underlie these differences. For example, with respect to sociability, young children who remain aloof and are reluctant to play with novel toys display increased heart rate and muscle tension in unfamiliar situations compared with children who are more outgoing and spontaneous (Kagan, 1994).

KEY THEME
Individual Differences

Consistent racial and ethnic differences have been reported and attributed to genetic differences in temperament as well. When Daniel Freedman (1979) compared Caucasian and Chinese American newborns, he found that Caucasian babies were more irritable and harder to comfort than Chinese American infants. Similarly, research with four-month-olds from Boston, Dublin, and Beijing indicated that American infants were more active and fretful than those in Dublin, who in turn were more reactive than those in China (Kagan et al., 1994).

temperament Stable, early-appearing constellation of individual personality attributes believed to have a hereditary basis; includes sociability, emotionality, and activity level.

Does the environment also play some important role in temperament and other personality differences? Studies comparing the personalities of unrelated children in

the same household report that the correlations are fairly low and often approach zero, especially in later childhood and adolescence (Plomin et al., 2001). Behavioral geneticists have uncovered some other surprising findings. *Shared environment,* the kinds of experiences children in a home or community bear in common and that are assumed to foster similarity in children within the same family, simply does not have much effect for many aspects of personality (Goldsmith, Buss, & Lemery, 1997; Plomin et al., 2001). In contrast, *nonshared environment,* the experiences unique to individual children as they interact with parents, peers, and others, can play a powerful role in development, one that tends to make children in the family *different* rather than similar (Harris, 1998; Plomin et al., 2001).

How can this be? One possibility is that peers, as well as genes, play a pivotal role in determining the differences in personality and social adjustment displayed by children reared in the same family. This view has sometimes escalated to suggest that parenting matters relatively little with respect to offspring's personality and social adjustment. Needless to say, such a conclusion has been heatedly debated (Collins et al., 2000; Harris, 2000; Vandell, 2000).

Perhaps, too, parents actually do treat their children differently, even though they claim not to. Although mothers and fathers typically do not report much differential treatment, siblings frequently express another take on the matter. Observations of how parents interact with their sons and daughters seem to provide some ammunition in support of the children's perceptions. For example, the toys boys and girls are encouraged to play with may differ considerably in some families (Lytton & Romney, 1991). And when siblings perceive that they are being treated unfairly by their parents, the quality of their relationships with one another deteriorates (Kowal & Kramer, 1997).

Research Applied to Parenting

Treating Siblings Fairly

As the party was nearing its end, the twins began opening their presents. Jasmine, just as she approached many of her other ventures, impulsively grabbed one of the brightly wrapped packages and without hesitation tore it open; Alyssa was more refined and systematic in her procedure, opening each box carefully, as carefully as any excited five-year-old might be expected to in such a situation. Their mother looked on with pride but with some apprehension as well. Should she encourage Jasmine to be less impetuous and Alyssa to be more spontaneous? Or should she promote their differences, respecting the individual styles each child showed even if it meant that each twin was treated very differently? She knew that Jasmine's impish streak would require more disciplining and that Alyssa's more cautious character would lead to fewer tests of limits. But she did not want her children to think that she favored one of the twins more than the other.

KEY THEME
Individual Differences

Should parents treat their children differently or in the same way? The answer is far from clear. For example, historically, identical twins were often dressed alike and treated almost as if they were one and the same individual. Today parents are advised to recognize their twins' individuality by encouraging them to gain unique experiences and to "pick out" their own niche in the family and community. As a consequence, they may be treated differently by parents and others. Other siblings, of course, share only about half of their genes and are quite distinct in physical appearance, personality, intellectual ability, and other characteristics. When is differential treatment a good thing? When is it a source of conflict? Research indicates parents might consider some of these points when confronting this matter.

1. *Expect siblings to kindle different reactions from others.* Because all children, with the exception of identical twins, are born with different genotypes, they are likely to evoke distinctive kinds of responses from family members, teachers, peers, and others. These responses can, in turn, serve to magnify existing differences in the behaviors of siblings.

2. *Assume siblings will actively search out ways to be different from one another.* Children very likely will engage in niche picking to set themselves apart from others within the family—a process called *sibling deidentification* (Feinberg & Hetherington, 2000). Such efforts may be observed even in identical twins such as Jasmine and Alyssa, as one twin takes on a more active role in leadership, becomes more studious, or engages in more social interactions than the other. Whether it stems from biology or experience, the opportunity to find one's niche within the family, as well as the larger community, is an important aspect of development for all children.

KEY THEME
Child's Active Role

3. *Anticipate that siblings will experience family events in different ways and will need to be treated differently in some circumstances.* Siblings within the family—by virtue of their birth order, spacing, and unique events such as an illness, the death of a grandparent, or a move to a new neighborhood—will necessarily receive unique experiences because of their specific developmental status. In fact, distinct socialization practices for children of different ages is the norm because treatment by caregivers could otherwise be developmentally inappropriate. Younger children typically need more nurturance and care, older children increased independence and greater responsibility. Caregivers recognize that differential treatment is sometimes necessary (Dunn & McGuire, 1994). Children as young as five years of age notice these differences. Moreover, younger children would prefer being the oldest in the family; that is, in their eyes older siblings seem to have higher status (McHale et al., 1995). Furthermore, children and adolescents perceive greater differential treatment of same-sex siblings than brother-sister dyads by parents (McHale et al., 2000). Stress within the family may magnify these differences (Crouter, McHale, & Tucker, 1999). Children do not always find differential treatment to be unfair. In fact, when children experience differential treatment on the part of their parents but feel it is justified, that is, equitable even though not equal, sibling relationships are generally positive (Kowal & Kramer, 1997)

KEY THEME
Individual Differences

4. *Treat siblings impartially when possible and appropriate.* Impartial treatment of siblings by parents, to the extent that it is appropriate, is associated with less conflict between siblings and between children and their parents (Brody & Stoneman, 1994; McHale et al., 1995). In other words, parents who maintain a balance between socialization practices that are equitable within the limits of age-appropriate differences have children who interact effectively and in constructive ways with others, including their siblings. Differences in parenting arising from the individual needs of children can have positive consequences, but only as long as these differences do not reflect a form of parental "favoritism."

Parents with more than one child often want to treat each of them equally but need to fine-tune their parenting efforts to the needs and age-appropriate activities of individual children. Fine motor skills demonstrated by the older sibling may permit her to color or carry out other activities that are beyond the capacity of the younger sibling, who, as shown here, has found her own way to keep busy. By providing individual support and avoiding "favoritism," parents seem to help siblings learn to appreciate each other's abilities.

KEY THEME
Nature/Nurture

● **Behavioral and Personality Disorders** Table 3.6 summarizes the concordance rate for identical and fraternal twins for a variety of behavioral and personality disorders. None of these findings indicates 100 percent concordance even in identical twins, despite the identity of their genetic makeup. Thus environmental factors play an important role in the manifestation of each of these problems. In fact, concordance measures for *conduct disorders* (fighting and aggressive behavior, failure to accept parental discipline) in both identical and fraternal twins are relatively high, suggesting that environmental factors contribute substantially to their appearance in both groups.

The genetic contribution to *bipolar disorder,* a disorder characterized by rapid and wide mood swings between feverish activity and withdrawn, depressed behaviors, appears to be substantial. Family studies reveal that children of a parent with bipolar disorder are at far greater risk for displaying the illness than children without such a parent. Research on adoptees provides further evidence that genotype plays a role; the risk for adopted children whose biological parents have the illness is about three times greater than for adopted children whose biological parents do not have it (Rutter et al., 1999b). Recent work on *autism* (more fully described in the chapter titled "Cognition: Piaget and Vygotsky"), a disorder that historically was assumed to be largely the consequence of improper caregiving, also reveals an ample hereditary contribution (Rutter et al., 1999b).

Alcoholism, although not linked to a single gene, shows a modest genetic component. In fact, more recent studies now put the concordance rate at over .50 for identical twins and over .30 for fraternal twins who are male, somewhat higher than shown in Table 3.6 for the general population (Kendler et al., 1997; McGue, 1999). *Schizophrenia,* a form of psychopathology that includes disturbances in thoughts and emotions, such as delusions and hallucinations, also exhibits some genetic contribution. As the biological relationship to someone diagnosed as having schizophrenia increases, an individual's risk for the same diagnosis rises. When one twin has schizophrenia, the other twin is about three times more likely to display schizophrenia if identical than if fraternal. Adoption studies further confirm a role for the genotype; schizophrenia is far more prevalent among adopted children who are the biological offspring of a schizophrenic parent than among those of a nonschizophrenic parent (Gottesman & Shields, 1982). At the present time, no single gene seems to be responsible for the findings; instead, polygenic contributions are more likely.

Recent behavioral genetic research raises the possibility that the same genes could have some role in different manifestations of psychopathology, including anxiety and depression or aggressive behavior and antisocial behavior (Eley, 1997). These "general" genes, in other words, may contribute to wide variations in characteristics and behaviors that are often problematic for the individual. Although psychologists have long assumed that environmental conditions play a part in different expressions of

Evidence for the genetic basis of behavioral and personality disorders is often supported by studies of twins. For these disorders, the likelihood that both members of a pair will display the disorder, if exhibited by one, is greater when the pair are identical twins rather than fraternal twins. However, environmental factors may contribute to high concordance rates for both identical and fraternal twins, as is likely for conduct disorders.

TABLE 3.6 The Genetic Basis of Selected Behavioral and Personality Disorders: Twin Data

Twin Concordances	Identical Twins	Fraternal Twins
Conduct disorder	.85	.70
Bipolar disorder	.65	.20
Autism	.65	.10
Unipolar depression	.45	.20
Alcoholism—males	.40	.20
Schizophrenia	.40	.10
Alcoholism—females	.30	.25

Source: Data from Plomin, 1994.

psychopathologies, the finding that a subset of genes may contribute to this phenomenon raises interesting questions about how biochemical processes affect behavioral and personality disorders and their development.

● **Other Characteristics** A host of other characteristics, including empathy, reading disabilities, sexual orientation, obesity, susceptibility to various illnesses such as heart disease and cancer, and even a propensity to watch television, have a genetic linkage (Castles et al., 1999; LeVay & Hamer, 1994; Pérusse & Chagnon, 1997; Plomin, Corley, et al., 1990; Plomin, Owen, & McGuffin, 1994). In addition, how individuals perceive their family environments also appears to be influenced by the genotype (Hur & Bouchard, 1995; Plomin et al., 1994), as does the way caregivers parent and how children adjust to that parenting (Neiderhiser et al., 1999). The influence of the genotype affects the entire breadth of human behavior, although exactly how it does so remains uncertain.

FOR YOUR REVIEW

- What methods do behavior geneticists use to investigate the extent to which behavior is influenced by combinations of genes versus experience? Why are identical and fraternal twins—as well as adopted children—important in work designed to evaluate the heritability of behavior?
- How do concordance rate and correlation differ as measures in investigating genetic and environmental contributions to development?
- How are conceptualizations of the interaction between genotype and environment advanced by notions of a range of reaction and canalization?
- How do passive, evocative, and active niche-picking correlations between the genotype and experience differ from one another?
- To what extent are behavorial phenotypes such as intelligence, personality and temperament, personality disorders, and other characteristics influenced by heredity?
- To what extent are shared and nonshared environments important in accounting for similarities and differences in children's behavior?
- Why might parents treat siblings differently, and how do children interpret these differential treatments?

CHAPTER RECAP

Summary of Developmental Themes

■ **Nature/Nurture** ***What roles do nature and nurture play in development?***

The phenotype, the observable behaviors and characteristics of an individual, is the product of a complex interaction between genotype and environment. Environment includes biological contexts, such as the foods we eat, but more frequently we consider it to be the experiences provided by caregivers and others. The relationship between genotype and environment is complicated by their interaction with each other and by passive, reactive, and niche-picking correlations. As a consequence, experiential factors are tightly interwoven with genotype to produce the range and variety of behaviors and characteristics an individual displays. Both genotype and environment are indispensable to development.

■ **Child's Active Role** ***How does the child play an active role in the effects of heredity on development?***

Researchers recognize the child's active efforts to seek out environments that support and maintain behavioral orientations and preferences influenced by hereditary factors. As the child achieves greater control over the environment, he or she has increasing opportunities to find a niche. In other words, behaviors, activities, and skills the child displays not only are a consequence of imposed social and physical experiences but also reflect the selective efforts of the child to discover interesting, challenging, and supportive environments. Inherited and environmentally imposed influences may be met with eager support or active resistance to determine each child's unique life history.

■ **Individual Differences** ***How prominent are individual differences in development?***

Individual differences are pervasive in intellectual, temperamental, and a host of other cognitive, social, and emotional aspects of development. Hereditary and environmental factors determine these differences. Alleles of genes contribute to the wide range of physical, cognitive, emotional, and social adaptations displayed by individuals. These individual differences are not solely produced by genes; they are also the product of a rich medley of physical, social, and cultural contexts in which each individual matures. A distinctive combination of genes and experiences promotes the abundant diversity we observe in human abilities and behavior.

Summary of Topics

Principles of Hereditary Transmission

- The observable and measurable characteristics of an individual are the result of his or her *genotype,* or inherited endowment, and experience. A *phenotype* refers to the traits and behaviors displayed by an individual.

The Building Blocks of Heredity

- The structures associated with the principles of heredity must be examined at several different levels.
- An individual's body is composed of trillions of cells. Twenty-three pairs of *chromosomes* (a total of forty-six), consisting of *deoxyribonucleic acid* or *DNA,* are located in the nucleus of most cells in the human body.
- The central unit of hereditary information is the *gene,* a segment of a chromosome. *Nucleotides* are two different sets of pairs of repeating molecules that form the biochemical building blocks for the genes and the basic structure of the chromosomes.
- Males and females differ in the composition of the twenty-third pair of chromsomes. In females, both members of the pair normally are *X chromosomes*. In males, one is normally an X chromosome and the other is a *Y chromosome.*
- The entire inventory of nucleotide base pairs in humans, which has recently been mapped, is called the *human genome.*

Gene Expression

- Variants of genes on the twenty-three pairs of chromosomes, or *alleles,* often interact with one another in a *dominant-recessive* pattern or in other ways to establish different probabilities of inheritance for particular traits or characteristics.
- Cell division in the human body takes place in two different ways. *Mitosis* is the process of cell division by which the forty-six chromosomes are duplicated in the body cells. The *gametes*, or sperm and egg cells, are formed by *meiosis*, a process of a cell division that results in twenty-three chromosomes in each of these cells.
- The random process by which a member of each of the twenty-three pairs of chromosomes is selected for the gametes, combined with *crossing over* during meiosis, ensures that every individual, with the exception of identical twins, has a unique hereditary blueprint.

Gene and Chromosomal Abnormalities

- A number of inherited diseases and abnormalities associated with chromosomes and alleles can lead to severe disruptions in physical and behavior development.

Gene Variations

- The likelihood of inheriting a genetic disease or disorder such as Williams syndrome, sickle cell disease, or phenylketonuria depends on whether it is caused by a dominant or a recessive allele. Gene disorders such as fragile X syndrome that are associated with the XX or XY chromosomes are said to be *sex-linked.*

Chromosome Variations

- The most common disorder contributing to mental retardation, trisomy 21 (Down syndrome), is associated with inheritance of an extra chromosome.
- Variations in number of sex chromosomes also occur but do not always contribute to behavioral or other developmental problems, especially if a supportive environment is available.

Genetic Counseling

- *Genetic counseling* provides prospective parents with information on the probability of having children affected by birth defects.

Prenatal Diagnosis

- Various tests, including *amniocentesis, chorionic villus sampling, fetal blood sampling, maternal blood screening,* and *ultrasonography,* can be carried out to assess the likelihood of the developing fetus having a genetic defect or one of many other abnormalities.
- Controversial ethical and social issues, including the opportunity to carry out sex preselection, confront parents and professionals as a result of advances in prenatal tests.

Developmental and Behavioral Genetics

- *Behavior genetics* is a method of attempting to determine the relative contribution of heredity and environment to traits and behaviors that are often the result of combinations of genes.

The Methods of Behavorial Geneticists

- To determine contributions from combinations of genes, behavioral geneticists frequently engage in selective breeding with lower organisms or compare findings between various family members, such as *identical twins, fraternal twins,* siblings, and adopted children, because these groups differ in the extent to which they share a common genotype. The *concordance rate* refers to the extent that both members of pairs of twins display the same characteristics.
- *Heritability* refers to the extent to which the variability on some characteristic in a sample of individuals is accounted for by genetic differences among those individuals.

Conceptualizing the Interaction Between Genotype and Environment

- The concept of *range of reaction* highlights the fact that a trait or behavior influenced by a person's genotype may be unique to a particular kind of environment.
- The principle of *canalization* emphasizes that some traits and characteristics are highly determined by the genotype or, conversely, that some environments may have a powerful influence in how these are displayed.

Conceptualizing the Correlation Between Genotype and Environment

- Determining the relative contribution of genotype and environment to variations in behavior is made complicated by interactions and correlations between these two factors, as well as by other limitations in our understanding of their relationship to one another.
- Correlations between genotype and environment may be *passive* in that caregivers with specific genotypes are likely to provide environments supportive of their children's genotypes; *evocative* in that parents, peers, and others are likely to react in ways that accommodate genetic inclinations; and *active* in that children may attempt to find or create environments that support their individual genetic propensities, that is, engage in *niche picking.*

Hereditary and Environmental Influences on Behavior

- Intelligence, temperament and other personality variables, social adjustment and behavioral disorders, and other traits and characteristics often display considerable heritability, suggesting that the genotype contributes substantially to variability among children for many aspects of development.
- The shared environment provided by the family increases similarity among siblings, often to a limited extent. However, the nonshared environment that children within the same family experience contributes substantially to individual differences among children.
- Siblings perceive differential treatment from their parents, but when the differential treatment is interpreted as equitable, even if not equal, positive relationships between siblings are fostered.