

## Chapter Objectives

- To describe the biochemical basis of genetic information and the process through which it is transmitted from one generation to the next.
- To identify the contributions of genetic factors to individuality through their role in controlling the rate of development, their contributions to individual traits, and the genetic sources of abnormalities.
- To trace fetal development through three trimesters of pregnancy, including an understanding of critical periods when normal fetal development can be disrupted.
- To describe the birth process and factors that contribute to infant mortality.
- To analyze the reciprocity between the pregnant woman and the developing fetus, focusing on ways that pregnancy affects a childbearing woman and expectant father, as well as basic influences on fetal growth such as maternal age, drug use, nutrition, and environmental toxins.
- To examine the impact of culture on pregnancy and childbirth.
- To analyze abortion from a psychosocial perspective, including the legal context, its social and emotional impact on women, and men's views.

*The fetus develops within the context of the pregnant woman's body, her family, and her culture. During the prenatal period, the pattern of development is guided largely by genetic information, but the outcome of the pregnancy and the robustness or vulnerability of the newborn are also influenced by the kind of environment the pregnant woman provides. In this chapter, we take the perspective of both the developing fetus and the expectant parents. For the parents, their role begins at the decision to have children, the experiences of pregnancy, and the events of childbirth. For the child, the process of growth begins at the moment of fertilization.*

*During the prenatal period, genetic factors guide the tempo of growth and the emergence of individual characteristics. As the human fetus grows, sensory and motor competencies emerge. The psychosocial environment provides resources for and challenges to healthy development. Cultural attitudes toward pregnancy and childbirth, poverty and the associated stressors, support from the child's father and other significant family members, maternal nutrition, and the use of obstetric drugs are among the factors that affect fetal growth. ❖*

## GENETICS AND DEVELOPMENT

From the time of fertilization, genetic information provides a set of guidelines for the individual's development. Given one's genetic potential, a wide range of individual variation is possible, depending on the quality of the environment and one's interaction with it.

### Genes and Chromosomes as Sources of Genetic Information

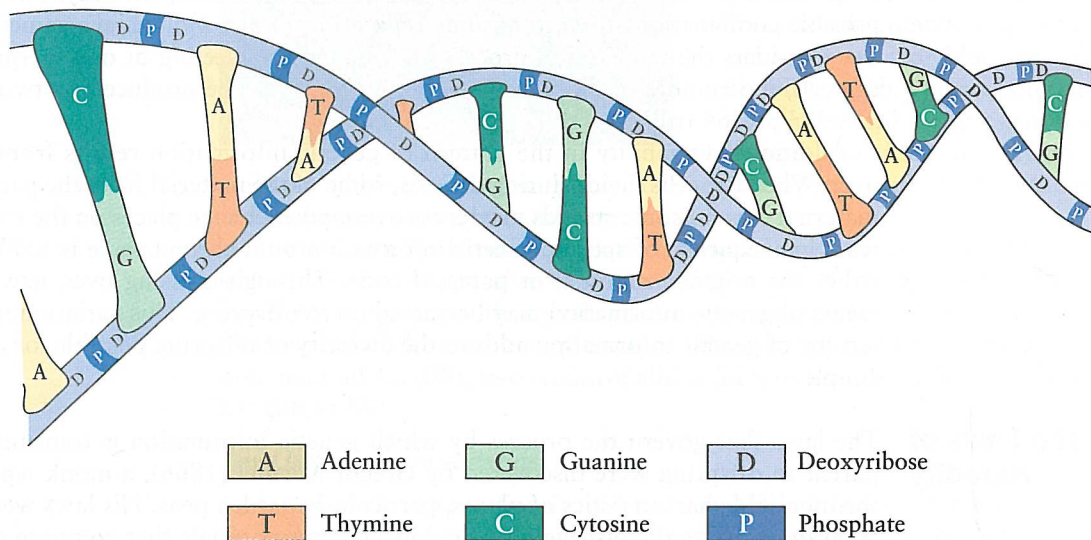
When we talk about inherited characteristics, we are really referring to two different kinds of heredity. The first includes all the genetic information that comes to us as members of the human species. We inherit genetic information that is shared by all human beings, such as patterns of motor behavior (walking upright, for instance), brain size, and body structure, including the proportional size of the head, torso, and limbs. Two of the most relevant of these species-related characteristics are the readiness to learn and the inclination to participate in social interaction. All humans share these attributes.

The second kind of heredity consists of characteristics that have been transmitted through a specific gene pool. Such traits as hair color, skin color, blood group, and height result from the genetic information passed on from one generation to the next. The principles of genetics that we describe here refer primarily to the products of a spe-

cific gene pool (Thompson et al., 1991). Genetic information links each new person to the human species in general and also to a specific genetic ancestry.

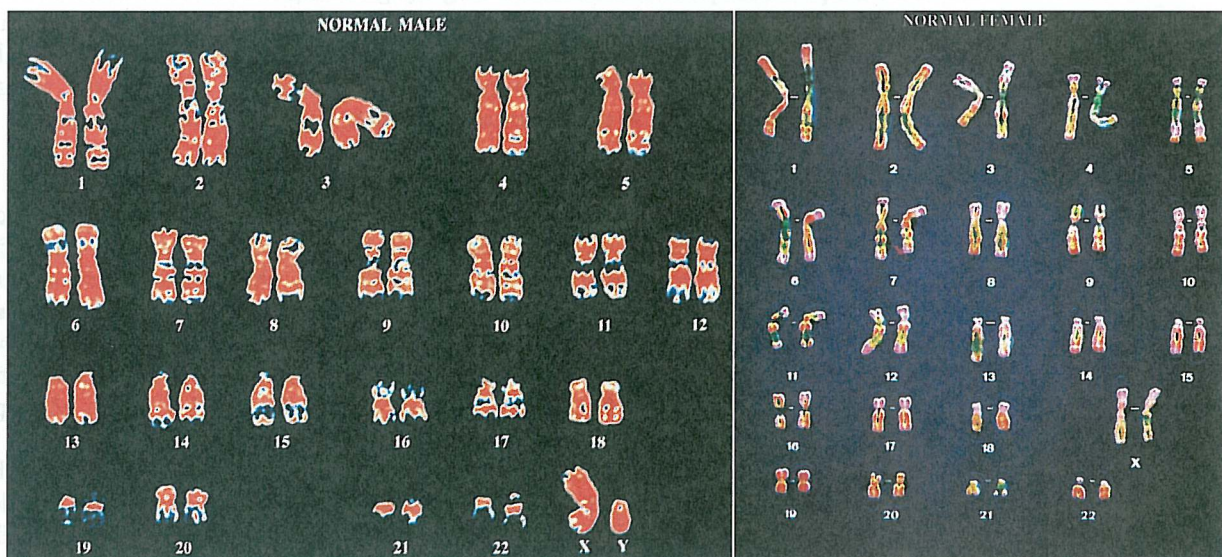
**Chromosomes**—long, thin strands of genetic material located in the cell nucleus—are formed from chains of DNA molecules (Figure 4.1). A gene is a portion of DNA that codes for one hereditary characteristic and occupies a specific place on a chromosome. Human beings have about 100,000 functional genes distributed along the 23 pairs of chromosomes (Figure 4.2). Genetic mapping involves identifying the specific location of each gene on a specific chromosome, which is an enormous task: The smallest human chromosome bands that can be recognized under the microscope contain from 2 to 5 million base pairs of DNA and many genes (Patterson, 1987; NIH/CEPH Collaborative Mapping Group, 1992).

The biochemical basis of genetic information is the DNA (*deoxyribonucleic acid*) molecule, which is in the shape of a double helix and looks something like a twisted rope ladder. The sides of this genetic ladder are composed of alternating units of sugar



**FIGURE 4.1**

Diagram of a small part of a DNA molecule



**FIGURE 4.2**

The 23 pairs of chromosomes in a normal human male (left) and female (right)

(deoxyribose) and phosphate, and the rungs are made up of pairs of nitrogen bases. Nitrogen bases are so named because they include the element nitrogen as well as the elements hydrogen and carbon. Four nitrogen bases are involved: *adenine (A)*, *guanine (G)*, *cytosine (C)*, and *thymine (T)*. These bases are often referred to by their initial letters, and A, G, C, and T are called the genetic alphabet.

In each chromosome pair, one chromosome comes from the father and one from the mother. The chromosome pairs differ in size, some containing over 1000 genes and others containing 2000. In 22 pairs of chromosomes, both members are similar in shape and size. They also contain the same kinds of genes. The 23rd pair of chromosomes is a different story: Females have two X chromosomes, and males have one X and one Y chromosome. The X and Y notation is used because these chromosomes differ in shape and size (the X chromosome is longer than the Y chromosome; see the last pair in Figure 4.2). There are very few similarities in the genes present on the X and Y chromosomes.

The same group of chromosomes does not appear in each gamete (egg or sperm cell). When the cells divide, the chromosomes separate independently. There are  $2^{23}$  possible combinations of chromosome separation in any individual's gametes. When one considers the fertilization process and the chance meeting of one sperm and one egg cell, the number of different individuals that might be produced by two adults is  $2^{23} \times 2^{23}$ , or 64 trillion.

Additional variability in the pattern of genetic information results from crossing over. When the cells divide during meiosis, some of the material from the paternal and maternal chromosome strands may cross over and exchange places on the strand. The resulting sequence of specific genetic information on a chromosome is unlike that in either the original maternal or paternal code. Through crossing over, new arrangements of genetic information may be passed on to offspring. This variation in the patterning of genetic information adds to the diversity of offspring possible for any single couple.

## The Laws of Heredity

The laws that govern the process by which genetic information is transmitted from parent to offspring were discovered by Gregor Mendel (1866), a monk who studied the inherited characteristics of plants, particularly garden peas. His laws were formulated long before the discovery of the biochemical materials that compose genes and chromosomes.

### Alleles

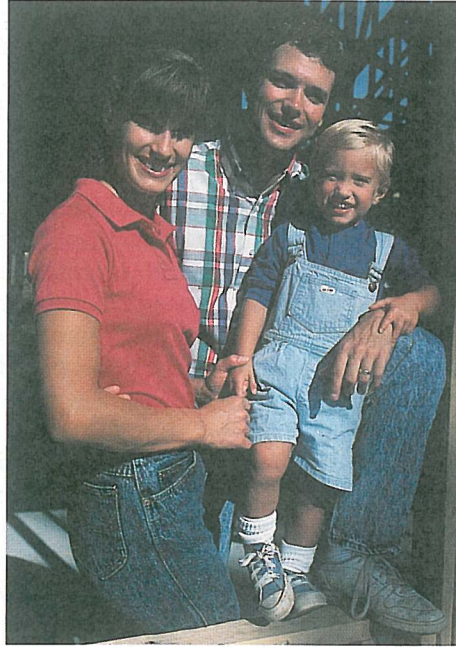
In the 22 pairs of identical chromosomes, each gene has at least two states or conditions, one on each chromosome strand in the pair. These alternative states are called **alleles**. Whatever the allelic state of the gene from one parent, the other parent's allele for that gene may be either the same or different. If both alleles are the same, the gene is said to be **homozygous**. If the alleles are different, the gene is **heterozygous**.

### Genotype and Phenotype

The genetic information about a trait is called the **genotype** (for example, the genetic information that guides skin color). The observed characteristic (for example, one's actual skin color) is called the **phenotype**. Genotype influences phenotype in three different ways. First, the differences in the allelic states of a gene sometimes result in a **cumulative relation**, in which more than one pair of genes influences the trait. An example of this kind of relation is the genetic contribution to height. A person who receives mostly "tall" genes will be tall; a person who receives mostly "short" genes will be short. Most people receive a mix of "tall" and "short" genes and are of average height.

Second, the differences between alleles may result in **codominance**, a state in which both genes are expressed in the new cell. An example of codominance is the AB blood type, which results from the joining of an A blood-type allele and a B blood-type allele. This blood type is not a mixture of A and B, nor is A subordinate to B or B to A; instead, a new blood type, AB, is formed.

Two dark-haired parents can have a blond child if both parents are heterozygous for blond hair color.



child have blue eyes. As the figure shows, on the average only 25% of the offspring of heterozygous parents have blue eyes.

In the case of a dominance relation, genetic information is not always observed in some outward characteristic. For example, people with alleles BB and Bb may both have brown eyes, even though they have different genetic information. For brown and blue eye color, there are two phenotypes (brown and blue) but three genotypes—two dominant alleles (BB), two recessive alleles (bb), or one dominant and one recessive allele (Bb or bB).

### Sex-Linked Characteristics

Certain genetic information is said to be **sex-linked** because the gene for the specific characteristic is found on the sex chromosomes. The female ova carry only X chromosomes. Half of the male sperm carry Y chromosomes, and half carry X chromosomes. Male children can be produced only when a sperm carrying a Y chromosome fertilizes an egg, and the result is an XY combination in the 23rd chromosome pair. All sperm carrying X chromosomes will produce female children.

Sex-linked traits are more likely to be observed in males, even though they are present in the genotype of females. You will understand this more readily if you visualize the XY chromosome pair. When a trait is carried on the Y chromosome, it will be inherited and transmitted only by males, since only males have the Y chromosome.

**FIGURE 4.3**

Probability of heterozygous parents producing a blue-eyed offspring

		Mother		
		B	b	
Father	B .5	BB .25	Bb .25	← Probability of passing the allele to offspring
	b .5	bB .25	bb .25	← Probability of the pairing
				← Probability of blue eyes

Note: Whenever B allele is present, eyes will be brown.

Third, the differences in the allele states of a gene may result in a dominance relation. **Dominance** means that, if one allele is present, its characteristic is always observed whether or not the other allele of the allelic pair is the same. The allele that dominates is called the **dominant gene**. The other allele that is present, but whose characteristic is masked by the dominant gene, is called the **recessive gene**. Eye color is the result of a dominance relation. The gene for brown eyes (B) is dominant over the gene for blue eyes (b). The probability that the recessive trait of blue eyes will emerge in the offspring of two heterozygous parents is illustrated in Figure 4.3. The possible combinations of the gene related to brown or blue eye color are BB, Bb, bB, and bb. Only if both parents carry the b allele and it is present in each of the gametes that form the offspring will the

Interestingly, the Y chromosome is quite small, and very few exclusively Y-linked traits have been identified. One of the key genes that has been identified on the Y chromosome is referred to as **testis-determining factor (TDF)**. This gene (or genes) is responsible for setting into motion the differentiation of the testes during embryonic development. Once the testes are formed, they begin to produce hormones that account for the further differentiation of the male reproductive system.

In addition to the Y-linked genes, sex-linked traits that are carried on the X chromosome are more likely to be observed in males than in females, because males do not have a second X chromosome with which to offset the effects of an X-linked trait. Currently, a map of the human X chromosome is being constructed through a coordinated, international research effort. The X chromosome comprises about 160 million base pairs. Genes associated with 26 inherited diseases have already been reproduced, and the location of genes associated with 50 others has been identified (Mandel et al., 1992).

Hemophilia is an example of a sex-linked trait. **Hemophiliacs** lack a specific blood protein that causes blood to clot after a wound (Lawn & Vohar, 1986). The allele for hemophilia is carried on the X chromosome. If the allele is either heterozygous or homozygous for the dominant characteristic (normal clotting), a female child will have normal blood-clotting capability. Only if she is homozygous for the recessive characteristic (a very rare occurrence) will she be hemophilic. The male, on the other hand, has only one allele for the blood-clotting gene, which he inherits from his mother. If that allele is dominant, his blood will clot normally; if it is recessive, he will be hemophilic (Figure 4.4).

There are other genes that are expressed exclusively in one sex but are not found on the sex chromosomes per se. For example, the genes for male beard development and female breast development are not located on the sex chromosomes. However, these characteristics will emerge only in the presence of the appropriate hormonal environment, which is directed by the sex chromosomes.

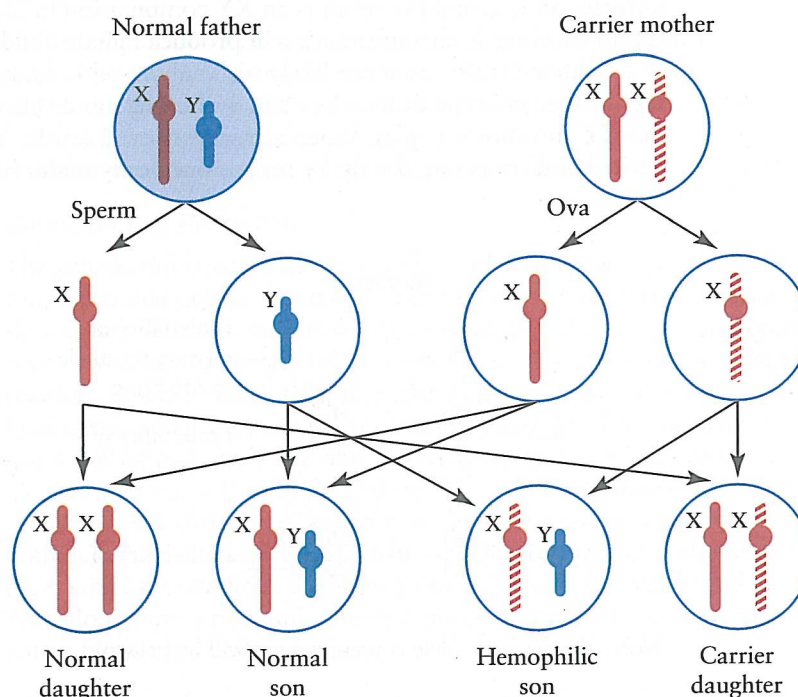
### Genetic Sources of Individuality

The study of genetics reveals that individual variability is due to more than the many variations in environment and experience that confront a growing person. Variability is built into the mechanisms of heredity. Each adult couple has the potential for producing a great variety of genetically distinct children. Three areas in which genetic determinants contribute to individual variability are rate of development, individual traits, and abnormal development.

**FIGURE 4.4**

*Sex-linked inheritance of hemophilia. Sex-linked inheritance of hemophilia results from the location of the factor VIII gene on the X chromosome. A male carrying a mutant factor VIII gene lacks normal factor VIII and is hemophilic. A female carrier is protected by the normal gene on her second X chromosome, but half of her daughters will be carriers and half of her sons will be hemophilic. When a father is hemophilic (not shown), his sons will not be hemophilic, because they receive his Y (not his X) chromosome, but his daughters will be carriers.*

SOURCE: Lawn & Vohar, 1986.



### **Genetic Determinants of Rate of Development**

Genes regulate the rate and sequence of maturation. The concept of an epigenetic plan for growth and development is based on the assumption that a genetically guided system promotes or restricts the growth of cells over the life span. Genetic factors have been found to play a role in behavioral development, including the onset of various levels of reasoning, language, and social orientation.

Considerable evidence of the role of genetics in guiding the rate and sequence of development has been provided by studies of identical twins (who have the same genetic structure). The rates at which identical twins develop are highly correlated, even when those twins are reared apart. A number of characteristics, including the timing of the acquisition of motor skills, personality development, changes in intellectual capacity among aged twins, and the timing of physical maturation, show a strong genetic influence (Holden, 1987).

Genes can be viewed as internal regulators that set the pace for maturation. They signal the onset of significant developmental changes throughout life, such as growth spurts, the eruption of teeth, puberty, and menopause. They also appear to set the limits of the life span. A small number of genes influences how many times the cells of a specific organism can divide and replicate (Marx, 1988). Research on three different animal species—fruit flies, worms, and mice—shows that when the most long-lived of a species are bred, the offspring have a longer life than average (Barinaga, 1991).

Differences in the rate of development contribute to our understanding of psychosocial growth. These differences bring different children into contact with new aspects of their environments and provide them with changing capacities at different chronological ages. Thus, the genetic processes that regulate readiness for certain kinds of growth and vulnerabilities to particular kinds of stress contribute to systematic differences among individuals. For example, adult expectations for the accomplishment of such specific tasks as toilet training, getting dressed without help, and learning to write interact with the child's developmental level. Disappointment may be conveyed to developmentally "late" children, and pride and approval may be conveyed to developmentally "accelerated" children.

### **Genetic Determinants of Individual Traits**

Genes contain specific information about a wide range of human characteristics, from eye color and height to the ability to taste a particular substance called **phenylthiocarbamide** (which to tasters is bitter, but to nontasters has no taste at all). Some characteristics are controlled by a single gene. However, most significant characteristics, such as height, weight, blood group, skin color, and intelligence, are controlled by the combined action of several genes. When multiple genes are involved in the regulation of a single trait, the possibilities for individual differences in that trait increase. Since many characteristics are regulated by multiple genes, the variety of human genotypes is enormous.

Genetic factors also play a substantial role in individual differences in personality (Plomin, 1990, 1994; Loehlin, 1992; Holden, 1987). Traits such as sociability (a tendency to be outgoing), inhibition (a tendency to be cautious and socially shy or withdrawn), and neuroticism (a tendency to be anxious and emotionally sensitive) are pervasive dimensions of personality that appear to have strong genetic components. Research on the biological basis of sexual orientation suggests that genes may influence the development of the part of the brain that guides sexual behavior (LeVay, 1991). Identical twins are more likely to have the same sexual orientation than are fraternal twins or adoptive siblings (Bailey & Pillard, 1991; 1994). Even in rather specific areas of personality, such as political attitudes, aesthetic preferences, and sense of humor, identical twins show greater similarity than fraternal twins, even when the identical twins are reared apart from each other.

Extending the analysis of the impact of genetics on individual differences, Sandra Scarr (1992) suggested at least three ways in which genetic factors influence the environments of individuals, thus increasing the impact of genetics on the expression and



*The many possible combinations of genetic information can produce tremendous individual variation. In this painting, Picasso shows how basic geometric shapes can be combined in a variety of ways to produce human-like figures, similar and yet each unique.*

elaboration of individual differences. First, most children are raised by their parents in environments created by their parents. Thus, children receive both their genes and their environment from a common genetic source—their parents. As an example, a parent who is temperamentally sociable is more likely than a withdrawn or timid parent to have lots of people at the house, to enjoy the companionship of others, and therefore to expose his or her children to more companionate adults. Second, people draw out responses from others that are related to their own personality characteristics. Thus, broad, genetically based aspects of one's individuality will affect the kinds of social responses one receives from others, including one's parents. Third, as people mature and become increasingly assertive in selecting certain experiences and rejecting others, their own temperaments, talents, intelligence, and level of sociability will guide the kinds of environments they select and will strengthen certain genetic predispositions, while dampening others.

### **Genetic Determinants of Abnormal Development**

In addition to characteristics such as physical appearance, temperament, talent, and intellectual capacity, a wide variety of abnormalities, or **anomalies**, have a genetic cause. The most dramatic anomalies result in a spontaneous abortion of the fetus early in the pregnancy. It is estimated that a majority of the spontaneous abortions that occur early in pregnancy are due to results of chromosomal abnormalities in the fertilized zygotes (the developing organism formed from the father's sperm and the mother's egg) (Clayman, 1989).

Of those infants who survive the neonatal period, an estimated 3–5% of newborns have one or more major recognizable anomalies (Cunningham et al., 1997). The incidence of anomalies increases to 6 or 7% as some disorders are diagnosed later in childhood. Some birth defects are linked to a specific chromosome or a single gene. Other birth defects are linked solely to environmental factors, such as drugs, medications, and fetal and maternal infections. The majority of malformations, however, result from the interaction of genetic vulnerabilities in the presence of environmental hazards or are of unknown origin (Moore, 1993).

Some examples of genetic and chromosomal disorders are listed in Table 4.1. The disorders are presented in two broad categories: those associated with specific genes and those associated with chromosomal abnormalities. Within those categories, some disorders are found on 1 of the 22 pairs of autosomal chromosomes (chromosomes other than the sex chromosomes), and others are on one of the sex chromosomes. Among the genetic disorders that result from a dominant gene, about 300 have been identified; among those that result from a recessive gene, about 250 have been identified. Molecular biologists have begun to identify the chromosomal site of a number of genetic disorders. This work will lead to a clearer understanding of the molecular mechanisms that account for these disorders and, eventually, to the development of gene therapies for many genetically caused abnormalities (Anderson, 1992; Crystal, 1995).

Certain genetic diseases are linked directly to our ancestry; therefore, the incidence of some genetic diseases is higher in certain populations than in others (Thompson et al., 1991; Emery & Mueller, 1992). For example, Tay-Sachs disease is present in 1 out of 3900 infants born to Ashkenazi Jews (Jews who settled in eastern Europe) but only

TABLE 4.1 Examples of Genetic and Chromosomal Disorders

- I. Genetic disorders
- A. Autosomal dominant gene
1. *Huntington's chorea*: rapid jerky, involuntary movements; deterioration of muscle coordination and mental functioning. Symptoms usually do not appear until age 35–50. Caused by genetic defect on chromosome 4.
  2. *Marfan's syndrome*: Elongated fingers; deformed chest and spine; abnormal heart. Tendons, ligaments, and joint capsules are weak.
- B. Autosomal recessive gene
1. *Albinism*: Hair, skin, and eyes lack the pigment melanin. Often accompanied by visual problems and a susceptibility to skin cancer.
  2. *Cystic fibrosis*: Certain glands do not function properly. The glands in the lining of the bronchial tubes produce excessive amounts of thick mucus, which lead to chronic lung infections. Failure of the pancreas to produce enzymes necessary for the breakdown of fats and their absorption from the intestines leads to malnutrition. Sweat glands are also affected. Often fatal by age 30. Caused by missing base pairs on chromosome 7.
  3. *Sickle-cell anemia*: Malformation of red blood cells reduces the amount of oxygen they can carry. Results in fatigue, headaches, shortness of breath on exertion, pallor, jaundice, pain, and damage to kidneys, lungs, intestine, and brain.
  4. *Tay-Sachs disease*: Absence of a certain enzyme results in the buildup of harmful chemicals in the brain. Results in death before age 3.
- C. X-linked recessive
1. *Color blindness*: Defect of light-sensitive pigment in one or more classes of cone cells in the retina of the eye and/or an abnormality in or reduced number of cone cells themselves. The two common types are reduced discrimination of light wavelengths within the middle (green) and long (red) parts of the visible spectrum.
  2. *Hemophilia*: Absence of a blood protein (factor VIII) reduces effectiveness of blood clotting. Severity of disorder varies. Bleeding episodes likely to begin in toddlerhood.
- II. Chromosomal disorders
- A. Autosomal abnormality
1. *Down syndrome*: Additional 21st chromosome; also called trisomy 21. The excess chromosome results in physical and intellectual abnormalities, including IQ in the range of 30–80; distinctive facial features, heart defects, intestinal problems, hearing defects; susceptibility to repeated ear infections. Tendency to develop narrowing of the arteries in adulthood, with attendant increase in risk of heart disease.
- B. Sex-chromosome abnormalities
1. *Turner's syndrome*: Usually caused by a lack of one X chromosome in a girl; sometimes one of two X chromosomes is defective; occasionally some cells are missing on an X chromosome. These abnormalities result in defective sexual development and infertility, short stature, absence or retarded development of secondary sex characteristics, absence of menstruation, narrowing of the aorta, and a degree of mental retardation.
  2. *Klinefelter's syndrome*: One or more extra X chromosomes in a boy. This abnormality results in defective sexual development, including enlarged breasts and small testes, infertility, and often mental retardation.

SOURCE: Based on information provided in Clayman, 1989.

1 in 112,000 other U. S. infants. *Thalassemia*, a disease involving faulty production of hemoglobin (which carries oxygen in the blood), is found most often in people of Mediterranean, Middle Eastern, and Southeast Asian origins. The variety of genetic abnormalities serves to broaden the range of individual variability. Many of the irregularities pose a challenge both to the adaptive capacities of the affected person and to the care-giving capacities of the adults involved.

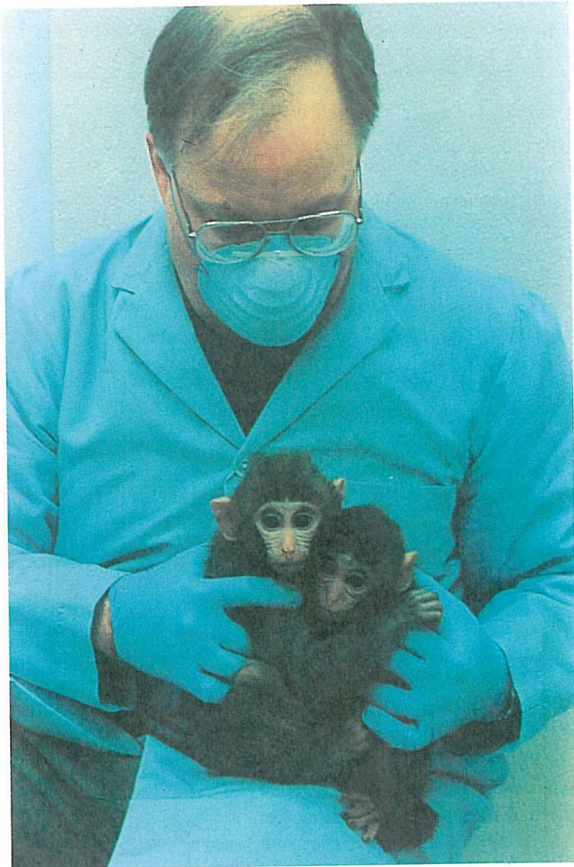
## Genetic Technology and Psychosocial Evolution

Psychosocial evolution has typically been differentiated from biological evolution in that change is accomplished by social mechanisms rather than incorporated in the genetic structure. As a result of scientific knowledge, however, we are entering an era when it is possible to intervene to influence the genotype. One such intervention is **genetic counseling**. Individuals and couples with a family history of a genetic disease, or who for some other reason worry about the possibility of transmitting a genetic disease to their children, can have a blood test to identify genes that may result in the inherited disorder. The location of the genes that account for such abnormalities as Tay-Sachs disease, sickle-cell anemia, Duchenne muscular dystrophy, and cystic fibrosis have been identified. Recent discoveries make it possible to identify genes responsible for certain forms of prostate, breast, and colon cancers. Couples who have reason to believe that they may carry genes for one of these diseases can be advised about the probability of having children who may be afflicted (Somerson, 1997). If significant numbers of the carriers of genetic diseases decided not to reproduce, the incidence of these diseases in the population would decline over time. Thus, a psychosocial intervention would modify the gene pool.

### Mapping the Genome

In the future, genetic technology promises to take us even further in the direct modification of the genetic structure of an individual. A project to map the **human genome**—to identify and list in order all of the genome's approximately 3 billion base pairs—was





*Cloning of animals for research and medical purposes has raised new questions about the ethics of genetic research, especially its applicability to humans. What are some of the ethical considerations that come to mind as you think of cloning human beings?*

### Evaluating the Impact of Heredity on Behavior

enhance aspects of normal development in humans. Some people are finding that even the advances in identifying genetic markers for specific diseases can lead to ethical dilemmas. Individuals are turning down the opportunity to be tested for possible genetic diseases for fear that they will be denied health insurance, life insurance, or employment (Beardsley, 1996).

The possibility of reproducing genetically identical clones from adult human tissue has stirred the conscience of the religious, political, and medical communities. Is it ethical to clone human genes so that great scholars, scientists, and artists can walk the earth again? Should cloning become an approved technology for coping with infertility? Of course, based on what we know of the dynamic interaction between genetics and environment, the similarity of an adult human clone to its genetic parent may not be as great as that of an adult cloned sheep or calf to its genetic parent. Through discussion, debate, research, observation of events, and court cases, a set of ethics is being hammered out that not only deals with specific issues but also sets the tone for the way life itself is conceptualized (Kluger, 1997; Woodward, 1997).

One way to summarize the influences of genetics on behavior is to view the genotype as establishing a **reaction range**—that is, a range of possible responses to environmental conditions, the limits of which are determined by one's genotype. Under similar environmental conditions, genetic differences are most likely to be expressed; however, when environmental conditions vary, the advantages of one genotype over another may be masked by the adversities or opportunities of the situation. Figure 4.5 shows the hypothetical reaction ranges of three children with respect to intelligence. Child A has greater genetic potential for intelligence than Child B, who has greater potential than Child C. When all three children are in unstimulating environments, their IQs develop at the lower end of their potential range. When all three children are in stimulating environments, their IQs develop toward the upper end of their potential range. If the three children are in different environments, the differences in genetic potential

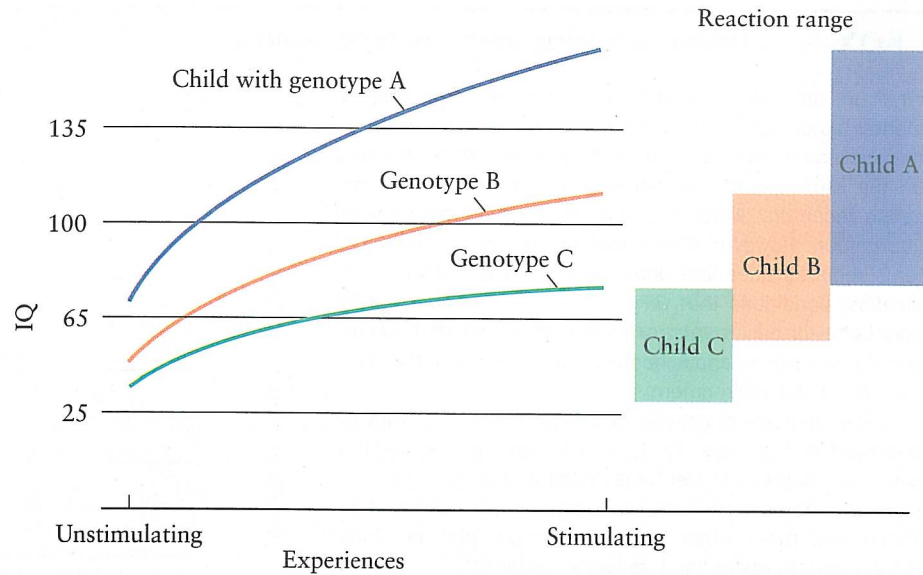
initiated in January 1989 by the National Institutes of Health (NIH). The project, expected to be completed between 2002 and 2005, will permit specialists to predict an individual's susceptibility to genetic diseases, to treat genetically caused diseases, and possibly to "enhance" a person's genetic potential through the introduction of gene modifications (NIH/CEPH Collaborative Mapping Group, 1992; Beardsley, 1996). As an example, the gene that is defective in patients with *cystic fibrosis*, a condition that occurs in children and young adults in which a thick, sticky mucus obstructs the airways and increases vulnerability to infection, has recently been identified (Collins, 1992). Cystic fibrosis is the most common and potentially lethal of the autosomal recessive diseases among Caucasians. Now that the biochemical process associated with the defective gene has been analyzed, a wide variety of therapeutic techniques are being developed, including both drug and gene therapy.

### Ethical Considerations

Gene transfer, the patenting of new life forms created through genetic engineering; genetic fingerprinting, which is used to help identify criminal suspects; and, most recently, cloning from an adult mammal are topics that are raising new ethical concerns. There is a general consensus that gene therapy used to treat serious diseases such as cystic fibrosis or cancer is ethical. However, there is less agreement about the use of intervention to alter the genetic code at the level of the zygote or to attempt to introduce genes intended to

**FIGURE 4.5**

Hypothetical reaction ranges of intelligence for three genotypes



may be masked by the way the environments act on this potential. If Child B and Child C are in stimulating environments and Child A is in an unstimulating environment, Child B may have the highest measured IQ, and the IQs of Children C and A may be lower and very similar. Each child's intellectual ability can be expressed as a range that is a product of the interaction of genetic potential and environment (Box 4.1).

The concept of the reaction range can be seen clearly in the outlook for children with **Down syndrome** (Patterson, 1987). This condition, which occurs in 1 of every 700 live births, is the most common genetic cause of mental retardation in the United States. In the early part of this century, children born with Down syndrome had a life expectancy of 9 years. Today, the life expectancy of a child with Down syndrome is 30 years, and 25% live to age 50. Medical care, early and constant educational intervention, physical therapy, and a nurturing home environment have significant, positive results for children with Down syndrome. Under optimal conditions, individuals with Down syndrome are able to achieve a moderate degree of independence and to participate actively in the life of their schools, communities, and families.

*The concept of reaction range is illustrated in the high level of functioning evidenced by this girl with Down Syndrome. With optimal home and school support, she can enjoy interacting with peers and learn many skills required for self-sufficiency.*

