2 Genes and Prenatal Development

I was picking up my daughter Rachel from kindergarten when another mother, Stephanie, pulled me aside. She whispered that she had seen Rachel fall on the sidewalk; her little finger was slightly bent and might need medical attention. A month earlier, Stephanie said, her son had fallen. He had had a similarly bent finger, which turned out to be broken.

Rachel was playing happily, but I saw that her finger was indeed crooked. I walked her home and consulted my husband. He smiled and spread out his hands, revealing a similarly bent little finger. Aha! An inherited abnormality, not an injury. I was relieved that we could avoid the emergency room, but I faulted myself for not noticing that finger earlier.

STEPHANIE AND I HAD BOTH mistaken a genetic quirk (heredity) for a potentially serious injury (environment). Many people experience this kind of confusion because they don't think about genes unless a problem appears.

As you remember, both genes and the environment affect every human characteristic. In this chapter, we begin with genetics. We

CHAPTER OUTLINE

then look at prenatal growth and the miracle of birth. We conclude with the interaction between nature and nurture in nearsightedness, alcoholism, and certain birth defects.

Problems are not the main theme of the chapter, however. Above all, this chapter describes an amazing process—how a single tiny cell becomes a wiggling, squalling, 7-pound human being.

CHAPTER INTRODUCTION

The Embryonic Period: From Two Through Eight Weeks

The Fetal Period: From Nine Weeks Until Birth

Birth

The Process of Birth

Traditional and Modern Birthing Practices

Not Waiting for Nature

Low Birthweight

Mothers, Fathers, and a Good Start

Nature, Nurture, and the Phenotype

Nearsightedness

Alcoholism

Prenatal Teratogens

Resolving Uncertainties

2.1 The Beginning of Life

Every person starts life as a single cell, which from the beginning is distinct from all other cells. As this section describes, conception is a prime example of both the universal processes and the unique characteristics that are evident in human life.

The Beginning of Life

2.1.1 Chromosomes and Genes

All living things are composed of cells that promote growth and sustain life according to instructions in their molecules of **DNA (deoxyribonucleic acid)**. Each molecule of DNA is called a **chromosome** (see **Figure 2.1**). Human cells have 46 chromosomes arranged in 23 pairs, with one important exception.

That exception is the reproductive cell, called a **gamete.** Each gamete —*sperm* in a man and *ovum* in a woman—has only 23 chromosomes, one from each of a person's 23 pairs of chromosomes. Each man or woman can produce 2^{23} different gametes, or more than 8 million versions of their 46 chromosomes.

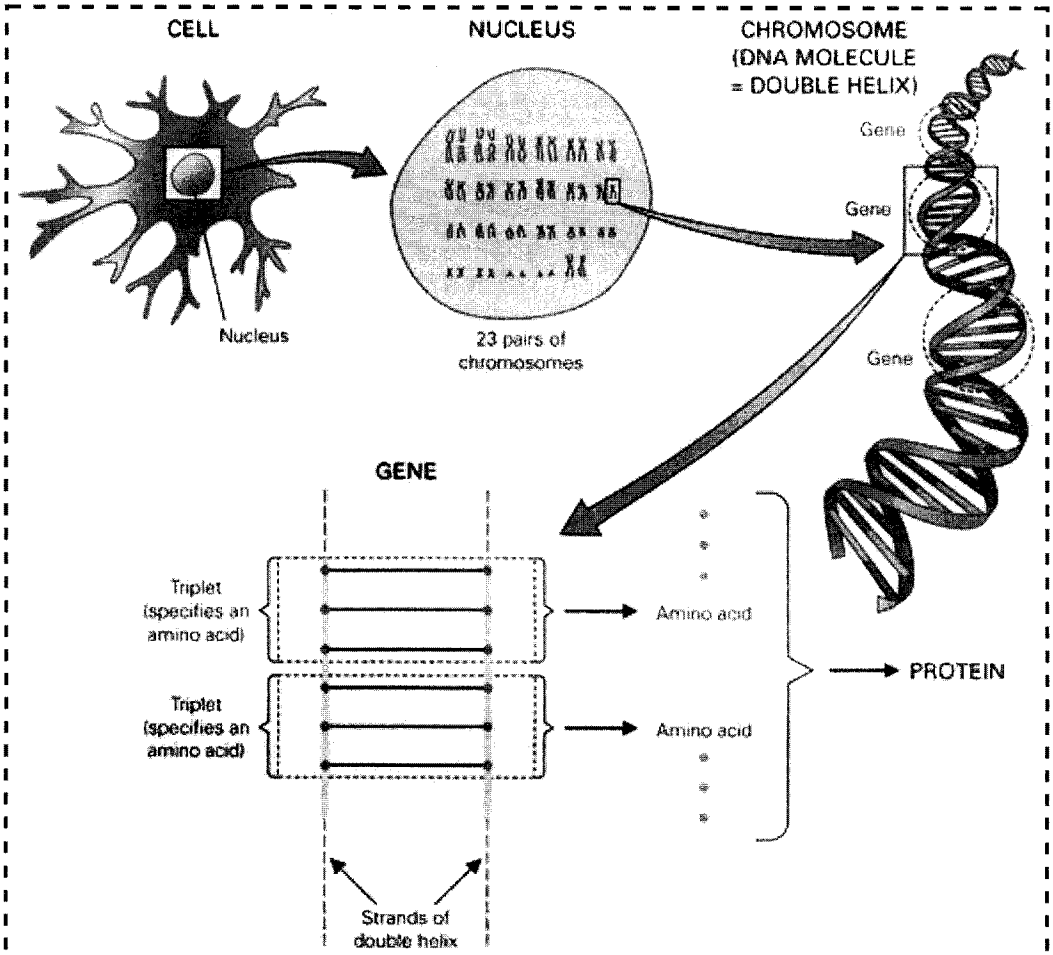


Figure 2.1 How Proteins Are Made The genes on the chromosomes in the nucleus of each cell instruct the cell to manufacture the proteins needed to sustain life and development.

When a sperm and an ovum combine, they create a new cell in which each of the 23 chromosomes from one parent is paired with a gamete with 23 from the other, for a total of 46 chromosomes. This new cell is called a **zygote.** In each zygote, half the chromosomes are from the mother and half are from the father. Each person began as a zygote, with all the DNA instructions for development contained in that one cell.

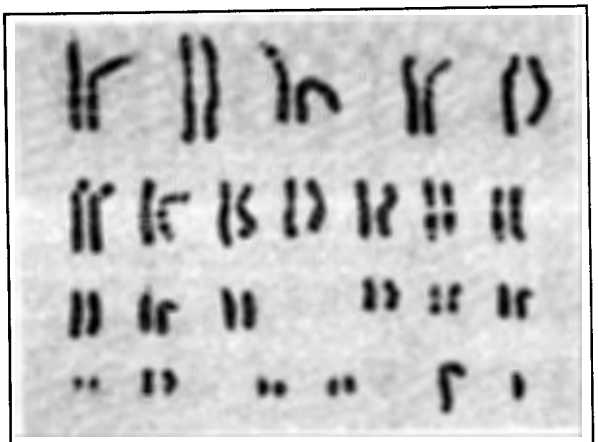
These instructions are organized into units called **genes,** with each gene located on a particular one of the 46 chromosomes. Thus, every gene is a separate section of a chromosome, and each gene contains certain instructions for development.

Genes carry their instructions via four chemicals, whose names are abbreviated A, T, C, and G. These four chemicals occur in four possible pairs —AT, TA, CG, and GC—that are strung together in triplets (sets of three pairs) resembling the rungs of a ladder. Each person has about 3 billion pairs, which carry the instructions to form that person.

GENETIC VARIATIONS AND SIMILARITIES

Genes are passed down from generation to generation. Half of a person's genes come from each parent, which means one-fourth come from each grandparent, one-eighth from each great-grandparent, and so on.

The Moment of Conception This ovum is about to become a zygote. It has been penetrated by a single sperm, whose nucleus now lies next to the nucleus of the ovum. Soon, the two nuclei will fuse, bringing together about 20,000 genes to guide development. COURTESY OF LENNART NILSSON/BONNIER FAKTA/STOCKHOLM



Mapping the Karyotype A karyotype portrays a person's chromosomes. To create a karyotype, a cell is grown in a laboratory, magnified, and then usually photographed. The photo is cut into pieces and rearranged so that the matched pairs of chromosomes are lined up from largest (top left) to smallest (bottom row, fourth pair from the left). Shown at the bottom right is the 23rd chromosome pair: These two do not match, meaning that this karyotype shows a male (XY). L. WILLATT/EAST ANGLIAN REGIONAL GENETICS SERVICE/PHOTO RESEARCHERS, INC

Together, all the genes make up the organism's genetic inheritance, or **genotype**, which is unique for each person. The genotype is quite different from the **phenotype,** which is a person's actual appearance and manifest behavior.

Many genes are identical for every human being. However, some genes vary slightly in their codes from one person to another. Each such variation of a gene is called an **allele.** For example, the two versions of the MAOA gene you read about in **Chapter 1** are alleles of the same gene. Some alleles

result in life-threatening conditions, others merely differentiate one person from another, and still others have no effect on the phenotype that scientists can detect (Marcus, 2004).

The Human Genome Genetic diversity not only distinguishes each person (you can immediately spot a close friend in a crowd) but also teaches us to appreciate human differences. As one expert said, "What's cool is that we are a mosaic of pieces of genomes. None of us is truly normal" (Eichler, quoted in J. Cohen, 2007b, p. 1315).

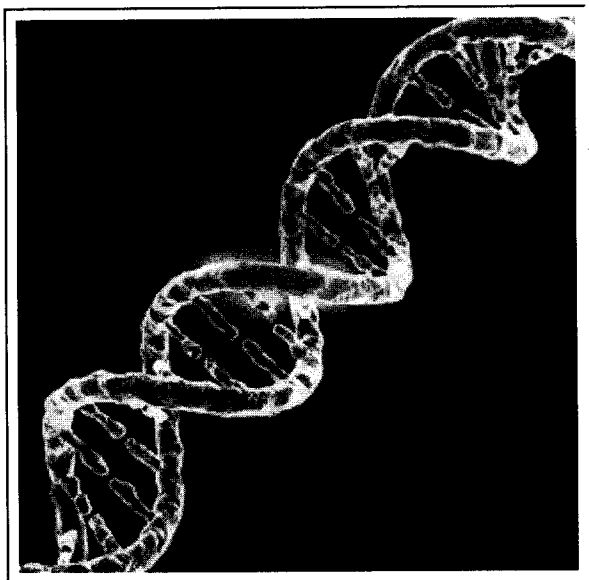
Moreover, genetic diversity enables the human species to adapt to many climates, from the frigid Arctic to the steamy tropics. Even devastating diseases do not necessarily kill us all, because of our diverse genes. For instance, a few people have an allele that protects them from the HIV virus that causes AIDS (E. Gonzalez et al., 2005). Similarly, genotype differences allowed some of our ancestors to survive tuberculosis, malaria, the Black Death, and other scourges.

The entire packet of instructions to make a living organism is the **genome.** There is a genome for every species, even for every kind of plant. The human genome is 99.5 percent identical for any two persons (J. Cohen, 2007b); the genomes for humans and chimpanzees are 98 percent the same; the genomes for humans and every other mammal are at least 90 percent the same. All these shared genes allow scientists to learn about human genetics from other creatures, especially mice, by transposing, deactivating, enhancing, and duplicating their genes.

The more scientists experiment with genes, the more they are amazed. A worldwide effort to map all the human genes led to the *Human Genome Project*, which was virtually completed in 2003 and which continues to reveal surprises. One surprise was the discovery of how many genes humans actually have.

Until 2001, scientists thought humans had about 100,000 genes, but that turned out to be a gross overestimate. The Human Genome Project found only about 20,000 genes. The precise count is still unknown, partly because of another surprise: It is not always clear exactly where one gene ends and another begins (Pennisi, 2007).

A more recent international project, called the *HapMap*, aims to spot all the variations in the human genome. The HapMap has noted 11 million variations among the 3 billion chemical pairs that constitute the human genome (Hinds et al., 2005).



Twelve of 3 Billion Pairs This is a computer illustration of a small segment of one gene, with several triplets. Even a small difference in one gene, such as a few extra triplets, can cause major changes in a person's phenotype. HYBRID MEDICAL ANIMATION/PHOTO RESEARCHERS, INC

Sibling Similarities Full siblings get half their genes from each parent, but the particular half depends on which of the millions of possible gametes combined to start their development. Two siblings may have the exact same gene (a 50/50 chance), but in a family with several children, about half will have that particular gene and half will not; those who share one gene do not necessarily share the next one.

This is evident when looking at a family with many children. For example,

two siblings may both happen to inherit their father's red hair, but one may have blue eyes (like the mother and the father's father) and the other, brown eyes (like the father and the father's mother). Given that each child has about 10,000 genes from each parent, with half a million common variations and millions more uncommon ones, billions of combinations are possible.

Of course, both parents might have some identical gene pairs; thus, all their children will have the same pair, one from each parent. For instance, if both parents have a pair of blue-eye genes, both will have blue eyes—and so will all their children. But for the many genes and alleles that differ, full siblings are not necessarily alike, even though they all come from the same man and woman.

Twins There is one major exception to genetic diversity. Although every zygote is genetically unique, about once in every 250 human conceptions, the zygote not only duplicates but splits apart completely, creating two, or four, or even eight separate zygotes, each identical to that first single cell. If each of those separated cells implants and grows, multiple births occur. One separation results in **monozygotic twins,** from one *(mono)* zygote (also called *identical twins*). Two or three separations create monozygotic quadruplets or octuplets. (An incomplete split creates *conjoined twins*, formerly called Siamese twins.)

Same Birthday, Same (or Different?) Genes

Twins who are of different sexes or who have obvious differences in personality are dizygotic, sharing only half of their genes. Many same-sex twins with similar temperaments are dizygotic as well. One of these twin pairs is dizygotic; the other is monozygotic.

DAVID YOUNG-WOLFF/PHOTOEDIT JOHNER IMAGES/GETTY IMAGES

Because monozygotic multiples originate from the same zygote, they have the same genotype, with identical genetic instructions for physical appearance, psychological traits, vulnerability to diseases, and everything else. However, because nurture always affects nature, even before birth, identical twins do not have exactly the same phenotype.

OBSERVATION QUIZ

Can you tell which pair is monozygotic? (see answer below)

Dizygotic twins, also called *fraternal twins*, occur about twice as often as monozygotic twins. They began life as two *(di)* zygotes created by two ova fertilized by two sperm at roughly the same time. (Usually, only one ovum leaves the ovaries per month, but sometimes two or more ova are released.) Triplets can be trizygotic, and so on.

Dizygotic twins, like any other siblings, have half their genes in common. They can look quite different (about half are male/female pairs) or they can

look quite similar, again like other siblings.

The incidence of dizygotic twins is genetic, because some women inherit the tendency to release more than one ovum when they ovulate. This genetic tendency varies by ethnicity. For example, dizygotic twins occur about once in every 11 births among the Yoruba in Nigeria, once in 100 among the British, and once in 700 among the Japanese (Gall, 1996; Piontelli, 2002). On the contrary, the incidence of monozygotic twins is not genetic; they are equally rare in every ethnic group.

Age also matters. Older women are more likely to release more than one ovum at a time and thus to have dizygotic twins.

MALE AND FEMALE

In 22 of the 23 pairs of human chromosomes that each person inherits, the chromosomes of each pair are closely matched. These 44 chromosomes are called *autosomes*, which means that they are not connected to the sex chromosomes (the other two).

The 23rd pair of chromosomes is a special case. In females, it is composed of two large X-shaped chromosomes. Accordingly, it is designated **XX.** In males, the 23rd pair has one large X-shaped chromosome and one smaller Y-shaped chromosome. It is called **XY.**

Because a female's 23rd pair is XX, every ovum that her body creates contains either one X or the other—but always an X. Because a male's 23rd pair is XY, half of his sperm carry an X chromosome and half carry a Y. The Y chromosome has a gene that directs the developing fetus to make male organs. Thus, the sex of the developing organism depends on which sperm penetrates the ovum—either an X sperm, which creates a girl (XX), or a Y sperm, which creates a boy (XY) (see **Figure 2.2**).

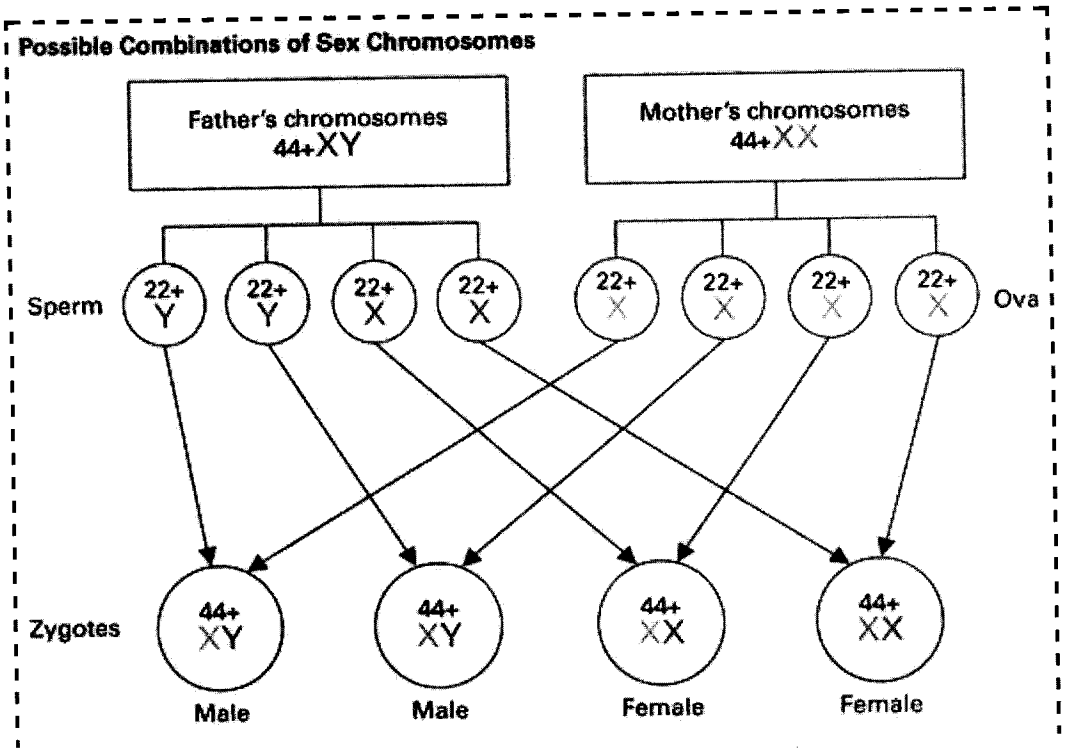


Figure 2.2 Determining a Zygote's Sex Any given couple can produce four possible combinations of sex chromosomes; two lead to female children and two, to male. In terms of the future person's sex, it does not matter which of the mother's Xs the zygote inherited. All that matters is whether the father's Y sperm or X sperm fertilized the ovum. However, for X-linked conditions, it matters a great deal because typically one, but not both, of the mother's Xs carries the trait.

The natural sex ratio at conception is close to 50/50, since each sperm has one chromosome of the father's 23rd pair of chromosomes, equally often a Y and an X. The sex ratio at birth is affected by serious adversity (such as famine) because males are more likely to be spontaneously aborted early in prenatal development.

Notice that if the 23rd pair is XY (male), it includes genes that are not paired. In XX (female) zygotes as well, one of the two X chromosomes is deactivated early in prenatal development, so genes on one X cannot pair with those on the other X.

Likewise, autosomes can carry genes that are not paired, or that dominate the other member of the pair, or that are activated only if particular other genes or nongenetic influences (the nutrients and transcription factors surrounding each gene) are present. Thus, for many reasons, the interaction among and between genes varies. Some of these variations are now described.

ANSWER TO OBSERVATION QUIZ

(from above) The Japanese American girls are the monozygotic twins. If you were not sure, look at their teeth, their eyebrows, and the shape of their faces, compared with the ears and chins of the boys.

Chromosomes and Genes

2.1.2 Genetic Interactions

No gene functions alone. Almost every trait is **polygenic** (affected by many genes) and **multifactorial** (influenced by many factors). The same gene produces the legs of butterflies, cats, centipedes, and humans. Similarly, the eyes of flies, mice, and people all originate from one gene, called *Pax6*. Other genes direct the human fetus to grow only two legs and to develop human visual acuity.

To differentiate humans from other animals, the action of those other genes, called **regulator genes,** is crucial. Regulator genes direct polygenic interactions, controlling the genetic expression, duplication, and transcription of the other genes (Marcus, 2004). Human regulator genes allow distinctively human modes of talking, walking, and thinking to spring from the same genes that control the equivalent functions in other creatures.

ADDITIVE HEREDITY

Some genes are **additive genes.** Their effects *add up* to make the phenotype. When genes interact additively, the phenotype reflects all the genes that are involved. Height, hair curliness, and skin color, for instance, are influenced by additive genes. Indeed, height is affected by an estimated 100 genes, each contributing a small amount (Little, 2002). (Of course, nutrition and illness also affect the phenotype for height.)

Centuries of marriage between people with different ancestries means that most families include members who vary in height, hair curliness, skin color, and other additive physical traits. Thus, a child's phenotype may not reflect either parent's phenotype, although it always reflects their genotype.

DOMINANT–RECESSIVE HEREDITY

Less common are *nonadditive* genes, which do not contribute equal shares. In one nonadditive form of heredity, alleles interact in a **dominant–recessive pattern,** in which one allele, the *dominant gene*, is far more influential than the other, the *recessive gene*.

ESPECIALLY FOR Future Parents

Suppose you wanted your daughters to be short and your sons to be tall. Could you achieve that? (see response, page 50)

In the most unequal match-up, the dominant gene completely controls the phenotype, preventing the recessive gene from having any obvious effect. For example, the genes for blood type B and the Rh-positive blood factor are both dominant; the genes for blood type O and Rh-negative blood factor are both recessive. That means the blood of a person whose genotype is B-negative from one parent and O-positive from the other will be B-positive.

The reason for the B-positive blood is that, since the genes for both O and Rh-negative are recessive, neither is evident in the phenotype. The dominant genes for B and Rh-positive prevent the recessive genes from being expressed (see Appendix A, p. A-3, for more on heredity of blood types). A person can have type O blood or the Rh-negative factor only by inheriting the recessive gene from *both* parents.

Contrary to this example, however, many recessive genes are not *completely*

hidden when paired with a dominant gene. For example, because the gene for brown eyes is dominant and the gene for blue eyes is recessive, most people who have one gene for blue eyes and one for brown end up with brown eyes. However, some people with this pairing have hazel eyes, a color that hints at their recessive blue-eye gene.

Genetic Interactions

2.1.3 Carriers and Genetic Expression

As you learn more about the interactions among genetic and nongenetic influences on development, remember to distinguish between a person's *genotype*, or genetic *potential*, and *phenotype*, or the actual *expression* of that genetic inheritance in physical appearance, health, intelligence, and actions.

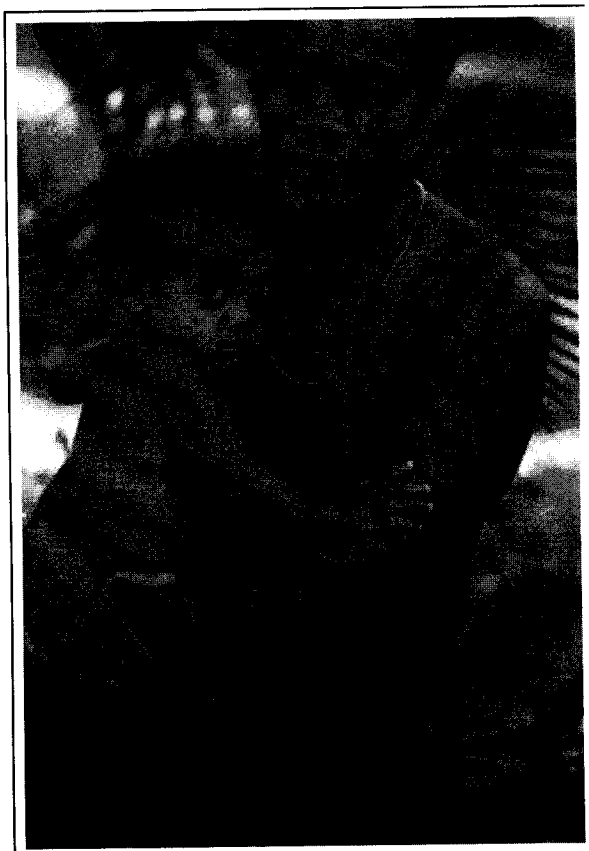
Everyone has many recessive genes in his or her genotype that are not expressed in the phenotype. In genetic terms, each of us is a **carrier** of unexpressed genes; that is, we "carry" a gene that will be transmitted to half our sperm or ova. Because we are merely carriers, we do not express that trait in our phenotype.

Since each gamete (reproductive cell) has half of each parent's genes, there is a 50/50 chance that a carried gene will be passed on to a child. If a child inherits a carried gene, whether or not that gene is expressed depends on many other genetic and nongenetic factors. Thus, the child could also be just a carrier, with the gene unexpressed, or could express that gene in the phenotype.

Heritability is a statistical term that indicates what portion of the variation in a particular trait within a particular population is inherited. Height, for instance, is highly heritable. Thus, when children are well nourished, about 90 percent of the height differences among children of the same age is genetic. Making a short 5-year-old drink his milk and eat his dinner will never make him a tall adult (unless he has tall genes but was previously severely malnourished).

Heritability is a useful measure within a population. But it is not accurate when applied from one population to another, or even to one individual in a particular population. For example, Rachel's bent little finger, mentioned in the chapter's opening, is the product of genes; finger shape is highly heritable. However, nurture always affects nature. As her mother, I wonder if that anomaly appeared because of something I did during early pregnancy, perhaps involving my diet, or stress, or sleep. I know that she inherited her finger from her father, but I will never know how much nurture affected heritability in that one case.

There is a general point to be made here. Every trait, action, and attitude has a genetic component: Without genes, no behavior could exist. But without environment, no gene could be expressed. The specifics are complicated, requiring longitudinal and multicultural research. Heritability is useful, but prediction in any one instance is not entirely certain.



Shyness Is Universal Inhibition is a psychological trait that is influenced by genetics. It is more common at some ages (late infancy and early adolescence) and in some gene pools (natives of northern Europe and East Asia) than others. But every community includes some individuals who are unmistakably shy, such as this toddler in Woleai, more than 3,000 miles west of Hawaii. CORBIS/THE PURCELL TEAM

KEY Points

- Humans have 46 chromosomes that match up in 23 pairs and about 20,000 genes.
- Genetic diversity is protective of the human species. Full siblings (except monozygotic twins) have only half their genes in common.
- Both parents' genes contribute equally to a child's genotype, but only sperm, not ova, determine the child's sex.
- Genes interact in many ways; the phenotype differs from the genotype. Everyone is a carrier of genes for some traits that are not expressed in the phenotype. Heritability is a useful statistic for a population group, but it does not predict individual differences.

Carriers and Genetic Expression

2.2 Genetic Problems

The chromosomal and genetic interactions just described are fascinating in themselves, but this knowledge has very practical implications. Sometimes a part of a gene has too many repetitions of a base pair; or a recessive allele pairs with another, similar allele; or a zygote does not have exactly 23 chromosomes. These anomalies can cause various disorders, some of which are discussed here.

Genetic Problems

RESPONSE FOR Future Parents

(from page 48) Yes, but you wouldn't want to. You would have to choose one mate for your sons and another for your daughters, and you would have to use sex-selection methods. Even so, it might not work, given all the genes on your genotype. More important, the effort would be unethical, unnatural, and possibly illegal.

2.2.1 Chromosomal Abnormalities

Gametes (sperm or ova) may have more or fewer than 23 chromosomes. This happens for many reasons, both inherited and environmental (such as a parent's exposure to radiation). The variable that most often correlates with chromosomal abnormalities is the age of the mother, presumably because ova become increasingly fragile by midlife. The father's age is also relevant (Crow, 2003).

Most abnormal zygotes formed by such gametes do not duplicate, divide, and differentiate (K. L. Moore & Persaud, 2003) or are spontaneously aborted early in pregnancy. Nonetheless, about once in every 200 births, an infant is born with 45, 47, or even 48 or 49 chromosomes instead of the usual 46.

Each chromosomal abnormality produces a recognizable *syndrome*, a cluster of distinct characteristics that occur together. Typically, the cause is that three chromosomes, instead of the usual two, occupy a particular location—a condition called a *trisomy*. Traditionally, such problems were recognized by physical signs such as malformed eyes, hands, or inner organs, but now chromosomal analysis can detect anomalies that do not affect appearance (Hamerton & Evans, 2005).

DOWN SYNDROME

Most embryos with chromosomal abnormalities are aborted spontaneously. Among those that survive, however, the most notable problem is three copies of chromosome 21, which results in **Down syndrome** (also called *trisomy-21*). Most individuals with Down syndrome have specific facial characteristics (thick tongue, round face, slanted eyes). Many also have hearing losses, heart abnormalities, muscle weakness, and short stature. All are slow to develop language.

Adults with Down syndrome age faster than other adults. By age 40, most have one or more problems of aging, including cataracts, dementia, and certain forms of cancer. Nonetheless, children and young adults with Down syndrome can live for many happy years. One such person advised others with Down syndrome:

> You may have to work hard, but don't ever give up. Always remember that you are important. You are special in your own unique way. And one of the best ways to feel good about yourself is to share yourself with someone else.

[Christi Todd, quoted in Hassold & Patterson, 1999]

Universal Happiness All young children delight in painting brightly colored pictures on a big canvas, but this scene is unusual for two reasons: Daniel has trisomy-21, and this photograph was taken at the only school in Chile where normal and special-needs children share classrooms. REUTERS/CLAUDIA DAUT

ABNORMALITIES OF THE SEX CHROMOSOMES

About 1 in every 500 infants has only one X and no Y chromosome (the X stands alone) or has three or more sex chromosomes, not just a pair. Although the normal 22 pairs of autosomes allow fetal growth and newborn

survival, these abnormalities at the 23rd pair hinder cognitive and psychosocial development as well as sexual maturation.

A newborn with only one X has *Turner syndrome*. As a result, she is unusually short, her female organs are underdeveloped, and she cannot conceive and bear children. Usually she is slow to develop, particularly in spatial understanding.

If there are three, four, or five sex chromosomes instead of two, a child may seem normal until puberty. If he is XXY, he has *Klinefelter syndrome*, whose symptoms include learning problems. Klinefelter syndrome may not be recognized until puberty, when the double X keeps a boy's penis from growing and fat accumulates around his breasts. Sometimes the condition is not recognized until adulthood, when a man discovers that he is infertile.

Many people with sex-chromosome abnormalities have "productive and healthy lives" (Hamerton & Evans, 2005, p. 631). Hormone therapy, special education, and psychological counseling for them and their families can help these people develop normally.

Chromosomal Abnormalities

2.2.2 Gene Disorders

Everyone carries genes or alleles that *could* produce serious diseases or handicaps in the next generation (see **Table 2.1** on pages 52–53). Given that most genetic disorders are polygenic and that the mapping of the human genome is recent, the exact impact of each allele is not yet known (Hinds et al., 2005). However, single-gene disorders have been studied for decades (S. M. Miller et al., 2006).

DOMINANT DISORDERS

Most of the 7,000 *known* single-gene disorders are dominant and easy to identify as such: Half the offspring of parents with a dominant disorder will also have the disorder (in other words, it will be expressed in their phenotype). If the condition is fatal in childhood, it will, of course, never be transmitted. Thus, all the dominant disorders either begin in adulthood or have relatively mild or variable symptoms.

One example of a disorder thought to be dominant is *Tourette syndrome*. Some who inherit the Tourette gene exhibit uncontrollable tics and explosive outbursts of verbal obscenities. But most have milder symptoms, such as an occasional twitch that is barely noticeable or a postponable impulse to speak inappropriately (Olson, 2004).

RECESSIVE DISORDERS

The number of recessive conditions is probably in the millions. Recessive genes are usually carried on the autosomes, which means that men and women are equally likely to carry them and offspring of both sexes are equally likely to inherit them. For example, cystic fibrosis, thalassemia, and sickle-cell anemia are equally devastating in both sexes and are also fairly common; about 1 in 12 North Americans is a carrier for one of them.

Lethal recessive conditions are usually rare. The three conditions just mentioned are exceptions. Carriers of the recessive gene for cystic fibrosis, thalassemia, or sickle-cell anemia are likely to survive and reproduce because the very gene that causes illness when two recessive genes are inherited is protective if a person is only a carrier.

For example, carriers of the sickle-cell trait have some protection against malaria, a deadly killer in central Africa. As a result, over the centuries, African carriers were more likely than noncarriers to survive. Similarly, the single cystic fibrosis gene is more common among people whose ancestors came from northern Europe because carriers of that gene may have been protected against cholera.

Some recessive conditions are sex-linked, usually carried on the X chromosome. Males are more likely to be affected by such conditions because they have no second X with a healthy dominant gene (see **Table 2.2** on page 54).

One X-linked condition, called **fragile X syndrome,** is caused

On the Autism Spectrum Ryan, age 11,

by a single gene that has more than 200 repetitions of a triplet (Plomin et al., 2008). (It is normal to have some repetitions, but not this many.) Fragile X syndrome is the most common form of *inherited* mental retardation (many other forms are not inherited). In addition to having cognitive problems, children with fragile X often have muscle weakness and are shy, with poor social skills (Hagerman, 2002).

prefers his action figures to real people. He has Asperger syndrome, a pervasive developmental disorder that is largely inherited, especially by boys. Ryan's two older brothers have it, too. JOHN AMIS/AP

Table 2.1 Common Genetic Diseases and Conditions

Name	Description	Prognosis	Probable Inheritance	Incidence*	Carrier Detection?†	Prenatal Detection?
Albinism	No melanin; person is very blond and pale	Normal, but must avoid sun damage	Recessive	Rare overall; 1 in 8 Hopi Indians is a carrier	No	No
Alzheimer disease	Loss of memory and increasing mental impairment	Eventual death, often after years of dependency	Early onset—dominant; after age 60—multifactorial	Fewer than 1 in 100 middle-aged adults; perhaps 25 percent of all adults over age 85	Yes, for some genes; ApoE4 allele increases incidence	No
Cancer	Tumors that can spread	With early diagnosis and treatment, most are cured; without them, death usually within 3 years	Multifactorial; almost all cancers have a genetic component	More than half of all people develop some form of cancer; about one-fourth die of it	No	No
Cleft palate, cleft lip	The two sides of the upper lip or palate are not joined	Correctable by surgery	Multifactorial	1 in every 700 births; more common in Asian Americans and American Indians	No	Yes
Club foot	The foot and ankle are twisted	Correctable by surgery	Multifactorial	1 in every 200 births; more common in boys	No	Yes
Cystic fibrosis	Mucous obstructions, especially in lungs and digestive organs	Most live to middle adulthood	Recessive gene, also spontaneous mutations	1 in 3,200; 1 in 25 European Americans is a carrier	Sometimes	Yes, in most cases
Diabetes	Abnormal sugar metabolism because of insufficient insulin	Early onset (type 1) fatal without insulin; for later onset (type 2), variable risks	Multifactorial; for later onset, body weight is significant	Type 1: 1 in 500 births; more common in American Indians and African Americans. Type 2: 1 adult in 6 by age 60	No	No
Deafness (congenital)	Inability to hear from birth on	Cochlear implants and/or sign language contribute to leading a normal life	Multifactorial; some forms are recessive	1 in 1,000 births; more common in people from Middle East	No	No
Hemophilia	Absence of clotting factor in blood	Death from internal bleeding; blood transfusions prevent damage	X-linked recessive; also spontaneous mutations	1 in 10,000 males; royal families of England, Russia, and Germany had it	Yes	Yes
Hydrocephalus	Obstruction causes excess fluid in the brain	Brain damage and death; surgery can make normal life possible	Multifactorial	1 in every 100 births	No	Yes
Muscular dystrophy	Weakening of muscles	Inability to walk, move; wasting away	Recessive, x-linked, or	1 in every 3,500 males develops Duchenne's	Yes, for some forms	Yes, for some
Neural-tube defects (open spine)	Anencephaly (parts of the brain missing) or spina bifida (lower spine not closed)	Anencephalic—severe retardation; spina bifida—poor lower body control	Multifactorial; folic acid deficit and genes	Anencephaly—1 in 1,000 births; spina bifida—3 in 1,000; more common in Welsh and Scots	No	Yes
Phenylketonuria (PKU)	Abnormal digestion of protein	Mental retardation, preventable by diet begun by 10 days after birth	Recessive	1 in 100 European Americans is a carrier, especially Norwegians and Irish	Yes	Yes
Pyloric stenosis	Overgrowth of muscle in intestine	Vomiting, loss of weight, eventual death; correctable by surgery	Multifactorial	1 male in 200, 1 female in 1,000; less common in African Americans	No	No
Rett syndrome	Neurological developmental disorder	Boys die at birth. At 6–18 months, girls lose communication and motor skills	X-linked	1 in 10,000 female births	No	Sometimes
Schizophrenia	Severely distorted thought processes	No cure, drugs, hospitalization, psychotherapy ease symptoms	Multifactorial	1 in 100 people develop it by early adulthood	No	No
Sickle-cell	Abnormal blood	Possible painful	Recessive	1 in 11 African	Yes	Yes

anemia	cells	crisis ; heart and kidney failure; treatable with drugs		Americans and 1 in 20 Latinos are carriers		
Tay-Sachs disease	Enzyme disease	Healthy infant becomes weaker, usually dying by age 5	Recessive	1 in 30 American Jews and 1 in 20 French Canadians are carriers	Yes	Yes
Thalassemia	Abnormal blood cells	Paleness and listlessness, low resistance to infections, slow growth	Usually recessive, occasionally dominant	1 in 10 Americans from southern Europe, northern Africa, or south Asia is a carrier	Yes	Yes
Tourette syndrome	Uncontrollable tics, body jerking, verbal outbursts	Appears at about age 5, worsens and then improves with age	Dominant, but variable penetrance	1 in 250 children	Sometimes	No

*Incidence statistics vary from country to country; those given here are for the United States. All these diseases can occur in any ethnic group. Many affected groups limit transmission through genetic counseling; for example, the incidence of Tay-Sachs disease is declining because many Jewish young adults obtain testing and counseling before marriage.

**"Yes" refers to carrier detection. Family history can also reveal genetic risk.

Sources: Benaceraf, 2007; Briley & Suber, 2001; M. G. Butler & Meaney, 2005; Haydon, 2007; Hemminki et al., 2008; Klug et al., 2006; McKusick, 2007; Mange & Mange, 1998; K. L. Moore & Persaud, 2007; Shahin et al., 2002.

Another sex-linked condition is *hemophilia*, in which the blood does not clot normally. In the past such children often bled to death; today blood transfusions can save their lives.

The most common X-linked condition is *color blindness*, an inability to distinguish certain hues, especially red and green. The reason color blindness is so common is that the condition is not debilitating and thus is often passed down from father to daughter and then from mother to son. All these conditions are much more common in boys than girls, because girls are likely to be protected by their second X.

OBSERVATION QUIZ

Is there any ethnic group that does not have a genetic condition that is more common among its members than among the general population? (see answer, page 54)

◀ Gene Disorders ▶

2.2.3 Advising Prospective Parents

Genetic counseling helps people know how likely they are to conceive a child with a severe genetic or chromosomal condition. Counselors need to be carefully trained, because emotional reactions to such information can lead many clients to misinterpret the words *facts, risk*, and *probability* (O'Doherty, 2006).

ANSWER TO OBSERVATION QUIZ

(from page 52) No. As you see, many major ethnic groups are mentioned in **Table 2.1**. In fact, even much smaller groups whose members tend to marry within the group also have higher rates of particular conditions.

Table 2.2 The 23rd Pair and X-Linked Color Blindness

X indicates an X chromosome with the X-linked gene for color blindness

23rd Pair	Phenotype	Genotype	Next Generation
1. XX	Normal woman	Not a carrier	No color blindness from mother
2. XY	Normal man	Normal X from mother	No color blindness from father
3. XX	Normal woman	Carrier from father	Half her children will inherit her X. The girls with her X will be carriers; the boys with her X will be color-blind.
4. XX	Normal woman	Carrier from mother	Half her children will inherit her X. The girls with her X will be carriers; the boys with her X will be color-blind.
5. XY	Color-blind man	Inherited from mother	All his daughters will have his X. None of his sons will have his X. All his children will have normal vision, unless their mother also had an X for color blindness.
6. XX	Color-blind woman (rare)	Inherited from both parents	Every child will have one X from her. Therefore, every son will be color-blind. Daughters will be only carriers, unless they also inherit an X from the father, as their mother did.

ESPECIALLY FOR Historians

Some genetic diseases may have changed the course of history. For instance, the last czar of Russia had four healthy daughters and one son with hemophilia. Once called the royal disease, hemophilia is X-linked. How could this rare condition have affected the monarchies of Russia, England, Austria, Germany, and Spain? (see response below)

Genetic counselors try to follow two ethical guidelines: (1) Their clients' test results are kept confidential, beyond the reach of insurance companies and public records, and (2) decisions regarding sterilization, adoption, abortion, or carrying a pregnancy to term are made by the clients, not by the counselor.

PRESENTING THE FACTS

Genetic counselors must present facts, not opinions, but facts are not always straightforward. If one parent has a dominant gene for a particular disease or if both members of a couple carry the same recessive gene, odds are that half their children will inherit the dominant gene or that one out of four will have the double recessive. But before conception, those are merely odds. Some, all, or none of their offspring *could* inherit the disease. Each pregnancy is a risk, another roll of the same dice.

To add to the uncertainty, the specific effects of many regulator genes are not yet known, and medical treatments for many conditions are not yet

proven effective (Gustafson, 2006). Prospective parents need to decide when to take the risk, when to wait, when to avoid pregnancy, and when to opt for prenatal testing.

More problems arise because tests during pregnancy often merely reveal that more tests are needed (see **Table 2.3**). For instance, the level of alpha-fetoprotein (AFP, found via a blood test) may be too high (possibly indicating a neural-tube defect) or too low (possibly indicating Down syndrome), but high and low levels are usually false alarms. Amniocentesis and chorionic villi sampling (CVS) are more definitive, but they are more invasive.

Furthermore, amniocentesis may reveal not an extra chromosome with known effects but a small segment that is added to a chromosome—with unknown implications. Modern technology often finds problems that were previously undetectable. Some test results are *false positives*, suggesting an abnormality that is not actually present. Others are *false negatives*, providing unfounded reassurance when a fetus is actually impaired.

Newborns are also tested for many genetic diseases; here, too, false positives cause needless worry. Moreover, accurate results (true positives) may reveal a problem for which no treatment exists, or a marriage may be strained if a spouse is told that he or she is, or is not, a carrier (F. A. Miller et al., 2009). Nonetheless, most health professionals, and many organizations that are concerned with children's health (such as the March of Dimes), advocate the testing of prospective parents and newborns for dozens of conditions (N. S. Green et al., 2006).

"The Hardest Decision I Ever Had to Make" That's how this woman described her decision to terminate her third pregnancy when genetic testing revealed that the fetus had Down syndrome. She soon became pregnant again with a male fetus that had the normal 46 chromosomes, as did her two daughters and her fourth child, not yet born when this photo was taken. Many personal factors influence such decisions. Do you think she and her husband would have made the same choice if they had had no other children? ROBERT SPENCER/THE NEW YORK TIMES

Table 2.3 Methods of Postconception Testing

Method	Description	Risks, Concerns, and Indications
Pre-implantation testing	After in vitro fertilization, one cell is removed from each zygote at the four- or eight-cell stage and analyzed.	Not entirely accurate. Requires surgery, in vitro fertilization, and rapid assessment. This delays implantation and reduces the likelihood of successful birth. It is used only when couples are at high risk of known, testable genetic disorders.
Tests for pregnancy-associated plasma protein (PAPPA) and human chorionic gonadotropin	Blood tests are usually done at about 11 weeks to indicate levels of these substances.	Indicate normal pregnancy, but false-positive or false-negative results sometimes occur.
Alpha-fetoprotein assay	The mother's blood is tested for the level of alpha-fetoprotein (AFP). Now usually done at mid-pregnancy, often combined with other blood tests and repeat sonogram.	Indicates neural-tube defects, multiple embryos (both cause high AFP), or Down syndrome (low AFP). Normal levels change each week; interpretation requires accurate dating of conception.
Sonogram (ultrasound)	High-frequency sound waves are used to produce a "picture" of the fetus as early as 6 weeks. Sonograms are more accurate later in pregnancy to detect less apparent problems, to confirm earlier suspicions, and to anticipate birth complications.	Reveals problems such as a small head or other body malformations, excess fluid accumulating on the brain, Down syndrome (detected by expert looking at neck of fetus), and several diseases (for instance, of the kidneys). Estimates fetal age and reveals multiple fetuses, placental position, and fetal growth, all of which are useful to know in every pregnancy. Sometimes sex is apparent. No known risks, unlike the X-rays that it has replaced.
Chorionic villi sampling (CVS)	A sample of the chorion (part of the placenta) is obtained (via sonogram and syringe) at about 10 weeks and analyzed. Since the cells of the placenta are genetically identical to the cells of the fetus, this can indicate many chromosomal or genetic abnormalities.	Provides the same information as amniocentesis but can be performed earlier. Not 100 percent accurate. Can cause spontaneous abortion (rare).
Amniocentesis	About half an ounce of the fluid inside the placenta is withdrawn (via sonogram and syringe) at about 16 weeks. The cells are cultured and analyzed.	Spontaneous abortion is rare (less than 0.05 percent). Detects chromosomal abnormalities and other genetic and prenatal problems. Is done later in pregnancy than other tests, and it takes a week before results are known.

Sources: Eddleman et al., 2006; Malone et al., 2005; K. L. Moore & Persaud, 2003; Reece & Hobbins, 2007; D. Wright et al., 2005.

PKU: A SUCCESS STORY

One routine test is for **phenylketonuria (PKU),** a recessive condition for which northern Europeans are particularly at risk (Welsh & Pennington, 2000). (The incidence is 26 times higher among the Irish than among the Japanese.) Newborns with the double-recessive PKU gene become severely retarded if they consume phenylalanine, an amino acid found in many foods. Parents of a baby with PKU need to feed their child a special diet.

Thousands of children who would have died in institutions now live fairly normal lives because of early testing and this special diet. A full-grown adult with PKU may eat foods containing phenylalanine; the exception is that a woman with PKU who wishes to become pregnant must go back to the special diet in order not to harm her embryo (Plomin et al., 2008).

The PKU case is straightforward: No professional would suggest skipping PKU testing, nor would any parent potentially cause damage to the baby's brain by refusing testing. The test for PKU is quite accurate, the treatment is fairly simple, and the result is a normal child instead of a severely impaired one.

However, PKU has many variants, some of which cause mild retardation even when the diet is followed. Some other consequences were not anticipated by the scientists who discovered the PKU gene; for example, some couples with a PKU child avoid a second pregnancy, and some children with PKU rebel and eat forbidden food, while others become overly dependent on their parents.

RESPONSE FOR Historians

(from above): Hemophilia is a painful chronic disease that (before blood transfusions became feasible) killed a boy before adulthood. Though rare, it ran in European royal families, whose members often intermarried, which meant that many queens (including England's Queen Victoria) were carriers of hemophilia and thus were destined to watch half their sons die of it. All families, even rulers of nations, are distracted from their work when they have a child with a mysterious and lethal illness. Some historians believe that hemophilia among European royalty was an underlying cause of the Russian Revolution of 1917 as well as of the spread of democracy in the nineteenth and twentieth centuries.

Despite these problems, a cost-benefit analysis makes it obvious that PKU testing is wise. However, the situation is not always so clear-cut for other conditions. Often the diagnosis is less certain and the consequences more variable (Sandel, 2007), as the following explains.

A VIEW FROM SCIENCE

Genetic Testing and Parental Choice

At the moment, genetic testing occurs primarily when there is a known risk of a particular condition that would result in the birth of a severely impaired child, often one who would become less and less capable and die before reaching adulthood. However, as more genetic traits are discovered, prospective parents may seek testing in order to abort those embryos that do not have the exact traits that they prefer.

This practice was common in China and India because a sonogram or amniocentesis can easily discover the sex of a fetus. In some cities in China, selective abortions became an unexpected consequence of a government policy begun in 1979 (Greenhalgh, 2008). A "one child per couple" campaign reduced poverty but led to abortions of female fetuses because many parents wanted their only child to be a boy.

Since 1993, the Chinese government has allowed many families to have a second child and forbidden prenatal testing to determine sex. But that "law has been spottily enforced": In one city with careful records (Guiyang), 75 girls are born for every 100 boys (H. W. French, 2005). Similar imbalances have occurred in other nations.

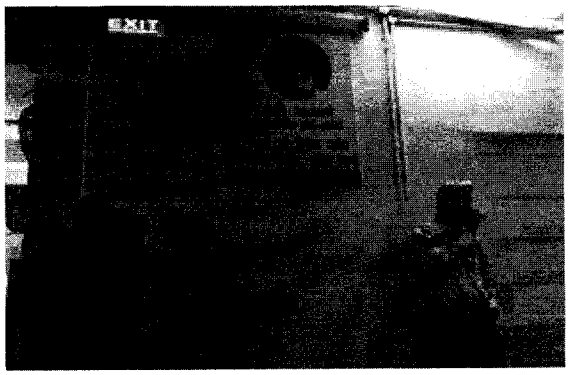
The United Nations opposes prenatal sex selection. Some nations allow it, but others—including China, India, Australia, and Canada—outlaw it except to prevent the birth of a child with severe disabilities (such as hemophilia, which affects only boys).

If you were a prospective parent, would you welcome a baby who had the following genetic tendencies?

- *Diseases:* heart attack, constricted arteries, diabetes, high blood pressure, Alzheimer disease
- *Responses to substances:* general substance abuse, alcoholism, tobacco addiction, lactose intolerance (poor digestion of milk)
- *Personality:* seeks novelty, evening preference, antisocial behavior, conduct disorder
- *Appearance:* male, blue eyes, fair skin, sticky earwax

An embryo with genes for all these traits would become a difficult, curious, and hard-to-control boy, likely to become a risk-taking drug addict and then to develop heart disease. If he survived to old age, he might become demented.

All these traits did, in fact, exist in one person, although when he was in the uterus, genetic testing was not yet available. True to his genetic heritage, he was a troublesome, disobedient boy who became a rebellious adolescent. As a young adult, he was drafted to fight in Vietnam. To avoid front-line combat, he got himself assigned to a field hospital, where he treated many soldiers who later died. He once tried to commit suicide by swimming out into the China Sea, but he changed his mind and swam back to shore. His father had a fatal heart attack at age 59 (J. Cohen, 2007b; Wade, 2007).



"Save the Girl Child" That's the slogan on this poster, which appears on the wall outside a New Delhi hospital. Sex selection, while illegal, is still widespread in India. In 2008, in response to the threat of a lawsuit, Google and Yahoo removed all advertisements for sex-selection products and services from their Indian connections. PEDRO UGARTE/AFP/Getty Images

Eventually, this man's military experiences convinced him that he needed more education. He earned a PhD and began to do research in genetics. He soon antagonized other scientists, who bristled at his enormous ego and his maverick ways. With a combination of brilliance, charm, and perseverance (all partly genetic traits, although the genes that predict them are not yet known), he founded a company called Celera Genomics, which succeeded at deciphering the human genome in a commercial project that ran parallel to the Human Genome Project.

This man's name is Craig Venter. He was one of five people whose DNA was used to generate the complete sequence of the human genome—which is how the information about his genetic tendencies was obtained. The entire world is benefiting from his work. The timing of events in his life helped him: Not only did his service in Vietnam change his direction, but his own genome was unknown before he became famous and successful. If it had been known, his fate might have been quite different.

Thanks partly to Venter, it will soon be possible to decode a person's genome for $1,000 (Winstein, 2007). That possibility raises many ethical questions. Some parents may knowingly risk having children with crippling diseases or severe mental illness. That may be unfair to the children or to the society that pays for their care; but it may also be unfair for anyone to prevent adults from bearing as many children, with or without genetic diseases, as they choose.

No reputable scientist advocates abortion of fetuses with the kinds of traits indicated in Venter's genome. Indeed, pediatricians whose patients suffer from conditions that are thought to be genetic often hesitate to advise the parents to be tested, having in mind the ethical implications for those parents' subsequent pregnancies (Pagon & Trotter, 2007).

Experts are well aware of these and other moral risks of genetic testing (Ekberg, 2007). Many doctors in clinics that offer pre-implantation testing would like more guidance about when not to test (Baruch et al., 2008). Now is the time for these ethical questions to be debated, before all the possibilities become realities.

KEY Points

- Everyone carries some genetic problems. The impact and inheritance patterns are variable: Some are dominant, some recessive, some additive.
- Chromosomal abnormalities include Down syndrome (trisomy-21) and an unusual number of sex chromosomes. Some people with such problems develop well.
- Gene disorders may be either dominant (e.g., probably Tourette syndrome) or recessive (e.g., cystic fibrosis, thalassemia, sickle-cell anemia). Some recessive gene disorders are sex-linked (e.g., fragile X, hemophilia, color blindness).
- Genetic testing can determine the risk of conceiving a child with many serious diseases and conditions. Decisions are made by parents, not professionals.

◀ Advising Prospective Parents ▶

FOR DEEPER REFLECTION

Why might someone want to know a baby's sex before birth?

2.3 From Zygote to Newborn

The beginning of human life is astonishing. A tiny cell, smaller than the period at the end of this sentence, turns into a person in a mere nine months. In less than a year, the most extensive transformation of the entire life span occurs.

To make it easier to study, prenatal development is often divided into three main periods. The first two weeks are called the **germinal period**; the third through the eighth week is called the **embryonic period**; the longest stretch, from the ninth week until birth, is called the **fetal period.** (For alternative terms, see **Table 2.4**.)

Table 2.4 Timing and Terminology

Popular and professional books use various phrases to segment pregnancy. The following comments may help to clarify the phrases used.

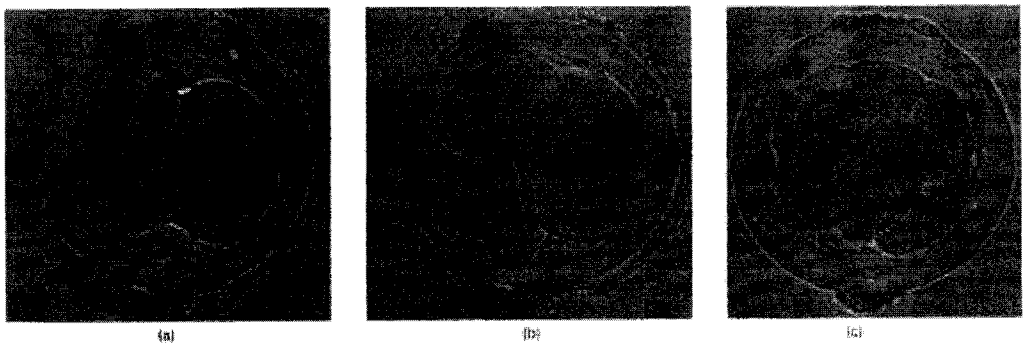
- *Beginning of pregnancy:* Pregnancy begins at conception, which is also the starting point of *gestational age.* However, the organism does not become an embryo until about two weeks later, and pregnancy does not affect the woman (and cannot be confirmed by blood or urine testing) until implantation. Paradoxically, many obstetricians date the onset of pregnancy from the date of the woman's last menstrual period (LMP), about 14 days *before* conception.
- *Length of pregnancy:* Full-term pregnancies last 266 days, or 38 weeks, or 9 months. If the LMP is used as the starting time, pregnancy lasts 40 weeks, sometimes expressed as 10 lunar months. (A lunar month is 28 days long.)
- *Trimesters:* Instead of *germinal period, embryonic period,* and *fetal period,* some writers divide pregnancy into three-month periods called *trimesters.* Months 1, 2, and 3 are called the *first trimester;* months 4, 5, and 6, the *second trimester;* and months 7, 8, and 9, the *third trimester.*
- *Due date:* Although doctors assign a specific due date (based on the woman's LMP), only 5 percent of babies are born on that exact date. Babies born between three weeks before and two weeks after that date are considered "full term" or "on time." Babies born earlier are called *preterm;* babies born later are called *post-term.* The words *preterm* and *post-term* are more accurate than *premature* and *postmature.*

From Zygote to Newborn

2.3.1 The Germinal Period: The First Two Weeks

Within hours after conception, the zygote begins *duplication and division*. First, the original 23 pairs of chromosomes duplicate and the single cell divides into two cells, each of which contains a complete set of chromosomes. Those two cells soon duplicate and divide, becoming four identical cells. Those four duplicate and divide, becoming eight, and so on.

In the process of becoming a baby, the one-celled zygote multiplies into about 10 trillion cells. By adulthood, the number of cells in a human body increases to more than 100 trillion. Almost every one of those cells carries an exact copy of the complete genetic instructions inherited by the zygote. This explains why DNA testing of any cell, even from a drop of blood or a snip of hair, can identify "the real father," or "the guilty criminal," or "the long-lost brother."



First Stages of the Germinal Period The original zygote as it divides into (a) two cells, (b) four cells, and (c) eight cells. Occasionally at this early stage, the cells separate completely, forming the beginning of monozygotic twins, quadruplets, or octuplets. ALL:PETIT FORMAT/NESTLE/SCIENCE SOURCE/PHOTO RESEARCHERS, INC.

Until about the eight-cell stage, the organism consists of *stem cells*, which could develop into any body part. But then a third process, *differentiation*, is added to duplication and division. Following genetic instructions, cells begin to specialize, taking different forms and reproducing at various rates, depending on where they are located. As one expert explains the impact of specialization, "We are sitting with parts of our body that could have been used for thinking" (G. Gottlieb, 1992/2002, p. 172).

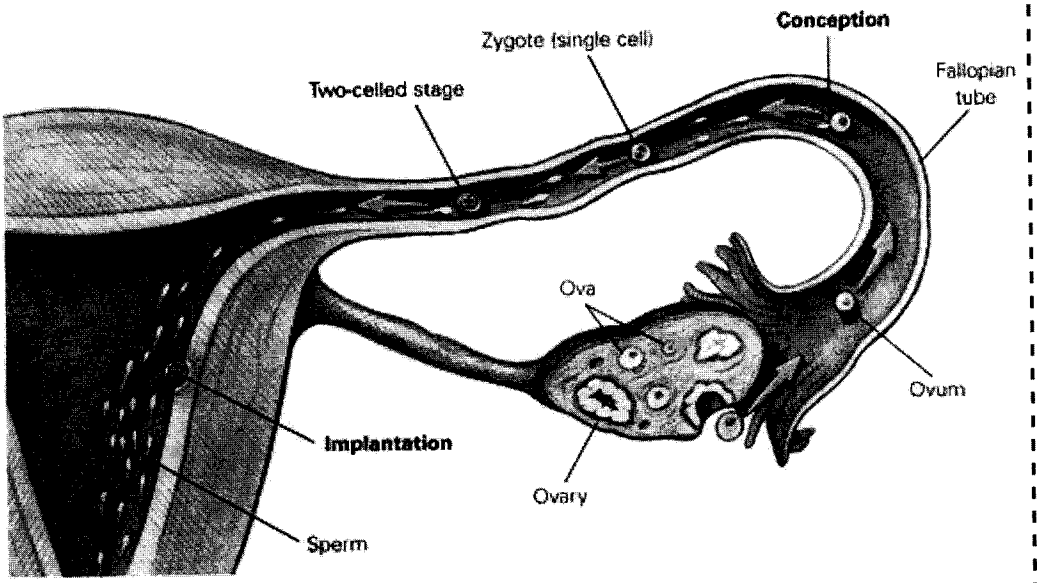


Figure 2.3 The Most Dangerous Journey In the first 10 days after conception, the organism does not increase in size because it is not yet nourished by the mother. However, the number of cells increases rapidly as the organism prepares for implantation, which occurs successfully about less than half the time.

One consequence of differentiation is that cells on the outer side of the developing mass become the **placenta,** the organ that will support the developing life. **Implantation,** the process by which the developing placenta connects to the nurturing environment of the uterus (see **Figure 2.3**), usually begins about 10 days after conception and takes about a week (K. L. Moore & Persaud, 2003).

About 60 percent of all conceptions *in vivo* (literally, "in life," inside the woman's body) and 70 percent of all conceptions *in vitro* (literally, "in glass," outside the body) fail to implant (see **Table 2.5**). Typically, a woman is unaware of a failed implantation; her period might merely arrive a few days later than usual.

Table 2.5 Vulnerability During Prenatal Development

The Germinal Period	An estimated 60 percent of all developing organisms fail to grow or implant properly and thus do not survive the germinal period. Most of these organisms are grossly abnormal.
The Embryonic Period	About 20 percent of all embryos are aborted spontaneously, most often because of chromosomal abnormalities.
The Fetal Period	About 5 percent of all fetuses are aborted spontaneously before viability at 22 weeks or are stillborn, defined as born dead after 22 weeks.
Birth	About 31 percent of all zygotes grow and survive to become living newborn babies.

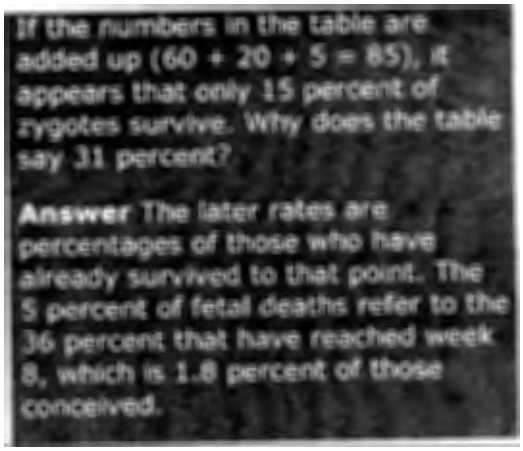
Sources: Bentley & Mascie-Taylor, 2000; K. L. Moore & Persaud, 2003.

UNDERSTANDING THE NUMBERS

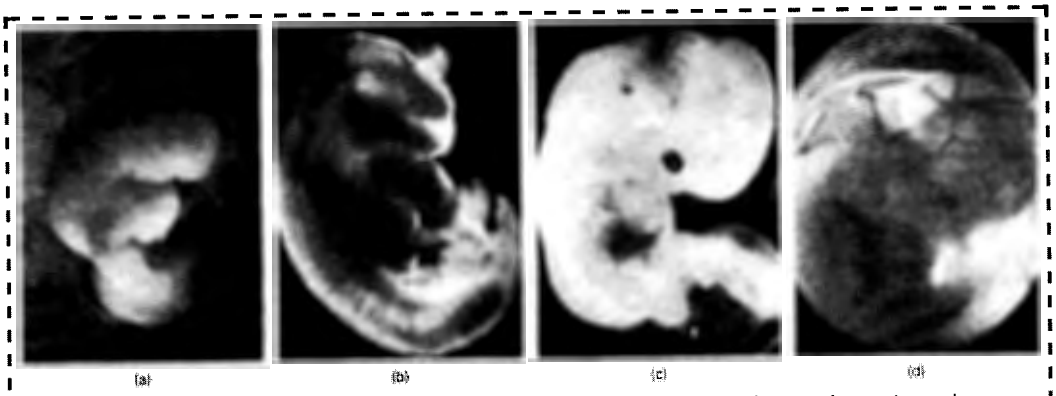
If implantation is successful, the organism begins to grow in size because the placenta is connected to the umbilical cord, which

brings nourishment and carries away waste. The first noticeable signs of pregnancy occur after implantation, when substances begin to enter a pregnant woman's body via the placenta, affecting her hormones and digestion. Pregnancy tests at that point (but not earlier) may detect new chemicals in her urine. After implantation, the woman is literally eating and urinating for two.

The Germinal Period: The First Two Weeks...



2.3.2 The Embryonic Period: From Two Through Eight Weeks



The Embryonic Period (a) At 4 weeks after conception, the embryo is only about ½ inch (3 millimeters) long, but already the head (top right) has taken shape. (b) At 5 weeks after conception, the embryo has grown to twice the size it was at 4 weeks. Its primitive heart, which has been pulsing for a week now, is visible, as is what appears to be a primitive tail, which will soon be enclosed by skin and protective tissue at the tip of the backbone (the coccyx). (c) By 7 weeks, the organism is somewhat less than an inch (2½ centimeters) long. Eyes, nose, the digestive system, and even the first stage of leg formation can be seen. (d) At 8 weeks, the 1-inch-long organism is clearly recognizable as a human fetus.[A, B] PETIT FORMAT/NESTLE/SCIENCE SOURCE/PHOTO RESEARCHERS, INC.[C] Carolina Biological Supply Company/Phototake USA, Inc.[D] LookatSciences/Phototake USA, Inc.

About two weeks after conception, the inner cells of the developing organism (now surrounded by the placenta) show the first sign of body shape and structure when a thin line called the *primitive streak* appears down the middle of the cell mass. That event occasions a new name for the developing organism: **embryo.** The primitive streak becomes the *neural tube* 22 days after conception, eventually becoming the brain and spine of the central nervous system (K. L. Moore & Persaud, 2003).

The head begins to take shape in the fourth week, as eyes, ears, nose, and mouth form. Also in the fourth week, a minuscule blood vessel that will become the heart begins to pulsate, making the cardiovascular system the first part of the embryo to show any activity. At that point, the embryo appears to have a tail because the spine is not yet enclosed.

Soon the body systems and parts grow, according to the schedule set by the genes. For example, by the fifth week, buds that will become arms and legs emerge. Upper arms and then forearms form, followed by hands, then palms, and finally webbed fingers. Legs, feet, and webbed toes, in that order, emerge a few days later, each with the beginning of a skeletal structure. At the end of the embryonic period, the fingers and toes separate (52 and 54 days after conception, respectively). At this point, the entire sexual-reproductive system is similar

in both sexes, preparing to produce gametes and developing the body structures necessary to eliminate waste.

The Embryonic Period: From Two Through E...

The Fetus At the end of 4 months, the fetus, now 6 inches long, looks fully formed but out of proportion—the distance from the top of the skull to the neck is almost as long as that from the neck to the rump. For many more weeks, the fetus must depend on the translucent membranes of the placenta and umbilical cord (the long white object in the foreground) for survival. S. J. ALLEN/INTERNATIONAL STOCK PHOTO

2.4 Birth

For a full-term fetus and a healthy mother, birth can be simple and quick. At some time during the last month of pregnancy, most fetuses change position, turning upside down so that the head is low in the mother's pelvic cavity. They are now in position to be born in the usual way, head first.

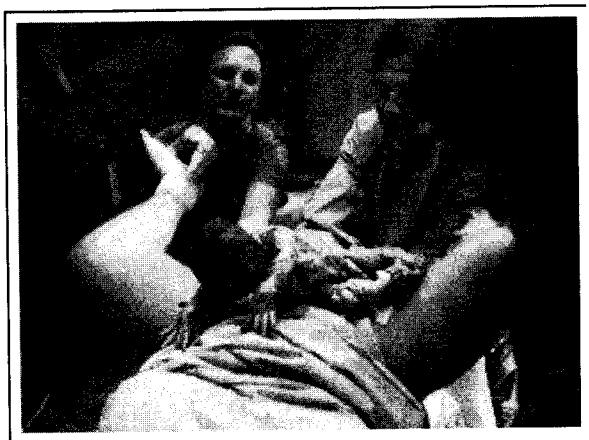
Birth

2.4.1 The Process of Birth

At about 38 weeks after conception, the fetal brain signals the release of hormones to trigger the woman's uterine muscles to begin contracting and relaxing. This is the start of *labor*, the birth process. A first baby is born, on average, 12 hours later (K. L. Moore & Persaud, 2003), although it is not unusual for labor to take twice, or half, that long. Labor is usually quicker for later-born babies, especially if the mother is still relatively young (under age 30).

The same hormones that trigger labor also protect the fetal brain (Tyzio et al., 2006). The typical head-first delivery enables newborns to breathe before they have fully emerged. (Some babies end up in the *breech* position, with the buttocks or, rarely, the feet first. To prevent anoxia [oxygen deprivation], breech babies are often delivered via cesarean section.)

The first breaths of oxygen cause the infant's color to change from bluish to pinkish. ("Bluish" and "pinkish" refer to the blood color, visible beneath the skin, and apply to newborns of all skin colors.) The eyes open wide; the tiny fingers grab; the tinier toes stretch and retract. The newcomer is instantly, zestfully ready for life.



No Doctor Needed In this Colorado Springs birthing center, most babies are delivered with the help of nursemidwives. This newborn's bloody appearance and bluish fingers are completely normal; an Apgar test at five minutes revealed that the baby's heart was beating steadily and that the body was "entirely pink." SEAN CAYTON/THE IMAGE WORKS

Although many newborns have misshapen heads, body bruises, and splotchy skin, appearance is not a reliable indicator of health. Beyond a birthweight of at least 5½ pounds (2,500 grams), the quickest measure of newborn vitality is the **Apgar scale** (see **Table 2.6**). The examiner checks five vital signs—heart rate, breathing, muscle tone, color, and reflexes—at one minute and again at five minutes after birth, assigning each a score of 0, 1, or 2 (Moster et al., 2001).

RESPONSE FOR Biologists

(from page 62) Only one of the 46 human chromosomes determines sex, and the genitals develop last in the prenatal sequence. Sex differences are apparent before birth, but they are relatively minor.

Oxygen level affects color, and at one minute, many healthy newborns are bluish, scoring a 0 or 1. The oxygen level quickly rises (Kamlin et al., 2006). If the five-minute Apgar total is 7 or above, all is well.

Table 2.6 Criteria and Scoring of the Apgar Scale

	Five Vital Signs				
Score	Color	Heartbeat	Reflex Irritability	Muscle Tone	Respiratory Effort
0	Blue, pale	Absent	No response	Flaccid, limp	Absent
1	Body pink, extremities blue	Slow (below 100)	Grimace	Weak, inactive	Irregular, slow
2	Entirely pink	Rapid (over 100)	Coughing, sneezing, crying	Strong, active	Good; baby is crying

Source: Apgar, 1953.

The Process of Birth

Printed Page 64

[Notes/Highlighting]

2.4.2 Traditional and Modern Birthing Practices

As modern medicine is introduced in the developing areas of the world, a conflict may arise between traditional home births attended by a midwife and hospital births attended by an obstetrician. Home births risk complications, and hospital births risk too much intervention. All too often, women must choose one or the other, rather than combining the best features of each. It need not be so, as in this example from the Inuit people of northern Canada:

> Until thirty or forty years ago every woman, and most men, learned midwifery skills and knew what to do to help at a birth if they were needed.... Since the 1950s, as the medical system took control in the belief that hospital birth was safer, more and more pregnant women were evacuated by air to deliver in large hospitals in Winnipeg and other cities....
>
> Around three weeks before her due date, a woman is flown south to wait in bed and breakfast accommodation for labor to start, and to have it induced if the baby does not arrive when expected. Anxious about their children left at home, mothers become bored and depressed.... Women...deliver in a supine position [on their back] instead of an upright one, which was part of their tradition, and also describe being tied up while giving birth. Many women say that children who have been born in a hospital are different and no longer fit into the Inuit lifestyle....
>
> Several new birth centres have now been created [in the Inuit homeland] and nurse-midwives are bringing in traditional midwives as assistants during childbirth, training some Inuit midwives to work alongside them, and at the same time learning some of the old Inuit ways themselves.
>
> *[Kitzinger, 2001, pp. 160–161]*

OBSERVATION QUIZ

Is the woman at left in the left-hand photo the pregnant woman's doctor? (see answer, page 66)

The Same Situation, Many Miles Apart: Preparing for Birth Both of these pregnant women are carrying twins, but their prognoses are quite different. The American woman in Lamaze class (left) is practicing breathing during labor. The pregnant woman in Afghanistan (right) and her doctors discuss why labor will be induced: One of her twins is not developing normally. Neither of her babies is expected to live. Virtually all newborns in developed nations survive; the Afghan woman has already lost two children at birth. BOB LARSON/CONTRA COSTA

Another example of a traditional custom incorporated into a modern birth is the **doula.** Long a fixture in many Latin American countries, a doula is a woman who helps with labor, delivery, breast-feeding, and newborn care. Traditionally, doulas were the only attendants at birth, but today they often work alongside the medical staff in hospitals.

In North as well as South America, many prospective parents find a doula to help at birth. Recently, a study enlisted middle-class couples in which the husbands planned to be with their wives during labor. Half were randomly assigned to be accompanied by a doula, while the other half had only the usual nurses and doctors. Not only were all the husbands and wives happy to have

doulas with them, but the births were easier, as evidenced by a significantly lower rate of cesarean sections (McGrath & Kennell, 2008).

TIMES/MCT/NEWSCOMAP PHOTO/LYNNE SLADKY

Every culture has some birthing practices that are beneficial and some that are destructive. From a developmental perspective, combining traditional and modern birthing practices is likely to be an improvement over wholesale adoption or rejection of either.

Traditional and Modern Birthing Practice...

2.4.3 Not Waiting for Nature

Under some circumstances, birth may occur before spontaneous labor begins. Prenatal checkups sometimes discover something seriously amiss, such as high blood pressure in the mother, slow fetal growth, or twins that are destroying each other (called twin transfusion syndrome). Sometimes medical professionals deliver babies by inducing labor (starting it before hormones do so naturally) or by performing surgery.

CESAREAN SECTION

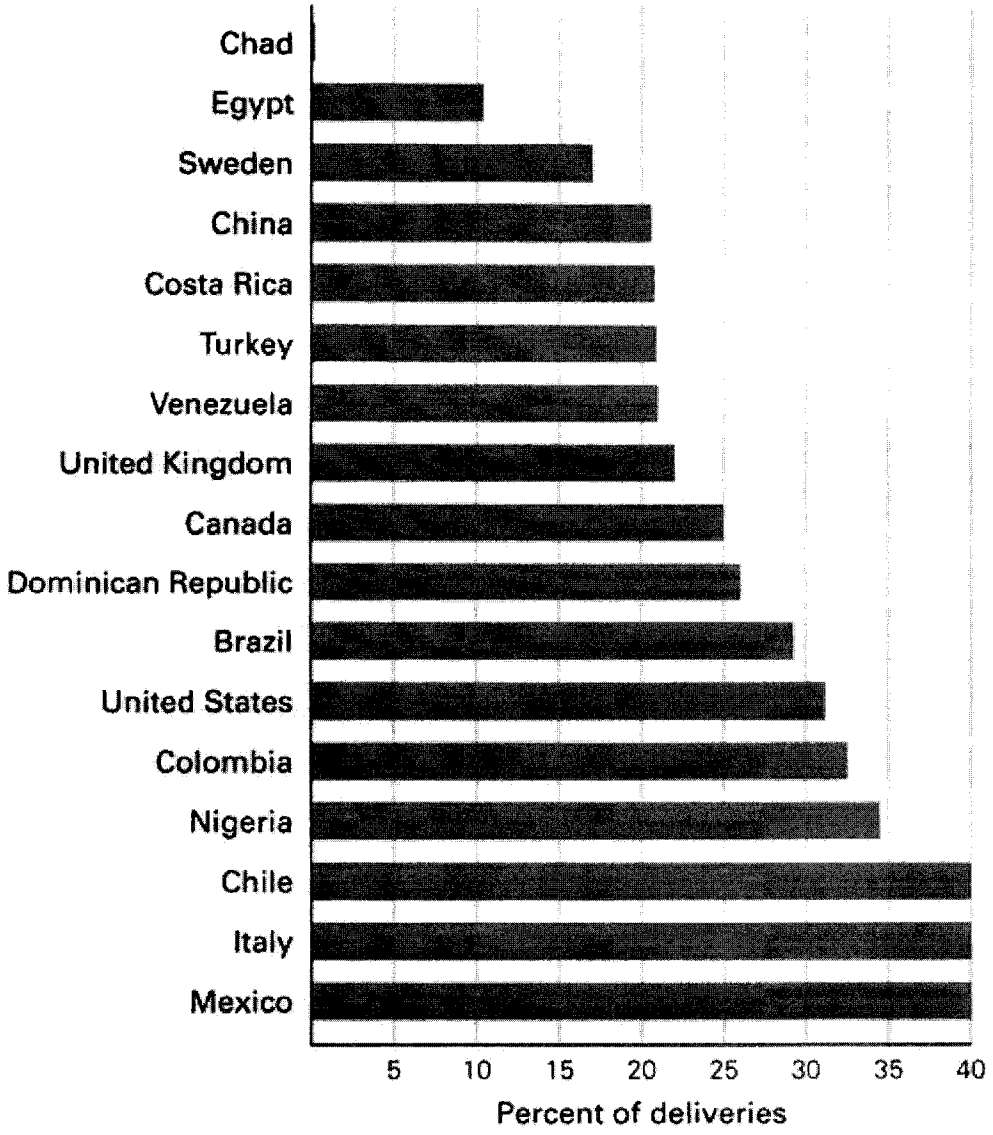
In a **cesarean section (c-section)**, the fetus is removed through surgery (B. Hamilton et al., 2004). Such births may occur if labor has begun but does not progress. More often, a cesarean is scheduled before the first contraction.

The rate of surgical birth varies markedly from place to place (see **Figure 2.5**). In the poorest nations, cesareans are performed only in an emergency. In richer nations, possible problems are anticipated and cesarean sections are planned. Some c-sections are performed for the convenience of the mother or the obstetrician—a practice widely regarded as dangerous (Jukelevics, 2008). About one-third of births in the United States are via c-section, a rate that has been rising for decades.

A c-section means less trauma for the newborn, who can immediately receive advanced care. However, surgery and anesthesia slow down the mother's recuperation, and this delay may impair her ability to breast-feed and care for the baby. Another possible drawback is that once a woman has had a cesarean, subsequent deliveries may also be by cesarean; this idea is controversial (Landon et al., 2004).

From Day One For various reasons, some countries have much higher rates of cesarean deliveries than others. These new mothers in Brazil, which has a high cesarean rate, have safely delivered their babies and, with the encouragement of the hospital, are breast-feeding them from the very beginning. DOUGLAS ENGLE/MCT

Rates of Cesarean Sections in Selected Countries



Sources: Organisation for Economic Cooperation and Development (OECD health data 2008, available at http://loweroecd.org/health/health data; retrieved March 2, 2009); Armson, 2007; Belizán et al., 1999; C. Black et al., 2005; Khawaja et al., 2004; C.K. Stanton & Holtz, 2006; Guo et al., 2007; C.-H. Tang et al., 2006.

Figure 2.5 Too Many Cesareans or Too Few? Rates of cesarean deliveries vary widely from nation to nation. In general, cesarean births are declining in North America and increasing in Africa. Latin America has the highest rates in the world (note that 40 percent of all births in Chile and Mexico are by cesarean), and sub-Saharan Africa has the lowest (the rate in Chad is less than half of 1 percent). The underlying issue is whether some women who should have cesareans do not get them, while other women have unnecessary cesareans.

MEDICAL INTERVENTION

A similar dilemma arises with every type of intervention. For instance, microsurgery in the days before and after birth on tiny, malformed hearts, lungs, and digestive systems prevents death. Eighty years ago, my nephew David (whose story is told in

FOR DEEPER REFLECTION

Do people's attitudes about medical intervention at birth reflect their attitudes about medicine at other points in the life span, in such areas

Chapter 1) would have died, as did 5 percent of all babies born in the United States in 1900 (De Lee, 1938). Now fewer than 1 newborn in 200 dies.

as assisted reproductive technology (ART), immunization, and life support?

Every year, obstetricians, midwives, and nurses save millions of lives. A lack of medical attention during birth and during illegal abortions is the major reason motherhood is still hazardous in the least developed nations; about 1 in 20 women in the poorest nations die from complications of pregnancy (Daulaire et al., 2002).

However, while modern medical care during childbirth has undeniable benefits, it also puts healthy women in hospitals, with attendants, equipment (such as an IV), birth positions, and surgery that may not be in their best interest (Jukelevics, 2008). For example, in Pelotas, Brazil, most births are by cesarean (82 percent for private patients in 2004); furthermore, the rate of low-birthweight infants in Pelotas is rising (from 11 to 16 percent in 10 years), because many infants are born before they are ready (Barros et al., 2005).

ANSWER TO OBSERVATION QUIZ

(from page 64) No; she is the pregnant woman's mother. Doctors are unlikely to attend their patients' Lamaze classes, but every pregnant woman in the class is supposed to have a helper—usually the baby's father, but sometimes a close relative or friend.

As an example of the alternative to hospital deliveries, consider this account of a home birth in Ghana:

> Huddled in a corner of the hut, she was lying on the floor.... She lay curled into a small ball on her left side, her pregnant and contracting uterus protruding from her thin frame. No sound came from her. No sound came from the midwife either. She was seated in the corner of the dark, hot hut, waiting. Suddenly, Emefa gave a low whimper and hauled herself into a sitting and then squatting position. The midwife crept over to her and gently supported Emefa's back as she bore down. No words, no commands, no yelling....The baby's head appeared gradually, slowly making its progress into the world. How did the midwife know that it was time?...A soft whoosh and the baby's body was born into the steady and confident hands of the midwife. And still there was no sound. The baby did not cry, not because there was any problem, but because it was a gentle birth. The baby was breathing as he was handed to his mother.
>
> *[Hillier, 2003, p. 3]*

Thinking About Her Baby This woman may be more fortunate than others in Ghana, which has a high death rate among mothers and babies from birth complications. The wooden structure behind her is a field hospital, so she will receive medical care when she gives birth. PHOTO BY DWIGHT CENDROWSKI

The idea of a "gentle birth" is appealing, but this newborn may have been lucky. The infant mortality rate in Ghana is 10 times that of the United States, where 99 percent of births take place in hospitals or birth centers, with emergency help at the ready. What is needed is a balance between nature and intervention. One sign of balance will be a decrease in the number of low-birthweight newborns.

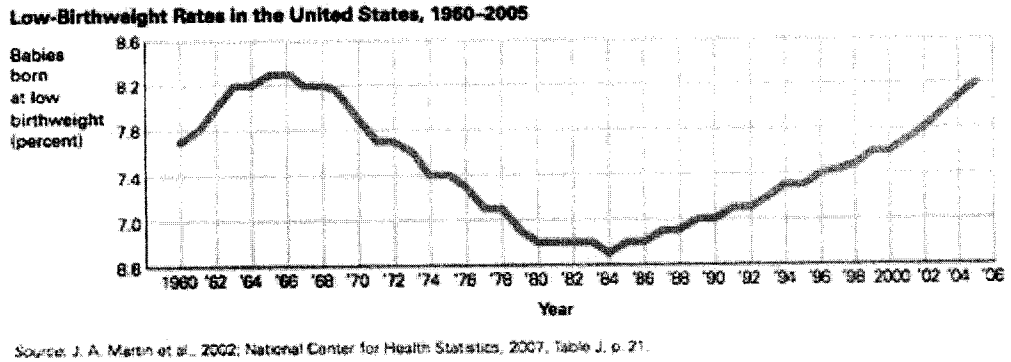
Not Waiting for Nature

2.4.4 Low Birthweight

Low birthweight (LBW) is defined internationally as a body weight of less than 2,500 grams (5½ pounds) at birth. In the United States, low birthweight has slowly increased over the past 25 years (see **Figure 2.6**) and is now the leading cause of infant mortality. About 8 percent of all newborns in the United States are seriously underweight—not the world's highest rate, but far from the lowest (see **Figure 2.7**).

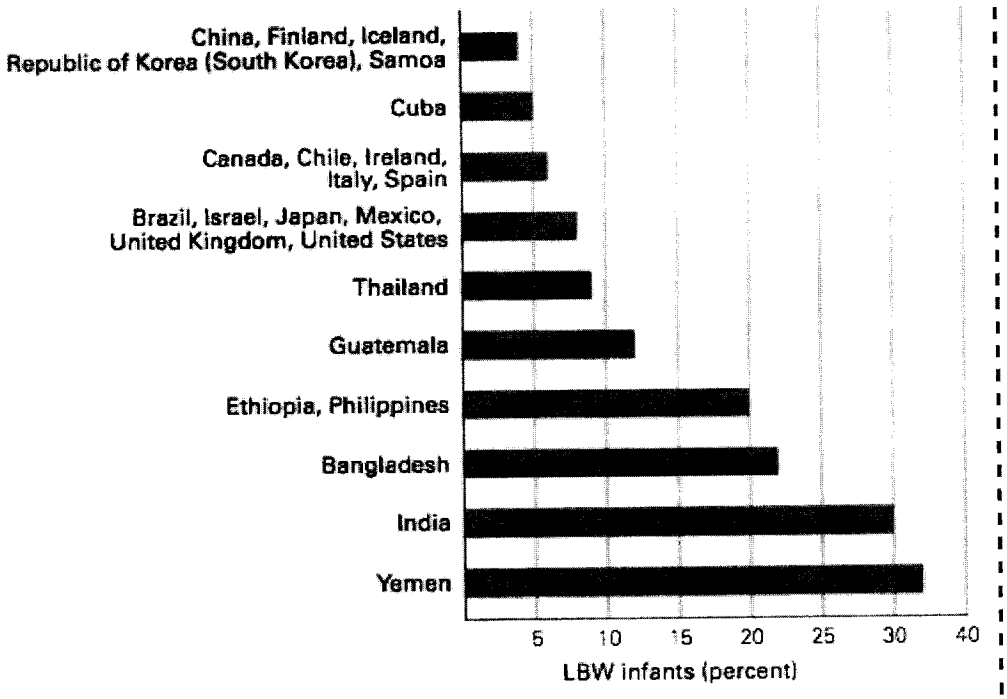
Low birthweight increases the danger of harm from prenatal teratogens (discussed later), adds to birth risks, and makes survival less certain. Every extra gram of weight at birth is protective (unless birthweight is over 4,000 grams, almost 10 pounds).

The LBW babies at greatest risk are the smallest. To focus on them, some underweight newborns are classified as **very low birthweight (VLBW)**, under 1,500 grams (3 pounds, 5 ounces), or **extremely low birthweight (ELBW)**, under 1,000 grams (2 pounds, 3 ounces).



Source: J. A. Martin et al., 2002; National Center for Health Statistics, 2007, Table J, p. 21.

Figure 2.6 What Is Changing? Several changes have occurred in the United States over the past 25 years that should reduce the rate of low birthweight, including fewer teen parents, more Mexican immigrants, and better prenatal care. However, the rate has increased instead, so other factors must be at work. Possible causes are drug use, more multiple births, and environmental pollution.



*Data are for the most recent year available during this period.
Source: United Nations Children's Fund (UNICEF), 2007; United Nations Development Programme, 2008, Table 7, pp. 251–254.

Figure 2.7 Some Better, Some Worse Some of these nations (e.g., Mexico, Bangladesh) have far fewer low-birthweight newborns than they did 25 years ago, and some have more (e.g., United States). The changes as well as the range indicate that birthweight is affected more by nurture than by nature. Very few newborns weigh less than 5½ pounds (2,500 grams) because most are genetically destined to have a healthy weight.

PRETERM OR SLOW-GROWING?

There are two reasons a newborn might be too small. If birth occurs early, before 35 weeks of gestation instead of at the usual 38, the baby is **preterm** and usually (though not always) weighs less than 2,500 grams (5½ pounds). Babies born before 32 weeks are always low birthweight.

The second reason for low birthweight is slow gain throughout pregnancy. Such infants are described as *small-for-dates*, or **small for gestational age (SGA)**, meaning that the baby's birthweight is significantly lower than expected, given the time since conception. SGA suggests impairment throughout prenatal development—a sign that something is seriously amiss.

CAUSES OF LOW BIRTHWEIGHT

Behind these two reasons for low birthweight are a wide variety of specific causes. Some are genetic; for example, small women tend to have small babies. However, nurture, not nature, is responsible for almost all cases of low birthweight.

Maternal illness, exhaustion, infection, and malnutrition all cause low birthweight. The importance of nutrition is starkly evident in data from

Gambia, one of the poorest nations in Africa. Preterm births are highest (17 percent) in July, when women usually work long hours in the fields. However, SGA births are more common (31 percent) in November, the end of the "hungry season," when most women have been undernourished for three months or more (Rayco-Solon et al., 2005). November newborns are most likely to die.

Severe malnutrition during pregnancy is rare in developed nations, where prenatal care is readily available; weighing the pregnant woman is part of every prenatal visit, and underweight women are encouraged to eat more. This advice is particularly important if the woman is under age 16 and still growing herself. Note, however, that the U.S. teen pregnancy rate has been falling while low birthweight has been increasing; young pregnancy is not the most common cause.

Instead, drug use may be to blame for most low-birthweight newborns in the United States. Fetal growth is slowed by every psychoactive drug, including tobacco, which is the most prevalent.

Another common reason for LBW is multiple births, which now often result from assisted reproduction (discussed in **Chapter 12**). Many doctors note that singletons are healthier; still, many infertile couples welcome multiple births.

Low Birthweight

2.4.5 Mothers, Fathers, and a Good Start

Fathers, other relatives, neighbors, and cultures can help to reduce the incidence of birth complications. Consider statistics about low birthweight in the United States. For mothers born in Mexico, the rate of LBW is only 5 percent, compared with 7 percent for mothers of Mexican or European heritage who were born in the United States (a significant difference, since the statistics are based on the total number of recorded births).

Why does a low-income group have fewer LBW infants than a high-income group? The probable reason is that the partners and families of pregnant Mexican-born women encourage them to eat well and avoid drugs (Lara et al., 2005).

THE FATHER'S ROLE

As is shown by Mexican American husbands who safeguard their wives' pregnancies, a supportive father-to-be helps a mother-to-be stay healthy, well nourished, and drug-free. For example, a woman's education level and employment status do not correlate with decreased alcohol consumption during pregnancy, but her marital status does (MMWR, April 5, 2002).

Not only through the example of their behavior but also more directly, fathers and other family members can decrease or increase a mother's stress, which in turn affects her circulation, diet, rest, digestion, and, ultimately, the fetus. One study in northern India found that 18 percent of fathers abused their wives during pregnancy; the result was a doubling of the rate of fragile newborns and infant death (Ahmed et al., 2006).

OBSERVATION QUIZ

Why is a green sheet draped in front of this new mother's face? (see answer, page 71)

Another way to interpret this research is that, even in a poor nation, 82 percent of fathers took good care of their wives. In fact, worldwide, many men are solicitous and helpful to their pregnant wives, who might be demanding or tearful.

The need for social support is mutual. Fathers need reassurance, just as mothers do. Levels of cortisol, a stress hormone, correlate between expectant fathers and mothers: When one parent is stressed, often the other is, too (S. J. Berg & Wynne-Edwards, 2002). Pregnant women should encourage fathers to feel fetal movement, listen to the heartbeat, participate in labor, hold the newborn. A close father–infant connection helps every member of the family.

His Baby, Too This new father's evident joy in his baby illustrates a truism that developmental research has only recently reflected: Fathers contribute much more than just half their child's genes. BUBBLES PHOTOLIBRARY/ALAMY

THE IMPORTANCE OF CLOSE CONTACT

Birth complications can have a lingering impact. Low-birthweight babies, for example, are more likely to become adults who are overweight, diabetic, and have disorders of the heart (Hack et al., 2002). Infants in intensive care have higher levels of stress hormones, and their parents treat them differently than they treat newborns who go home soon after birth, ready to eat, sleep, and react without any special equipment.

Once developmentalists became aware of these differences, policies changed. If a newborn must stay in the hospital for weeks after birth, both mother and father are encouraged to help with early caregiving. Doing so benefits both the baby and the parents, who are understandably deprived, depressed, stressed, or angry (Eriksson & Pehrsson, 2005).

One technique for developing close parental involvement is **kangaroo care,** in which the mother of a low-birthweight infant holds her newborn between her breasts, skin-to-skin, allowing the tiny baby to hear her heartbeat and feel her body heat. Fathers can also benefit their newborn and themselves by cradling the baby next to their chests (Magill-Evans et al., 2006).

A Beneficial Beginning These new mothers in a maternity ward in Manila are providing their babies with kangaroo care. ALEX BALUYUT/ONASIA.COM

A comparison study (Feldman et al., 2002) found that newborns given kangaroo care slept more deeply and spent more time alert than did those in standard care. At 6 months, infants who had received kangaroo care were more responsive. Other research confirms the benefits of kangaroo care (Ludington-Hoe et al., 2006; Tallandini & Scalembra, 2006).

Another way parents can help their premature newborn is by regularly performing massage therapy, gently stroking the baby several times a day. Tiny babies who are caressed seem less irritable and more relaxed (Hernandez-Reif et al., 2007). Such soothing massage of tiny babies is routine in India; it is a radical innovation in the United States.

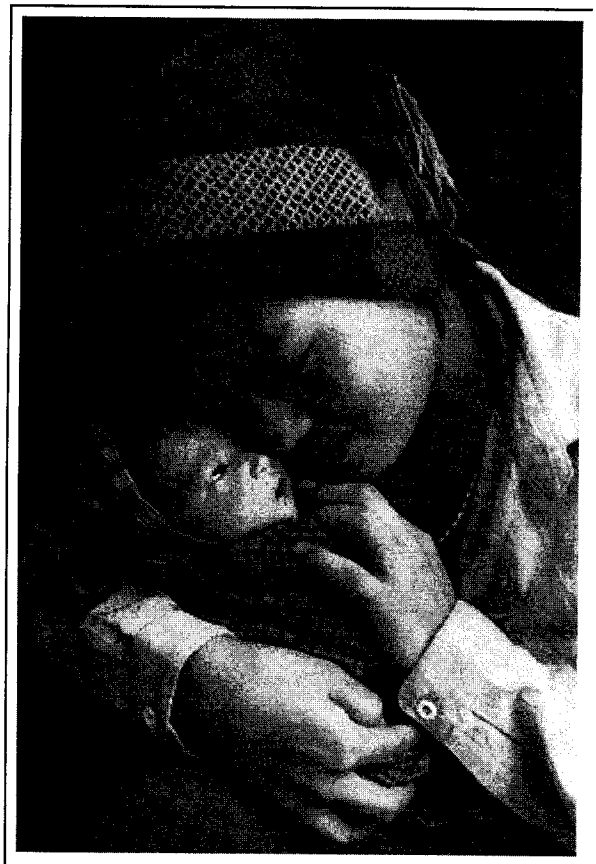
POSTPARTUM DEPRESSION

In the days and weeks after birth, between 8 and 15 percent of mothers experience **postpartum depression,** a sense of inadequacy and sadness (called *baby blues* in the mild version and *postpartum psychosis* in the most severe form) (Perfetti et al., 2004). These rates are for the United States; other nations may have higher rates. For example, a standard postpartum scale found rates of 36 percent in Pakistan (Husain et al., 2006).

Depressed mothers find baby care—feeding, diapering, bathing—burdensome. They may think about mistreating the infant. Postpartum depression can have a long-term impact on the child; it should be promptly treated.

When a mother experiences postpartum depression, the father plays a particularly crucial role. His active caregiving allows the baby to thrive and the mother to recover. Unfortunately, some fathers become depressed themselves. One study found that, even when the mothers were not depressed, if fathers were depressed after birth, their sons had notable behavior problems as toddlers (Ramchandani et al., 2005).

From a developmental perspective, some causes of depression predate the pregnancy; others occur during pregnancy; and still others arise from coping with a newborn who has health,



A Teenage Mother This week-old baby,

feeding, or sleeping difficulties. Birth itself—with its accompanying anesthesia, changing hormones, and pain-can precipitate depression (Ashman & Dawson, 2002; I. Jones, 2006). No matter what the cause, someone must help new parents adjust.

born in a poor village in Myanmar (Burma), has a better chance of survival than he might otherwise have had because his 18-year-old mother has bonded with him. SHEHZAD NOORANI/PETER ARNOLD, INC.

ESPECIALLY FOR Scientists

Research with animals can benefit people, but it is sometimes used too quickly to support conclusions about people. When does that happen? (see response, page 72)

BONDING

Focusing on the parents' emotions raises the question: Are the first hours crucial for formation of the **parent–infant bond,** the strong, loving connection that forms as parents hold, examine, and feed their newborn? Some claim that this bond must develop soon after birth when a mother touches her naked baby, just as sheep and goats must immediately smell and nuzzle their newborns if they are to nurture them (M. H. Klaus & Kennell, 1976).

However, research does not find that early skin-to-skin contact is essential for humans (Eyer, 1992; Lamb, 1982). Unlike sheep and goats, most mammals do not need immediate contact for parents to nurture their offspring. In fact, substantial research on monkeys focuses on *cross-fostering*, in which newborns are removed from their biological mothers in the first days of life and raised by another female or even a male. A strong and beneficial relationship does sometimes develop (Suomi, 2002).

KEY Points

- After about 38 weeks of gestation, birth occurs. A healthy newborn should weigh at least 5½ pounds (2,500 grams) and score at least 7 out of 10 on the Apgar scale.
- Medical intervention during pregnancy and birth is sometimes necessary to save lives, but it may occur too soon or too intensely.
- Prenatal care reduces the incidence of low birthweight, which is associated with poor nutrition, multiple births, and drug use.
- The newborn's well-being is affected by the father's participation, close physical contact with the parents, and postpartum depression in the mother. The parent-infant connection begins before birth and endures throughout life.

◀ Mothers, Fathers, and a Good Start ▶

2.5 Nature, Nurture, and the Phenotype

One goal of this chapter is to enable readers to understand the interaction between genes and experiences. As you remember, *nature* refers to genotype, a person's genetic inheritance. *Nurture* refers to everything that surrounds the person, including the prenatal environment. For every trait, scientists try to figure out how much of the phenotype results from nature and how much from nurture.

Hundreds of researchers in dozens of nations have studied thousands of twins—monozygotic and dizygotic, some raised together and others raised in different homes-and tens of thousands of other siblings, including stepsiblings, adopted siblings, and biological siblings. Furthermore, taking a multidisciplinary approach, biologists and biochemists have analyzed the molecules of DNA. The results of all their research have led to four general conclusions (B. J. Ellis & Bjorklund, 2005; Gottesman & Hanson, 2005; Plomin et al., 2003):

Too Cute? This portrait of the Genain sisters was taken 20 years before they all developed schizophrenia. However, from their identical hair ribbons to the identical position of their feet, it is apparent that their unusual status as quadruplets set them apart as curiosities. Could their life in the spotlight have nurtured their potential for schizophrenia? There is no way to know for sure. COURTESY OF EDNA MORLOK

- Genes affect every aspect of human behavior, including personality and learning.
- Nongenetic influences begin at conception and continue lifelong, sometimes altering genetic instructions.
- Most environmental influences on children raised in the same home are *not* shared, partly because parents treat each child differently.
- Children, adolescents, and, especially, adults "niche-pick," choosing environments that are compatible with their genetic inheritance.

In this section, to illustrate these conclusions and to highlight the central role that the nature—nurture interaction plays in development, we examine its influence on three conditions: nearsightedness, alcoholism, and birth defects caused by prenatal teratogens.

ANSWER TO OBSERVATION QUIZ

(from page 69) The drape prevents the mother from watching as her doctor stitches up the incision of a c-section. The sight is likely to be upsetting to many women, even though anesthesia allows them to remain awake without feeling pain.

Nature, Nurture, and the Phenotype

2.5.1 Nearsightedness

How well people can see depends on many factors, among them age, nutrition, and experience. Genes are certainly crucial. Nearsightedness, or *myopia*, is evident in more than 150 genetic syndromes (I. G. Morgan, 2003). Often the problem is "high" nearsightedness, which is very severe. More common among the general population is "low" nearsightedness, which renders distant objects blurry. Low nearsightedness runs in families, which indicates that it may be genetic.

A study of British twins found that minor variations in the Pax6 gene are a common cause of low nearsightedness (Hammond et al., 2004). This research found the heritability of myopia to be about 90 percent, which means that if one monozygotic twin is nearsighted, the other twin is almost always nearsighted, too.

But we must interpret that 90 percent carefully. Remember from page 49 that heritability is determined by calculating inherited variations *within* a particular group, in this case British twins. When we study only one group, all from the same era and region, it may seem as if genes are the major cause of poor vision (Farbrother & Guggenheim, 2001). However, historical and multicultural research finds that environment plays a powerful role.

A dramatic example comes from Africa, where at least 100,000 people have poor eyesight because their diet (and their mothers' diet during pregnancy) lacks sufficient vitamin A. For them, the heritability of nearsightedness is low because their environment is so influential. Scientists are working to develop a strain of maize (a staple of the African diet) that is high in vitamin A (Harjes et al., 2008). If they succeed, and if everyone eats the new strain, not only will eyesight improve among Africans but the heritability of nearsightedness will also increase.

Good Students These young Korean children are already learning to read a second language, Chinese. They are probably also advanced in math and science, compared with 8-year-olds in the United States. Their accomplishments may have come at a price: Many of them are nearsighted. REUTERS/KIM KYUNG-HOON/LANDOV

Nearsightedness among well-nourished children may also be affected by the environment. Among East Asian schoolchildren, nearsightedness has increased dramatically over the past two decades: from 12 to 84 percent among children in Taiwan; from 28 to 44 percent in Singapore; from 10 to 60 percent in Hong Kong (cited in Grosvenor, 2003). These numbers from cross-sectional samples have been confirmed by a sequential study in Singapore. The eyesight of every 17-year-old male in that city-state is tested as part of the exam for military service. In 1980, 26 percent of them were nearsighted; in 1990, 43 percent were (Tay et al., 1992).

Parents of these children are less often nearsighted. Since a Pax6 allelle makes a person vulnerable, and since children have the same genes as their parents, something in the children's environment must have interacted with

their Pax6 genes.

Schoolwork is one possibility. Fifty years ago, most East Asian children did not attend school; now most spend long hours studying. As their young eyes focus on their books, they may lose acuity for objects far away, becoming nearsighted.

Ophthalmologists suggest that if these children spent more time outside playing, fewer would need glasses (Goss, 2002; Grosvenor, 2003; I. G. Morgan, 2003). This explanation may be wrong, but something in nurture, not just nature, affects visual acuity.

RESPONSE FOR Scientists

(from page 71) Animal research tends to be used too quickly whenever it supports an assertion that is popular but has not been substantiated by research data, as in the social construction about physical contact being crucial for parent-infant bonding.

Nearsightedness

2.5.2 Alcoholism

Alcoholism was once thought to be a sinful habit, a sign of moral weakness (Kobler, 1973/1993). Today, however, most people realize that alcoholism is a disease that originates with the genes. Instead of blaming alcoholics for a lack of moral fiber, developmentalists now blame their biochemistry. Genes can cause an overpowering addictive pull in some people (Heath et al., 2003).

As with most other genetic conditions, alcoholism is "likely to be conferred by multiple genes of small to modest effects, possibly only apparent in gene–environment interactions" (Enoch, 2006, p. 193). Those multiple genes include some that affect the body, influencing how the person metabolizes and digests alcohol, and some that affect the mind.

Certain genetic traits (a quick temper, sensation seeking, high anxiety) in certain contexts make it hard to abstain; conversely, other traits (such as a need for self-control) and other contexts (such as a church social in a "dry" county) make it hard to drink. Note that context is crucial: Alcoholism is genetic, but the environment interacts with the genes. This is apparent even before birth.

ESPECIALLY FOR Future Drug Counselors

Is the wish for excitement likely to lead to addiction? (see response, page 75)

In 1973, scientists first realized that some pregnant women who drank heavily had babies with distorted facial features, including small eyes and thin upper lips. Those infants had what is now known as **fetal alcohol syndrome (FAS)**; as children, they were mentally retarded, impulsive, and hyperactive (Calhoun & Warren, 2007).

Yes, But... An adopted boy points out something to his father-a positive interaction between the two. The shapes of the boy's eyes, ears, and upper lip indicate that he was born with fetal alcohol syndrome (FAS). Scientists disagree about the strength of the correlation between FAS and drinking alcohol during pregnancy. © DAVID H. WELLS/CORBIS

Alcohol is definitely the cause of FAS, but some infants born to drinking mothers seem unharmed. One reason they are protected is genetic, as is indicated when dizygotic twins are affected differently before birth by their mother's drinking. Because it is impossible to predict which fetuses will and will not be affected, almost all obstetricians in the United States (fewer in Europe) advise women who are, or may become, pregnant to abstain from alcohol completely.

Since 1998, four states have authorized "involuntary commitment" (jail or forced residential treatment) for pregnant women who do not stop drinking. Advocates for women consider such laws discriminatory, especially since they are more often enforced for minority women. Some experts believe that the threat of jail may cause women who most need prenatal care to avoid getting it. In that case, nurture would intensify, rather than moderate, the effects of nature.

Genes are crucial in cultures where alcohol is readily available and being drunk is considered acceptable, but they are irrelevant for people who live where alcohol is unavailable (rural Saudi Arabia, for instance). In such places, no one can become an active alcoholic,

so it does not matter what genes a person has for alcohol metabolism. Nature *and* nurture must combine to create an alcoholic.

Consider the story of baseball superstar Mickey Mantle. He mistakenly thought his family's genetic patterns were destiny, not a warning (Jaffe, 2004). Several of his male relatives died before middle age, including Mantle's father, who died of cancer at age 39. Many relatives on both sides of his family were heavy drinkers of alcohol. Knowing his genetic background, Mantle might have chosen to avoid alcohol. Instead, he became "a notorious alcoholic [because he] believed a family history of early mortality meant he would die young" (Jaffe, 2004, p. 37). At age 46, Mantle purportedly said, "If I knew I was going to live this long, I would have taken better care of myself." He kept on drinking. Despite a liver transplant, liver damage killed him at age 63, 15 years short of the life expectancy for men of his time.

FOR DEEPER REFLECTION

How much protection, if any, should the legal system provide for fetuses? Should alcoholic women who are pregnant be jailed to prevent them from drinking? What about people who enable them to drink, such as their partners, their parents, bar owners, bartenders?

◀ Alcoholism ▶

2.5.3 Prenatal Teratogens

Most pregnant women in the modern world are exposed to **teratogens.** Teratogens are substances (such as drugs and pollutants) and conditions (such as severe malnutrition and extreme stress) that increase the risk of prenatal abnormalities. For many reasons, the risks are much higher for some pregnancies than for others. **Table 2.7** below lists some teratogens and their effects, as well as protective measures.

The possible harm that a teratogen may do to a developing organism (whether zygote, embryo, or fetus) is more extensive than people once thought. Structural abnormalities (such as malformed faces) are obvious at birth, but many substances are *behavioral teratogens*. These affect the child's developing brain, making him or her slow to talk, hyperactive, or learning-disabled. The effects of behavioral teratogens do not become evident until months or years after birth.

Pregnant women in the twenty-first century cannot avoid all teratogens: At some point during pregnancy, everyone is exposed to a virus, or breathes a pollutant, or is emotionally stressed. Through the mother, these circumstances may affect the fetus; the specifics depend on both nature and nurture.

UNDERSTANDING THE NUMBERS

What was the average life expectancy for men when Mickey Mantle died?

Answer Age 78 (63 + 15), the average for U.S. men who had reached age 50. Mantle may have misunderstood cohort effects as well as genetics. When he was born in 1931, the average male life span was 63 because one of every 15 boys died in infancy, and his father's early death was not unusual. When Mantle died in 1995, the average newborn lived to age 70, and most men his age lived to 80.

Table 2.7 Teratogens: Effects of Exposure and Prevention of Damage

Teratogens	Effects on Child of Exposure	Measures for Preventing Damage
Diseases		
Rubella (German measles)	In embryonic period, causes blindness and deafness; in first and second trimesters, causes brain damage	Get immunized before becoming pregnant
Toxoplasmosis	Brain damage, loss of vision, mental retardation	Avoid eating undercooked meat and handling cat feces, garden dirt
Measles, chicken pox, influenza	May impair brain functioning	Get immunized before getting pregnant; avoid infected people during pregnancy
Syphilis	Baby is born with syphilis, which, untreated, leads to brain and bone damage and eventual death	Early prenatal diagnosis and treatment with antibiotics
AIDS	Baby may catch the virus. Without treatment, illness and death are likely during childhood.	Prenatal drugs and cesarean birth make AIDS transmission rare
Other sexually transmitted infections, including gonorrhea and chlamydia	Not usually harmful during pregnancy but may cause blindness and infections if transmitted during birth	Early diagnosis and treatment; if necessary, cesarean section, treatment of newborn
Infections, including infections of urinary tract, gums, and teeth	May cause premature labor, which increases vulnerability to brain damage	Get infection treated, preferably before becoming pregnant
Pollutants		
Lead, mercury, PCBs (polychlorinated biphenyls), dioxin, and some pesticides, herbicides, and cleaning compounds	May cause spontaneous abortion, preterm labor, and brain damage	Most common substances are harmless in small doses, but pregnant women should avoid regular and direct exposure, such as drinking well water, eating unwashed fruits or vegetables, using chemical compounds, eating fish from polluted waters
Radiation		
Massive or repeated exposure to radiation, as in medical X-rays	In the embryonic period, may cause abnormally small head (microcephaly) and mental retardation; in the fetal period, suspected but not proven to cause brain damage. Exposure to background radiation, as from power plants, is usually too low to have an effect.	Get sonograms, not X-rays, during pregnancy; pregnant women who work directly with radiation need special protection or temporary assignment to another job
Social and Behavioral Factors		
Very high stress	Early in pregnancy, may cause cleft lip or cleft palate, spontaneous abortion, or preterm labor	Get adequate relaxation, rest, and sleep; reduce hours of employment; get help with housework and child care
Malnutrition	When severe, may interfere with conception, implantation, normal fetal	Eat a balanced diet (with adequate vitamins and minerals, including,

	development, and full-term birth	especially, folic acid, iron, and vitamin A); achieve normal weight before getting pregnant, then gain 25–35 lbs (10–15 kg) during pregnancy
Excessive, exhausting exercise	Can affect fetal development when it interferes with pregnant woman's sleep, digestion, or nutrition	Get regular, moderate exercise
Medicinal Drugs		
Lithium	Can cause heart abnormalities	Avoid all medicines, whether prescription or over-the-counter, during pregnancy unless they are approved by a medical professional who knows about the pregnancy and is aware of the most recent research
Tetracycline	Can harm the teeth	
Retinoic acid	Can cause limb deformities	
Streptomycin	Can cause deafness	
ACE inhibitors	Can harm digestive organs	
Phenobarbital	Can affect brain development	
Thalidomide	Can stop ear and limb formation	
Psychoactive Drugs		
Caffeine	Normal use poses no problem	Avoid excessive use: Drink no more than three cups a day of beverages containing caffeine (coffee, tea, cola drinks, hot chocolate)
Alcohol	May cause fetal alcohol syndrome (FAS) or fetal alcohol effects (FAE)	Stop or severely limit alcohol consumption during pregnancy; especially dangerous are three or more drinks a day or four or more drinks on one occasion
Tobacco	Reduces birthweight, increases risk of malformations of limbs and urinary tract, and may affect the baby's lungs	Stop smoking before and during pregnancy
Marijuana	Heavy exposure may affect the central nervous system; when smoked, may hinder fetal growth	Avoid or strictly limit marijuana consumption
Heroin	Slows fetal growth and may cause premature labor; newborns with heroin in their bloodstream require medical treatment to prevent the pain and convulsions of withdrawal	Get treated for heroin addiction before becoming pregnant; if already pregnant, gradual withdrawal on methadone is better than continued use of heroin
Cocaine	May cause slow fetal growth, premature labor, and learning problems in the first years of life	Stop using cocaine before pregnancy; babies of cocaine-using mothers may need special medical and educational attention in their first years of life
Inhaled solvents (glue or aerosol)	May cause abnormally small head, crossed eyes, and other indications of brain damage	Stop sniffing inhalants before becoming pregnant; be aware that serious damage can occur before a woman knows she is pregnant

Note: This table summarizes some relatively common teratogenic effects. As the text makes clear, many individual factors in each pregnancy affect whether a given teratogen will actually cause damage and what that damage might be. This is a general summary of what is known; new evidence is reported almost daily, so some of these generalities will change. Pregnant women or women who want to become pregnant should consult with their physicians.
Sources: Mann & Andrews, 2007; O'Rahilly & Muller, 2001; Reece & Hobbins, 2007; Shepard & Lemire, 2004; L. T. Singer et al., 2002.

GENETIC VULNERABILITY

Some zygotes carry genes that make them vulnerable. For example, Japan has excellent preconception and prenatal care and, consequently, lower-than-average rates of almost every birth defect. However, babies with cleft lip are born in Japan at three times the rate in Canada (World Health Organization, 2003), suggesting that cleft lip is influenced by genes and that those genes are more common in Japanese people than in Canadians.

The neural tube, which develops into the central nervous system, is the first part of the embryo to form. Neural-tube defects, which lead to abnormalities of the spine and brain, are caused by abnormally low levels of folic acid in a pregnant woman's body. This deficiency is often dietary, but it may also have a genetic cause: Some women do not metabolize folic acid well.

RESPONSE FOR Future Drug Counselors

(from page 72): Maybe. Some people who love risk become addicts; others develop a healthy lifestyle that includes adventure, new people, and exotic places. Any trait can lead in various directions. You need to be aware of the connection so that you can steer your clients toward healthy adventures.

A woman would not know about her metabolism of folic acid, so all women are urged to take vitamin supplements containing folic acid *before* becoming pregnant. However, only one-third of U.S. women of childbearing age do so (Suellentrop et al., 2006). In response, folic acid is now added to many foods in the United States and Canada, and the rate of spina bifida (a serious condition in which

the spinal column is not completely closed) has decreased.

TIMING OF EXPOSURE

Each body part has a *critical period* when that body part develops, followed by a *sensitive period* when teratogens can interfere with recent growth (see **Figure 2.8**). Teratogens are most harmful to whatever part of the embryo is forming at the time of exposure (K. L. Moore & Persaud, 2003).

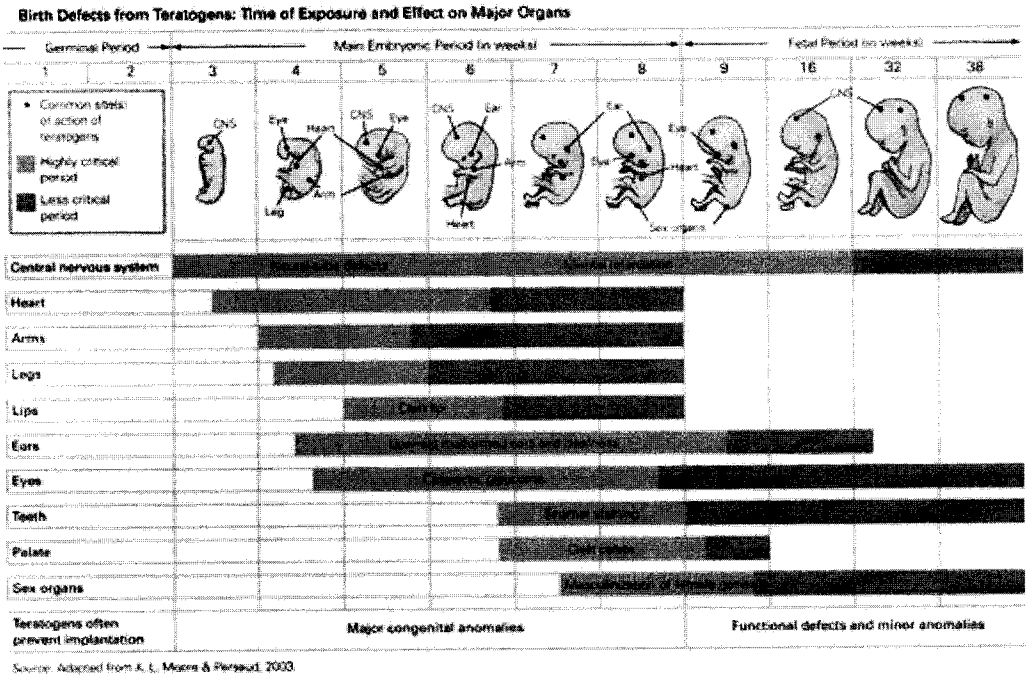


Figure 2.8 Critical Periods in Human Development The most serious damage from teratogens (orange bars) is likely to occur early in prenatal development. However, significant damage (purple bars) to many vital parts of the body, including the central nervous system (CNS, the brain and spinal cord), eyes, and genitals, can occur during the last months of pregnancy as well.

For structural abnormalities, the critical period is the first two months, often before the woman knows that she is pregnant. This is also when most spontaneous abortions occur, typically because something is severely amiss.

However, body functioning and the cortex continue to mature throughout the embryonic and fetal stages of development. Consequently, teratogens that cause preterm birth (notably cigarettes) are particularly harmful in the second half of pregnancy; others (notably psychoactive drugs) are harmful to the brain at any time, even in the last weeks before birth.

AMOUNT OF EXPOSURE

The critical period is an aspect of heredity (nature); the other crucial factor causing birth defects is the amount of exposure to a teratogen (nurture). Some teratogens have a **threshold effect**; they are virtually harmless until exposure reaches a certain level, when they "cross the threshold" and become damaging (O'Rahilly & Müller, 2001).

For instance, bisphenol A (BPA) is a chemical compound used to make clear

plastic. Everyone has trace amounts in their bodies; that seems harmless. But too much is teratogenic in mice, causing infertility, obesity, diabetes, and cancer. Scientists do not know the threshold for humans, but some advise pregnant women to avoid plastic containers and plastic dishware, since the embryo and the fetus are more vulnerable than the adult (Vandenberg et al., 2007).

Similar uncertainties exist for dozens of other chemicals that were unknown 50 years ago, including several of the plasticizing chemical compounds known as phthalates and dozens of chemicals found in pesticides, prescription drugs, cosmetics, and herbicides. Note that these substances are found in most modern societies. The *nature* of prenatal development has not changed much for thousands of years, but *nurture*, or environmental influences, includes many new substances.

Some teratogens reach the threshold of harm only when a pregnant woman ingests them. Accordingly, pregnant women should avoid taking prescription drugs unless their doctor is aware that they are pregnant, is familiar with the research on the drug's effects, and is convinced that the drug is necessary. Exposure to other teratogens is literally unavoidable because they are present in the air and water. Trace amounts are harmless, but it is not known when a trace becomes a teratogen.

Indeed, some substances are beneficial up to a point but are fiercely teratogenic in large quantities. For example, vitamin A is an essential part of the prenatal diet, but 50,000 or more daily units of vitamin A can cause the development of abnormal body structures in the fetus. Some of the nutrients in fish are beneficial to fetal brain development; but fish may also contain pollutants that are harmful when ingested in sufficient quantities.

Only research on pregnant humans—thousands of them, studied longitudinally—can determine the exact threshold for a specific teratogen. Such a study would need to be comprehensive, since substances interact to raise or lower the risk. Furthermore, since each woman and each fetus is unique, the threshold varies from one person to another.

Prenatal Teratogens

2.5.4 Resolving Uncertainties

This point highlights a problem with all advice and warnings about nature and nurture. Pregnancy need not be an anxious time, filled with restrictions and fears about diet, diseases, drugs, and other possible dangers. The anxiety itself may reduce sleep, impair digestion, and raise blood pressure—all of which may hinder development.

Good prenatal care teaches women what they can do—what vitamins to take, what drugs to avoid, what foods to eat—to help ensure that they will have healthy babies. Pregnant women who drink and smoke, for instance, are more likely to quit or at least cut down with professional medical advice than without it.

Sometimes prenatal care literally saves a life. For example, if testing in the first two months discovers maternal syphilis or HIV, treatment can prevent transmission of the virus to the fetus. Prenatal care can also protect the fetus if a woman is anemic, has high blood pressure, or develops gestational diabetes.

Ideally, prenatal care reassures prospective parents that all is well, thus helping to ensure a healthy pregnancy by reducing stress (see **Table 2.8**). Both future parents can listen to the heartbeat and view a sonogram of the fetus. Hearing and seeing their developing baby are likely to increase their motivation to reduce the fetus's exposure to risks. Thus, one more developing person will be ready for 80 years or so of life on Earth.

Table 2.8 Before Pregnancy

What Prospective Mothers Should Do	What Prospective Mothers Really Do (U.S. data)
1. Plan the pregnancy.	1. About 60 percent of pregnancies are intended.
2. Take a daily multivitamin with folic acid.	2. About 40 percent of women aged 18 to 45 take vitamins.
3. Avoid binge drinking (defined as four or more drinks in a row).	3. One-eighth of all women who might become pregnant binge drink.
4. Update immunizations against all teratogenic viruses, especially rubella.	4. Because of laws regarding school admission, most young women in the United States are well immunized.
5. Gain or lose weight, as appropriate.	5. Babies born to underweight women are at risk for low birthweight. Babies born to obese women have three times the usual rate of birth complications. About half of all women begin pregnancy at an appropriate weight.
6. Reassess use of prescription drugs.	6. Eighty-five percent of pregnant women take prescription drugs (not counting vitamins).
7. Maintain exercise habits.	7. Most women do not exercise, especially in the third trimester.

Sources: Downs & Hausenblas, 2007; Suellentrop et al., 2006; Tsai et al., 2007.

KEY Points

- Nature and nurture always interact; neither operates in isolation.
- Virtually all physical and psychological conditions–including nearsightedness, alcoholism, and the effects of prenatal teratogens —have genetic roots.
- Environmental influences allow genes to affect the phenotype or not —as when a genetic tendency toward alcoholism is irrelevant because the person never drinks.
- Good prenatal care can help to ensure a healthy baby, assure the

prospective parents that all is well, and even save lives.

Resolving Uncertainties

SUMMARY

The Beginning of Life

1. Genes are the foundation for all development. Human conception occurs when two gametes (an ovum and a sperm, each with 23 chromosomes) combine to form a zygote, 46 chromosomes in a single cell.

2. Every cell of every human being has a unique genetic code made up of about 20,000 genes, some in variations called alleles. The environment interacts with the genetic instructions for every trait.

3. The sex of an embryo depends on the sperm: A Y sperm creates an XY (male) embryo; an X sperm creates an XX (female) embryo. Twins occur if a zygote splits in two (monozygotic, or identical, twins) or if two ova are fertilized by two sperm (dizygotic, or fraternal, twins).

4. Genes interact in various ways; sometimes additively, with multiple genes contributing to a trait, and sometimes in a dominant–recessive pattern. Heritability refers to the impact of genes on a trait within a specific population, not necessarily within an individual.

Genetic Problems

5. Often a zygote has more or fewer than 46 chromosomes. Such zygotes usually do not develop; the main exceptions are those with three chromosomes at the 21st location (Down syndrome, or trisomy-21) or an odd number of sex chromosomes.

6. Genetic testing and counseling can help many couples learn whether their future children are at risk for a chromosomal or genetic abnormality. The odds depend on what types of genes are involved (dominant, recessive, or additive). The final decisions are made by the couple after learning the facts and possibilities.

From Zygote to Newborn

7. The first two weeks of prenatal growth are called the germinal period. The cells differentiate, as the developing organism implants itself in the lining of the uterus.

8. The period from the third through the eighth week after conception is called the embryonic period. The heart begins to beat, and the eyes, ears, nose, and mouth form. By the eighth week, the embryo has the basic organs and features of a human, with the exception of the sex organs.

9. The fetal period extends from the ninth week until birth. By the 12th week, all the organs and body structures have formed. The fetus attains viability at 22 weeks, when the brain is sufficiently mature to regulate basic body functions.

10. The average fetus gains approximately 4½ pounds (2,000 grams) during the last three months of pregnancy. Maturation of brain, lungs, and heart

ensures survival of virtually all full-term babies.

Birth

11. Medical intervention can speed contractions, dull pain, measure health via the Apgar scale, and save lives, but it is sometimes impersonal and unnecessary. The goal is a balance, protecting the baby but also allowing parental involvement and control.

12. Birth complications, such as unusually long and stressful labor that includes anoxia (a lack of oxygen to the fetus), have many causes. Low birthweight (under 5½ pounds, or 2,500 grams) may arise from multiple births, placental problems, maternal illness, malnutrition, smoking, drinking, drug use, and age.

13. Kangaroo care is helpful when the newborn is of low birthweight. Mother-newborn interaction should be encouraged, although the parent–infant bond depends on many factors in addition to birth practices.

14. Many women feel unhappy, incompetent, or unwell after giving birth. Postpartum depression gradually disappears with appropriate help; fathers are particularly crucial to the well-being of mother and child, although they, too, are vulnerable to depression.

Nature, Nurture, and the Phenotype

15. Nature and nurture interact to cause virtually all human problems, including alcoholism and nearsightedness. Whether a teratogen harms an embryo or fetus depends not only on genes but also on timing and amount of exposure.

SUMMARY

KEY QUESTIONS

1. How and when is the sex of a zygote determined?

2. Genetically speaking, what are the similarities and differences between individuals and between humans and other animals?

3. What are the differences among monozygotic twins, dizygotic twins, and other siblings?

4. What are the causes and effects of a zygote with more or fewer than 46 chromosomes?

5. Genetic testing for various diseases is much more common now than it once was. What are the advantages and disadvantages?

6. What are the major differences between an embryo at 2 weeks and at 8 weeks after conception?

7. Since almost all fetuses born at 30 weeks survive, why don't women have an elective cesarean at that time?

8. How and when does a pregnant woman's husband influence pregnancy?

9. How have medical procedures helped *and* harmed the birth process?

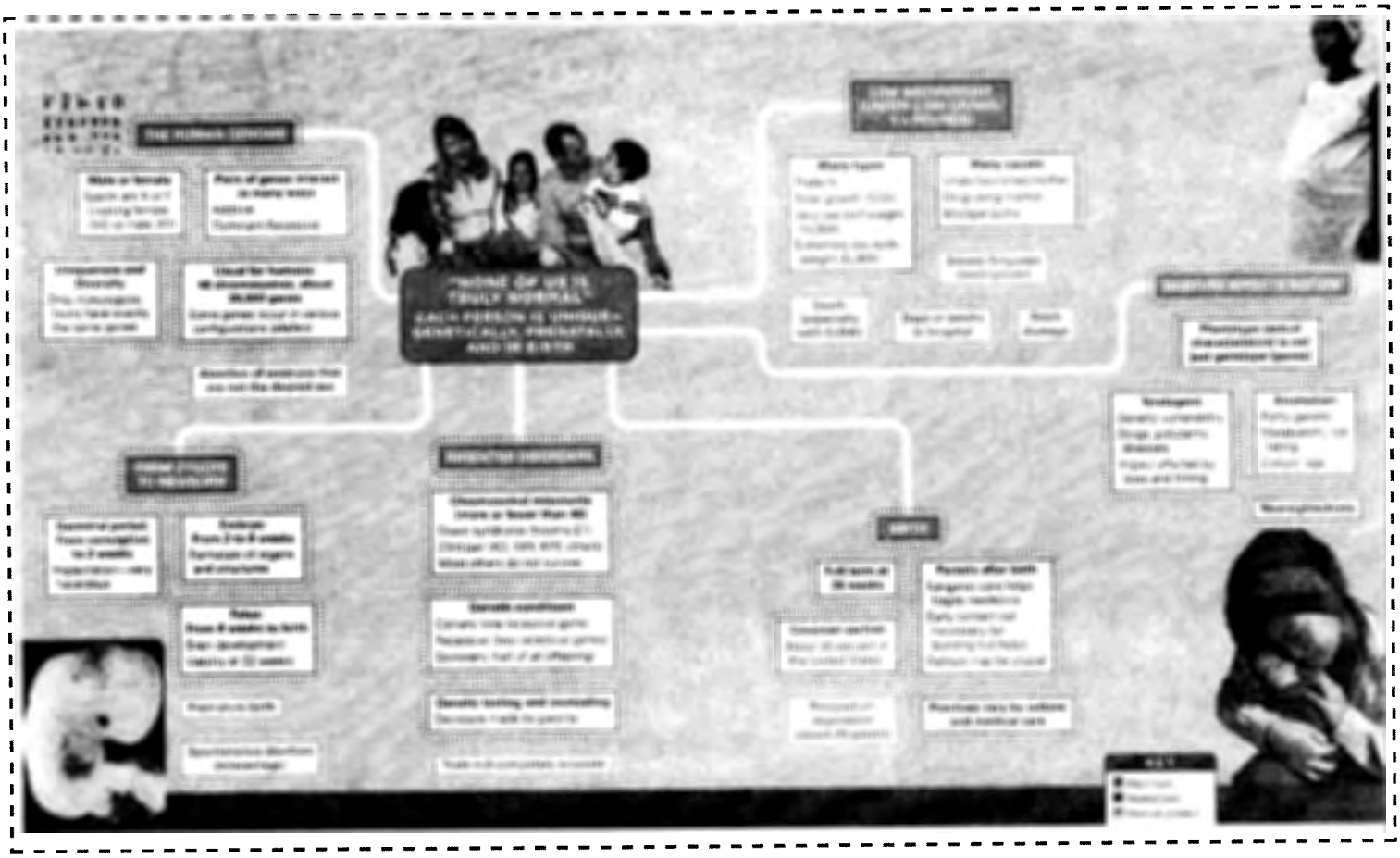
10. Why do hospitals encourage parents of fragile newborns to provide some care, even if the newborn is in critical condition?

11. Name four causes of low birthweight. What can be done to prevent each one?

12. What can be done to relieve the effects of postpartum depression on the mother, the father, and the infant?

KEY QUESTIONS

CHAPTER 2: CONCEPT REVIEW



CHAPTER 2: CONCEPT REVIEW

KEY TERMS

DNA (deoxyribonucleic acid) (p. 43)

chromosome (p. 43)

gamete (p. 43)

zygote (p. 44)

gene (p. 44)

genotype (p. 44)

phenotype (p. 44)

allele (p. 45)

genome (p. 45)

monozygotic twins (p. 46)

dizygotic twins (p. 47)

XX (p. 47)

XY (p. 47)

polygenic (p. 48)

multifactorial (p. 48)

regulator gene (p. 48)

additive gene (p. 48)

dominant-recessive pattern (p. 48)

carrier (p. 49)

heritability (p. 49)

Down syndrome (p. 50)

fragile X syndrome (p. 51)

genetic counseling (p. 53)

phenylketonuria (PKU) (p. 55)

germinal period (p. 57)

embryonic period (p. 57)

fetal period (p. 57)

placenta (p. 59)

implantation (p. 59)

embryo (p. 60)

fetus (p. 60)

sonogram (p. 61)

age of viability (p. 61)

Apgar scale (p. 63)

doula (p. 65)

cesarean section (c-section) (p. 65)

low birthweight (LBW) (p. 67)

very low birthweight (VLBW) (p. 67)

extremely low birthweight (ELBW) (p. 67)

preterm birth (p. 68)

small for gestational age (SGA) (p. 68)

kangaroo care (p. 69)

postpartum depression (p. 70)

parent-infant bond (p. 70)

fetal alcohol syndrome (FAS) (p. 73)

teratogens (p. 73)

threshold effect (p. 76)

KEY TERMS