CHAPTER 2

Genetics and prenatal development

For mothers-to-be worldwide, pregnancy is often experienced with a combination of joy, hope and fear.

Yet, here, as in other aspects of development, the experience differs substantially depending on the economic and cultural context. For most women in rural areas of developing countries, there is little in the way of technology or medical care to promote the healthy development of the fetus. Instead, pregnant women often rely on folk beliefs, a midwife's years of experience and social support from the extended family. For most women in developed countries, medical care and technological aids are available throughout pregnancy. Yet prospective mothers and fathers face formidable challenges in altering their lives to make room for the demands of raising a small child often while continuing to pursue their careers.

Pregnancy is experienced in many different ways around the world, but everywhere it is a momentous event. In this chapter we examine the process of prenatal development, from its genetic beginnings until the final months of pregnancy. The first section of the chapter covers the basics of genetics and how a new human life begins. In the second section, we examine prenatal development and prenatal care for both mother and baby to enhance the likelihood that all will go well. Sometimes problems arise in the course of pregnancy, so the final section of the chapter addresses prenatal complications as well as testing and counselling options.

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SECTION 1

GENETIC INFLUENCES ON DEVELOPMENT

LEARNING OBJECTIVES

- **LO 2.1** Distinguish between genotype and phenotype and identify the different forms of genetic inheritance.
- **LO 2.2** Describe the sex chromosomes and identify what makes them different from other chromosomes.
- **LO 2.3** Explain how behaviour geneticists use heritability estimates and concordance rates in their research.
- **LO 2.4** Describe how the concept of epigenesis frames gene–environment interactions and connect epigenesis to the concept of reaction range.
- **LO 2.5** Explain how the theory of genotype → environment effects casts new light on the old nature–nurture debate.
- **LO 2.6** Outline the process of meiosis in the formation of reproductive cells.
- **LO 2.7** Describe the process of fertilisation and conception.
- **LO 2.8** List the major causes of and treatments for infertility, and describe how infertility is viewed in different cultures.

LO 2.1

Distinguish between genotype and phenotype and identify the different forms of genetic inheritance.

chromosome
sausage-shaped structure in the nucleus of cells, containing genes, which are paired, except in reproductive cells

DNA (deoxyribonucleic acid)
long strand of cell material that stores and transfers genetic information in all life forms

gene
segment of DNA containing coded instructions for the growth and functioning of the organism

genome
entire store of an organism's hereditary information

genotype
organism's unique genetic inheritance

phenotype
organism's actual characteristics, derived from its genotype

GENETIC INFLUENCES ON DEVELOPMENT: GENETIC BASICS

In all organisms, humans included, individual development has a genetic beginning. To understand the role of genetics in human development, it is important to have a basic foundation of knowledge about genes and how they function.

Genotype and phenotype

Nearly all cells in the human body contain 46 **chromosomes** in 23 pairs, with one chromosome in each pair inherited from the mother and the other inherited from the father (see Figure 2.1). The chromosomes are composed of complex molecules known as **DNA (deoxyribonucleic acid)** (see Figure 2.2). The DNA in the chromosomes is organised into segments called **genes**, which are the basic units of hereditary information. Genes contain paired sequences of chemicals called *nucleotides*, and these sequences comprise instructions for the functioning and replication of the cells. There are about 23 000 genes in our 46 chromosomes, the total human **genome**, with altogether about 3 billion nucleotide pairs (Ezkurdia et al., 2014).

Not all 23 000 genes are expressed in the course of development. The totality of an individual's genes is the **genotype**, and the person's actual expressed characteristics are called the **phenotype**. In part, the difference between genotype and phenotype is a consequence of the person's environment. For example, if you were born with a genotype that included exceptional musical ability, this talent might never be developed if your environment provided no access to musical instruments or musical instruction. Consequently, the musical ability present in your genotype would not be apparent in your phenotype.

Another aspect of genetic functioning that influences the relation between genotype and phenotype is **dominant–recessive inheritance** (Jones & Lopez, 2014). On every pair of chromosomes there are two forms of each gene, one on the chromosome inherited

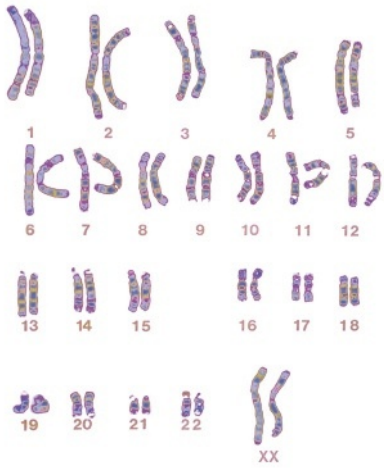

FIGURE 2.1 The human genome

The 46 chromosomes in the human genome are organised into 23 pairs. This genome is of a female; in a male the 23rd pair would be XY rather than XX.

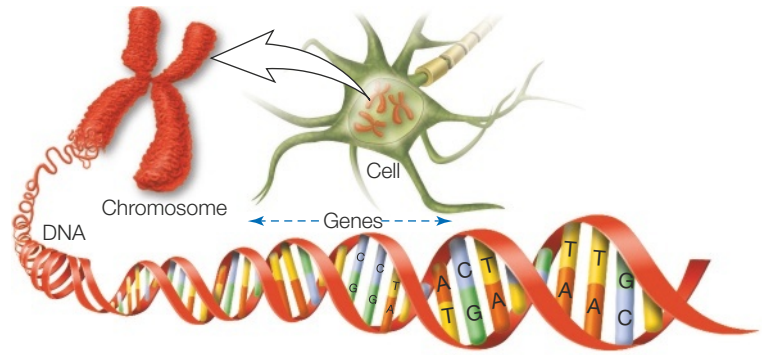

FIGURE 2.2 The chemical structure of DNA

DNA is composed of nucleotide pairs.

from the mother and one on the chromosome inherited from the father. Each form of the gene is called an **allele**. On many of these pairs of alleles, dominant–recessive inheritance occurs. This means that only one of the two genes—the *dominant gene*—influences the phenotype, whereas the *recessive gene* does not, even though it is part of the genotype. For example, if you inherited a gene for curly hair from one parent and a gene for straight hair from the other, you would have curly hair because curly hair is dominant and straight hair is recessive. Recessive genes are expressed in the phenotype only when they are paired with another recessive gene. A clear pattern of dominant–recessive inheritance is evident only for traits determined by a single gene, which is not true of most traits, as we will see shortly. Some other examples of dominant and recessive characteristics are shown in Table 2.1.

dominant–recessive inheritance
pattern of inheritance in which a pair of chromosomes contains one dominant and one recessive gene, but only the dominant gene is expressed in the phenotype

allele
on a pair of chromosomes, each of two forms of a gene

TABLE 2.1 TRAITS WITH SINGLE-GENE DOMINANT–RECESSIVE INHERITANCE

DOMINANT	RECESSIVE
Curly hair	Straight hair
Dark hair	Blonde hair
Facial dimples	No dimples
Normal hearing	Deafness (some forms)
Normal vision	Nearsighted vision
Freckles	No freckles
Unattached ear lobe	Attached ear lobe
Can roll tongue in U-shape	Cannot roll tongue in U-shape

Most characteristics in human development are not determined solely by a single pair of genes. Despite what you may have heard about the supposed existence of a 'gay gene' or 'religion gene' or 'crime gene', no such genes have been found, nor are they likely to be (Carr, 2010; Pinker, 2004). Although single gene pairs sometimes play a crucial role in development, more commonly the influence of genes is a consequence of

polygenic inheritance
expression of phenotypic characteristics as a result of the interaction of multiple genes

polygenic inheritance, the interaction of multiple genes (Lewis, 2005). This is true for physical traits such as height, weight and skin colour, as well as for traits such as intelligence, personality and susceptibility to various diseases (Hoh & Ott, 2003; Karlsson, 2006; Rucker & McGuffin, 2010).

LO 2.2

Describe the sex chromosomes and identify what makes them different from other chromosomes.

The sex chromosomes

sex chromosomes
chromosomes that determine whether an organism is male (XY) or female (XX)

Of the 23 pairs of chromosomes, one pair is different from the rest. These are the **sex chromosomes**, which determine whether the person will be male or female (Jones & Lopez, 2014). In the female, this pair is called XX; in the male, XY. The Y chromosome is notably smaller than other chromosomes and contains only one-third the amount of the genetic material. All eggs in the mother contain an X chromosome but sperm may carry either an X or a Y chromosome. So, it is the father's sperm that determines what the sex of the child will be. Ironically, people in many cultures mistakenly believe that the woman is responsible for the child's sex, and blame her if she fails to have sons (DeLoache & Gottlieb, 2000).

People in many cultures also have beliefs about how to predict the baby's sex (DeLoache & Gottlieb, 2000). Such beliefs demonstrate how important gender is to a child's future in most cultures, even before birth. Many cultures have a bias in favour of boys, and the use of sex-selective abortion to achieve this is resulting in gender ratios skewed towards boys, especially in Asian cultures where this bias is especially pronounced (Abrejo, Shaikh & Rizvi, 2009). In China, the birth ratio is the most skewed, with 120 males born for every 100 females (Hesketh, Lu & Xing, 2011). In Australia, the National Health and Medical Research Council bans the use of technology for sex selection, but this does not prevent an individual from choosing to have an abortion for any reason (Whittaker, 2015).

X-linked inheritance
pattern of inheritance in which a recessive characteristic is expressed because it is carried on the male's X chromosome

The sex of the developing organism also has biological consequences for prenatal development. Having only one X chromosome makes males more vulnerable than females to a variety of recessive disorders that are linked to the X chromosome (Narayanan & Warren, 2006). The reason for this is that if a female has one X chromosome that contains the recessive gene for a disorder, the disorder will not show up in her phenotype because the dominant gene on her other X chromosome will prevent the disorder from being expressed. She will be a carrier of the disorder to the next generation but will not have the disorder herself. In contrast, if a male receives one X chromosome containing the recessive gene for a disorder, he will definitely have the disorder because he has no other X chromosome that may contain a dominant gene to block its expression. His Y chromosome cannot serve this function. An example of this pattern of **X-linked inheritance** is shown in Figure 2.3 for haemophilia, a disorder in which the blood does not clot properly and the person may bleed to death from even a minor injury. Because of X-linked inheritance, males are at greater risk for a wide variety of genetically based conditions, including

In China, sons tend to be favoured over daughters and this contributes to a skewed ratio of boys to girls.

Jarun Ontakrai/Shutterstock

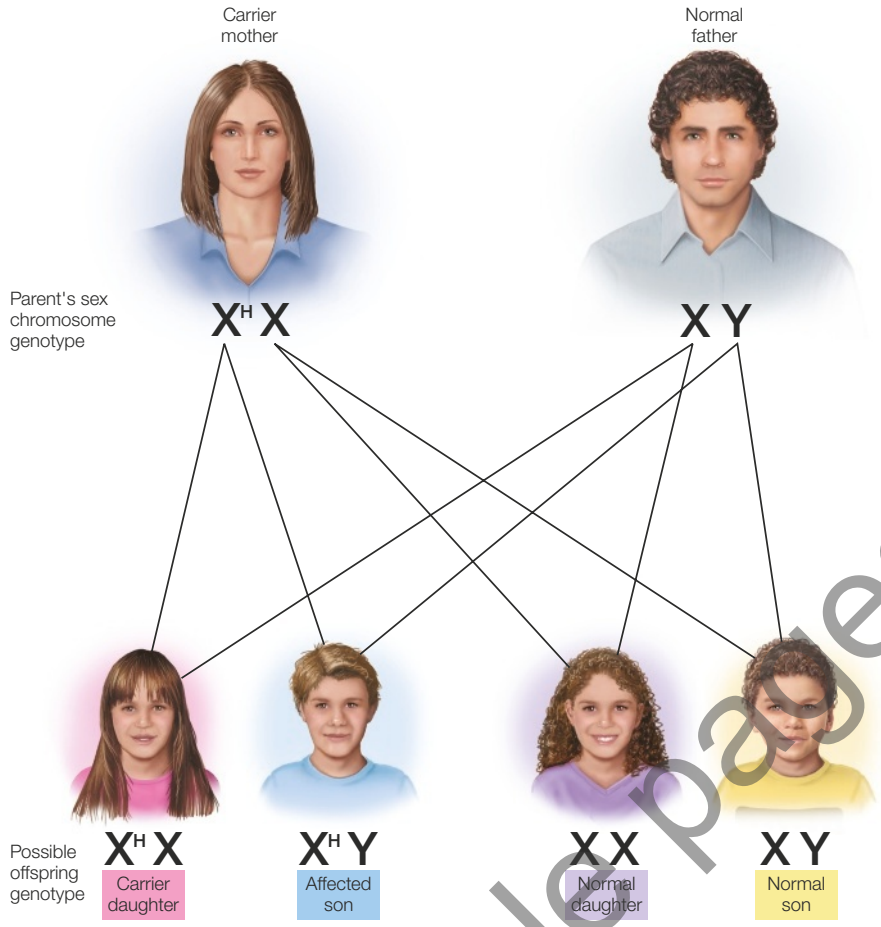

FIGURE 2.3 X-linked inheritance in haemophilia

Why are males more vulnerable to recessive disorders carried on the X chromosome?

learning disabilities and intellectual disability (Halpern, 2000; James, Hadley, Holtzman & Winkelstein, 2006). Humans naturally give birth to about 105 boys per 100 girls. Evidently, this is nature's way of compensating for the greater genetic vulnerability of males (World Health Organization [WHO], 2017a).

GENETIC INFLUENCES ON DEVELOPMENT: GENES AND THE ENVIRONMENT

There is no doubt that genes have some influence on human development—but how much? Scholars have long debated the relative importance of genes and the environment in human development. In this **nature–nurture debate**, some scholars have claimed that development can be explained by genes (nature) and that environment matters little, whereas others have claimed that development depends mainly on environmental factors (nurture) (compare Baumrind, 1993; Scarr, 1993). In recent years, most scholars have reached a consensus that both genes and environment play key roles in human development, although the relative strength of nature and nurture continues to be debated (Dodge, 2007; Lickliter & Honeycutt, 2015; Pinker, 2004).

nature–nurture debate
debate among scholars as to whether human development is influenced mainly by genes (nature) or environment (nurture)

behaviour genetics
field in the study of human development that aims to identify the extent to which genes influence behaviour, primarily by comparing people who share different amounts of their genes

Principles of behaviour genetics

The question of how much genes influence human development is at the heart of the field of **behaviour genetics** (Chabris, Lee, Cesarini, Benjamin & Laibson, 2015; Gottesman, 2004; Plomin, 2009). Researchers who work in behaviour genetics estimate the influence of genes on development by comparing people who share different amounts of their genes, mainly

LO 2.3

Explain how behaviour geneticists use heritability estimates and concordance rates in their research.

MZ twins have the same genotype.

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through twin studies and adoption studies. Identical or **monozygotic (MZ) twins** have 100% of their genes in common. Fraternal or **dizygotic (DZ) twins** and siblings have 40–60% of their genes in common. Consequently, when MZ twins are more similar than DZ twins or siblings, this indicates that genetics play a strong role. A study of Australian twins was able to determine that children's scores on the NAPLAN standardised tests of literacy and numeracy were primarily influenced by genetics, with a much smaller influence from the environment (Grasby, Coventry, Byrne, Olson & Medland, 2016). In situations where children are adopted by families who are strangers, adoptive children have no genetic resemblance to their adoptive families. Consequently, adoption studies can allow a researcher to study whether certain behaviours or traits of adoptive children are more similar to those of their biological parents (indicating a stronger genetic influence) or to those of their adoptive families (indicating a stronger environmental influence).

monozygotic (MZ) twins
twins who developed from a single ova and sperm, and who therefore have exactly the same genotype; also called identical twins

dizygotic (DZ) twins
twins who result when two ova are released by a female instead of one, and both ova are fertilised by different sperm; also called fraternal twins

heritability
statistical estimate of the extent to which genes are responsible for the differences among people within a specific population, with values ranging from 0 to 1.00

By comparing these different groups, behaviour geneticists are able to calculate a statistic called **heritability**. Heritability is an estimate of the extent to which genes are responsible for the differences among people within a specific population. The value of the heritability estimate ranges from 0 to 1.00. The higher the heritability, the more the characteristic is believed to be influenced by genetics.

Behaviour genetics has flourished in the past two decades, and heritability estimates have been calculated for a wide range of characteristics. For intelligence, heritability estimates for children and adolescents have been found to be about 0.50, meaning that about half the variation in their IQ scores has been attributed to genetic influences (Turkheimer, Harden, D'Onofrio & Gottesman, 2009). With regard to personality characteristics, heritability estimates range from 0.40 to 0.50 for a wide array of characteristics, such as sociability, activity level and even religiosity (Bouchard & McGue, 2003; Vukasović & Bratko, 2015).

Heritability estimates have been criticised for giving a misleading impression of the influence of genetics on development (Collins, Maccoby, Steinberg, Hetherington & Bornstein, 2000; Lerner, 2015). According to the critics, to state that a trait is heritable implies that we know with precision how much genes contribute to its development—but this is not so. Heritability estimates are simply estimates based on comparisons of people with different amounts of genetic material in common; they are not direct measures of the activity of genes. Heritability estimates are a measure not just of genetic influence but of *how much the environment allows the genes to be expressed*. In other words, heritability estimates measure phenotype rather than genotype.

This can be seen in the studies finding that heritability of intelligence increases from childhood to adulthood (Franić et al., 2015 ; McGue & Christensen, 2002). Obviously, genes do not change during this time, but the environment changes to allow greater expression of genetic potentials, as children grow into adolescence and become increasingly able to choose their own environments (e.g. whom they will have as friends). Other studies find that heritability of intelligence is higher in middle-class families than in poor families (McCartney & Berry, 2009; Turkheimer et al., 2009). This is not because middle-class families have different kinds of genes than poor families do, but because the greater economic resources of middle-class families make it more likely that children's genotypic potential for intelligence will be expressed in their phenotype.

concordance rate
degree of similarity in phenotype among pairs of family members, expressed as a percentage

Another statistic of genetic influence used in behaviour genetics is **concordance rate**. This is a percentage that indicates the degree of similarity in phenotype among pairs of family

members. Concordance rates range from 0 to 100%. The higher the concordance rate, the more similar the two people are.

In many studies, comparisons of concordance rates are made between MZ and DZ twins. When concordance rates are higher among MZ twins than among DZ twins, this indicates that the basis for the trait is partly genetic. For example, concordance rates for schizophrenia, a severe mental disorder involving hallucinations and disordered patterns of thinking and behaviour, are 50% for MZ twins and 18% for DZ twins (Insel, 2010). This means that when one MZ twin has schizophrenia, 50% of the time the other twin has schizophrenia as well. For DZ twins, when one twin has schizophrenia, the other twin has the disorder only 18% of the time. Adoption studies also sometimes use this statistic, comparing concordance rates between parents and adopted children, parents and biological children, and adoptive or biological siblings.

Gene–environment interactions: epigenesis and reaction ranges

Studies of heritability show not only that genes influence development but also that the environment influences how genes are expressed. A related idea is **epigenesis**, which means that development results from the bidirectional interactions between genotype and environment (Gottlieb, 2004; Gottlieb & Lickliter, 2007; Naumova & Taketo, 2016). According to epigenetic theory, the expression of genes is affected by and responds constantly to environmental influences. Development is influenced by genes but is not purely determined by them (Beach, Brody, Barton & Philibert, 2016). Experience may result in some genes getting expressed and others not.

Here is an example of epigenesis. Girls normally begin menstruating around age 11–16, towards the lower end of this range under healthy conditions and towards the higher end when nutrition is insufficient or the girl is suffering from medical problems (Neberich, Penke, Lenhart & Asendorph, 2010). Clearly, it is part of the human-female genotype for menstruation to be initiated somewhere in this age range, with the timing influenced by environmental conditions. Furthermore, when girls' environmental conditions change, their menstrual patterns may also change. Girls who experience severe weight loss often stop menstruating (Roberto, Steinglass, Mayer, Attia & Walsh, 2008). If their nutritional intake improves, they begin menstruating again. This demonstrates a continuous interaction between genotype and environment, with menstruation being 'turned on' genetically as part of puberty but 'turned off' if environmental conditions are dire, then turned on again once the nutritional environment improves.

As this example illustrates, often when genes influence human development it is by establishing boundaries for environmental influences rather than specifying a precise characteristic. In other words, genes establish a **reaction range** of potential expression, and environment determines where a person's phenotype will fall within that range (McCartney & Berry, 2009). To take another example, height is known to be influenced by genes. You can probably tell this just by looking at your own height in relation to other members of your family. However, the genes for height simply establish the reaction range's upper and lower boundaries, and where a person's actual height ends up—the phenotype—is determined by environmental influences such as nutrition and disease.

epigenesis
in development, the continuous bidirectional interactions between genes and environment

reaction range
range of possible developmental paths established by genes; environment determines where development takes place within that range

LO 2.4

Describe how the concept of epigenesis frames gene–environment interactions and connect epigenesis to the concept of reaction range.

Genes establish a reaction range for height, and environment determines where a person's height falls within that range. Shown here are sisters from the Hamer tribe in Ethiopia, a tribe whose members are known for being exceptionally tall.

dbtravel/dbimages/Alamy Stock Photo

Maternal stress during pregnancy can have negative effects on fetal development, including cognitive development.

Antonio Guillem/Shutterstock

Evidence for this is clear from the pattern of changes in height in societies around the world over the past century. In most Western countries, average height rose steadily in the first half of the 20th century as nutrition and health care improved (Freedman, Khan, Serdula, Ogden & Dietz, 2006). The genes of their populations could not have changed in just a generation or two; instead, the improving environment allowed them to reach a higher point in their genetic reaction range for height. In other countries, such as China and South Korea, improvements in nutrition and health care came later, in the second half of the 20th century, so increases in height in those countries have taken place only recently (Wang, Wang, Kong, Zhang & Zeng, 2010). However, people are unlikely ever to grow to be 3 metres tall. In recent decades in Western countries there has been little change in average height, indicating that the populations of these countries have reached the upper boundary of their reaction range for height.

Recently, scientists have explored the connections between stress and epigenetic effects. Stress during pregnancy can have negative effects on fetal development, including the development of the nervous system (Benoit, Rakic & Frick, 2015). Cognitive development of offspring may be affected by maternal stress during pregnancy (Dias, Maddox, Klengel & Ressler, 2015; Kleefstra, Schenck, Kramer & van Bokhoven, 2014). Epigenetic effects also occur after birth. Early life stress is associated with genetic changes that may be precursors to psychiatric disorders later in life (Boku et al., 2015). This evidence provides good motivation to help pregnant women and young children reduce their stress and develop coping mechanisms for stress.

LO 2.5

Explain how the theory of genotype → environment effects casts new light on the old nature–nurture debate.

The theory of genotype → environment effects

One influential theory of behaviour genetics is the **theory of genotype → environment effects** proposed by Sandra Scarr and Kathleen McCartney (Plomin, 2009; Plomin, DeFries, Knopic & Neiderhiser, 2013; Scarr, 1993; Scarr & McCartney, 1983). According to this theory, both genotype and environment make essential contributions to human development. However, the relative strengths of genetics and the environment are difficult to unravel because our genes actually influence the kind of environment we experience. That is the reason for the arrow in the term *genotype → environment effects*. Based on our genotypes, we *create our own environments*, to a considerable extent.

theory of genotype → environment effects
theory proposing that genes influence the kind of environment we experience

The three forms of genotype → environment effects

These genotype → environment effects take three forms: passive, evocative and active.

1 **Passive genotype → environment effects** occur in biological families because *parents provide both genes and environment for their children*. This may seem obvious, but it has profound implications for how we think about development. Take this father–daughter example. Dad has been good at drawing things ever since he was a boy, and now he makes a living as a graphic artist. One of the first birthday presents he gives to his little girl is a set of crayons and coloured pencils for drawing. As she grows up, he also teaches her a number of drawing skills as she seems ready to learn them. She goes to university and studies architecture, then goes on to become an architect. It is easy

passive genotype → environment effects
in the theory of genotype → environment effects, the type that results from the fact that, in a biological family, parents provide both genes and environment to their children

to see how she became so good at drawing, given an environment that stimulated her drawing abilities so much—right?

Not so fast. It is true that Dad provided her with a stimulating environment, but he also provided her with half her genes. If there are any genes that contribute to drawing ability—such as genes for spatial reasoning and fine motor coordination—she may well have received those from Dad, too. The point is that in a biological family, it is difficult to separate genetic influences from environmental influences because *parents provide both*, and they are likely to provide an environment that reinforces the tendencies they have provided to their children through their genes.

When parents and children are similar, is the similarity as a result of genetics or environment?

Sergey Novikov/Shutterstock

So, you should be sceptical when you read studies that claim parents' behaviour is the cause of the characteristics of their biological children. Remember from Chapter 1: correlation does not imply causation! Just because there is a *correlation* between the behaviour of parents and the characteristics of their children does not mean the parents' behaviour *caused* the children to have those characteristics. Maybe causation was involved, but in biological families it is difficult to tell. One good way to unravel this tangle is through adoption studies. These studies avoid the problem of passive genotype → environment effects because one set of parents provided the children's genes but a different set of parents provided the environment. An extraordinary case of adoption is presented in the *Research focus: Twin studies: the story of Oskar and Jack* feature.

2 **Evocative genotype → environment effects** occur when a person's inherited characteristics evoke responses from others in their environment. If you had a son who started reading at age 3 and seemed to love it, you might buy him more books. If you had a daughter who could sink jump shots at age 12, you might arrange to send her to basketball camp. Did you ever baby-sit or work in a setting where there were many children? If so, you probably found that children differ in how sociable, cooperative and obedient they are. In turn, you may have found that you responded differently to them, depending on their characteristics. This is what is meant by evocative genotype → environment effects—with the crucial addition of the assumption that characteristics such as reading ability, athletic ability and sociability are at least partly based in genetics.

evocative genotype → environment effects
in the theory of genotype → environment effects, the type that results when a person's inherited characteristics evoke responses from others in the environment

3 **Active genotype → environment effects** occur when people seek out environments that correspond to their genotypic characteristics, a process called *niche-picking*. The child who can run faster than her peers may be motivated to try out for a sports team; the adolescent with an ear for music may ask for piano lessons; the emerging adult for whom reading has always been slow and difficult may prefer to begin working full time after high school rather than going to university; in young adulthood a highly sociable person may seek a career that involves being around other people all day. The idea here is that people are drawn to environments that match their inherited abilities.

active genotype → environment effects
in the theory of genotype → environment effects, the type that results when people seek out environments that correspond to their genotypic characteristics

RESEARCH FOCUS

Twin studies: the story of Oskar and Jack

The interplay between genes and the environment is one of the most important, complex and fascinating topics in the study of human development. One approach that has been helpful in unravelling these interactions is twin studies, especially research on twins separated early in life and raised in different environments. Studies of twins reared apart provide a good example of a natural experiment, which is something that occurs without the intervention of a researcher but can provide valuable scientific information.

The Minnesota Study of Twins Reared Apart, led by Thomas J. Bouchard Jr of the University of Minnesota, has been studying separated twins since 1979, and the results have been groundbreaking and sometimes astounding.

Among the most remarkable cases in the Minnesota study is the story of identical twins, Oskar and Jack. They were born in Trinidad in 1933, but within 6 months of their birth, their parents split up.

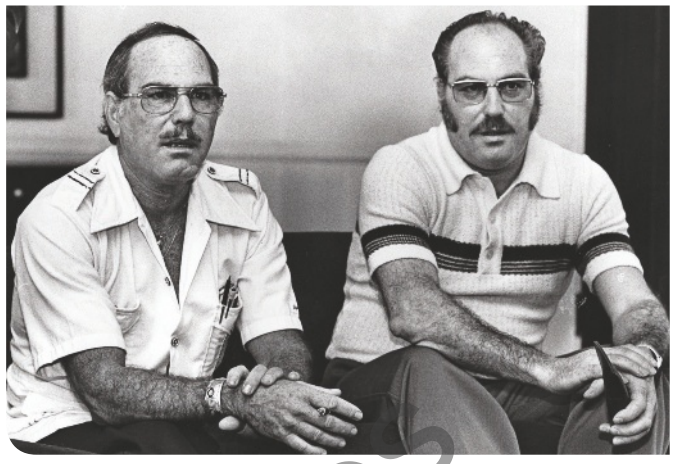
Oskar left for Germany with his Catholic mother, while Jack remained in Trinidad in the care of his Jewish father. Thus, unlike most separated twins, who at least remain within the same culture and country, Oskar and Jack grew up with the same genotype but with different cultures, different countries and different religions.

Furthermore, Oskar migrated with his mother to Germany in 1933, the year the Nazis rose to power. And Jack was raised as a Jew, at a time when Jews were targeted for extermination by the Nazis.

In some ways, the twins' childhood family environments were similar—as in similarly miserable. Oskar's mother soon moved to Italy and left him in Germany in the care of his grandmother, who was stern and harsh. Jack's father alternated between ignoring him and beating him. Despite these similarities, their cultures were about as different as could be. Oskar was an enthusiastic member of the Hitler Youth, and he learned to despise Jews and to keep his own half-Jewish background hidden. Jack was raised as a Jew and at 16 was sent by his father to Israel to join the navy, where he met and married an American Jew. At age 21 he and his wife moved to the United States.

What were the results of this extraordinary natural experiment in the two men's adult development? The extensive data collected by the Minnesota team, which included a week of tests and interviews with the men as well as interviews with their family members and others close to them, indicated that they had highly similar adult personalities.

Both were described by themselves and others as being short-tempered, demanding and absent-minded. In addition, they shared a remarkable range of unusual, quirky personal habits. Both read books from back to front, sneezed loudly in elevators, liked to wear rubber bands on their wrists, and wrapped tape around pens and pencils to get a better grip.



Robert Lachman/Los Angeles Times

However, their cultural identities and worldviews were as far apart as one might imagine, given the vastly different cultures they grew up in. Oskar repented his membership in the Hitler Youth as an adult and lamented the Holocaust that had taken millions of Jewish lives under the Nazis, but he considered himself very German, and he and Jack disagreed vehemently over the responsibility and justification for bombings and other acts of war conducted during World War II.

Thus, despite all their similarities in personality, because of their different cultural environments they ultimately had very different identities—starkly separate understandings of who they were and how they fitted into the world around them. As Oskar told Jack when they met again in adulthood, 'If we had been switched, I would have been the Jew and you would have been the Nazi.'

Review questions

1 Studies of twins raised apart provide a good example of _____.

- a reliability but not validity
- b validity but not reliability
- c experimental research
- d a natural experiment

2 Which of the following is NOT one of the ways in which Oskar and Jack were similar?

- a Both were absent-minded.
- b Both were short-tempered.
- c Both had a strong Jewish faith.
- d Both read books from back to front.

Genotype → environment effects over time

The three types of genotype → environment effects operate throughout childhood, adolescence and adulthood, but their relative balance changes over time (Plomin et al., 2013; Scarr, 1993). In childhood, passive genotype → environment effects are especially pronounced, and active genotype → environment effects are relatively weak. This is because the younger a child is, the more control parents have over the daily environment the child experiences and the less autonomy the child has to seek out environmental influences outside the family.

However, the balance changes as children move through adolescence and into adulthood (Plomin, 2009). Parental control diminishes, so passive genotype → environment effects also diminish. Autonomy increases, so active genotype → environment effects also increase. In adulthood, passive genotype → environment effects fade entirely (except in cultures where people continue to live with their parents even in adulthood), and active genotype → environment effects move to the forefront. Evocative genotype → environment effects remain relatively stable from childhood through adulthood.

CRITICAL-THINKING QUESTION

Think of one of your abilities and describe how the various types of genotype → environment effects may have been involved in your development of that ability.

GENETIC INFLUENCES ON DEVELOPMENT: GENES AND INDIVIDUAL DEVELOPMENT

LO 2.6

Outline the process of meiosis in the formation of reproductive cells.

When does individual human development begin? The answer may surprise you. The process of forming a new human being actually begins long before sperm and egg are joined. Sperm and eggs themselves go through a process of development. In this section, we look at the genetic basis of prenatal development, beginning with sperm and egg formation.

Sperm and egg formation

Most cells in the human body contain 46 chromosomes that reproduce by the process of **mitosis**, in which the chromosomes duplicate themselves and the cell divides to become two cells, each containing the same number of chromosomes as the original cell (Pankow, 2008). The only cells in the human body that do not contain 46 chromosomes are the reproductive cells or **gametes**: the sperm in the male and the egg or **ovum** (plural, *ova*) in the female. Gametes form in the testes of the male and the ovaries of the female through a process that is a variation of mitosis called *meiosis* (see Figure 2.4). In **meiosis**, cells that begin with 23 pairs of chromosomes first split into 46 single chromosomes, then replicate themselves and split into two cells, each with 23 pairs of chromosomes like the original cell. So far the process is just like mitosis. But then the chromosome pairs separate into single chromosomes and split again, this time into gametes that have 23 unpaired chromosomes instead of the original 46. So, at the end of the process of meiosis, from the original cell in the testes or ovaries, four new cells have been created, each with 23 chromosomes.

There are some important sex differences in the process of meiosis (Jones & Lopez, 2014). In males, meiosis is completed before sperm are released, but in females, the final stage of meiosis only takes place when and if the ovum is fertilised by a sperm (more on this shortly). Also, in males, the outcome of meiosis is four viable sperm, whereas in

mitosis
process of cell replication in which the chromosomes duplicate themselves and the cell divides into two cells, each with the same number of chromosomes as the original cell

gametes
cells, distinctive to each sex, that are involved in reproduction (egg cells in the ovaries of the female and sperm in the testes of the male)

ovum
mature egg that develops in ovaries, about every 28 days in human females

meiosis
process by which gametes are generated, through separation and duplication of chromosome pairs, culminating in the creation of four new gametes from the original cell, each with half the number of chromosomes of the original cell

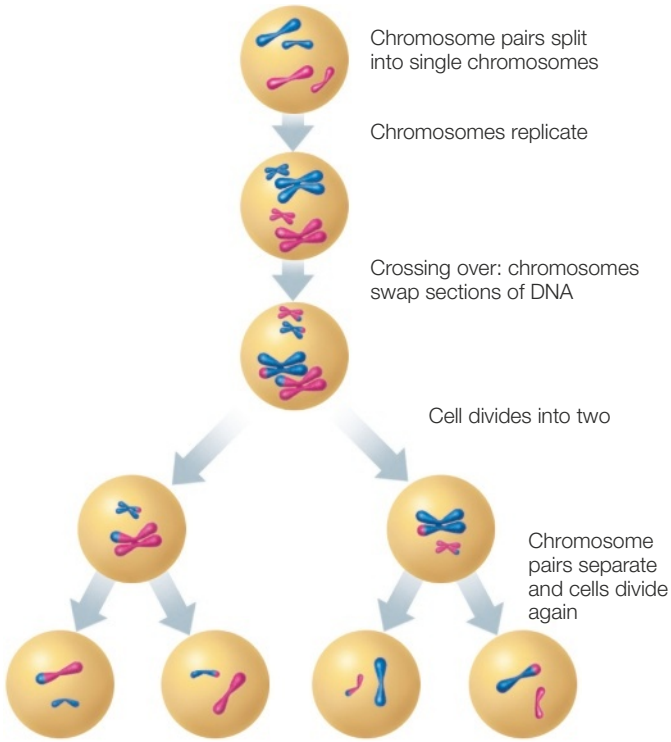


FIGURE 2.4 The creation of gametes through meiosis

How does meiosis differ from mitosis?

females, meiosis produces only one viable ovum along with three *polar bodies* that are not functional. The ovum hoards for itself a large quantity of **cytoplasm**, the fluid that will be the main source of nutrients in the early days after conception, whereas the polar bodies are left with little.

Did you ever think about why you are different from your brothers or sisters, even though both you and your sibling(s) have 23 chromosomes each from your mother and father? Even parents of fraternal twins are constantly amazed at how different the twins are. Here is the explanation for sibling diversity. Something fascinating and remarkable happens at the outset of the process of meiosis. After the chromosomes first split and replicate but before the cell divides, pieces of genetic material are exchanged between the alleles in each pair, a process called **crossing over** (refer again to Figure 2.4). Crossing over mixes the combinations of genes in the chromosomes, so that genetic material that originated from the mother and father (your grandparents) is rearranged in a virtually infinite number of ways (Pankow, 2008). Your parents could have had dozens, hundreds or even millions of children together (hypothetically!), and none of them would be exactly like you genetically (unless you have an identical twin).

cytoplasm
in an ovum, fluid that provides nutrients for the first 2 weeks of growth if the ovum is fertilised, until the fertilised ovum reaches the uterus and begins drawing nutrients from the mother

crossing over
at the outset of meiosis, the exchange of genetic material between paired chromosomes

Here is another interesting fact about the production of gametes. Upon reaching puberty, males begin producing millions of sperm each day. There are 100–300 million sperm in the typical male ejaculation (Johnson, 2016). In contrast, females have already produced all the ova they will ever have *while they are still in their own mothers' womb*. Because crossing over begins when ova are created, this means that the development of a unique genotype for each individual begins before the individual's mother is born!

Females are born with about 1 million ova, but this number diminishes to about 40 000 by the time they reach puberty, and about 400 of these will mature during a woman's childbearing years (Johnson, 2016; Moore, Persaud & Torchia, 2015). Most women cease ovulating at some time in their 40s or 50s, but men produce sperm throughout their adult lives (although the quantity and quality of the sperm may decline with age) (Finn, 2001).

LO 2.7

Describe the process of fertilisation and conception.

Conception

When sexual intercourse takes place between a man and a woman, many millions of sperm from the man begin making their way through the woman's reproductive organs—first into the vagina, then through the cervix, through the uterus and up the fallopian tubes towards the ovaries. Hundreds of millions of sperm may seem like more than enough, but keep in mind that sperm are composed of a single cell, not much more than 23 chromosomes and a tail, so they are not exactly skilled at navigation. The distance from the vagina to the ovaries is vast for such a small object as a sperm. Furthermore, the woman's body responds to sperm as a foreign substance and begins killing them off immediately. Usually only a few hundred sperm make it up the fallopian tubes to where fertilisation can take place (Jones & Lopez, 2014).

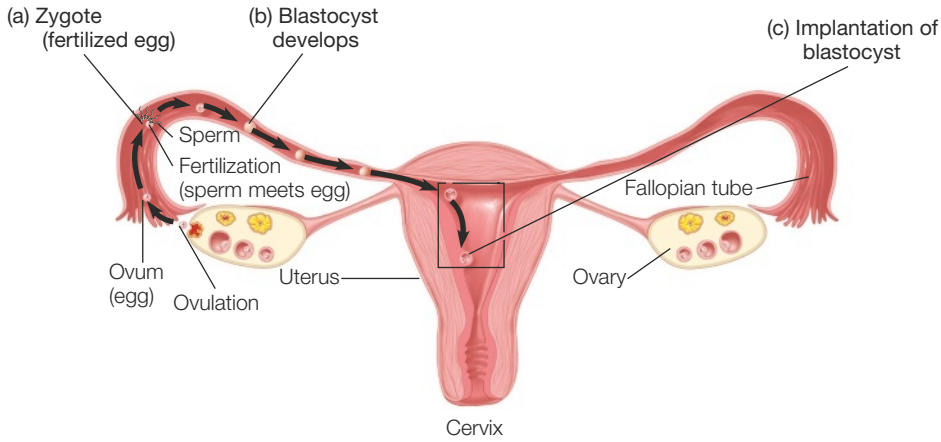


FIGURE 2.5 Ovulation process

The two ovaries alternate ovulation in each monthly menstrual cycle.

Within the woman, there are two ovaries that release an ovum in alternating months. During the early part of the woman's menstrual cycle, the ovum is maturing into a **follicle**. The follicle consists of the ovum plus other cells that surround it and provide nutrients. About 14 days into a woman's cycle, the mature follicle bursts and *ovulation* takes place as the ovum is released into the fallopian tube (see Figure 2.5). The ovum is 2000 times larger than a sperm because it contains so much cytoplasm (Johnson, 2016). The cytoplasm will provide nutrients for the first 2 weeks of growth if the ovum is fertilised, until the fertilised ovum reaches the uterus and begins drawing nutrients from the mother.

follicle
during the female reproductive cycle, the ovum plus other cells that surround the ovum and provide nutrients

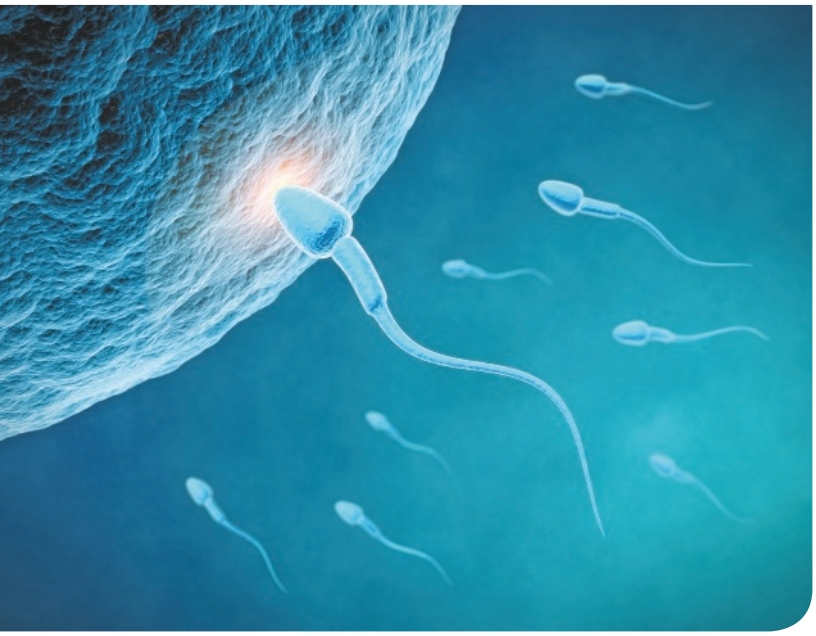
It is only during the first 24 hours after the ovum enters the fallopian tube that fertilisation can occur. It takes sperm from a few hours to a whole day to travel up the fallopian tubes, so fertilisation is most likely to take place if intercourse occurs on the day of ovulation or on the two previous days (Wilcox, Weinberg & Baird, 1995). Sperm can live for up to 5 days after entering the woman's body, but most do not last more than 2 days (Johnson, 2016).

When sperm reach the ovum they begin to penetrate the surface of the cell, aided by a chemical on the tip of the sperm that dissolves the ovum's membrane. Once the sperm penetrates the ovum's membrane, the head of the sperm detaches from the tail and continues towards the nucleus of the cell while the tail remains outside. The moment a sperm breaks through, a chemical change takes place in the membrane of the ovum that prevents any other sperm from getting in.

When the sperm head reaches the nucleus of the ovum, the final phase of meiosis is triggered in the ovum (Johnson, 2016). Fertilisation takes place as the 23 chromosomes from the ovum pair up with the 23 chromosomes from the sperm, and a new cell, the **zygote**, is formed from the two gametes. The zygote's 46 paired chromosomes constitute the new organism's unique genotype, set once and for all at the moment of conception.

zygote
following fertilisation, the new cell formed from the union of sperm and ovum

Although this is how conception usually takes place, there are occasional variations. One of the most common variations is that two ova are released by the woman instead of one, and both are fertilised by sperm, resulting in DZ twins (recall that DZ stands for *dizygotic*—two zygotes). This occurs overall about once in every 60 births, and this rate is consistent in Australia and New Zealand (Australian Bureau of Statistics [ABS], 2017b; Pison, Monden & Smits, 2015). However, there are substantial ethnic variations in the birth rate of DZ twins, ranging from 1 in every 25 births in Nigeria to 1 in every 700 births in Japan (Gall, 1996).



Fertilisation can take place only in the first 24 hours after the ovum enters the fallopian tube.

Sashkin/Shutterstock

In general, Asians have the lowest rates of DZ twins, and Africans the highest (Mange & Mange, 1998; Smits & Monden, 2011). In addition to ethnic background, some of the factors that increase the likelihood of DZ twins are a family history of twins, age (older women are more likely to release two eggs at once) and nutrition (women with healthy diets are more likely to have DZ twins) (Bortolus et al., 1999). Today, another common cause of DZ twins is infertility treatments, which we discuss in more detail shortly.

Twins can also result when a zygote that has just begun the process of cell division splits into two separate clusters of cells, creating MZ twins (recall that MZ stands for *monozygotic*—one zygote). MZ twins are less common than DZ twins, occurring about 1 in every 285 births (Zach, Pramanik & Ford, 2001). In contrast to DZ twins, MZ twins are not more common in some ethnic groups than in others. The birth rates for MZ twins are approximately the same all around the world. Also unlike DZ twins, MZ twins do not run in families and are not predicted by maternal age or nutrition.

LO 2.8

List the major causes of and treatments for infertility, and describe how infertility is viewed in different cultures.

Infertility

Most women of reproductive age (roughly ages 15–40) who have sexual intercourse on a regular basis will become pregnant within a year or two. However, for some couples, becoming pregnant is more problematic. **Infertility** is defined as the inability to attain pregnancy after at least a year of regular sexual intercourse without contraception. One in six couples in Australia is considered to have fertility problems based on this definition, and this represents an increase compared to previous decades (Department of Health and Ageing, 2010). A worldwide assessment of infertility between 1990 and 2010 also found that rates had stayed consistent at about 9–13% (Mascarenhas, Flaxman, Boerma, Vanderpoels & Stevens, 2012).

infertility
inability to attain pregnancy after at least a year of regular sexual intercourse

Sources of infertility

About half the time, the source of infertility is in the male reproductive system, and about half the time it is in the female reproductive system (Jones & Lopez, 2014). Among men, there are three main factors in infertility (Jequier, 2011): (1) too few sperm may be produced; (2) the quality of the sperm may be poor, due to disease or defects in the sperm manufacturing process in the testicles; or (3) the sperm may be low in *motility* (movement) and therefore unable to make it all the way up the fallopian tubes. These problems may be genetic or they may be caused by behaviour such as drug abuse, alcohol abuse or cigarette smoking. Or, they may simply be as a result of age—it takes three times longer for men over 40 to impregnate a partner than it does for men under 25, because the quantity and quality of sperm production decreases with age (Patel et al., 2015).

Among women, infertility is most often caused by problems in ovulation (National Women's Health Information Center, 2011). Inability to ovulate can be caused by disease, or it can be due to drug abuse, alcohol abuse or cigarette smoking, or to being extremely underweight or overweight. However, age is the most common cause of inability to ovulate (Maheshwari, Hamilton & Bhattacharya, 2008). Fertility decreases for women throughout their 20s and

30s but drops rapidly after age 40, when women become more likely to have menstrual cycles with no ovulation at all (see Figure 2.6).

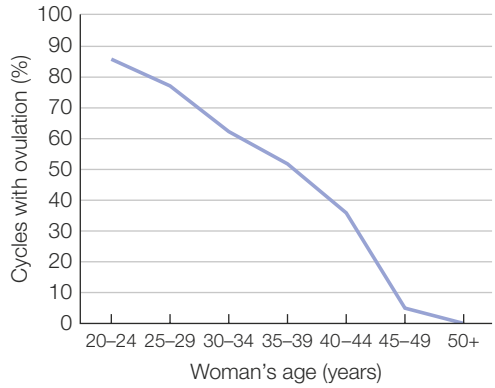


FIGURE 2.6 Fertility and maternal age

Why does fertility decline after the mid-20s?

Infertility treatments

We now know that men and women contribute equally to infertility. However, this knowledge is recent, coming in only about the past 50 years. For most of human history in most cultures, infertility has been regarded almost exclusively as a female problem, and women suffering from it were described not as infertile but as 'barren' (Marsh & Ronner, 1996). In the West, for more than 2000 years, from about the 4th century BCE to the 1800s, the reigning explanations for infertility were based on incorrect theories of how conception occurred. Thus, treatments were ineffective.

During the course of the 20th century, treatments for infertility became more scientifically based and technologically advanced. Today, there are a variety of approaches. These methods are used by infertile couples as well as by gay and lesbian couples and by single women. A variety of related methods for overcoming infertility are grouped under the term **assisted reproductive technologies (ART)**, including artificial insemination, fertility drugs and in vitro fertilisation (IVF). ART methods are used in response to a wide variety of infertility problems in either the male or female reproductive system, or both (Advisory Committee on Assisted Reproductive Technology, 2015). In 2014, 14 238 babies in Australia and New Zealand were born following ART treatments (Harris et al., 2016), and approximately 4.4% of all pregnant Australian mothers received some form of ART in 2013 (Australian Institute of Health and Welfare [AIHW], 2015a).

assisted reproductive technologies (ART)
methods for overcoming infertility that include artificial insemination, fertility drugs and IVF

The oldest effective treatment for infertility is **intrauterine insemination (IUI)**, which involves injecting the man's sperm directly into the woman's uterus, timed to coincide with her ovulation (Schoolcraft, 2010). This treatment was first developed in the 19th century when physicians believed the primary cause of infertility was a too-tight cervix (the opening between the vagina and the uterus). IUI most often occurs as *donor insemination*, in which a man other than the woman's husband or partner provides the sperm. Most often this approach is because of problems in the husband's or partner's sperm production, but increasingly this procedure is chosen by lesbian couples or single women who wish to have a child (Monseur, Franasiak, Sun, Scott & Kaser, 2017). Prior to IUI, the sperm are first 'washed' to remove the rest of the semen and enhance the likelihood of success (with 'success' being defined as a live birth). IUI is the simplest and least expensive reproductive technology, and has a success rate of about 10–20% per trial (Thijssen et al., 2017). Over several trials, success rates vary sharply with age, from nearly 40% for women under age 25 to 15% for women aged 42–43 (Schorsch et al., 2013).

intrauterine insemination (IUI)
procedure of injecting sperm directly into the uterus

If the primary problem is that the woman cannot ovulate properly, the most common approach is to stimulate ovulation through fertility drugs. The drugs mimic the activity of the hormones that normally provoke ovulation. Usually fertility drugs stimulate both the quality and the quantity of follicles in each cycle. More than half of the women who take the drugs become pregnant within six cycles (Schoolcraft, 2010).

Fertility drugs work for many women, but they also carry serious risks, including blood clots, kidney damage and damage to the ovaries, so women using the drugs should be closely monitored by their doctor (Schram, 2016). The purpose of the drugs is to stimulate the development of follicles in the ovaries, but often more than one follicle develops, resulting in the release of two, three or more ova. Consequently, use of fertility drugs produces high rates of multiple births, about 10–25% depending on the

Multiple births often receive extensive media attention, but the consequences of such births are often tragic, with higher risks of miscarriages, premature birth and serious developmental difficulties.

Yuri Mashkov/ITAR-TASS News Agency/Alamy Stock Photo

in vitro fertilisation (IVF)
form of infertility treatment that involves using drugs to stimulate the growth of multiple follicles in the ovaries, removing the follicles and combining them with sperm, then transferring the most promising zygotes to the uterus

drug (Diamond et al., 2015; Schoolcraft, 2010). Usually this means twins, but there is the possibility of triplets or more. You may have read magazine stories or seen television shows about multiple births of six, seven or eight infants and how adorable they are, but the consequences of multiple births are often tragic. The more babies conceived at once, the higher the risk for miscarriages, premature birth and serious developmental difficulties.

If fertility drugs are unsuccessful in achieving pregnancy, the next step in the ART method is **in vitro fertilisation (IVF)**. In IVF, after fertility drugs are used to stimulate the growth of numerous follicles in the woman's ovaries, the ripe ova are then removed and combined with the man's sperm so that fertilisation will take place. After a few days, it is possible to tell which of the zygotes have developed and which have not, so the most promising two or three are placed into the woman's uterus in the hope that one will continue to develop. IVF success rates have steadily improved in recent years, and are currently about 50% per attempt for women under age 35 (Society for Assisted Reproductive Technology [SART], 2017). However, the success rate declines with age to 24% for women aged 38–40 and to just 4% for women aged 42 and over.

Few people in developing countries have access to reproductive technologies such as fertility drugs and IVF. Women may try herbal remedies provided by a midwife. Others may seek supernatural remedies (Leonard, 2002).

Infertility worldwide

Across cultures, most people wish to have children, and infertility is experienced as a source of frustration and distress (van Balen & Inhorn, 2002). However, there are definite cultural differences in how seriously infertility is viewed and how it is framed socially. In the individualistic West, infertile couples often experience a sense of sadness and loss. In one Swedish study, couples seeking infertility treatments felt frustration over missing out on a major focus of life, and they experienced a negative effect on their sexual relationship (Hjelmstedt et al., 1999). Other studies have found that infertility often creates strains in the marital relationship; in the long run, however, about half of couples report that the experience of infertility made their relationship closer and stronger (Schmidt, Holstein, Christensen & Boivin, 2005).

Outside the West, cultures tend to be more collectivistic, and the social consequences of infertility are even more profound. Infertility is often deeply stigmatised. This is especially true for women, who are usually blamed for the problem and for whom motherhood is essential to their identity and their place within the social world (Inhorn & van Balen, 2002; Sembuya, 2010). In many cultures, infertility means much more than that the couple will miss out on the joys of raising a child. It may mean that there will be no one to continue the family tradition of remembering and worshipping the ancestors, a responsibility that often falls on the oldest son, especially in Asian and African cultures. It may also mean that the status of the wife is lowered in relation to her husband, her in-laws and the community because infertility is viewed more as being her failure than his. Even if a woman has a daughter, she may still be seen as inadequate if she fails to produce a son. This is misguided because, as discussed earlier in the chapter, biologically it is the father and not the mother who determines the sex of the child.

If infertility persists, it is viewed in many cultures as being grounds for the husband to divorce his wife or take another wife. For example, in Vietnam it is generally accepted that if a man's wife is infertile he will attempt to have a child with another 'wife', even though having more than one wife is illegal (Pashigian, 2002). In Cameroon, if a couple cannot conceive a child, the husband's family may encourage him to obtain a divorce and seek the return of the 'bridewealth' his family paid to the wife's family when they married (Feldman-Savelsberg, 2002).

SUMMARY GENETIC INFLUENCES ON DEVELOPMENT

LO 2.1 Distinguish between genotype and phenotype and identify the different forms of genetic inheritance.

There are 46 chromosomes in the human genome, organised into 23 pairs. The totality of an individual's genes is the genotype, and the person's actual characteristics are called the phenotype. Genotype and phenotype may be different as a result of dominant–recessive inheritance, incomplete dominance and environmental influences. Most human characteristics are polygenic, meaning that they are influenced by multiple genes rather than just one.

LO 2.2 Describe the sex chromosomes and identify what makes them different from other chromosomes.

The sex chromosomes determine whether the person will be male or female. In the female this pair is called XX; in the male, XY. Having only one X chromosome makes males more vulnerable than females to a variety of recessive disorders that are linked to the X chromosome.

LO 2.3 Explain how behaviour geneticists use heritability estimates and concordance rates in their research.

Heritability estimates indicate the degree to which a characteristic is believed to be influenced by genes within a specific population. Concordance rates indicate the degree of similarity between people with different amounts of their genes in common, for example MZ twins and DZ twins.

LO 2.4 Describe how the concept of epigenesis frames gene–environment interactions and connect epigenesis to the concept of reaction range.

Epigenesis is the concept that development results from bidirectional interactions between genotype and environment. The concept of reaction range also involves gene–environment interactions because it means that genes set a range for development, and environment determines where development falls within that range.

LO 2.5 Explain how the theory of genotype → environment effects casts new light on the old nature–nurture debate.

Rather than viewing nature and nurture as separate forces, this theory proposes that genes influence environments through three types of genotype → environment effects: passive (parents provide both genes and environment to their children); evocative (children evoke responses from those who care for them); and active (children seek out an environment that corresponds to their genotype). The three types of effects operate throughout the life span but their relative balance changes with time.

LO 2.6 Outline the process of meiosis in the formation of reproductive cells.

In meiosis, cells that begin with 23 pairs of chromosomes split and replicate repeatedly until they form four gametes, each with 23 individual chromosomes. In males, the outcome of meiosis is four viable sperm, whereas in females, meiosis produces only one viable ovum. Also, males produce millions of sperm daily beginning in puberty, whereas females produce all the eggs they will ever have while still in their mother's womb.

LO 2.7 Describe the process of fertilisation and conception.

About 14 days into a woman's menstrual cycle an ovum is released into the fallopian tube. For the next 24 hours, fertilisation can occur in which the 23 chromosomes from the ovum pair up with the 23 chromosomes from the sperm, and a new cell, the zygote, is formed from the two gametes. The zygote's 46 paired chromosomes constitute the new organism's unique genotype, set at the moment of conception.

LO 2.8 List the major causes of and treatments for infertility, and describe how infertility is viewed in different cultures.

Male infertility may be caused by too few sperm, poor quality of sperm or low motility of sperm. Female infertility is most often caused by problems in ovulation. Infertility in both men and women is often because of age, but it can also be genetic or caused by behaviour such as drug abuse, alcohol abuse or cigarette smoking. Treatments for infertility are termed *assisted reproductive technologies* (ART) and include artificial insemination, fertility drugs and in vitro fertilisation (IVF).

In developed countries, infertility often results in frustration and sadness, and presents a challenge to the couple's relationship, although it may ultimately make the relationship stronger. In developing countries, the woman is usually blamed for the infertility, and her social status is damaged.

SECTION 2

PRENATAL DEVELOPMENT AND PRENATAL CARE

LEARNING OBJECTIVES

- **LO 2.9** Describe the structures that form during the germinal period.
- **LO 2.10** Outline the major milestones of the embryonic period.
- **LO 2.11** Describe the major milestones of the fetal period and identify when viability occurs.
- **LO 2.12** Compare and contrast prenatal care in traditional cultures and developed countries.
- **LO 2.13** Identify the major teratogens in developing countries and developed countries.

PRENATAL DEVELOPMENT AND PRENATAL CARE: PRENATAL DEVELOPMENT

When sperm and ovum unite to become a zygote, a remarkable process is set in motion. If all goes well, about 9 months later a fully formed human being will be born. Here, we look closely at this process, from conception to birth (summarised in Figure 2.7).

germinal period
first 2 weeks after conception

LO 2.9

Describe the structures that form during the germinal period.

The germinal period (first 2 weeks)

The first 2 weeks after fertilisation are called the **germinal period** (Jones & Lopez, 2014). This is the period when the zygote travels down the fallopian tubes to the uterus and implants in the uterine wall. As it travels, the zygote begins cell division and differentiation. The first cell

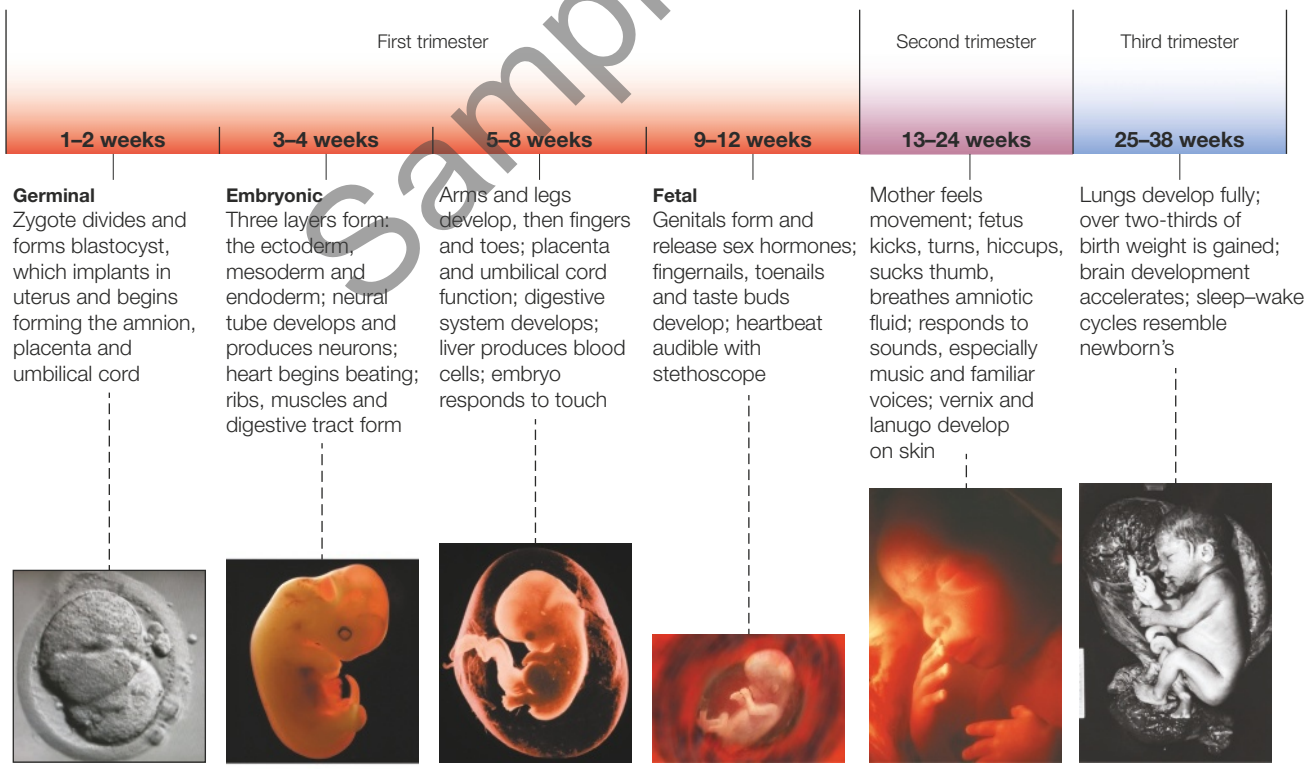
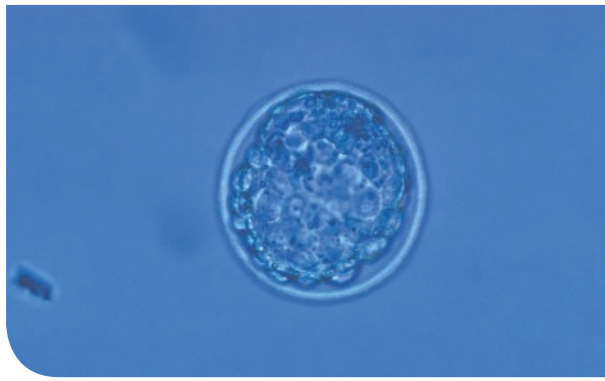


FIGURE 2.7 Milestones of prenatal development

BSIP/Newscom; Kage-Mikrofotografie/Alamy Stock Photo; Dr M.A. Ansary/Science Source; Steve Allen/Alamy Stock Photo; Neil Bromhall/Science Source; Photo Researchers Inc./Science Source

division does not occur until 30 hours after conception, but after that, cell division takes place at a faster rate. By 1 week following conception, there is a ball of about 100 cells known as a **blastocyst**. The blastocyst is divided into two layers. The outer layer of cells, called the **trophoblast**, will form the structures that provide protection and nourishment. The inner layer of cells, the **embryonic disk**, will become the embryo of the new organism.

During the second week after conception, implantation occurs as the blastocyst becomes firmly embedded into the lining of the uterus. Since the ovum was released from the ovary, the follicle from which it was released has been generating hormones that have caused the uterus to build up a bloody lining in preparation for receiving the blastocyst. Now the blastocyst is nourished by this blood.



Cell division begins about 30 hours after conception.

R. Rawlins PhD/Custom Medical Stock Photo/Newscom

blastocyst
ball of about 100 cells formed by about 1 week following conception

trophoblast
in the blastocyst, the outer layer of cells, which will go on to form structures that provide protection and nourishment to the embryo

embryonic disk
in the blastocyst, the inner layer of cells, which will go on to form the embryo

amnion
fluid-filled membrane that surrounds and protects the developing organism in the uterus

The trophoblast begins to differentiate into several structures during this second week. Part of it forms a membrane, the **amnion**, which surrounds the developing organism and fills with fluid, helping to keep a steady temperature for the organism and protect it against the friction of the mother's movements (Johnson, 2008). In-between the uterine wall and the embryonic disk, a round structure, the **placenta**, begins to develop. The placenta will allow nutrients to pass from the mother to the developing organism and permit wastes to be removed. It also acts as a gatekeeper, protecting the developing organism from bacteria and wastes in the mother's blood. In addition, it produces hormones that maintain the blood in the uterine lining and cause the mother's breasts to produce milk. An **umbilical cord** also begins to develop, connecting the placenta to the mother's uterus.

Implantation is the outcome of the germinal period if all goes well. However, it is estimated that more than half of blastocysts never implant successfully, usually as a result of chromosomal problems that have caused cell division to slow down or stop (Johnson, 2016). If implantation fails, the blastocyst will be eliminated from the woman's body along with the bloody uterine lining during her next menstrual period.

The embryonic period (weeks 3–8)

LO 2.10

Outline the major milestones of the embryonic period.

During the germinal period, the trophoblast differentiated faster than the embryonic disk, developing the structures to protect and nurture the organism during pregnancy. Now, differentiation occurs rapidly in the embryonic disk. Over the 6 weeks of the **embryonic period**, which covers 3–8 weeks' **gestation** (the time elapsed since conception), nearly all the major organ systems are formed (Fleming, 2006).

During the first week of the embryonic period—the third week after conception—the embryonic disk forms three layers. The outer layer, the **ectoderm**, will become the skin, hair, nails, sensory organs and nervous system. The middle layer, the **mesoderm**, will become the muscles, bones, reproductive system and circulatory system. The inner layer, the **endoderm**, will become the digestive system and the respiratory system.

The nervous system develops first and fastest (Johnson, 2016). By the end of week 3 (since conception), part of the ectoderm forms the **neural tube**, which will eventually become the spinal cord and brain. Once formed, the neural tube begins producing **neurons** (cells of the nervous system) in immense quantities, more than 250 000 per minute. In week 4, the shape of the head becomes apparent, and the eyes, nose, mouth and ears begin to form. The heart begins to beat during this week, and the ribs, muscles and digestive tract appear. By the end of week 4, the embryo is only 6 millimetres long but is already remarkably differentiated. Nevertheless, even an expert embryologist would have trouble at this point judging whether the embryo was to become a fish, a bird or a mammal.

placenta
in the uterus, the gatekeeper between mother and fetus, protecting the fetus from bacteria and wastes in the mother's blood, and producing hormones that maintain the blood in the uterine lining and cause the mother's breasts to produce milk

umbilical cord
structure connecting the placenta to the mother's uterus

embryonic period
weeks 3–8 of prenatal development

gestation
in prenatal development, the elapsed time since conception

ectoderm
in the embryonic period, the outer layer of cells, which will eventually become the skin, hair, nails, sensory organs and nervous system (brain and spinal cord)

mesoderm
in the embryonic period, the middle of the three cell layers, which will become the muscles, bones, reproductive system and circulatory system

During weeks 5–8, growth continues its rapid pace. Buds that will become the arms and legs appear in week 5, developing webbed fingers and toes that lose their webbing by week 8. The placenta and the umbilical cord become fully functional (Jones & Lopez, 2014). The digestive system develops, and the liver begins producing blood cells. The heart develops separate chambers. The top of the neural tube continues to develop into the brain, but the bottom of it looks like a tail in week 5, gradually shrinking to look more like a spinal cord by week 8.

By the end of week 8, the embryo is just 2.5 centimetres long and 1 gram in weight. Yet all the main body parts have formed, as have all of the main organs except the sex organs. Furthermore, the tiny embryo responds to touch, especially around its mouth, and it can move (Moore et al., 2015). Now the embryo looks distinctly human (Johnson, 2016).

The fetal period (week 9–birth)

LO 2.11

Describe the major milestones of the fetal period and identify when viability occurs.

endoderm
in the embryonic period, the inner layer of cells, which will become the digestive system and the respiratory system

neural tube
in the embryonic period, the part of the ectoderm that will become the spinal cord and brain

neuron
cell of the nervous system

fetal period
in prenatal development, the period from week 9 until birth

trimester
one of the three 3-month periods of prenatal development

vernix
at birth, babies are covered with this oily, cheese-like substance, which protects their skin from chapping in the uterus

During the **fetal period**, lasting from 9 weeks after conception until birth, the organs continue to develop, and there is tremendous growth in sheer size, from a weight of 1 gram and length of 2.5 centimetres at the beginning of the fetal period to an average (in developed countries) of 3.4 kilograms and 51 centimetres by birth.

By the end of the third month, the genitals have formed. After forming, the genitals release hormones that influence the rest of prenatal development, including brain organisation, body size and activity level, with male fetuses becoming on average somewhat larger and more active (Cameron, 2001). Also during the third month, fingernails, toenails and taste buds begin to develop. The heart has developed enough so that the heartbeat can now be heard through a stethoscope.

After 3 months, the typical fetus weighs about 85 grams and is about 7.6 centimetres long. Prenatal development is divided into three 3-month periods called **trimesters**, and the end of the third month marks the end of the first trimester.

During the second trimester, the fetus becomes active and begins to respond to its environment (Henrichs et al., 2010). By the end of the fourth month, the fetus's movements can be felt by the mother. Gradually over the course of the second trimester, the activity of the fetus becomes more diverse. By the end of the second trimester, it breathes amniotic fluid in and out; it kicks, turns and hiccups; it even sucks its thumb. It also responds to sounds, including voices and music, showing a preference (indicated by increased heart rate) for familiar voices, especially the voice of the mother. An oily white substance called **vernix** covers the skin, to protect it from chapping because of the amniotic fluid, and downy hair called *lanugo* helps the vernix stick to the skin. By birth, the fetus usually sheds its lanugo, although sometimes babies are born with lanugo still on, and then shed it in the early weeks of life.

By the end of the second trimester, 6 months after conception, the typical fetus is about 36 centimetres long and weighs about 900 grams. Although it seems well developed in many aspects of its behaviour, the fetus is still questionable in its *viability*, meaning its ability to survive outside of the uterus. A full-term fetus is defined as 38 weeks or more gestation. Babies born before 22 weeks rarely survive, even with the most advanced technological assistance. Survival rates vary depending on the country of birth and the corresponding availability of medical care. In high-income countries, 50% of babies born at 24 weeks survive, but in low-income countries, 50% of babies born at 32 weeks survive (WHO, 2012a). Globally the number of preterm births has been rising since 1990 in almost all countries. This trend could be due to improved data collection and dating of the pregnancies, but factors such as older maternal age and infertility treatments, especially

those related to multiple embryos, are also likely to be contributing to more preterm births (WHO, 2012a).

The main obstacle to viability at the beginning of the third trimester is the immaturity of the lungs. The lungs are the last major organ to become viable, and even a baby born in the seventh or early eighth month may need a respirator to breathe properly. Weight gain is also important. During the last trimester, the typical fetus gains over 2200 grams, and this additional weight helps it sustain life. Babies born weighing less than 2500 grams are at risk for a wide range of problems.

The brain is even less mature than the lungs in the third trimester, but its immaturity does not represent an obstacle to viability. Early brain immaturity was an evolutionary adaptation that enabled human beings to have an exceptionally large brain yet still fit through the birth canal. More than any other animal, humans are born with immature brains, which is why human babies are vulnerable and need parental care longer than other animals do. Nevertheless, more brain development occurs in the last 2 months of prenatal development than in any previous months. Neurons are created in vast numbers, up to 500 000 per minute, and the connections between them become increasingly elaborate (Gross, 2008).

By the third trimester, brain development has progressed to the point where, at 28 weeks, the sleep–wake cycles of the fetus are similar to those of a newborn infant. The fetus becomes increasingly aware of the external environment, especially in its ability to hear and remember sounds (James, 2010). In one study, mothers were asked to read Dr Seuss's *The Cat in the Hat* to their fetuses every day during the last 6 weeks of pregnancy (DeCasper & Spence, 1986). After birth, the babies showed a preference for a recording of their mother reading *The Cat in the Hat,* by sucking on a plastic nipple in order to turn the recording on. They sucked harder to hear *The Cat and the Hat* than they did for recordings of their mothers reading similar rhyming stories they had not heard before. Fetuses respond to their internal environment as well. When the mother is highly stressed, the fetus's heart beats faster and its body movements increase (DiPietro, Hilton, Hawkins, Costigan & Pressman, 2002).

PRENATAL DEVELOPMENT AND PRENATAL CARE: PRENATAL CARE

Because prenatal development carries risks for both mother and fetus, all cultures have developed customs and practices to try to promote a healthy outcome. First, we look at some of the practices of prenatal care in traditional cultures; we then look at the scientific approach to prenatal care that has developed recently.

Variations in prenatal care

LO 2.12

Compare and contrast prenatal care in traditional cultures and developed countries.

All cultures have a store of advice about what a woman should and should not do during pregnancy (Gottlieb & DeLoache, 2017). What kind of guidelines or advice have you heard? You might ask your mother, your grandmother and other mothers you know what advice they followed and where they obtained it.

Sometimes pregnancy advice seems practical and sensible. The practical advice reflects the collected wisdom that women pass down to each other over generations, based on their own experiences. Pregnant Aboriginal mothers in one anthropological study in the Northern Territory were mentored by other women and encouraged to continue to exercise (walking, digging, squatting) and to eat fresh bush food (such as yams, berries, goanna and crocodile) (Dunbar & Ford, 2011). Customs that seem peculiar to an outsider may arise because pregnancy is often perilous to both mother and fetus. Cultures sometimes

develop their prenatal customs out of the intense desire to ensure that pregnancy will proceed successfully, but without the scientific knowledge that would make such control possible.

Here are a few examples. Traditionally, a Māori ceremony called *whakatō tamariki* (planting the seed of a child) could be performed for a couple who were struggling to conceive (Mead, 2016). The belief is that the man provides the seed of life and the woman is the bed where life is nurtured. A number of incantations are associated with pregnancy to support the growth of the fetus. Māori believe the spirit is activated with the development of the eyes and that, soon after, the fetus is able to think. On the Indonesian island of Bali, 'hot' foods are to be avoided during pregnancy, including eggplant, mango and octopus (Diener, 2000). Also, a pregnant mother should not accept food from someone who is viewed as being spiritually impure, such as a menstruating woman or someone who has recently had a death in the family. Witches are believed to be especially attracted to the blood of a pregnant woman and her unborn child, so pregnant women are advised to obtain a magic charm and wear it on their belt or hang it on the gate of their yard, for protection (Diener, 2000).

Even in most developed countries, which have a long scientific tradition, not much was known about prenatal care from a scientific perspective until recent decades. As recently as the middle of the 20th century, women in some developed countries were being advised by their doctors to limit their weight gain during pregnancy to no more than 6.5 kilograms (Murkoff & Mazel, 2008). By now, scientific studies have shown that women who have a healthy weight before becoming pregnant should typically gain 11–16 kilograms during pregnancy, and women who gain less than 9 kilograms are at risk for having babies who are preterm and have low birth weight (Ehrenberg, Dierker, Milluzzi & Mercer, 2003).

There are also risks of gaining too much weight during pregnancy. These risks may affect both the mother and her child (Restall et al., 2014). The expectant mother who gains too much weight may experience hypertension, pre-eclampsia and diabetes (Chen, Keen, Rosander & Von Hofsten, 2010). A larger fetus may make for a more difficult delivery, and that poses a higher likelihood of caesarean delivery being required. There is also a greater risk of the child becoming overweight later in life (Restall et al., 2014).

In other areas, too, an extensive body of scientific knowledge has accumulated on prenatal care in recent decades. One key conclusion of this research is that pregnant women should receive regular evaluations from a skilled health care worker, beginning as soon as possible after conception, to monitor the health of mother and fetus and ensure that the pregnancy is proceeding well. Babies born to mothers who received no prenatal care are more likely to have a low birth weight or to die than babies born to mothers who receive prenatal care (Perinatal and Maternal Mortality Review Committee, 2016). Most women in developed countries have access to doctors, nurses or certified **midwives** who can provide good prenatal care. However, some poor women may not have access to such care, even in developed countries. In 2014, the percentage of women in New Zealand who began prenatal care early also varied by ethnicity and socioeconomic status. Māori and Pacific women, young women under age 20 and women from the most deprived neighbourhoods were less likely to begin prenatal care in the first trimester (Ministry of Health, 2015b).

midwife
person who assists in pregnant women's prenatal care and the birth process

Pregnant women in developing countries are much less likely than those in developed countries to receive prenatal care from a skilled health care worker. *Making Pregnancy Safer*, a global initiative developed by the World Health Organization (WHO), has focused on working with governments to set up programs that provide pregnant women with

such care (WHO, 2009a). Currently 99% of maternal and infant deaths occur in developing countries—only 1% occur in developed countries—and the WHO program is focused on the 70 countries with the highest death rates, mostly in Africa and South Asia.

Guidelines for prenatal care focus mostly on three key areas: diet, exercise and avoidance of potentially harmful influences called teratogens (see Table 2.2; WHO, 2016d). Prenatal behaviours was one of the areas that was included in the first data from the longitudinal Growing Up in New Zealand study that began in 2009 with around 7000 families, and which has been following these families for several years to date. Prior to pregnancy, 61% of the women reported they exercised regularly; once pregnant, 60% of these pregnant mothers continued exercising (Morton et al., 2010).

The *Cultural focus: Pregnancy and prenatal care across cultures* discusses the custom of prenatal massage in some cultures.

CRITICAL-THINKING QUESTION

Are there any beliefs in your culture about what a woman should eat or should avoid eating before or during pregnancy?

CULTURAL FOCUS

Pregnancy and prenatal care across cultures

Although many cultures have folk beliefs about pregnancy that have no scientific or practical basis, most also have customs that provide genuine relief to pregnant women. One helpful method of prenatal care common in many traditional cultures is massage (Field, 2010; Jordan, 1994). The prenatal massage is usually performed by a midwife in the course of her visits to the pregnant woman. While the massage is taking place, the midwife asks the woman various questions about how the pregnancy is going. As part of the massage, the midwife probes to determine the fetus's position in the uterus. If the fetus is turned in an unfavourable position, so that it would be likely to come out feet first rather than head first, the midwife will attempt an *inversion* to turn the fetus's head towards the vaginal opening. This is sometimes painful, but a head-first birth is much safer than a feet-first birth, for both baby and mother.

Prenatal massage has a long history in many cultures (Jordan, 1994). In New Zealand, Pacific caregivers in one study frequently mentioned having a traditional pregnancy massage called *milimili* (gentle rubbing), which they used when they were not feeling well and also more generally to promote wellbeing and to prepare for birth (Abel, Park, Tipene-Leach, Finau & Lennan, 2001). In recent years, it has also begun to be used by midwives, nurses and doctors in developed countries.

Anders Ryman/Corbis NX/Getty Images

By now, a substantial amount of research has accumulated to support the benefits of massage for mother and fetus. Benefits to the mother include lower likelihood of back pain, less swelling of the joints and better sleep (Field, 2004, 2010). Babies whose mothers received prenatal massage score higher on scales of their physical and social functioning in the early weeks of life (Field, Diego & Hernandez-Reif, 2006).

Review question

For pregnant women in developed and developing countries, are there different benefits to massage?

Moderate exercise is part of good prenatal care.

Anna Omelchenko/Shutterstock

TABLE 2.2 ESSENTIALS OF PRENATAL CARE

BEFORE PREGNANCY

- Have a medical examination to ensure there are no diseases that may affect prenatal development. If not fully vaccinated, obtain vaccinations for diseases, such as rubella, that can damage prenatal development. (Vaccinations may be unsafe during pregnancy.)
- Avoid tobacco, alcohol and other drugs, which may make it more difficult to become pregnant and are damaging to prenatal development.

DURING PREGNANCY

- *Diet.* Maintain a balanced diet, including protein, grains, fruits and vegetables. Avoid excessive fats, sugars and caffeine, and obtain sufficient iron and iodine. Gain 11–16 kilograms in total; avoid dieting as well as excessive weight gain. Women should also drink more fluids during pregnancy than they normally do because the fetus needs fluids for healthy development, and a pregnant woman's body also requires more.
- *Exercise.* Engage in mild to moderate exercise regularly, including aerobic exercise. *Aerobic exercise*, such as walking, jogging or swimming, is related to decreased back pain, lower risk of gestational diabetes, better sleep and more energy (Binkley, Binkley & Wise, 2015). However, it is important to avoid strenuous exercise and high-risk sports, such as long-distance running, contact sports, downhill skiing, waterskiing and horseback riding.
- *Teratogens.* Avoid tobacco, alcohol and other drugs. Avoid exposure to X-rays, hazardous chemicals and infectious diseases.

LO 2.13

Identify the major teratogens in developing countries and developed countries.

teratogen
behaviour, environment or bodily condition that can have damaging influence on prenatal development

Teratogens

An essential part of good prenatal care is avoiding **teratogens**, which are behaviours, environmental effects and bodily conditions that could be harmful to the developing organism (Haffner, 2007). Both the embryo and the fetus are vulnerable to a variety of teratogens. The embryonic period, especially, is a *critical period* for prenatal development, meaning that it is a period when teratogens can have an especially profound and enduring effect on later development, as Figure 2.8 illustrates. This is because the embryonic period is when all the major organ systems are forming at a rapid rate. However, some teratogens can do damage during the fetal period. Major teratogens include malnutrition, infectious diseases, alcohol, tobacco and drugs. Other potential teratogens include environmental pollution, radiation

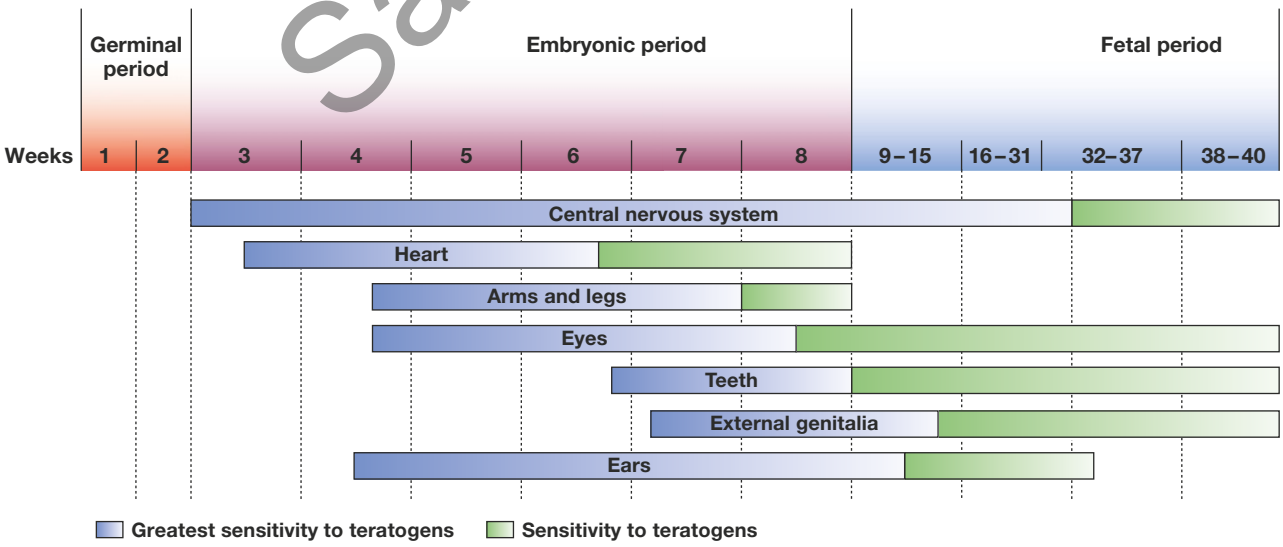


FIGURE 2.8 Timing of vulnerability to teratogens

Vulnerability to teratogens is greatest in the embryonic period.

Source: Based on Moore (1974).

and severe maternal stress. Certain kinds of work are best avoided during pregnancy if they involve exposure to teratogens such as X-rays, hazardous chemicals or infectious diseases.

Time of exposure, dosage and frequency of exposure to drugs and other chemicals, and genetic susceptibility all influence the extent of damage caused. The timing of exposure matters because specific teratogens may affect specific organs. Teratogens typically have their worst effects when those organs are developing. As you can see in Figure 2.8, the central nervous system may be affected any time during the embryonic and fetal periods because the central nervous system is developing the entire time. On the other hand, teratogens that affect the ears have their effects when the ears are developing, beginning in week 4 and ending around week 32 (Moore, Persaud & Torchia, 2011).

Malnutrition

Probably the most common teratogen worldwide is malnutrition. Medical experts recommend that pregnant women who have a healthy weight before becoming pregnant gain 11–16 kilograms, and that they eat a healthy, balanced diet of proteins, grains, fruit and vegetables (Martin, Park & Sutton, 2002; Ministry of Health, 2014). However, as discussed in Chapter 1, 40% of the world's population lives on less than US$2 a day, so you can imagine that most mothers who comprise part of that 40% receive a prenatal diet that falls far short of the ideal.

Furthermore, about half the world's population is rural, and the diet of people in rural areas often varies substantially depending on the time of year. They may eat fairly well during summer and autumn when their crops provide food, but less well during winter and spring when fresh food is unavailable. Consequently, prenatal health may depend greatly on when the child was conceived.

Malnutrition is a common teratogen in developing countries. Pictured here is a pregnant woman in rural Zambia.

Sue Cunningham Photographic/ Alamy Stock Photo

Dramatic evidence of this effect has been shown in recent decades in China (Berry et al., 1999). In the 1980s, China had the highest incidence in the world of two serious prenatal disorders, *anencephaly*, in which parts of the brain are missing or malformed, and *spina bifida*, which is an extreme distortion in the shape of the spinal column. It was discovered that for both of these disorders the main cause is a deficiency of folic acid, a nutrient found especially in fruits and vegetables. Furthermore, researchers observed that the traditional marriage period in China is January and February, and most couples try to conceive a child as soon after marriage as possible. Consequently, the early months of pregnancy typically take place in winter and early spring, when rural women are least likely to have fruits and vegetables as part of their diet. After this pattern was discovered, the Chinese government established a nationwide program to provide mothers with supplements of folic acid, and since that time the incidence of anencephaly and spina bifida has been sharply reduced (Centers for Disease Control and Prevention [CDC], 2011).

Many other countries have also taken steps to reduce folic-acid deficiencies in pregnant mothers. After research established that folic acid was the key to preventing anencephaly and spina bifida, governments in many countries passed laws requiring folic acid to be added to grain products such as cereals, bread, pasta, flour and rice. Almost immediately, the incidence of both disorders fell sharply (Honein, Paulozzi, Mathews, Erickson & Wong, 2001). Medical authorities now recommend that women begin taking folic acid supplements and eating plenty of fruits and vegetables even when they are trying to become pregnant because the damage from lack of folic acid can take place in the early weeks of pregnancy, before the woman knows for sure that she is pregnant (de Villarreal, Arredondo, Hernández & Villarreal, 2006).

Two other common nutritional deficiencies during pregnancy are iron and iodine. Iron-rich foods such as beef, duck, potatoes (including skin), spinach and dried fruits are important for building the blood supply of mother and fetus. The WHO estimates that nearly one-half of women worldwide are deficient in iron, placing them at risk for having preterm and low-birth-weight babies (WHO, 2016d). Even with a healthy diet that includes iron-rich foods, health authorities recommend pregnant women take an iron supplement from week 12 of pregnancy onward.

Iodine is also crucial because low-iodine intake during pregnancy increases the risks of miscarriage, stillbirth and abnormalities in fetal brain development. In most developed countries, salt has been iodised since the 1920s, so women receive adequate iodine as part of a normal diet. However, in many developing countries most women do not use iodised salt and consequently they often experience iodine deficiencies. The WHO and other major health organisations have made a strong push recently to make iodine supplements available in developing countries.

Infectious diseases

Infectious diseases are far more prevalent in developing countries than in developed countries (WHO, 2009a, 2009b). Many of these diseases influence prenatal development. One of the most prevalent and serious is *rubella* (also known as *German measles*). The embryonic period is a critical period for exposure to rubella. Over half of infants whose mothers contract the illness during this period have severe problems, including blindness, deafness, intellectual disability and abnormalities of the heart, genitals or intestinal system (Eberhart-Phillips, Frederick & Baron, 1993). During the fetal period, effects of rubella are less severe, but can include low birth weight, hearing problems and skeletal defects (Brown & Susser, 2002). Since the late 1960s, a vaccine given to children has made rubella rare in developed countries—girls retain the immunity into adulthood, when they become pregnant—but it remains widespread in developing countries where children are less likely to receive the vaccine (Plotkin, Katz & Cordero, 1999; WHO, 2017b).

AIDS (acquired immune deficiency syndrome)
sexually transmitted infection caused by HIV, resulting in damage to the immune system

Another common infectious disease of prenatal development is **AIDS (acquired immune deficiency syndrome)**, a sexually transmitted infection (STI) caused by the human immunodeficiency virus (HIV), which damages the immune system. HIV/AIDS can be transmitted from mother to child during prenatal development through the blood, during birth or through breast milk. HIV/AIDS damages brain development prenatally, and infants with HIV are unlikely to survive to adulthood unless they receive an expensive 'cocktail' of medications rarely available in the developing countries where AIDS is most common. In developing countries, mother–child transmission of HIV/AIDS has been dramatically reduced in recent years through four strategies: (1) effective antiretroviral drugs given to mothers before birth; (2) caesarean delivery for mothers infected with AIDS where it is safe to do so; (3) breastfeeding mothers who are HIV positive supported to adhere to antiretroviral drugs; and (4) the use of infant formula in place of breastfeeding when medical intervention is not possible (WHO, 2016a, 2016b). However, 95% of all HIV infections take place in Africa, and few African mothers or infants have access to these strategies that are effective against HIV/AIDS.

Pregnant women in developing countries who have AIDS rarely receive adequate medical treatment. Here, a woman is being treated at a clinic for HIV/AIDS patients in Lesotho.

Gideon Mendel/Corbis Historical/Getty Images

Alcohol

In developed countries, the teratogen that causes the most widespread damage to prenatal development is alcohol (Mattson et al., 2010; Sokol, Delaney-Black & Nordstrom, 2003). Although it used to be believed

that moderate alcohol use would cause no harm during pregnancy, recent research has shown that the only safe amount of alcohol for a pregnant woman is *none at all*. Even one or two drinks a few days a week puts the developing child at risk for lower height, weight and head size at birth, and for lower intelligence and higher aggressiveness during childhood (Willford, Richardson, Leech & Day, 2004). However, many fetuses are exposed to alcohol. A survey in New Zealand found that 34% of pregnant women drank alcohol at some point during their pregnancy, and 24% continued to drink once they knew they were pregnant (Mallard, Connor & Houghton, 2013). In 2013, 53% of pregnant Australian women abstained from drinking; many who drank did so before they knew they were pregnant, but 26% continued to consume alcohol when they knew they were pregnant (AIHW, 2016a).

When mothers drink during pregnancy, their infants are at risk for **fetal alcohol spectrum disorder (FASD)**, which includes facial deformities, heart problems, misshapen limbs and a variety of cognitive problems such as intellectual disability and attention and memory deficits (Mattson et al., 2010). Infants born with FASD face a lifetime of difficulty, and the more alcohol their mothers drank during pregnancy, the worse their problems are likely to be (Barr & Streissguth, 2001). A review of 25 studies found that adolescents exposed to alcohol in the uterus experienced cognitive, behavioural, social and emotional problems (Irner, 2012). These problems are severe and make it difficult for the adolescents to succeed academically or socially (Mattson et al., 2010). In Australia, the incidence of fetal alcohol syndrome is estimated to be as much as 39 times higher for Indigenous children as for non-Indigenous children, but this incidence is still far lower than the rates seen among Indigenous populations from other countries (Burns, Breen, Bower, O'Leary & Elliott, 2013).

fetal alcohol spectrum disorder (FASD)
range of effects that can occur in an individual whose mother drank during pregnancy, including facial deformities, heart problems, misshapen limbs and a variety of cognitive problems

Tobacco

Maternal cigarette smoking has a wide range of damaging effects on prenatal development. Women who smoke during pregnancy are at higher risk for miscarriage and premature birth, and smoking is the leading cause of low birth weight in developed countries (Espy et al., 2011). Maternal smoking raises the risks of health problems in infants, such as impaired heart functioning, difficulty breathing and even death (Jaakkola & Gissler, 2004). Prenatal exposure to smoking predicts problems in childhood and adolescence, including poorer language skills, problems with attention and memory, and behaviour problems (Cornelius et al., 2011; Sawnani, Jackson, Murphy, Beckerman & Simakajornboon, 2004).

Second-hand smoke from fathers' and others' smoking leads to higher risks of low birth weight and childhood cancer (Rückinger, Beyerlein, Jacobsen, von Kries & Vik, 2010a). Rates of smoking are generally higher in developed countries than in developing countries, but smoking rates are rising rapidly in developing countries around the world as their economies grow (WHO, 2011a). In Australia and New Zealand, about 12% of pregnant women report smoking during pregnancy (AIHW, 2016a; Ministry of Health, 2015b). However, 34% of Māori in the Growing Up in New Zealand study reported that they smoked during pregnancy (Morton et al., 2010). Two in 5 women who smoked before pregnancy continued to smoke while they were pregnant (AIHW, 2016b). Smoking is also associated with other risk factors, such as fewer visits to health professionals, being a teenager and living in a remote area.

Drugs

Alcohol and tobacco are the most common teratogens in developed countries, but other types of drugs also have a harmful effect on the developing child. Maternal use of recreational psychoactive drugs, such as cocaine, heroin and marijuana, causes physical, cognitive and behavioural problems in infants (Messinger & Lester, 2008; National Institute

on Drug Abuse, 2001). Specifically, marijuana can affect the development of the brain and central nervous system, and it can retard the growth of the fetus (Gunn et al., 2015; Wu, Jew & Lu, 2011). The use of cocaine can result in low birth weight due to both slow growth of the fetus and premature labour, as well as emotional reactivity from infancy through adolescence (Bridgett & Mayes, 2011; Eiden, Foote & Schuetze, 2007; Minnes et al., 2010). Like marijuana and cocaine, heroin can also slow fetal growth (Kaltenbach & Finnegan, 1989; Keegan, Parva, Finnegan, Gerson & Belden, 2010). Babies born to mothers who used heroin during pregnancy go through a painful withdrawal and are usually given methadone to cope with the pain. It is helpful to have these babies room in with their mothers, rather than being in a separate nursery (Abrahams et al., 2007).

Certain prescription drugs can also cause harm. Even non-prescription drugs such as medicines for the common cold can be damaging to prenatal development, so women who are pregnant or seeking to become pregnant should always check with their doctors about any medications they may be taking (Morgan, Cragan, Goldenberg, Rasmussen & Schulkin, 2010). Among prescription drugs, Accutane, a drug used to treat severe acne, can cause devastating damage to major organs such as the brain and heart during embryonic development (Honein et al., 2001). Many women take selective serotonin reuptake inhibitors (SSRIs) for depression or anxiety, and some of these drugs (e.g. fluoxetine and paroxetine) have been found in some studies to cause heart and other abnormalities (Myles, Newall, Ward & Large, 2013). However, many studies suggest that these drugs are safe overall (Casper, 2015). And, the effects of maternal depression on the developing fetus may be worse than the effects of any drugs that treat depression (Olivier, Åkerud & Sundström Poromaa, 2015). When it comes to any prescription or over-the-counter drugs, it is best to weigh potential risks and benefits in a discussion with a health provider.

SUMMARY PRENATAL DEVELOPMENT AND PRENATAL CARE

LO 2.9 Describe the structures that form during the germinal period.

During the germinal period, a ball of cells called the *blastocyst* forms and implants in the lining of the uterus. The blastocyst has two layers: the embryonic disk that will become the embryo of the new organism, and the trophoblast that will form the supporting structures of the amnion, placenta and umbilical cord.

LO 2.10 Outline the major milestones of the embryonic period.

During the embryonic period (3–8 weeks after conception) all the major organ systems are initially formed, except the sex organs. Rapid development of organs during this period makes it a critical period for the effects of teratogens.

LO 2.11 Describe the major milestones of the fetal period and identify when viability occurs.

During the fetal period (week 9–birth), organ systems continue to develop and there is immense growth in size. Viability is rare before the third trimester because of the immaturity of the lungs. By 28 weeks, the fetus has sleep–wake cycles similar to those of a newborn baby and can remember and respond to sound, taste and the mother's movements.

LO 2.12 Compare and contrast prenatal care in traditional cultures and developed countries.

In traditional cultures, prenatal care often includes massage as well as folk knowledge that may or may not have practical consequences. Essential elements of scientifically based prenatal care include regular evaluations by a health care professional and guidelines concerning diet, exercise and avoiding teratogens. Pregnant women of a healthy weight are advised to gain 11–16 kilograms in the course of pregnancy, and light to moderate exercise is encouraged.

LO 2.13 Identify the major teratogens in developing countries and developed countries.

The major teratogens are malnutrition and infectious diseases in developing countries, and alcohol and tobacco in developed countries. The embryonic period is a critical period for prenatal development because all the major organ systems are forming at a rapid rate. However, some teratogens can do damage during the fetal period as well.

SECTION 3

PREGNANCY PROBLEMS

LEARNING OBJECTIVES

- **LO 2.14** Explain how chromosomal disorders occur.
- **LO 2.15** Describe causes and symptoms of some common genetic disorders.
- **LO 2.16** Describe the main techniques of prenatal diagnosis.
- **LO 2.17** Explain who is likely to seek genetic counselling and for what purposes.

PREGNANCY PROBLEMS: CHROMOSOMAL AND GENETIC DISORDERS

Most pregnancies proceed without major problems and end with the birth of a healthy infant. However, many things can go wrong in the course of prenatal development. In this section, we'll look at some common chromosomal disorders and genetic disorders.

Chromosomal disorders

LO 2.14

Explain how chromosomal disorders occur.

In the course of the formation of the gametes during meiosis, sometimes errors take place and the chromosomes fail to divide properly. Consequently, instead of ending up with 46 chromosomes, the person has 45 or 47 (or even, in rare cases, 48 or 49), and problems occur. It is estimated that as many as half of all conceptions involve too many or too few chromosomes, but most of the zygotes that result either never begin to develop or are spontaneously aborted early in the pregnancy (Borgaonkar, 1997; Johnson, 2016). In 1 out of 200 live births, the child has a chromosomal disorder. There are two main types of chromosomal disorders: (1) those that involve the sex chromosomes; and (2) those that take place on the 21st pair of chromosomes, resulting in a condition known as Down syndrome.

Sex chromosome disorders

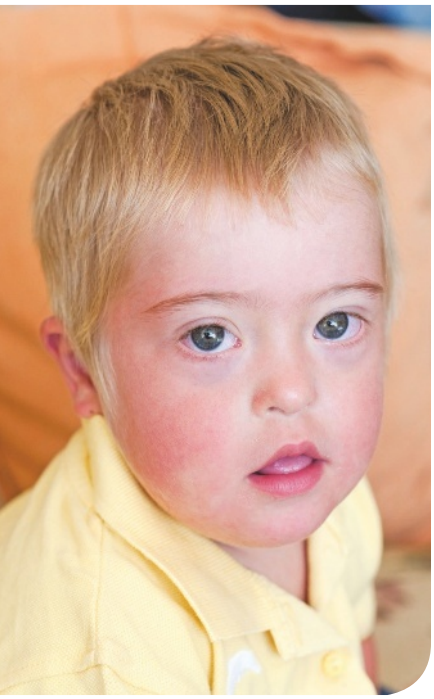
The sex chromosomes are especially likely to be involved in chromosomal disorders. A person may have an extra X chromosome (resulting in XXX or XXY), an extra Y chromosome (XYY) or may have only an X and no second sex chromosome. About 1 in every 500 infants has some type of sex chromosome disorder.

There are two common consequences of sex chromosome disorders (Batzer & Ravitsky, 2009). One is that the person has some type of cognitive deficit, such as intellectual disability (ranging from mild to severe), a learning disorder or a speech impairment. The other kind of problem is that the person has some abnormality in the development of the reproductive system at puberty, such as underdeveloped testes and penis in boys or no ovulation in girls. One of the functions of the sex chromosomes is to direct the production of the sex hormones, and having too few or too many sex chromosomes disrupts this process. However, treatment with hormone supplements is often effective in correcting the problem.

Down syndrome

When there is an extra chromosome on the 21st pair of chromosomes, the condition is known as **Down syndrome**, or *trisomy-21*. People with Down syndrome have distinct physical features, including a short, stocky build; an unusually flat face; a large tongue; and an extra fold of skin on the eyelids. They also have cognitive deficits, including intellectual disability

Down syndrome
genetic disorder due to carrying an extra chromosome on the 21st pair of chromosomes



People with Down syndrome typically face a wide range of physical and cognitive problems.

Alex Segre/Alamy Stock Photo

LO 2.15

Describe causes and symptoms of some common genetic disorders.

and speech problems (Pennington, Moon, Edgin, Stedron & Nadel, 2003). Many also have problems in their physical development, such as hearing impairments and heart defects.

Their social development varies widely. Some children with Down syndrome smile less readily than other people and have difficulty making eye contact, but others are exceptionally happy and loving. Supportive and encouraging parents help children with Down syndrome develop more favourably (Hodapp, Burke & Urdano, 2012; Sigman, 1999). Intervention programs in infancy and preschool have been shown to enhance these children's social, emotional and motor skills (Carr, 2002; Hodapp et al., 2012). In adulthood, with adequate support many are able to hold a job that is highly structured and involves simple tasks.

People with Down syndrome age faster than other people (Berney, 2009). Their total brain volume begins to decrease as early as their 20s. Various physical ailments that may develop for other people in late adulthood begin to afflict people with Down syndrome in their 30s and 40s, including leukaemia, cancer, Alzheimer's disease and heart disease (Hassold & Patterson, 1999). As a result, their life expectancy is considerably lower than in the general population. However, with medical treatment most are able to live into at least their 50s or 60s (Hodapp et al., 2012).

Parental age and chromosomal disorders

Children with chromosomal problems are almost always born to parents who have no disorder (Batzer & Ravitsky, 2009). Chromosomal problems occur not because the parents have an inherited problem that they pass on to their children, but usually because of the age of the parents, especially the mother. For example, the risk of Down syndrome rises with maternal age, from 1 in 1900 births at age 20 to 1 in 30 births at age 45 (Meyers, Adam, Dungan & Prenger, 1997). The risk of chromosomal disorders is low for mothers in their 20s and rises only slightly in the 30s, but rises steeply after age 40 (see Figure 2.9; Umrigar, Banajee & Tsien, 2014).

Recall that a woman's gamete production takes place while she is still in the uterus of her own mother. The older the woman gets, the longer the eggs have been in her ovaries. When conception takes place and the last part of meiosis is completed in the ovum, the older the woman, the greater the likelihood that the chromosomes will not separate properly because they have been suspended in that final stage of meiosis for so long. The father's sperm is the cause of the chromosomal disorder in 5–10% of cases, but it is unclear if the risk increases with the father's age (Crow, 2003; Fisch et al., 2003; Muller, Rebiff, Taillandier, Qury & Mornet, 2000).

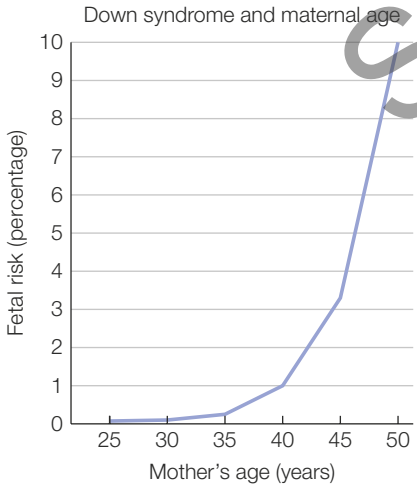


FIGURE 2.9 Down syndrome and maternal age

Why does the risk rise so steeply after age 40?

Source: Based on Umrigar, Banajee & Tsien (2014).

Genetic disorders

Genetic disorders may be caused by incomplete dominance or by mutations. A common example of incomplete dominance is sickle-cell trait. Two common examples of genetic mutations are fragile X syndrome and phenylketonuria (PKU).

In **incomplete dominance**, the phenotype is influenced primarily, but not exclusively, by the dominant gene. One example of incomplete dominance involves the sickle-cell trait that is common among Black Africans and their descendants on other continents (see Figure 2.10). Most blood cells are shaped like a disk, but when a person inherits two recessive genes for the sickle-cell trait, the blood cells become hook-shaped, like the blade of a sickle. This results in a condition called *sickle-cell anaemia*, in which the sickle-shaped blood cells clog up the blood vessels and cause pain, susceptibility to disease and early death. About 1 in 500 people of Black African heritage have this disorder, and it

incomplete dominance
form of dominant–recessive inheritance in which the phenotype is influenced primarily by the dominant gene but also to some extent by the recessive gene

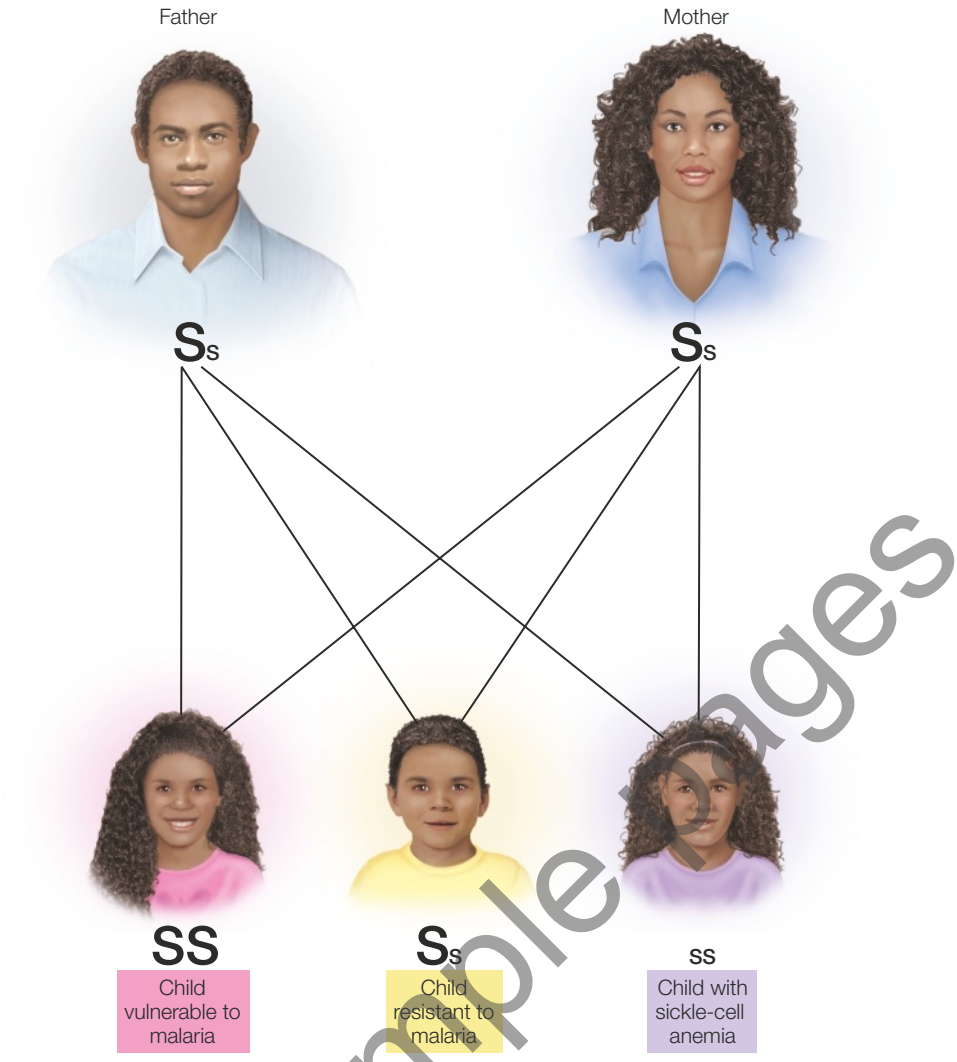


FIGURE 2.10 Incomplete dominance in sickle-cell inheritance
Two recessive genes for the sickle-cell trait results in sickle-cell anaemia, but having one dominant and one recessive gene provides protection against malaria.

also occurs (less commonly) in people whose ancestors are from India, Saudi Arabia, the Mediterranean region or Central America (WHO, 2017a).

However, if a person inherits only one recessive gene for the sickle-cell trait, along with a normal dominant gene, the dominance is incomplete, and a portion—but not all—of the person's blood cells will be sickle shaped. This portion is not large enough to cause sickle-cell anaemia, but it is large enough to make the person resistant to malaria, a blood disease that is spread by mosquitoes. Malaria is often fatal, and even when it is not, it can cause brain damage and other enduring health problems. It occurs worldwide in developing countries but is especially common in Africa, killing over a million people a year. In many central African countries, over 50% of children are affected (WHO, 2013).

This explains why the sickle-cell trait evolved especially among Africans. Because the effects of contracting malaria are so severe, in evolutionary terms it is an advantage genetically to have the sickle-cell trait to protect against malaria, even if it also raises the risk of sickle-cell anaemia.

Other fairly common genetic disorders include fragile X syndrome and phenylketonuria (PKU). Fragile X syndrome, the most common inherited intellectual disability worldwide, is caused by genetic mutations on the X chromosome (Kanwal et al., 2015). It is also the most common genetic disorder associated with autism spectrum disorder (Dalton, Holsen, Abbeduto & Davidson, 2008). Although the syndrome occurs in both males and females, males are more affected by the mutation and exhibit more symptoms. The majority of males have an intellectual disability, ranging from moderate to severe, but only about one-third of females with fragile X syndrome exhibit intellectual delays. Intellectual problems tend to include difficulty paying attention and impulsiveness (Hagerman & Hagerman, 2002). Males tend to exhibit physical symptoms of a long face, large ears, flat feet, soft skin and hyper-flexible joints. For females, the physical symptoms tend to be fewer and milder. There is no cure for fragile X syndrome.

PKU is a birth defect that causes the amino acid phenylalanine to build up in the body because people with PKU cannot process that amino acid effectively. Early, sustained treatment is usually effective and involves avoiding foods and drinks that contain phenylalanine. A low-protein diet is recommended into the teens and 20s (Nardecchia et al., 2015). Left untreated, PKU causes intellectual disability and poor information processing abilities (Weglage et al., 2013).

PREGNANCY PROBLEMS: TESTING AND COUNSELLING OPTIONS

LO 2.16

Describe the main techniques of prenatal diagnosis.

Prenatal testing is an important part of monitoring the development of the fetus. In fact, some couples seek testing and counselling before attempting conception to know the risks or difficulties they might face.

Prenatal diagnosis

ultrasound
machine that uses sound waves to produce images of the fetus during pregnancy

In developed countries, a variety of techniques are available to monitor the growth and health of the fetus and to detect prenatal problems. Common methods include ultrasound, amniocentesis, chorionic villus sampling (CVS) and non-invasive prenatal testing (NIPT).

- *Ultrasound.* In **ultrasound**, high-frequency sound waves are directed towards the uterus, and as they bounce off the fetus they are converted by computer into an image that can be viewed on a screen. Ultrasound technology has improved in recent years and the three- and four-dimensional (3D/4D) images are distinct enough to make it possible to measure the fetus's size and shape and to monitor its activities (Merz & Abramowicz, 2012). Studies have also found that viewing ultrasound images helps promote a feeling of parental involvement and attachment even before birth (Righetti, Dell'Avanzo, Grigio & Nicolini, 2005).

Ultrasound allows medical professionals and parents to monitor prenatal development.

Keith Brofsky/Photodisc/Getty Images

The WHO recommends ultrasounds before week 24 to estimate the age of the fetus, assess anomalies in development such as Down syndrome, and monitor multiple pregnancies (WHO, 2016d). Pregnancies that involve multiple fetuses are high risk, so it is important to assess the development of each fetus. Ultrasound is used for normal pregnancies in developed countries, not just for those that are high risk (Merz & Abramowicz, 2012). It is cheap, easy and safe, and it allows doctors to monitor fetal growth and gives parents the

enjoyment of seeing the fetus as it is developing in the uterus. It also allows parents to learn the sex of the child before birth, if they wish.

- *Amniocentesis*. In **amniocentesis**, a long hollow needle is inserted into the pregnant woman's abdomen and, using the ultrasound image for guidance, a sample of the amniotic fluid is withdrawn from the placenta surrounding the fetus (Alfirevic, Navaratnam & Mujezinovic, 2017). This fluid contains fetal cells sloughed off in the course of prenatal development, and the cells can be examined for information on the fetus's genotype. Amniocentesis is conducted 15–20 weeks into pregnancy. It is used only for women who are at risk for prenatal problems because it carries a small risk of triggering miscarriage. It can detect 40 different defects in fetal development with 100% accuracy (Alfirevic et al., 2017; Brambati & Tului, 2005).

amniocentesis
prenatal procedure in which a needle is used to withdraw amniotic fluid containing fetal cells from the placenta, allowing possible prenatal problems to be detected

- *Chorionic villus sampling (CVS)*. Like amniocentesis, **chorionic villus sampling (CVS)** entails sampling and analysing cells early in development to detect possible genetic problems (Bhatt, 2017). CVS takes place at 5–10 weeks into the pregnancy; the sample is obtained from the cells that are beginning to form the umbilical cord. Guided by ultrasound, a tube is inserted through the vagina and into the uterus to obtain the cell sample. CVS entails a slight but genuine risk of miscarriage or damage to the fetus, so, like amniocentesis, it is used only when there is a family history of genetic abnormalities or the woman is age 35 or over (Bakker et al., 2017). It is 99% accurate in diagnosing genetic problems.

chorionic villus sampling (CVS)
prenatal technique for diagnosing genetic problems, involving taking a sample of cells at 5–10 weeks' gestation by inserting a tube into the uterus

- *Non-invasive prenatal testing (NIPT)*. A method called **non-invasive prenatal testing (NIPT)** (or cell-free DNA analysis) was first introduced in 2013 in Australia. It is far less invasive and nearly risk-free; mothers give only a simple blood sample. Laboratories are able to examine fetal DNA that is in the mother's bloodstream for chromosomal abnormalities from very early in the pregnancy. There is evidence that the test is 99% accurate in detecting chromosomal abnormalities such as Down syndrome, and it is far less likely to get false positives than the procedures described above (Norton et al., 2015). NIPT has the potential to significantly change how pregnant women, their families and health practitioners approach prenatal testing (Wolfberg, 2016).

non-invasive prenatal testing (NIPT)
prenatal blood test that examines fetal DNA in the mother's bloodstream for chromosomal abnormalities

Genetic counselling

LO 2.17

Explain who is likely to seek genetic counselling and for what purposes.

Even before pregnancy, couples whose family history places them at risk for having children with genetic disorders may seek *genetic counselling*, which involves analysing the family history and genotypes of prospective parents to identify possible risks (Coughlin, 2009). Those with risks that merit genetic counselling include people who have an inherited genetic condition or a close relative who has one; couples with a history of miscarriages or infertility; and older couples (women over 35 and men over 40) (Fransen, Meertens & Schrander-Stumpel, 2006). The decision to obtain genetic counselling may be difficult because the results may require the couple to make the choice between trying to become pregnant and risking that the child will have a genetic disorder, or deciding not to pursue pregnancy. However, the knowledge obtained from genetic counselling enables people to make an informed decision.

In the first step of genetic counselling, the counsellor takes a comprehensive family history from each prospective parent, seeking to identify patterns that may indicate problematic recessive or X-linked genes. Then each partner provides a blood, skin or urine sample that can be used to analyse their chromosomes to identify possible problems. With the information obtained from genetic counselling, the couple can then decide whether or not they wish to attempt pregnancy (Coughlin, 2009).

CRITICAL-THINKING QUESTION

Do you think that genetic counselling will increase or decrease in the next 10 years? Provide reasons for your answer from an ecocultural point of view.

SUMMARY PREGNANCY PROBLEMS

LO 2.14 Explain how chromosomal disorders occur.

Chromosomal disorders occur when the chromosomes fail to divide properly during meiosis. These disorders may involve the sex chromosomes or may take place on the 21st pair of chromosomes, resulting in a condition known as Down syndrome. Risks of chromosomal disorders rise with parental age.

LO 2.15 Describe causes and symptoms of some common genetic disorders.

Common genetic disorders include sickle-cell anaemia, fragile X syndrome and phenylketonuria (PKU). Sickle-cell anaemia is a genetic disorder that causes misshapen red blood cells that cannot process oxygen correctly. Sickle-cell trait is caused by incomplete dominance, in which the dominant gene exerts most, but not all, of the influence over genetic expression. Fragile X syndrome is caused by a mutation on the X chromosome, and it is the most common genetic disorder linked to intellectual disability. In PKU, individuals are unable to digest the amino acid phenylalanine.

LO 2.16 Describe the main techniques of prenatal diagnosis.

Prenatal diagnosis may include ultrasound, amniocentesis and chorionic villus sampling (CVS). Ultrasound and non-invasive prenatal testing carry no risks. The risks of amniocentesis and CVS should be discussed with a doctor.

LO 2.17 Explain who is likely to seek genetic counselling and for what purposes.

Couples who may be at high risk for genetic disorders sometimes seek genetic counselling before attempting pregnancy. Information from family histories and blood, skin or urine samples is used to help couples make family planning decisions

Chapter quiz

1 The totality of an individual's genes is the _____.

a phenotype
b genotype
c allele
d dominant gene

2 Which of the following best describes the situation as to who has the greatest risk of developing haemophilia, which is an X-linked recessive disorder?

a A female who has one X chromosome that contains the gene for this disorder
b A male who has one X chromosome that contains the gene for this disorder
c Males and females with one X chromosome that contains the gene for the disorder will have equal risk.
d Only some Asians because of their unique genetic makeup

3 Which of the following questions would a behaviour geneticist be most likely to ask?

a 'Why are children in the same family so different from one another?'
b 'Are preterm babies more likely than full-term babies to have learning difficulties during the school years?'
c 'How can prenatal tests be used to detect Down syndrome?'
d 'What effects does alcohol have on the developing organism?'

4 **Why has there been little change in the average height of populations in Western countries over the last few decades?**

a The population has become overweight or obese, which negatively affects height.
b People in Western countries have been exposed to more diseases.
c People have reached the upper boundary of their reaction range for height.
d Evolutionary influences are causing all populations to decrease in height.

5 **John is short for his age and has good coordination. Although he is exposed to a variety of activities, none has particularly interested him. His father, who used to wrestle when he was younger, signs John up for wrestling, thinking this could be the perfect sport for John. He convinces John to give wrestling a try, and John goes on to become a champion wrestler. This is an example of _____.**

a passive genotype → environment effects
b evocative genotype → environment effects
c active genotype → environment effects
d heritability

6 **As a result of the process of crossing over, _____.**

a the risk of Down syndrome is increased
b boys are more likely to be born with a learning disability
c women are at increased risk for infertility
d each child born to a set of parents is genetically unique (with the exception of identical twins)

7 **Sasha is most likely to have DZ twins if _____.**

a she has Asian biological parents
b she is in her late teens
c she is concerned about gaining too much weight and severely restricts her kilojoule intake
d her mother had DZ twins

8 **In Australia, about one in _____ couples is infertile.**

a three
b four
c six
d eight

9 **The germinal period lasts _____.**

a about 1 day
b about 3 days
c about 7 days
d about 14 days

10 **During the embryonic period, _____.**

a the blastocyst forms
b the zygote is created
c the zygote attaches to the uterine wall
d the major organs develop

11 **Saad, a baby born 6 weeks prematurely, is more at risk of not surviving than Nona, a full-term baby, because Saad's _____ is(are) still immature.**

a small intestines
b heart
c lungs
d spleen

12 **Which is the following is true of good prenatal care?**

a Exercise should be avoided.
b Tobacco, alcohol and other drugs should be avoided.
c Women should drink fewer fluids than before pregnancy.
d Twenty to 25 kilograms should be gained.

13 **Louisa's baby was born blind, deaf and with intellectual disability. It is most likely that during her pregnancy, Louisa _____.**

a contracted AIDS
b had rubella
c had a severe nutritional deficiency
d ate foods that were too high in folic acid

14 **A child who has an X0 chromosomal make-up (where '0' denotes a missing chromosome where there is supposed to be a 23rd pair) will most likely _____.**

a be a male with Down syndrome
b be a female who will later experience problems in the development of the reproductive system
c be a typical female who will not experience cognitive or physical problems
d not survive past the age of 3

15 **Keisha has inherited one recessive gene for the sickle-cell trait along with one normal dominant gene. As a result of this _____, she is resistant to malaria and does not have sickle-cell anaemia.**

a dominant–recessive inheritance
b incomplete dominance
c polygenic inheritance
d reaction range

16 **Carissa has a family history of Down syndrome and is in week 5 of her pregnancy. She decides she would like to find out as early as possible whether her unborn child has Down syndrome or any other genetic abnormality. What test is she most likely to get?**

a Fetal monitoring
b Ultrasound
c Amniocentesis
d Non-invasive prenatal test

17 **The first step of genetic counselling is that each prospective parent provides a _____.**

a blood test
b comprehensive family history
c urine sample
d chorionic villus sampling