CHAPTER

2

Foundations of development

Genetics and prenatal development

LEARNING OBJECTIVES

After reading this chapter you should be able to:

2.1 Describe the basic principles of genetic transmission of normal and abnormal human attributes

2.2 Discuss disorders associated with chromosomal abnormalities

2.3 Discuss the ways in which psychologists employ behaviour genetics to distinguish between inherited and experiential factors in development

2.4 Outline the time course of prenatal development

2.5 Identify the principles governing the action of environmental agents that may cause harm to the developing embryo/foetus

2.6 Assess evidence of prenatal learning

2.7 Identify the adverse effects on infant survival and healthy development of prematurity/low birth weight

2.8 Conduct a research exercise examining the difference between related and unrelated pairs of children on a number of behavioural characteristics.

This chapter outlines the biological foundations of development, examining first the genetic bases of normal and abnormal development and the methods adopted by psychologists to investigate the genetic contribution to complex behaviours. Next we consider the rapid developmental changes that take place in the prenatal period and the abnormalities that can occur when environmental agents disrupt the normal pattern of development. Our main objectives are to introduce the factors that impinge on the formation of the individual prior to birth and to give a feel for the scope of individual variability that is possible as a result of both genetic and prenatal factors.

This chapter is not a detailed account of human genetics or behaviour genetics, nor does it attempt to detail the multitude of changes that occur prenatally. Such detail can be found in texts dealing with human genetics, embryology and teratology (e.g. O'Rahilly & Muller, 2001; Plomin, DeFries, Knopik & Neiderhiser, 2013).

GENETIC FOUNDATIONS

A knowledge of the genetic contribution to the emergence of abilities is central to understanding how development proceeds. Such understanding provides information about the nature side of the nature/nurture debate. Those attempting to differentiate the inherited from the environmental influences on the development of human behaviour have traditionally relied on twin studies and adoption studies for their evidence, but new advances in identifying the genetic make-up of individuals promise more direct evidence on this matter. The mapping of the human genome has opened up a vast potential for understanding the genetic bases of behaviour (see Research Focus 2.1). Research in this area promises some of the most exciting scientific discoveries of the 21st century.

In recent years it has become obvious that genetics plays a major role in behaviour. Some of the most important recent discoveries about behaviour have involved genetics, and many of these discoveries have related to genetic disorders. There are over 7000 known genetic disorders, many of which affect behaviour, with intellectual disability the most common behavioural symptom. Notable examples of these disorders include Alzheimer's disease, Down syndrome, Fragile-X syndrome and bipolar disorder.

Alzheimer's disease (see Chapter 12), the most common cause of dementia, is a progressive brain disease that causes severe memory loss and confusion. Dementia has become an increasing public health concern as the population ages, with the Australian Institute of Health and Welfare (AIHW) (2014) estimating that by 2020 there will be nearly 400 000 people with dementia in Australia. The incidence of dementia rises exponentially in elderly people, from 1 per cent at age 65 to approximately 40–50 per cent by the age of 95 (Corrada, Brookmeyer, Paganini-Hill, Berlau & Kawas, 2010). A large proportion of these cases is due to Alzheimer's disease. While its cause is not fully understood, it is clear that genetics plays a role in the onset of the disease.

Early-onset Alzheimer's (occurring before 60 years of age) is relatively rare, representing less than 5 per cent of all Alzheimer's cases. While for some of these cases the cause is unknown, most are inherited (familial Alzheimer's disease, or FAD). FAD is caused by any of a number of single-gene mutations on chromosomes 21, 14 and 1 (National Institutes of Health, 2011). These mutations cause the formation of abnormal proteins which result in the brain abnormalities associated with the disease (e.g. amalyoid plaques and neurofibrillary tangles).

Most cases of Alzheimer's are the late-onset form, developing after the age of 60. The causes of this form of the disease are not fully understood; however, as pointed out by the National Institutes of Health (2011), it is likely that a combination of genetic, environmental and lifestyle factors influence a person's risk of developing late-onset Alzheimer's. While not caused by the single genes associated with FAD, it is possible to identify genes that are associated with the risk for the disorder. Systematic meta-analysis of studies investigating the genetics of the disease has highlighted more than 20 gene loci that have significant effects on the risk of contracting Alzheimer's (Bertram & Tanzi, 2008). Additionally, later studies identified five new genes associated with Alzheimer's disease (Hollingworth et al., 2011) and four new genes linked to late-onset Alzheimer's disease (Naj et al., 2011).

Down syndrome
A genetic disorder resulting from the inheritance of an extra chromosome (chromosome 21). It is the single most important cause of mental retardation.

Down syndrome is the single most important cause of mental retardation (accounting for more than a quarter of institutionalised mentally retarded individuals). It is also caused by a genetic disorder, the inheritance of an entire extra chromosome (chromosome 21).

The second most common cause of mental retardation is a condition called **Fragile-X retardation**, so called because it results from the breaking of the X (sex) chromosome. In 1991, researchers identified a single gene on the X chromosome responsible for the disorder. It occurs nearly twice as often in males as in females.

Bipolar disorder is a relatively common and often devastating illness characterised by extreme mood dysregulation. Family, twin and adoption studies consistently indicate a strong genetic component, but the specific genes that contribute to the disorder remain unclear (Hayden & Nurnberger, 2006). The genetic basis of bipolar disorder is being studied by a team of Sydney-based researchers who have identified the likely location on a small section of chromosome 4 of a gene (or genes) that increases the risk of the disease developing (Blair et al., 2006). More recently,

Fragile-X retardation
A genetic disorder caused by a gene on the X chromosome that results in the breaking of the chromosome. It is the second most important cause of mental retardation.

bipolar disorder
A relatively common and severely debilitating depressive disorder that appears to have a strong genetic link.

RESEARCH FOCUS 2.1

The Human Genome Project

The Human Genome Project (HGP) has been described as 'the first major foray of the biological and medical research communities into "big science"' (Collins, Morgan & Patrinos, 2003, p. 286). Commencing in 1990 under the direction of the US Department of Energy and the National Institutes of Health, and joined in the mid-1990s by UK researchers funded by the Wellcome Trust, the project was a 13-year research effort to map the human genome.

The primary goal of the HGP was to provide researchers with the tools to understand the genetic factors in human disease, hence providing pathways to developing new diagnostic, treatment and prevention strategies. The specific goals, as outlined on the Human Genome Project Information website (US Department of Energy, 2011), were to identify all of the approximately 20 000–25 000 genes in human DNA and to determine the sequences of the three billion chemical base pairs that make up human DNA. Other stated goals were to store this information in databases; to improve tools for data analysis; to transfer related technologies to the private sector; and to address the ethical, legal and social issues arising from the project.

In April 2003 the project was successfully completed, and it has already contributed to the discovery of more than 1800 genes responsible for disease and to the development of more than 2000 genetic tests for human conditions. It has also resulted in the development of hundreds of biotechnology-based products for treatment of disorders, some 350 of which are currently undergoing clinical trials (National Institutes of Health, 2010).

The HGP was the first large scientific undertaking to dedicate a portion of its budget (3–5 per cent) for research into social, legal and ethical implications, resulting in 'the world's largest bioethics program' (US Department of Energy, 2011). Many issues have been (and are being) addressed in this research, including questions of fair use of the information (e.g. who should have access to personal genetic information?), privacy and confidentiality (e.g. who owns and controls this information?), psychological impact (e.g. what are the effects of genomic information on perceptions and treatment of individuals?), reproductive issues (e.g. should 'at risk' couples be counselled?) and clinical issues (evaluation of genetic tests for accuracy, reliability and utility). Others have looked at issues to do with the uncertainties associated with gene testing (e.g. should tests be performed when there is no treatment available?), at conceptual issues such as the implications for ascribing responsibility for behaviours that have a strong genetic basis (nature/nurture debate) and at the commercialisation of products resulting from research based on the HGP (e.g. will patenting DNA sequences limit their availability?).

The results of the HGP are likely to have a dramatic impact upon research into the future, with continued benefits in medical and behavioural science (e.g. identifying the genetic abnormalities seen in major types of cancer; National Institutes of Health, 2010). The results will also contribute to non-human research using the new techniques developed for the HGP in areas such as agricultural and environmental research (e.g. improved crops/livestock and better methods for cleaning up toxic materials).

the team established that serotonin 2A receptor gene is also associated with susceptibility to bipolar disease (McAuley et al., 2009).

It used to be thought that many mental illnesses, such as schizophrenia, autism and some forms of depression, were environmentally caused, and these were often attributed to poor parenting, but it has now been demonstrated that there is a strong genetic contribution to many of these disorders. For most, the gene(s) responsible have not yet been identified, but the race is on to locate them and Australian researchers are prominent in the search.

Just as genes contribute to disorders such as dementia, mental retardation and mental illness, they also play an important role in the normal variation in attributes such as cognitive ability, personality, school achievement and self-esteem. An understanding of the basic principles of genetics is essential to understanding the origins of behaviour.

PRINCIPLES OF GENETIC INHERITANCE

LO 2.1

Describe the basic principles of genetic transmission of normal and abnormal human attributes

The beginnings of modern genetic theory lie with the insights of Gregor Mendel who, in 1865, reported the findings of his extensive research program on the inheritance of colour and other attributes in flowering sweet peas. Mendel introduced a number of ideas that formed the basis of modern genetic theory:

- inheritance via 'factors' (now called genes);
- that genes come in pairs (one gene contributed by each parent);
- the notion of dominant and recessive genes; and
- the law of segregation (meiosis).

genes
Units of hereditary information that act as a blueprint for cells to synthesise the enzymes and proteins that build and regulate the body.

Genes synthesise the enzymes and proteins that build and regulate the body. They determine the type of body cell that will develop (bone, muscle, brain, etc.) and the timing of this development during ontogenesis. They also have long-term influences, such as switching on/off puberty and menopause and determining longevity.

chromosomes
Strings of deoxyribonucleic acid (DNA) in which is coded the genetic information in sequences of three of the four chemical bases adenine, thymine, guanine and cytosine. Humans have 23 pairs of chromosomes, one member of each pair coming from each parent.

We now know that the genes are carried on **chromosomes**. Chromosomes are made up of strings of deoxyribonucleic acid (DNA) and are located in the nucleus of every cell in the body. Genetic information is coded in DNA in sequences of three of the four chemical bases adenine, thymine, guanine and cytosine. Each three-base sequence is known as a **codon** because it is the code for one of the 20 amino acids that make up the thousands of specific proteins and enzymes that are the building blocks of living organisms. Each gene is a long sequence of these three-base codes.

codon
A three-base sequence that is the code for one of the 20 amino acids that make up the thousands of specific proteins and enzymes that are the building blocks of living organisms. Each gene is a long sequence of these three-base codes.

Each species has its own number of chromosomes. In humans the normal complement is 46 chromosomes, or 23 pairs, made up of a pair of **sex chromosomes** which determine the sex of the individual and 22 pairs of **autosomes** which carry the genetic information for almost all the other characteristics of the individual. Each chromosome is uniquely identifiable by microscopic examination, and geneticists refer to them by number (1 to 22 for the autosomes) or by their characteristic shape (X or Y for the sex chromosomes). As a result of the Human Genome Project we now have a map of the genes on the chromosomes (although we are a long way from knowing what all the genes do).

sex chromosomes
A single pair of chromosomes which determine the sex of the individual.

The replication of genetic information during cell multiplication and the transfer of genetic information during reproduction are processes central to an understanding of development and growth. The two basic processes are mitosis, the process involved in normal body cell replication, and meiosis, the process involved in germ cell (ova/sperm) production. Errors during meiosis are the source of many of the genetically related abnormalities.

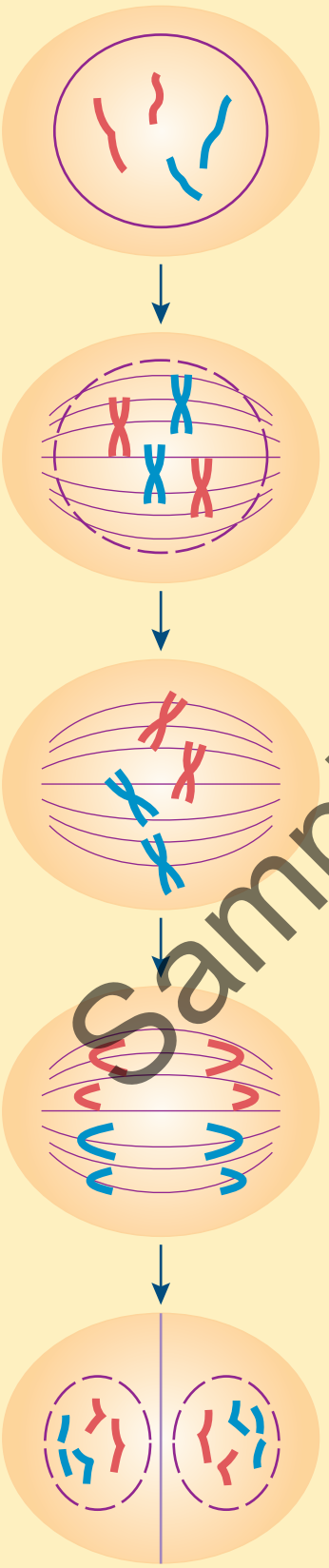
autosomes
The 22 pairs of chromosomes (other than the sex chromosomes) which carry the genetic information for almost all of the other characteristics of the individual.

Mitosis is the process that ensures that a duplicate cell is identical in genetic make-up to the original. It occurs in the normal cell replication process involved in growth and body maintenance. A simplified version of this process is shown in Figure 2.1. More detail can be obtained from genetics texts or from websites such as <www.accessexcellence.org/AB/GG/mitosis.html>.

Meiosis is essentially a process of reduction/division. In the ovaries and testes a cell with 23 pairs of chromosomes divides in such a way that the resultant cells (eggs and sperm) include only one member of each chromosome pair. This ensures that at fertilisation, when the egg and sperm unite, the fertilised ovum (**zygote**) contains the normal 23 pairs, with half of each pair contributed

FIGURE 2.1

A simplified description of mitosis



Parent cell – showing two of the 23 pairs of chromosomes

Step 1 (prophase) – the four chromosomes replicate (split lengthwise)

Step 2 (metaphase) – the four replicated (but still joined) chromosomes line up at the cell midline

Step 3 (anaphase) – the replicated chromosomes separate and migrate to opposite poles of the cell

Step 4 (telophase) – two identical 'daughter' cells are formed when the parent cell divides in two

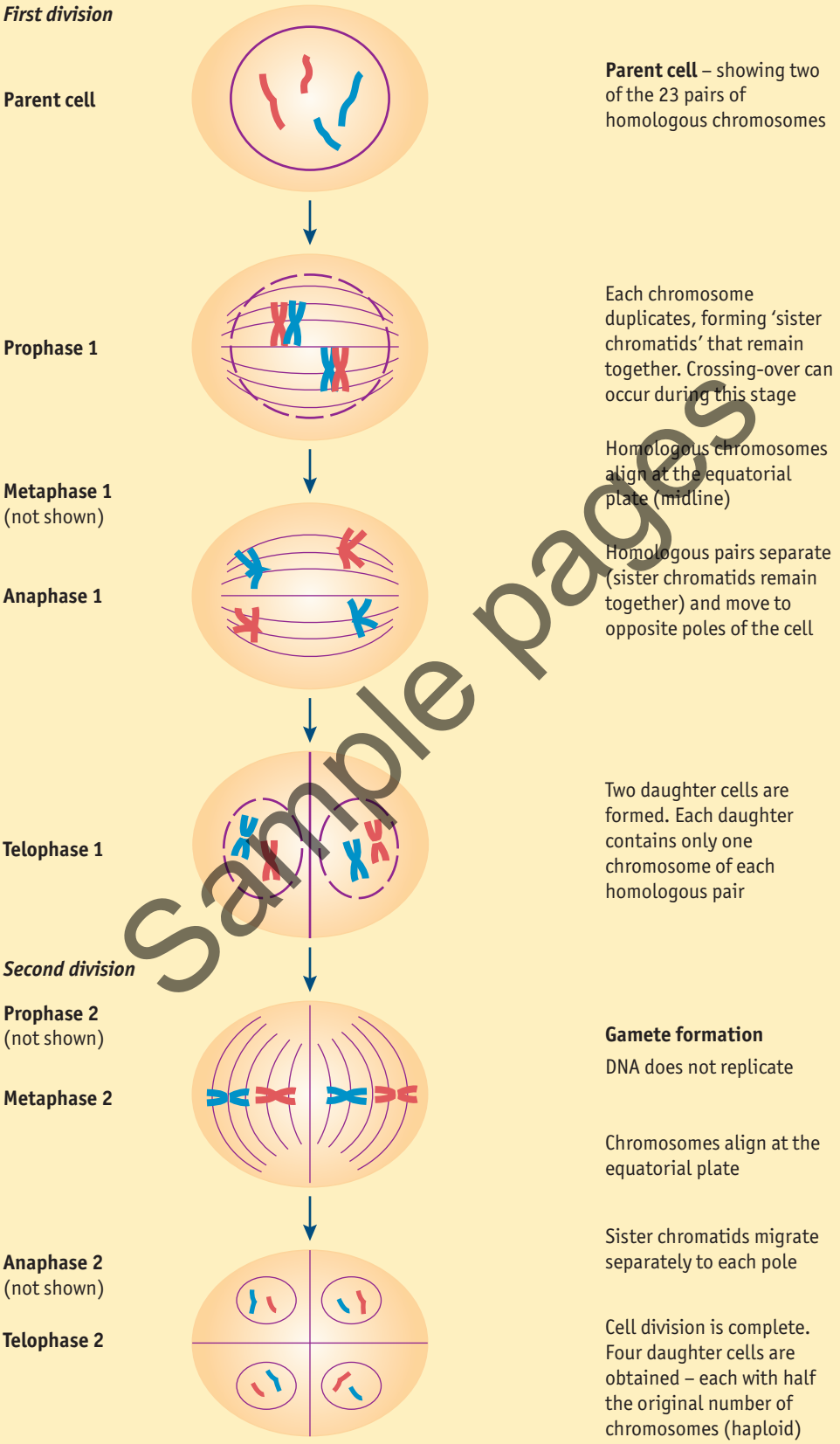
mitosis
The process involved in normal body cell replication – the process that ensures that a duplicate cell is identical in genetic make-up to the original.

meiosis
The process involved in germ cell (ova/sperm) production – a process of reduction/division which ensures that at fertilisation, when the egg and sperm unite, the fertilised ovum contains the normal 23 pairs of chromosomes.

zygote
A single cell formed by the fertilisation of an ovum by a sperm.

FIGURE 2.2

A simplified description of meiosis



by each parent. A simplified version of this process is shown in Figure 2.2. More detail can be obtained from genetics texts or from websites such as <www.accessexcellence.org/AB/GG/meiosis.html>.

Each egg and sperm has one of over eight million possible combinations of the 23 chromosome pairs. This genetic variability is further increased during meiosis by a phenomenon called **crossing-over**, where genetic material is exchanged between pairs of matching chromosomes (one from each parent). Such matching pairs are referred to as **homologous chromosomes**. At the beginning of meiosis, when the pairs of homologous chromosomes are aligned, equivalent sections of each chromosome in the pair break away and reattach to the other member of the pair. This process is shown in Figure 2.3.

Once fertilisation takes place the new zygote begins to divide and multiply by the process of mitosis, ensuring that each cell in the body carries the same genetic code as the original cell.

In Gregor Mendel's original work, many of the attributes he studied were ones in which a single pair of genes seemed largely responsible for the characteristic (e.g. flower colour). He discovered that the combination of genes in the gene pair (the **genotype**) determined the way the characteristic was expressed (the **phenotype**). It is now recognised that most human psychological characteristics (such as intelligence or personality) are determined by more than one pair of genes (**polygenically determined**). However, a number of physical traits (e.g. eye colour, blood type, curliness of hair) are influenced by a single pair of genes and a number of heritable diseases are attributable to single gene pairs. The phenotype expressed is the result of the characteristics of the pair of genes at the particular position on the chromosome (**gene locus**).

crossing-over
The process by which genetic material is exchanged between pairs of matching chromosomes.

homologous chromosomes
Matching chromosomes (one from each parent).

genotype
The genetic make-up (types of genes carried on the pairs of chromosomes) of the individual.

phenotype
The way the genotype expresses itself as a characteristic of the individual (e.g. hair colour, blood type).

polygenically determined characteristics
Characteristics (intelligence, personality, etc.) that are determined by more than one pair of genes.

gene locus
A particular position on a chromosome.

FIGURE 2.3

A simplified description of the process of crossing-over

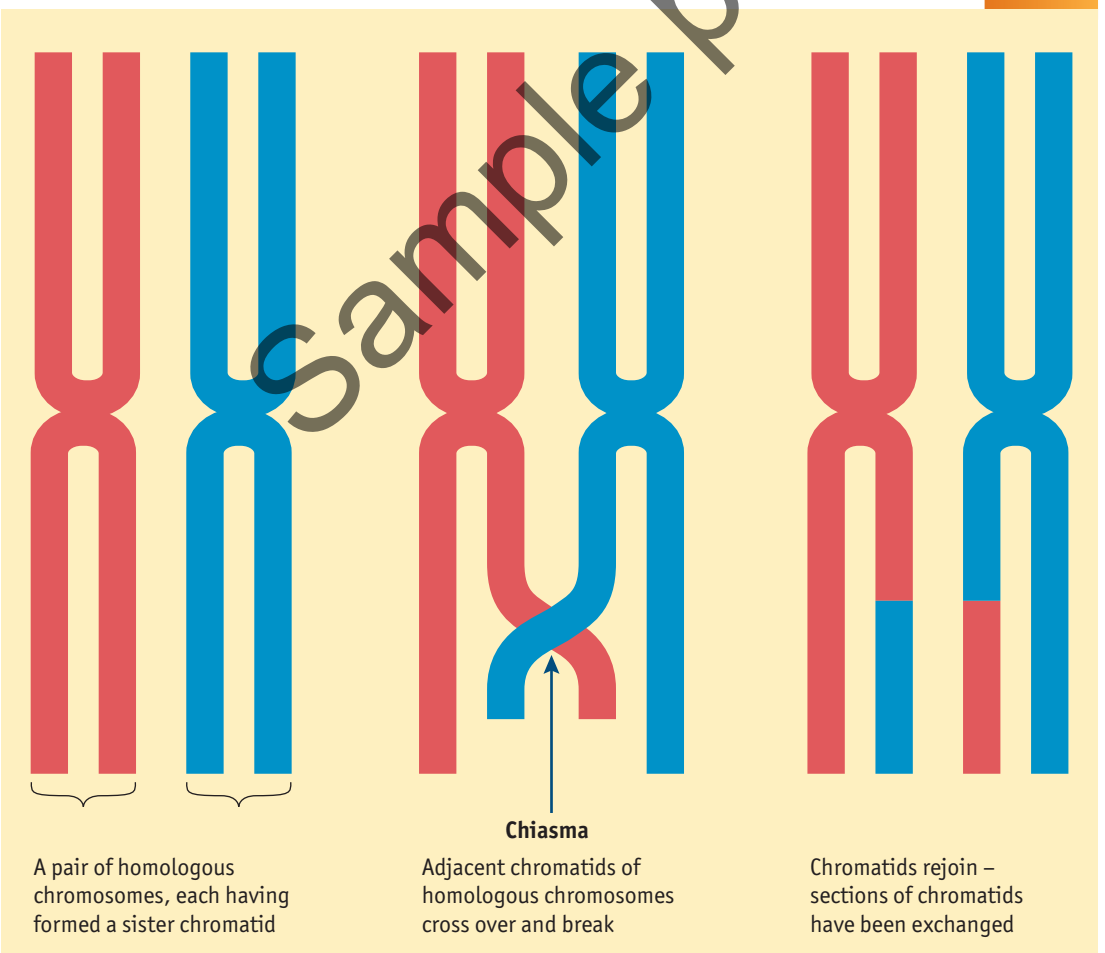
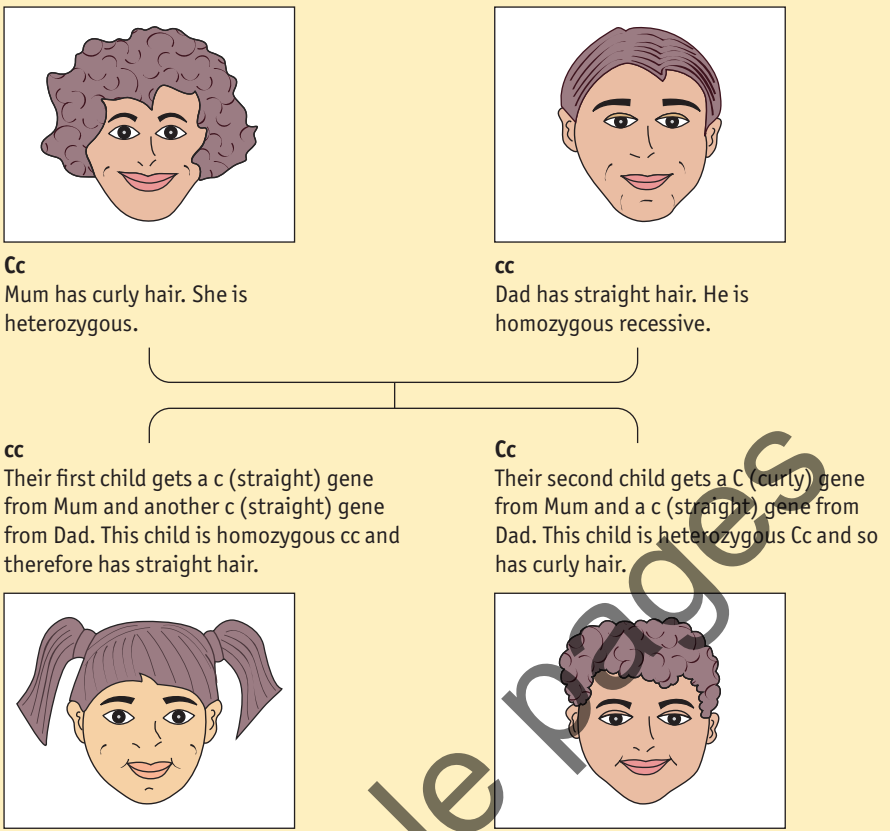


FIGURE 2.4

Inheritance of hair type



alleles
Alternative forms of a gene.

homozygous
When the alleles on the homologous chromosomes from both parents are the same.

heterozygous
When the alleles from the two parents are different.

dominant genes
Genes which express in the phenotype even in the heterozygous state.

recessive genes
Genes which express in the phenotype only when homozygous for that allele.

At each gene locus there may be two (or more) alternative forms of the gene (**alleles**). If the alleles from both parents are the same, the child is **homozygous** at that locus. If the alleles from the two parents are different, the child is **heterozygous** at that locus. An example, as shown in Figure 2.4, is that of curly versus straight hair type. If a child has the curly-haired allele from both parents she is homozygous for curly hair and will be phenotypically curly-haired. Similarly, if she receives the straight-hair allele from both parents she will be homozygous for straight hair and will have straight hair. However, if she receives the curly allele from one parent and the straight allele from the other, she will be heterozygous for hair type. What hair type will she have? In this case, the gene for curly hair is **dominant** over that for straight hair (the gene for straight hair is **recessive**); hence, the child will have curly hair.

Dominant–recessive relationships

Some of the characteristics determined by single-gene-pair dominant and recessive genes are shown in Table 2.1. As this list shows, a number of disorders are caused by genes at a single locus. Whether the disorder is expressed in the phenotype depends on whether they are dominant or recessive and present in the heterozygous or homozygous form.

Disorders caused by dominant genes

Disorders that are dominant traits express in every individual carrying the allele, whether heterozygous or homozygous. Dominant autosomal genes that cause severe (life-threatening) problems are likely to disappear from the gene pool, as those carrying the gene are unlikely to reproduce. In a few cases, severely disabling dominant genes are passed on because they do not become active until relatively late in the affected individual's life.

TABLE 2.1

Some common single-gene-pair dominant and recessive characteristics

DOMINANT	RECESSIVE
Curly hair	Straight hair
Brown eyes	Grey or green eyes
Normal hair	Baldness
Dark hair	Light or blonde hair
Thick lips	Thin lips
Roman nose	Straight nose
Double-jointedness	Normal joints
Normally pigmented skin	Albinism
Facial dimples	No dimples
Normal vision	Myopia, nearsightedness
Farsightedness	Normal vision
Normal colour vision	Red–green colour blindness
Type A blood	Type O blood
Type B blood	Type O blood
Rh positive blood	Rh negative blood
Normal respiratory and gastrointestinal function	Cystic fibrosis
Normal blood clotting	Haemophilia
Normal metabolism	Phenylketonuria (PKU)
Normal red blood cells	Sickle cell anaemia
Normal central nervous system (CNS) development	Tay-Sachs disease
Huntington's disease	Normal CNS function in adulthood

An example of such a trait is **Huntington's disease**. This disorder is rare, affecting one Australian in 17 000. The age of onset of this disease varies, but it usually strikes after 40 years of age. Quite suddenly the nervous system begins to deteriorate, resulting in uncontrollable muscular movements and disordered brain function. There is gradual deterioration, both physically and behaviourally, until death, usually within 10 to 15 years. In 1983 the gene responsible for Huntington's disease was identified on chromosome 4, and there is now a test for determining whether a person has inherited the gene. People at risk know that they are at risk because a parent developed the disease. Being a dominant trait, all carriers develop the disease. Unfortunately, there is no cure, and this raises a dilemma for those who are at risk. A detailed protocol has been developed for those at risk to decide if they want to know the result of the test.

Huntington's disease
A disorder caused by a dominant gene resulting in nervous system deterioration (usually after 40 years of age).

While dominant disorders are readily detected, the situation is not as simple for recessive disorders, which express only when the individual is homozygous for the recessive allele; hence, the disorder may skip generations.

Disorders caused by recessive genes

A number of diseases produced by recessive genes result in inborn errors of metabolism. One such disease is **phenylketonuria** (PKU). Phenylketonuria is the most widely cited example of a recessive disorder, one for which all newborn infants are routinely screened. It is caused by a single autosomal

phenylketonuria (PKU)
A disorder caused by a recessive gene resulting in an inability to metabolise phenylalanine in individuals who are homozygous.

recessive gene which, in homozygous individuals, results in an inability to properly metabolise phenylalanine, an amino acid found in many foods. These people accumulate phenylalanine in their blood, and this results in the symptoms of the disease. Symptoms include severe cognitive deficits, convulsive disorders, exaggerated reflexive responses, postural abnormalities and a short lifespan (usually less than 30 years). When PKU is detected the child is placed on a phenylalanine-free diet.

Phenylketonuria is often used as an example of how the environment can be manipulated to alleviate a genetic problem. However, given the wide range of foods containing phenylalanine it is often very difficult for children to maintain this regimen. Phenylalanine is contained in most protein-rich foods, including dairy products, peanuts, almonds, seeds, avocados and lima beans.

STOP & REVIEW

A dominant gene determines the presence of facial dimples. Jane's father has dimples, but neither Jane's mother nor her sister has them. What are the odds that Jane has facial dimples?

Other diseases produced by recessive genes that result in metabolic errors include Tay-Sachs disease and cystic fibrosis.

Co-dominance

co-dominance
Where two different alleles are present and both influence the characteristic.

sickle cell anaemia
A recessive disorder affecting the shape of red blood cells that manifests in homozygous recessive individuals. It occurs most often in people of African descent.

carriers
(In genetics) Heterozygous individuals carrying a recessive gene (e.g. for a disorder such as PKU).

In some cases the typical dominant–recessive relationship does not hold. It is replaced by a situation in which both alleles influence the characteristic. This is called **co-dominance**. Sickle cell trait is an example of such co-dominance. **Sickle cell anaemia** is a recessive disorder that manifests in homozygous recessive individuals. It causes the usually round blood cells to become sickle-shaped, especially under low-oxygen conditions. The sickled cells clog the blood vessels, blocking blood flow and causing tissue damage, swelling and intense pain. Such individuals typically have a much reduced lifespan, with only about 50 per cent surviving to 40 years and only 1 per cent to 60 years of age (Ashley-Koch, Yang, & Olney, 2000). In the heterozygous state, the sickle cell allele still affects the phenotype.

The allele is particularly prevalent in populations from areas in which malaria is common. Those who are heterozygous for the recessive gene (**carriers**) are more resistant to malaria than those who are homozygous for the gene for normal round blood cells. These carriers thus survive and reproduce more frequently than others, ensuring that the sickle cell allele persists in the population. In areas where the risk of malaria is low, there is no advantage for carriers and the frequency of the gene is steadily declining (e.g. the US National Heart, Lung, and Blood Institute (2011) reports that only 8 per cent of African Americans carry the allele; for Indigenous Africans, the figure is 20 per cent).

Another example of a characteristic that demonstrates co-dominance is AB blood type, where AB individuals have both A-antigens and B-antigens in their bloodstream.

Sex-linked traits

sex-linked genes
Genes found only on the sex chromosomes.

An apparent exception to Mendel's laws appears in a group of characteristics that are determined by genes found only on the sex chromosomes. These characteristics do not follow the classic Mendelian inheritance pattern but depend on whether the mother or the father displayed the characteristic. Such traits often skip generations. They are called **sex-linked genes**. The vast majority of these sex-linked traits are produced by recessive genes that are found only on X chromosomes (X-linked). The Y chromosome is the smallest of the chromosomes and carries very few genes other than those responsible for maleness.

The inheritance patterns for sex-linked attributes differ according to the child's sex. Females have two X chromosomes (one from each parent), while males have an X (from the mother) and a Y (from the father). If a girl inherits a recessive X-linked trait from one parent, it is usually blocked

by a dominant gene on the X chromosome from the other parent and hence the child has a normal phenotype (but is a carrier of the recessive gene). Only when homozygous for the recessive gene will girls have the recessive attribute. When boys receive a sex-linked recessive gene from their mother, there is no matching gene on the Y from their father that might counter the effect of the recessive gene, so the trait is expressed. For this reason, sex-linked traits are much more common in males than in females.

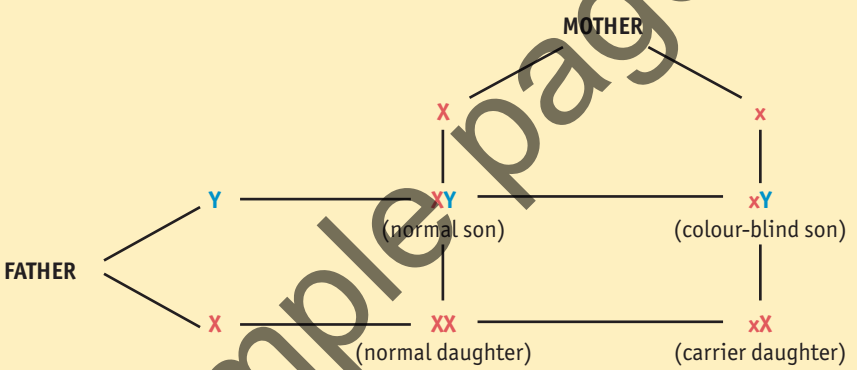
There are more than 100 sex-linked characteristics, and most are disabling (Plomin et al., 2013). These include haemophilia, two kinds of muscular dystrophy, and forms of deafness and night blindness. One familiar sex-linked trait that is troublesome rather than disabling is red-green colour blindness. Most of us know someone (almost always male) who is colour blind. Figure 2.5 shows the way in which red-green colour blindness is passed on.

In this example, where the mother is a carrier and the father has normal colour vision, there is a 50 per cent chance of sons being colour blind and a 50 per cent chance of daughters being carriers.

FIGURE 2.5

Sex-linked inheritance of red-green colour blindness

Red-green colour blindness is a sex-linked trait that is usually passed onto sons from a carrier mother in the following manner:



X = X chromosome carrying the dominant gene for normal colour vision
x = X chromosome carrying the recessive gene for colour blindness
Y = Y chromosome (carries no genes for colour vision)

STOP & REVIEW

What would be the probability of a daughter being colour blind if the father was colour blind and the mother was a carrier?

DNA repeat sequences

In recent years, other genetic phenomena that do not appear to conform to Mendel's laws (in the sense that they are not inherited in a simple way through the generations) have been described. One such phenomenon is caused by DNA repeat sequences.

In many instances the sequence of codons that makes up a gene carries repeats of a short sequence of code (2, 3 or 4 codons). Sometimes, during cell replication, these repeat sequences grow, repeating many more times than is normal, and this can cause problems. The severity of the problem is related to the number of repeats.

The most common of such problems is Fragile-X syndrome (mentioned at the beginning of the chapter). This results from abnormal repeats on the X chromosome in women. In some families the X repeat sequence expands over generations. Up to 1 per cent of women have an abnormal number of repeats. In the general population the number of repeats is typically less than 60, but in some women the number rises from generation to generation until the full form of the disorder is manifest when the number exceeds 200 (Hay, 2008). The defective gene at the repeat site expresses when it is passed on from mother to child. A major effect of Fragile-X is mental retardation, and the extent increases as the number of repeats grows.

Early research into this effect by Australian researchers (Hay, 1994; Hay & Loesch, 1989) found that the effects are quite specific, with some aspects of intelligence being much more affected than others. They report that it is not unusual to find differences of 30 or more IQ points between verbal and nonverbal performance, the latter being much poorer. Recent research has attempted to look more closely at the cognitive impairments associated with Fragile-X. Cornish and colleagues (2008) found that an inhibitory deficit is associated with the disorder, while Krueger, Osterweil, Chen, Tye & Bear (2011) found a deficit in cognitive flexibility. Such deficits point to abnormal frontal-lobe function, a conclusion supported by the findings of Peng, Kelly, Quinton, Ramon, Thompson and Reiss (2014).

Being a sex-linked trait, Fragile-X is much more common in boys than in girls. Girls who inherit an affected X chromosome from their mother usually have an unaffected X from the father which counters the effect of the abnormal chromosome. Boys have only the X from their mother.

Genetic imprinting

genetic imprinting
Where the expression of an allele depends on whether it is inherited from the mother or the father.

Another exception to Mendel's laws is called **genetic imprinting,** where the expression of an allele depends on whether it is inherited from the mother or the father. Diabetes, for example, is more likely to occur in children whose fathers suffer from the disease than in children whose mothers have the disease. A striking example of such imprinting involves deletions of a small part of chromosome 15 that leads to two very different disorders, depending on whether a deletion is inherited from the mother or the father. If from the mother, it results in a group of problems including severe mental retardation and is called *Angelman syndrome*. If inherited from the father, it causes other behavioural problems such as overeating, temper tantrums and depression, as well as physical problems such as obesity and short stature. This is called *Prader-Willi syndrome*.

LO 2.2
Discuss disorders associated with chromosomal abnormalities

CHROMOSOMAL ABNORMALITIES

Accidents during meiosis can disrupt the normal configuration of the chromosomes. If the disruption is too great, development cannot proceed (as evidenced by the high rate of chromosome abnormalities in miscarriages). However, a number of disorders have been identified as resulting from chromosomal disruption, and these can affect the autosomes or the sex chromosomes.

Autosome abnormality

non-disjunction
Failure of a chromosome pair to separate during meiosis.

One of the earliest identified and the most common autosomal disorder is Down syndrome, which occurs in one out of 800 live births. The most frequent form is Trisomy 21, where there is an extra copy of chromosome 21, one of the smallest of the autosomes. This arises from the failure of the chromosome 21 pair to separate during meiosis (**non-disjunction**). Characteristics of this disorder are mental retardation (average IQ 55), memory and speech problems, limited vocabulary and slow motor development, as well as distinct physical features that include a short, stocky build, a flattened face, a protruding tongue, almond-shaped eyes and an unusual crease running across the palm of the hand. Additionally, children with Down syndrome often have congenital eye, ear, heart and intestinal defects.

Although intellectually impaired, and more difficult to care for than unaffected children, those with Down syndrome often reach many of the same developmental milestones as unaffected

children, but at a slower pace (Fidler, Most & Philofsky, 2009). Most learn to care for their basic needs, and some even learn to read and write. Developmental progress appears to be best when family members provide them with high levels of stimulation and emotional support.

Down syndrome is more common in children of older mothers. This has long been thought to reflect a gender difference in the way in which the eggs and sperm are produced. Sperm production continues throughout the life of an adult male, while egg production is thought to be fairly complete before birth. Thus the older a woman is when she ovulates, the older the egg at her child's conception. It is believed that, as eggs age, they become more prone to errors in cell division, and the chromosomes are more likely to fail to separate properly as they complete the process of meiosis. Recent research on animals has questioned this explanation. Researchers at Harvard Medical School have shown that female mice produce new eggs well into adulthood. Another explanation for the higher rate of Down syndrome in older mothers is that the ability to reject and abort abnormal foetuses may diminish with age (O'Rahilly & Muller, 2001). Hence, foetuses with chromosomal abnormalities such as trisomy, which in younger women might be naturally aborted, may in older women proceed to full term and birth.

Down syndrome is a rare example of an autosome disorder. In most cases, abnormal autosomes result in severe disruption to development and miscarriage occurs. In contrast, individuals with abnormal sex chromosomes are much more likely to develop to term and to develop fewer problems. In some cases, sex chromosome disorders may not be detected until adolescence.

Abnormalities of the sex chromosomes

Abnormalities are usually the result of non-disjunction during meiosis so that germ cells carry abnormal numbers of the sex chromosome. Hence, at fertilisation, the resultant zygote could be XO (Turner's syndrome), XXX (Poly-X syndrome), XXY (Klinefelter's syndrome) or XYY ('super-male' syndrome). The typical characteristics associated with these syndromes are shown in Table 2.2. Most children with sex chromosome disorders do not suffer from mental retardation. Instead, their intellectual problems tend to be very specific. An extra X chromosome (Triple-X girls and Klinefelter's boys) produces a tendency for verbal problems (reading and vocabulary), while girls lacking an X (Turner's syndrome) are likely to have spatial problems (drawing pictures, telling right from left, etc.). The reason for this difference is still unknown.

Syndromes associated with sex chromosome abnormalities: Typical characteristics and incidence TABLE 2.2

SYNDROME	TYPICAL CHARACTERISTICS	INCIDENCE
Turner's syndrome (XO)	Female, short stature, webbed neck, heavy build, infertility, failure to develop sexually at puberty (may require hormone treatment), tendency for poor memory and spatial ability (may require special education)	1 in 5000 female births
Poly-X syndrome (XXX)	Female, very tall, normal sexual development and fertility, tend to have impaired verbal intelligence (may require special education)	1 in 1000 female births
Klinefelter's syndrome (XXY)	Male, very tall, sterile, female body type associated with low levels of male hormones (may require hormone therapy at puberty), atypical sexual development, impaired verbal intelligence is likely (may require special education)	1 in 900 male births
XYY syndrome (XYY)	Male, unusually tall, normal sexual development and fertility, near-normal intelligence	1 in 1000 male births

STOP & REVIEW

Given what you now know about the sex of individuals with different numbers of X and Y chromosomes, what is it that determines whether a person will be male or female?

LO 2.3

Discuss the ways in which psychologists employ behaviour genetics to distinguish between inherited and experiential factors in development

POLYGENIC INHERITANCE: THE ROLE OF BEHAVIOUR GENETICS

The study of single-gene disorders and disorders arising from identifiable chromosomal abnormalities provides clear evidence that there is a strong genetic contribution to human attributes. However, most of the attributes of concern to psychologists – attributes to do with behaviour – are not determined by genes at a single locus but are polygenically determined. Many genes (mostly unidentified) are thought to contribute to the outcome, and multiple environmental influences are usually also involved. This raises the question of how significant a role is played by heredity in such complex behavioural traits. How are the contributions of genes and environment differentiated?

behavioural genetics
The study of the contributions of nature and nurture to behavioural diversity.

heritability
The proportion of the difference between people that is due to genetic rather than environmental factors.

Behavioural genetics is the study of the contributions of nature and nurture to behavioural diversity. The term **heritability** is often used to summarise what proportion of the total difference between people is due to genetic rather than environmental factors. How do we determine the heritability of any behavioural trait?

strain comparison studies
Studies comparing the behaviour of different strains of animals that have been inbred for a particular characteristic.

selective breeding
Breeding for a particular characteristic by crossing animals with that characteristic (e.g. maze-bright rats).

Experimental studies of the inheritance of behavioural traits have necessarily (for practical and ethical reasons) involved animal studies. These have included **strain comparison studies** of inbred strains and **selective breeding** experiments such as the classic study by Tryon (1942), in which he showed that maze-learning ability is a heritable attribute in rats. He selected 'maze-bright' rats and 'maze-dull' rats; then, across many generations, he mated bright rats with bright rats and dull rats with dull rats. The difference in maze-learning ability of the two groups became progressively greater across generations. Similarly, people have for centuries been breeding dogs for particular temperaments and behaviours (e.g. terriers for aggressive/hunting behaviour, spaniels as non-aggressive companions, sheepdogs for complex human-supervised tasks). Such animal studies provide powerful evidence for the importance of genetic influence on behaviour.

We cannot perform selective breeding experiments with humans, so the field of human behavioural genetics has to rely on correlational methods rather than experiments. The method is to compare individuals who are more or less related genetically and who are raised in more or less similar environments on particular attributes (such as intelligence or personality). If the attributes are heritable, the similarity between any two individuals who live in the same environment should increase as a function of their **kinship** (the extent to which they have the same genes).

kinship
The extent to which individuals have the same genetic make-up.

Twin studies

monozygotic twins
Twins that develop from a single fertilised egg that splits into two identical replicas (identical twins).

dizygotic twins
Twins that are the product of the independent fertilisation of two eggs by two sperm and hence share only half their genes (fraternal twins).

Identical or **monozygotic twins** result from the splitting of a single fertilised egg; hence, they have identical genetic make-up. Fraternal or **dizygotic twins** are the product of the independent fertilisation of two eggs by two sperm and hence share only half their genes. Dizygotic twins are no more similar genetically than non-twin siblings. However, the environments experienced by twins who are reared together are likely to be more similar than for siblings who are of different ages. If aspects of behaviour (e.g. personality, IQ, cognitive ability) are heritable, we would expect monozygotic twins to be more similar than same-sex dizygotic twins on such behaviour. On the other hand, if monozygotic twins differ consistently from their co-twin on particular attributes, it would indicate that such attributes are governed largely by environmental factors. Similarly, if dizygotic twins are more alike than non-twin siblings, this points to an environmental effect.

The twin comparison methodology has been employed by many researchers over several decades as a means of examining the heritability of cognitive and personality variables. They

STOP & REVIEW

In what ways might the environments experienced by twins be more similar than for siblings of different ages?

typically administer standardised tests and/or inventories to pairs of monozygotic and dizygotic twins and compare co-twin scores on those measures to evaluate differences between them. By comparing the correlations between monozygotic co-twin scores and the correlations between dizygotic co-twin scores, an estimate of the heritability of the characteristic can be made. Examination of a number of such studies (e.g. Haworth et al., 2009; Bleidorn, Kandler, Riemann, Angleitner & Spinath, 2009) shows that, for monozygotic twins, the correlations between IQ scores for twin pairs is about 0.85 and for personality about 0.50; for dizygotic twins, these values are 0.55 and 0.30, respectively. These results suggest a genetic role in both characteristics.

The genetic contribution to such behavioural characteristics is often expressed as a **heritability coefficient** (H), which is calculated by using the formula:

heritability coefficient
A numerical estimate (ranging from .00 to 1.00) of the heritability of an attribute.

$$H = 2\{r\ (MZ\ pairs) - r\ (DZ\ pairs)\}.$$

Hence, based on the above results, the heritability for intelligence is: $H = 2(0.85 - 0.55) = 0.60$; and for personality is: $H = 2(0.50 - 0.30) = 0.40$. It is clear that, while genetics contribute significantly to these characteristics, the environment also plays a crucial role in these behavioural traits and that this role is greater for personality than for IQ.

A basic assumption is made when calculating heritability in this manner. It is assumed that the environmentally induced differences between identical twin pairs are no different from the environmentally induced differences between same-sex fraternal twin pairs (i.e. identical and same-sex fraternal twin pairs are treated in the same manner by those around them). This assumption has been questioned. It has been pointed out that identical twins are expected by those around them, including parents, to behave more similarly than non-identical twins, and that these differences in expectations are likely to influence the way in which the twins are treated. They are more likely to spend time together, to dress alike, to play together, to share a bedroom and to be treated alike by their parents (Plomin et al., 2013).

Hence, there is some evidence that the assumption of equivalent environmental influence cannot be supported, and this brings into question heritability estimates. This view is supported by researchers such as Richardson and Norgate (2005), who suggest that there is a need for more thorough examination of such confounding effects in the interpretation of twin data. Perhaps identical twins are treated more similarly and therefore behave more similarly, rather than the converse.

The ultimate check of the strength of inherited factors would seem to be to compare identical twins who have been separated and brought up in different family environments. Bouchard, Lykken, McGue, Segal and Tellegen (1990) located over 100 such twin pair adults who had been separated in infancy and measured them on standard IQ tests. They found that the correlation between twin pairs was 0.76. While this is somewhat lower than that found for monozygotic twins reared together ($r = 0.85$), it was nevertheless higher than found for dizygotic twins reared together ($r = 0.55$). This provided strong evidence that heredity contributes substantially to IQ and that the more similar environments normally experienced by monozygotic twins cannot account for the higher correlation between identical co-twins than between fraternal co-twins.

As pointed out by Busjahn and Hur (2006), twin research is proving increasingly valuable in studying the interplay of genes and environment. This contribution is in large part due to the establishment of large-scale twin registers in many countries, including Australia. The Australian Twin Registry has, since the late 1970s, enrolled more than 33 000 pairs of twins of all zygosity types and ages who are willing to participate in research studies (Hopper, Treloar, deKlerk & Morley, 2006). By 2010 this resource had facilitated more than 678 peer-reviewed studies using a variety of designs, including both longitudinal and intervention studies (Australian Twin Registry, 2011).

Adoption studies

Adoption produces family members who share the family environment but do not share a genetic background, and also genetically related individuals who do not share a common family environment. Examination of adoptive and non-adoptive (nuclear) families allows us to look at the correlations in behavioural measures between parents and their offspring, and between siblings at various levels of genetic and environmental relatedness: *genetic plus environmental* (related individuals living together as a nuclear family); *genetic only* (related individuals living separately); and *environmental only* (unrelated individuals living in the same family). For most psychological characteristics that have been assessed in this way, genetic factors appear to be important.

Table 2.3 illustrates the correlations in cognitive ability scores found by Loehlin, Horn and Willerman (1989) using an adoption design. This indicates that genetically related individuals show some resemblance with each other even if adopted and reared apart, and that genetics accounts for about half of the similarity between related individuals reared together in the same family (G + E).

TABLE 2.3 **Correlations in cognitive ability scores between: related individuals reared apart (G); unrelated individuals reared together (E); and related individuals reared together (G + E)**

	G+E	G	E
Parent and offspring	0.42	0.24	0.20
Siblings	0.47	0.24	0.32

Adoption studies also indicate that for many psychological traits other than cognitive ability, genetic factors play a greater role than environment in determining familial similarity (Plomin et al., 2013). This appeared to be the case for inheritance of schizophrenia in a classic adoption study by Heston (1966). He demonstrated that the risk of schizophrenia for the children of schizophrenic parents is just as great whether raised by those parents or by adoptive (non-schizophrenic) parents.

Plomin and colleagues (2013) discuss three potentially confounding factors that must be taken into consideration when assessing the results of adoption studies. The first of these is the representativeness of the biological parents, adoptive parents and adoptive children of the rest of the population. If not representative, the generalisability of the results is limited. Second, the common prenatal environment of adopted and non-adopted siblings may account for similarities between them (but such similarity might be interpreted as genetically based). A third source of possible confounding is the tendency for adoption agencies to selectively place adopted children (e.g. to match characteristics such as the educational and social background of the biological parents). This may introduce a bias towards similar genetic background and hence confound the interpretation of environmental contribution.

Despite such complications, adoption and other family studies are the only methods available to evaluate the relative contributions of environmental and genetic factors to complex human behaviour. Behavioural geneticists acknowledge that there are problems involved with these methods, whether twin studies, adoption studies or studies involving nuclear families. In recent years they have begun to use designs that combine the family, adoption and twin methods in order to bring more power to these analyses. The use of converging evidence from the different approaches, evaluating the consistency of findings across techniques, decreases the likelihood that the problems associated with each individual approach will lead to misinterpretation. The Colorado Adoption Project has been using such a combined design to examine behavioural development (Rhea, Bricker, Wadsworth & Corley, 2013). A finding from this long-running and large-scale study is that there is an increase in the genetic influence on cognitive ability during infancy and childhood.

The interaction of genotype and environment

It is clear that human development involves an interplay between heredity and environment and that this interplay is a complex and dynamic process. A number of concepts bear on this interplay.

Canalisation

canalisation
The process by which some characteristics are genetically restricted to a limited range of outcomes and follow a genetic growth plan. Only strong environmental forces can change these characteristics.

Some characteristics appear to be restricted genetically to a limited range of outcomes. A behaviour that is strongly canalised follows a genetic growth plan, and only strong environmental forces can change it. An example of a highly canalised human attribute is infant babbling. All infants, even deaf ones, babble in pretty much the same way over the first eight or so months of life. The environment appears to have little effect on this attribute. In contrast, attributes such as IQ, personality and temperament are much less canalised and more open to environmental influences.

Reaction range

reaction range
The genetically determined boundaries within which environmental influences might affect a particular attribute.

Gottesman (1963) used the term *reaction range* to describe how, for many attributes, the genes appear to set the boundaries within which environmental influences have their effects. Each individual reacts to the environment in a unique way because of their genetic make-up. Thus, depending on the level of appropriate environmental stimulation, a child might have a higher or lower IQ. The level of stimulation required to maximise IQ score will differ from individual to individual but will always be within the bounds set by their genetic endowment.

Niche-picking

niche-picking
Where individuals actively seek out environments that suit their genetic make-up.

This concept was introduced by Scarr and McCartney (1983) to describe how children actively seek out environments that complement their heredity. Thus, children are not simply passive recipients of environmental input but active seekers of the sort of environmental stimulation that suits their genetic make-up. For example, a child who has an active sociable temperament will seek out activities to match this temperament (e.g. sport, drama). A more introverted child will seek out activities that better suit their temperament (more solitary pursuits, e.g. reading and drawing). These environmental choices, initially influenced by genetic factors, will then affect the way development proceeds, as they dictate the types of interaction the child experiences. Hence, even children who appear to be experiencing the same environment (e.g. all in the same class at school or all in the same family) will have quite different experiences from this environment. This adds to the difficulty of studying environmental effects on development.

The difficulty (impossibility) of isolating the hereditary from the environmental factors contributing to development will ensure that the nature/nurture debate continues. It is clear, however, that both contribute to development and that this interrelationship is complex.

PRENATAL DEVELOPMENT

LO 2.4
Outline the time course of prenatal development

The period of prenatal development (between conception and birth) is the time of the most rapid and dramatic change in the entire human lifespan. At conception a single undifferentiated cell resulting from the fusion of the egg and sperm begins the rapid transition towards a fully formed human. At birth, nine months later, the individual consists of millions of cells, differentiated into all the specialised organs and structures of the body and functioning much as they will for the rest of life. The transition from single cell to functional human is influenced by numerous factors, including the genetic make-up of the foetus, the genetic make-up of the mother, and innumerable environmental factors that impinge on both of them. Whether or not the pregnancy will proceed to full term and, if so, whether the foetus will develop normally, is affected by these factors.

Three prenatal periods

germinal period (period of the zygote)
The prenatal period from conception until implantation of the zygote in the wall of the uterus by the end of the second week.

Prenatal development is typically divided into three periods, each of which is characterised by a distinctive pattern of growth and distinctive interaction between the developing organism and its environment. These three periods are the *germinal period*, from conception until implantation of the zygote in the wall of the uterus by the end of the second week; the *embryonic period*, from implantation to about the end of the eighth week; and the *foetal period*, from the ninth week until birth at about 38/39 weeks.

Germinal period

blastocyst
A ball of cells consisting of two layers of cells resulting from rapid cell replication after fertilisation.

trophoblast layer
The outer cell layer of the blastocyst, enclosing a fluid-filled cavity. It develops into tissues that support, protect and nourish the developing embryo.

embryonic disk
The inner cell layer of the blastocyst, containing the cells that will become the embryo.

implantation
The process by which the blastocyst attaches to the wall of the uterus and connects to the mother's blood vessels.

amnion
A membrane that grows from the trophoblast after the implantation of a fertilised egg in the uterus. It forms as a sac that fills with fluid from the mother's tissues (amniotic fluid).

amniotic fluid
Fluid that cushions a developing foetus from jolts, helps maintain constant temperature, and provides support and a medium in which the foetus can move.

yolk sac
The structure that produces blood cells until an embryo is capable of producing its own.

chorion
A membrane that forms around the amnion by the end of the second week after fertilisation of an egg. This becomes the foetal part of the placenta.

The **germinal period**, or **period of the zygote**, is a period of rapid cell replication (multiplication) so that by the time the zygote reaches the uterus it consists of hundreds of cells. By this time it is a ball of cells known as a **blastocyst**, consisting of two cell layers – an outer cell mass (the **trophoblast layer**) which encloses a fluid-filled cavity, and an inner cell mass called the **embryonic disk**. The trophoblast layer will develop into tissues that support, protect and nourish the developing embryo, while the embryonic disk contains the cells that will become the embryo. When the blastocyst reaches the uterus, trophoblast cells put out tiny branches that burrow into the spongy wall of the uterus until they come into contact with the maternal blood vessels. This is termed **implantation**.

Implantation is a significant step in development. Only about half of all fertilised ova become firmly implanted and, of these, as many as half are either genetically abnormal and fail to develop, or burrow into a site incapable of sustaining them and are miscarried (Suzumori & Sugiura-Ogasawara, 2010).

The development of support systems begins immediately after implantation. Membranes grow rapidly from the trophoblast layer to ensure that the developing embryo is provided with nutrients and protection from environmental trauma. An inner membrane, the **amnion**, forms as a watertight sac that fills with fluid from the mother's tissues (**amniotic fluid**). This cushions the developing organism from jolts as the mother moves around, helps to maintain constant temperature, and provides support and a medium in which it can move. Within the amniotic fluid a **yolk sac** forms, and this produces blood cells until the embryo is capable of producing its own (when the developing spleen, liver and bone marrow take over this function).

An outer membrane, the **chorion**, forms around the amnion by the end of the second week. This becomes the foetal part of the **placenta**, a complex organ made up of tissue from both the mother and the embryo. The placenta and embryo become linked by the umbilical cord, which is formed by another membrane, the **allantois**. These are shown in Figure 2.6.

Until birth the placenta acts simultaneously as a barrier that prevents the bloodstreams of the mother and embryo/foetus from coming into direct contact and as a filter that allows nutrients, oxygen and waste products to be exchanged.

Embryonic period

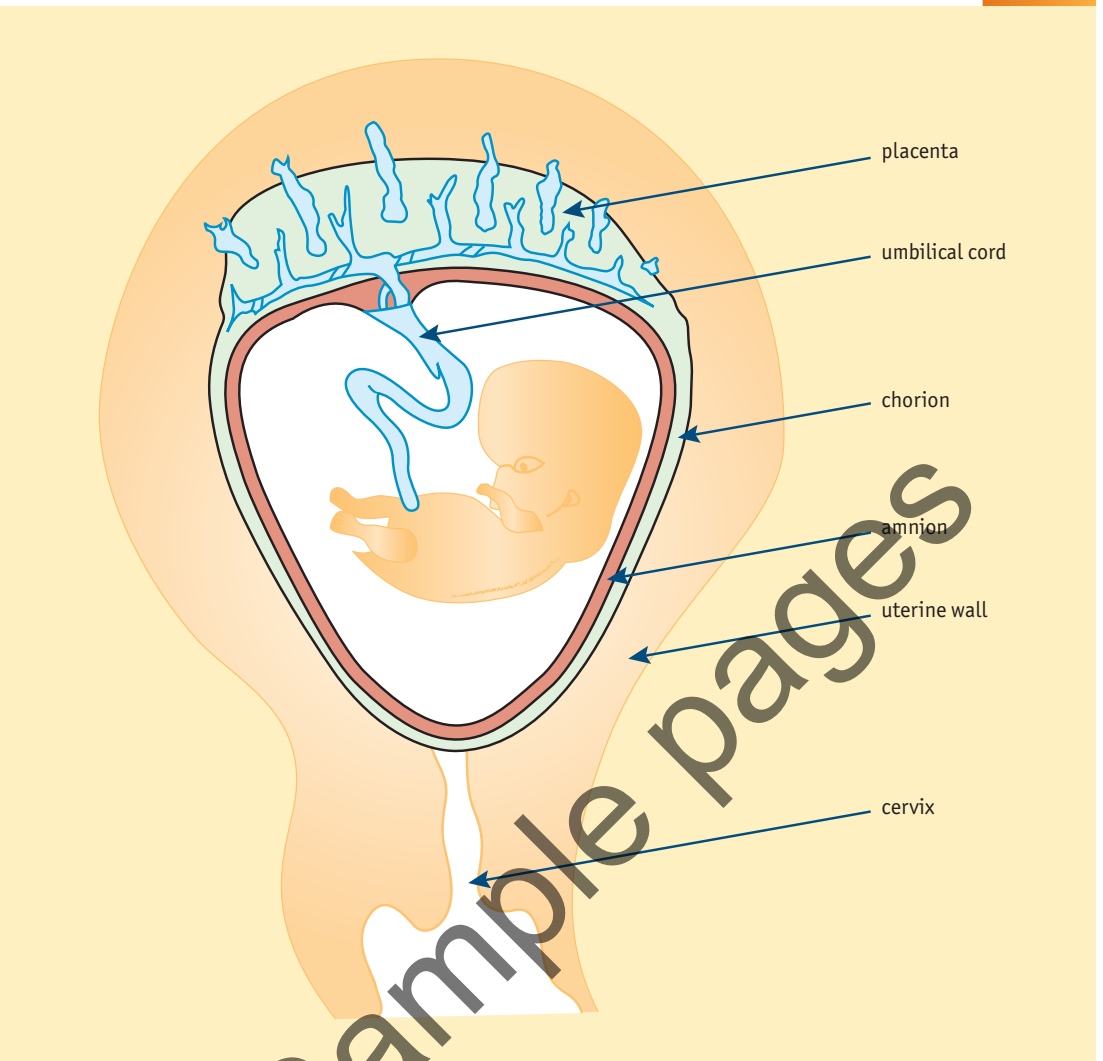
The **embryonic period**, or **period of the embryo**, lasts from implantation until about the end of the eighth week of gestation. It is a period when all the basic organs of the body are formed (**organogenesis**) and the embryo begins to respond to direct stimulation. This is a period of very rapid development.

By the third week after conception the embryonic disk is rapidly differentiating into three cell layers that will give rise to all parts of the body. The **ectoderm** will form the nervous system, skin and hair. The **mesoderm** will form the muscles, bones, circulatory system and other internal organs. The **endoderm** will form the digestive system, lungs, urinary tract and glands.

The first tissue to form is the neural tube, or primitive spinal cord, and by the end of the first month the brain has begun to develop, the heart has formed and begun to beat, muscles are forming, and the backbone, ribs and digestive system have begun to form. Limb buds, the beginnings of arms and legs, have also appeared. At this stage the embryo is about 6 millimetres long and consists of millions of cells organised into groups with specific functions.

FIGURE 2.6

Diagram of the developing embryo showing the cell layers and membranes



Rapid growth continues in the second month and the embryo becomes much more human in appearance. Eyes, ears, nose, mouth and neck develop, and limbs rapidly form from the limb buds. By the seventh week the embryo has a rudimentary skeleton and in the eighth week movement begins and bones begin to harden. By the end of the second month all the basic tissues and organs exist in rudimentary form. The brain can direct primitive muscle contractions, sexual development has begun, and the embryo's circulatory system is functioning on its own as the liver and spleen have begun producing blood cells. (The yolk sac is now defunct.)

The remaining seven months of gestation are devoted to refining, interconnecting and making functional the structures laid down in the first two months. This is the final period of prenatal development, the foetal period.

Foetal period

In the **foetal period**, or **period of the foetus**, the organs take on their final form and begin to function. (It is sometimes referred to as the period of **histogenesis**.)

The prenatal period is often divided into **trimesters** (three periods of three months each). The period of the foetus begins two-thirds of the way through the first trimester and occupies the remainder of the prenatal period.

placenta
A complex organ made up of tissue from both the mother and the embryo. It acts simultaneously as a barrier, preventing the bloodstreams of the mother and embryo/foetus from coming into direct contact, and as a filter, allowing nutrients, oxygen and waste products to be exchanged.

allantois
The foetal membrane that forms the umbilical cord.

embryonic period (period of the embryo)
The prenatal period from implantation until about the end of the eighth week of gestation.

organogenesis
The formation of the basic organs of the body in a developing embryo.

ectoderm
The outer cell layer of the embryonic disk that will form the nervous system, skin and hair.

mesoderm
The middle cell layer of the embryonic disk that will form the muscles, bones, circulatory system and other internal organs.

endoderm
The inner cell layer of the embryonic disk that will form the digestive system, lungs, urinary tract and glands.

foetal period (period of the foetus)
The prenatal period from the ninth week after conception until birth.

histogenesis
The development of function of the organs as they begin to sense, secrete, move, etc.

trimesters
The three periods of three months each into which the nine months of gestation are often divided when discussing prenatal development.

By the end of the first trimester (13 weeks) the foetus is about 75 millimetres long and weighs about 25 grams. The spinal cord is recognisable, the kidneys are able to secrete, the sexes are externally different and the foetus responds reflexively to a touch on the face. At this stage the head is about half the total size of the foetus.

By the end of the second trimester (about 26 weeks) the foetus is about 360 millimetres long and weighs about 900 grams. The mother's abdomen is distended and she can feel the foetus move, reporting kicks. Suck and swallow reflexes are present, the brain is differentiating and the sheathing of nerve fibres has begun. The foetus can now hear sounds, and the eyes open and shut regularly and appear to shift their gaze. The internal genitals are formed, and hair is forming on the body and head. At the end of this period the lungs are beginning to produce *surfactin*, a substance that enables the lungs to inflate. At this point almost all the organs, except the central nervous system, are fully formed, and survival outside the womb is possible with exceptional medical care.

The third trimester is a period of rapid weight gain (50 per cent in the last month) as a layer of brown fat is deposited beneath the skin. This will keep the newborn warm. The nervous system is now functioning well enough to support life outside the womb, the lungs are able to extract oxygen from air, and the eyes have become responsive to light. Electrical activity in the brain indicates periods of sleep and wakefulness. The reflexes are now highly responsive. Normal birth occurs at the end of the third trimester (week 38/39).

TERATOGENIC INFLUENCES

LO 2.5

Identify the principles governing the action of environmental agents that may cause harm to the developing embryo/foetus

During the course of prenatal development many agents may raise the incidence of deviations or produce malformations in the foetus. These agents are called **teratogens** (from the Greek *teras*, meaning 'monster'). They include maternal diseases and blood disorders, diet, irradiation, drugs of many kinds, and abnormal temperature and oxygen levels. In addition, maternal characteristics such as age, emotional state and the number of children previously borne can influence prenatal development. The adverse effects of these agents include congenital anomalies, embryonic or foetal death, intrauterine growth retardation and mental dysfunction.

teratogens
Any agents that cause a birth defect. From the Greek word *teras*, meaning 'monster'.

The thalidomide disaster in the early 1960s brought into public awareness the dangers of taking drugs during pregnancy. **Thalidomide** was widely prescribed to pregnant women as a sedative and to control nausea in the early stages of pregnancy. It was very effective and produced no adverse effects for the women taking it. Some, however, who took the drug between the fourth and sixth weeks after conception went on to have children with an unusual group of abnormalities. Most common were deformities of the arms and legs, including fused fingers and toes, dislocated hips and phocomelia (an abnormality where the limbs are missing and the hands and feet are attached directly to the torso). Less frequently there were also facial abnormalities (eye defects, cleft palate, depressed bridge of the nose, small ears and facial palsy) and malformed organs (heart, digestive and genitourinary tracts).

thalidomide
A drug that was prescribed to pregnant women as a sedative and to control nausea in the early stages of pregnancy. It had a teratogenic effect, causing deformities in many foetuses.

About 7000 children worldwide were deformed by the drug before the link between thalidomide and the birth defects was identified by an Australian researcher (William McBride) and the drug was withdrawn from the market. The effects of thalidomide exemplify a number of principles that apply to the effects of teratogenic agents on prenatal development.

Effects of teratogenic agents on prenatal development

Teratogens have different effects on different organs at different times in development, but there are some general principles that apply to the action of teratogenic agents (Moore, Persaud & Torchia, 2013).

- *Sensitive periods for action of teratogens.* The effects of a teratogen vary with the developmental stage during which it is present. As the various organ systems begin and end their prenatal development at different times, their sensitivity to agents varies over time (i.e. there is a sensitive period for teratogenic effects on each developing organ system). Table 2.4 summarises some of the prenatal sensitive periods. Overall, the gravest danger to life is during the first two

TABLE 2.4

Sensitive periods for developing organ systems

DEVELOPING ORGAN SYSTEM	PERIOD OF SENSITIVITY TO TERATOGENS			EXAMPLES OF POSSIBLE DEFECTS
	Overall sensitivity	Highly sensitive	Less sensitive	
Central nervous system (CNS)	3 weeks – birth	3–16 weeks	16 weeks – birth	Neural tube defects; retardation
Heart	3–9 weeks	3–7 weeks	7–9 weeks	Malformed aorta and pulmonary artery; defects (e.g. hole in heart) between left and right atriums (ASD) or left and right ventricles (VSD)
Upper limbs	4–9 weeks	4–6 weeks	6–9 weeks	Missing, shrunken or deformed arms
Lower limbs	4–9 weeks	4–6 weeks	6–9 weeks	Missing, shrunken or deformed legs
Upper lip	5–9 weeks	5–7 weeks	7–9 weeks	Cleft lip
Ears	4–30 weeks	4–10 weeks	10–30 weeks	Malformed ears; deafness
Eyes	4 weeks – birth	4–9 weeks	9 weeks – birth	Abnormally small eye(s); glaucoma; cataracts
Teeth	6 weeks – birth	6–9 weeks	9 weeks – birth	Deficient and stained enamel
Palate	6–16 weeks	6–9 weeks	9–16 weeks	Cleft palate
External genitalia	7 weeks – birth	7–9 weeks	9 weeks – birth	Masculinised female genitals

Source: Adapted from K. L. Moore, T. V. N. Persaud & M. G. Torchia, *The Developing Human: Clinically Oriented Embryology* (9th ed.). Philadelphia: Saunders, 2013, p. 473.

weeks, well before cell differentiation. Teratogens present during this stage will either completely destroy the zygote or have no lasting effect. The embryonic period is the most vulnerable in terms of teratogens producing structural abnormality, as this is the time when all the structures are being laid down. Teratogens acting in the foetal period are more likely to affect function than structure, as this is the period when the various systems are becoming functional. Thalidomide had its devastating effect in the early embryonic period (weeks 4 to 6), when limbs are particularly vulnerable.

- *Tissue-specific effect.* Each teratogen acts in a specific way on specific developing tissue, and thus causes a particular pattern of abnormal development (e.g. the drug thalidomide caused deformation of arms and legs, while mercury causes brain damage).
- *Dose–response relationship.* In general, the greater the concentration of the teratogenic agent to which the organism is exposed, the greater the risk of abnormal development.
- *Individual differences in effect.* Not every child will be equally affected by a given amount of exposure to a particular teratogen (e.g. less than a quarter of pregnant women who used thalidomide during the critical time when limbs were forming gave birth to malformed babies). The way in which the developing organism responds to the teratogenic agent depends to some degree on its genotype and that of its mother. The mother's age, nutrition, uterine condition and hormonal balance can all affect the action of teratogens.
- *Effects on mother.* Mothers may suffer no adverse effects from teratogens that can produce defects in the developing embryo. In fact, some of the agents are taken by the mother because they have a beneficial effect on her (e.g. thalidomide).

We now look at some teratogenic agents and their effects.

Maternal diseases and disorders

It is estimated that 5 per cent of women catch an infectious disease while pregnant. Most of these have no effect on the embryo or foetus, but a few diseases can cause considerable damage. Some of these are shown in Table 2.5 and discussed below.

rubella (German measles)
A viral infection that can cause severe developmental defects in babies born to mothers who contract the disease in the first months of pregnancy.

- The connection between **rubella** (**German measles**) and birth defects was first suspected and reported in 1941 by an Australian ophthalmologist, Norman Gregg. He had noted a sudden increase in children born blind and was able to link this to a rubella outbreak in the summer of 1940. Rubella is a virus that causes developmental defects in more than 50 per cent of all babies born to mothers who suffer from the disease during the first month of pregnancy (i.e. before pregnancy is detected). This drops to 17 per cent by the third month. The disease results in congenital heart disease; cataracts; deafness; abnormalities of genital, urinary and intestinal tracts; and mental retardation (although this may be a result of a lack of visual and auditory stimulation). Infection in the second trimester is less harmful but may lead to hearing loss and bone defects. A vaccine for rubella was developed in 1969 and this is routinely given to children in infancy and childhood. However, as the incidence of the disease has dropped, so too has parental insistence on vaccination, so the disease persists and there are periodic outbreaks.

human immunodeficiency virus (HIV)
A viral infection that leads to AIDS. About a quarter of pregnant women who are infected with HIV pass it on to their child.

- Another virus that is currently having an enormous impact on populations throughout the world is the **human immunodeficiency virus (HIV)**, which leads to acquired immune deficiency syndrome (AIDS). About a quarter of pregnant women who are infected with the virus pass it on to their child. AIDS progresses rapidly in children, and most babies born with AIDS survive for only about six months after the symptoms appear. The antiviral drug zidovudine (ZDV) reduces prenatal AIDS transmission by up to 95 per cent with no apparent harmful consequences to the child. This has led to a marked reduction in prenatally acquired AIDS in countries where the drug is available.

TABLE 2.5 Maternal diseases and disorders that can produce teratogenic effects

MATERNAL DISEASE	POSSIBLE EFFECTS ON CHILD
Maternal disorders	
Anaemia (iron deficiency)	Death; brain impairment
Scarlet fever	Early death
Diabetes mellitus	Death; stillbirth; respiratory problems; metabolic disorders
Viral infections	
Chickenpox	Physical malformation; mental retardation
Cytomegalovirus*	Stillbirth; death; CNS damage; mental retardation; blood disorders; microcephaly
(most frequent prenatal source of infection)	Miscarriage; physical malformation; blindness; death
Genital herpes*	Brain damage; repeated illness; death
HIV/AIDS*	Heart defects; blindness; deafness; genital, urinary and intestinal abnormalities; mental retardation
Rubella	
Bacterial infections	
Syphilis*	Death; blindness; deafness; mental retardation
Tuberculosis	Death; lowered resistance to tuberculosis
Parasitic infections	
Malaria	Miscarriage; low birth weight/prematurity
Toxoplasmosis	Brain defects; mental retardation; heart defects; death

* Sexually transmitted diseases

- Chronic infections such as venereal diseases (gonorrhoea and syphilis), which invade the developing foetus and remain active, have their worst effects at later stages of prenatal development. Syphilis, for example, has its effects after the 18th week of gestation and can result in miscarriage or mental retardation, blindness and physical abnormality. The effects may not be obvious at birth but emerge in the early years of development as cognitive deficits, a decline in motor and mental ability, and even death.

  Other maternal conditions such as high blood pressure, diabetes, and blood incompatibilities between the mother and the foetus may also affect development.
- **Rh incompatibility** is the most frequent and destructive of a number of blood incompatibilities. Rh is a complex substance on the surface of red blood cells. A dominant gene determines one of the components of Rh. People who have the gene are Rh-positive. Fewer than one in five people are homozygous for the recessive form and are Rh-negative. The child of an Rh-positive man and an Rh-negative woman is likely to be Rh-positive. During birth some of the baby's blood cells usually pass into the mother's bloodstream and her immune system creates antibodies to fight this foreign substance. The antibodies stay in her bloodstream and, if she becomes pregnant with another Rh-positive child, they attack and destroy its red blood cells. Rh incompatibility is routinely tested for and, when detected, the mother is injected with anti-Rh serum within 72 hours of the birth. This prevents the formation of the antibodies.

  Other maternal factors such as levels of emotional stress, nutrition, maternal age, previous births and even exercise can have an effect on the developing embryo and foetus.

Rh incompatibility
The most frequent and destructive of a number of blood incompatibilities. It occurs when an Rh-negative mother gives birth to an Rh-positive child.

Prescription and non-prescription drugs

Any drug that has a molecule small enough to cross the 'placental barrier' is capable of entering the bloodstream of the developing embryo or foetus. Only relatively few drugs have been studied well enough to determine whether they are safe for pregnant women. The effects of thalidomide were discussed earlier. Other prescription drugs that have been identified as causing problems include *quinine*, which can cause congenital deafness; *reserpine*, a tranquilliser that can cause respiratory problems; *tetracyclines*, which can depress skeletal growth; and certain *anticonvulsant drugs* that might result in cleft lip and palate.

Even aspirin, one of the most commonly taken drugs, has been linked to problems in the infants of mothers who took it regularly during pregnancy. These problems include low birth weight, infant death around the time of birth, poorer motor development and lower IQ scores in early childhood. There is some dispute about the effects of aspirin, as some research has failed to find the effects. This exemplifies the difficult decision faced by pregnant women. The evidence is clear that some drugs have extremely deleterious effects, but it is equivocal about others. It is obvious that pregnant women need to exercise extreme caution about the drugs they take.

Many non-prescription drugs are also known to cause risk. Alcohol, cocaine, heroin, methadone, tobacco and even caffeine have been shown to affect development. *Alcohol* is the non-prescription drug we know most about.

When a pregnant woman drinks, the alcohol in her blood crosses the placenta into the foetus's bloodstream and the amniotic fluid. Hence the foetus imbibes both directly via its blood and by ingesting the amniotic fluid. The foetal blood-alcohol level rapidly reaches that of the mother but, given the less efficient foetal metabolic processes, persists much longer in the foetus's system.

Many children of alcoholic mothers are affected by foetal alcohol syndrome, described by Jones and Smith (1973).

Foetal alcohol syndrome

Foetal alcohol spectrum disorder is an umbrella term for a range of adverse effects caused by prenatal alcohol exposure. Children with the full spectrum of symptoms are defined as having **foetal alcohol syndrome (FAS)** (Australian National Council on Drugs, 2012). FAS is one of the leading causes of preventable birth defects and developmental disabilities (Itthagarum, Nair, Epstein & King, 2007). The most noticeable characteristics of FAS are defects such as microcephaly

foetal alcohol syndrome (FAS)
A complex of abnormalities found in many children of mothers who drank alcohol heavily during pregnancy.

(small head), and malformations of the heart, limbs, joints and face. The facial abnormalities are the most noticeable external anomalies, including a smooth upper lip, short nose, and narrow, widely spaced eyes. The brains of FAS children are typically smaller and show a lack of cortical convolutions (tend to be smoother). An affected baby is likely to exhibit abnormal behaviours such as excessive irritability, hyperactivity, seizures and tremors. At birth, FAS children are smaller and lighter than normal and their physical growth lags behind that of normal age mates. They do not catch up. The majority of these children score well below average in intelligence tests and many are mentally retarded.

How much alcohol does it take to harm a foetus? Even moderate alcohol consumption (one drink per day) during early pregnancy can retard prenatal growth and produce minor physical and behavioural abnormalities (Khalil & O'Brien, 2010; O'Leary et al., 2010b). Infants born to mothers who are heavy drinkers (more than five drinks a day) have a 30 per cent chance of suffering FAS. O'Leary and colleagues (2010a) reported a fourfold increased risk of birth defects after heavy prenatal alcohol exposure in the first trimester (see Research Focus 2.2). Binge drinking (occasionally having many drinks over a short period of time) has also been identified as potentially very harmful (Moore, Persaud & Torchia, 2013; O'Leary et al., 2010a, 2010b; O'Leary & Bower, 2012).

Not all foetuses are equally susceptible to the effects of alcohol exposure. As outlined earlier, other factors, both genetic and environmental, can make some foetuses more vulnerable than others. Many children who were exposed to high levels of alcohol as foetuses do not have full-blown

RESEARCH FOCUS
Effects of prenatal alcohol exposure

A team of researchers based in Western Australia has been conducting population-based studies into the effects of prenatal alcohol exposure upon postnatal physical and behavioural development.

Colleen O'Leary and colleagues (2010a) investigated the associations between dose, pattern and timing of prenatal alcohol exposure, and alcohol-related birth defects. Between 1995 and 1997, data from a randomly selected cohort of non-Indigenous women in Western Australia (n = 4714) who gave birth to a live infant were linked to birth information recorded in the Western Australian Birth Defects Registry. Alcohol use before and during pregnancy was categorised as low (fewer than 7 standard drinks per week and no more than 1–2 on any one day), moderate (as for the low group but with greater amounts per occasion, with 2–5 on any one day), or heavy (more than 14 drinks per week, or more than 7 drinks per week and binge-drinking more than twice per week). The comparison group consisted of women who abstained from drinking alcohol.

When compared to the abstinent group, the group who engaged in heavy alcohol use in the first trimester had four times the likelihood of having an infant with a birth defect that was classified as alcohol-related. In contrast, no association was found in this study between low or moderate prenatal alcohol exposure and alcohol-related birth defects.

O'Leary and her colleagues (2010b) also reported on childhood behaviour problems as a function of prenatal alcohol exposure in an eight-year longitudinal study of this Western Australian sample. They reported that heavy levels of prenatal alcohol exposure at any stage of pregnancy resulted in increased prevalence of problems for the child. Even moderate exposure in the first trimester resulted in elevated levels of internalising behaviours (anxiety/depression), while high exposure in later pregnancy resulted in externalising and aggressive problems.

The researchers concluded that their studies indicate the importance of making women of childbearing age aware of the risks to their child of even moderate alcohol consumption in early pregnancy. While low levels appeared not to increase risk in this study, they pointed out the fine distinction between low and moderate consumption and concluded that 'the safest message for pregnant women is abstinence' (O'Leary et al., 2010a, p. 84).

FAS but show a variety of neuropsychological deficits, including hyperactivity and attention problems (Guerri, Bazinet & Riley, 2009). Given the severe physical and behavioural effects that can result from prenatal alcohol consumption and the wide range of response to even quite small amounts, there is no uniformly safe level of alcohol consumption during pregnancy. Hence, the advice now offered to pregnant women is to avoid the consumption of alcohol.

One Western Australian study (Elliott, Payne, Morris, Haan & Bower, 2008) indicated a lack of knowledge about FAS by health professionals, resulting in under-diagnosis of the condition and a lack of advice to pregnant women. This has concerning implications for the health of Australian children (particularly Indigenous children, who were over-represented in the WA sample).

So far we have mentioned maternal diseases and disorders, drugs and blood incompatibilities as factors that can result in developmental deviations. Other factors, including maternal diet, environmental pollution (e.g. lead and mercury), radiation and the emotional state of the mother, have also been shown to affect prenatal development. As well, very low birth weight and anoxia during birth put the infant at risk. O'Rahilly and Muller (2001) discuss the effects of such factors.

It is clear that an appropriate supportive prenatal environment is essential for normal development and that an abnormal environment can produce abnormal behaviour. We also know that infants learn from environmental events shortly after birth (e.g. they will habituate to repetitive stimuli). This has led investigators to ask whether learning takes place before birth.

FOETAL LEARNING

LO 2.6
Assess evidence of prenatal learning

The study of prenatal cognitive functions is limited by the stimuli that can be presented reliably to the foetus and by the responses that can be obtained to those stimuli. Kisilevsky and Hains (2010), in reviewing the literature linking foetal heart rate and cognition, state that while there are many measures available for studying infant cognition, heart rate is the only measure available for use in foetal studies. Changes in heart rate are taken as indications of cognitive processing.

Kisilevsky and Muir (1991) established that foetuses habituate to repetitive sounds heard while *in utero*. They repeatedly played two different types of sound at low volume via a microphone placed on the mother's abdomen close to one of the foetus's ears (located by ultrasound). One sound was a pure tone, while the other was a steady vibration noise. They recorded the foetus's responsiveness to the sounds by measuring movements (kicks) and heart rate immediately before, during and after the sounds were played. They found that the foetus initially responded vigorously to the new sound, but that the response declined with repeated presentation of the same sound until, after about eight repeats, the sound failed to elicit a response. This indicated that the foetus had habituated to the sound. When a novel sound was played, the vigorous response appeared again (indicating discrimination between the sounds) and this too declined with repetition.

While such results clearly indicate prenatal learning, they did not provide any evidence that the learning persists after birth. Some evidence for such persistence (memory) has been provided in other studies where neonatal behaviour has been linked to prenatal experiences. Prenatal exposure to chemosensory stimuli via the mother's diet has been shown to lead to taste preferences in the neonate in a number of mammalian species, including humans (Wells & Hepper, 2006). Schaal, Marlier and Soussignan (2000) found that neonates born to mothers who had consumed anise flavour during pregnancy demonstrated a preference to anise odour, whereas neonates of mothers who did not consume anise during pregnancy had an aversion response to the odour. Mennella, Jagnow and Beauchamp (2001) demonstrated a similar taste preference for carrot-flavoured cereal in infants of mothers who had drunk carrot juice during pregnancy.

One of the first attempts to examine foetal learning was that of Lee Salk (1973). Salk predicted that the foetus would have habituated to the sound of the mother's normal heartbeat and that they would find this sound comforting after birth. Salk exposed groups of newborn infants to (a) the sound of the mother's normal heartbeat (80 beats per minute); (b) the sound of mother's heartbeat sped up to 120 beats per minute; or (c) no special sounds. Group (a) gained more weight and cried less than the controls during the four-day experiment. Salk reported that the group that heard the abnormal heartbeat became so distressed that this part of the study was abandoned. Salk concluded

that the infant's prior experience in the womb had made the sound of the normal heartbeat familiar and reassuring (i.e. the foetus learned from experience *in utero*).

DeCasper and Spence (1986) provided evidence that infants can remember learning that took place *in utero*. In their study, 12 pregnant women read a passage from the Dr Seuss story *The Cat in the Hat* aloud twice a day for the last six weeks of pregnancy (a total of about 3.5 hours). Two days after birth the babies were tested for their preference for either the familiar *Cat in the Hat* passage or a novel passage (either *The King, the Mice and the Cheese* by Gurney & Gurney or *The Dog in the Fog*, a modified *Cat in the Hat*). The passages were read by either the mother or another woman. A special dummy (pacifier) was used to measure the infants' sucking rates. Baseline sucking was recorded for two minutes; any change in their sucking rate would then switch on a recording of either the familiar passage or the new passage. Thus, infants could control the frequency with which they heard the passage by changing their sucking rate. It was found that infants modified their sucking rate when rewarded by the familiar passage but not when they heard the unfamiliar passage (regardless of whether read by their mother or another woman). DeCasper and Spence concluded that the infants had heard the stories read to them *in utero* and that this influenced the sounds they found rewarding after birth.

STOP & REVIEW

Given the background noises present in the womb (from the mother's digestive processes, blood flow, heartbeat, etc.), how clearly would the foetus be likely to hear the reading of *The Cat in the Hat*?

LO 2.7
Identify the adverse effects on infant survival and healthy development of prematurity/low birth weight

THE BIRTH PROCESS

The prenatal stage ends with a series of traumatic events resulting in the birth of the child. Approximately 38 weeks after conception, in response to a complex series of hormonal changes, the uterine wall begins irregular contractions that mark the beginning of labour. Typically, the baby has rotated to a head-down position by this stage (the first stage of labour). Over the next 10 to 20 hours the contractions become stronger and more regular, and the mother's cervix (opening of the uterus) gradually dilates. Full dilation and the beginning of the baby's movement through the birth canal mark the beginning of the second stage of labour, in which strong contractions lasting up to 60 seconds push the baby to the outside world. The final stage of labour is the expulsion of the placenta, which occurs within minutes of the baby's birth.

Assessment of the child at birth

Apgar scale
A standardised assessment of the child's physical condition that is administered immediately after birth.

Immediately after birth (at one and five minutes after birth), the child's physical condition is assessed by doctors and nurses using a standardised assessment tool known as the **Apgar scale** (Apgar, 1953). This assesses five physiological functions (respiration, heart rate, reflex response, muscle tone and colour), each on a three-point scale (a score of 0 indicating an absence of function, a score of 1 indicating weak or poor function, and a score of 2 indicating good function). The scores over all five functions are summed to give an overall score, where a score of 7 or better indicates that the infant is in good physical condition, while a score between 4 and 6 indicates that some assistance may be required to establish respiration and other vital signs. An Apgar score below 4 signals that the infant is in serious danger and immediate medical assistance is required. The second test is given to infants who did not score well at one minute, as often they take a little time to adjust to life outside the womb.

Effects of prematurity and low birth weight

Most infants are born on time (within two weeks of the due date of 38 weeks post-conception) and of appropriate size (over 2500 grams). A minority are premature (three weeks or more prior to due date) or, while full-term, are less than 2500 grams ('small-for-dates'). Such children typically

require more medical care than those born full-term and of normal weight. The earlier and smaller, the greater the intervention required and the poorer the prognosis for the child. Birth weight is the best predictor of infant survival and healthy development.

An Australian Institute of Health and Welfare report (2014), based upon Australian Bureau of Statistics data from 2011, indicated that, in Australia in 2011, 9 per cent of all births were preterm (less than 37 weeks' gestation) and 6.3 per cent of babies were of low birth weight (under 2500 grams). While this figure is low compared with many other countries, the rate was not uniformly low. The proportion of babies of Indigenous mothers that were of low birth weight was 12 per cent, double the rate for babies of non-Indigenous mothers.

The AIHW (2014) report also indicated that babies born in remote areas were more likely to be of low birth weight than those born in major cities. This is particularly evident in babies born to Indigenous mothers, as was shown in a study by Mackerras and colleagues (2003). Their study of urban and remote-dwelling Aboriginals in the Northern Territory showed that there is an even greater discrepancy between these groups. They measured the **body mass index** (BMI) of these infants (using a formula that takes into account weight and length/height) and compared this with data on all Australian neonates from the 1985 Australian Urban Fitness survey. They found that the Aboriginal infants were well below the Australian norm for BMI, with 11 per cent of the urban group and 37 per cent of the remote group below the fifth percentile on these norms. These data point to the marked differences between groups within the Australian population. This discrepancy is also evident in the infant mortality data.

body mass index
A standard measure of body mass, taking into account both height/length and weight, that is used for assessing a child's physical development.

Infant mortality

The rate of infant mortality (death in the first year after birth) is very much related to general social factors such as economic level, availability of health care and level of social support. An ABS report (2002) indicated that there are wide discrepancies in the rates of infant mortality in different countries, varying from three deaths per 1000 live births in countries such as Singapore, Japan and Sweden to much higher levels of 27 in Indonesia and 60+ in Papua New Guinea. Australia and New Zealand have approximately 5/1000, slightly lower than the United States with 6/1000. Those with the lowest rates have the highest levels of support for expectant parents and provide good medical, social service and maternal support to most members of the population. In less developed countries where health and social support is minimal the infant mortality rates can be staggering. (For example, Scheper-Hughes, 1992, cites the example of a small city in Northern Brazil where the rate was 90 per cent.) While Australia's infant mortality rate is among the lowest in the world, the death rate for Indigenous infants is not so good. The AIHW (2014) report on Australia's health indicated that while the rate of Indigenous infant deaths had dropped significantly in the last decade (by 62 per cent), the rate in 2012 was still double that of the general population.

It is clear that, just as genetic factors affect developmental outcome, environmental influences within the womb are also critical in determining the success (or otherwise) of development. The child enters the world as the product of these dual influences, and development throughout life will continue to be affected by both.

CRITICAL THINKING

Imagine that you are a school counsellor to whom a concerned teacher has referred a child with very poor reading and spelling but with good number skills. The child's mother reports that it is not something that can be fixed as 'dyslexia runs in her family' and therefore 'must be genetic'. What evidence is there for a genetic basis for dyslexia? What other factors could be contributing to this familial problem? How would you design a study to test the inheritance of such a characteristic?

CRITICAL THINKING FOR GROUP DISCUSSION

1. Based on your knowledge of X-linked inheritance, explain why males are more likely to be miscarried than females. (LO 2.1, 2.2)
2. Why is it difficult to interpret the results of studies of resemblance in intelligence between relatives of differing degrees of genetic similarity? (LO 2.3)
3. What is a 'sensitive period'? Discuss this concept in relation to the effects of teratogenic agents. (LO 2.4, 2.5)
4. What is foetal alcohol syndrome? How many alcoholic drinks are 'safe' for an expectant mother? Outline the case against drinking during pregnancy. (LO 2.4, 2.5, 2.7)
5. Does any of the evidence of foetal learning indicate that it plays an important role in later development? (LO 2.6)

PRACTICAL EXERCISE

LO 2.8

Conduct a research exercise examining the difference between related and unrelated pairs of children on a number of behavioural characteristics.

Familial effects on behaviour

This is a class exercise in which students will compare related and unrelated children on a number of behavioural characteristics and then evaluate the data for evidence of a familial association in these characteristics. Ethics approval from the institution's human ethics committee will be required before proceeding.

Hypothesis

Related children will be more alike than unrelated children.

Method

Students will work in pairs. In each pair, Student A will collect data on two children aged between two and six years who are siblings (and living in the same family). Student B will collect data on two children who are unrelated (living in different families) but of the same age and sex as the children who are Student A's participants.

The data will be collected using questionnaires to be completed by the mother of each child.

Mothers will be told: 'I am going to read some statements. Please indicate how well these fit your child – what percentage of the time your child would be like this: never (0%), sometimes (25% of the time), about half the time (50%), most of the time (75%) or always (100%).'

Record the mother's response on a 5-point scale where 1 = 0% and 5 = 100%.

1. My child is active and extroverted, always wants to be the centre of attention.
2. My child prefers boisterous, vigorous activities to quiet, sedentary ones.
3. My child talks well and uses language to communicate her/his needs.
4. My child is easily upset.
5. My child persists when faced with a difficult problem – he/she doesn't give up easily.
6. My child has good fine-motor skills and often plays with toys requiring good manual dexterity.
7. My child is very sociable and outgoing, likes to be with adults and reacts well to strangers.
8. My child follows rules.
9. My child gets on well with friends.
10. My child gets involved in trouble.