CHAPTER

15 Genotypes and phenotypes

FIGURE 15.1 Different phenotypes can be seen in these dogs' chocolate, black and cream coat colours. In this chapter we will explore visible phenotypes as the expressions of underlying genotypes, examine how gene expression can be influenced by environmental and epigenetic factors, and identify differences between monogenic and polygenic traits.

KEY KNOWLEDGE

This chapter is designed to enable you to:

- become familiar with the use of symbols to denote genotypes
- identify dominant and recessive phenotypes and understand their differences
- recognise that the expression of genes can be influenced by environmental factors and by epigenetic factors
- develop knowledge and understanding of polygenic inheritance.

Baby Rose and the CFTR gene

Baby Rose, daughter of Sarah and Daniel, was the third addition to the Trengarth family. Her arrival was especially welcome as Rose was the first sister for her two male siblings, James and Trent, and the first daughter for Sarah and Daniel. At birth Rose seemed a healthy baby. About a day after her birth, a heel prick blood sample was taken from Rose. This was done as part of the routine screening test that is carried out on newborn babies Australia-wide. The test result showed that she had **cystic fibrosis** (CF).

CF is an inherited disorder and the gene responsible is the **CFTR** gene on the number-7 chromosome. The CFTR gene encodes a trans-membrane protein that controls the transport of chloride ions across the plasma membrane. (Refer to chapter 1, pp. 36–7 for more details on the role of this transporter protein and the effects of a faulty transporter protein, in particular one that causes the mucus of the lungs to be thick and sticky.) The diagnosis of CF was unexpected as neither parent had ever heard of CF and each stated 'No one in our family has ever had it'.

Cystic fibrosis is the most common inherited single-gene disorder seen in Caucasians of northern European descent and in their derived populations in Australia, Canada and New Zealand. CF occurs equally in females and in males. The incidence of CF in Caucasians is generally stated to be about 1 baby in every 2500 live births, but in other populations, such as Asian and Pacific Islander populations, the incidence is much lower.

CF cannot be cured but the development of affected babies is carefully monitored, watching for any bacterial infections of the lungs that are rapidly treated. Other treatments include pancreatic enzyme replacement and physiotherapy sessions to assist in removing the thick sticky mucus from the lungs.

The **CFTR** gene has several different forms, or alleles. The various alleles result from small differences in the base sequence of the **CFTR** gene which affect the ability of the protein that it encodes to perform its normal transporter function.

For the shorthand notation of different alleles of one gene, the practice is to use variants of the same letter(s) of the alphabet, as follows:

- Where a gene has two phenotypic expressions or alleles, such as 'trait present' and 'trait absent', or 'red' and 'white' flower colour, symbols such as ***A*** and ***a***, or ***R*** and ***r*** or ***D*** and ***D'*** might be used, depending on the dominance relationship between the two alleles. Usually, the letter chosen relates to one of the phenotypic expressions of the gene, such as ***R*** for red flower colour.
- Where a gene has multiple alleles, each having a different phenotypic expression, a common letter is still used, but with the addition of superscripts to the common letter, for example, I^A, I^B and ***i***, or ***C***, c^b, c^s and c^a.

So, in the case of the **CFTR** gene, the allele that produces the normal transporter protein can be denoted by the symbol ***C***. We can group the alleles which produce a defective transporter protein that causes a person's mucus to be thick and sticky, and these can be denoted using the symbol ***c***. Because the **CFTR** gene is an autosomal gene, located on the number-7 chromosome, baby Rose has two copies of this allele and so is genotype ***cc***.

ODD FACT

Australia-wide, newborn babies are screened for several conditions including cystic fibrosis, phenylketonuria, galactosemia and primary congenital hypothyroidism (see p. 590).

What is a phenotype?

Baby Rose's cystic fibrosis is her **phenotype**. **A phenotype is the visible or measurable expression of the genetic make-up of an organism's structure and/or functioning,** often in terms of one gene. In baby Rose's case, her phenotype for the **CFTR** gene is an abnormal transporter protein that produces the clinical signs of cystic fibrosis, including the thick and sticky mucus in her lungs and pancreatic ducts.

Other examples of phenotypes in various organisms include:

- red, green, orange and yellow fruit colours in capsicums (refer to figure 13.42, p. 517)
- short leg length and normal leg length in sheep (*Ovis aries*) (see figure 15.2a)
- blood types A, B, AB and O in people
- purple, white and yellow kernel colour in corn (*Zea mays*)
- beaked, round, flattened and elongated fruit shapes in tomatoes (*Solanum lycopersicum*) (see figure 15.2b)
- presence and absence of fur and eye pigment in kangaroos (*Macropus* spp.)
- ability and inability to differentiate between the colours, red and green, in people
- requirement or non-requirement for the amino acid arginine for growth in yeast (*Saccharomyes cerivisiae*).

FIGURE 15.2 A phenotype is the visible, measurable or detectable effect of a gene on the structure and/or functioning of an organism. Examples of phenotypes include **(a)** leg length in sheep, with the abnormally short legs of an Ancon sheep seen here in comparison with the normal leg length and **(b)** shape and colours of tomato fruits.

Phenotypic differences can be seen not only in eukaryotic species, but also in microbial species. One significant phenotypic difference that affects human health is the emergence of resistance to antibiotics in many bacterial species. For example, *Staphylococcus aureus* (also known as golden Staph) includes some strains that are sensitive to one or more antibiotic drugs and some that are resistant to virtually all the current useful antibiotics. These antibiotics include methicillin, which interferes with the synthesis of a component of the bacterial cell wall, and erythromycin and streptomycin, which attack the bacterial ribosomes.

Different strains of one bacterial species as well as different bacterial species can be distinguished as being either drug-sensitive or drug-resistant using **antibiotic sensitivity testing** in a laboratory (see figure 15.3). In this test, a specific bacterial species is allowed to grow across the surface of a gel in a Petri dish. The gel contains all the nutrients required for growth and the bacterial growth appears as a white film. Discs impregnated with different antibiotics (or with different concentrations of the same antibiotic) are placed on the plate and the antibiotic will diffuse from the disc into the gel. If the bacteria are sensitive to the antibiotic on a disc, the bacteria in the area will die and this appears as a clear area around the disc.

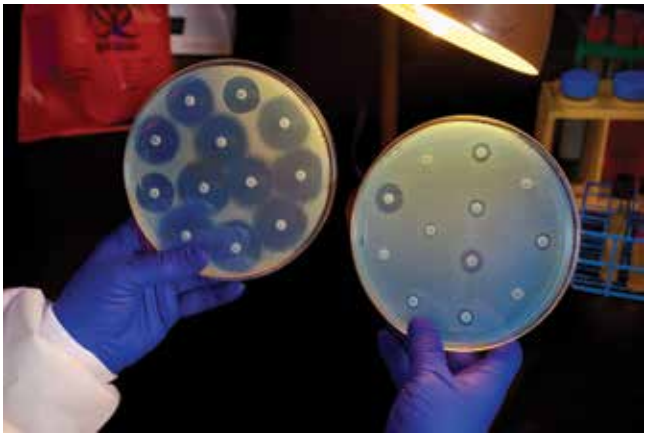

FIGURE 15.3 Results of antibiotic sensitivity testing of various antibiotics against two strains of bacteria. Which strain of bacteria (left or right plate) is more sensitive to the antibiotics?

A phenotype may be expressed as a single feature of an organism's structure or functioning, such as purple kernel colour in corn. In other cases, however, the responsible gene produces a phenotype that shows multiple effects on several organs. This is the case for CF where the lungs, pancreas and sweat glands all show phenotypic effect. The CF phenotype is seen in the lungs which produce abnormally thick and sticky mucus that causes breathing difficulties and increases the risk of lung infections. The CF phenotype is also seen in the pancreas where the thick mucus blocks the pancreatic ducts preventing the release of its digestive enzymes into the gut so that digestion is impaired. And the CF phenotype is seen in the sweat glands that produce abnormally salty sweat.

Many phenotypes are the physical or physiological expression of a single gene. However, in some cases, a visible phenotype may be the result of an interaction between genes at two or more gene loci. For example, all cats are genetically tabby cats as they all have the **TABBY** gene that determines the pattern of tabby striping, either mackerel or blotched. However, these patterns are only displayed if the cat also has at least one copy of the agouti allele (***A***) of the **ASIP** gene. If the cat has two identical alleles of the recessive non-agouti allele (***aa***), the tabby pattern will not show — instead the cat will be one solid colour (see table 15.1).

The genetics of cat colour involves many genes and many interactions between these genes.

TABLE 15.1 One example of the interaction between two genes in cats. Which gene determines whether or not a cat will display its phenotype in terms of tabby striping?

Genotype at ASIP locus	Genotypes at TABBY locus	Phenotypes	
AA homozygous agouti	Ta^MTa^M Ta^Mta^b ta^bta^b	mackerel pattern mackerel pattern blotched pattern	
Aa heterozygous agouti	Ta^MTa^M Ta^Mta^b ta^bta^b	mackerel pattern mackerel pattern blotched pattern	
aa homozygous non-agouti	Ta^MTa^M Ta^Mta^b ta^bta^b	solid colour (no stripes) solid colour (no stripes) solid colour (no stripes)	

What is a genotype?

The underlying genetic make-up that determines an organism's phenotype is called its **genotype**. A genotype is not visible, only its phenotypic effects can be seen or measured. A genotype is the combination of the particular alleles of a gene or genes that are present and active in a cell or in an organism and determine a specific aspect of its structure or functioning.

In diploid organisms, the genotype is typically written as a pair of alleles. This is the case for autosomal genes for both females and males. If the two alleles of a gene are identical, for example, ***CC***, the genotype (and the organism concerned) are said to be **homozygous**. If the two alleles are different, the genotype is described as **heterozygous**, for example, ***Cc***.

The only exceptions are the genotypes of males for **X-linked** and **Y-linked genes**, that is, genes on the X chromosome and genes on the male-exclusive major segment of the Y chromosome. For these genes only, the genotype of a normal male consists of just one allele and is described as being **hemizygous** (*hemi* = half). In baby Rose's case, the genotype that underlies her cystic fibrosis phenotype is homozygous recessive genotype ***cc***. The encoded instruction at the **CFTR** gene locus on both of her number-7 chromosomes is 'make defective transporter protein'. We will see later that the genotypes of both of baby Rose's parents are heterozygous ***Cc***. Baby Rose inherited one ***c*** allele from her mother and another ***c*** allele from her father. When an autosomal gene has two alleles, for example, ***F*** and ***f***, the maximum number of different genotypes possible in a diploid organism is three, namely ***FF***, ***Ff*** and ***ff***. However, when a gene has multiple alleles, more genotypes are possible; if a gene has three multiple alleles, six different genotypes are possible, if four multiple alleles exist then 10 different genotypes are possible. Refer to table 13.7 on page 515 to review some examples of genes with multiple alleles. Can you identify the six genotypes possible for the three multiple alleles of cats shown in table 13.7?

Next we will look more closely at genotypes, using mainly human examples.

Genotypes for autosomal genes

Both females and males have two copies of each gene located on an autosome; these are termed **autosomal genes**. While people share identical genes, they may differ in the specific alleles they possess.

Each person has two copies of the **ABO** gene on their pair of number-9 chromosomes. However, people may have different alleles of that gene. One person may have two copies of the ***i*** allele, another person may have one copy of the I^A allele and one copy of the ***i*** allele, yet another person may have one copy of the I^B allele and one copy of the ***i*** allele. The particular combination of alleles of a gene is that person's genotype in terms of that gene.

The gametes produced by a homozygous person, such as genotype I^AI^A, will be identical, with all having the same allele. The gametes produced by a heterozygous person, such as genotype I^AI^B, will be of two kinds, with half having the I^B allele and half having the I^A allele.

Genotypes for genes on the sex chromosomes

The sex chromosomes are the X and the Y chromosomes. The DNA of each chromosome contains genes, and because of their size differences, the X chromosome has many more gene loci than the Y chromosome (see figure 15.4).

FIGURE 15.4 The human sex chromosomes magnified 10 000 times. The X chromosome (left) is much larger than the Y chromosome and carries about 800 genes; in contrast, the Y chromosome carries about 50 genes.

ODD FACT

In AD 200, a Jewish edict exempted a boy from circumcision if his two brothers had bled to death. Their male cousins were also exempted.

People have been aware for a long time that some conditions, such as certain colour vision defects and a blood-clotting disorder (haemophilia) that occur in particular families, appear more often in males than in females. Why?

This is because the genes controlling colour vision and blood clotting are located on the X chromosome and are not represented on the Y chromosome. To be affected, females must inherit two copies of the particular allele; males are affected if they have just one allele so males more commonly show the trait. When a gene is located on a sex chromosome, the traits controlled by its alleles do not appear equally in both sexes.

FIGURE 15.5 Children with muscular dystrophy take part in play group therapy.

Genes on the X chromosome

Many genes are located on the human X chromosome (X-linked), including the **DMD** gene, located on the short arm of the X chromosome (refer to figure 14.32, p. 545). This gene controls production of the muscle protein dystrophin, which is found on the membrane of skeletal muscle cells. The **DMD** gene has two alleles. When only abnormal dystrophin is produced by muscle cells, a disorder called Duchenne muscular dystrophy results. This disorder is extremely rare in females but it affects about 1 in every 3500 male babies. Affected males suffer slow but irreversible loss of the muscle function and are confined to a wheelchair usually before they reach their teens (see figure 15.5). Death usually occurs before or in early adulthood.

Males have only one X chromosome and so have one allele of this gene. A male has a hemizygous genotype, and is either ***M*** or ***m***. (These genotypes are shown as ***M*** (Y) and ***m*** (Y), where the (Y) denotes the Y chromosome.) Males affected by Duchenne muscular dystrophy have the hemizygous genotype ***m*** (Y).

Normal females with two X chromosomes have two alleles of the **DMD** gene. A female with the heterozygous ***Mm*** genotype is not affected by muscular dystrophy, nor is a female with a genotype ***MM***. Affected males invariably inherit the ***m*** allele from a heterozygous mother, but not an affected father. (Why?)

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Other genes on the X chromosome include the:

- **OPN1LW** gene, responsible for the protan form of red–green colour blindness that results from defective red receptors in the retina of the eye
- **F8** gene, responsible for the classical blood-clotting disorder, haemophilia A, that results from defective or absent factor VIII
- **F9** gene, responsible for a second blood-clotting disorder, called Christmas disease, that is due to defective or absent factor IX
- **IDS** gene, responsible for Hunter syndrome in which complex carbohydrates (mucopolysaccharides) build up in cells producing deleterious physical and mental effects
- **GLA** gene, responsible for Fabry disease, a lysosome storage disease
- **BTK** gene, responsible for an X-linked form of agammaglobulinemia, a condition in which antibody-forming cells of the immune system are not formed
- **G6PD** gene, responsible for deficient production of enzyme glucose-6-phosphate-dehydrogenase, which can result in bouts of severe anaemia, often precipitated by environmental factors.

All genes that are termed X-linked genes occur *exclusively* on the X chromosomes.

Genes on the Y chromosome

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The Y chromosome has a number of distinctive features. Small segments of DNA at each of the ends of the Y chromosome are homologous with DNA segments on the X chromosome. Can you suggest why this might be the case? During prophase of meiosis, homologous chromosomes synapse (pair up) in preparation for migrating to different poles of the spindle. The presence of regions of the Y chromosome that are homologous to regions on the X chromosome means that during meiosis in males the sex chromosomes can pair and then separate (disjoin) correctly.

Most (more than 95%) of the Y chromosome is not shared with any other chromosome — it is specific to males only. This DNA consists of about 23 million base pairs and it is the location of about 50 genes, all Y-linked. Y-linked genes occur exclusively on the Y chromosome.

Included among the genes located on the Y chromosome is the **SRY**, or sex-determining, gene that controls production of a protein involved in testis formation in the human embryo. (This gene is also known as the testis-determining factor.) The early embryo has undifferentiated gonads — they are neither testes nor ovaries. The testis-forming protein takes effect in about the sixth week of embryonic development, causing the gonads to develop as testes. In the absence of this protein, the gonads differentiate to form ovaries.

Very rarely, a person who is chromosomally 46, XY has an altered and inactive form of the **SRY** gene. What sex will this person display? Such a person cannot be male because of the lack of the testis-determining factor that is the product of the normal **SRY** gene. The XY combination of sex chromosomes results in a normal male only if the SRY that controls the production of testis-determining factor is active. This person is a rare XY female.

At birth such a baby appears with the external characteristics of a normal female. However, she has no ovaries; instead she has traces of gonadal tissue only. It is not until this girl shows delayed puberty that this condition, known

as **Swyer syndrome**, would be discovered. Hormone replacement therapy is used to initiate the development of female secondary sex characteristics including breast development.

Included among genes located on the male-specific portion of the Y chromosome are the:

- **AMELY** gene that controls the organisation of enamel during tooth formation
- **SOX21** gene that encodes a protein SOX-21, which controls some hair loss conditions
- **AZF** gene that encodes a protein which is important in sperm formation. Deletions of part or all of this gene result in **azoospermia**, with males having no sperm in their semen.

KEY IDEAS

- A phenotype is the visible or measurable expression of the genetic make-up of the structure and/or functioning of an organism.
- A phenotype is often the physical or physiological expression of a single gene.
- A phenotype may sometimes result from interactions between genes at two or more gene loci.
- A genotype is the genetic make-up of an organism that determines its phenotype.
- Genes and the traits they control can be described in terms of their chromosomal locations, that is autosomal or X-linked or Y-linked.
- Genotypes are homozygous or heterozygous, except for the hemizygous genotypes of males for Y-linked and X-linked genes.

QUICK CHECK

1 Identify the key difference between the terms:
   a phenotype and genotype
   b homozygous, heterozygous and hemizygous.

2 Identify whether each of the following statements is true or false.
   a The **CFTR** gene has its locus on the number-7 chromosome.
   b The cystic fibrosis phenotype involves just the lungs.
   c The genotype of a person who is affected by cystic fibrosis could be heterozygous ***Cc***.
   d A person's genotype in terms of hair colour can be observed and measured.

Relationship between expression of alleles

If you are simultaneously given two instructions that are mutually exclusive, such as 'Turn left at the next corner!' and 'Turn right at the next corner!', you can carry out only one instruction. However, for two other instructions, such as: 'Paint the door yellow' and 'Paint the door green', you might carry out both instructions by producing a bi-coloured yellow and green door. Similar situations can be recognised for the phenotypes produced by genes.

Complete dominance

Rose, the baby of Sarah and Daniel, was diagnosed as having cystic fibrosis. The **CFTR** gene on number-7 chromosome that encodes the transporter

protein involved in cystic fibrosis has two alleles, ***C*** and ***c***. The ***C*** allele gives the instruction 'Make normal active transporter protein', while the ***c*** allele gives the instruction 'Make an abnormal inactive transporter'.

A person with the heterozygous ***Cc*** genotype has two mutually exclusive genetic instructions, but such a person produces an active transporter protein, makes normal mucus secretions and so is free of cystic fibrosis. From this, we can conclude that the disease-free condition with normal mucus secretions is **dominant** to cystic fibrosis with abnormal thick mucus. The relationship between the alleles of the **CFTR** gene are shown in table 15.2. Note that baby Rose with her ***cc*** genotype can produce only an inactive abnormal transporter protein and so has cystic fibrosis.

TABLE 15.2 Relationship between allele of the **CTRF** gene

Genotype	Phenotype
homozygous ***CC***	normal mucus
heterozygous ***Cc***	normal mucus
homozygous ***cc***	thick, sticky mucus (cystic fibrosis)

To decide whether a trait is dominant or recessive, the phenotype of a heterozygous organism is identified. The trait that is expressed in this phenotype is the dominant trait. Alleles that control dominant traits are usually symbolised by a capital letter; for example, the allele ***S*** controls the dominant short fur length trait in cats. Alleles that control recessive traits are symbolised by the lower case of the *same* letter; for example, the allele ***s*** controls the recessive long fur length in cats.

Being a carrier

In genetics, the term **carrier** refers to a heterozygote that has the allele for a recessive trait but does not show the trait. In people, alleles may be carried for hidden recessive traits that do not affect normal functioning, such as straight hairline and blood type O. However, some alleles that are carried by heterozygotes are for recessive disorders, such as cystic fibrosis or albinism (see table 15.3).

TABLE 15.3 Some dominant and recessive human traits. Heterozygotes carry alleles for recessive traits but their effects are not expressed. What genotype is necessary for a recessive trait to be expressed for an autosomal gene?

Dominant trait	Recessive trait
peaked hairline (widow's peak) (***W***) (see figure 15.6)	straight hairline (***w***)
free ear lobes (***F***)	attached ear lobes (***f***)
mid-digital hair present (***G***)	mid-digital hair absent (***g***)
shortened fingers (brachydactyly) (***S***)	normal length fingers (***s***)
normal pigmentation (***A***)	pigmentation lacking (albinism) (***a***)
non-red hair (***R***)	red hair (***r***)
normal secretions (***C***)	cystic fibrosis (***c***)
dwarf stature (achondroplasia) (***N***)	average stature (***n***)
Rhesus positive (Rh +ve) blood (***D***)	Rhesus negative (Rh –ve) blood (***d***)

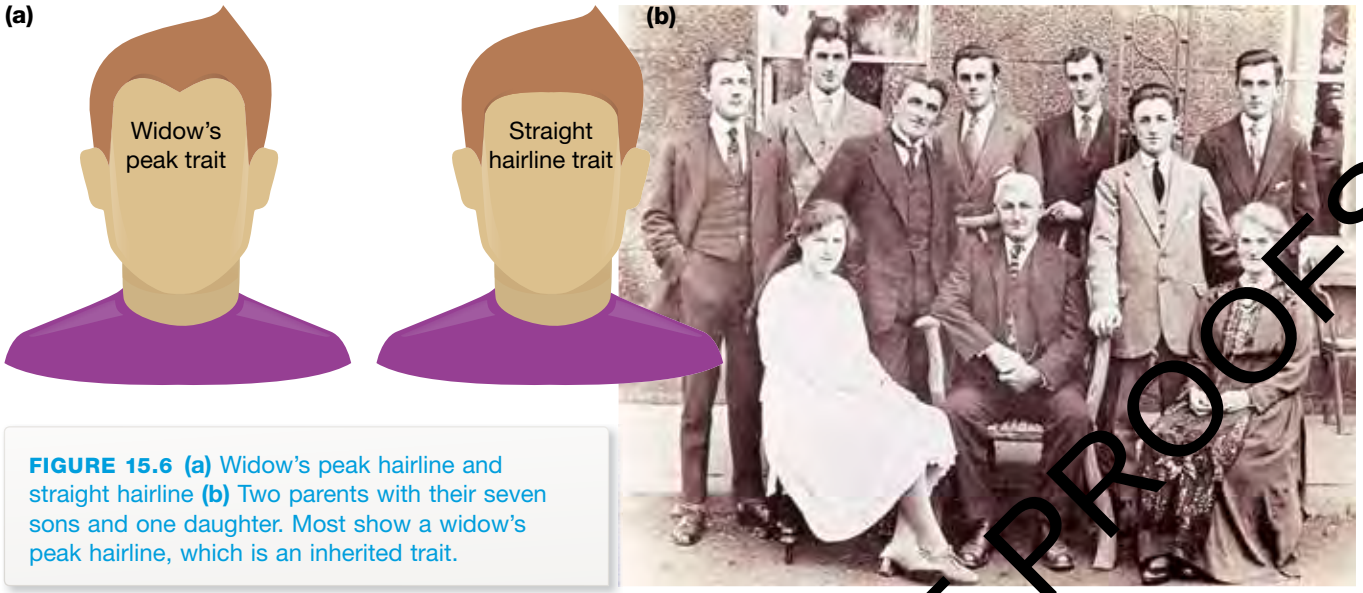

FIGURE 15.6 **(a)** Widow's peak hairline and straight hairline **(b)** Two parents with their seven sons and one daughter. Most show a widow's peak hairline, which is an inherited trait.

Most often, heterozygotes are *not* aware of their carrier status for an allele controlling a recessive trait. Parents may realise they are carriers only when they have a baby with a recessive disease. Sarah and Daniel, the parents of baby Rose, were unaware that they were both genotype ***Cc***, and so were carriers of an allele that resulted in cystic fibrosis until Rose was born. People with heterozygous genotypes sometimes know that they are carrying a 'hidden' allele. Consider the case of Maria. Both Maria's sister and her cousin have thalassaemia, a recessive inherited disorder affecting the haemoglobin in red blood cells. Maria wondered if she was a carrier. A simple test done through the Thalassaemia Society in Melbourne confirmed that Maria was a carrier of thalassaemia with the heterozygous genotype ***Tt***.

If the genotype of an individual is unknown but could be either ***Aa*** or ***AA***, this may be denoted as ***A***– (where ' – ' represents either allele).

Co-dominance

The **ABO** gene, located on the number-9 chromosome, has three alleles that determine antigen production (see table 13.6, p. 514). Antigen A and antigen B occur on the surface of the red blood cells of some people. Depending on which antigens are present, blood is typed as group A, B, AB or O. The presence (or absence) of a particular antigen is inferred by adding specific antibodies and observing the result (see figure 15.7). Antibodies used to type blood are *anti-A antibodies* and *anti-B antibodies*. Anti-A antibodies cause clumping or agglutination of red blood cells with antigen A. Anti-B antibodies cause clumping of red blood cells with antigen B.

Look at the reaction of the blood sample from Tran (column 3). His blood clumped when anti-A antibodies were added, so his red blood cells have antigen A and he has the allele I^A. Tran's blood also clumped when anti-B antibodies were added, so his red blood cells have antigen B and he must also have the allele I^B. Tran's genotype is heterozygous I^AI^B. Because **both traits are expressed in the heterozygote, these two alleles show co-dominance**. Alleles showing co-dominance are denoted by a capital letter with a superscript added to distinguish between them. Table 15.4 shows that the phenotypic actions of both the I^A and I^B alleles are dominant to the action of the ***i*** allele, and so blood group O is recessive to the other blood types.

As well as co-dominance, you may see terms such as partial dominance and incomplete dominance in various genetics books.

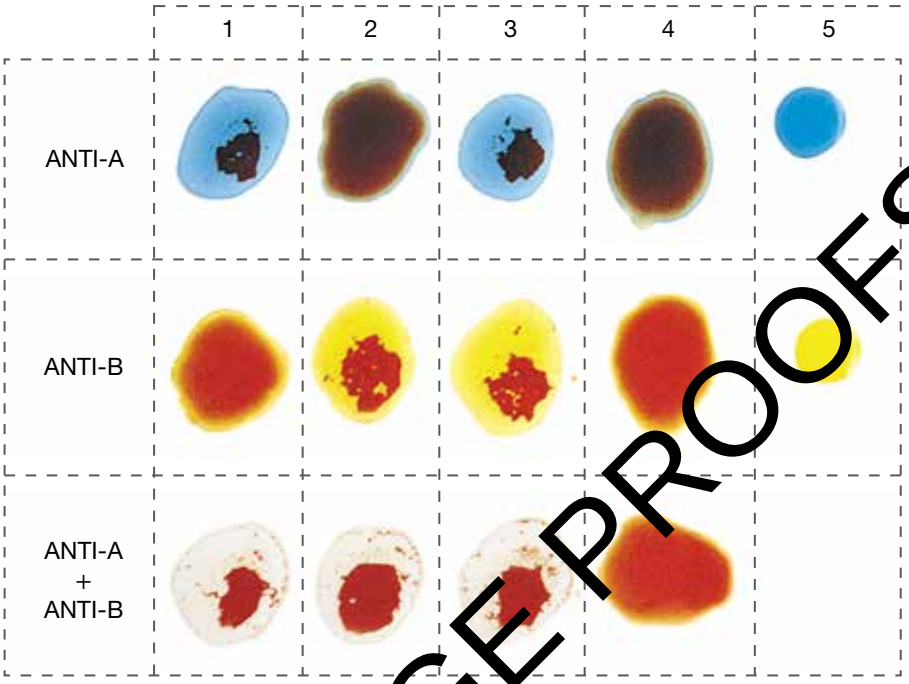

FIGURE 15.7 Addition of specific antibodies to blood samples causes cells to 'clump' or agglutinate when the corresponding antigen is present on the surface of the red blood cells.

TABLE 15.4 Relationship between genotypes and phenotypes for the **ABO** gene. What relationship exists between the I^A and the I^B alleles? Between the I^B and the i alleles?

Genotype	Instructions carried by alleles	Phenotype
homozygous I^AI^A	'produce antigen A'	blood type A
homozygous I^BI^B	'produce antigen B'	blood type B
homozygous ii	'produce neither antigen A nor B'	blood type O
heterozygous I^AI^B	'produce antigen A' and 'produce antigen B'	blood type AB
heterozygous I^Ai	'produce antigen A' and 'produce neither antigen'	blood type A
heterozygous I^Bi	'produce antigen B' and 'produce neither antigen'	blood type B

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Co-dominance
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Genes and alleles in domestic animals

Like people, plants and other animals carry genetic instructions or genes on their chromosomes. Figure 15.8 identifies some phenotypes due to the action of common alleles in several domestic mammals: cat (*Felis catus*), dog (*Canis familiaris*) and horse (*Caballus equus*).

Look at the alleles of the three genes identified in the cats. Can you suggest why only the third cat shows a pattern of tabby stripes? What is the phenotypic action of the ***G*** allele in horses? What interaction appears to exist between the three genes in the horses?

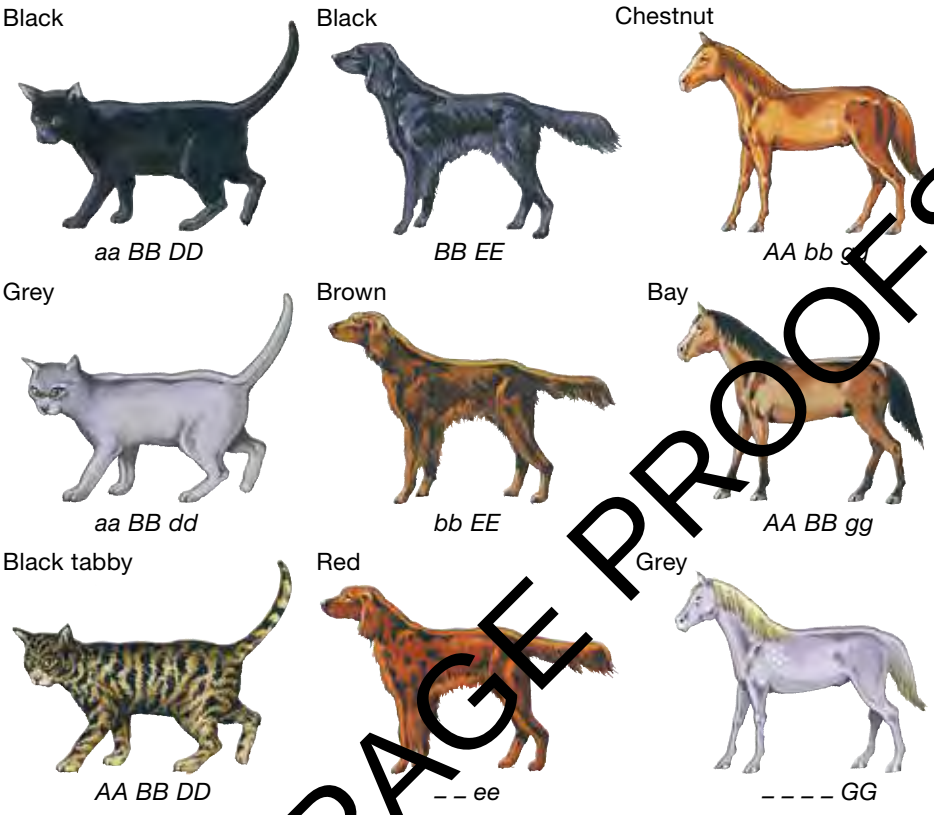

FIGURE 15.8 Some phenotypes in domestic mammals and their possible genotypes. The dash symbols cover any and all combinations of the alleles in the position occupied by the dashes. So, for the third dog in the row, the – – symbol can mean ***BB*** or ***Bb*** or ***bb***.

Environment interacts with genotype

The phenotypes shown in figure 15.8 are expressions of the underlying genotypes. However, a phenotype is not always produced exclusively by the genotype. In some cases, the phenotype is a result of the interaction between the genotype and environmental factors:

genotype + environment ⟶ phenotype

Many environmental factors, both external and internal, act on an organism so that its final phenotype is due to varying contributions of genotype and environment.

The phenotype due to a particular genotype may appear only in one specific environment. In a different environment, another phenotype may appear. Let's examine some examples of how environmental factors can affect the phenotype.

ODD FACT

For a fetus, its environment is the mother's uterus. A mother's actions — drinking alcohol, smoking — can change this environment and affect the phenotype of the fetus.

PKU and dietary-controlled phenotype

The inherited disorder, phenylketonuria (PKU) results from the action of the gene that controls production of an enzyme known as *phe hydroxylase*. Babies that inherit the homozygous recessive genotype ***pp*** from their parents cannot produce this enzyme. If these babies are fed diets that include proteins that contain normal quantities of the amino acid, *phe*, the babies will suffer brain damage and become mentally retarded, so early diagnosis is critical. However, if these babies are fed a special diet that includes proteins with very low levels of *phe*, the babies will not suffer brain damage and **will show a normal phenotype.**

In this case, the phenotype of a child with genotype ***pp*** depends on the internal environment that is controlled by the diet:

genotype pp + HIGH phe diet ⟶ phenotype: PKU
genotype pp + LOW phe diet ⟶ phenotype: normal

Figure 15.9 shows dietary items that are low in the amino acid phenylalanine (Phe). The availability of these special foods make it easier for families to cater for a child with PKU.

FIGURE 15.9 Products available for a wide range of metabolic disorders, including PKU. Each product has a particular use and excludes one or more amino acids or other compounds. In the case of PKU, the restricted amino acid is phenylalanine (Phe). Special low-protein bread, biscuits, mixes and pasta are also available.

Cats and temperature

Figure 15.10a shows some Siamese kittens. At birth, these kittens are all white. A few weeks later, the kittens begin to develop pigmentation, first along the edges of their ears. Gradually the pigment spreads until the kittens show the characteristic colouring on the face, ears, feet and tail. This pattern of colour change is due to an interaction between the cats' genotypes and their environment.

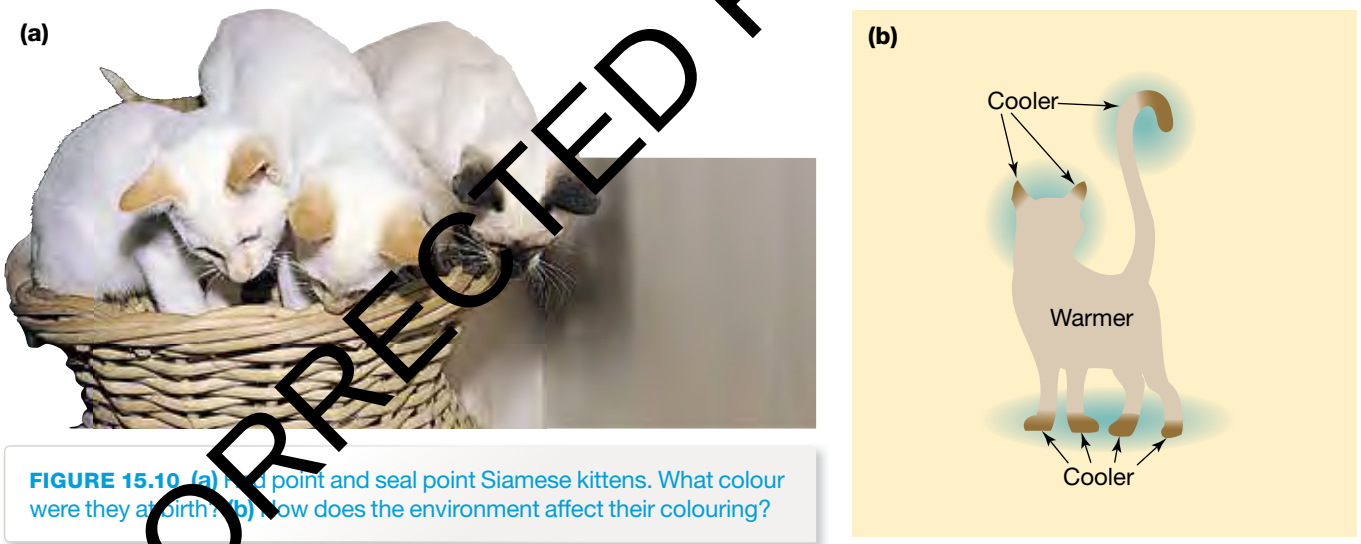

FIGURE 15.10 (a) Red point and seal point Siamese kittens. What colour were they at birth? (b) How does the environment affect their colouring?

Siamese cats have a particular form of a gene that codes for the production of tyrosinase. This enzyme catalyses one step in the production of pigment:

$$\text{precursor} \xrightarrow{\text{tyrosinase}} \text{pigment}$$

In Siamese cats, the particular form (allele) of this gene produces a tyrosinase enzyme that is heat sensitive. **This enzyme can catalyse the step in the production of pigment when the temperature is lower than the core body temperature only.** Siamese kittens undergo embryonic development in a warm uterine environment and so are born unpigmented (white). Pigment appears first on the coolest parts of their bodies — the ear margins — and then on other extremities (figure 15.10b).

Plants and soil pH

Hydrangea plants (see figure 15.11) produce **blooms with colours that depend on the acidity or alkalinity (pH) of the soil** in which they are growing. The colour is due to pigments known as anthocyanins, which are located in membrane-bound sacs within the petal cells. In soil with an acidic pH these pigments are a bright blue, while at alkaline pH they are a pink or red.

FIGURE 15.11 When grown in acid soil hydrangeas produce deep blue flowers. In contrast, when grown in alkaline soil the flower colour is pink or red. Is variation in flower colour in hydrangeas under genetic or environmental control?

Broad beans and black urine

The **G6PD** gene, an X-linked gene, encodes the protein that acts as the enzyme glucose-6-phosphate dehydrogenase. Females with the homozygous ***gg*** genotype and males with the hemizygous ***g*** genotype are deficient in this enzyme. Normally, these persons do not display phenotypic effects of this deficiency. However, exposure to certain environmental factors can produce marked phenotypic effects. One such environmental factor is a substance in broad beans (*Vicia fava*). If persons with the enzyme-deficient genotype eat broad beans, this can cause their red blood cells to break down so that they suffer severe anaemia and the products of this breakdown can appear in their urine, causing it to be blackened. A similar reaction can occur if these persons are exposed to other compounds, including the anti-malarial drug primaquine and naphthalene, the compound found in moth balls.

KEY IDEAS

- One gene can exist in a number of different forms called alleles, each identified with a specific phenotype.
- When two different alleles are both expressed in the phenotype of a heterozygote, they are said to be co-dominant.
- The phenotype of an organism can be the result of an interaction between its genotype and the environment.
- The phenotype may on occasion be the result of an interaction between two or more genes.
- To decide whether a phenotypic trait is dominant or recessive the phenotype of the relevant heterozygous organism must be identified.
- Phenotypes may be due to an interaction between genotypes and environmental factors.

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Unit 2 AOS 2 Topic 3 Concept 3 — **Factors affecting phenotypes** Concept summary and practice questions

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Interactivity
Generating the phenotype
int-0178

QUICK CHECK

3 Identify a key difference between the following.
   a Autosomal and X-linked
   b X-linked and Y-linked genes
   c Compete dominance and co-dominance
   d Dominant and recessive phenotype
4 List an example of:
   a an X-linked gene
   b a phenotype that is the result of a interaction between the genotype and the environment
   c a Y-linked gene.
5 What information would you need in order to decide which of two alleles produced the dominant phenotype?

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Unit 2 AOS 2 Topic 3 Concept 4

Polygenic inheritance
Concept summary and practice questions

Polygenic inheritance

So far in this chapter we have dealt mainly with **monogenic** traits (*mono* = one). These are traits that are controlled by a single gene. Monogenic traits typically show **discontinuous variation**; that is the expression of the single gene involved produces just a few discrete and non-overlapping phenotypes, often just two categories. So, for the **CFTR** gene, two phenotypes are seen: unaffected and cystic fibrosis. For Mendel's sweet peas, flowers are white or purple and for tabby cats, the pattern of stripes is either mackerel or blotched. Traits that show discontinuous variation are typically qualitative and are described in words.

In contrast, some inherited traits are controlled by a several genes, each having a small, but cumulative effect on the phenotype. These genes are called **polygenes** and the traits that they control are said to be **polygenic** (*poly* = many). Typically, the phenotypes produced by polygenes form many classes that show **continuous variation**. If a large sample is taken, the values form a continuum. Human traits under polygenic control that show continuous variation include height and skin colour. Traits that show continuous variation are typically quantitative and are often described in numerical values, such as a height of 168 cm, 150 cm or more than 170 cm.

Figure 15.12 shows a simplified representation of discontinuous and continuous variation.

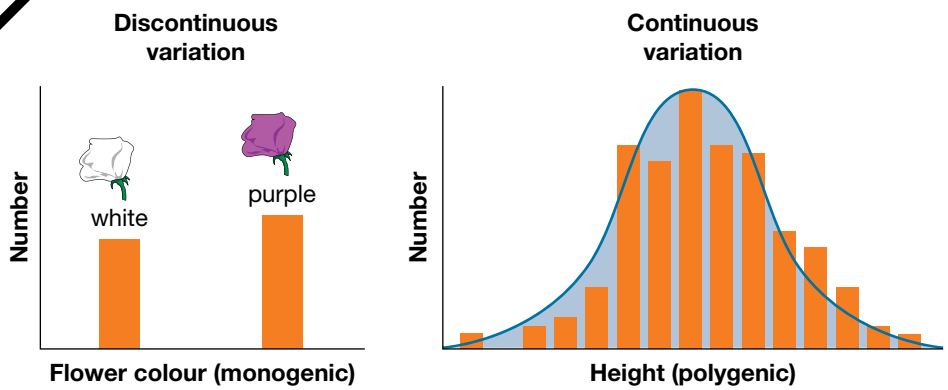

FIGURE 15.12 **(a)** Discontinuous variation is seen when the phenotypes for a particular trait fall into a few — often just two — non-overlapping distinct groups or classes. This type of variation is seen in traits controlled by a single gene. **(b)** Continuous variation is seen when phenotypes for a trait fall into many classes. This type of variation is seen in traits that are under polygenic control.

Other examples of polygenic traits are the fat content of cows' milk, the mass of bean seeds, maximum speed of thoroughbred horses and plant height in tobacco plants. It is interesting to note that, unlike tobacco plants, plant height in Mendel's pea plants is a monogenic trait with his plants falling into two categories 'tall' and 'short'.

Polygenes and human height

Adult human height exhibits a large range of phenotypes. Most typically, height phenotypes range from under 1.5 m to over 2.1 m. Adult height can be affected by factors such as the occurrence of illnesses in childhood and childhood diet. However, adult height is known to have a strong genetic component. Given that this trait does not show discontinuous variation but rather shows continuous variation across a range, it is reasonable to conclude that height is not controlled by a single gene.

Height is a polygenic trait with a number of genes, each having a small but additive effect on adult height. We can summarise a simple model of polygenic inheritance of height as follows:

- A number of polygenes contribute to adult height.
- Each polygene has two alleles, the plus (+) allele that contributes a small amount to an increase in height above a base value, and the minus (–) allele that does not add to the trait.
- The phenotypic action of each polygene is equal so that, for example, the + + – – genotype shows the same phenotype as the + – + – genotype.
- The greater the number of + alleles, the greater the increase in height above the base value.
- Polygenes do not show dominance or recessiveness, but act in a cumulative manner.

So, for the polygenic trait of height in human adults, it is reasonable to predict that the distributions of height in a large sample of adults will show a **bell-shaped distribution**. Figure 15.13 shows such a bell-shaped distribution of heights in female and male adults.

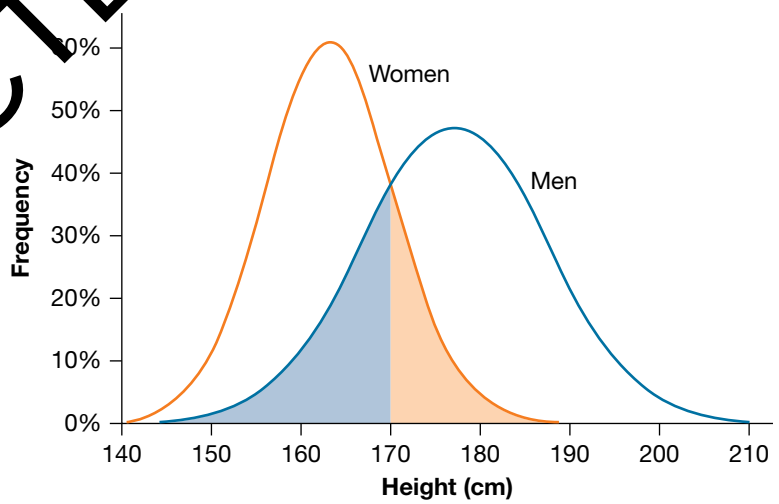

FIGURE 15.13 Bell-shaped distribution of height in human female and male adults. The frequency of each height is shown on the vertical axis. What is the most common height in females? In males? What is the approximate height range for males?

Eye colour is a polygenic trait

The assumptions for the operation of polygenic inheritance as outlined in the previous section are ideal. In real life, there will be some deviations from these assumptions, for example, in human eye colour. In the determination of eye colour, all polygenes do not contribute equally to the phenotype. As we will see, one gene makes a major contribution to the eye colour phenotype.

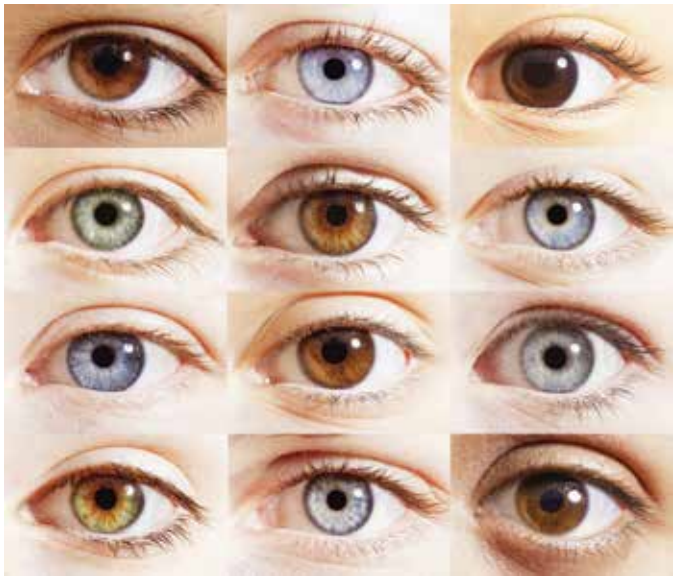

FIGURE 15.14 Eye colour variation in people. Eye colour is not simply either brown or blue — many other colours exist.

Eye colour in people depends on (1) the amount of **melanin** pigment present in cells of the iris of the eye, known as **melanocytes** and (2) the numbers of these melanocytes. At one end of a spectrum is 'blue eye colour' that occurs in an iris with small numbers of melanocytes, each with low levels of pigment. At the other end of the spectrum is 'brown eye colour' that occurs when high levels of melanin pigment are present in larger numbers of melanocytes. In between are various eye colours including hazel and green (see figure 15.14).

The gene that plays a major role in the determination of eye colour is the **OCA2** gene on chromosome 15. This gene encodes a protein involved in melanin production and it has the alleles ***B*** and ***b***. Persons with the ***bb*** homozygous genotype at this locus are *usually* blue eyed and those with the genotypes, either ***BB*** or ***Bb*** are *usually* brown eyed. Clearly, that is not the end of the story, because people do not have either blue or brown eye colour — a range of other eye colours exist.

In fact, eye colour in humans is a polygenic trait. In addition to the involvement of the **OCA2** gene, eye colour is now known to be influenced by at least seven other genes that are involved in some aspects of melanin production. The action of these genes can affect the amount of melanin pigment in the eye. These genes can interact with the **OCA2** gene and, in some cases, reduce its expression so that, less commonly, a person with the ***BB*** or ***Bb*** genotype will have blue eyes instead of the expected brown eye colour. This explains why on rare occasions two blue-eyed persons can have a brown-eyed child. Such a brown-eyed child is a result of the action of polygenes, not marital infidelity.

Explaining polygenic inheritance

Skin pigmentation is another polygenic trait. If we use the assumptions outlined on page 562 and assume that two genes are involved in skin colour, we can see how polygenic inheritance can create many phenotypes.

Table 15.5 shows a simple model of polygenic inheritance of skin colour.

TABLE 15.5 A simple model of polygenic inheritance of skin colour

Number of plus (+) alleles	Possible genotype(s)		Degree of pigmentation
	Gene 1	Gene 2	
4	+ +	+ +	very dark
3	+ + + –	+ – + +	dark
2	+ + + – + –	– – + – ++	intermediate dark
1	+ – – –	– – + –	slightly dark
0	– –	– –	light

In reality, skin colour is more complex than this model suggests and probably involves at least four polygenes. The degree of skin pigmentation can also be affected by environmental factors, such as exposure to UV radiation.

In general, as the number of polygenes increases, the number of possible variations in a trait also increases, as shown in table 15.6.

TABLE 15.6 Increase in the number of polygenes correlates to an increase in the number of possible variations in a trait.

Number of polygenes	Number of variations possible
2	5
3	7
4	9
n	$2n + 1$

Where the number of polygenes is n, the number of possible variations is $2n + 1$. So, if $n = 8$ polygenes, the number of possible variants in a population is 17. The following box describes a case study of polygenes in action.

POLYGENES IN ACTION

As a result of ultra-sonography, Kym and her husband, Joe, knew that she would soon give birth to twins. Kym's ancestors are Irish and she has fair skin; Joe has intermediate dark skin, which he says came from his West Indian father. Joe's mother is English.

The couple have a son, Jimmy, whose skin colouring is midway between that of the parents. They expected that the twins would also have skin colouring midway between their own colouring.

Healthy dizygotic (non-identical) twin girls were born. One girl, Jasmine, is fair and her skin colour is as light as her mother's. The other girl, Carly, has a dark complexion, with a skin colour as dark as her father's. Can this event be explained?

Figure 15.15 shows the genotypes of Kym and Joe. Joe can produce three kinds of sperm:

+ + and + − and − −

In contrast, all of Kym's eggs are − −.

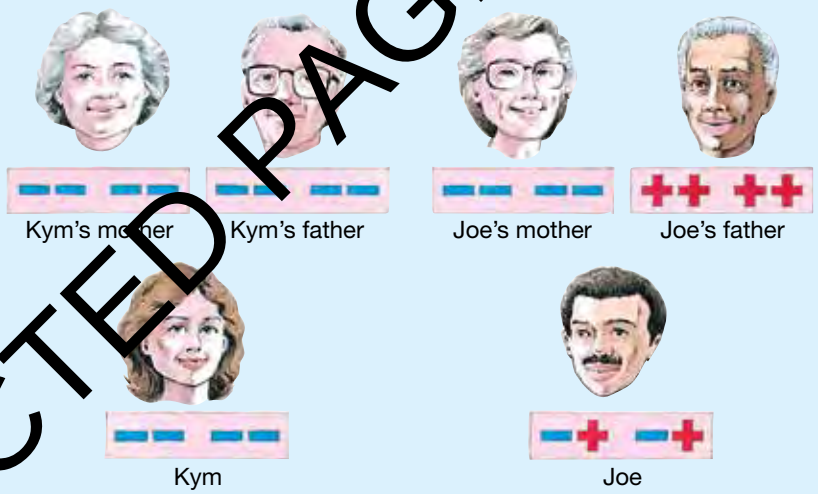

FIGURE 15.15 Simple model of inheritance of skin pigmentation

Jasmine resulted from the fertilisation of an egg (− −) by a sperm (− −). Carly resulted from the fertilisation of an egg (− −) by a sperm (+ +). Can you identify a possible combination of egg and sperm that produced Jimmy?

We have seen how the variation in human skin pigmentation can be explained on the basis of polygenic inheritance. Polygenic inheritance also applies to traits in other organisms, such as butterfat content of milk in cows, length of cobs in corn, size of hens' eggs, body mass in poultry and extent of white spotting in Holstein cattle.

The alleles of each polygene are inherited in a Mendelian fashion; that is, the members of each pair of alleles segregate to different gametes and the assortment of one polygene is independent of that of other polygenes. So, for example, an organism with the genotype + + − − + + for a polygenic trait can only produce gametes carrying the information + − +. In contrast, an organism with the genotype + − + − + − can produce a large number of different kinds of gametes including + + +, − − −, + − + and − + −. Can you identify other kinds?

Polygenes in action in corn

The length of a corn kernel can range from about 5 cm to about 21 cm in different varieties of corn. We can account for this range of variation if we assume that:

- four polygenes are involved in determining length of a mature corn cob
- the base length of a cob is about 5 cm for corn grown under standard conditions
- each polygene has two alleles: a plus (+) allele and a minus (–) allele
- each plus (+) allele adds 2 cm to the base length when corn is grown under standard conditions
- each minus (–) allele contributes nothing to the base length

The maximum number of plus alleles that a corn plant can have is eight and the minimum is zero. Table 15.7 shows how this model can generate a range of variation in cob length seen in different corn varieties.

TABLE 15.7 A model for cob length in corn based on a system of four polygenes. Sample genotypes only are shown for the alleles at each of the four loci. Can you write another genotype for the three plus allele situation?

Number of plus (+) alleles	Sample genotypes				Length of mature cob (cm)
	Locus 1	Locus 2	Locus 3	Locus 4	
0	– –	– –	– –	– –	5
1	– –	– –	– –	– –	7
2	+ –	– –	– –	– –	9
3	+ –	– +	– +	+ –	11
4	+ +	– –	+ –	+ –	13
5	+ –	+ +	+ –	+ +	15
6	+ +	– –	+ +	+ +	17
7	– +	+ +	+ +	+ +	19
8	+ +	+ +	+ +	+ +	21

KEY IDEAS

- Monogenic traits show discontinuous variation, with a few discrete classes.
- Inherited variation in some traits is due to the action of polygenes.
- Polygenic traits are controlled by a number of genes.
- Polygenic inheritance generates many phenotypes that show continuous variation.
- Simple models of polygenic inheritance can be constructed.

QUICK CHECK

6 Identify whether each of the following statements is true or false.
- **a** Polygenic traits typically show discontinuous variation.
- **b** The greater the number of polygenes, the greater the number of possible phenotypic classes.
- **c** All the gametes of an organism with the genotype + + – – would be + –.
- **d** Polygenic traits measured over a large sample of people would be expected to show a bell-curve type distribution.

7 If skin colour were controlled by four polygenes, how many phenotypic classes would be possible?

Epigenetics

What is **epigenetics**? The term *epi*genetics literally means 'above' genetics or 'in addition to' genetics or 'on top of' genetics. Epigenetics is the study of how cells with identical genotypes can show different phenotypes. Such phenotypic differences are not due to differences in the base sequences of the DNA of their genes. In addition, these differences are stable within an organism and, in some cases, these differences can be transmitted across generations. So, as well as traditional Mendelian inheritance, another kind of inheritance exists, namely **epigenetic inheritance**.

FIGURE 15.16 Identical twins have been shown to have identical genomes. This means that, as expected, they have identical genotypes. In some cases, differences in their phenotypes may be due to the action of epigenetic factors.

Epigenetics refers to all changes to genes, apart from changes to their base sequences, that bring about phenotypic changes. Epigenetic factors are factors that can bring about these changes. Such factors are external to DNA, but act on DNA and turn genes permanently 'on' or 'off'. Epigenetic factors may underlie some of the differences seen the phenotypes of identical twins, since these differences cannot be explained by differences in their genotypes (see figure 15.16).

Epigenetic factors

Epigenetic factors can change how DNA in cells is packaged or how it is labelled.

Packaging of DNA in cells may be tight or may be open. (Refer to figure 13.23, p. 498 to review how DNA is packaged around histone proteins.) Genes in segments of DNA that are tightly packaged are silenced, while genes in segments of DNA with open packaging are active in transcribing polypeptide gene products.

Labelling DNA is like adding a 'tag' that does not alter the base sequences of genes but can either silence genes or make them active. **Methyl groups** (–CH3) are one example of an epigenetic tag. Methyl groups can be added to any C base alongside a G base in DNA. (Addition of methyl groups is called methylation.) Active genes are found to have fewer methyl groups than inactive genes, so it appears that tagging genes by the addition of methyl groups to their C–G bases can change gene expression and permanently switch those genes 'off'.

Once established, epigenetic tags remain for the life of a cell and are transmitted to all daughter cells derived from that cell. Usually they are not passed on to the next generation. Typically the DNA of a fertilised egg is cleared of the epigenetic tags. In some cases, however, the epigenetic tags on the DNA are not erased but instead are conserved and passed to the next generation(s).

Examples of epigenetic inheritance

Some examples of epigenetic inheritance include:

- *Cell differentiation*. The cells of a human embryo, and later a fetus, are all derived from a single fertilised egg by a series of mitotic cell divisions. During embryonic development, cells will develop along different pathways; for example, some cells will differentiate as brain cells (neurons), some will develop into smooth muscle cells and some into liver cells. All the various

cell types — more than 200 cell types in total — have the same genotypes, but different sets of genes are active in each cell type. Epigenetic factors produce the changes that start various stem cells down different developmental paths.

- *X-inactivation.* The somatic cells of normal female mammals have two copies of the X-chromosome. Early in embryonic development, one of the X chromosomes in each somatic cell is inactivated, switching off all its genes. The epigenetic tags that cause the inactivation of a particular X chromosome are transmitted to all the daughter cells produced by subsequent mitotic cell divisions, so that the same X chromosome remains inactive.
- *Imprinted genes.* Imprinted genes refer to genes whose expression is affected by their parental origin. Children born with a small deletion of one of their number-15 chromosomes will show one of two different phenotypes. Which phenotype is displayed depends on whether the chromosome with the deletion came from the mother or from the father. If from the father, the clinical phenotype is that of Prader-Willi syndrome; if from the mother, the clinical phenotype that is displayed is Angelman syndrome (see figure 15.17).

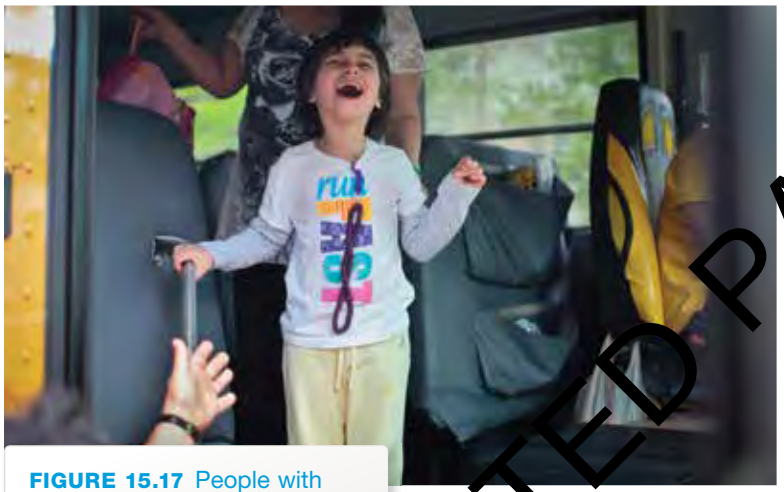

FIGURE 15.17 People with Angelman syndrome have jerky movements, smile frequently and laugh often.

- *Chemical action.* Vinclozolin is a commercial fungicide. If ingested by mammals, it interferes with sperm formation. In an experiment, pregnant rats were injected with vinclozolin and, as expected, the male offspring produced reduced numbers of sperm with lower than normal mobility. However, an interesting finding was that if these male rats managed to mate their sons showed the same defect, and this defect was then passed to males in the next generation. The chemical did not cause a mutation of the DNA of the original male. What was observed in the three generations of male rats was a change in the level of methylation of their DNA.

KEY IDEAS

- Epigenetics is the study of how cells with identical genotypes can show different phenotypes.
- Epigenetic factors act on DNA but do not change the DNA base sequence.
- Methylation of C bases in DNA is one kind of epigenetic factor.
- Packaging of DNA is another means by which epigenetic factors can act.

QUICK CHECK

8 Identify whether each of the following statements is true or false.
- **a** Epigenetic inheritance is an alternative name for Mendelian inheritance.
- **b** Once an epigenetic tag is in place within a cell it will remain for the life of that cell.
- **c** Some epigenetic tags can pass across generations.
- **d** Development of different cell types in an embryo involves the action of epigenetic factors.

9 Give an example of an epigenetic change that can be transmitted across generations.

BIOCHALLENGE

1 a Identify the gender, in terms of external features, of each of the following.

i Person A — XY chromosomes, with a normal **SRY** gene
ii Person B — normal XX chromosomes
iii Person C — XY chromosomes, but an inactive **SRY** gene
iv Person D — XY chromosomes with a deletion of the entire **SRY** gene

Add your decisions to table 15.8.

b Now, for a challenge. Predict the gender, in terms of external features, of the following.

i Person E — XX chromosomes. This person is the result of fertilisation of a normal egg by a sperm formed by meiosis during which a crossing-over event between the X and Y chromosome occurred that caused the **SRY** gene on the Y chromosome to be relocated to the X chromosome.
ii Person F — XY chromosomes. This person has a very rare condition known as complete androgen insensitivity syndrome because of a change in the **AR** gene on the X chromosome. The normal allele of this gene makes the proteins that are the androgen receptors on cells. These receptors are needed for cells to respond to androgen hormones, including testosterone. The changed allele stops the androgen receptors from working so that the cells are non-responsive to testosterone. (Testosterone is essential for the development of testes and for the appearance of the male physical characteristics.)

Add your decisions to table 15.8.

TABLE 15.8

Person	Predicted gender
A	
B	
C	
D	
E	
F	

2 For the 1992 Olympic Games, the International Olympic Committee (IOC) introduced a new gender identification test to prevent males, disguised as females, from competing in women's events. This test was intended to 'prove' that competitors were indeed female. The test was a 'blanket' test as it covered all female competitors.

The new test was one for the presence of the **SRY** gene. The test for the **SRY** gene detects the presence of sequences of DNA from that gene. All women entrants in Olympic events were required to undergo this test.

For each of the six persons identified in question 1, identify the gender that would be assigned to each as a result of the SRY gene test.

Add your decisions to table 15.9.

TABLE 15.9

Person	Gender from SRY testing
A	
B	
C	
D	
E	
F	

3 Blanket gender testing was abolished before the Sydney 2000 Olympics because of many concerns.

Suggest why this happened.

Chapter review

Key words

antibiotic sensitivity testing
autosomal gene
azoospermia
bell-shaped distribution
carrier
co-dominance
continuous variation
cystic fibrosis
discontinuous variation
dominant
epigenetic inheritance
epigenetics
genotype
hemizygous
heterozygous
homozygous
melanin
melanocytes
methyl group
monogenic
phenotype
polygene
polygenic
Swyer syndrome
X-linked gene
Y-linked gene

Questions

1 Making connections → Use at least eight of the chapter key words to draw a concept map relating to genotypes and phenotypes. You may use other words in drawing your map.

2 Linking concepts from earlier chapters → Phenotypes seen in bacterial species include sensitivity to particular antibiotic drugs. The antibiotic streptomycin interferes with bacterial ribosomes.

- **a** Identify a reason that streptomycin is effective in killing some bacteria.
- **b** Would you predict that streptomycin would be effective for treating just one infection caused by one kind of bacteria only or would you predict that it would be generally effective against several bacterial infections? Briefly explain your decision.

3 Applying knowledge and understanding → The genetic control of height differs in tobacco plants and pea plants.

- **a** What are the differences in the genetic control of height in these two plant species?
- **b** Make a rough sketch that shows height classes in a group of pea plants.
- **c** Now make a rough sketch that shows height classes in tobacco plants.

4 Demonstrating understanding and suggesting explanations → In tobacco plants (*Nicotiana* sp.), flower length is under the control of four polygenes, with each plus allele adding an equal amount to the baseline value of this trait and each minus allele having no effect. The genotypes of three plants follow.

Plant P + + + + − − + −
Plant Q + − + − + − + −
Plant R + + + + − − − −

The plants were grown under comparable conditions.

- **a** Which plant would be expected to have the longest flowers?
- **b** How many different kinds of gametes could plant R produce?
- **c** How many different kinds of gametes could plant Q produce?

5 In dairy cattle, the level of butterfat in milk varies from a high value of about 6.6 per cent to a low of about 2.6 per cent and this trait is under the control of polygenes. Develop a simple model to account for this range of butterfat content based on four polygenes.

- **a** What baseline value have you assumed?
- **b** What is the contribution of each plus (+) allele?
- **c** Peach, the cow, has a genotype of + − + − + − + − for butterfat level. Samson, the bull, has an identical genotype. Which, if any, of the four calves below could result from the mating of Peach and Samson? Explain.

Calf 1 + + + + + + + +
Calf 2 − − − − − − − −
Calf 3 + − + − + − + −
Calf 4 − − + + − − + +

7 Explain the following observations.

- **a** Most children with cystic fibrosis are born into families with no history of this disorder.
- **b** Normal males have just a single copy of the DMD gene that encodes the dystrophin protein, while females have two copies.
- **c** A polygenic trait that is controlled by a greater number of polygenes shows more phenotypes than a polygenic trait that is controlled by fewer genes.
- **d** A monogenic trait would be expected to show just a few non-overlapping phenotypic classes.

8 Demonstrating knowledge → Using standard notation, write a possible genotype for an X-linked gene for each of the following.

- **a** A female who is homozygous at the gene locus concerned
- **b** A normal male
- **c** A female with an XXX sex chromosome make-up
- **d** A male with Klinefelter syndrome

9 Developing explanations ➜ Leaves of white oak trees (*Quercus alba*) can show two different phenotypes (see figure 15.18).

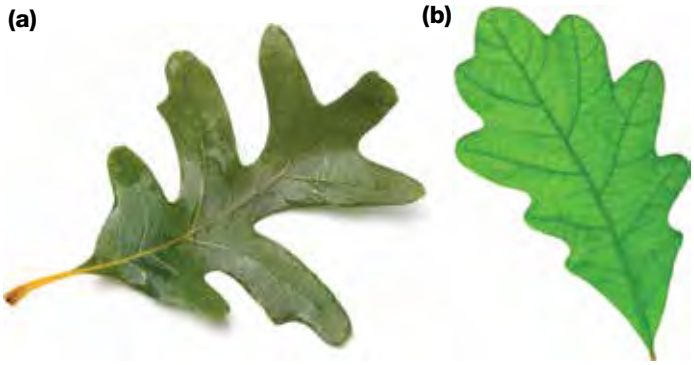

FIGURE 15.18 Two phenotypes of white oak trees (*Quercus alba*)

Further examination revealed that the leaves with the shape shown in figure 15.18a grow on areas of trees that are exposed to full sunlight, while the leaves shaped as in figure 15.18b are located in areas shaded from the sun.

a Do the leaf cells on areas of the same tree exposed to sunlight have the same genotype as those growing in shaded areas? Briefly explain.

b Which of the following is the best biological explanation for the variation in leaf shape on the same tree?

i The tree wants to increase the surface area of the shaded leaves in order to maximise photosynthesis.

ii The phenotype of the leaves is due not only to the genotype, but is also influenced by an environmental factor, namely, light intensity.

iii The genotypes of the leaves change in response to the different environments in which the leaves are growing.

10 Recognising and using allelic notation ➜

a Identify, as fully as possible and including genders, the phenotypes that correspond to the following genotypes. The allelic symbols are those from table 13.7 on page 515.

i Hh ii Cc iii aa

iv Mm v M

b Using the allelic notation in table 13.7, write possible genotypes for the following persons.

i A male hemizygous for red–green colour blindness

ii A male with normal pigmentation, but whose child has the albinism phenotype

iii A female carrier of Duchenne muscular dystrophy

iv A female homozygous for Rhesus positive blood type

v A male with group O blood type

11 Formulating explanatory models ➜ You are given the following information about a particular species of flowering plant:

- Flower colour in this plant species is under genetic control.
- Two flower colours, red and white, are seen in flowers of this plant.

a Based on this information, identify a probable explanation for the inheritance of flower colour in this plant.

b You are later told that, as well as white and red flower colours, other colours are seen in this plant species, including pale pink, medium pink and intense pink.

Given this additional information, how, if at all, would you change your explanation for the inheritance of flower colour in this plant species?

12 Using skills of understanding and analysis ➜ Consider the small number of genes that are located on the short regions of the Y chromosome that are homologous to corresponding regions of the X chromosome.

a For any one of these genes, how many copies are present in a human cell?

b Can these genes be correctly labelled as Y-linked genes? Briefly explain your decision.

c Can these genes be correctly labelled as X-linked genes? Briefly explain your decision.

d Can these genes be correctly labelled as autosomal genes? Briefly explain your decision.

e There is another term 'pseudo-autosomal' that describes genes that are not located on an autosome, but behave as if they were autosomal genes. Might this be an appropriate label for the few genes that are present on both the X and the Y chromosome? Briefly explain your decision.