CHAPTER

9 Changes in genetic make-up of populations

FIGURE 9.1 **(a)** A flock of wild budgerigars in flight in inland Australia. Budgerigar populations in the wild do not vary in colour, with all birds being the so-called light green colour. **(b)** Other colour variants in budgerigars have arisen by mutation from time to time in captive stock. Much of the variation seen in populations of plants and animals has a genetic basis, as is the case for these budgerigar varieties. In this chapter, we will examine inherited variations in populations, their different causes and how the genetic make-up of a population can change under the influence of agents such as selection (both natural and artificial), chance and migration.

KEY KNOWLEDGE

This chapter is designed to enable students to:

- understand the concept of the gene pool of a population
- appreciate the range of genic and chromosomal variants that can exist in a population
- identify the factors that can act to change the allele frequencies in a population's gene pool
- recognise that selection pressure on phenotypes varies with changes in environmental conditions
- gain knowledge and understanding of how gene pools can be manipulated through selective breeding programs and the consequences for genetic variation
- become aware of the process of allopatric speciation
- appreciate the value of genetic diversity in a population and the importance of its preservation.

ODD FACT

Milk from dairy goats, dairy sheep and even buffalo is in demand in Australia for use in the manufacture of specialist cheeses. A market for goats' milk also exists for people with allergies to cows' milk.

Can you drink milk?

A glass of cold milk — plain or flavoured — is a refreshing drink for many people (see figure 9.2). In the year 2014–2015 in Australia, sales of drinking milk, principally cows' milk, totalled 2481 million litres. Unflavoured fresh milk comes as regular (at least 3.2% fat), reduced fat (no more than 1.5% fat) and 'no fat' or skim milk (contains a maximum of 0.15% fat).

FIGURE 9.2 Some of the many fresh milks on sale — regular, reduced fat and low fat, and plain and flavoured.

Like other baby mammals, human babies can be sustained on a diet exclusively of breast milk for a significant period after birth. In the case of human babies, the World Health Organization identifies this as a six-month period after which complementary foods are added to a baby's diet.

Milk contains lactose, a disaccharide sugar, at concentrations of about 5 grams per 100 mL. Lactose must be broken down to its monosaccharide components, glucose and galactose, before it can be absorbed from the gut. This reaction is catalysed by the enzyme **lactase**, which is released from cells lining the small intestine:

$$\text{lactose} \xrightarrow{\textit{lactase}} \text{glucose} + \text{galactose}$$

In humans, the lactase enzyme is encoded by the **LCT** gene on the number-2 chromosome.

Human infants up to about three to four years of age can digest lactose. However, after that time, the majority (about 70%) of the world's population start to lose their lactase enzyme activity and, by about seven or eight years of age, cannot digest lactose. This situation is seen in many people whose ancestry is African, Asian or southern European. For such people, drinking fresh milk can result in abdominal cramps, bloating and watery diarrhoea. Such people are said to be **lactase nonpersistent** (sometimes termed lactose intolerant). Lactase nonpersistence is a recessive trait.

Reminder: The ending for an enzyme name is ***-ase***, for example, lactase. The ending for a sugar is ***-ose***, hence glucose, galactose and lactose.

FIGURE 9.3 A glass of milk being enjoyed by a lactase persistent adult.

ODD FACT

Cats and dogs are commonly lactase nonpersistent. Pet food manufacturers produce a special milk for these family pets that is low in lactose, such as Whiskas® Milk Plus, a lactose-reduced milk for cats.

In contrast, the majority of people whose ancestry is from northern Europe retain their lactase enzyme activity into adulthood and are said to be **lactase persistent**. For lactase persistent people, milk can be a source of nutrition throughout their lives (see figure 9.3). Lactase persistence is an autosomal dominant trait.

When several discrete inherited variations are present in the members of a population, that population is said to be **polymorphic** for the trait concerned. The existence of the lactase persistent/nonpersistent variants in human populations is one example of a polymorphism.

Distribution of lactase persistence

All young non-human mammals, under natural conditions, do not drink milk again after being weaned. For these mammals, no disadvantage is associated with the absence of lactase enzyme activity when they are adults. A similar situation also existed in human prehistory. The ancestral state in human populations was lactase nonpersistence, meaning that all members of early modern human populations were lactase nonpersistent. The appearance of lactase persistence in human populations is a more recent occurrence.

Figure 9.4 shows current regional levels of nonpersistence of lactase enzyme activity (lactose intolerance). Note that in some regions, such as east Asia, the population contains a high percentage of lactase nonpersistent individuals. Note that indigenous populations in Australia and North America are lactase nonpersistent. What does this suggest about their traditional diets?

Table 9.1 provides data for various population groups for lactase persistence/nonpersistence. Note, in particular, the contrasting situation between some African indigenous groups and those from North America and Australia.

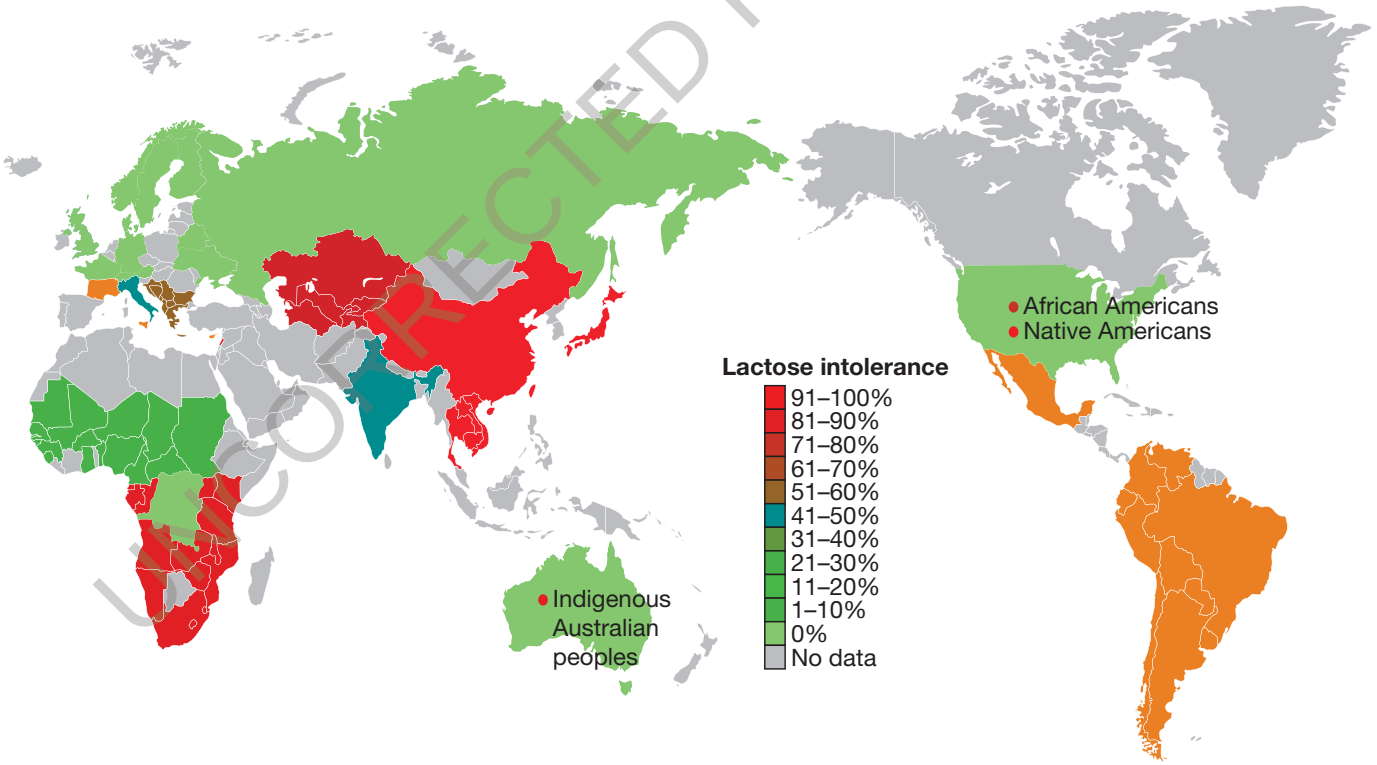

FIGURE 9.4 Map showing the levels of lactose intolerance (lactase nonpersistence) in various regions of the world. The green-shaded areas indicate regions where the majority of people are lactose tolerant (lactase persistent) and the red-shaded areas are regions where the majority of people are lactose intolerant (lactase nonpersistent). Note that within the Australian population, the Indigenous peoples are lactose intolerant.

TABLE 9.1 Percentages of lactase nonpersistent individuals in samples of various human population groups. Of the African ethnic groups — Igbo, Yoruba, Tutsi and Fulani — some come from regions where cattle cannot survive because of tsetse fly infestation. Can you identify the ethnic groups from these regions?

Population sample	Lactase nonpersistence (%)
Native Americans	100
Thai	99
Igbo (West Africa)	99
Yoruba (West Africa)	99
Bantu (Central to Southern Africa)	99
Chinese	88
Indigenous Australian peoples	85
African Americans	70
Bedouin (North Africa to the Middle East)	25
Tutsi (Central Africa)	20
Fulani (West Africa)	18
French	17
White Americans	15
Swiss	10
Swedish	6
Australians (European)	4
Danes	4
Dutch	1

Spread of lactase persistence

Perhaps about 10 000 years ago, a mutation occurred in a hunter-gatherer population that produced the allele ***C*** for persistence of lactase enzyme activity into adulthood. When lactase persistence first appeared in human populations, this mutation gave no benefit.

Soon afterwards, a cultural change occurred in some human populations that would give a selective advantage to people with lactase persistence. This cultural change was the shift from a hunter-gatherer lifestyle to that of farming.

This transition began in the Middle East about 11 000 years ago and then spread to Europe. Farming with animals — cows, sheep or goats — provided not only meat, but also a source of milk (see figure 9.5). At first, raw milk could not be tolerated by the members of early farming communities because they were all lactase nonpersistent. Archaeological evidence of early cheesemaking indicates that the early farmers may have made fermented milk products such as cheese. The fermentation process involved in making cheese, and other dairy products such as yogurt, breaks down most of the lactose, and so these dairy products could have been consumed by lactase nonpersistent people.

Perhaps about 7000 years ago, the lactase persistent mutation began to spread through farming communities. **Agricultural herding of animals that produced milk for human consumption became an agent of positive selection for individuals with the mutant allele for lactase persistence.** A mutant allele that had previously been of no value became an asset in the new environment of dairy farming. Raw milk provided an additional source of nutrition for lactase persistent members of the population. Groups of lactase persistent farmers with their herds began to spread across Europe.

ODD FACT

One speculation is that a large proportion of Europeans may be descendants of the early lactase persistent dairy farmers in Europe.

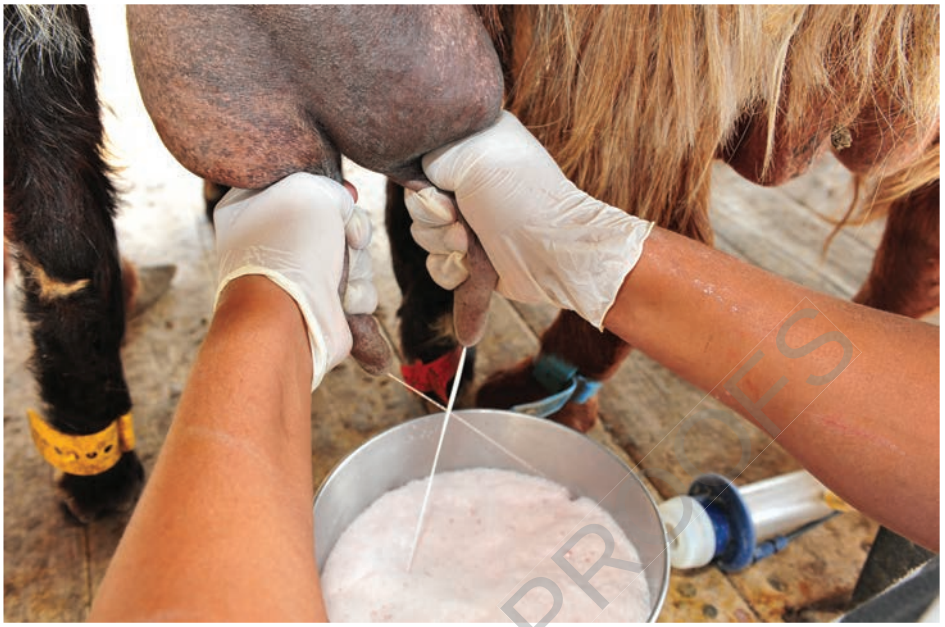

FIGURE 9.5 Goats are one example of a herd animal that can provide milk. Here we see a goat being milked.

The spread of dairy farming across Europe is associated with the spread of the ***C*** allele for lactase persistence. In populations that subsisted on dairying, individuals with even a single copy of the ***C*** allele were able to consume milk and milk products into adulthood, giving them an additional source of nutrition that was not available to lactase nonpersistent people, genotype ***cc***, in the same population. Within a dairy-farming environment, lactase persistent individuals have a **selective advantage** over those individuals who are lactase nonpersistent.

In populations where dairy farming was practised, lactase persistence increased in frequency over successive generations, producing the high levels seen today in Europe and in some African ethnic groups. This increased frequency is evidence of the selective advantage conferred by the presence of the lactase persistence allele, either in a single copy in the heterozygous condition (***Cc***) or as two copies in the homozygous condition (***CC***). A selective advantage confers greater genetic fitness on people possessing the lactase persistence allele. Greater genetic fitness is seen in the greater survival rate to reproductive age and a greater reproductive success as expressed in the contribution of genes to the next generation (see figure 9.6).

Refer back to table 9.1. The link between dairy farming and a high level of lactase persistence in a population is also seen in some African ethnic groups, such as the Tutsi people and the Fulani people, whose traditional diet includes fresh milk from their herds of cattle or goats. This link is also seen in nomadic Bedouin ethnic groups whose camels provide both transport and a source of milk.

So, the pattern that emerged is:

- populations that have not had fresh milk as part of their historical diet have high proportions of lactase nonpersistent individuals, and the presence of the lactase persistence allele is of no benefit in a non-dairying environment
- populations that have had fresh milk as part of their historical diet have high proportions of lactase persistent individuals because the lactase persistence allele conferred a selective advantage in a dairy farming and herding environment.

FIGURE 9.6 Who is fitter (in a genetic sense) — the Olympic weightlifter who is sterile or the relative 'weakling'? Genetic fitness relates to the contribution of genes to the next generation. So, the organism that hides and survives to reproduce is fitter in a genetic sense than the organism that fights and dies before reproducing.

Populations have a pool of inherited (genetic) variation. A change in the environmental conditions in which a population lives can act as an **agent of selection**. The action of an agent of selection can produce differential survival and reproduction rates in members of a population, depending on differences in their inherited physical, physiological or behavioural characteristics. Where members of a population, because of their particular genetic make-up or genotype, have a higher survival and reproductive rate compared to other members of the population, they are said to have a selective advantage.

The lactase persistence/nonpersistence variation that exists in human populations today is just one example of the inherited variation that exists in plant and animal populations. In the next section, we will explore other examples of inherited variations in populations.

KEY IDEAS

- Human populations from various regions show a genetic polymorphism in terms of the persistence/nonpersistence of lactase activity.
- During their childhoods, individuals who are lactase nonpersistent lose the ability to digest milk, while those who are lactase persistent retain this ability into adulthood.
- Lactase nonpersistence is the ancestral state, and lactase persistence arose as a result of a later mutation.
- The emergence of dairy herding was an agent of positive selection for lactase persistence.

QUICK CHECK

1 What prediction may be made about the relative proportions of lactase persistence and nonpersistence in a population that, early in its history, developed dairy farming?

2 Identify the following statements as true or false:

a The more common state in the human global population is lactase nonpersistence.

b Lactase persistence is a result of a mutation that disables the gene that encodes the lactase enzyme.

c The frequency of lactase persistent individuals in the Tutsi population is greater than in a Chinese population.

d Lactase persistence is a dominant trait.

e The lactase persistence allele (***C***) gives any person possessing it a selective advantage.

Inherited variation in populations

The raw material of evolution is the presence in populations of inherited (genetic) variation. **Charles Darwin recognised that if evolutionary change was to occur in a population, it could only do so if inherited variation was present in that population.** Inherited variation may be expressed as structural, physiological, biochemical or behavioural traits.

Inherited variation in a population may have:

- a genic basis: that is, variation that arises from the action of one or more genes
  - action of a single gene
  - action of several genes (polygenes).

- a chromosomal basis: that is, variation that arises as a result of changes in chromosomal make-up
  - gain of entire set(s) of chromosomes (polyploidy)
  - gain or loss of a single chromosome (aneuploidy), where a gain produces a chromosomal trisomy, and a loss produces a monosomy.

Let us look first at variations in population that arise from the action of a single gene. You have already met one of these inherited variations, namely, the lactase persistence/nonpersistence states in people, which are due to the action of alleles of a single gene.

Variation due to a single gene

The relationships between the three alleles of the ABO gene are detailed in *Nature of Biology Book 1 Fifth Edition*, page 557.

Inherited traits that exist as two or three discrete (non-overlapping) variants in a population are typically controlled by a single gene; these single genes often have just two common alleles. When the relationship between the expression of these alleles is one of simple dominance and recessiveness, two variants of the trait can exist, as, for example, long and short fur length in cats, and black or liver coat colour in dogs. When the relationship between the two alleles is one of co-dominance, three variants will exist, as, for example, red, pink and white petal colour. In the case of the ABO blood type gene, four ABO blood type variants (A, B, AB and O) are produced because this gene has three alleles.

Let's look first at examples of inherited discrete variations that are controlled by a single gene.

Variation in people (see figure 9.7a):

- lactase activity — persistent and nonpersistent
- ABO blood types — A, B, AB and O
- hair colour — red and non-red (non-red covers all other colours)
- haemoglobin types — Hb A and Hb S
- tasting ability — able to taste PTC and unable to taste PTC.

Variation in cats (see figure 9.7b):

- pattern — striped (tabby) and non-striped
- colour — black and ginger; grey and cream
- skeletal structure — tailed and no tail (Manx).

Variation in domestic dogs (see figure 9.7c; refer also to figures 9.50 and 9.64 on pages 426 and 440):

- colour — black and liver
- hair length — long and short.

Variation in insects

- response to pesticides — resistant and sensitive
- wing pattern — spotted and solid (see figure 9.7d).

Variation in plants (see figures 9.7e and 9.7f)

- flower colour (many species) — red, pink and white, etc.
- growth habit — prostrate (ground hugging) and erect.

Inherited traits that differ in their expression, such as all the traits shown in figure 9.7, are called **polymorphisms** (*poly-* = many, *morphe* = shape, form). These polymorphic traits are encoded by one gene that has two or more alleles. When more than one variant of a trait is seen in a population, the population can be said to be polymorphic. In contrast, when all members are identical in terms of a particular trait, the population is said to be **monomorphic** (*mono-* = alone, single).

In each case, members of a population can be separated into a small number of discrete non-overlapping classes that can be readily identified, and the number in each class can be quantified.

In this section, the focus is on inherited variants that are discrete or non-overlapping; in other words, the variation is **discontinuous**, with a clear demarcation between the different classes. (It is easy to separate red from pink and white.)

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FIGURE 9.7 Examples of discrete inherited variations controlled by single genes in several different species. **(a)** At left: in people, the A, B, AB and O blood groups are variations that result from various combinations of the three alleles of the ABO gene; at right: the ability to taste bitter substances, such as the chemical phenylthiocarbamide (PTC) as well as the chemically related isothiocyanates (ITC) present in vegetables such as cabbage and broccoli, is under the control of a single gene, with some people able to detect the bitterness while others are unable to detect any bitterness. (Right-hand image in (a) courtesy of Judith Kinnear.)

(continued)

FIGURE 9.7 *(continued)* **(b)** Single gene variants in cats include fur colour (black and orange), colour density (black and grey), white spotting (extensive and limited) and fur length (short and long haired). (Images in (b) courtesy of Judith Kinnear.) **(c)** At left and centre: single gene variants in dogs showing variation in white spotting pattern (Irish pattern and piebald), coat colour (yellow and black) and coat length (long and short haired); at right: short-haired dogs showing colour variation (black and yellow). (Right-hand image in (c) courtesy of Judith Kinnear.) **(d)** Wing pattern variation in the peppered moth, *Biston betularia*, typica variety and solid or melanic variety. **(e)** Petal colour variation in the harlequin flower, *Sparaxis tricolor*. (Images in (e) courtesy of Judith Kinnear.) **(f)** Variation in growth habits as seen in juniper species. *Juniperus chinensis* has an erect growth habit while *J. procumbens* has a prostrate growth habit (at front). In several plant species, this variation has been shown to be due to the action of a single gene. (Image (f) courtesy of Judith Kinnear.)

Let's now look at inherited variation under the control of many genes (polygenes), producing variation that tends to be continuous.

Variation due to polygenes

Inherited variation between adult members of a population can involve subtle differences, such as slight variations in body dimensions. Red-winged blackbirds, for example, show variation in the lengths of their beaks, bodies, tails and wings. Figure 9.8a shows an image of a red-winged blackbird.

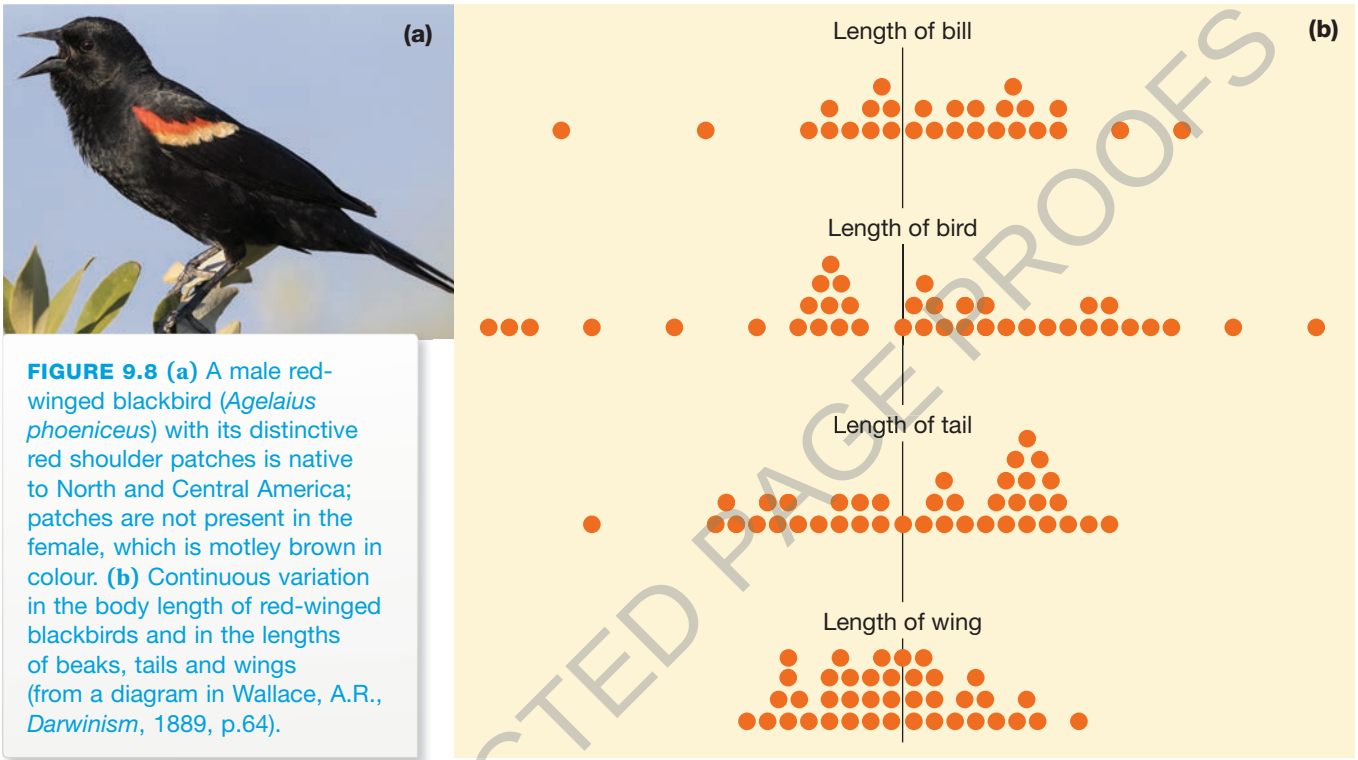

FIGURE 9.8 (a) A male red-winged blackbird (*Agelaius phoeniceus*) with its distinctive red shoulder patches is native to North and Central America; patches are not present in the female, which is motley brown in colour. **(b)** Continuous variation in the body length of red-winged blackbirds and in the lengths of beaks, tails and wings (from a diagram in Wallace, A.R., *Darwinism*, 1889, p.64).

Variation of the type shown in figure 9.8b is **continuous**. Other examples of continuous variation include fat content of cows' milk, mass of pea seeds, and many human traits including adult height, body mass index, waist-hip ratio, blood lipid levels and, in females, age at first menstrual bleeding (menarche). Variation of this continuous type is often controlled by a large number of **polygenes**, each with a small but additive effect. The greater the number of polygenes, the greater the number of possible variants. (Many of these traits are also influenced by environmental factors including nutrition, level of activity, and disease incidence.)

For traits controlled by polygenes, the variation seen in members of a population does not fall into a few non-overlapping classes. Instead, the variants are distributed across a continuous range. Figure 9.9 shows the contrast between the continuous distribution of variants for adult male height, a polygenic trait, and the discrete distribution of the same group of people for lactase persistence and nonpersistence, traits controlled by a single gene.

Polygenic inheritance is discussed in *Nature of Biology Book 1 Fifth Edition*, pages 561–563.

In one group of birds, commonly called Darwin's finches, subtle differences in the height and width dimensions of their beaks have been recorded. In this case, the variation is the result of the action of just one gene, the **BMP4** master gene. Shifts in the timing, the level of expression and where this gene is expressed generate a range of different beak shapes (see chapter 11, pages 533–534. This is a different way of generating variants compared to that resulting from the action of polygenes.

Chromosomal variation: polyploidy

Polyploidy refers to a condition in which an organism has more than two matched sets of chromosomes. When just the two matched sets of chromosomes are present, an organism (and its cells) are said to be **diploid**. The presence of one or more additional sets of chromosomes to the base diploid condition (2 sets) produces organisms that are termed triploid (3 sets), tetraploid (4 sets), hexaploid (6 sets) and even dodecaploid (12 sets).

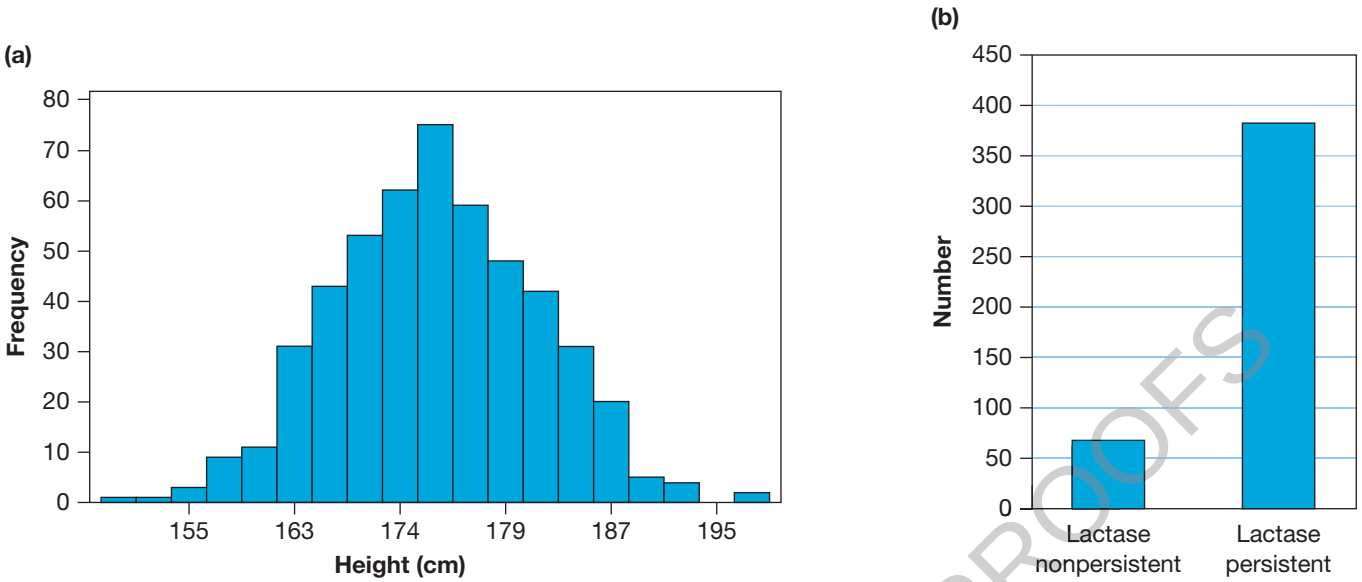

FIGURE 9.9 (a) Continuous distribution of heights in a sample of adult males. **(b)** Discontinuous distribution of lactase persistence/nonpersistence status of the same group. Which distribution shows variation controlled by one gene with two alleles with a simple dominant/recessive relationship?

Polyploidy is very common in flowering plants and ferns. As well as occurring naturally, polyploid plants can also be artificially created. This is deliberately done because polyploid flowering plants tend to be bigger than their diploid relatives and have larger flowers. This has occurred in several ornamental gardens, such as tetraploid snapdragons (*Antirrhinum majus*) $4\times = 64$ (see figure 9.10).

FIGURE 9.10 Tetraploid ($4\times = 64$) snapdragons are bigger, showier and hardier than their diploid ($2n = 32$) close relatives.

Polyploidy is much rarer in animals. Among polyploid animals are the various species of tetraploid salmon; for example, the Chinook salmon (*Ororhynchus tshawytscha*) has a chromosome number of 4× =68. Goldfish (*Carassius auratus*) and carp (*Cyprinus carpio*) are both tetraploid, each with a chromosome number of about 104.

Table 9.2 shows some of the polyploid flowering plants and their numbers of chromosome sets. Note that various cordgrass species in the genus *Spartina* have different levels of polyploidy, up to dodecaploid (12× = 120).

TABLE 9.2 Polyploidy in flowering plants.

Plant	Chromosome number	Description
watermelon, *Citrullus lanatus*	2× = 22, 3× = 33, and 4× =44	diploid, triploid and tetraploid
banana, *Musa acuminata*	3× = 33	triploid
durum wheat, *Triticum turgidum*	4× = 28	tetraploid
emmer wheat, *Triticum dicoccum*	4× = 28	tetraploid
cotton, *Gossypium hirsutum*	4× = 52	tetraploid
potato, *Solanum tuberosum*	4× = 48	tetraploid
kiwifruit, *Actinidia deliciosa*	6× = 174	hexaploid
bread wheat, *Triticum aestivum*	6× = 42	hexaploid
oat, *Avena sativa*	6× = 42	hexaploid
cordgrasses, *Spartina* spp.	4× = 40, 6× = 60, and 12× = 120	tetra-, hexa- and dodecaploid

In some cases, a polyploid individual will have additional complete sets of chromosomes from its own species — this is a situation termed **autopolyploidy**, as seen in snapdragons, bananas and watermelon. The polyploidy condition produces plants with larger cells and larger organs, such as flowers, fruits, leaves and even chloroplast counts in leaf cells.

In other cases, the additional chromosome sets in a polyploidy may come from another species. In this case, the resulting organism is called an **allopolyploid**. Many allopolyploids show 'hybrid vigour', indicating that they are more robust and display superior qualities as compared to the parents from which they were derived.

Bread wheat (*Triticum aestivum*) is an allopolyploid (6× = 42) that arose spontaneously in about 7500 BC. Research has shown that its sets of chromosomes came from three different species, not at once, but in a pathway involving several crosses between different species (hybridisations) (see figure 9.11).

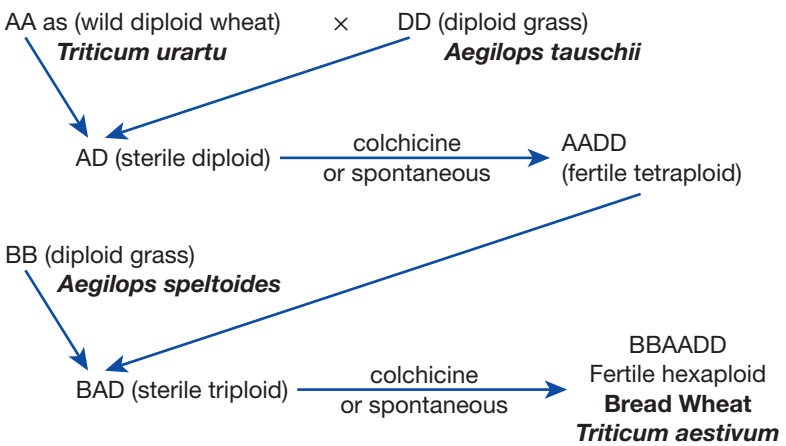

FIGURE 9.11 Formation of hexaploid bread wheat, *Triticum aestivum* (6× = 42) from three antecedent species, including a wild diploid wheat, *Triticum urartu*, and two wild species of grasses.

How are polyploids formed?

Figure 9.12 summarises the various ways in which polyploid plants can be produced, both autopolyploid and allopolyploids. The red circle denoted by A indicates the basic (haploid) chromosome set of the diploid species AA, while the blue square indicates the basic (haploid) chromosome set of diploid species BB.

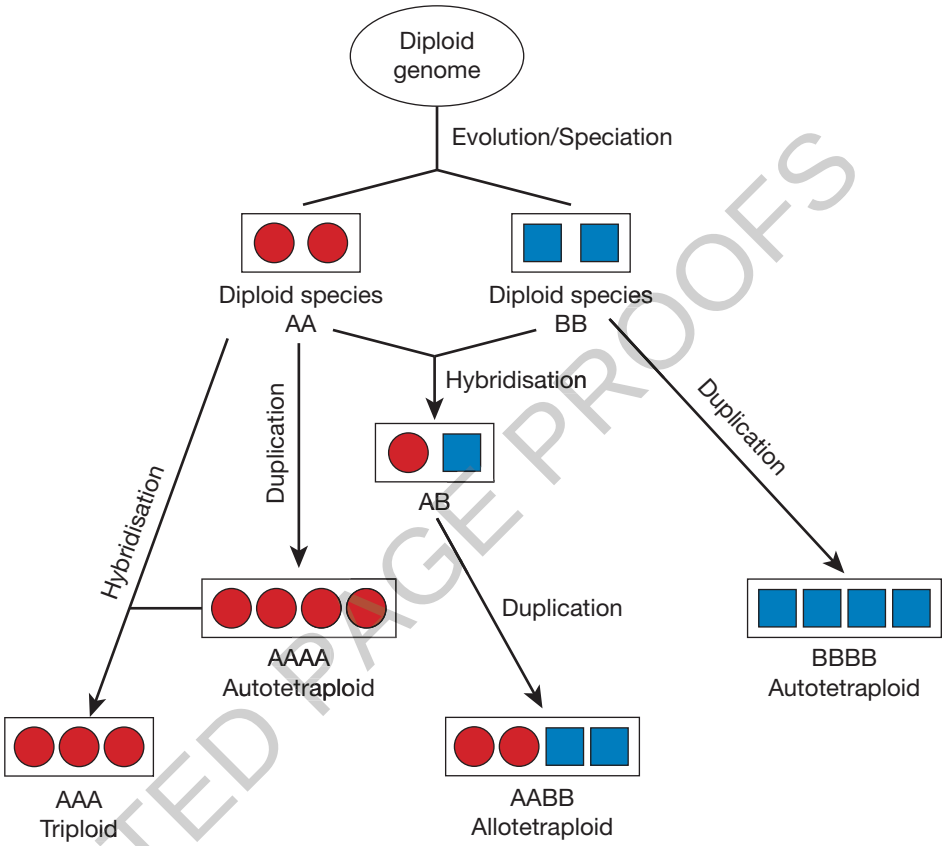

FIGURE 9.12 Various strategies to make polyploidy plants.

The various processes involved in producing polyploidy organisms include:

- duplication that involves chemical treatment to produce a doubling of the chromosome number; this is most commonly achieved through colchicine treatment
- **hybridisation** that involves crossing parents belonging to two different species, as for example:
  - crossing two different diploid species to produce a diploid hybrid progeny
  - crossing a tetraploid and a diploid to produce triploid progeny.

Polyploidy has changed populations by creating new chromosomal combinations and producing new species that display new variations.

Chromosomal variation: aneuploidy

An **aneuploid** organism is one that has a chromosome number that differs by a small number from the standard chromosome number for its species. The difference is typically one or two chromosomes more than the standard number or one or two chromosomes less.

Loss of chromosome(s): The loss of one of a pair of chromosomes or both members of a pair of chromosomes would be expected to create a lethal condition in diploid organisms because of the loss of the function of the genes carried on those chromosomes. However, polyploid organisms can tolerate the loss of one or even two members of a pair of chromosomes because of the presence of other chromosomes.

Gain of chromosome(s): The gain of a chromosome would not be expected to benefit the organism concerned because of the chromosomal imbalance

it would produce. In humans, for example, the only autosomal trisomy that survives beyond infancy is Down syndrome, with three copies of the number-21 chromosome. All other autosomal trisomies either result in spontaneous miscarriages or, in the case of trisomy 13 and trisomy 18, the birth of a baby with severe abnormalities, both physical and mental. In both trisomy 13 and trisomy 18, the expected life span is very short, measured in days or weeks.

However, one plant shows an interesting range of variation for fruit shape resulting from chromosomal trisomy events. Thorn apple (*Datura stramonium*), an introduced plant, is a declared noxious weed in many Australian states. The thorn apple is an annual herb that grows to about one metre in height and has large white, bell-shaped flowers. Its fruits, about half the size of an apple, are covered by thorns.

Thorn apples have twelve pairs of chromosomes ($2n = 24$) and show variation in fruit shape. The variations in fruit shape are chromosomal in origin. Each different shape is due to the presence of one specific additional chromosome. Some of the different fruit shapes are shown in figure 9.13, and the additional chromosome that causes the particular shape is identified by a letter.

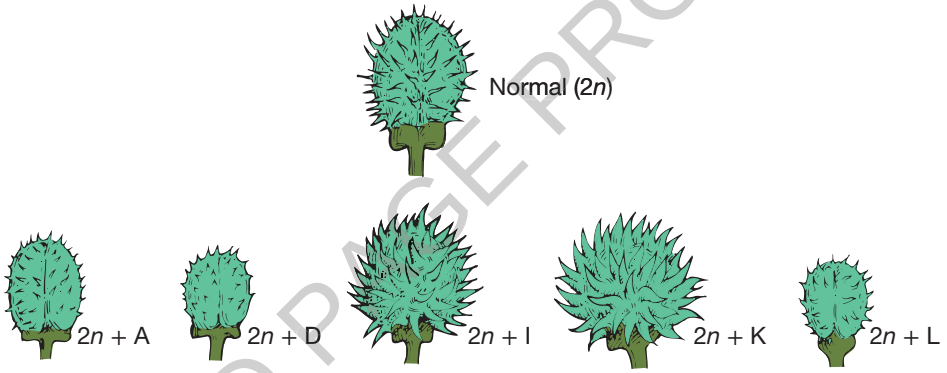

FIGURE 9.13 Some variations seen in fruit shapes in thorn apple. The fruit of the normal diploid thorn apple is shown for comparison. In the case of an additional copy of the D chromosome, how has the fruit shape been affected?

HOW TO MAKE A SEEDLESS WATERMELON

Seedless watermelons are the fruits of a sterile triploid ($3\times = 33$) hybrid plant. As such, these watermelon plants cannot reproduce, and viable seeds are not present in their fruits (see figure 9.14). Seedless watermelons were first marketed in the 1990s and they now dominate the market. No-one wants to bite into a watermelon with large hard black seeds!

FIGURE 9.14 Seedless watermelons are triploid ($3\times = 33$). (Image courtesy of Judith Kinnear.)

To make a seedless triploid watermelon, two plants are needed:

1. A standard diploid ($2n = 22$) watermelon plant to be a source of pollen.
   This pollen is formed by the usual process of meiosis so that the pollen is haploid, with a single set of 11 chromosomes.
2. A tetraploid ($4\times = 44$) plant to be a source of eggs.
   Tetraploid watermelons are autopolyploids that are produced artificially by colchicine treatment. These eggs are formed by the normal process of meiosis, and each egg has half the number of chromosomes as the tetraploid parent. Each egg of course has 22 chromosomes, the diploid number, and they comprise two sets of watermelon chromosomes.

To make a triploid watermelon plant, artificial pollination is required. Pollen grains, each with a single set of 11 chromosomes, are dusted on the stigma of a female flower of the tetraploid plant. In the ovary of this plant are diploid eggs, each with two sets of chromosomes, 22 chromosomes in total. The resulting fertilisation produces seeds, each with three sets of chromosomes, totalling 11 + 22 = 33 chromosomes (see figure 9.15).

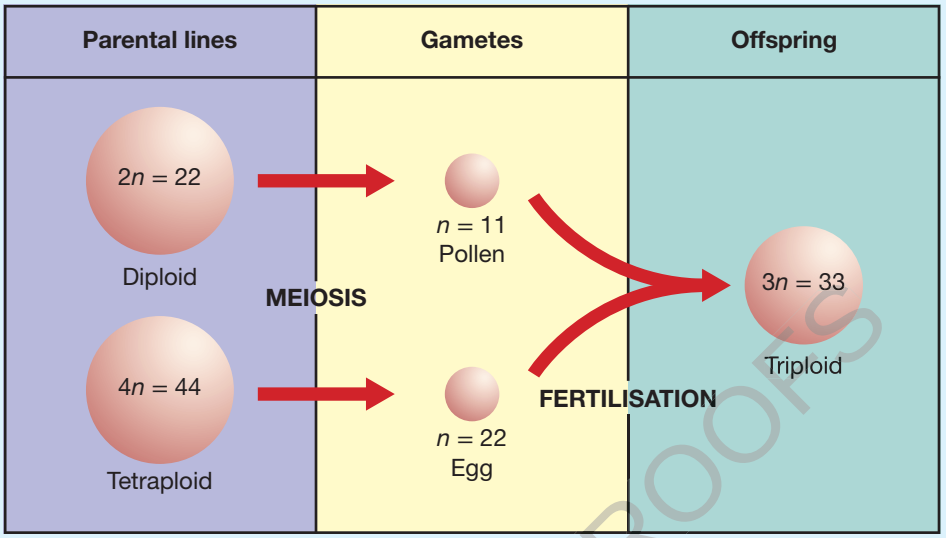

FIGURE 9.15 Diagram showing a simplified version of the process required to produce triploid watermelons with seedless fruit. The tetraploid (4× = 44) parent watermelon is an autopolyploid that is produced artificially using colchicine treatment.

When three copies of each chromosome are present, as in a triploid watermelon, the process of meiosis cannot proceed in an orderly manner. The seedlings that germinate from these triploid seeds cannot produce viable eggs and pollen, and their fruits have small soft white traces instead of large hard black seeds.

Each new generation of triploid watermelon plants must be created anew by seed companies. These companies maintain diploid and tetraploid parental lines of watermelons, which they must pollinate by hand to produce more triploid seeds.

KEY IDEAS

- Variation can exist among members of a population in the wild and under domesticated conditions.
- Inherited variation in a trait may result from the action of different alleles of one gene or from the action of polygenes.
- Variation in a trait may be classified as continuous or discontinuous.
- Polyploidy, either spontaneous or artificially created, results in new chromosomal combinations and new species.
- Aneuploid changes in chromosome numbers are generally poorly, if at all, tolerated in animals but can be tolerated in polyploidy plants.

QUICK CHECK

3 An inherited trait in a population shows two variations only. How many genes are likely to be involved?

4 A population of cattle varies in coat colour, being red, roan or white. Is this variation continuous or discontinuous?

5 What is the difference between the members of the following pairs:
- **a** aneuploidy and polyploidy
- **b** continuous and discontinuous variation
- **c** autopolyploid and allopolyploid organisms
- **d** tetraploid and hexaploid?

6 Identify the following statements as true or false:
- **a** Polyploidy is far more common in plants than in animals.
- **b** Salmon is an example of a polyploidy animal.
- **c** A triploid plant would be expected to be fertile.
- **d** Hybrids are the result of crossing two different species.

Genes in populations

A **population** refers to an interbreeding group of organisms of the same species living in the same region at the same time. So, we can talk about the present population of lions (*Panthera leo*) on the Serengeti Plains or the human population of Milan during the period of the Black Death. A population may be as small as a group of one species on a remote island or isolated on a mountain top or it may be as large as the gene pool of one species on a continent.

Each individual in a population has a total set of genetic information known as its genotype. For example, in sheep, the gene that controls wool colour has the alleles white (***W***) and black (***w***), with white being dominant to black. Sheep genotypes at this gene locus are expressed as a combination of two alleles: for example, either ***WW*** or ***Ww*** for a white sheep, and ***ww*** for a black sheep (see figure 9.16a).

Populations have gene pools

An individual organism has a genotype that identifies the alleles present at each gene locus. A population has a **gene pool** that **consists of the sum of the genotypes of all the members of that population**. Gene pools are described by the frequencies (proportions) of the alleles of each gene present.

To understand allele frequencies, look at figure 9.16b and work through the following information, which will give us the frequencies of the alleles for the gene for wool colour in sheep:

1. There are ten sheep, each with two alleles, so the total number of alleles is $10 \times 2 = 20$.
2. Using the genotypes of the sheep as shown, we can count the number of each allele:

   Number of ***w*** alleles = 6, and number of ***W*** alleles = 14.

3. So, the frequency of the ***w*** allele = 6/20 = 3/10 = 0.3, and the frequency of the ***W*** allele = 14/20 = 7/10 = 0.7.

The frequency of the allele that determines the dominant phenotype is typically denoted by the letter 'p' and that of the allele that determines the recessive phenotype is denoted by the letter 'q'.

So, for this limited gene pool, we can write: freq (***W***) = p = 0.7 and freq (***w***) = q = 0.3.

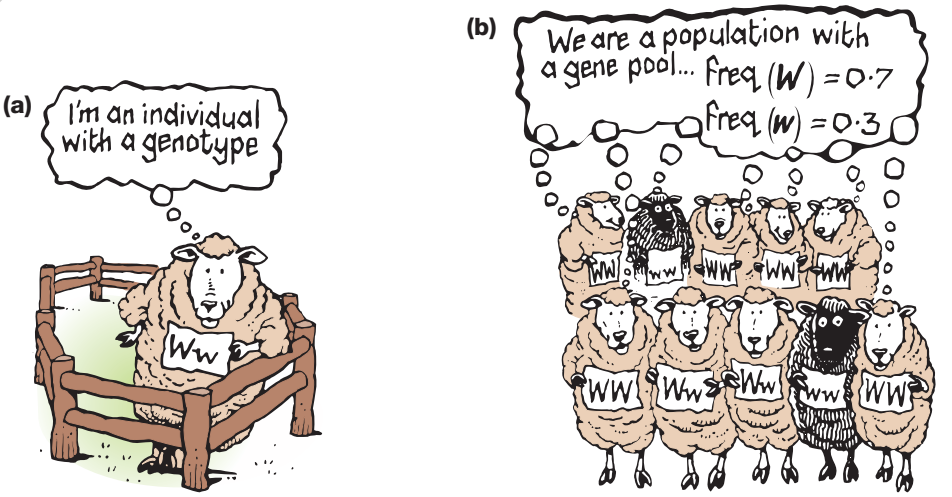

FIGURE 9.16 **(a)** Genotype of individual sheep. **(b)** Gene pool of sheep population.

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Note that an allele frequency in a population can have any value from 0 to 1. When the frequency of an allele is equal to 1, all organisms in that population are homozygous at that particular gene locus and show the same phenotype for the trait concerned. When this happens, the allele is said to be **fixed** and, in this case, the frequency of that allele is 1 and that of the alternative allele must be 0. When an allele becomes fixed in a population, the genetic diversity of the gene pool is reduced by the loss of the alternative allele.

Figure 9.16b involves some sleight of hand. What is it? You cannot simply look at a white sheep in a mixed flock and identify its genotype with certainty — it could be ***WW*** or it could be ***Ww***. (No problem with black sheep as they can only be ***ww***.) In reality, in a population where two alleles exist with a simple dominant/recessive relationship, it is not possible to do a direct count of the two alleles such as we have just done above. However, a neat equation overcomes this difficulty. Refer to the box titled *Using the Hardy–Weinberg equation* on page 394.

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Allele frequencies tend to stay constant

In 1908, Godfrey Hardy (1877–1947), a British mathematician working at Cambridge, and Wilhelm Weinberg (1862–1937), a German physician, working independently, identified an important feature of populations. Hardy and Weinberg recognised that allele frequencies in populations tend to remain constant, generation after generation, provided a particular set of conditions is met. This concept is known as the **Hardy–Weinberg principle**.

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The conditions that apply in a population if allele frequencies are to remain constant over generations are as follows:

- The population must be large.
- Members of the population must mate at random; **random mating** contrasts with non-random or assortative mating in which matings are restricted to those between organisms of like phenotypes, such as blue × blue and green × green. (In random mating, all kinds of crosses are equally likely, such as blue × blue, and blue × green.)
- All matings are equally fertile, producing equal numbers of viable offspring.
- The population must be closed, with no migration either into or out of the population.

Figure 9.17a shows a population in H–W equilibrium in which the allele frequencies for a particular gene remain constant, generation after generation. The ***B*** allele stays at a frequency of about 0.7 (that is, freq (***B***) = 0.7) in the gene pool, and the ***b*** allele at a frequency of about 0.3 (that is, freq (***b***) = 0.3). Figure 9.17b shows a population in which the allele frequencies are changing over generations.

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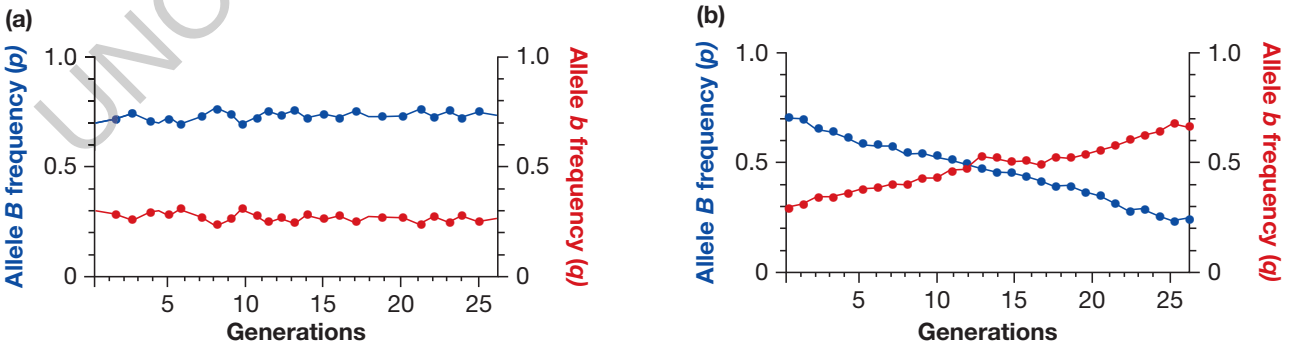

FIGURE 9.17 Frequency of the ***B*** and ***b*** alleles at a particular gene locus over several generations in two populations (**a** and **b**) under different environmental conditions. Which population is in H–W equilibrium?

USING THE HARDY–WEINBERG EQUATION

In this box, we are looking at variation in a population determined by two alleles of a particular gene, and where the relationship between the alleles is one of simple dominance/recessiveness.

The Hardy–Weinberg equation is: $p^2 + 2pq + q^2 = 1$.

1. What do the terms in this equation denote?
2. How can we estimate the values of the various terms?

1. **Identifying the terms:**
We can take as a given that p and q denote the frequencies in the gene pool of the two alleles of a particular gene. In particular, p denotes the frequency of the allele producing the dominant trait, and q denotes the frequency of the allele controlling the recessive trait. We also known that $p + q = 1$.

Check out figure 9.18, which shows what the terms in the Hardy–Weinberg equation denote.

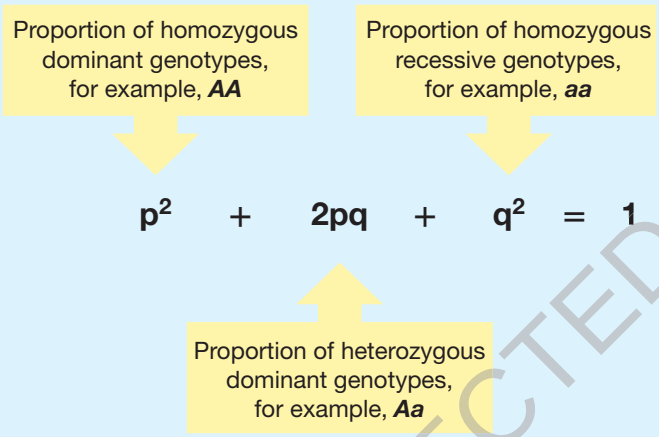

FIGURE 9.18 The Hardy–Weinberg equation can be used to estimate the allele frequencies in a population and, from these values, to estimate probable proportions of unknown genotypes in a population.

2. **Estimating the value of terms:**
The key starting value is q^2. This is the proportion of homozygous recessive individuals in the population under consideration. The homozygous recessive members of the population can be recognised because of their distinctive phenotype, and so they can be counted, making it possible to obtain a value for q^2.
   - Then, knowing q^2, it is easy to obtain the value of q.
   - Then, since $p + q = 1$, it is easy to obtain the value of p.
   - Finally, knowing both p and q, it is possible to calculate estimates of q^2 and 2pq.

Now, you have all the details of the population, both the estimated allele frequencies, and the proportions of the three genotypes in the population.

3. **Applying the principles:**
Consider a population (n = 100) that is tested for lactase persistence/nonpersistence. The results show that 36 individuals are lactase persistent and 64 are lactase nonpersistent. Lactase persistence, ***C***, is dominant to nonpersistence, ***c***.

We are told that the 36 individuals in this population sample are lactase persistent, but how many of these are homozygous ***CC*** and how many are heterozygous ***Cc***?

We can now write:

Given the frequency of ***cc*** individuals = $q^2 = 64/100 = 0.64$

Therefore, $q = \sqrt{0.64} = 0.8$

Then, since $p + q = 1$, $p = 0.2$

We already know that the frequency of individuals with the ***cc*** genotype = $q^2 = 0.64$, that is 64 per cent.

Now that we have the allele frequencies, we can estimate the frequencies (proportions) of the two remaining genotypes in this sample population:

- Frequency of individuals with the ***CC*** genotype = $p^2 = 0.2 \times 0.2 = 0.04$, that is 4 per cent are homozygous persistent.
- Frequency of individuals with the ***Cc*** genotype = $2pq = 2 \times 0.2 \times 0.8 = 0.32$, that is 32 per cent are heterozygous persistent.

Note that the majority of individuals who are lactase persistent are heterozygous ***Cc*** and carry the nonpersistent allele.

So, we can show the lactase status of our sample population as a piechart:

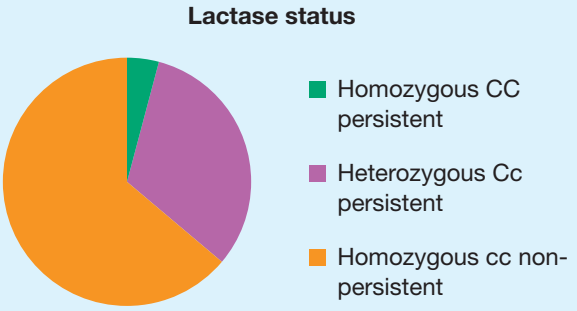

FIGURE 9.19

KEY IDEAS

- The genetic information in an individual is a genotype, while the genetic information in a population is a gene pool.
- A gene pool is described in terms of the allele frequencies (proportions) of each gene.
- Allele frequencies in populations in Hardy–Weinberg equilibrium tend to remain constant generation after generation.
- A population in Hardy–Weinberg equilibrium will be expected to be large, closed and showing random matings, with all matings equally fertile.

QUICK CHECK

7 Identify what is key between a genotype and a gene pool.
8 A population consists of 50 fish.
  a What is the total number of alleles for one autosomal gene in this population?
  b How many different alleles could be present in one fish?
9 Two large populations (M and N) of the same mammalian species contain some members that are pink-eyed and some that are yellow-eyed. Eye colour is under the control of an autosomal gene. Organisms in population M tend to mate with organisms with the same eye colour. However, this mating preference does not occur in population N. One of these populations is in Hardy–Weinberg equilibrium. Explain which population (M or N) this is likely to be.
10 List two conditions that apply to a H–W population.
11 True or false? A closed population is one in which migration, both in and out, is absent.

Change agents in populations

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Unit 4 AOS 1 Topic 1 Concept 8
Outcome of changes in allele frequencies
Summary screen and practice questions

The allele frequencies in a population in Hardy–Weinberg equilibrium tend to remain unchanged, generation after generation. However, in reality, many populations show changes in allele frequencies in their gene pools over time. What agents can bring changes in allele frequencies of these populations?

Agents that can cause allele frequencies to change over time include:

- selection
- gene flow or migration
- chance, as seen in genetic drift, bottlenecks and founder effects.

Let's look at each of these change agents in turn.

Natural selection as an agent of change

Selection that acts on a population can be of two kinds, depending on whether the population that is being acted upon is living under natural conditions or is a captive population. The two kinds of selection are natural selection and artificial selection. In the following section, we will explore natural selection. Artificial selection is discussed later in this chapter (see page 423) in the context of selective breeding.

The action of selection on populations in the wild is termed **natural selection**. We can define **natural selection as the process by which new heritable traits, whether morphological, physiological or behavioural, evolve and persist in a population**.

Natural selection is brought about by a range of **selecting agents** that act on populations. Examples include:

- physical agents, such as climate change or food shortages
- biological agents, such as infectious diseases, predation or competition
- chemical agents, such as a pollutants in soil or water.

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Change agent: natural selection
Summary screen and practice questions

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See more
Change agents and natural selection

In the wild, members of a population must compete with each other for access to living space, energy supplies and mating partners in their habitat. Members of a population are also exposed to competition from other species, to predation, to parasites and to disease-causing microorganisms. Variants within a population react differentially to these pressures, with some being more successful than others. **Natural selection occurs when any selecting agent acts on a population in the wild and produces differences in the survival and reproduction rates of variants in that population.**

Differential reproduction occurs when one variant (phenotype) in a population produces more viable offspring than other variants, and so makes a greater contribution to the gene pool of the next generation. The action of natural selection over successive generations results in an increase in the frequency of this phenotypic variety in the population relative to other varieties. In other words, the frequencies of alleles present that are associated with this phenotype will increase in the gene pool of the population as the frequencies of alternative alleles decrease.

Darwin and Wallace (see chapter 10, p. 444, The Darwin–Wallace view) recognised natural selection as a causative agent of evolution, based on a number of simple observations and resulting deductions, as follows:

Observation 1
In the wild, the numbers of offspring produced by plants and animals over their lifetimes is greater than the number of parents. For example, a single *Eucalyptus* tree produces an enormous number of seeds every year and one female rabbit produces a large number of kits over her lifetime.

Deduction 1
A struggle for survival occurs.

Observation 2
Over time, the size of natural populations tends to remain fairly constant, although population size may fluctuate depending on environmental factors such as drought, disease and plentiful food supplies.

Deduction 2
In this struggle, some variants in a population have a greater chance of survival than others. These variants are favoured under the conditions in a particular environment and are reproductively more successful.

Observation 3
Variation exists in populations of plants and animals; that is, no two organisms in a population are identical, and some variations are inherited.

Deduction 3
Inherited traits present in surviving parents are passed on to their offspring so that the genetic composition of populations can change over time.

The effects of an agent of natural selection acting on a population depend on the time over which it acts. Over a few generations, natural selection can cause a variant that is at a selective advantage to become more common in a population. When the relative numbers of different variants in a population change over a few generations as a result of natural selection, the outcome is **intra-specific evolution** or evolution within a species. Over longer periods, natural selection can result in the formation of subspecies (see figure 9.31 on page 409); and, over geological time, natural selection can result in the formation of new species.

Selection acts on phenotypes

Members of a polymorphic population living *under a particular set of environmental conditions* are exposed to a range of selecting agents and the selection pressures that they produce. Under these circumstances, the various phenotypes (variants) may show different survival and reproductive rates. **A phenotype that makes the greater contribution to the gene pool in the next generation has a higher fitness value and is said to be 'at a selective advantage'.** Phenotypes that make lesser contributions to the gene pool of the next generation are 'less fit' or are said to be 'selected against'.

An observed fitness value for a phenotype is not fixed, but applies under a particular set of environmental conditions. Table 9.3 shows examples of how different agents of natural selection can act on populations and give a selective advantage to one variant over another under a particular set of environmental conditions.

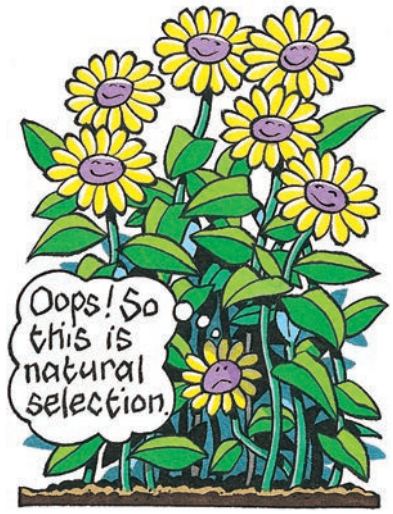

FIGURE 9.20 Natural selection acting on inherited variations in height can result in differential survival of variants. Who will survive — shorter or taller plants?

TABLE 9.3 Action of natural selection in populations with members showing inherited variations. Which plants are at a selective advantage — the faster or the slower growers? In the absence of viral disease, what happens to the selective advantage of resistant rabbits?

Variants present in population	Agent of natural selection	Outcome of natural selection
plants with a noxious chemical and plants lacking chemicals	grazing by herbivores	chemical-producing plants are more likely to survive and reproduce than those lacking chemicals (their leaves and reproductive structures (flowers and fruits) are more likely to be eaten by herbivores)
plants with faster growth rate and plants with slower growth rates	crowding and limited space	faster growing seedlings are more likely to survive and reproduce than slower growers (that are shaded from light and die)
viral-resistant rabbits and non-resistant rabbits	presence of virus in environment	resistant rabbits are more likely to survive and reproduce than non-resistant rabbits (that will be incapacitated or killed by viral disease)

Consider the following example of natural selection in action on a plant population that consists of a small number of copper-tolerant plants and a majority of copper-sensitive plants. Under normal conditions, this inherited variation does not affect the survival or numbers of offspring produced by each plant variant. What happens if the environment changes and the plant population is exposed to the pressure of high concentrations of copper salts in the soil because of seepage from wastes at a copper mine?

The copper-sensitive plants come under strong selection pressure and will be harmed or killed, but the copper-tolerant plants will survive and reproduce at relatively higher rates. Over time, if high levels of copper remain in the soil, the population will eventually consist of copper-tolerant plants only. *Under this particular set of environmental conditions*, the copper-tolerant plants have a selective advantage relative to the copper-sensitive plants in the population. Table 9.4 summarises this situation.

ODD FACT

A study of copper-tolerant plant species growing on sites of copper mine waste found that some copper-tolerant species excluded copper from their shoots by immobilising it in their roots. Neat!

TABLE 9.4 Possible results in a plant population of continuous exposure to copper.

Outcome of introduction of copper pollution	
variations present in plant population	copper-resistant and copper-sensitive plants
environmental change	introduction of copper pollution
agent of natural selection	environmental change
plants with selective advantage	copper-resistant plants
plants with selective disadvantage	copper-sensitive plants
outcome of natural selection over several generations	copper-resistant plants become more common

Now consider the following example of natural selection in action on an insect population.

Figure 9.21a shows a large random mating insect population that includes a small proportion of members that are resistant to a particular pesticide (these insects are shown in red), with the majority being sensitive to the pesticide. Assume that this variation is due to the action of one gene with the alleles ***r*** (sensitive) and ***R*** (resistant). In an environment where there is no pesticide, both the resistant and the sensitive phenotypes survive and reproduce equally. The allele frequencies, as well as the genotype and phenotype frequencies, would be expected to stay constant generation after generation. Why?

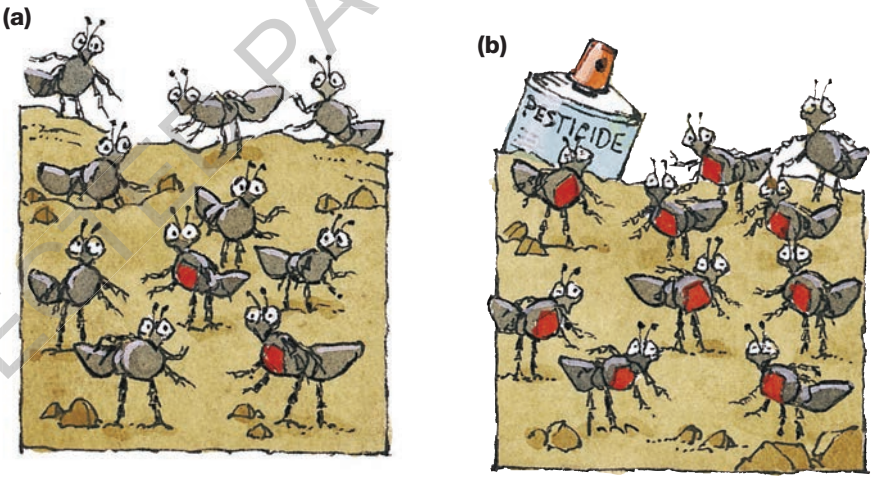

FIGURE 9.21 **(a)** Sample of insect population in pesticide-free environment. The sensitive phenotype (shown as plain grey) is more common than the resistant phenotype (shown as red). **(b)** Sample of insect population after several generations in the presence of pesticide. The resistant phenotype (red) has become more common. Have the allele frequencies changed?

However, assume that the insects' habitat is cleared for farming and pesticide is introduced into the environment. Under these conditions, many of the sensitive insects are killed by the pesticide spray. The resistant insects are now at a selective advantage and **will survive to form the majority of parents to produce the next generation**. What is the agent of selection?

After several generations, with pesticide in the environment, the population changes so that resistant insects form the majority of the population (see figure 9.21b). The pesticide does not cause the resistant phenotype. Resistant insects were present in the original population, and, in the presence of the pesticide, their proportion increased through the action of selection.

Genetic fitness varies in different environments

In human history, important selecting agents affecting populations have included widespread infectious diseases that are major causes of death. Malaria is one such disease (see figure 9.22). For people living in areas of the world affected by the malarial parasites, their fitness depends on whether they

are able to produce haemoglobin S. The parasite cannot survive in red blood cells containing haemoglobin S, which is due to the H^S allele. However, the malarial parasite thrives in normal red blood cells, which contain haemoglobin A, produced through the action of the H^A allele, so this group is at risk of death from malaria. On the other hand, when red blood cells contain only haemoglobin S, this causes the disease sickle-cell anaemia (see table 9.5).

FIGURE 9.22 Coloured scanning electron micrograph (SEM) image of red blood cells. The lower cells are infected with malarial parasites as indicated by the yellowish swollen lumps on the cells, which eventually rupture and spread the infection. The lumps cause the cells to adhere in the brain and other tissues, producing symptoms of malaria, such as recurrent fevers. The normal cells, above, lack these lumps because a disrupting gene in the parasite has reduced their ability to adhere.

Table 9.5 shows how genetic fitness can vary in different environments. Note that the H^AH^S individual is at a selective advantage relative to the others in a malaria-affected environment, but loses that advantage in a malaria-free environment.

TABLE 9.5 The fitness of a phenotype depends on the environment. Is the fitness of people with normal red blood cells containing only haemoglobin A similar in the two environments?

Genotype	Phenotype	Relative fitness
In malaria-affected environments:		
H^AH^A	usual red blood cells with haemoglobin A only	can die from malaria *reduced fitness*
H^AH^S	cells capable of sickling with haemoglobins A and S	minor effects of sickling but resistant to malaria *most fit*
H^SH^S	severe sickle-cell anaemia with haemoglobin S only	resistant to malaria but can die from sickle-cell anaemia *least fit*
In malaria-free environments:		
H^AH^A	usual red blood cells	no effects of sickling *most fit*
H^AH^S	cells capable of sickling	minor effects of sickling *reduced fitness*
H^SH^S	severe sickle-cell anaemia	can die from sickle-cell anaemia *least fit*

Levels of selection

The levels of selection against phenotypes in a population can vary.

- **Complete selection** against a phenotype occurs when any organism with a given phenotype cannot reproduce because of death before reproductive age is reached or because of sterility.
- **Partial selection** against a phenotype occurs when matings involving that phenotype produce on average fewer viable and fertile offspring relative to other matings.

Case studies in selection

The following section includes details of two examples of natural selection in an Australian setting.

Case study 1: Selection in cattle tick populations

The cattle tick (*Boophilus microplus*) is a major pest in northern Australia (see figure 9.23). It carries the parasite that causes tick fever in cattle.

FIGURE 9.23 Cattle showing a heavy infestation of ticks. (Image courtesy of CSIRO Publishing.)

The first attempt to control ticks used arsenic-based cattle dips. At first, this killed many ticks. By 1936, the tick populations were showing resistance to this poison so arsenic use gradually disappeared. By 1946, the insecticide DDT was introduced to control cattle ticks and, at first, this reduced the tick populations. By the early 1960s, however, the tick populations had become resistant to DDT insecticide. The next chemicals brought into the attack on cattle ticks were organophosphate pesticides. Soon after these were introduced, the first resistant ticks were detected and, by the 1980s, resistance to many of these compounds had developed in tick populations. How?

Resistance to each chemical pesticide involves a different gene. Before a particular pesticide was widely used, the frequency in the cattle tick gene pool of the various alleles that confer resistance to these pesticides was low. Once a pesticide became widely used, however, it acted as a selecting agent that produced an increase in the survival and reproduction rates of resistant ticks relative to those of nonresistant ticks. As a result of this selection pressure, the proportion of resistant ticks increased and the frequency of the resistant allele in the gene pool of the population increased. This recurred in turn with different alleles as each new pesticide was introduced. A pesticide does not produce the resistant allele. The resistant allele is already present in the gene pool of the tick population. So, the pesticide positively selects resistant members of the tick population, with the result that resistant members have a greater genetic fitness than the nonresistant variety and, over a few generations, will dominate the tick population.

Other control measures for cattle tick infestation are now being used, including strict controls on the movement of cattle from tick-infested regions to tick-free areas.

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Case study 2: 1080 resistance in native mammals

In the southwestern corner of Western Australia, several species of native plants produce sodium fluoroacetate, a poison known commercially as 1080 (ten-eighty). These plants include species within the genera *Gastrolobium* and *Oxylobium*(see figure 9.24). These genera are members of the pea family (Papilioaceae) and typically grow as perennial shrubs.

FIGURE 9.24 One of Western Australia's native toxic plants, sandplain poison *(Gastrolobium microcarpum)*, that produces fluoroacetate poison, a substance that can cause the death of introduced farm stock.

Populations of the common brushtail possum, *Trichosurus vulpecula*, include some living in regions where these toxic species grow. When their response to fluoroacetate is measured, these possums are found to be resistant to the poison. Other possum populations live outside the range of fluoroacetate-producing plants. Not surprisingly, when these possums are tested for resistance to fluoroacetate, they show symptoms of poisoning at low concentrations. Data on the resistance of native mammals to 1080 poison are shown in table 9.6.

The presence of fluoroacetate in the diet of marsupials living within the range of the toxic plants has **acted as an agent of selection so that, over time, resistant possums have higher survival and reproduction rates than nonresistant possums**. Because of this increased genetic fitness of resistant possums in the presence of fluoroacetate, the allele frequencies in the possum gene pool have changed over several generations so that the resistant allele has increased in frequency.

TABLE 9.6 Approximate mean LD_{50} for fluoroacetate poison for different populations living in environments either with exposure to the toxin or without known exposure. LD_{50} is a measure of toxicity: the higher the number, the more resistance to the poison.

Animal group	Prior exposure to 1080 LD_{50} (mg/kg)	No prior exposure to 1080 LD_{50} (mg/kg)
introduced herbivores: rabbit, goat	n/a	0.3
introduced carnivores: fox, cat	n/a	0.2
native marsupial herbivores: possum, wallaby	42.0	0.3
native marsupial carnivores: quoll, antechinus	8.3	2.7

Table adapted from McIlroy, J.C., 1986, *Australian Wildlife Research*, Vol. 13, p. 39.

ODD FACT

When myxomatosis virus was first released via mosquito vectors to control rabbits in 1950, the death rate for rabbits in the Corowa region of New South Wales was more than 99 per cent but by the second year had dropped to 85 per cent. Why?

KEY IDEAS

- Various phenotypes in a population may differ in their fitness value.
- The fitness value of phenotypes varies according to the environment.
- Selecting agents act on populations and change allele frequencies.

QUICK CHECK

12 A herd of wild mountain goats moves permanently to a higher region and over many generations shows an increase in average body size. If this change is a result of selection, is it natural or artificial? Explain.
13 What is meant by 'fitness' in a genetic sense?
14 In a plant population, some phenotypes survive periods of drought better than other phenotypes. What might be expected if this population was exposed to drought conditions for several generations?
15 Would complete selection have a greater effect against a dominant trait in a population or a recessive trait? Explain.

Gene flow as an agent of change

In the first half of the twentieth century, the allele for beta thalassaemia was very rare in the populations of Australian capital cities. Today, however, this allele is common. What has caused this change in allele frequency?

Gene flow, also known as **migration**, can change the allele frequencies of a population. Unlike selection, which requires several generations to have an effect, **changes due to migration can occur very quickly**.

Regional human populations have differences between their gene pools that reflect the long-term effects of natural selection in the prevailing environmental conditions, as well chance events and gene mutations. The gene pool of western Europeans has a higher frequency of the allele for cystic fibrosis compared to the gene pool of southern Europeans. The gene pool of that population has a relatively higher frequency of the allele for beta thalassaemia. The **immigration** of groups of people from one region to a new population in a different region can introduce new alleles into the gene pool of the new population or can alter its existing allele frequencies. This is what occurred in the post–World War II period when large-scale migration to Australia, including that from southern European countries such as Italy and Greece, resulted in an immediate increase in the frequency of the beta thalassaemia allele in the gene pool of the Australian population. At the same time, this mass immigration produced a decrease in the frequency of the allele for cystic fibrosis in the same gene pool.

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FIGURE 9.25 Photo showing Ron Cooke, his wife and 10 children on Southampton Dock prior to boarding a ship in early 1969 to emigrate to Australia. More than one million Britons emigrated to Australia on an assisted scheme between 1947 and 1982. Immigration or gene flow can have an immediate effect on the gene pool of a receiving population. Emigration may also have an effect on the gene pool of the source population.

ODD FACT

In the period from 1945 to 1975, the population of Australia increased from about 7.5 million to 13 million, and this increase included about 3 million migrants and refugees.

Emigration can also change allele frequencies if the emigrant group is not a representative sample of the original population. Imagine a small hypothetical population that comprises mainly blue-furred organisms, both homozygous ***BB*** and heterozygous ***Bb***, and a few homozygous ***bb*** pink-furred organisms. If a group emigrates from that population that is not representative of its existing allele frequencies, the gene pool of that population will immediately change. If, for example, all the pink-furred organisms emigrate, the frequency of the ***b*** allele will decrease. Because the pink-furred phenotype is no longer present in the population, it may appear that the ***b*** allele has disappeared. However, this allele is 'hidden' in the remaining heterozygous ***Bb*** blue-furred organisms and can reappear in the next generation as a result of the mating of two heterozygotes.

Gene flow can increase the diversity of a gene pool. If, for example, the group of pink-furred organisms emigrates to a new population that consists entirely of ***BB*** blue-furred organisms, the gene pool of their new population immediately becomes more diverse.

With the development of mass transport, human migration continues. As human groups move, their new environmental conditions may be very different from those in their place of origin. Phenotypes that were selected for in the 'home' environment may be disadvantageous to the migrants in their new setting, as seen in the migration of fair-skinned Celtic people from Ireland and Scotland to Australia.

In the northern high-latitude home environment of Celtic people, sunlight is not intense, even in summer, and, during winter, the days are short. Fair skin is favoured by natural selection because it allows the absorption of sunlight that is needed for the synthesis of essential supplies of vitamin D. In the Australian environment, with its more intense sunlight, people with fair skin are poorly protected against UV radiation. People of Celtic ancestry are at higher risk of skin cancer than people who have heavily pigmented skin.

The forcible movement of people from west Africa to the United States as part of the slave trade introduced the sickle-cell H^S allele into the American population. However, since malaria was not prevalent in the new environment, people carrying this allele were no longer at a selective advantage. Over time, the incidence of this allele in the African American population has decreased.

Australia is now a multicultural population. Each of the groups that migrated to Australia has brought with it specific alleles that are common in the home regions of each group. The high incidence of cystic fibrosis in Australia is due to the migration here of people from western Europe, where the allele that causes cystic fibrosis is common. Since World War II, migration of people from southern Europe, the Middle East, India and South-East Asia introduced the beta thalassaemia (***t***) allele into the gene pool of the Australian population. This allele gives a selective advantage to the heterozygous ***Tt*** individuals because this genotype protects against malaria. In the Australian setting, however, this heterozygote advantage no longer exists.

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Chance as an agent of change

Chance factors can cause allele frequencies in a population to change over time. In this section we will explore chance factors that impact on the genetic make-up of a population, that is, change the gene pool of a population. These chance events include:

- genetic drift
- bottleneck events
- founder effects.

Genetic drift can change gene pools

When chance operates on the allele frequencies in a population, the direction of the change is unpredictable and can vary from one generation to

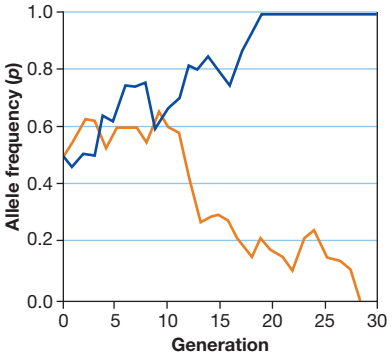

FIGURE 9.26 Changes in allele frequency in replicate populations due to chance. Is the direction predictable from one generation to the next? What has happened in population number 1 (blue line) by the nineteenth generation? Now check population number 2 (orange line). Contrast what has happened in the period from generation 0 to 7 with what happened in the period from about generation 10 to 17.

the next. The resulting pattern of random change is known as **genetic drift** (see figure 9.26). The change in allele frequency from one generation to the next in genetic drift is random, and this is in marked contrast to the directional change that occurs in natural selection.

The smaller the population size, the greater the potential impact of genetic drift. Unlike the action of natural selection, genetic drift does *not* favour one allele over another; both are equally subject to being affected by genetic drift. In a very small population, genetic drift can lead to the decrease, and eventual loss, of favourable alleles from the gene pool. For this reason, when a species is reduced to one or a few small populations, the species is at great risk of extinction. It may become extinct in spite of conservation measures.

Bottleneck effects can change gene pools

Bottleneck effects come into operation when the size of a population is drastically reduced for at least one generation. This reduction may be the result of a major disaster, such as a widespread bushfire or flood, or the introduction of a new disease to which the population has not previously been exposed. **The few survivors that reproduce to give the next generation may by chance be an unrepresentative sample of the gene pool of the original population**. This is known as the **bottleneck effect**.

An evolutionary bottleneck occurs when some disaster or disease causes a rapid decrease in the size of a large population. The original large population may have included a diverse set of alleles in its gene pool. The small post-disaster population is likely, by chance, to have a much less diverse set of alleles in its gene pool (see figure 9.27).

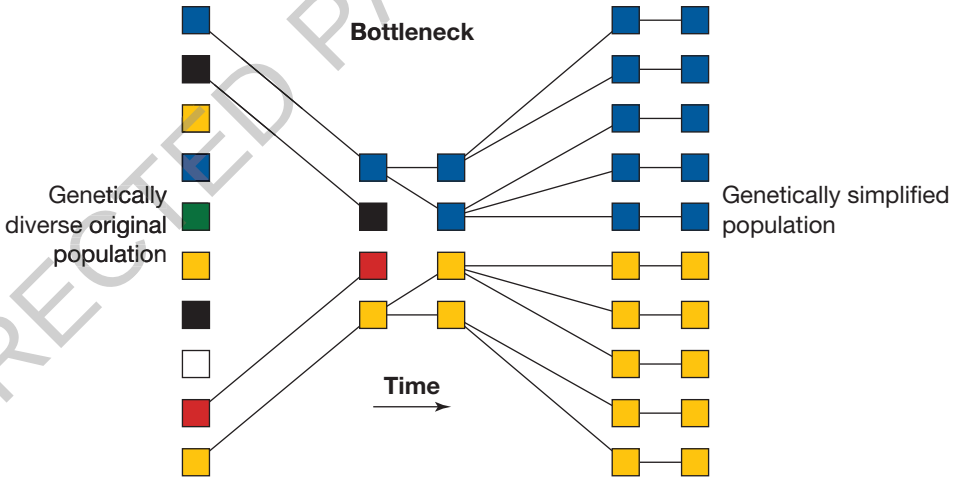

FIGURE 9.27 Diagram illustrating the concept of an evolutionary bottleneck that occurs when the population size is very sharply reduced by a disaster of some kind. Note that the survivors at the bottleneck have only some of the genetic diversity present in the original population. Note also that, by chance, only some of these survivors contribute to the gene pool of the 'recovery' population that rebuilds after the disaster.

After the disaster has ceased, the population will rebuild. However, the allele frequencies can be very different from those in the original (pre-disaster) population, and the genetic diversity that is the population's insurance policy may be greatly reduced.

Founder effects can change gene pools

A founder effect occurs when a new colony is started by a few members from a larger population. This small population size means that the gene pool of the

new colony is highly likely (i) to have reduced genetic variation and (ii) to be a non-random sample of the original population (see figure 9.28).

To understand the influence of sample size, think about netting samples of fish from a large tank that contains 50 orange fish and 50 black fish. If a sample of only four fish is netted, this sample might comprise fish of one variety only. If the sample size is larger, say 14, the sample will almost certainly include fish of both varieties and be representative of the tank population.

With a sample of 14, the chance that the fish will all be black is about one in 16 000.

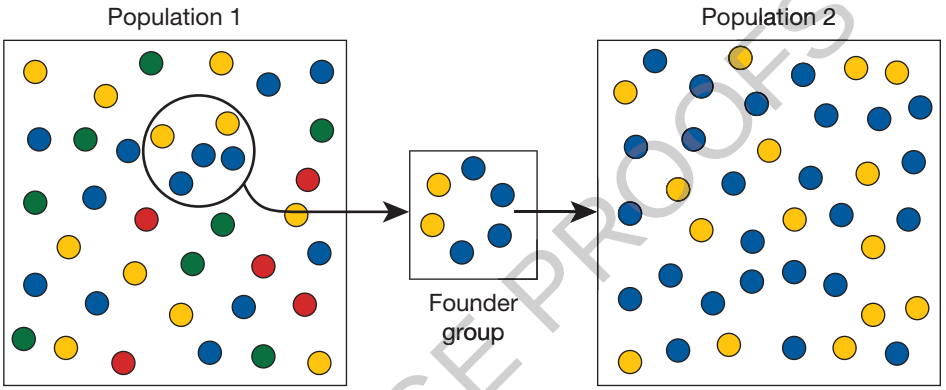

FIGURE 9.28 Diagram showing the concept of the founder effect. Population 1 is more genetically diverse than the small founder group that leaves it. In consequence, population 2, which develops over time from the founder group, is also less diverse than population 1.

FIGURE 9.29 A macaroni penguin. Is this a white-faced or a black-faced variant?

Populations of macaroni penguins (*Eudyptes chrysolophus*) live on subantarctic islands and on the Antarctic Peninsula (see figure 9.29). Most have black faces but a small proportion have white faces. On Macquarie Island, however, the population is composed almost entirely of the white-faced variety. How did this occur? It may be by chance that the small **founder population** of Macaroni penguins that first occupied Macquarie Island consisted of the white-faced variety only. So, **when a small unrepresentative or non-random sample of a population leaves to colonise a new region, this is known as the founder effect**.

Chance effects, including founder effect, can be significant in small populations, such as in zoos, where captive breeding programs must be carefully managed to avoid inbreeding and to maximise genetic diversity.

Founder effects may also be the cause of unusually high incidences of particular inherited diseases in a particular geographic area. One person comes into a region and reproduces there, introducing into the local gene pool the allele that determines a particular disease. Years later, or generations later, **the founder effect is made visible in a cluster of a particular disease in a restricted region**. One Australian example can be seen with Huntington's disease (HD).

A medical historian has shown that virtually all the families in Tasmania, and in the south-eastern states of Victoria and South Australia who are affected by HD can trace their family trees back to an English woman who migrated to Tasmania in 1842 with her husband and seven children. This woman, 'Mary' was born in 1806 in Somerset, England, and she inherited the HD allele from her father. Mary and her husband had a further seven children who were born in Tasmania. Nine of Mary's fourteen children developed HD, and all of these produced children.

A similar founder effect lies behind the high prevalence of a form of early-onset Alzheimer disease in the town of Yarumal and surrounding district in Colombia, South America. About 5000 people presently carry a particular mutation that causes this disease that typically shows its effects when people are in their early forties. This early-onset form of Alzheimer disease is a dominant trait. Researchers analysed the sequence of the DNA segment involved in the affected people and found that all these people carried the same mutation. It has been estimated that this particular mutation was introduced to the population about 375 years ago. Further, it has been concluded that this allele was brought into the district by a Spanish conquistador in the early sixteenth century. This conquistador did not remain in the district but, as a result of his sexual activities, his genetic material remained through at least one child whom he fathered with a local woman. This child survived to pass this DNA segment to the next generation. As mentioned, about 5000 people are now known to carry this DNA and will develop the destructive symptoms of early-onset Alzheimer disease.

BIOLOGIST AT WORK

Dr Kate Charlton-Robb — researching a population

I am a dolphin biologist and conservation geneticist. My work involves researching populations of dolphins in southern Australia and takes me into the field, into museums across Australia and into laboratories to process and analyse dolphin DNA. I was the principal investigator in the discovery of a new dolphin species that is endemic to southern and south-eastern Australia. I was fortunate enough to name the new dolphin, *Tursiops australis*, with the common name Burrunan dolphin, following aboriginal narrative. Only four new dolphin species have been discovered since the late 1800s. This was not only incredibly exciting but also quite a scientific journey for me.

My research began in 2003 when, as a Bachelor of Science undergraduate student at Monash University, I examined different techniques for obtaining skin samples from live dolphins and extracting and assessing their DNA. I completed my Honours year working with the dolphin population in Port Phillip Bay, Victoria. I used a common biopsy sampling technique that let me take a small piece of skin using a biopsy dart at a distance from the dolphins. This technique is not harmful to the dolphin and, in many cases, the dolphins were inquisitive about the dart floating in the water after the biopsy had been collected. The skin samples allowed me to investigate many things about the dolphins. Using their DNA signatures, previously unknown information about the dolphins can be revealed, including kinship relationships, gender, movement and breeding patterns and importantly the genetic variability and thus the genetic health of the dolphins.

During this initial project we discovered that their mitochondrial DNA differed substantially from that of the other two recognised bottlenose dolphin species, *Tursiops truncatus* (common bottlenose dolphin) and *Tursiops aduncus* (Indo-Pacific bottlenose dolphin). The discovery of unique mtDNA first highlighted that the smaller 'inshore' dolphins in Port Phillip Bay and in the Gippsland Lakes might potentially represent a new dolphin species. In 2006 I received the Dean's Postgraduate Research Scholarship to continue my research at Monash University as a part of my PhD studies. During this time I investigated numerous other DNA regions and, at each of these regions, these dolphins differed from the other bottlenose dolphin species. However, the taxonomic description of a new dolphin species requires that more than one line of evidence be presented (not just DNA), so I investigated the dolphins' skulls, colouration and external morphology (body, beak and flipper length, etc.). We then used numerous statistical tests to compare our findings with those of the other recognised bottlenose dolphin species. Our multiple lines of evidence showed that the 'inshore' dolphins were in fact different enough to represent a new dolphin species.

This work involved many years of dedicated research during which I collaborated with many fascinating researchers, from specialists in ancient DNA to taxonomists, and learned many different fields of science. Along with my collaborators, we put forward a taxonomic paper 'A new dolphin species, the Burrunan dolphin *Tursiops australis* sp. nov., endemic to southern Australian coastal waters.' This paper formally identified, described and named the new dolphin species and was scrutinised by our scientific peers and The International Commission on Zoological Nomenclature (ICZN), which is

responsible for the International Code that governs the naming of animals. We were delighted that in 2011 our paper was accepted and published in a leading scientific journal, *PLoS ONE*.

I love my work because I get to be out on the water with these amazing animals, work with museum collections dating back to the early 1900s and use applied genetic technologies. The Burrunan dolphin is now listed as 'Endangered' under Victoria's Flora and Fauna Guarantee Act, and I continue my research on the Burrunan dolphin as Head of Research at the Australian Marine Mammal Conservation Foundation. My main aim for future research is to find out where else these dolphins occur in Australia, to further assess the genetic variability, population demographics and, most importantly, to work to conserve and protect this amazing new species for future generations to enjoy.

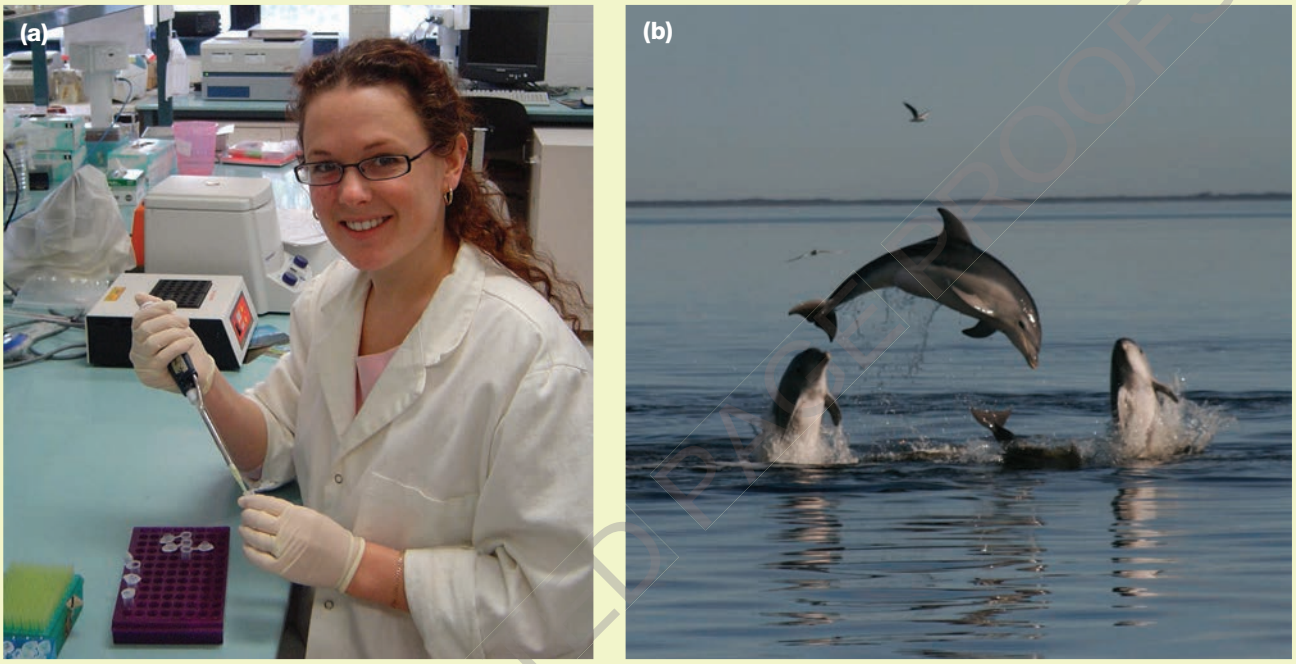

FIGURE 9.30 **(a)** Kate Charlton-Robb during her PhD at Monash University. **(b)** Burrunan dolphins (*Tursiops australis*) in Port Phillip Bay, which were recognised as a new species in 2011. (Images courtesy of Dr Kate Charlton-Robb.)

KEY IDEAS

- In addition to selection pressure, other agents of change in populations are gene flow and several chance factors.
- Gene flow can produce an immediate change in the gene pools of populations.
- Chance factors that can alter the gene pool of a population are genetic drift, bottleneck events and founder effects.
- Genetic drift effects on allele frequencies in gene pools are not directional, but are random.
- Gene flow, such as the migration of a group into an existing population, may result in an increase in the diversity of the receiving population.
- Bottleneck events typically result in a decrease in the diversity of the gene pool of the recovery population.
- Founder effects occur when a small sample of one population becomes the foundation of a new population.

QUICK CHECK

16 The frequency of a particular allele in the gene pool shows a steady decline over each successive generation. Is this change the result of genetic drift? Briefly explain.

17 Bottleneck events and founder effects are both chance factors that can act on the gene pool of a population.
- **a** Identify one way in which they are similar.
- **b** Identify a key difference between these two factors.

18 Identify the following statements as either true or false:
- **a** Genetic drift is expected to produce an increase in the gene pool of those alleles that confer greater genetic fitness.
- **b** Gene flow can produce an immediate increase in the diversity of a gene pool.
- **c** The smaller the population size, the greater the impact of genetic drift.
- **d** Bottleneck effects can occur when a population undergoes a sudden decrease in size.
- **e** A high prevalence of an inherited disease in a restricted region might be explained as being due to a founder effect.

Speciation

We have seen how the gene pools of large populations living under particular environmental conditions can change in response to selection pressures acting on the different variants present in the population. In small populations, change agents such as genetic drift can also produce changes in their gene pools.

So, populations of the same species living under different environmental conditions are likely to be subject to different selection pressures so that the allele frequencies in their gene pools diverge. In addition, any new alleles added by mutation to these gene pools are likely to be dissimilar.

The Darwin–Wallace theory of evolution by natural selection proposes that populations of the same species living in different locations under different conditions can evolve in different directions. Over many generations, under different selection pressures, these populations can become increasingly different from each other in structure, physiology and behaviour. Eventually they can become so different that they form two distinct species.

The process of formation of new species is termed **speciation**. For living organisms that are sexually reproducing organisms, species are recognised as different when they are not able to interbreed under natural conditions, or, if they interbreed, the offspring are either inviable or, if they survive, are sterile.

Several factors keep different species separated and prevent interbreeding or gene flow. These may come into operation before mating takes place — these are often called pre-mating isolation mechanisms — or they may operate after mating — these are post-mating isolating mechanisms.

Pre-mating isolation mechanisms include the following factors that are barriers to finding and securing a mate:
- Isolation in time: one species may be active by day and the other species is active at night — so, they are unlikely to meet.
- Isolation in space: one species may live on mountain tops and the second in valleys — again, they are unlikely to meet.
- Isolation by behaviour: one species does not recognise the signs of sexual readiness in a second species or is unable to perform the correct courtship rituals — so they meet, but nothing comes of it.
- Isolation by anatomy: mating may be attempted but, because of physical differences, it is not successful — so they meet, they try, but the parts don't fit.

Post-mating isolation mechanisms operate after the transfer of sperm from male to female and are effective because of chromosomal and chemical imbalances between the different species. Example include:

- Incompatibility of gametes: sperm cannot penetrate the outer coats surrounding the egg of the second species, so fertilisation does not occur.
- Zygote mortality: fertilisation occurs, but the zygote fails to develop.
- Inviability of zygote: zygote develops into an embryo but does not develop beyond that.
- Sterility of hybrid: hybrid offspring survive, but are sterile.

Subspecies

Before becoming different species, separated populations may be identified as distinct subspecies. At this stage, members of geographically separated populations are recognisably different from each other. However, if the populations come together again, members of the different subspecies groups can still interbreed and produce viable and fertile offspring.

Subspecies are denoted by the addition of a third part to the scientific name. Figure 9.31 shows some of the different subspecies of the crimson rosella (*Platycercus elegans*):

A red-headed subspecies (*Playtycercus elegans elegans*) is found mainly on the south-east coastal regions of Queensland, New South Wales and Victoria. The orange-headed subspecies (*Platycercus elegans subadelaidae*) is found mainly in the Gulf region of South Australia. The yellow-headed subspecies (*Platycercus elegans flaveolus*) is found in the Murray–Darling Basin of New South Wales, Victoria and South Australia.

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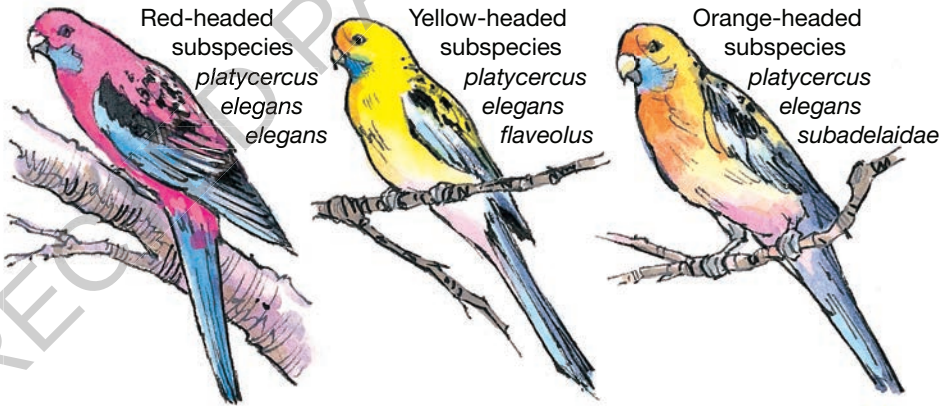
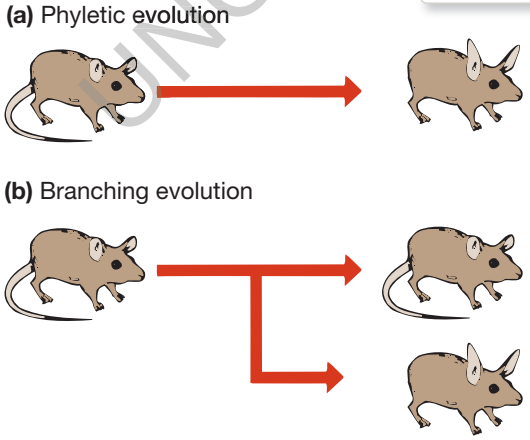


FIGURE 9.31 Three subspecies of the rosella (*Platycercus elegans*). If colour is important in attracting a mate and in pre-mating rituals, what is likely to happen to these subspecies over time?



Two patterns of speciation are possible:

- phyletic evolution in which one population progressively changes over time to become a new species (see figure 9.32a)
- branching evolution in which a population of one species splits and one part of the population evolves separately to form a new species distinct from the original species (see figure 9.32b).

FIGURE 9.32 Two different patterns of speciation: **(a)** phyletic evolution; **(b)** branching evolution. Which pattern is more common?

A pattern of branching evolution is more common than phyletic evolution. Branching evolution typically involves:

1. splitting of a smaller group from an original population so that the small group becomes geographically isolated
2. over many generations, the isolated population being subjected to different selection pressures because of different environmental conditions and to other change factors such as genetic drift
3. the isolated population changing over time such that, even if the two populations were to come together again, their members are unable to reproduce successfully — they are now two distinct gene pools and two different species (see figure 9.33).

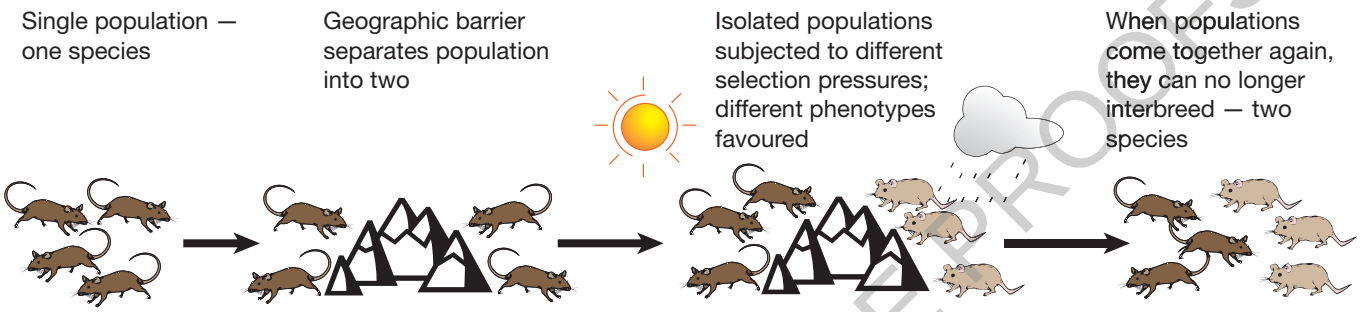


FIGURE 9.33 Natural selection can, over time, produce two different species from a single ancestral species. This most commonly occurs when a population is split into two isolated groups, a situation called allopatric speciation.

When speciation occurs under circumstances where populations become geographically separated, it is termed **allopatric speciation** (allopatric = 'different homeland'). Factors that can split a population into geographically isolated groups may be:

- quick acting, such as habitat fragmentation owing to clearing or construction
- slow acting, such as the change of a river course
- even slower geological processes, such as the uplift of mountains or rising sea levels.

Another concept is **sympatric speciation** (sympatric = 'same homeland'). This occurs when members of a population living in the same area diverge sufficiently to become two different species. It was thought for some time that speciation could not occur under sympatric conditions. However, more recent evidence indicates the cichlid fish living in Lake Nicaragua are in the process of becoming a new species.

FIGURE 9.34 Charles V: a person with Hapsburg lip. The transmission of the autosomal dominant Hapsburg lip through many generations is revealed in portraits, allowing the trait to be identified.

Mutations: source of new alleles

Generally, the DNA of the genetic material is stable, both in its base sequence and in its chromosomal location, and is passed unchanged from generation to generation. For example, members of one European family, the House of Hapsburg, have been immortalised in art over several generations. Their portraits show that many members of the family had an excessively developed lower jaw that projected in front of the face (see figure 9.34). This condition, known as prognathism (also called Hapsburg lip), can be followed from Ernest the Lion (1377–1424) through ten generations to Maria Theresa (1717–1780).

However, the DNA of the genetic material can change. This change is very commonly a change in the base sequence of a gene. When this happens, a new form of a gene, called an allele, is created. Such a change is known as a **gene mutation** and it can alter the instructions that are encoded in the DNA.

FIGURE 9.35 Laila El Garaa of Morocco is a short-statured woman who competed in the 2008 Beijing Paralympics where she won a bronze medal in the women's sports class F40 shot put event. Achondroplasia is due to the action of one allele at the **ACH** gene locus, with small stature being dominant to common stature. While aspects of their skeletal structure are affected, short-statured people (a preferred description) are otherwise unaffected and play standard roles in society.

This may be seen in the unexpected birth of a baby with achondroplasia, a form of dwarfism, to two parents of average height (see figure 9.35). This event is the visible expression of one change in the base sequence of a gene in a gamete (either egg or sperm) of one of the parents. Other gene mutations result from a change in the **location of a gene, such that the DNA moves to a new chromosomal neighbourhood and this move alters its expression**.

Earlier in this chapter, you saw examples of inherited variation in several organisms (refer back to page 383). These phenotypic variations are expressions of just a tiny sample of the different forms (alleles) of each gene that have arisen by mutation of ancestral genes.

Spread of mutations in populations

Some mutations spread locally, regionally and even globally. For example, we know that prehistoric modern humans were all lactase nonpersistent. The gene mutated, giving rise to a new allele that conferred lactase persistence and has spread where dairying cultures were established. Archaeological evidence and ancient DNA studies support the inference that in Europe this occurred about 7500 years ago.

The ancestral form of the ABO gene that determines ABO blood types is believed to be the I^A allele that determines group A blood. This allele was already present in the first modern humans, having evolved much earlier in their remote ancestors. Sometime later in human prehistory, other mutations of this gene occurred that gave rise, first to the I^B allele, and later to the i^o allele. These alleles are spread globally. Worldwide, the most common blood type is group O, with more than 60 per cent of the world's population having this type (see figure 9.36).

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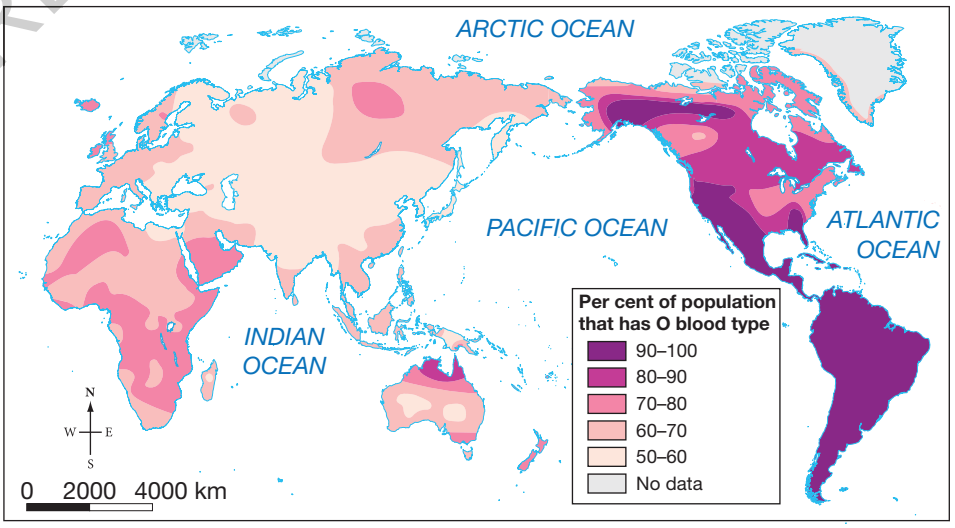

FIGURE 9.36 Distribution of blood group O in indigenous populations worldwide. Note the very high percentage in Central and South America.

Source: Anthro Palomar

Table 9.7 shows the current distribution of the ABO blood types in several populations.

ODD FACT

Karl Landsteiner was the first to identify three of the four ABO groups when, in 1901, he discovered the A, B and O blood groups. Landsteiner originally called them groups A, B and C, but later renamed group C as group O (= Ohne, German for 'zero' or 'without').

TABLE 9.7 Distribution of the four ABO blood groups in a number of human populations.

People group	O	A	B	AB
American (Caucasian)	45	41	10	4
Australian (other)	49	38	10	3
Bororo (Brazil)	100	0	0	0
British	47	42	8	3
Chinese (Beijing)	29	27	32	13
Danes	41	44	11	4
Dutch	45	43	9	3
East Indian (Bangalore)	40	24	30	6
East Indian (Banjara caste)	28	23	37	12
Indigenous Australian peoples	61	39	0	0
Japanese (Ainu)	17	32	32	18
Maori (New Zealand)	46	54	1	0
Mayan (Central America)	98	1	1	1
Navajo (American Southwest)	73	27	0	0
Pakistani (NW Frontier)	35	25	32	8
Peruvian (Indians)	100	0	0	0
Spanish	38	47	10	5
Sudanese	62	16	21	0
Thai	37	22	33	8

Data adapted from bloodbank.com, *Asian Journal of Transfusions Science, Journal of Pakistan Medical Association, The Internet Journal of Biological Anthropology,* and *A Contribution to the Physical Anthropology and Population Genetics of Sweden* (Lund: Sweden, 1959).

FIGURE 9.37 Wall painting from tomb of Nebamun in Thebes, Egypt, about 1350 BC, showing Nebamun accompanied by his wife, Hapshetsut, hunting birds in the marshes of the Nile river. Note the striped (tabby) cat near the reeds that is looking up at the birds.

Mutations in domestic pets

We can track the appearance of some mutations in the domestic cat (*Felis catus*) from historical records. For example, wall paintings and papyrus manuscripts from ancient Egypt show that the first domesticated cats of Egypt (and indeed the world) were striped (tabby) cats with their stripes arranged in the so-called mackerel tabby pattern. Figure 9.37 shows part of a wall painting, dated more than 3000 years ago, showing such a cat.

Since their domestication and spread from Egypt, many other mutations have spontaneously occurred in cat populations around the world, including:

- a blotched mutation that produces a different pattern of tabby striping that is known to have first appeared in England (see figure 9.38b)
- a non-agouti mutation that produces a solid-coloured cat, such as an all-black cat (see figure 9.38c)
- an X-linked orange mutation that produces ginger cats, both male and female, and female tortoise-shell cats
- a dilute mutation that produces grey, rather than black cats, and cream, rather than orange (ginger) cats
- a Manx mutation in which the cat has either no tail or has a greatly reduced stumpy tail (see figure 9.39a).

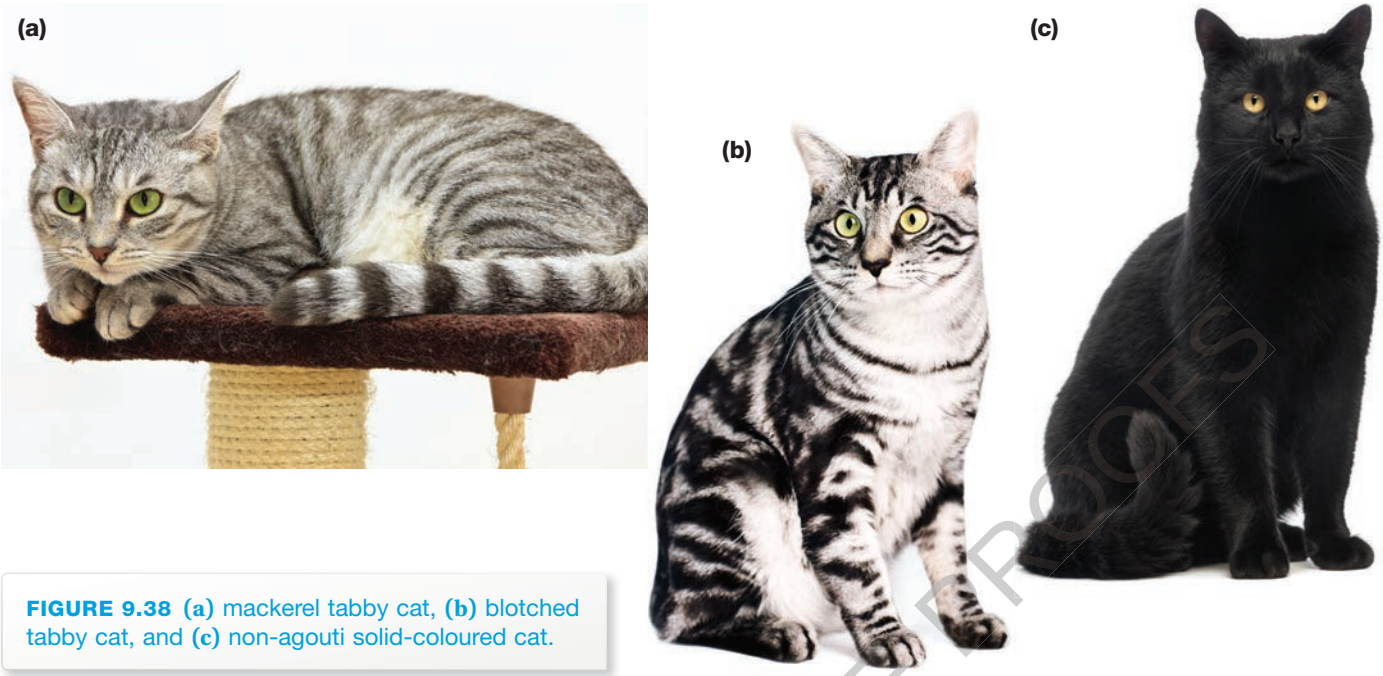

FIGURE 9.38 **(a)** mackerel tabby cat, **(b)** blotched tabby cat, and **(c)** non-agouti solid-coloured cat.

Mutations continue to occur in cats, as in other species. In 1950, in Cornwall, England, the rex mutation appeared, in which the cats' coats consisted of the soft and curly undercoat with the normal overcoat missing (see figure 9.39b). In 1966, in New York, the 'wirehair' mutation was detected in which the cats' fur is very coarse.

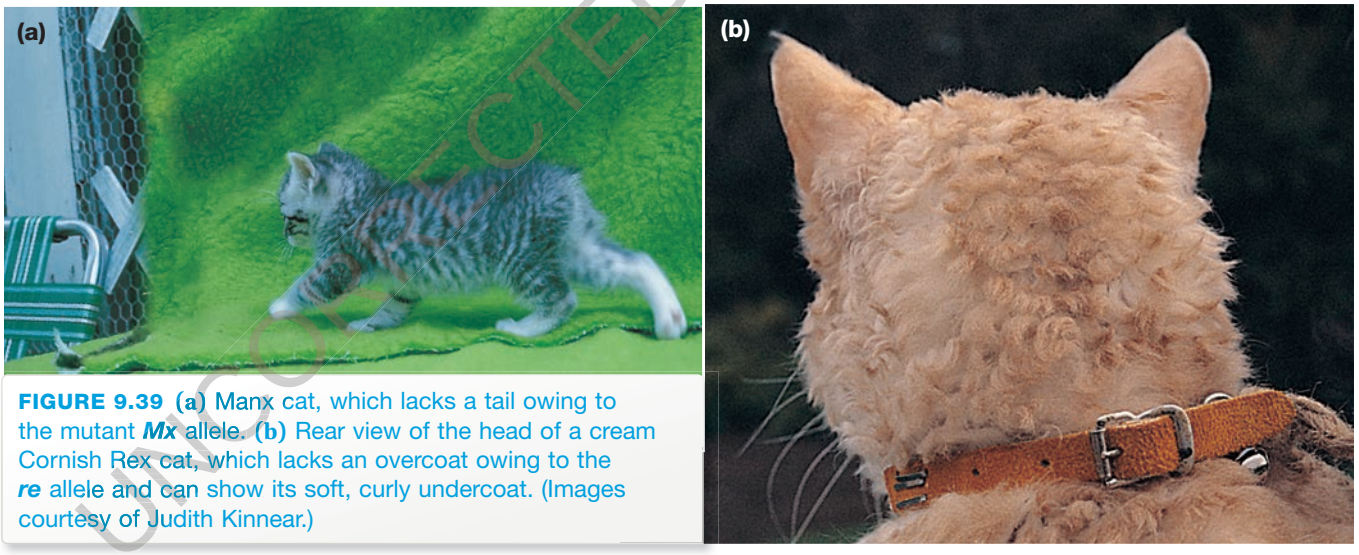

FIGURE 9.39 **(a)** Manx cat, which lacks a tail owing to the mutant ***Mx*** allele. **(b)** Rear view of the head of a cream Cornish Rex cat, which lacks an overcoat owing to the ***re*** allele and can show its soft, curly undercoat. (Images courtesy of Judith Kinnear.)

The budgerigar, *Melopsittacus undulatus*, is the world's most popular cage bird. In the wild, populations of these birds are predominantly green. However, many gene mutations have occurred in captive breeding populations of these birds (refer back to figure 9.1 on page 377).

The first budgerigars were taken from Australia to England in 1840. For about 30 years, light green was the only colour seen in these pet birds. Breeders found that, generation after generation, these birds bred true. However, in 1872, the first colour mutant appeared in birds in Belgium and Germany; this was the

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dilute mutation. Soon afterwards, in 1878, the *light-blue* mutation appeared — this caused quite a sensation — green birds now came in a blue variety, and these blue budgerigars initially commanded very high prices. Later, other colour variants appeared, including the *ino* mutation, which produces all yellow and all white colours (1879), the dark mutation (1915), the *cinnamon* mutation (1931), and the *yellow-faced blue* mutation (1934), and, to date, about 30 different colour variants have appeared in captive budgerigar populations as a result of gene mutations.

The continued appearance of mutations reflects the fact that DNA is a large macromolecule and that errors can occur during processes in meiosis, including DNA replication and synapsis and crossing over.

Fate of new mutations

In order for a gene mutation to be sustained in a population, it is first necessary that the mutation occurs in the germline of an organism so that it may be passed to the next generation. Somatic mutations, that is, mutations in body cells of an organism, disappear with the death of the organism in which the mutation occurred.

If the germline mutation produces an allele that controls a dominant trait, its phenotypic expression will appear in the next generation. If, however, the new allele controls a recessive trait, it may be several generations before the trait is expressed phenotypically in the population. Why? The first appearance requires the mating of two heterozygous carriers of the trait, and if the allele is rare, as would be the case for a new allele, such matings would also be rare.

After a mutation is expressed in a phenotype, it will be the actions of change agents, in particular natural selection, that determine whether the allele is lost from the population or whether it becomes established in the gene pool of a population. Some gene mutations disappear after one or a few generations, because of strong selection pressure acting against them. In addition, in a small population, even advantageous mutations may be lost through the action of genetic drift.

If a new allele created by mutation confers a strong selective advantage in terms of survival and reproduction rate under the prevailing environmental conditions, it is reasonable to assume that this allele has a high probability of becoming established in a population and increasing its frequency in the gene pool over successive generations. This occurred in the case of the lactase persistence allele of the **LCT** gene in populations with a dairying culture, and with the H^S mutant haemoglobin allele in environments where malaria is a major killer.

Does a very high frequency of a particular allele in the gene pool of a population always indicate that the corresponding phenotype has a selective advantage as compared to other variants in a population? Not necessarily. This could be a founder effect. For example, indigenous populations in South America are 100 per cent blood group O. It has been suggested that the founder groups of these populations might all have been group O, genotype $i^o\ i^o$.

ODD FACT

In the 1920s, flies were taken up in a balloon to a height of more than 18 kilometres and it was found that gene mutation in these flies occurred greater than five times more rapidly at this height than at sea level. Can you suggest a possible explanation?

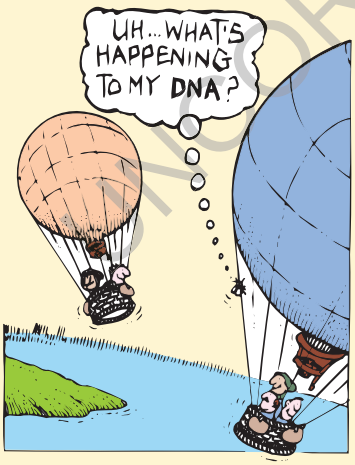

FIGURE 9.40

Causes of mutations

Mutations may be spontaneous or may be induced by known exposure to **mutagenic agents**, such as irradiation (for example, X-rays and gamma rays), some chemicals (for example, benzene, dioxane and mustard gas) and some viruses.

Protection against mutagenic agents is necessary to protect the DNA in both body cells and germline cells. Have you had an X-ray during a dental

check-up? The use of a lead apron during this procedure is to protect body tissues from exposure to radiation. Doses received by people working with radiation are monitored by wearing badges that contain radiation-sensitive devices (see figure 9.41).

FIGURE 9.41 Nuclear decontamination workers removing their radiation protection. Note the protective breathing apparatus and the radiation-sensitive dosimeter that can be seen on the chest one of the workers.

Mutation rates are not equal for all genes and the spontaneous mutation rate for achondroplasia, one form of inherited dwarfism, is much higher than that for some other inherited disorders (see table 9.8). This is in accord with the fact that about 90 per cent of the cases of achondroplasia are sporadic, and are due to mutation. On average, more sporadic cases occur in association with increased paternal age at the time of conception.

TABLE 9.8 Spontaneous mutation rates at different gene loci in various human population samples. Estimated mutation rates are based on frequencies of births of babies showing these dominant conditions, to unaffected parents. Note the high rate of mutation at the achondroplasia gene locus.

Mutation	Locus	Mutations per 100 000 gametes
retinoblastoma	number-13 chromosome	
United Kingdom		1.2
United States		1.8
achondroplasia	number-4 chromosome	
Northern Ireland		14.3
Japan		12.2
Huntington's disease	number-4 chromosome	
United States		0.5

Autosomal dominant conditions that have a serious deleterious effect on an individual are usually the result of a new mutation. Can you suggest why?

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Gene mutations
Summary screen and practice questions

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Do more
Mutations

Types of mutations

In this section, we will examine mutations, both gene mutations and chromosomal (block) mutations.

Gene mutations

Gene mutations are changes in the genetic material or DNA. Several kinds of change in DNA can be identified, such as:
point mutations that include

- base substitution
  - 'silent' mutation
  - nonsense
  - missense
    - conservative
    - non-conservative.
- base insertion
  - frameshift mutation.
- base deletion
  - frameshift mutation.

Point mutations

Point mutations affect a single base in DNA, either by substitution, insertion or deletion. Figure 9.42, on page 418, shows some of the possible point mutations in the coding sequence of a gene.

These mutations commonly affect the base sequence of a structural gene. However, these mutations may also affect a segment of DNA that regulates the expression of a structural gene. This was the case for the lactase persistence allele. We now know that the mutation that produced the lactase persistent phenotype is a single base substitution in the region upstream of the **LCT** gene. This base substitution enhances the promoter of the **LCT** gene.

Point mutations: base substitutions

In a substitution, one base in a DNA triplet is replaced by another. Three kinds of base substitutions are recognised: silent, nonsense and missense, as follows:

1. Silent mutations: A silent mutation is a base substitution in DNA that does not result in a change in the polypeptide product encoded by this gene. This is because the genetic code is redundant and many amino acids are encoded by several DNA triplets. For example, the amino acid valine (*val*) is encoded by four DNA triplets: CAA, CAG, CAT and CAC

...ATG CAA GGT... → ...ATG CAT GGT...

This DNA substitution mutation will not affect the expression of the gene because the mutated DNA triplet (CAT) still codes for the same amino acid (*val*) as the original DNA triplet (CAA), hence the label 'silent'. Silent mutations most commonly occur in the third base of a DNA triplet.

While these mutations are 'silent' in terms of their polypeptide product, they are of course visible when the DNA sequence is examined.

2. Nonsense mutations: A nonsense mutation in DNA is a base substitution that changes a DNA triplet so that, instead of coding for an amino acid, it codes for a STOP signal. The stop or termination signals in DNA are the triplets ATT and ATC. Look at the following substitution mutation:

...ATG TTT GGT... → ...ATG ATT GGT...

Such a mutation will have a serious effect on the polypeptide product of the gene because transcription will stop at the mRNA codon transcribed by the mutated triplet DNA sequence. In turn, this will shorten the polypeptide

chain encoded by the gene. The degree of shortening of the polypeptide gene product will depend on the position of the point mutation relative to the start of the coding sequence — the earlier in the DNA sequence, the greater the shortening of the polypeptide.

3. Missense mutations: A missense mutation is a base substitution in DNA that results in a single amino acid alteration in the polypeptide product of the gene. Missense mutations may be conservative or non-conservative.

When a missense mutation occurs and a different amino acid is incorporated into a polypeptide chain, the outcome depends on whether the mutated polypeptide can carry out its normal function. This is likely to be the case in a **conservative missense mutation** and such a mutation can be tolerated. A **conservative missense** mutation of DNA results in the substitution of one amino acid by a different amino acid with similar chemical properties, for example:

. . . ATG CAA GGT . . . → . . . ATG CGA GGT . . .

This mutation results in the replacement of the amino acid valine (*val*) with alanine (*ala*), both amino acids having hydrophobic side chains.

In other cases, the missense mutation results in the substitution of an amino acid by one that has different chemical properties. Such a mutation is said to be a **non-conservative missense mutation.** In this case, the resulting polypeptide cannot carry out its normal function and such a mutation will have severe clinical effects.

A non-conservative missense mutation of DNA results in the substitution of an amino acid with an amino acid containing a different class of side chain and different chemical properties. For example, the replacement of histidine (*his*), which contains a charged side chain, with tyrosine (*tyr*), which contains a hydrophobic chain:

. . . ATG GTA GGT . . . → . . . ATT ATA GGT . . .

One example of a non-conservative missense mutation is haemoglobin M disease. Haemoglobin M results from a missense mutation that results in the substitution of histidine with tyrosine in the alpha chain of haemoglobin. Haemoglobin M can carry oxygen but cannot release it to body cells where it is needed. Another example of a missense mutation results in achondroplasia (refer back to pages 411 and 415). This condition is the result of the replacement of the amino acid glycine (*gly*) with arginine (*arg*) at amino acid number 380 in the receptor protein for a fibroblast growth factor. We can write this replacement as Gly380Arg. This replacement is caused by a single base substitution in the DNA triplet that encodes the 380th amino acid in this protein that mutates as **CCA → GCA**. Just one specific base substitution among hundreds of bases in a segment of DNA has this major effect.

ODD FACT

Changes in one of the first two bases of a DNA triplet are more often a source of nonsense and missense mutations than changes in the third base.

Point mutations: Frameshift mutations

Single base insertions have a major effect on the genes involved because they alter the DNA triplet at the point of the insertion and also affect every DNA triplet following that point. Likewise, single base deletions have the same effects. These single base insertions or deletions are known as **frameshift** mutations.

Frameshift mutations alter the base sequence of a gene so that the message it encodes no longer makes sense. Remember that the genetic information in DNA is organised into groups of three bases or triplets that are then read in order, with one triplet coding for one amino acid.

Consider the sentence: **the boy hit the dog** with each word representing a DNA triplet. In this form, this sentence makes sense.

Now, add an extra letter — this is like a base insertion: **the bxo yhi tth edo g**

The addition of the x causes the sense to be lost at the point of the addition and beyond.

Now delete the first e from the original sentence — this is like a base deletion: **thb oyh itt hed og**

An insertion occurs when one nucleotide base is added to a DNA strand. This type of mutation is more damaging than a point mutation because they cause a frameshift with all the bases from the point of insertion being moved down by one position to make room for the extra base.

A base deletion also causes a frameshift with all bases from the point of the deletion being moved back by one position to compensate for the deletion. In addition, both insertion and deletion may form a stop codon where one should not exist, having drastic consequences by creating a premature cessation of translation of a polypeptide chain. Figure 9.42 shows both of these types of frameshift mutations.

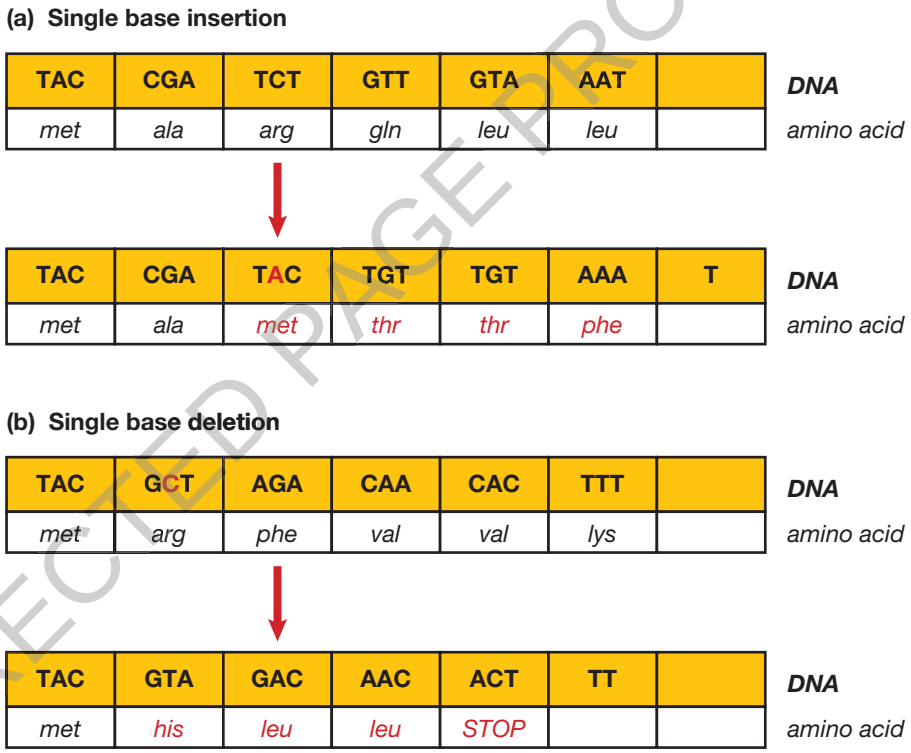

FIGURE 9.42 Diagram showing the effects of **(a)** a base addition or insertion into a DNA sequence and **(b)** a base deletion. Both mutations cause a significant change in the polypeptide that is encoded by this DNA by changing the amino acids from the point of addition or deletion and beyond as a result of the changes in the DNA triplets.

The point mutation discussed above involves single base changes. However, some mutations, known as trinucleotide repeats involve mutations that repeat a large number of bases. Go to the box at the end of this section to read about these mutations, which are the cause of two major inherited disorders, fragile X syndrome and Huntington's disease (HD).

Chromosomal or block mutations

Block mutations are chromosomal changes affecting large segments of a chromosome. These block mutations most commonly arise as a result of spontaneous errors in crossing over during meiosis or they may be induced by mutagenic agents, such as X-rays.

These block mutations include:

- deletion of part of a chromosome
- duplication or gain of part of a chromosome
- translocation or reciprocal exchange between non-homologous chromosomes
- inversion when a segment of a chromosome rotates through 180 degrees.

Figure 9.43 shows some of these block mutations. In each case, chromosome breakages occur followed by rearrangement and rejoining.

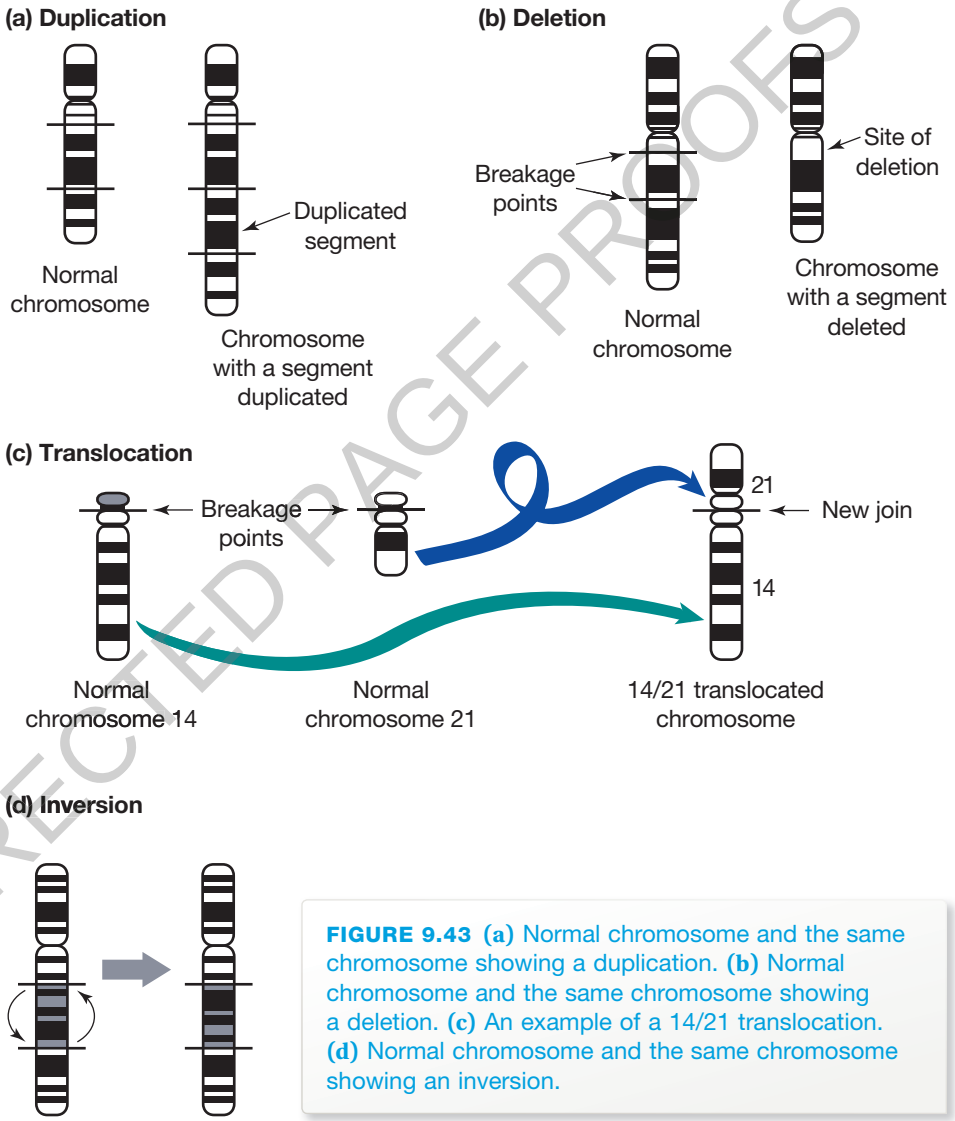

FIGURE 9.43 **(a)** Normal chromosome and the same chromosome showing a duplication. **(b)** Normal chromosome and the same chromosome showing a deletion. **(c)** An example of a 14/21 translocation. **(d)** Normal chromosome and the same chromosome showing an inversion.

None of these chromosomal block mutations involves a loss of genetic material, but they do alter the location of some genes, relocating them in a new position on a chromosome, giving them different next-door neighbours. Research has shown that relocating can alter the activity or the expression of a gene — such effects are termed **position effects**. In some cases, a gene at a breakpoint may be silenced or its expression may become unstable or, in the case of some reciprocal translocations, abnormalities such as cancers occur. Figure 9.44 shows the details of a reciprocal translocation that results in chronic myeloid leukaemia.

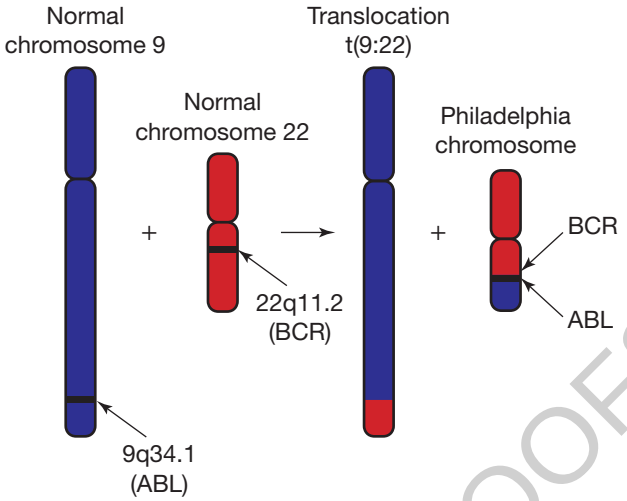

FIGURE 9.44 Diagram showing the reciprocal exchange that occurs between chromosome 9 and chromosome 22 in chronic myeloid leukaemia. The relocation of the **ABL** and the **BCR** genes in close proximity on the Philadelphia chromosome has been shown to be the cause of chronic myeloid leukaemia.

Figure 9.45 shows the unstable expression of a gene for eye colour in the compound eye of a fruit fly (*Drosophila melanogaster*) after relocation of the gene involved to a new chromosomal location by an inversion. Note that some eye facets are dark where the gene is active but others are pale where the gene is not expressed.

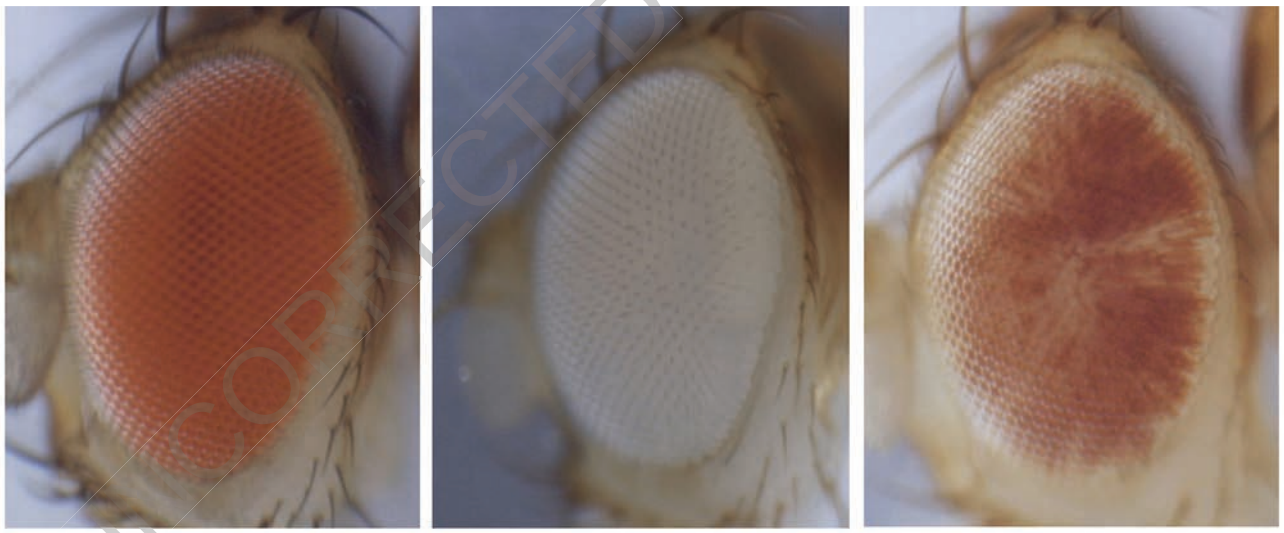

FIGURE 9.45 Images of the compound eye of the fruit fly *Drosophila melanogaster*, showing the normal red eye (left), the normal white eye (middle) and the variegated eye (at right). The variegation results from the unstable expression of the gene determining red eye colour because of a chromosomal inversion that has relocated it. (Images courtesy of A.M. Bauer and S.C.R. Elgin, Washington University, St Louis.)

Earlier in this chapter (refer back to page 389), you were introduced to chromosomal changes involving whole chromosomes (aneuploidy) or the gain of whole sets of chromosomes (polyploidy). Refer back to page 386.

Table 9.9 summarises some of these aneuploid changes in humans. Note that polyploidy does not occur in humans.

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Trisomy 21

TABLE 9.9 Some examples of chromosome changes and approximate incidence rates. Which syndrome is an example of a trisomy? A monosomy? The XYY condition does not have a clinical name.

Chromosome change	Resulting syndrome	Approximate incidence rate
Addition: whole chromosome		
extra number-21 (47, +21)	Down syndrome	1/700 live births
extra number-18 (47, +18)	Edwards syndrome	1/3000 live births
extra number-13 (47, +13)	Patau syndrome	1/5000 live births
extra sex chromosome (47, XXY)	Klinefelter syndrome	1/1000 male births
extra Y chromosome (47, XYY)	n/a	1/1000 male births
Deletion: whole chromosome		
missing sex chromosome (46, XO)	Turner syndrome	1/5000 female births
Deletion: part chromosome		
missing part of number-4	Wolf-Hirschhorn syndrome	1/50 000 live births
missing part of number-5	cri-du-chat syndrome	1/25 000 live births

TRINUCLEOTIDE REPEAT EXPANSION MUTATIONS

One class of gene mutations involves the addition or deletion of a large number of short sequences of bases.

Several normal human genes contain multiple copies of a three-base (trinucleotide) sequence, such as **CCG** and **CAG**. One kind of mutation, known as a **trinucleotide repeat expansion (TRE)**, involves additional repeats of these sequences beyond the normal range. These TRE mutations are the cause of two inherited disorders: fragile X syndrome (FMR1) and Huntington's disease (HD) (see table 9.10).

TABLE 9.10 Examples of trinucleotide repeat expansion mutations

Gene	Mutant condition	Trinucleotide repeats
FMR1	fragile-X syndrome	CCG: 200 to 1000+ repeats
HD	Huntington's disease	CAG: 36 to 120 repeats

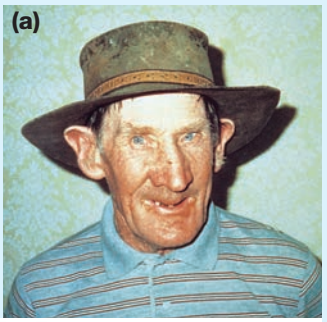
Compared with the normal alleles that have a small number of trinucleotide repeats, the mutant alleles that determine these disorders have many more repeats. This can result in a significant increase in the total number of bases compared to the number in the normal allele.

Fragile X syndrome is the most common inherited form of mental retardation in males and occurs at a frequency of about 1 in 1250 male births. Affected males often have long faces, large ears and, after puberty, may have enlarged testes (macro-orchidism). In many cases, this mutation can be visibly seen when the X chromosome is examined using light microscopy.

In 1991, it was recognised that males affected by fragile X syndrome (see figure 9.46) have from 200 to 1000 plus repeats of a CCG trinucleotide within their mutant **FMR1** allele. In contrast, an unaffected person has from 6 to about 50 repeats of the CCG trinuceotide.

Both males and females affected by Huntington's disease have from 36 to over 100 repeats of the CAG trinucleotide in their mutant **HD** allele, while unaffected people usually have from 6 to about 35 copies of this trinucleotide.

Trinucleotide repeat expansion mutations tend to be unstable so that the number of repeats can change from one generation to the next. For example, the number of trinucleotide repeats has been observed to increase when the mutant **HD** allele is transmitted from an affected male parent to his children.



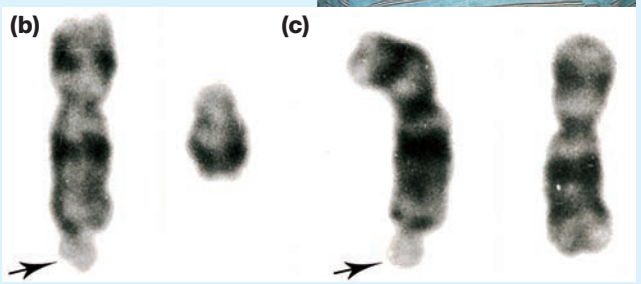


FIGURE 9.46 **(a)** A male with fragile X syndrome. **(b)** Fragile X (left) and Y chromosomes from an affected male. **(c)** A fragile X (left) and normal X chromosome from a female who is a heterozygous carrier of the fragile X. (Images courtesy of Marjory Martin.)

KEY IDEAS

- Mutations are the source of new allelic forms of genes.
- Mutations continue to occur in the gene pool of populations.
- Mutations may be either spontaneous or induced by exposure to a known mutagenic agent.
- Spontaneous mutation rates are not identical for all genes.
- Mutations may be classified as point mutations or chromosomal (block) mutations.
- Relocation of genes can produce changes in their expression.

QUICK CHECK

19 Give two examples of:
- **a** a point mutation
- **b** a block mutation.

20 Identify the following statements as either true or false:
- **a** Gene mutation can occur in the DNA of structural genes only.
- **b** The relocation of a gene may cause its expression to be changed.
- **c** Chronic myeloid leukaemia is caused by a reciprocal translocation.
- **d** A frameshift mutation would be expected to have more significant effects on a gene product than a missense mutation.

21 Identify the difference between the members of the following pairs:
- **a** a gene mutation and a block mutation
- **b** a frameshift mutation and a substitution mutation
- **c** a chromosomal deletion and a chromosomal translocation
- **d** an induced and a spontaneous mutation.

Manipulating gene pools

Lambs in spring

Early spring is the lambing season in Australia. In certain sheep breeds, such as merino, a female sheep or ewe typically produces just a single lamb each season. However, these days, it is not uncommon to see merino ewes with twins, sometimes triplets and, in some cases, even quads. What has caused this change? It is human intervention through artificial selection of an allele for increased fecundity by selective breeding with rams and ewes that have that particular allele.

The gene concerned is known as the Booroola gene, formally designated **BMPR-1B**. Its action in producing increased fecundity was recognised by the Seears brothers in the merino flock at their 'Booroola' property, near Cooma, New South Wales in the 1950s. The Booroola gene is located on the number-6 sheep chromosome and its rare B^B allele increases ovulation rate while its more common B^+ allele has no effect on ovulation rate. In heterozygous B^BB^+ ewes, the ovulation rate is increased by a factor of 1.5, while in homozygous B^BB^B ewes, their ovulation rate is increased by a factor of 3.0. (The ovulation rate in homozygous B^+B^+ ewes is 1.0.)

Using selective breeding techniques, the B^B allele has now been incorporated into other sheep breeds that normally produce a single lamb from each mating. This was done by crossing these breeds with sheep known to be homozygous for the B^B allele. The process in which specific animals are used in breeding programs to introduce particular alleles of a gene into the gene pool of a population and to fix the preferred alleles in the homozygous state in members of the population is termed **selective breeding**.

The Booroola gene is formally known as **BMPR-1B** *and its alleles are denoted as* $FecB^B$ *and* $FecB^+$ *but, for simplicity, they are shown here as* B^B *and* B^+.

ODD FACT

The Booroola gene appears to have been introduced by Bengal sheep imported to Australia from India in 1792. Records show that the Booroola merino flock can be traced back to the merinos owned by Rev. Samuel Marsden (1764–1838), who also owned a Bengal flock.

FIGURE 9.47 Sheep with four lambs. The high ovulation rate of this ewe may be assumed to be due to the presence of a double dose of the B^B allele of the Booroola gene.

Selective breeding is an example of artificial selection. In this procedure, only those animals that display a particular trait in their phenotype or are known carriers of the trait are chosen to reproduce. Selective breeding is an example of how humans can intervene in natural evolutionary processes by artificially manipulating the gene pool of a population. Let's look at some other examples of selective breeding.

Selective breeding in action

From early times, artificial selection was carried out to improve herd quality. Farmers selected the best males for mating with their breeding females. In the case of beef cattle, bulls were chosen for their genetic superiority in terms of desirable market characteristics, such as meat yield and non-fatty carcass. For dairy cattle, desirable market characteristics included milk yield and butterfat content. Merino rams were chosen for the fineness (thinness) of their wool fibre and the yield of greasy fleece. Males were also selected based on other inherited features, including good conformation (form, outline or shape), high fertility based on sperm counts and the absence of any known genetic defects. Through artificial selection, farmers could improve the quality of their herds.

The deliberate selection by a breeder of specific animals to provide the genetic material for the next generation is a process known as selective breeding or artificial selection. This is in contrast to the random breeding that occurs when any male animal in a population has an equal chance of mating with any female.

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Selective breeding in action: sheep

More than 200 recognised breeds of sheep exist, derived from wild sheep or mouflons (*Ovis musimon*) that were domesticated at least 6000 years ago. Mouflon sheep are native to parts of Europe and central Asia and are still living today.

About 75 per cent of the sheep in Australia today are merino sheep, which are prized for their wool quality. The first merinos were imported to Australia by John Macarthur in 1796 — these were Spanish merinos from South Africa. In Australia, several strains (breeds) of merino are recognised, including the

Peppin merino, the South Australian merino and the Saxon merino. Selective breeding (artificial selection) over generations has enhanced particular features of these breeds and their survival in particular climatic conditions. For example, the large South Australian merino, which has been bred to thrive in the arid regions of that state and in similar regions in others, produces greasy wool with strong (thick) fibres. In contrast, the much smaller Saxon merino thrives in wetter regions and has been bred for wool of increasing fineness (thin fibres).

Table 9.11 shows some of the different wool characteristics of several sheep breeds, including some merino breeds.

TABLE 9.11 Wool characteristics of some sheep breeds. A staple is a collection of a large number of wool fibres. Typically, the fleece grows to a fixed length (like the coats of cats and dogs). In contrast, the fleece of Drysdale sheep continues to grow (like human hair) and so the staple length is given as growth per year. Which breed produces the finest (thinnest) wool?

	Breed of sheep					
Trait	Drysdale	Saxon merino	Peppin merino	Strong Wool merino	Polwarth	Corriedale
body mass (kg)	116	60	132	147	130	104
greasy fleece weight (kg)	12.0	5.0	18.6	20.0	12.0	13.0
diameter of fibre (μm)	40.0	17.4	22.0	23.5	24.6	28.0
length of staple (mm)	400 (annual)	75	150	95	150	175

Data from the National Wool Museum (Geelong, Victoria).

The value of a wool fleece is determined principally by two traits, namely, clean fleece weight — the heavier the better — and fibre diameter — the thinner the better! These are inherited traits but, because they are polygenic, the rate of change that can be achieved through selective breeding over many generations is slower than with monogenic traits. (Can you suggest why?) For example, selection programs at the Triangle Research Station in New South Wales using several merino strains produced gains in these two important traits. The results of selective breeding, with an equal emphasis on increasing clean fleece weight and decreasing fibre diameter, are shown in table 9.12. Although the gains may appear small, they represent significantly increased profits for woolgrowers.

ODD FACT

The first merino sheep brought to the Australian colony in 1796 had fleeces that yielded about 1.5 to 2.0 kilograms of wool per year. Today, fleeces of merino sheep can commonly yield up to 10 kilograms of wool per sheep per year.

TABLE 9.12 Results of a 10-year selective breeding program on two strains of merino sheep with an equal emphasis on increasing clean fleece weight and decreasing fibre diameter.

Merino strain	Group	Clean fleece weight (kg)	Fibre diameter (microns)
Fine wool merino	Experimental	4.0	18.3
	Control	3.5	19.7
Medium wool merino	Experimental	5.0	19.3
	Control	4.7	20.8

Data from NSW DPI Prime Facts 579, March 2007.

Several sheep breeds have been developed in Australia, including the Poll Dorset, a meat producer, and the White Suffolk, a dual-purpose wool and meat producer. Suffolk sheep have black faces and legs (see figure 9.48a). This breed produces superior, fast-growing lean lambs but growers were not receiving an appropriate market price for these lambs because their wool included some dark fibres. By selective breeding over many generations involving crosses of black-faced Suffolks and white-faced Poll Dorsets, a new breed, the White Suffolk, was produced in Australia. This new breed has all the features of the Suffolk breed except for the black face and legs (see figure 9.48b).

FIGURE 9.48 Sheep breeds **(a)** Suffolk sheep with their black faces and legs. **(b)** White Suffolk, which has the features of the Suffolk except for the black face and legs. The White Suffolk was derived from a program of selective breeding and artificial selection, and was developed for a range of Australian pastoral conditions.

Selective breeding: Australian cattle

The many different breeds of cattle are believed to have been derived by artificial selection from a wild species known as the auroch (*Bos taurus*), which was domesticated at least 6500 years ago.

The original cattle brought to Australia were British breeds that were suited to the temperate regions but did not thrive in tropical Australia. Selective breeding has produced a number of breeds for the tropics. These new breeds include the Australian Braford, which was developed in Queensland in the period from 1946 to 1952. The Australian Braford breed combines features of the Brahman, such as the hump, loose skin, short coat and heat and tick resistance, with features of the Hereford, including its colour markings (see figure 9.49). Another cattle breed developed in Australia is the Australian Milking Zebu, which resulted from selected crosses between Asian dairy cattle and European dairy cattle. This breed is a reliable milk producer in tropical regions, it is heat resistant because it is able to sweat and its loose skin makes it resistant to ticks, which it can easily shed.

ODD FACT

Aurochs are now extinct. The last member of this species was killed by a Polish poacher in 1627.

FIGURE 9.49 Australian Braford cattle. Note the folds of loose skin. (Image courtesy of the Australian Braford Society.)

Selective breeding by fanciers

Selective breeding of animals is not restricted to agricultural stock, such as cattle and sheep. Selective breeding has been used by animal fanciers and hobbyists to produce the great variety of breeds that may be seen in various pets, such as dogs, and in other domestic animals, such as poultry. This selective breeding continues today.

Dogs: The Australian National Kennel Council recognises 200 different breeds of dog, from Affenpinschers to Yorkshire Terriers. Figure 9.50 shows three dog breeds that show striking variation in terms of their coats — from almost zero hair in the Chinese Crested Dog to the Puli with its profuse corded coat.

FIGURE 9.50 Selective breeding of dogs has produced a remarkable range of variation seen in the diversity of breeds recognised by **(a)** the Chinese crested dog, which is almost hairless with only fine tufts of hair on its head, **(b)** shih tzu, a toy-sized variety with long fine hair, and **(c)** puli, with its profuse corded coat hair. Note other structural differences, such as muzzle length. (Images courtesy of Judith Kinnear.)

Poultry: Frizzle, Indian game, Houdan, Cochin, Hamburgh, Australorp, Silkie, Andalusian, Dorking, Scots dumpy . . . what are they? These are all breed or varieties of the domestic fowl (*Gallus domestica*). The domestic fowl is believed to have descended from the wild red jungle fowl (*Gallus gallus*), which is native to a region of south Asia extending from India to Indonesia.

Over time, many mutations have arisen in the domestic fowl that have produced features that are not present in their wild type ancestor, the jungle fowl (see figure 9.51a), for example:

- Most domestic fowls have legs covered with scales, but some breeds, such as the Cochin, have feathered legs (see figure 9.51b).
- Most domestic fowls have four toes, but some breeds, such as the Dorking, have five toes (see figure 9.51c).
- Dark lacing on the edges of feathers can be seen in several fowl breeds (see figure 9.51d) in contrast to solid-coloured feathers in other breeds.
- Some fowl breeds have crests on their heads (see figure 9.51d) unlike most other breeds.

These mutations have been preserved in fowls by fanciers through their selective breeding practices.

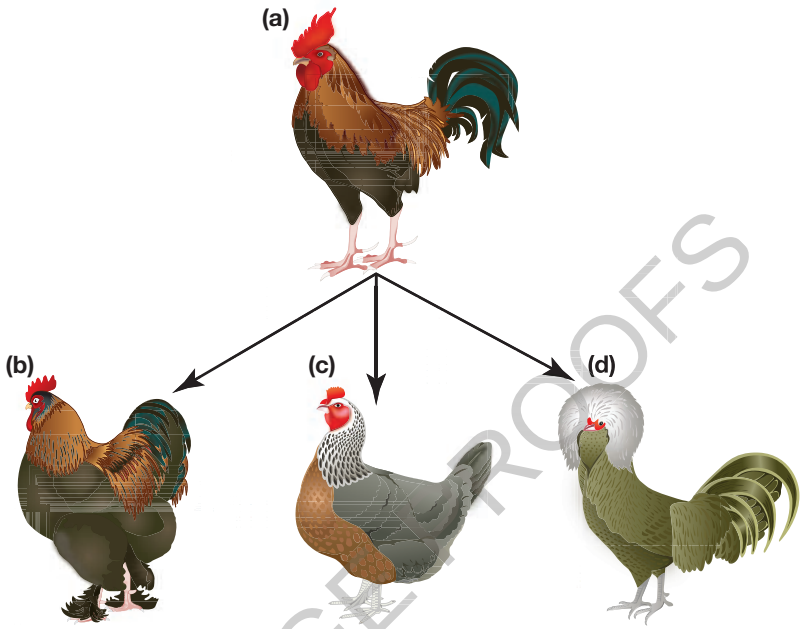


FIGURE 9.51 **(a)** Red jungle fowl, the presumed ancestor of the domestic fowl. **(b)** Feathered legs in the Cochin breed of domestic fowl. **(c)** The Dorking breed with its five-toed feet. **(d)** The Polish breed of domestic fowl with crest and dark lacing on the feather edges. (Images (b), (c) and (d) courtesy of Judith Kinnear.)

The jungle fowl has a simple comb that is also seen in many domestic fowls, including the Australorp and the Leghorn. However, other comb shapes have arisen by mutation and have been retained through selective breeding. These different comb shapes include the V-shaped comb of the Crevecoeur, the rose comb of the Dominique, the walnut comb of the Silkie and the pea comb of the Brahma (see figure 9.52).

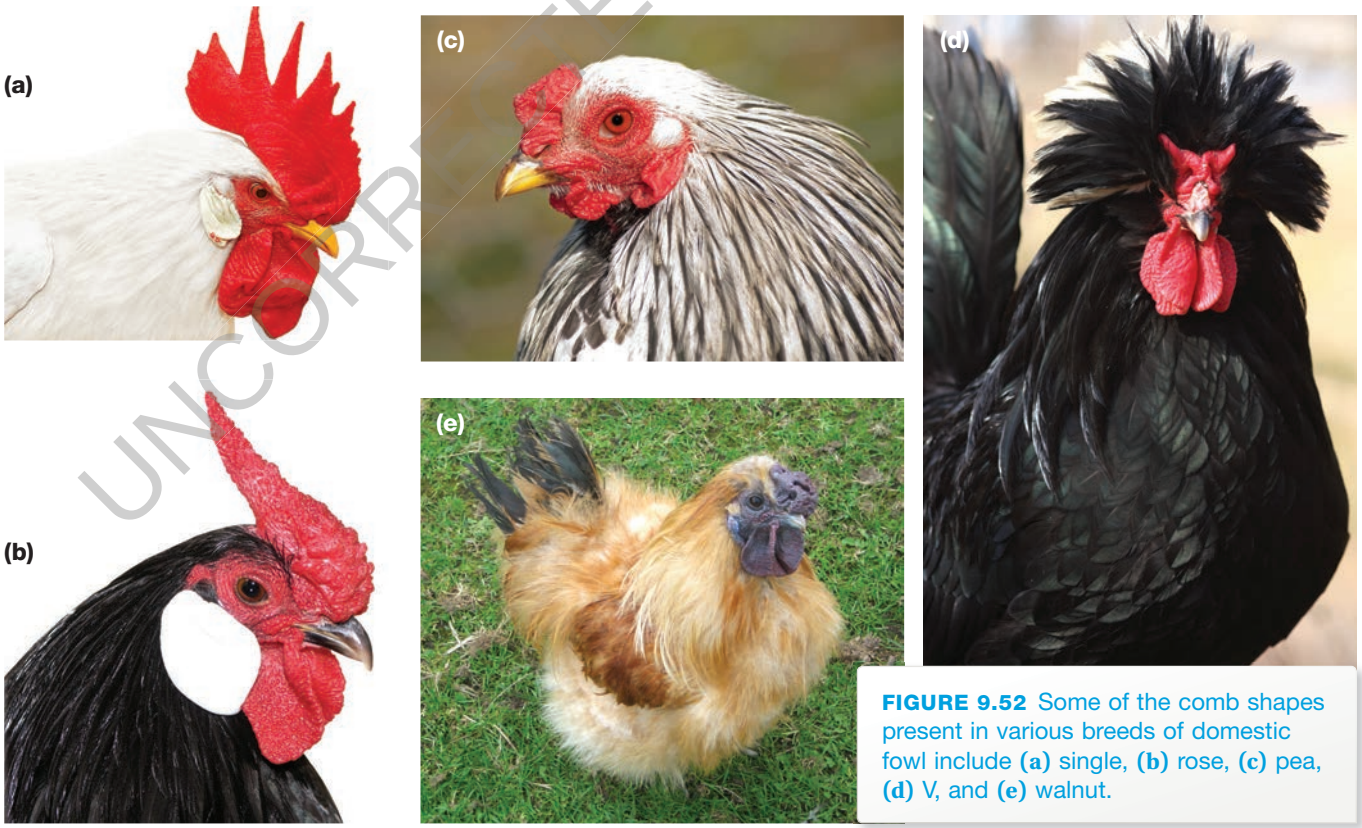


FIGURE 9.52 Some of the comb shapes present in various breeds of domestic fowl include **(a)** single, **(b)** rose, **(c)** pea, **(d)** V, and **(e)** walnut.

The diversity of breeds of various farm and pet animals is the result of selective breeding. This selective breeding may have been part of formal breeding programs or the breeding activities of animal fanciers. The power of selective breeding is seen, for example, in chickens. Selective breeding over many generations has transformed wild fowls into farmed chickens with very different egg production capabilities. Wild fowls produce a total of only about 12 eggs during their short breeding season each year. In contrast, domesticated chicken varieties lay eggs continuously throughout the year and annually produce very large numbers of eggs.

Effect of selective breeding on genetic fitness

Selective breeding or **artificial selection** occurs when breeders, fanciers and farmers prefer particular inherited features in their show animals and livestock because of their economic value or aesthetic appeal, and use selective breeding to enhance those features and increase their frequency in the breeding stock. In doing this, breeders manipulate the gene pool of their breeding stock.

Artificial selection is in contrast to natural selection, which favours only those inherited features that enhance survival and reproduction in the wild. Artificial selection can maintain features in a population that are economically important or aesthetically appealing but which are disadvantageous in terms of survival and reproduction. In natural populations in the wild, these features would come under negative selection pressure so that individuals with these features would not be maintained, and the alleles responsible would tend to be lost from the gene pool.

So, inherited characteristics that are the goal of selective breeding programs are not necessarily those features that best equip animals for survival and reproduction under natural conditions in the wild.

Artificial selection in domesticated species, particularly in so-called show varieties, can favour features that are clearly disadvantageous for survival and reproduction and would be positively selected against in the wild. Examples of features maintained only by artificial selection can be seen in domesticated animals such as:

- Jacobin pigeons with a distinctive arrangement of neck feathers that forms a ruff, masking their faces except from immediately in front (see figure 9.53a). Continued selective breeding over many generations has enhanced the size of the feather ruff that surrounds the face of the pigeon. Compare this image with figure 10.9, which shows a Jacobin pigeon in 1835. Because the large ruffs make it difficult for these pigeons to see, many breeders clip the ruff short during the breeding season to assist these birds to mate and ensure that they can feed their hatchlings.
- hairless Sphynx cats (see figure 9.53b) and dogs (see figure 9.50a above). The lack of an insulating fur coat in these pets means that, in cold environments, the homeostatic mechanisms that regulate core body temperature are at risk of failing.
- English bulldogs with greatly shortened muzzles that result in breathing problems (see figure 9.53c).

Other examples of disadvantageous features maintained thorough selective breeding can be seen in the short-legged dog breeds, such as dachshunds. These dogs are at a greatly increased risk of a disc herniation, in which one of the discs between the spine vertebrae is displaced. This creates pressure on the spinal cord, resulting in pain and, in some cases, temporary or permanent nerve damage.

Disadvantageous features such as these are maintained in the gene pools of these populations only through human intervention and selective breeding.

FIGURE 9.53 **(a)** Jacobin pigeon. Compare this image with figure 10.9b on page 449, which shows a Jacobin pigeon in 1835. Continued selective breeding over many generations has enhanced the size of the feather ruff that surrounds the face. **(b)** Sphynx cat. These cats are essentially hairless but most have some very fine soft hair on their noses and ears. Hairlessness in cats has arisen as a spontaneous mutation several times in different parts of the world. **(c)** English bulldog. The characteristic short muzzle of this breed has several disadvantages, including impaired breathing.

Technologies in selective breeding

In commercial herds and flocks, new reproductive technologies resulting in selective breeding include:

- artificial insemination
- sex selection through sperm sorting
- multiple ovulation and embryo transfer
- oestrus synchronisation.

ODD FACT

To collect sperm from a stud animal, a 'teaser' animal is used to entice the stud to mate. When the stud tries to mount the teaser, his penis is redirected into a warmed tube that is used to collect the semen.

ODD FACT

By means of AI, a single ram can 'mate' with 2000 ewes as compared to a maximum of 200 ewes through natural matings.

Artificial insemination

Natural breeding in mammals can be modified by **artificial insemination (AI)**. This technique changed herd management by altering the 'how', 'when' and 'where' of breeding.

AI involves collecting **semen** from a selected stud animal and then introducing this semen by artificial means into the reproductive tract of females of the same species. When first developed, AI involved the use of fresh semen only.

The use of AI increases the number of offspring that one stud animal can produce. One ejaculate from a male bull contains sufficient sperm to fertilise ten eggs. This volume of semen can be divided into ten portions and used to artificially inseminate ten cows and so can produce ten offspring. In contrast, in a natural one-to-one mating, that same volume of ejaculate would normally produce just one offspring.

In 1949, a successful technique was developed for freezing semen. 'Successful' meant that, when the frozen semen was thawed, it contained motile sperm that could fertilise eggs. The freezing technique involves adding

FIGURE 9.54 Transport of frozen semen and artificial insemination has geographically extended the reproductive capacity of prize studs.

semen to a special solution with a controlled pH and that comprises a mixture of various chemicals, including glycerol. Samples of this diluted semen (0.25 mL volume) are taken up in 'straws,' frozen rapidly and stored in liquid nitrogen at –196 °C. Under these circumstances, semen samples can be stored for many years and still retain their ability to fertilise an egg after thawing.

Through AI technology using frozen semen, physical and temporal barriers to mating are removed. This technology means that one prize stud animal can:

- fertilise many more females than under natural conditions
- fertilise females located hundreds or thousands of kilometres distant from the stud animal because its frozen sperm can be easily transported over great distances (see figure 9.54)
- fertilise female animals and produce offspring long after its death.

A process similar to AI is used by plant breeders with populations of cultivated plants. In plants, the process is termed **artificial pollination** and it is another example of human intervention in the evolutionary process. Unlike AI, artificial pollination has been used for centuries.

Embryo transfer in livestock (MOET)

AI and the use of frozen semen can greatly increase the contribution of particular bulls and rams to the genetic make-up of herds and flocks. In a similar way, a technique known as **multiple ovulation and embryo transfer (MOET)** allows high-quality cows and ewes to make a much greater than usual contribution to the future generations.

- **Multiple ovulation** refers to a process whereby a female receives injections of the follicle-stimulating hormone (FSH), which stimulates her to super-ovulate, or produce multiple eggs. An injection of gonadotrophin-releasing hormone (GnRH) is also given to make all the eggs mature at the same time.
- **Embryo transfer** refers to the process through which embryos at six to seven days of development are removed from the reproductive tract of a female and transplanted into the tracts of other females of the same species. These females act as surrogate mothers and carry the embryos to term and give birth.

MOET is another example of human intervention in the evolutionary process. In the process of MOET in sheep, for example, a high-quality donor ewe is treated so that she super-ovulates. When her eggs are released, they are fertilised, typically through AI with sperm from a selected ram. The fertilised eggs develop within the ewe's uterus for about six days. At the end of that time, the embryos are flushed from the ewe's uterus. On average, about seven embryos can be collected from a single flush. These embryos are immediately transferred directly into the uteruses of young recipient ewes (see figure 9.55), which will be the surrogate mothers of these embryos. Embryos not transferred to recipient ewes are frozen in liquid nitrogen and stored for later use. (Frozen embryos can be stored indefinitely.)

ODD FACT

In sheep, AI can be achieved through either an intra-uterine or a cervical technique. In intra-uterine AI, semen is placed directly in the uterus of a ewe through use of a laparoscope, with average pregnancy rates of 70 to 90 per cent. With cervical AI, semen is introduced via the vagina, with average pregnancy rates of 55 to 65 per cent.

ODD FACT

MOET has made the movement of genetic stock between countries easier because quarantine regulations are less stringent for frozen embryos than for live animals.

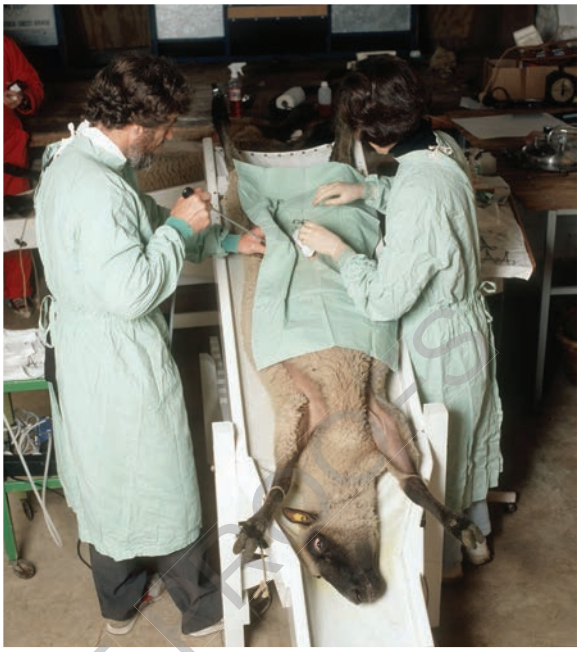


FIGURE 9.55 Transfer of an embryo into the uterus of a recipient, or surrogate ewe.

The same donor ewe can be used for MOET procedures several times during a breeding season. Over a normal reproductive lifetime, one ewe might produce 30 eggs. Multiple ovulation, however, greatly increases this egg output. The advantages of embryo transfer are that genetically important female lines can be multiplied at much faster rates than can occur through normal reproduction and that valuable embryos can be stored. Under conditions of natural selection, this would not be possible.

ODD FACT

The sex of an embryo produced by IVF can be identified at a very early stage of its development before it is transferred to a female. A single cell is taken from the embryo, cultured and the sex chromosome make-up is identified.

Sex selection through sperm sorting

Under normal circumstances, a sex ratio of about one male to one female is expected in live-born mammals. In the beef industry, male calves are preferred because they have more beef (muscle) on their carcasses at a given age than females. In contrast, in the dairy industry, female calves are necessary for milk production and are preferred (see figure 9.56).

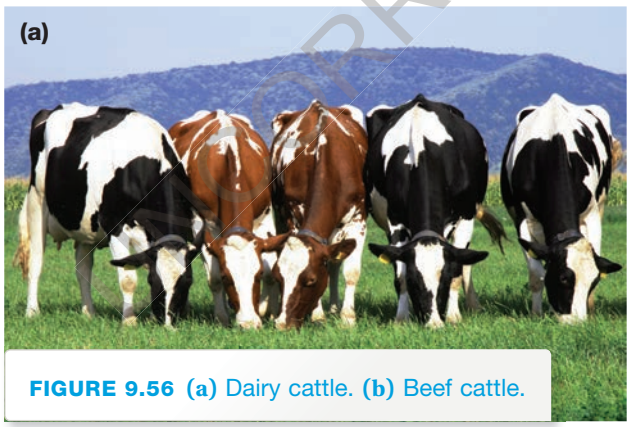


FIGURE 9.56 **(a)** Dairy cattle. **(b)** Beef cattle.

Sex selection is now possible. After semen has been collected from a stud bull, for example, it is possible to treat the semen and separate sperm with X chromosomes from those with Y chromosomes. Sperm cells are first labelled with a harmless fluorescent dye that binds to the DNA. The X chromosome in mammals is larger and contains more DNA than the Y chromosome.

As a result, the sperm with X chromosomes fluoresce more brightly than those with Y chromosomes. After labelling, the sperm are then separated into two groups depending on their fluorescence. The use of this sperm separation technique has allowed sex selection to occur on a large scale (see figure 9.57).

FIGURE 9.57 A technician at work with a FACStarPLUS flow cytometer used for the rapid and accurate separation of cells. Cells are stained with a fluorescent dye and this instrument uses a laser beam to sort cells according to differences in their fluorescence. If sperm cells were being sorted using fluorescence from a DNA-binding dye, which sperm would fluoresce more — sperm with an X or a Y chromosome?

Manipulating breeding cycles

It is now possible to **synchronise the time of oestrus** or sexual receptivity of female farm animals, such as cattle and sheep. **Oestrus synchronisation** results in all sexually mature females being in oestrus within a predictable and narrow time frame, with the result that the time of fertilisation in a herd or flock, either by AI or natural mating, can be more efficiently managed. Synchronisation is also necessary for MOET procedures so that the intended embryo donor and the recipient surrogate mothers come into oestrus at the same time.

Advantages of synchronisation include:

- less time (and hence lower labour costs) needed to test animals to see if they are in oestrus
- higher fertilisation rates and birth rates
- more uniform and manageable crops of calves or lambs, since all young are born within a short period
- lower mortality rates because greater oversight of all newborns is possible.

Oestrus synchronisation can be achieved in a number of ways. One method depends on the fact that the hormone progesterone inhibits ovulation by stopping production of another hormone, oestrogen, which is needed to bring female animals into oestrus. By adding an external source of progesterone to female livestock, oestrus production and the associated ovulation are suppressed. When the source of progesterone is simultaneously removed from a group, mature females go into oestrus and ovulate within a short time period. How is this external progesterone delivered?

ODD FACT

Release of the hormone oestrogen from ovarian follicles stimulates the release of luteinising hormone (LH) from the pituitary gland. Release of LH in turn causes ovulation and suppresses oestrogen production.

Methods of supplying progesterone to farm livestock include:

- feeding using a dietary supplement
- implants under the skin
- sponges inserted into the vagina
- CIDRs (controlled internal drug-releasing devices) inserted into the vagina.

In the case of CIDR (pronounced cee-dar) use in cattle, the insert is left in place for seven days. When the insert with its supply of progesterone is removed, the level of circulating progesterone drops and oestrus begins within three days.

Genetic impact of selective breeding

Genetic variation refers to the genetic differences present in the members of a population. In the gene pool of a population, the greater the number of different alleles of each gene, the greater the genetic variation in that population.

Selective breeding of domesticated animals, such as cattle and sheep, occurs when the parental input to the next generation is restricted to a small number of animals, selected on the basis of their superiority for a limited number of inherited traits. This process of artificial selection can result in a loss of genetic variation in the gene pool of the herd or flock concerned. The loss of genetic variation may not be noticed immediately and may become apparent only when there is a change in the environmental conditions. If, for example, members of an animal population have never been exposed to a particular disease-causing virus, the loss of an allele that confers resistance to that disease would not be noticed. The value of the allele in question would be realised only after the virus comes into the environment.

Genetic variation can also be lost when plant crops are cultivated from commercially produced seeds that come from just one, or a small number, of the many varieties of that plant species.

Loss of genetic variation can be an unintended consequence of the use of reproductive technologies, such as artificial insemination (AI) combined with the transport of frozen semen, which increases the genetic influence of a small number of stud males in animal herds and flocks. Similarly, the use of multiple ovulation and embryo transfer (MOET) restricts the number of breeding females in animal herds and flocks. Likewise, the use of artificial pollination with a limited number of plants as the pollen source may reduce the genetic variation of a plant population under cultivation.

Genetic variation is lost when habitats are destroyed in a region with the local loss of plant and animal species. Although other populations may exist in other regions, what will be lost is the genetic variation that is unique to those local gene pools.

Does loss of genetic variation matter?

The death of more than one million Irish peasants from starvation in the period from 1845 to 1849 is a tragic reminder that genetic variation matters. Potatoes were the staple diet of Irish peasants. When an outbreak of late potato blight occurred, caused by the fungus *Phytophthora infestans*, potato tubers turned black and rotted in the ground.

Potatoes are endemic to South America, where hundreds of species grow wild. This food crop was introduced to Europe, first by the Spaniards in 1570 and later, in about 1590, by the English. All potatoes grown in Europe were plants descended from the 'European' potato (*Solanum tuberosum*). When potato blight broke out in Ireland in 1846, this potato variety was genetically uniform for susceptibility to fungal infection so that all the potato tubers rotted. This produced a mass famine that resulted in the deaths of an estimated

FIGURE 9.58 Sculpture by Rowan Gillespie commemorating the Great Famine, located on Custom House Quay in Dublin's Docklands. (Image courtesy of Judith Kinnear.)

one million people, either directly from starvation or from starvation-related diseases. About another one million people emigrated, the majority to the United States, but significant numbers came to Australia. This tragic event is commemorated in a sculpture on the Custom House Quay beside Dublin's Liffey River (see figure 9.58).

Natural populations of the wild relatives of plants can be the source of alleles for potentially valuable traits. For example, in the 1940s, resistance to the late potato blight disease was identified in *Solanum demissum*, which grows wild in the Andes and is a close relative of the European potato. Crossbreeding was used to introduce this allele into the commercial varieties of potato, providing them with resistance to blight.

Saving genetic variation

As wild populations are destroyed through land clearance and as larger areas are devoted to the cultivation of smaller numbers of commercial crop varieties, the safeguarding of genetic variation in wild populations is critical. In the case of plant varieties, some contribution to this is being achieved through the establishment of seed banks.

Gene banks and seed banks

In 1992, Western Australia became the first Australian state to establish a genetic diversity seed bank when the *Threatened Flora Seed Centre* was established at the WA Herbarium. In 2011, the centre relocated to state-of-the-art facilities within the WA Conservation Science Centre. Since starting operations in 1993, the centre has built a store of seeds from many threatened plant species. Seeds of rare and endangered plant groups are collected from various populations of the native plant groups. (Why collect from more than one population?) After collection, the seeds are carefully cleaned and counted in a laboratory (see figure 9.59a). The seeds are then dried to reduce their water content, then stored in sealed aluminium foil packs at low temperatures (see figure 9.59b). The centre also engages in research and has reintroduced many threatened species back into the wild.

FIGURE 9.59 **(a)** Processing and testing of seed in the laboratory prior to storage. **(b)** The freezer. Extracted and dried seed is sealed in laminated aluminium foil packages and frozen at –18 °C for the long term. (Images courtesy of Threatened Flora Seed Centre, Science and Conservation Division Department of Parks and Wildlife, Western Australia.)

Other states have also established seed banks to store seeds from native plants and together they form the Australian Seed Bank Partnership. The Victoria node of this partnership is located in the National Herbarium of Victoria at the Royal Botanic Gardens Victoria in Melbourne.

Other facilities have a major focus on the seeds of crop plants. In March 2014, the Australian Grains Genebank was opened in a new facility in Horsham, in country Victoria. This centre combines the temperate field crops collection from Horsham, the cereals crop collection from Tamworth in New South Wales, and the tropical crops collection from Biloela in Queensland. The gene bank houses seeds from pulses (pea and bean species), cereals, oilseeds and, importantly, from their wild relatives from temperate and tropical regions of Australia. Seeds from indigenous wild crop varieties also form part of this collection. This facility will hold up to 180 000 samples from Australia and around the world. Samples of 1000 seeds are dried to 6 per cent moisture and then stored in vacuum-sealed, triple-foil aluminium pouches at –20 °C. Small seed samples of 100 seeds are distributed free on request to plant breeders, scientists and research students, and are also exchanged with other gene banks internationally. When stocks become low, seeds are grown for regeneration. This is not only a means of replenishing seed stocks, but it also provides an opportunity to collect relevant data on seed performance, such as time to flowering, height, and grain yields.

An important step in the preservation of the genetic variation and biodiversity of the world's major crop plants and their wild relatives occurred in February 2008 when the Svalbard Global Seed Vault (SGSV) was opened on remote Spitsbergen Island close to the North Pole (see figure 9.60). The SGSV facility has large underground storage areas dug into the side of a mountain and these are maintained at a temperature of −18 °C (see figure 9.61). Here, duplicate samples of seeds from seed banks throughout the world can be placed in secure long-term storage at no cost. This storage acts as an insurance against the loss of the valuable genetic variation in food crop plant varieties and their wild relatives.

FIGURE 9.60 The Norwegian island of Spitsbergen, north of the Arctic Circle, is the location of the Svalbard Global Seed Vault.

At capacity, SGSV can store more than 4 500 000 samples, each containing up to 500 seeds. During the first year of its operation, various national and international groups deposited more than 400 000 unique seed samples. The first seeds from Australia were deposited in the SGSV in early 2011.

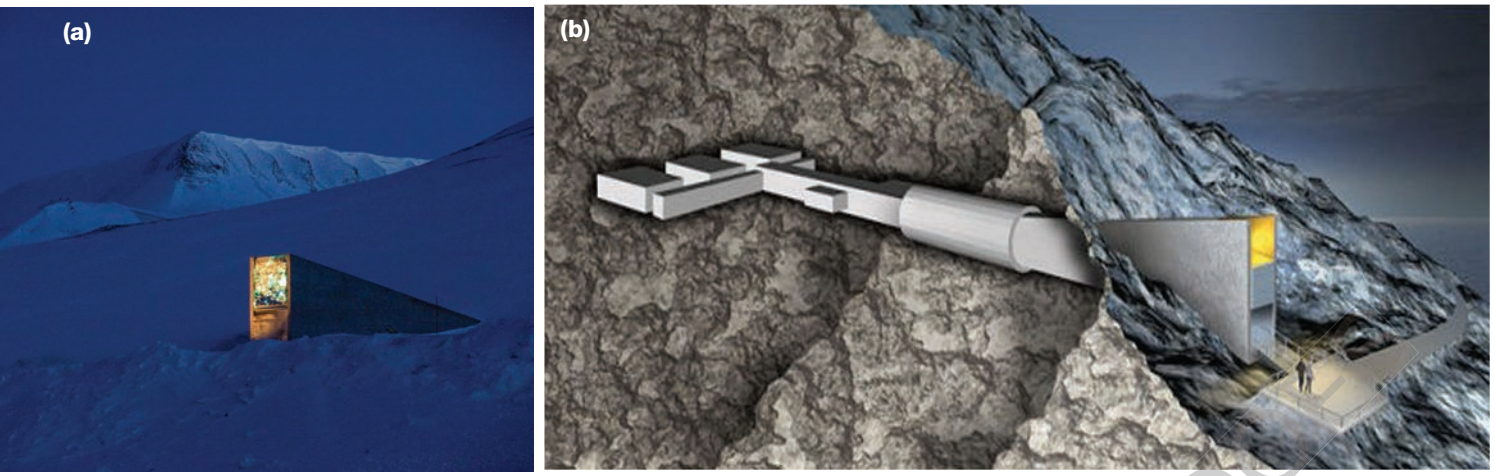


FIGURE 9.61 **(a)** Entrance to the Svalbard Global Seed Vault on the island of Spitsbergen. **(b)** Diagram showing the three storage areas at the end of a tunnel deep within a mountain. This facility will ensure that the genetic variation in plant crops is preserved for future generations. (Images courtesy of the Global Crop Diversity Trust.)

BIOLOGISTS AT WORK

Catherine Cavallo and Sonia Sanchez are PhD students with Monash University and Phillip Island Nature Parks. Together, they study little penguins in their environment, in an effort to better understand and protect marine ecosystems for the future.

Cathy and Sonia write: 'Marine ecosystems are under increasing stress from climate change and other pressures such as pollution and overfishing. We rely on these ecosystems for food, oxygen production, employment and recreation, so it is critical that we protect them. But first, we have to understand how they function. In marine environments, this is a difficult and expensive task, requiring an innovative approach.'

'To understand what goes on beneath the waves, we monitor little penguins at Phillip Island. As top predators, penguins reflect changes in the food web below them, and are useful indicators of ecosystem health. We use an automated monitoring system to count and weigh penguins as they enter and exit the colony over a specially designed bridge. This tells us how long penguins have to spend hunting to find enough food, and how successful they are. We also check nests during the breeding season, counting eggs and chicks and weighing chicks to monitor their growth rates. Little penguin breeding success is very sensitive to prey availability, so we can learn a lot about the ecosystem by observing how quickly their chicks grow and how many survive to leave the burrow.'

Cathy continues: 'My specific role is to use DNA analysis to understand penguin diet, by identifying prey DNA in their poo. It's a smelly task, but an important one, which gives us invaluable information about the abundance and availability of little penguin prey, and, consequently, health and change in the penguins' environment.

'Several times a year, I get to travel to the Australian Antarctic Division in Tasmania to analyse my samples alongside scientists who work in Antarctica. I also go to conferences to share ideas on how we can protect the planet with our colleagues from all over the world. My PhD project involves my working alongside government (Australian Antarctic Division), tourism (Phillip Island Nature Parks) and media, and means working in some incredible locations.

'As a kid, I always knew I wanted to work with animals, but as I grew I fell deeply in love with the sea, and felt compelled to understand and protect it. Through my research, I feel that I am helping to protect our precious marine environments and species from the growing pressures humans place on them. My study path included a Bachelor of Science (majoring in zoology) and a Master of Science studying sea turtles and climate change, as well as taking every opportunity that was offered to me to increase my research experience and networks.'

Sonia adds: 'Currently, less than 4 per cent of our oceans are protected. Tracking marine top predators at sea can be an important tool to identify important sites to protect within marine protected areas, as they feed in areas where food is available and abundant. I am trying to find out where the feeding hotspots of little penguins from Phillip Island are. I attach two miniaturised devices to their backs, a GPS and an accelerometer, to record their location and

behaviour at sea. With this technology I can see where the penguins go and where they find food. By protecting these areas, we protect not only the food source for the penguins, but the wider ecosystem as a whole. I spend many nights waiting for my penguins to return to their burrows, so I can retrieve their tracking devices.

'When I was a kid I used to spend all my summers swimming and exploring at the beach and I guess that is when I decided I wanted to study marine sciences, to protect and understand that sea that made me so happy. I did a Bachelor of Science and a Master in Marine Sciences in Barcelona (Spain), my home city, where I used to work on anchovies and sardines, and spent time living and working with other researchers on an oceanographic vessel.'

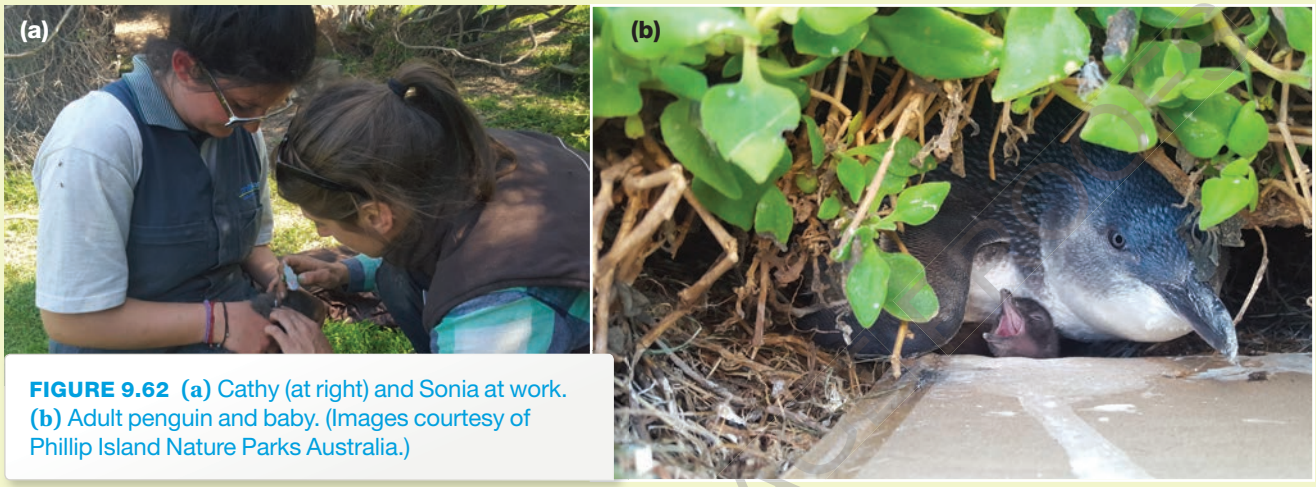


FIGURE 9.62 **(a)** Cathy (at right) and Sonia at work. **(b)** Adult penguin and baby. (Images courtesy of Phillip Island Nature Parks Australia.)

KEY IDEAS

- Selective breeding is one form of artificial selection used in plant and animal breeding.
- Artificial selection using selective breeding can maintain inherited features in a population that, under natural conditions, would be selected against.
- Several new reproductive technologies are available to assist selective breeding in commercial flocks and herds.
- Reproductive technologies, such as AI, artificially increase the contribution of selected animals to the gene pool of the next generation.
- Use of reproductive technologies can result in reduced genetic variation in a gene pool.
- Loss of genetic variation in a population may result in failure of the population to survive environmental change.
- Seed banks and gene banks are among the facilities that seek to conserve the genetic diversity of plant species.

QUICK CHECK

22 Define the following terms:
- **a** artificial insemination
- **b** artificial pollination.

23 What is the likely effect on the genetic variation in the gene pool of herds where reproduction involves AI using semen from one prize stud?

24 Identify two consequences for AI when technology allowed semen to be successfully frozen.

25 What is meant by the abbreviation MOET?

26 **a** Why did the potato crop in Ireland fail in the 1840s?
- **b** What does this show about genetic variation?

BIOCHALLENGE

1 Consider the diagrams (A to D) below. Match each diagram to one of the chapter keywords giving a brief explanation for your choice. Note that each coloured dot represents an individual organism.

2 Explain what change, if any, each of these events would produce on the gene pool of the population denoted on the right-hand side of each diagram.

3 The two generations shown in D below have been living under a particular set of environmental conditions. The release of a pollutant changes the environmental conditions so that individuals denoted by the pink dots are at a selective advantage. Draw a possible representation of the generation 3 population.

4 A normal protein has 141 amino acids.
A single gene mutation produces a mutant form of the protein that is 31 amino acids longer.
Describe what kind of mutation could have caused this.

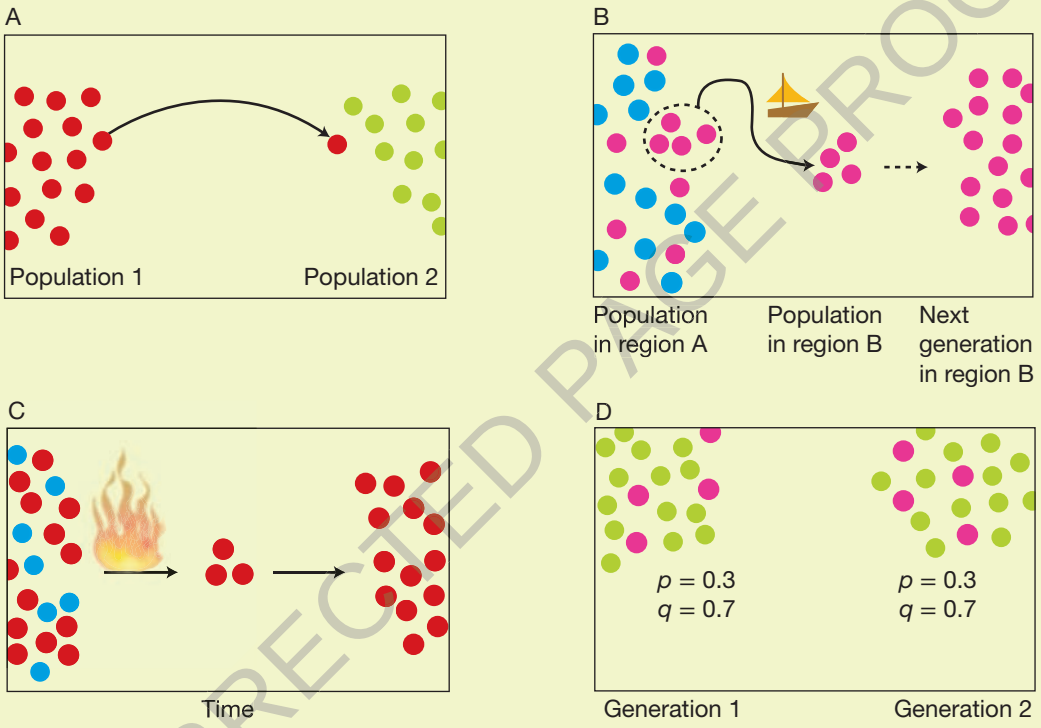


FIGURE 9.63

Chapter review

Key words

agent of selection
allopolyploid
aneuploid
artificial insemination (AI)
artificial pollination
autopolyploidy
bottleneck effect
complete selection
conservative missense
conservative missense mutation
continuous
differential reproduction
discontinuous
embryo transfer
fixed
founder effect
founder population
frameshift
gene mutation
gene pool
genetic drift
genetic variation
Hardy–Weinberg principle
hybridisation
immigration
intra-specific evolution
lactase nonpersistent
lactase persistent
LCT
monomorphic
multiple ovulation
multiple ovulation and embryo transfer (MOET)
mutagenic agents
natural selection
non-conservative missense mutation
oestrus
oestrus synchronisation
partial selection
polygenes
polymorphic
polymorphisms
population
position effects
post-mating isolation mechanisms
pre-mating isolation mechanisms
random mating
selecting agents
selective advantage
selective breeding
semen
sex selection
speciation
trinucleotide repeat expansion (TRE)

Questions

1 **Making connections** ➜ Draw a concept map for 'genetic changes in populations' incorporating key words from this chapter. You may add any other concepts that you wish.

2 **Demonstration knowledge and understanding** ➜ Identify the difference between the members of the following pairs:
- **a** gene flow and gene pool
- **b** genic change and chromosomal change (in the make-up of a population)
- **c** natural selection and artificial selection
- **d** germline mutation and somatic mutation.

3 **Demonstrating knowledge and understanding** ➜ In an animal species, body colour is inherited with yellow being dominant over purple. Consider two populations of this species: Population A consists of 9 purple and 1 yellow individual; population B consists of 900 purple and 100 yellow individuals.
- **a** In which population would genetic drift be more likely to lead to loss of the allele controlling the yellow colour? Explain.

  Population C consists of 500 purple and 500 yellow individuals.

  A sample of ten individuals is chosen at random from each of populations B and C. These small groups will be the founder groups of new populations in two different regions.
- **b** Which founder group is more likely to have less genetic diversity in terms of body colour. Explain.

4 **Demonstrating knowledge and understanding** ➜ Consider the following mutations in the coding strand of DNA and identify the kind of mutation that they represent. Assume that the code will be read starting from the first base shown.
- **a** ...TTT TCT AGG GTC ➜
  ...TTT T**G**T AGG GTC
- **b** ...TTT TCT AGG GTC ➜
  ...TTT TCT **A**AG GGT C
- **c** ...TTT TCT AGG GTC ➜
  ...TTT CTA GGG TC
- **d** ...TTT ATT GTC CCT ➜
  ...TTT AT**C** GTC CCT

5 **Developing valid biological explanations** ➜ Suggest a reasonable explanation for the following observations:
- **a** Indigenous populations in Australia and North America show a high percentage of lactase nonpersistence, but some indigenous populations in Africa exhibit a low percentage lactase nonpersistence.
- **b** Populations descended from groups that do not have a history of herding dairy animals have only a small proportion of lactase persistent individuals.

6 **Demonstrating knowledge and understanding** ➜ An autosomal gene in a population of diploid organisms has eight different alleles. How many different alleles could be present in:
- **a** one member of this population
- **b** the gene pool of this population?

7 **Analysing data and applying principles** ➜ In an imaginary large population of sexually reproducing

organisms, foot colour is a monogenic trait with green feet (***G***) being dominant to yellow (***g***). Both green-footed and yellow-footed organisms occur in this population. These short-lived organisms normally breed and then die. Assume that all organisms with green feet are prevented from breeding.

a Under these conditions, how rapidly will the ***G*** allele be lost from the gene pool of this population?

b What are values of *p* and *q* when the ***G*** allele is removed from the gene pool?

Return to the original population with organisms of both foot colours and assume that all yellow-footed organisms are prevented from breeding.

c Under these conditions, will the ***g*** allele be lost from the gene pool as quickly as the ***G*** allele? Explain.

d Which genotype acts as the hidden reservoir of an allele that controls a recessive trait?

8 Developing valid explanations ➜

FIGURE 9.64 Two adult dogs, a Great Dane and a Chihuahua.

This figure above shows examples of inherited variation in dogs.

Identify, giving an explanation for your decision, the most likely genetic basis for:

a the observed variation in body size between the two dogs

b the observed variation (black/yellow) in the base colour of the two dogs (ignore the white spotting).

Consider the proposal that these size differences emerged in wild dog populations by natural selection.

c Is this assumption reasonable or not? Explain your choice.

d If you decided that the assumption is not reasonable, suggest an alternative explanation.

9 Analysing information ➜ Read the following abstract of a research report by Iannuzzi, M.C. *et al.* published in the *American Journal of Human Genetics* vol. 48m, page 227 (1991):

'Cystic fibrosis (CF) is a recessive disease caused by mutations in the CF transmembrane conductance regulator (CFTR) gene. We have identified in exon 7 two frameshift mutations, one caused by a two-nucleotide insertion and the other caused by a one-nucleotide deletion; these mutations–CF1154insTC and CF1213delT, respectively, are predicted to shift the reading frame of the protein and to introduce termination codons at residues 369 and 368.'

a What is a frameshift mutation?

b Two frameshift mutations are identified in this case of cystic fibrosis. Note that CF means cystic fibrosis and the following number identifies a base position in the DNA of that gene. What do you think is meant by the shorthand CF1154insTC and CF1213delT?

c What effect did these frameshifts have on the protein encoded by this mutated gene?

10 Demonstrating understanding and communication ➜ The sheep strike blowfly, *Lucilia cuprina*, is a major pest for sheep farmers. In 1955, the insecticide dieldrin was first introduced to control this pest. The introduction was initially highly successful, but, within two years, this pesticide began to be less effective against these blowflies.

a Explain carefully what was happening in this two-year period?

b Outline what would be expected to happen if dieldrin continued to be used.

11 Analysing data and making predictions ➜ Populations of giraffe (*Giraffa camelopardalis*) from different regions on the African continent show distinctive differences in inherited traits, including colour and pattern. In all, up to nine **subspecies of giraffe** are recognised. Figure 9.65 shows the geographic distributions of the various groups.

a What might be expected to happen over a very long period if the populations remain geographically separated?

b Suggest a likely origin for the different colours and patterns in the different subspecies.

c What test would tell if the giraffe populations are the same or different species?

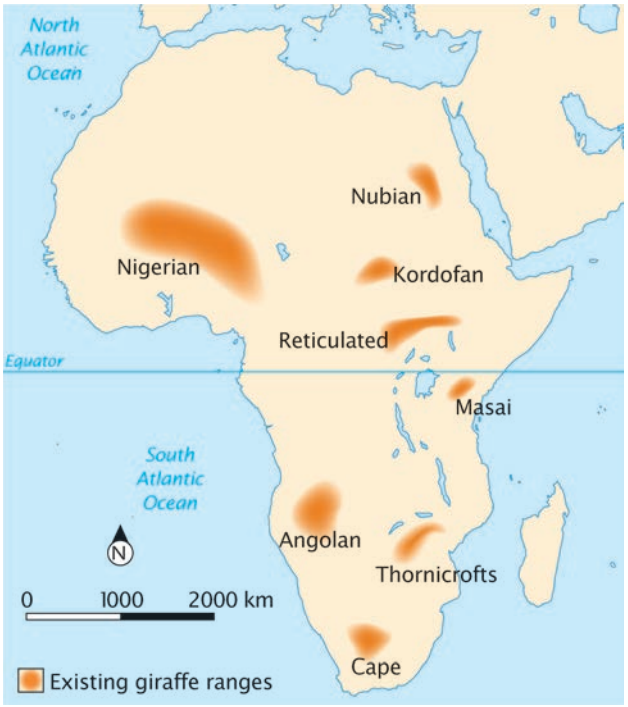
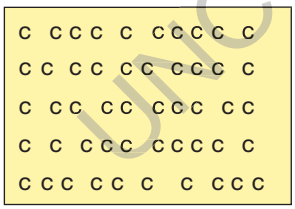


FIGURE 9.65 (Map provided by MAPgraphics Pty Ltd, Brisbane.)

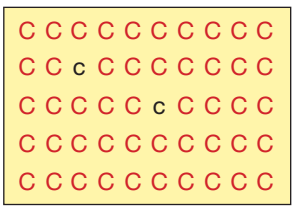
12 Demonstrating understanding and communication ➜ Examine figure 9.66, which shows stylised representations of the gene pool of the same population at three different times. The time periods are sufficiently long for several generations to have passed. The letters denote the alleles, C and c, of one gene.

The images are not in any particular order. Place the boxes in an order of your choice that you think is feasible and tell the story of what has happened in the population over the time period involved, consistent with your chosen sequence of gene pool images.

Population (A)



Population (B)



Population (C)

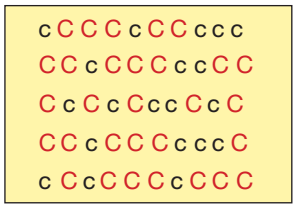
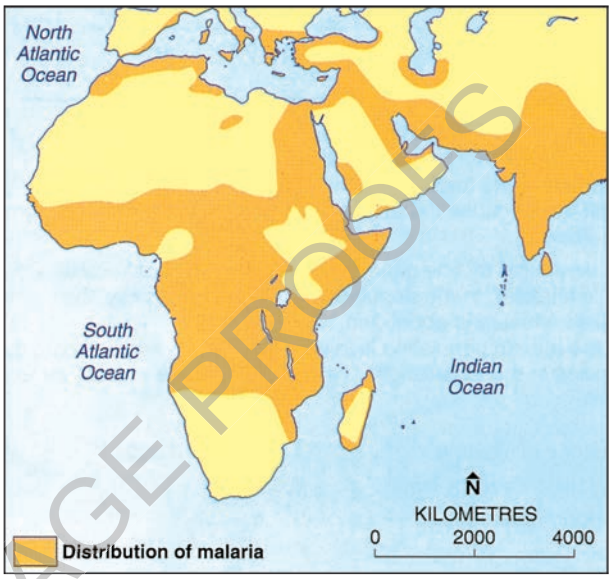


FIGURE 9.66

13 Analysing data and drawing conclusions ➜ Refer back to table 9.5 on page 399. The areas of Africa, south-west Asia and southern Europe subject to malaria are shown in figure 9.67a. The frequency of the H^S allele that is responsible for sickle-cell anaemia is shown in figure 9.67b.

(a)



(b)

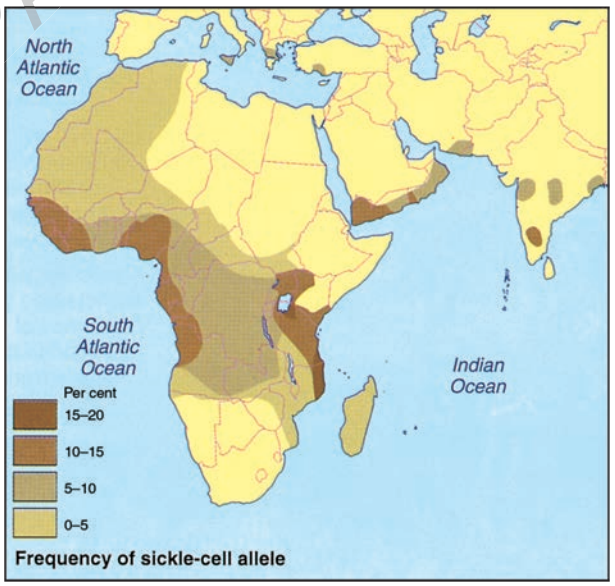


FIGURE 9.67 **(a)** Map showing malaria-prone areas. **(b)** Map showing frequency of H^S allele. (Maps provided by MAPgraphics Pty Ltd, Brisbane.)

a What is the most likely origin of the H^S allele?
b Is the high frequency of the H^S allele in areas of Africa best explained by the action of natural selection or by genetic drift? Explain.
c If malaria were eradicated from this planet, what changes in the frequency of the H^S allele might be expected to occur over time? Explain.

d The frequency of the H^S allele is not rare in the United States, yet that country is not significantly affected by malaria. Suggest a possible explanation for this observation.

e The H^S allele is extremely rare in Australian populations and has a frequency far less than the frequency found in the United States. Suggest a possible explanation for this observation.

14 Discussion question ➜

Consider the following statement:

Selection pressures can change — it is likely that the predation pressure acting on hominins was reduced by cultural development such as toolmaking and the controlled use of fire.

(Hominins include the early human species *Homo habilis* and a later species, *Homo erectus* (whom you will meet in chapter 12.) Note that cultural practices are not inherited traits.)

a What is meant by the term 'predation pressure'?

b What is meant by 'the controlled use of fire'?

The first humans to exercise the controlled use of fire are believed to be the human species *Homo erectus*.

c Discuss how the ability to exercise the controlled use of fire might contribute to a reduction in predation pressure on populations of *Homo erectus*.

d Identify other benefits that could been generated by the controlled use of fire in these populations.

Simple stone tools first appeared in association with *Homo habilis* and then came more sophisticated tools made by *Homo erectus*. Populations of early human species survived by hunting and gathering.

e Discuss how the development of various tools might have resulted in more efficient and more successful hunting and gathering practices of early humans.