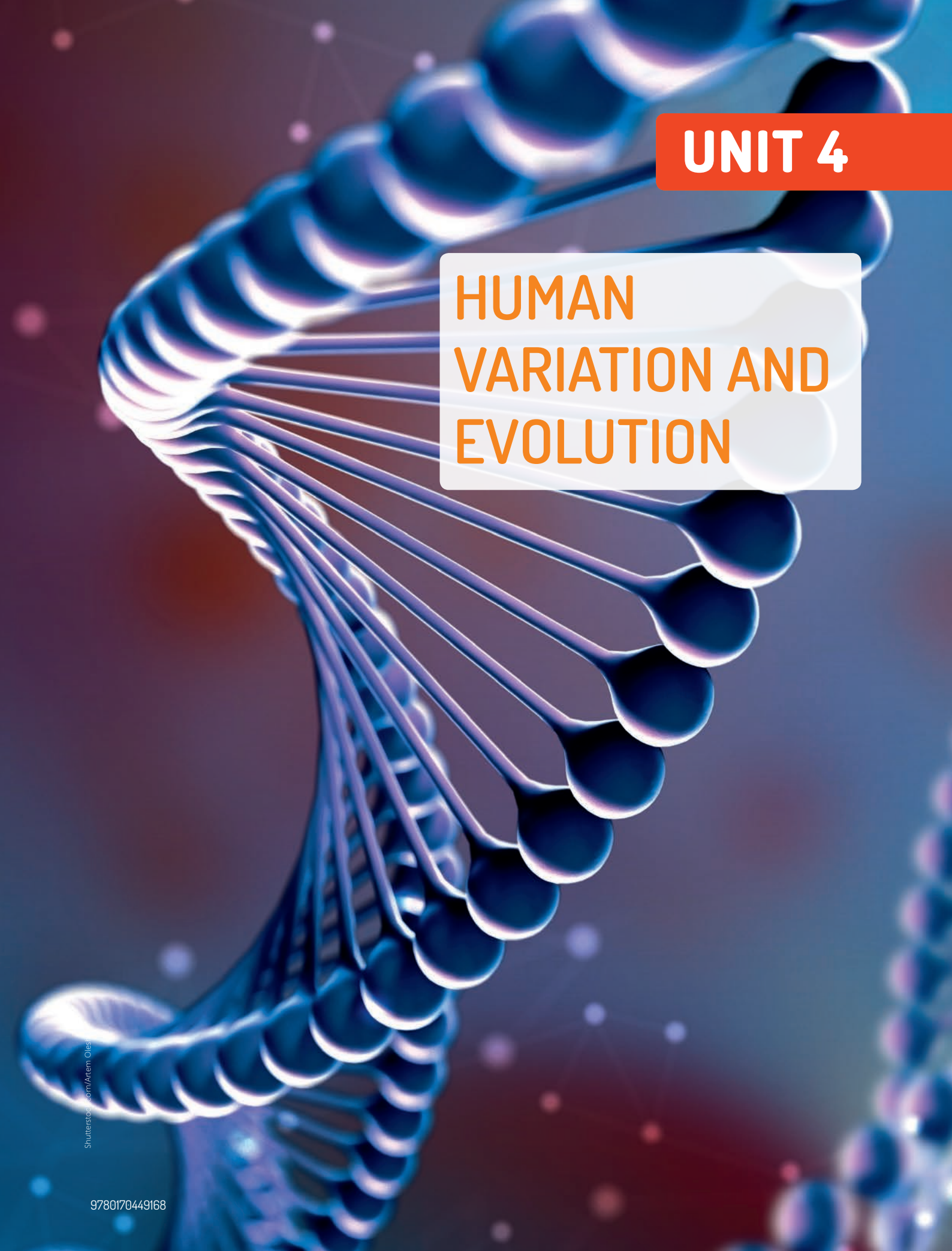

UNIT 4

HUMAN VARIATION AND EVOLUTION

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9 EVOLUTION PRODUCES CHANGES ACROSS GENERATIONS

UNIT 4 CONTENT

SCIENCE INQUIRY SKILLS

- conduct investigations, including the use of virtual or real biotechnological techniques of polymerase chain reaction (PCR), gel electrophoresis for deoxyribonucleic acid (DNA) sequencing, and techniques for relative and absolute dating, safely, competently and methodically for valid and reliable collection of data
- interpret a range of scientific and media texts, and evaluate models, processes, claims and conclusions by considering the quality of available evidence; and use reasoning to construct scientific arguments
- select, use and/or construct appropriate representations, including phylogenetic trees, to communicate conceptual understanding, solve problems and make predictions

SCIENCE UNDERSTANDING

Mutations

- mutations in genes and chromosomes can result from errors in DNA replication, cell division or from damage caused by mutagens
- different genotypes produce a variety of phenotypes, which are acted on differently by factors in the environment, producing different rates of survival
- mutations are the ultimate source of variation introducing new alleles into a population: new alleles may be favourable or unfavourable to survival

Gene pools

- populations can be represented as gene pools that reflect the frequency of alleles of a particular gene; gene pools can be used to compare populations at different times or locations
- gene pools are dynamic, with changes in allele frequency caused by:
  - mutations
  - differing selection pressures
  - random genetic drift, including the founder effect
  - changes in gene flow between adjoining groups
- the incidence of genetic diseases in particular populations illustrates the effects of different factors on the dynamics of gene pools, including the incidence of Tay-Sachs disease, thalassemia (α and β) and sickle-cell anaemia
- natural selection occurs when factors in the environment confer a selective advantage on specific phenotypes to enhance survival and reproduction
- the mechanisms underpinning the theory of evolution by natural selection include inherited variation, struggle for existence, isolation and differential selection, producing changes to gene pools to such an extent that speciation occurs

Source: School Curriculum and Standards Authority, Government of Western Australia

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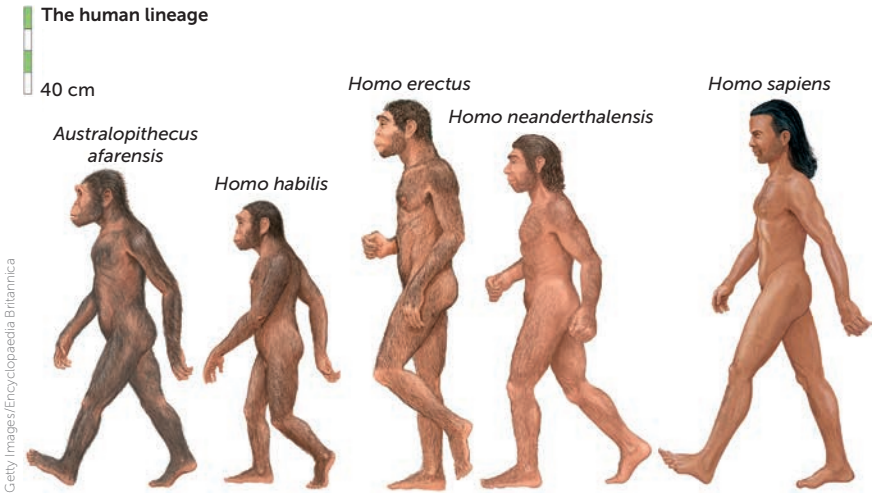

Getty Images/Encyclopaedia Britannica

FIGURE 9.1 An artist's representation of different hominin species as they have changed through evolution

In Chapter 7 you learnt about bacteria that have become resistant to antibiotics. Some bacteria may have a mutated gene that allows them to survive antibiotic treatment. These bacteria are the ones to reproduce and, therefore, there are more resistant bacteria in the next generation. Over time, more and more bacteria are resistant and so the species has **evolved**.

Evolution is the change in characteristics of a species over time. It is a gradual change that occurs over a number of generations, rather than the change of a particular individual or generation. The **phenotypes**, or set of characteristics, of individuals are a result of the alleles, or **genotype**, for each trait. Therefore, evolution reflects the changes in allele frequency in populations.

Changes to the alleles present in a population may be due to new alleles forming as a result of mutations or being introduced to a population through migration. The frequency of these alleles may alter because of a selective pressure in natural selection or by chance in genetic drift. We will be looking at these mechanisms of evolution in this chapter.

Antibiotic resistance
This article has more information about bacteria evolving to become antibiotic resistant through natural selection.

9.1 MUTATIONS

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FIGURE 9.2 Variation occurs in humans

Gene pool

A **population** is a group of organisms of the same species living together in a particular place at a particular time. When studying populations, **geneticists** – scientists who specialise in the study of inheritance – prefer to consider the characteristics of the population as a whole and not those of the individuals that make up the population. They find it convenient to pool the genotypes of all the individuals capable of reproducing and refer to this as the gene pool. Thus, the **gene pool** is the sum of all the alleles in a given population.

When studying a population, geneticists are interested in how often each allele of a gene occurs in the gene pool for that population. These are called the **allele frequencies** for the population. For example, an allele for cystic fibrosis is found on chromosome number 7. If the frequency of the cystic fibrosis allele in a given population is 5%, then among population members, five in every 100 of chromosome 7 will carry that allele. Ninety-five out of 100 chromosome 7s will have the normal form of the gene.

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FIGURE 9.3 The frequency of the allele for blue eye colour would be higher in the Scandinavian population than in the African population

Populations that differ in the characteristics they possess are likely to have different frequencies of the various alleles of a gene in their respective gene pools. For example, Scandinavians commonly have blue eyes, whereas black Africans have brown eyes. The frequency of the allele for blue eyes would be much higher in the Scandinavian gene pool than in the African gene pool. Thus, any two populations having differing characteristics are likely to have different gene pools.

Mutations

Offspring may show variations that do not resemble either parent, and have never occurred before in the history of the family. Therefore, they are due not to an allele being passed down from the parents, but to a new allele being formed. This can happen when the DNA is changed by a **mutation**, resulting in a different variation of the trait. Not all mutations are harmful, but many are. An organism with a characteristic resulting from a mutation is called a **mutant**. There are two main types of mutations:

- **gene mutations**, which are changes in a single gene so that the traits normally produced by that gene are changed or destroyed
- **chromosomal mutations**, in which all or part of a chromosome is affected.

If a mistake occurs spontaneously when the DNA molecule is copied during mitosis or meiosis, or when the chromosomes are separated during meiosis, the change may have significant effects on the functioning of the cell. However, many mutations are repaired, and therefore don't cause a problem. If they do remain, when the cell divides the mutated DNA will be copied and passed on to daughter cells. If the daughter cells are gametes, the mutation may be passed on from generation to generation.

There are relatively few mutations in human populations, considering the millions of cell divisions that occur. Those that do occur sometimes result in traits better suited to a particular environment, and so may contribute to human survival.

Mutations
This website provides more information on mutations.

Causes of mutations

Mutations occur without any known cause, but a number of agents are known to increase the rate at which they occur. These are called **mutagenic agents** or **mutagens**. Some known mutagens are mustard gas, formaldehyde, sulfur dioxide and some antibiotics. All kinds of ionising radiation, including ultraviolet light, X-rays, cosmic rays, radiation from radioactive waste, and fallout from atomic and nuclear explosions, are also mutagenic. If a woman is treated with large doses of X-rays during the first three months of her pregnancy, the child may be born with intellectual disability, skeletal malformations, or microcephaly (a condition where the head is small in relation to the rest of its body). For this reason, doctors try to avoid using X-rays early in pregnancy.

Activity 9.1
Investigating the effect of ultraviolet radiation on UV-sensitive and wild-type forms of the yeast *Saccharomyces cerevisiae*

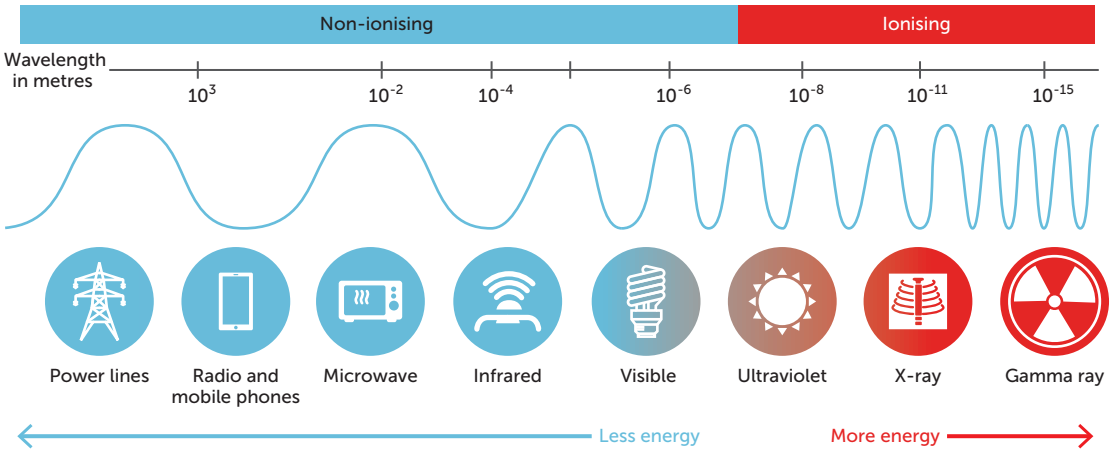

FIGURE 9.4 Ionising radiation is an example of a mutagen

Key concept

Mutations are changes in the DNA resulting in a variation in the associated trait. They may occur spontaneously, but are often due to exposure to a mutagen.

Types of mutations

DNA is composed of a double helix, each side of which is a long string of four types of nucleotides. Each nucleotide possesses identical sugar–phosphate groups that contribute to the DNA framework but differs in the base that links the two frameworks. Within genes, the sequence of the bases in the DNA is the code for the amino acids used to build a protein. Each group of three bases codes for an amino acid.

When it was recognised that genetic information is contained in the sequence of bases in the DNA, it became possible to understand the chemical nature of gene mutations. A change in the bases could change the amino acid and so could alter a protein. It is possible that a mutation could have no effect at all, or it may alter the protein or prevent it from being produced. Thus, if the DNA of a particular gene is altered, the protein for which it codes may be missing or abnormal. Just one missing or abnormal protein can have an enormous effect on the entire body.

Albinism, for instance, is the result of one missing protein. **Albinism** is marked by an absence of pigment from the hair, skin and eyes. The hair of a person with albinism tends to be whitish blond, the skin extremely pale and the eyes pinkish.

iStock.com/Vesnaandjic

FIGURE 9.5 Albinism is an inherited condition caused by a mutation that results in just one missing protein

Mutations can be classified by a number of different characteristics. It is the sum of these characteristics that determines the overall impact the mutation will have on the individual.

Cause of the mutation

You have already learnt that mutagens in the environment can increase the chance of mutations occurring. These mutations are known as **induced mutations**. Other mutations occur due to a random error in a biological process such as mitosis or meiosis. These are called **spontaneous mutations**.

Heritability of the mutation

One way of classifying mutations is by the type of cells where they occur, such as a person's body cells or reproductive cells. When the body cells, or somatic cells, are involved with a mutation, it is known as a **somatic mutation**. In this situation, only the individual with the somatic mutation is affected. Each time the mutant body cell divides, the mutation is passed on to the daughter cells. However, as the reproductive cells are not affected, once the individual dies the mutation is lost. Somatic mutations are involved in many cancerous growths that may be a result of a mutagenic agent.

If the reproductive cells are affected, the mutation can occur in the gametes and may then be passed on to the next, and subsequent, generations. These are known as **germinal** or **germline mutations**. In this case, the individual in whom the mutation occurs is not usually affected. However, that individual produces gametes with changed DNA. If conception occurs involving one of the affected gametes, the embryo is often naturally aborted early in the pregnancy. Diseases such as **phenylketonuria (PKU)** can arise through a mutation during the formation of gametes and can be passed on to offspring.

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FIGURE 9.6 Lead aprons are used to protect the cells, particularly gametes, during exposure to ionising radiation

Effect of the mutation

Another way that mutations can be classified is based on their effect.

- **Missense mutations** cause a change in the amino acid, and therefore in the protein produced.
- **Nonsense mutations** change the base sequence to the code to STOP. This means that the synthesis of the protein will stop, and so a shorter protein is produced that is unlikely to be able to fulfil its function.
- **Neutral mutations** cause a change in an amino acid; however, the amino acid is of the same type and does not change the structure of the protein enough to change its function.
- **Silent mutations** do not cause any change in the amino acid, and therefore in the protein produced. This is possible, as most amino acids are coded for by more than one base sequence.

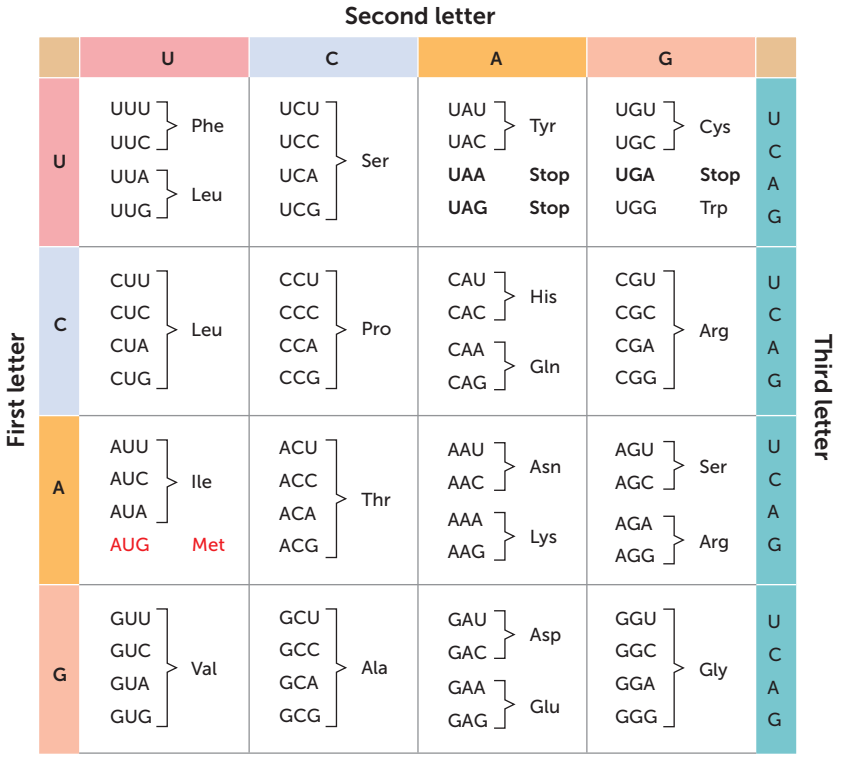

FIGURE 9.7 The codons that code for each amino acid. Note that the codon is the sequence of three bases on mRNA. This will be complementary to the base sequence on the DNA molecule

Extent of the mutation

Another characteristic that is used to classify mutations is the amount of DNA affected. This could range from a single base to a whole chromosome. A gene mutation affects only a single gene, while a chromosomal mutation affects a number of genes. It may change the structure of a chromosome or the number of chromosomes. Chromosomal mutations often cause abnormalities so severe that miscarriage often occurs early in the pregnancy.

Change in the DNA

Mutations vary in the change in the DNA. **Point mutations** are due to changes in a single nucleotide; therefore, only one base is changed. These mutations may be due to a nucleotide being:

- *inserted* – a new nucleotide is added to the DNA strand
- *substituted* – an existing nucleotide is replaced with another one, with a different base
- *deleted* – a nucleotide is removed from the DNA strand.

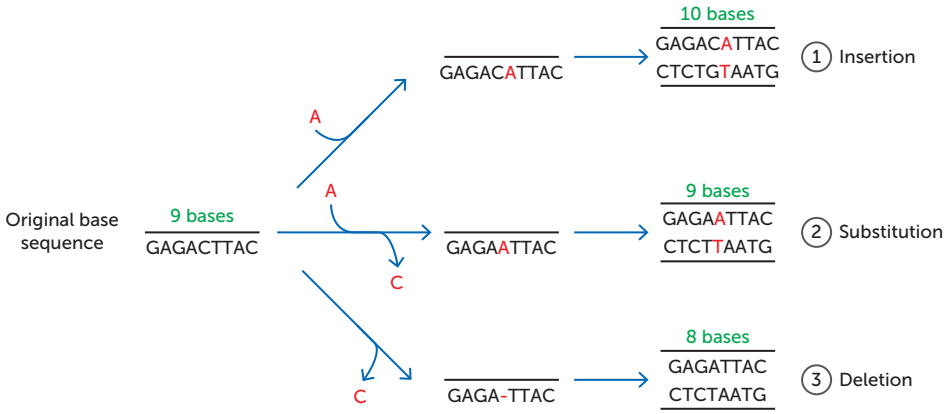

FIGURE 9.8 Insertion, substitution and deletion mutations

Some mutations will result in a frameshift. A **frameshift** occurs when bases have been added or removed. This results in the series of three bases that code for an amino acid starting at a different base. Therefore, although the mutation may have only altered a single base, frameshift mutations affect the outcome for all the DNA from that point on.

Frameshift mutations will not occur when three bases are added or deleted. In these instances, the DNA will simply code for one more, or one less, amino acid, but the rest of the amino acids will be the same. Therefore, it would still be a mutation, just not a frameshift mutation.

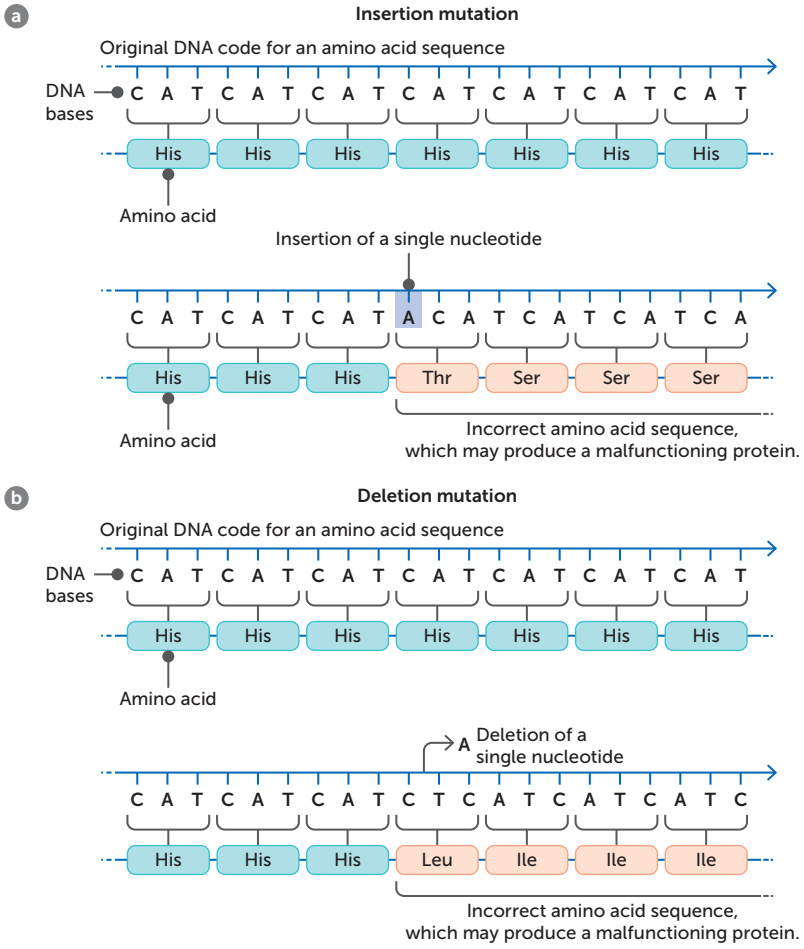

FIGURE 9.9 Frameshift mutations due to the **a** insertion and **b** deletion of a nucleotide

Other mutations affect a larger section of DNA. They may be due to:

- *duplication (or insertion)* – a section of chromosome occurs twice
- *deletion* – a piece of DNA is removed
- *inversion* – breaks occur in a chromosome and the broken piece joins back in, but the wrong way around
- *translocation* – part of a chromosome breaks off and is rejoined to the wrong chromosome
- *non-disjunction* – during meiosis, a chromosome pair does not separate and so one daughter cell has an extra chromosome and one daughter cell has one less than the normal number. These are sometimes referred to not as 'mutations', but as **aneuploidy** – a change in the chromosome number.

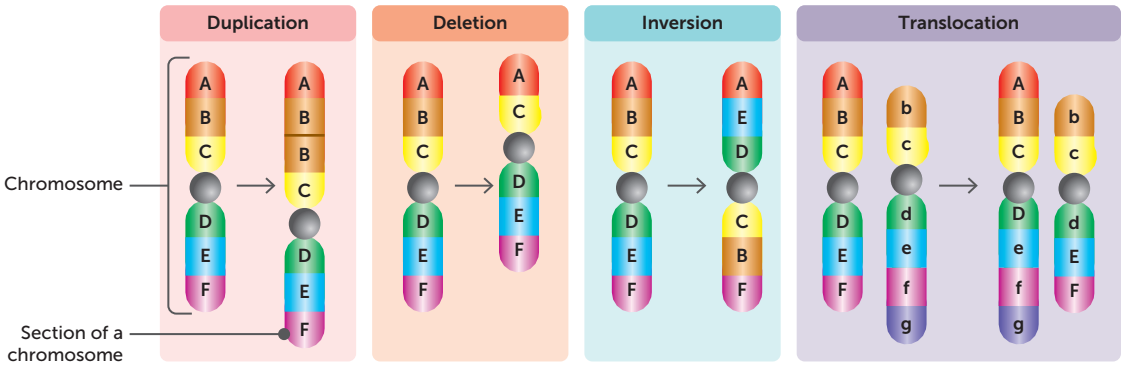

FIGURE 9.10 Mutations due to duplication, deletion, inversion and translocation of sections of DNA

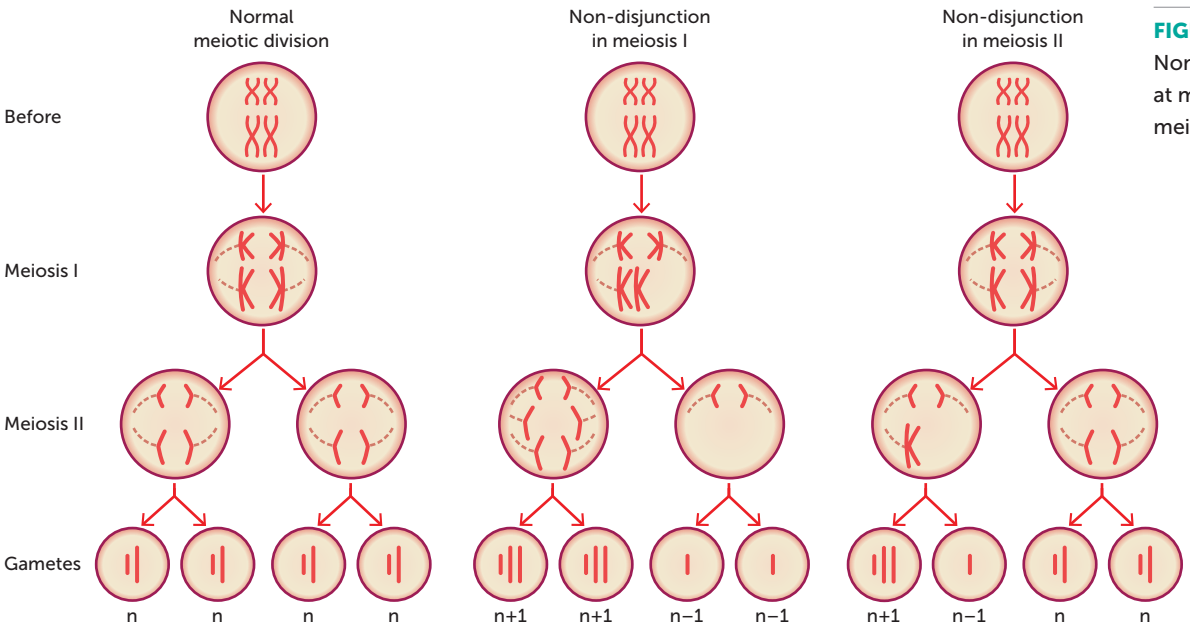

FIGURE 9.11 Non-disjunction at meiosis I and meiosis II

Key concept

Mutations can be classified based on their cause, where they occur, their effect, the amount of DNA they affect and the change in the DNA.

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Conditions due to mutations

Gene mutations

Duchenne muscular dystrophy may occur through gene mutation. This may arise through a mutation in the mother, which can then be inherited by her sons. The mutation may also occur in a male zygote so that the child develops the disease. This disease results in a wasting of the leg muscles and later the arms, shoulders and chest. Duchenne muscular dystrophy usually becomes apparent around the age of three to five years, when muscle weakness becomes evident. Eventually, death occurs due to failure of the respiratory muscles. Boys with the Duchenne form of muscular dystrophy are unlikely to live for more than 20–25 years.

Cystic fibrosis is another genetically determined disease caused by a mutation. The mutation occurs in a huge gene on chromosome number 7. The gene has the code for 1480 amino acids that make up a protein that regulates the passage of chloride ions across the cell membrane. Without the correct protein the affected person suffers from a variety of symptoms: salty-tasting skin; persistent coughing, wheezing or pneumonia; and digestive and other problems. The mutant allele is recessive, so a sufferer must inherit it from both parents.

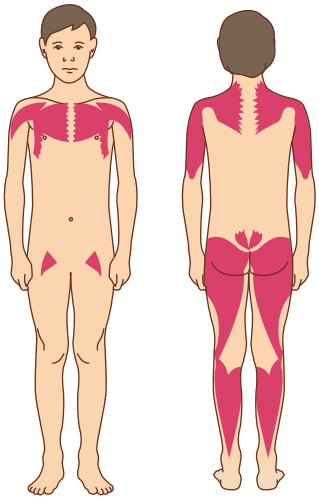

FIGURE 9.12 The muscles that are affected by Duchenne muscular dystrophy

Chromosomal mutations: Trisomy

Trisomy is a result of non-disjunction, failure of one or more chromatids to separate in the second division of meiosis. The eggs or sperm formed when non-disjunction occurs have one chromosome too many, or one chromosome is missing.

A chromosomal mutation that occurs relatively frequently, especially in children of older mothers, is **Down syndrome**, or **trisomy 21**, where the child has three of chromosome 21 instead of the normal two. People with Down syndrome have a characteristic facial expression, intellectual disability and weak muscles. They may also suffer from some birth defects such as heart defects or digestive abnormalities.

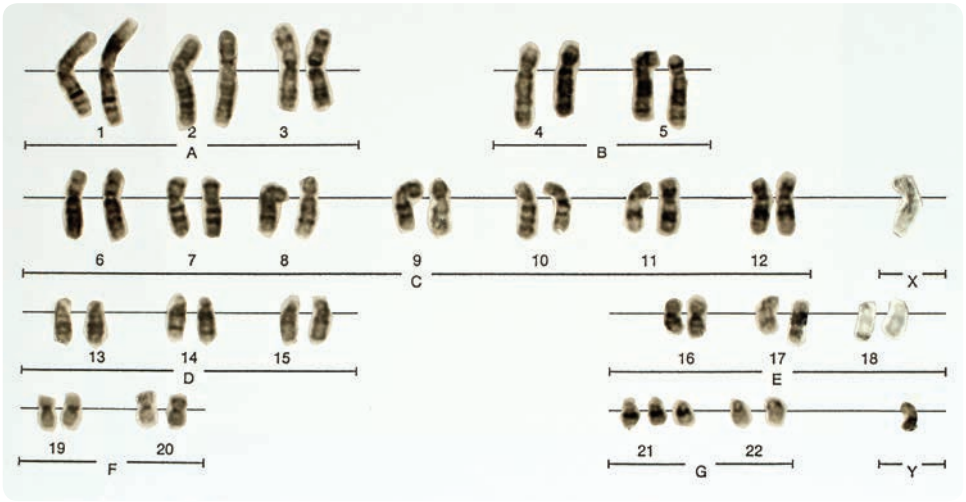

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FIGURE 9.13 Karyotype of Down syndrome (note the extra chromosome 21)

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FIGURE 9.14 Down syndrome children have a characteristic facial expression

Many of the symptoms of Down syndrome can also occur when part of an extra copy of chromosome 21 is attached to one of the other chromosomes. This is called partial trisomy.

Trisomy also occurs with other human chromosomes. **Patau syndrome** is when an extra chromosome 13 produces individuals with intellectual disability, microcephaly, an extra finger on each hand, a cleft palate and/or cleft lip, and malformations of the ears and eyes.

The extra chromosome 13 can come from either the mother's egg cell or the father's sperm cell. The features of trisomy 13 result from having this extra chromosome in each of the body's cells. Trisomy 13 occurs in about one out of every 5000 live births. However, more than 80% of children with trisomy 13 die within a month of birth.

Trisomy can also occur with the sex chromosomes. In males, non-disjunction may occur during either the first or the second meiotic division, producing individuals with either an extra X chromosome (*XXY*) or an extra Y chromosome (*XYY*). Individuals with trisomy *XXY* are normal as boys but develop **Klinefelter syndrome** as adults. They have small testes that do not produce sperm, the breasts are enlarged and body hair is sparse. Occasionally, the individual has an intellectual disability.

Chromosomal mutations: Monosomy

Monosomy is where an individual is missing a chromosome. If an autosome is completely missing, monosomy usually results in severe malformations and miscarriage. If only part of a chromosome is missing, it is referred to as **partial monosomy**. Part of the chromosome has two copies, but part has only one copy.

An example of partial monosomy is **Cri-du-chat syndrome** (from the French for 'cry of the cat'), a rare genetic disorder due to a missing portion of chromosome 5. The syndrome gets its name from the characteristic cry of infants born with the disorder. The infant sounds just like a meowing kitten, due to problems with the larynx and nervous system.

Monosomy can also occur with the sex chromosomes. Individuals with a chromosome set with only one *X* chromosome (monosomy *X*) suffer from **Turner syndrome**. These females are short in stature, lack secondary sexual characteristics and are infertile.

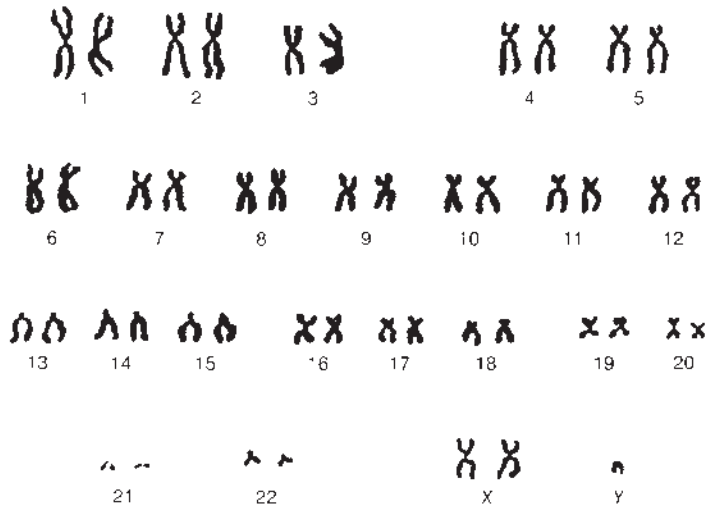

FIGURE 9.15 Karyotype for Klinefelter's syndrome

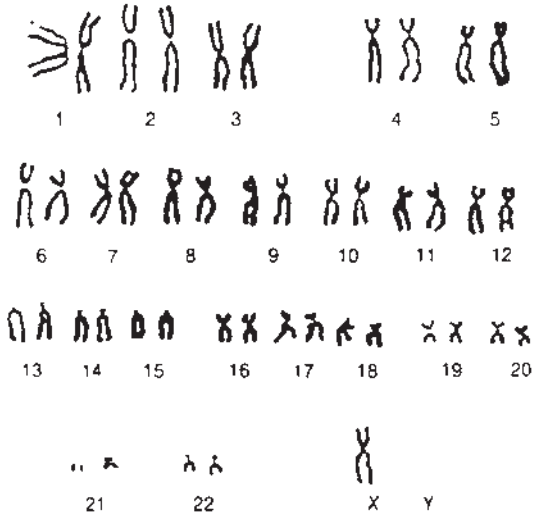

FIGURE 9.16 Karyotype for Turner syndrome

Lethal recessives

Most gene mutations produce a recessive allele because they prevent the gene from producing a protein that will be able to function in the body. A person could therefore have large numbers of mutations in the genes and be totally unaware of them. If the person reproduces with a partner who has the same recessive mutation, the recessive condition could appear in their offspring. This is what happens when couples unexpectedly have a child with cystic fibrosis.

Some recessive mutations are lethal if they are not masked by a dominant normal allele. These **lethal recessives** cause the death of the embryo or foetus by a miscarriage or spontaneous abortion, or the early death of the child.

It is easy to see how a lethal recessive mutation could cause changes in the composition of a gene pool. People who inherit two such alleles would die before their alleles could be passed on to the next generation, so the proportion of lethal recessive alleles in the gene pool would gradually be reduced.

Tay-Sachs disease (TSD) is a disorder of lipid metabolism that is inherited in an autosomal recessive pattern. It is a lethal recessive condition caused by a mutation in the HEXA gene that codes for the enzyme beta-hexosaminidase. This enzyme is responsible for breaking down toxic substances, including a fatty substance called GM2 ganglioside, in the brain and spinal cord. The missing enzyme results in the accumulation of GM2 ganglioside in the nervous system, which destroys the neurons. A baby with two recessive alleles for TSD develops normally for the first few months, and then deterioration that causes intellectual and physical disabilities begins. Death usually occurs in early childhood.

Examples of unbalanced chromosomal arrangements
This website provides more information on some chromosomal mutations.

WS
9.1 Mutations

Questions 9.1

RECALL KNOWLEDGE

1. Define 'mutagen' and list three examples.
2. Describe the effects of these different types of mutation:
   - **a** missense mutation
   - **b** nonsense mutation
   - **c** neutral mutation
   - **d** silent mutation.
3. Draw and label diagrams to demonstrate how an inversion changes the base sequence on DNA.
4. Explain why only germline mutations are passed on to the next generation.
5. List two conditions due to:
   - **a** gene mutations
   - **b** chromosomal mutations.

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6 Is Down syndrome an example of a gene mutation or a chromosomal mutation? Explain your answer.

APPLY KNOWLEDGE

7 Use a diagram to explain why the insertion of a single nucleotide can cause a frameshift mutation, resulting in a change in many amino acids.

8 The original base sequence of a small section of DNA is shown below.

C	C	T	A	G	T	C
G	G	A	T	C	A	G

Name the point mutation that has occurred in each of the following.

a

C	C	A	A	G	T	C
G	G	T	T	C	A	G

b

C	C	T	A	G	G	T	C
G	G	A	T	C	C	A	G

c

C	T	A	G	T	C
G	A	T	C	A	G

9 Use Tay-Sachs disease as an example to explain how a gene mutation can be lethal.

10 Some mutations result in a STOP codon, preventing any more amino acids being added to the chain. These are known as nonsense mutations. Figure 9.7 shows the base sequence on the mRNA, which is a copy of the coding strand of the DNA but containing the base uracil instead of thymine. Describe the mutation that could have occurred to each of the following original base sequences on the coding strand to result in a nonsense mutation.

a AAA
b TCGA
c TAT

9.2 MIGRATION

Changes in allele frequencies in a gene pool can also be due to gene flow brought about by migration. **Gene flow** is the movement of genetic material from one population to another. When individuals move between populations, they enable gene flow. This movement is known as **migration**. Therefore, if immigrants to a certain country bring alleles that are not already in the population, the frequencies for the alleles of that gene will be altered. This has occurred in China, for example. In the past, the Chinese population all had the Rh-positive blood group. The Rh, or Rhesus factor, is an antigen found on the surface of red blood cells. People with this antigen are referred to as Rh+; those who do not have the antigen are Rh−. When European countries began trading with China in the 16th century, European immigrants and sailors introduced the Rh− allele into the Chinese population. However, the frequency of the allele is still very low in China compared to other countries.

An example of how the distribution of ABO blood groups has been influenced by migration is the change in the frequency of the I^B allele across Europe and Asia. The inhabitants of East Asia, the Mongols, have a proportionately higher frequency of the allele I^B than those living to their west in Europe. In fact, it is thought that most Western Europeans originally did not have the I^B allele at all. In the 12th and 13th centuries, the Mongols invaded Europe on a number of occasions, spreading not only their culture but their genes as well. Today, there is a steady decrease in the I^B allele from Central Asia to Western Europe. Interestingly, the lowest concentrations of the I^B allele are now in the Pyrenees mountains and a few isolated locations in Scandinavia.

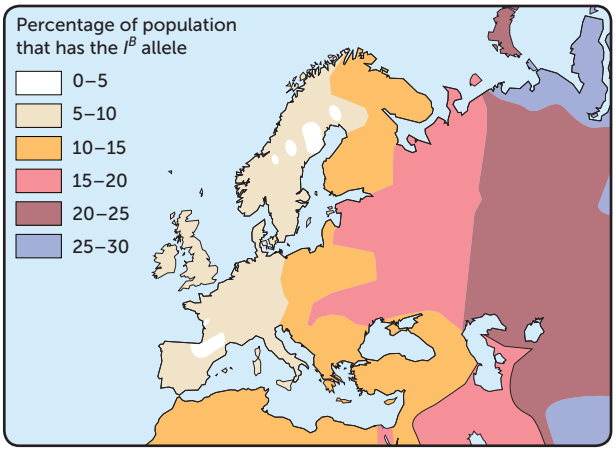


FIGURE 9.17 Distribution of the allele for the B blood group in Europe

Barriers to gene flow

Populations are often kept apart by barriers that inhibit the amount of interbreeding between them. This isolation leads to separate gene pools forming. Barriers to gene flow can be classified based on their cause.

- **Geographical barriers** include oceans, mountain ranges, large lake systems, deserts and expansive ice sheets. For example, the original inhabitants of Australia were isolated for thousands of years by ocean barriers that formed as sea levels rose.
- **Sociocultural barriers** such as economic status, educational background and social position are barriers to interbreeding. For example, statistics indicate that Australians tend to marry people of similar educational background, and members of particular religious groups favour partners who have the same faith. Religion and language can also be barriers to gene flow. Some religions do not allow marriages outside the religion, and it is unlikely that people who cannot communicate with one another will marry.

Key concept

Gene flow is the movement of genetic material from one population to another. It is facilitated by migration, but hindered by barriers such as geography and sociocultural factors.

Questions 9.2

RECALL KNOWLEDGE

1. Define 'gene flow'.
2. Describe how migration facilitates gene flow.
3. List four barriers to gene flow.
4. Describe how religion may be a barrier to gene flow.

APPLY KNOWLEDGE

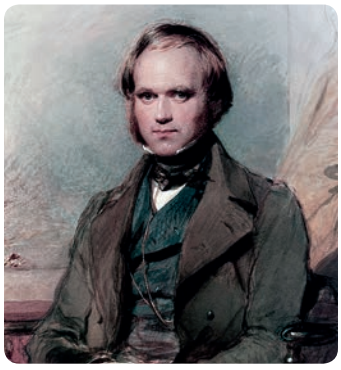
5. Explain why geographical barriers have less influence on gene flow in today's populations than in previous times.
6. Describe an example where barriers to gene flow would be:
   - a an advantage
   - b a disadvantage.

9.3 NATURAL SELECTION

Development of the theory of evolution

There are countless millions of species of plants, animals and micro-organisms living on Earth today. How has this multitude of species come into existence? Until the 1800s, it was widely believed that God, or a supreme being, had individually created each species. This is known as **special creation** and it is still the belief of members of some religious groups. Evolution is a gradual change in the characteristics of a species. The theory of evolution through natural selection was put forward independently by Charles Darwin and Alfred Russel Wallace in 1858. However, it is Darwin's name that is usually associated with this theory because of the massive amount of supporting evidence he collected.

Darwin was a keen amateur naturalist and as a young man he joined a surveying expedition as its biologist. He voyaged on HMS *Beagle*, visiting, among other places, the Galapagos Islands, New Zealand and Australia. This voyage, and the material Darwin



Alamy Stock Photo/FineArt

FIGURE 9.18 Charles Darwin. Together with Alfred Russel Wallace, Darwin put forward the theory of evolution through natural selection in 1858

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collected, was to be the preparation for all his later work. The Galapagos Islands were especially important for his research. On these islands, Darwin was able to observe the differences and similarities between animals separated by:

- geography – those living on the mainland of South America and those on the various islands
- time – animals recently extinct and species still alive.

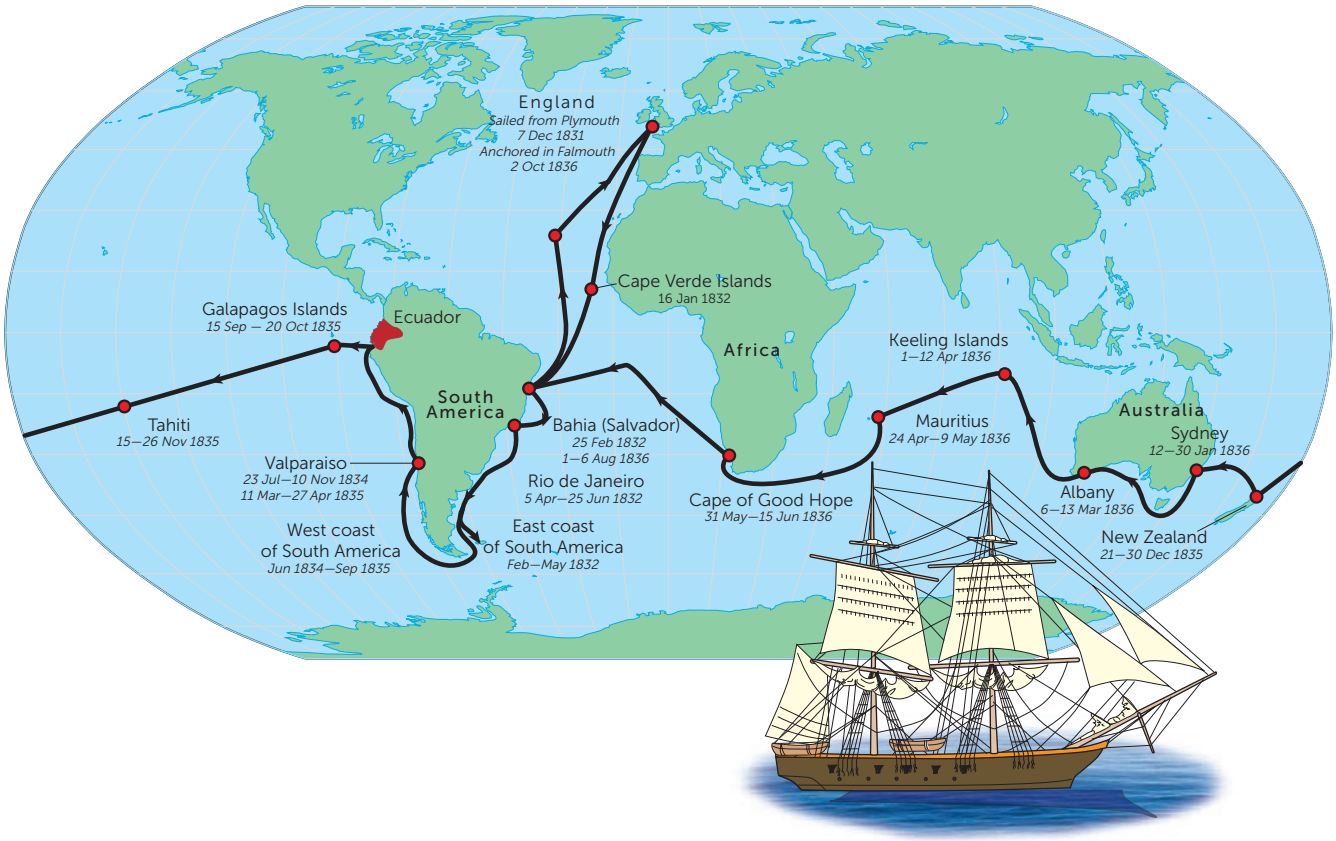

FIGURE 9.19 The route taken by HMS *Beagle* from December 1831 to October 1836. Darwin's observations on this journey were crucial to his later ideas on natural selection

These observations led Darwin to question the commonly held belief that living species had always been exactly the same as they then appeared. He became convinced that species did change. But how did the changes take place?

Darwin was greatly influenced by the works of other people. Carolus Linnaeus (1707–78) established the basis of our present system of classification and the binomial system of naming organisms using the generic (genus) and specific (species) names. This system was important to Darwin as it enabled him to classify and organise the material he collected.

Another major influence on Darwin was a book, *The Principles of Geology*, written by his friend Charles Lyell. Lyell hypothesised that the natural forces existing in the past were much the same as those existing in his own time. This hypothesis implied that Earth's surface had been gradually moulded over a very long period of time, by such simple forces as changes in temperature, running water and earth movements. Lyell's ideas provided Darwin with a concept of constant change against which he could view his own work.

Thomas Malthus, a British clergyman and political economist, provided the idea for the foundation of Darwin's theory of natural selection. Malthus, in 'An Essay on the Principle of Population', pointed out that the human population was increasing at a rate far exceeding the rate of food production. Drawing on examples from natural populations of plants and animals, he demonstrated that natural reproduction rates exceeded the available resources; that is, more plants

and animals are produced than can possibly survive. Darwin realised that under these circumstances a struggle for existence would occur, with favourable variations being preserved and unfavourable ones being gradually lost from the population.

In 1858, Darwin received a copy of an essay by Alfred Russel Wallace, a naturalist then on the island of Ternate in Indonesia (then the Dutch East Indies). Wallace's essay, 'On the Tendency of Varieties to Depart Indefinitely from the Original Type', covered the same ideas that Darwin had been working on. Darwin had been collecting evidence and refining his ideas for 20 years, but Wallace's essay was the stimulus for him to publish his views. A joint essay was prepared by Darwin and Wallace and read before the Linnean Society in 1858.

A year later, Darwin published his first book, *On the Origin of Species*. The book created a storm of controversy, but with the support of other scientists Darwin's ideas became firmly established.

Darwin's theory of natural selection

Darwin's theory of **natural selection** was based on three observations.

- *Variation:* Darwin noted that all members of a species vary. He made no attempt to explain the source of this variation. However, he did point out that these variations were passed on from one generation to the next, with characteristics displayed by the parents being passed on to their offspring.
- *Birth rate:* Inspired by Malthus, Darwin realised that all living organisms reproduce at a rate far greater than that at which their food supply and other resources increase. This would normally result in overcrowding.
- *Nature's balance:* Darwin observed that, although the birth rate of organisms was very high, each species' numbers tended to remain at a relatively constant level.

From these three observations, Darwin made a number of interpretations. First, he realised that, because of the excessive birth rate and limited resources, there must be a **struggle for existence**; and, second, because there was a range of variations in any species, those with characteristics best suited to their environment were more likely to survive. This second point became known as **survival of the fittest**: organisms with favourable characteristics survived, while many of those with unfavourable characteristics died before they had an opportunity to reproduce and pass on the trait.

Charles Darwin
This website provides an interesting perspective on Charles Darwin and the impact of his work.

Survival of the fittest is possible because there is **variation** within any species. That is, the members of a species differ from one another in their physical characteristics, body functioning and behaviour.

With knowledge of the mechanisms of inheritance, scientists building on the work of Darwin were able to explain the process of natural selection far more satisfactorily. Today, natural selection can be viewed as the selection of those alleles in a population that give an organism a greater survival advantage. The environmental factor acting on the population is known as the **selective agent**. Those organisms that survive will pass on favourable alleles to their offspring. Gradually, over a period of time, the characteristics of a population change so that it becomes better suited to its environment. In addition, where the environment is gradually changing, characteristics that enhance survival enable succeeding generations to gradually adapt to it. An important point to note is that individual organisms do not adapt. Instead, the species adapts to its environment by natural selection, and the process of adaptation takes many generations.

Alleles and natural selection

Natural selection can be looked at in terms of the frequencies of alleles in the gene pool of a population. If the environment tends to favour a particular characteristic, more of the alleles for that trait will be passed on to the next generation. This will result in a change in the frequency of that allele in the gene pool. Over time, that characteristic becomes more frequent in the population.

Activity 9.2
Venusians: investigating natural selection

The principles of evolution through natural selection can be summarised as follows:

- There is variation of characteristics within a species.
- More offspring of a species are produced than can possibly survive to maturity.
- Because of excessive birth rate and limited resources, there is a struggle for existence or competition for survival.
- Individuals with characteristics best suited to the environment have more chance of surviving and reproducing. This is known as survival of the fittest.
- Favourable characteristics are passed on to the next generation.
- In the gene pool, the proportion of alleles that produce favourable characteristics gradually increases.

Activity 9.3
Modelling natural selection

Key concept

Natural selection is the change in allele frequency in populations as a result of a selective agent.

Examples of natural selection

Body stature

Initially, the human gene pool would have contained alleles for a whole range of statures, from the short-bodied, long-limbed physique of present-day black Africans, to the long-bodied, short-limbed stature of the Inuit people of today. Individuals with long bodies and short limbs have a smaller surface area in relation to body volume than those with short bodies and long limbs. Such individuals lose less heat in very cold environments and would therefore have a survival advantage. When individuals of this type reproduced, they would have passed on the alleles for long bodies and short limbs to their children. They, too, would have had a survival advantage and would pass on the favourable alleles to their offspring. As fewer of the short-bodied, long-limbed individuals would survive in the extreme cold, fewer of the alleles for these characteristics would have been passed on. Many individuals with less-favourable characteristics would have died before reproductive age, so the frequency of unfavourable alleles in the gene pool would gradually decrease. Over time, those alleles would have decreased to such an extent that the unfavourable characteristics would no longer occur in the population. In this way, the frequency of alleles controlling body stature in the population would have changed. Those controlling long bodies and short limbs would have increased, while those for short bodies and long limbs would have decreased. Thus, evolution or genetic change has taken place. Within a particular gene pool, the frequencies of the alleles have changed over time.

a

Alamy Stock Photo/wanderluster

b

Getty Images/Mitch Kezar

FIGURE 9.20
a Short limbs and long bodies would have evolved in very cold climates, such as where Inuits live; **b** Long limbs and short bodies would have evolved in very hot climates, such as where Africans live

Sickle-cell anaemia

The incidence of **sickle-cell anaemia** in different parts of the world is another example of natural selection operating in human populations. The *Anopheles* mosquito, which transmits the malarial parasite, is not normally an inhabitant of tropical forests. It needs quiet, stagnant pools of water for breeding sites. This habitat is more often found in open areas. As humans began to clear the forests of Africa for agriculture, they changed the environment in a manner that created additional breeding areas for *Anopheles* mosquitoes. The increased food supply from agricultural production allowed the human population to increase, providing more bodies on which the mosquitoes could feed. Thus, the incidence of malaria increased. Figure 9.21 shows the distribution of malaria throughout the world.

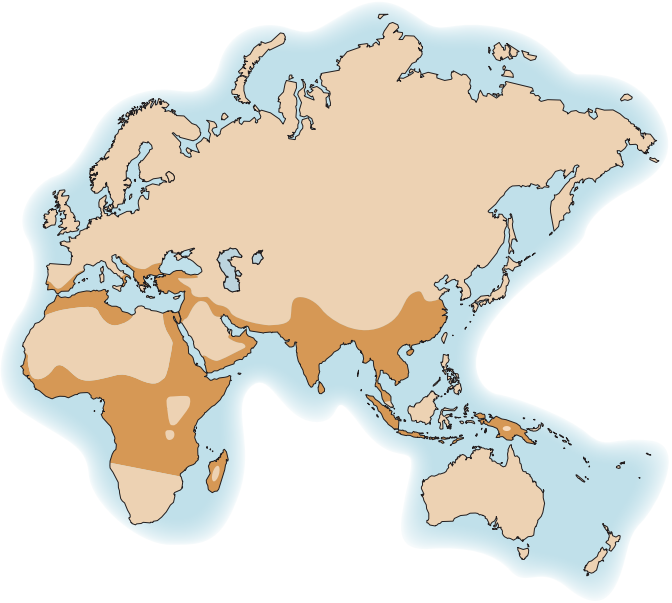

FIGURE 9.21 The distribution of malaria

In 1910, a young West Indian student living in Chicago visited his doctor with a variety of symptoms, including clogged blood vessels, pneumonia, rheumatism, heart disease, inflammation of the hands and feet, and anaemia. His doctor took a blood sample and observed it under a microscope. When air was excluded from the sample, the red blood cells showed a dramatic change in their shape from round to a crescent-like, or sickle, shape.

Subsequent investigation showed that sickle-cell disease, or sickle-cell anaemia, results when a person is homozygous for a particular recessive allele. We now know that this allele is due to a point mutation in the DNA sequence of the HBB gene. This gene codes for one of the beta-globulin proteins that make up haemoglobin in the red blood cells. The different base sequence means that the amino acid valine is added instead of glutamic acid. This results in a different form of the protein, altering the haemoglobin produced which distorts the shape of the red blood cell.

People who are heterozygotes normally show no ill effects unless oxygen is in short supply. When this occurs, their red blood cells show mild sickling. These individuals are carriers and are said to have sickle-cell trait. Individuals homozygous for the normal dominant allele have blood that shows no signs of the sickling phenomenon.

Science Photo Library/Eye of Science

FIGURE 9.22 The crescent shape of red blood cells of someone with sickle-cell anaemia

Sickle-cell anaemia
These websites provides more information on the changes to haemoglobin with sickle-cell anaemia.

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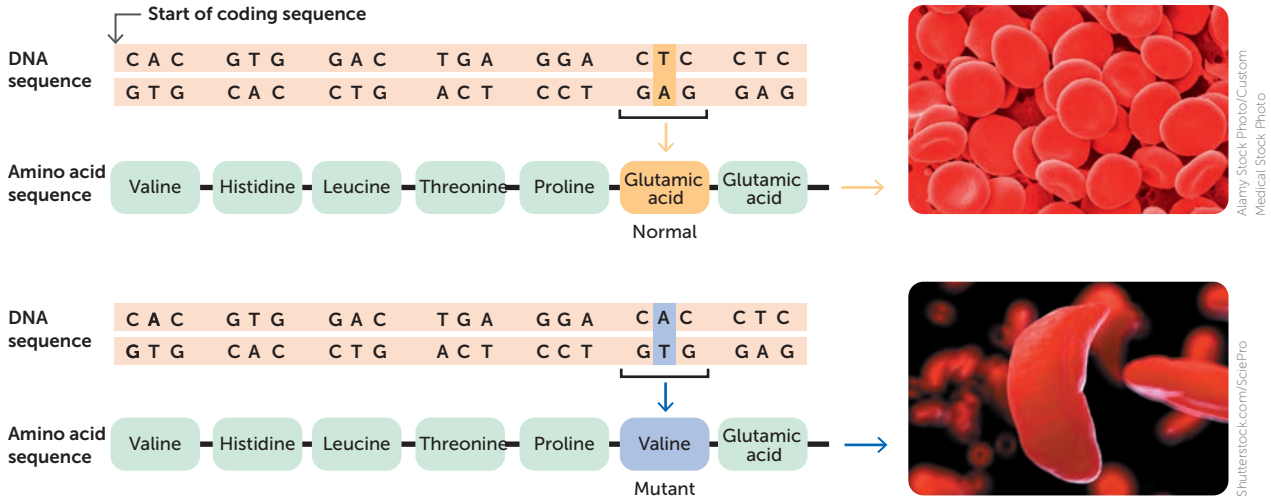

FIGURE 9.23 The point mutation of a substitution of one base pair results in sickle-cell anaemia

The sickle-shaped red blood cells often die early, resulting in **anaemia**. They are also inflexible and can become stuck in the blood vessels, causing a blockage. Other complications of sickle-cell anaemia include fatigue, jaundice, organ damage (such as to the kidneys, lungs and brain), high blood pressure and heart failure.

If a person with sickle-cell anaemia dies before reproducing, the allele that causes the disease is not passed on to the next generation. Therefore, you would expect that over many generations the frequency of the sickle-cell allele would gradually decrease until it was eliminated from the population altogether. On the other hand, if the ratio of mutation of normal alleles to sickle-cell alleles was great enough, it could cancel out the loss of alleles through the death of affected individuals. However, this is not the case: investigations have shown that the rate of alleles being lost from the population is about 100 times greater than the average rate of mutation at any point along a human chromosome. Some other mechanism must be at work to maintain the sickle-cell allele in the population. Figure 9.24 shows places in the world where the sickle-cell allele occurs in the population. When this is compared with Figure 9.21, you can see that the sickle-cell allele occurs only in areas where malaria is prevalent.

FIGURE 9.24 The distribution of sickle-cell anaemia

Anthony Allison was one of the first to notice the relationship between sickle-cell anaemia and malaria. He reported his observations in the *British Medical Journal* in 1954, noting that the sickling allele tended to have its highest frequency in areas where the risk from malarial parasites was greatest. He suggested that individuals with one sickle-cell allele were more resistant to malaria than those with normal haemoglobin in their red blood cells. This conclusion was based on Allison's observations that malarial patients who were also 'sicklers' had fewer malarial parasites than did malarial patients who were 'non-sicklers'. To gather evidence in support of these observations, Allison conducted a number of experiments. He inoculated both sicklers and non-sicklers with malaria and then treated those individuals in whom the disease developed. His results confirmed that the heterozygotes were less susceptible to infection from

malaria than individuals homozygous for normal haemoglobin. Further studies since Allison's pioneering experiments have supported his findings. It is now generally accepted that individuals heterozygous for the sickle-cell allele have a survival advantage in areas where malaria is prevalent.

The sickle-cell example shows how natural selection occurs in human populations. A mutation established a new allele in the population. Having one of these alleles gave individuals living in malarial-prone areas a survival advantage. This is known as a **heterozygote advantage**. Individuals with two sickle-cell alleles, those with sickle-cell anaemia, usually die. Those who are homozygous for normal haemoglobin are more susceptible to malaria. Therefore, the presence of malaria acted as a selective agent for the sickle-cell allele.

Activity 9.4
Investigating sickle-cell haemoglobin

Tay-Sachs disease

In section 9.1 you learnt that Tay-Sachs disease is a lethal condition caused by a mutation that results in the absence of the enzyme beta-hexosaminidase. This condition leads to a deterioration of the nervous system, and death usually occurs at a young age.

Tay-Sachs disease is a recessive condition found only in individuals who are homozygous recessive. People who are heterozygous have a reduced amount of beta-hexosaminidase. It appears that these individuals have some protection from tuberculosis, an infectious disease that primarily affects the lungs. This means that people who are:

- homozygous recessive die before reproducing, due to Tay-Sachs, and therefore do not pass on the recessive allele
- heterozygous survive tuberculosis, reproduce and therefore pass on both alleles
- homozygous dominant are affected by tuberculosis and may die prior to reproducing.

The heterozygote advantage provided by a heterozygous genotype increases the percentage of the recessive allele in the gene pools in areas affected by tuberculosis.

Thalassemia

Haemoglobin is made up of four protein chains that fit together. Two of these are alpha globin chains and the other two are beta globin chains. You have already learnt that sickle-cell anaemia affects haemoglobin molecules. Another disorder that alters the structure of haemoglobin is thalassemia. There are two forms of thalassemia.

- Alpha thalassemia is due to a mutation in the HBA gene on chromosome 16. This reduces the level of alpha globin in haemoglobin.
- Beta thalassemia is due to a mutation in the HBB gene on chromosome 11. This reduces the level of beta globin in haemoglobin.

Both of these conditions are inherited in an autosomal recessive manner. People with thalassemia have less haemoglobin in their red blood cells and, therefore, cannot carry as much oxygen in their blood. The severity of the disorder varies, depending on the number of affected genes, ranging from mild anaemia and fatigue to an enlarged liver and heart.

Thalassemia is more common in areas affected by malaria. Alpha thalassemia is more prevalent in South-east Asia, while beta thalassemia is more prevalent in the Mediterranean basin. Research has shown that malaria can act as a selective agent, resulting in an increased frequency of the alleles of alpha thalassemia. It is thought that the lower amount of haemoglobin gives some protection against malaria. Patients also seem to recover more quickly than those without thalassemia, possibly due to the increased number of red blood cells. It is possible that this is also the case with beta thalassemia, however, this has not been supported by research. Therefore, other factors may also influence the allele frequency. These include migration, genetic drift and founder effect.

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Questions 9.3

RECALL KNOWLEDGE

1 Define 'natural selection'.
2 Describe the phenotype of individuals who have each of the following genotypes for sickle-cell anaemia:
   a homozygous b heterozygous.
3 List the steps involved in evolution through natural selection.
4 Describe what a selective agent is, and give an example.

APPLY KNOWLEDGE

5 Explain why variation is crucial for natural selection.
6 Use a flow chart to summarise the history of our understanding of natural selection.
7 Describe how natural selection may have led to an increased occurrence of Tay-Sachs disease in areas where tuberculosis occurs.
8 Suggest the selective agent for each of the following characteristics.
   a Bacteria becomes resistant to antibiotics.
   b Inuit people tend to be short limbed and long bodied.
   c Prickly pear cacti have thorns on their flesh.
9 Explain how malaria can lead to an increased frequency of the allele for sickle-cell anaemia.
10 Use a Venn diagram to compare and contrast alpha thalassemia and sickle-cell anaemia.

9.4 GENETIC DRIFT

In any generation there is always the chance that some individuals will be more, or less, likely to pass on their alleles. This produces **genetic drift**, the random, non-directional change in allele frequency between generations. Genetic drift is not affected by whether an allele is beneficial or harmful; instead, it is purely by chance. Genetic drift occurs in populations of all sizes; however, it is unlikely to have a significant effect in large populations. It can, however, play an important role in evolution in small populations.

Genetic drift is also known as **random genetic drift** or the **Sewall Wright effect**, after the man who first recognised its significance in causing changes to allele frequencies. It is much the same as if you had 50 red balls and 50 black ones, with each ball able to reproduce itself periodically. If these 100 balls were placed in a bag and 50 balls were selected at random from it, the expected result would be 25 balls of each colour. It would not be surprising, though, to find that your sample contained 30 black balls and only 20 red ones. In this case, after

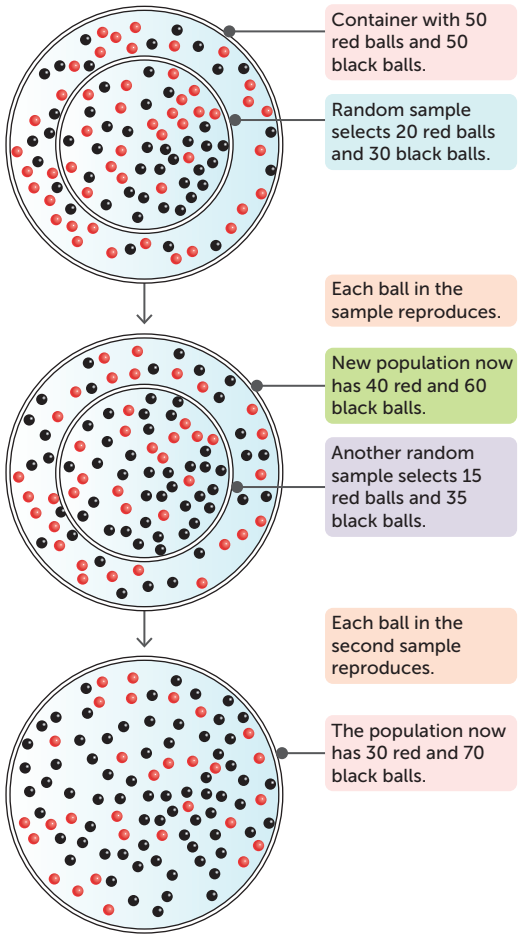

FIGURE 9.25
A model showing how random sampling of a pair of alleles can, by chance, increase the frequency of one allele and decrease the frequency of the other

reproducing, your new population would have 60 black balls and 40 red ones. If 50 balls were again selected at random, you would now expect the black ones to be favoured. This is how random genetic drift works in small human populations.

Studies have been carried out on isolated populations of Aboriginal Australians. One study investigated the isolated populations of Bentinck and Mornington Islands in the Gulf of Carpentaria. Originally these islands were part of the mainland, but rising sea levels cut them off and their populations became isolated. However, the Mornington Islanders maintained some contact with the mainland by using the smaller islands in between as 'stepping stones'. The blood group frequencies of the islanders have been studied and compared with those of the population occupying Bayley Point on the mainland. The occupants of Bentinck Island show allele frequency values for blood groups that fall outside the range for Aboriginal people in the rest of Australia. They show a very high proportion of the I^B allele and a complete absence of the I^A allele, unlike the mainland population, which has a low proportion of the I^B allele and a relatively high proportion of the I^A allele.

Key concept

Genetic drift is the random, non-directional change in allele frequency that occurs by chance. It is particularly significant in small populations.

Founder effect

An extreme example of genetic drift is the **founder effect**. This effect occurs when a small group moves away from its homeland to a totally new area and establishes a population, which later expands. Because of its small size, chance can cause the new groups to have:

- a different allele frequency from the original population
- decreased genetic variation.

This means that the new population may show a frequency of features that are not typical of the original homeland population.

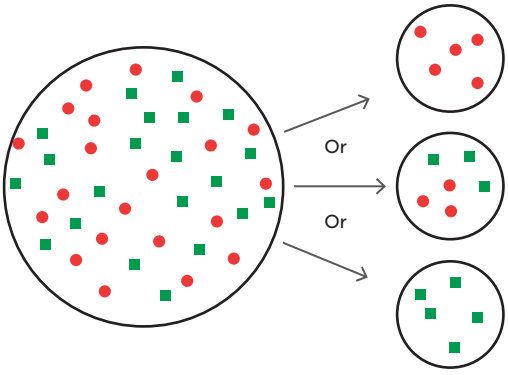

FIGURE 9.26
A model illustrating the founder effect. The original population is on the left and three different possible founding populations are on the right. It is possible for the founders to be quite unrepresentative of the original population. The chance selection of the founders will have a marked effect on the gene pool of later populations

Studies have been done on isolated groups to demonstrate this effect. One early and well-known study was by Bentley Glass and his co-workers in the 1950s on an isolated population in the United States. This group, known as the 'Dunkers', lives in Pennsylvania but originally came from Hesse, Germany. They are descended from Old German Baptist Brethren who came to the United States in the early 18th century. Their religion does not allow them to marry outside their group, and thus they constitute an isolated breeding population within the total population of the United States. The study investigated a number of easily measured physical traits, including the frequency of the ABO, Rh and MN blood groups, mid-digital hair, left- or right-handedness, and attached or free earlobes. For most of the traits studied, the Dunkers varied in allele frequency from the present-day population of Hesse and also from the surrounding American population. The environment for both the Dunkers and the surrounding American population is essentially the same, so there would not have been any natural selection to account for the differences in allele frequencies. Therefore, Bentley Glass concluded that genetic drift was responsible for this variation as the small size of the Dunker population allowed certain characteristics to become more common purely by chance.

Founder effect
This website provides more information about the founder effect.

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iStock.com/golibo

FIGURE 9.27
Left-handedness was one of the physical traits observed by Bentley Glass in his study of the Dunkers

Another example of the founder effect is the incidence of Tay-Sachs disease in the population of Ashkenazi Jews. Earlier in this chapter, Tay-Sachs disease was discussed as an example of a lethal recessive disease. Approximately 1 in 27 Ashkenazi Jews carries the altered allele, compared to 1 in 300 in non-Ashkenazi Jews. Ashkenazi Jews descended from a small number of individuals from Central or Eastern Europe. This group was initially isolated geographically. There was additional isolation through the custom of endogamy (only marrying within the community). This created a founder effect. The incidence of the mutated allele for Tay-Sachs disease was higher in the ancestors, and hence genetic drift is responsible for the prevalence of the disease in the current Ashkenazi Jew population.

The five most common Ashkenazi genetic diseases
This website has more information about the prevalence of disease in Ashkenazi Jews.

Genetic drift simulation
Use the simulation on this website to investigate the founder effect and the effect of a bottleneck.

Bottleneck effect

The **bottleneck effect** is another extreme example of genetic drift. In this situation, an event such as a natural disaster severely reduces the size of the population. The allele frequency after the disaster may, by chance, be different from before the event. It is important to note that, in the bottleneck effect, the chance of survival is by chance and not due to a specific trait.

An example of the bottleneck effect occurred in 1775 when a typhoon reduced the population of Pingelap, an island in Micronesia, to only 20. These survivors formed the founding population for the current inhabitants. Interestingly, among the survivors was a person heterozygous for achromatopsia. **Achromatopsia** is an inherited form of total colour blindness. The allele for achromatopsia is recessive. Today, after a number of generations, the incidence of achromatopsia on Pingelap is 5% of the population. In other parts of the world it is 0.0033%. Furthermore, 30% of the Pingelap population are carriers; they are not colour blind but they do have the affected allele. This is another example of how allele frequencies can change in small, atypical populations.

Key concept

The founder effect and bottleneck effect are two extreme examples of genetic drift that occur when a small group moves away from the main group or an event leaves only a small number of survivors.

Questions 9.4

RECALL KNOWLEDGE

1 Define:
   - **a** genetic drift
   - **b** founder effect
   - **c** bottleneck effect.

2 Explain how genetic drift is different from natural selection.

3 Use an example to explain how the founder effect may result in a population having different characteristics from another population of the same species.

APPLY KNOWLEDGE

4 Explain why genetic drift is unlikely to have a significant effect on the allele frequency of a large population.

5 Explain how it is possible that bushfires that affected areas of Victoria and New South Wales in early 2020 may reduce the genetic variation in koalas.

9.5 SPECIATION

All humans, from whatever part of the world and whatever ethnic background, have basic similarities and are capable of interbreeding to produce fertile offspring. That is, all humans belong to the same species. A **species** is a group of individuals that share many characteristics and are able to interbreed under natural conditions to produce fertile offspring.

Earlier in this chapter, isolation was mentioned as a barrier to gene flow. Reproductive isolation may lead to the development of separate gene pools. No two environments are exactly the same, so it would be expected that certain alleles would be favoured in one environment more than another. Therefore, over time the allele frequencies of each gene pool will change, depending on which characteristics are favoured for survival. Over many generations, the populations will become less and less alike as they develop characteristics that better suit them to their respective environments.

Speciation
This website provides an animation about the mechanisms of speciation.

If two populations are isolated for a very long period of time, and the environmental influences on each are different enough, major changes in the allele frequencies within each population could occur. In such a situation, the members of those populations may become so different that, even if the barriers to reproduction were removed, interbreeding would no longer be possible. If this occurred, the two populations would be regarded as separate species. The process of producing two species in this way is referred to as **speciation**.

The steps involved in speciation can be summarised as follows:

1 *Variation:* There is variation between individuals of a species.
2 *Isolation:* Populations of the same species are isolated without gene flow.
3 *Selection:* Each population is subjected to different selective agents.
4 *Speciation:* The allele frequency changes until they become so different that the two groups are no longer able to interbreed.

9.2 Evolutionary mechanisms

Key concept

Speciation, or the formation of new species, occurs due to variation, isolation and selection leading to two groups becoming so different that they can no longer interbreed.

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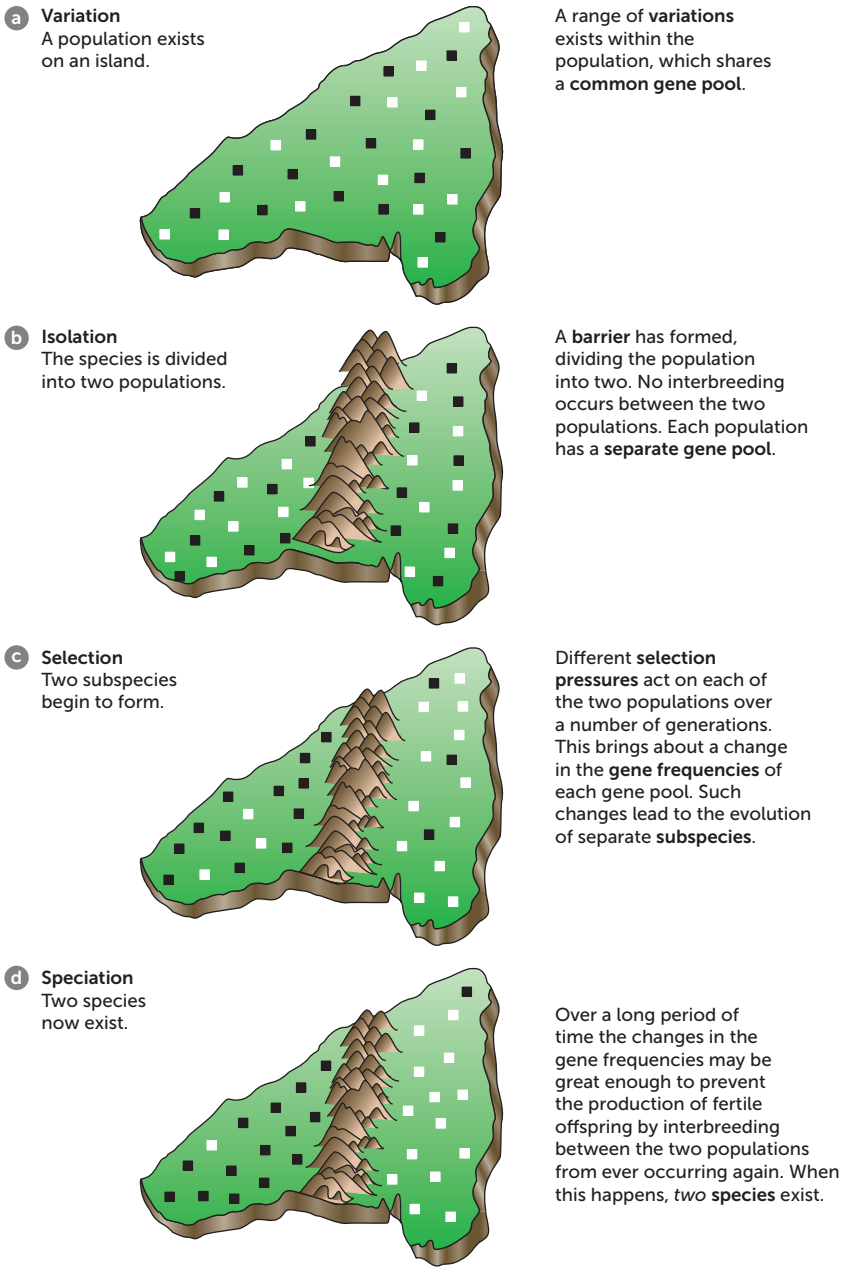

FIGURE 9.28
A diagrammatic representation of variation, isolation, selection and speciation

Questions 9.5

RECALL KNOWLEDGE

1 Define 'speciation'.
2 List the steps involved in speciation.
3 List three situations that may lead to isolation of groups.

APPLY KNOWLEDGE

4 Explain why variation is necessary for speciation.
5 Do you think it is likely that humans will form a new species in the future? Explain your answer.
6 Is mutation, natural selection or genetic drift the most important process in speciation? Justify your answer.
7 Would two groups of a species that are isolated in environments that are similar form new species? Explain why or why not.

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CHAPTER 9 ACTIVITIES

SOUTHERN Biological Developed by Southern Biological

ACTIVITY 9.1 Investigating the effect of ultraviolet radiation on *Saccharomyces cerevisiae*

We classify the broad spectrum of electromagnetic radiation from the sun into segments according to the effects we experience. For example, the warm sensation of sunshine on our skin is caused by invisible infrared radiation with wavelengths ranging from 700 nm to 1 000 000 nm (1 mm). Visible light is composed of wavelengths of between 400 nm (violet) and 700 nm (red). Radiation with wavelengths shorter than 400 nm but longer than 10 nm is classified as ultraviolet (UV) radiation. Radiation with wavelengths shorter than 10 nm is classified as X-rays. Some exposure to UV radiation is necessary for humans to produce vitamin D, but a careful balance is required because X-rays and UV radiation are destructive to many biological molecules, including DNA. Fortunately, the earth's atmosphere acts as a protective screen and filters out almost all the sun's radiation with wavelengths shorter than 290 nm. Nevertheless, the narrow UV band from 290 nm to 400 nm that can penetrate the atmosphere and reach the surface of the earth is capable of causing photochemical damage to DNA that can lead to skin cancer, so it is important to avoid over-exposure. As a defence against too much UV exposure, most organisms that are subject to the sun's rays have evolved to incorporate some level of DNA repair in their cell mechanisms. This confers a limited amount of inherent UV resistance.

Aim

To determine how ultraviolet radiation can be destructive for many biological molecules
Time requirement: 50 minutes

You will need

UV-sensitive yeast *Saccharomyces cerevisiae* starter plate; wild-type yeast starter plate; 8 sterile swabs; 8 YED agar plates; 4 plastic pipettes; 2 sterile culture tubes; Bunsen burner; permanent marker; sterile water; 2 sterile inoculation loops; ethanol or bleach; disposable gloves

Risks

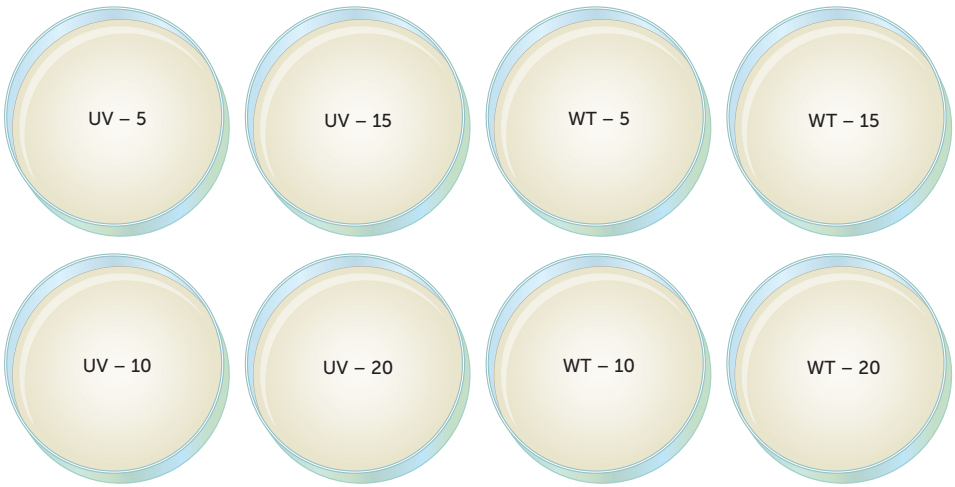
WHAT ARE THE RISKS IN THIS INVESTIGATION?	HOW CAN YOU MANAGE THESE RISKS TO STAY SAFE?
While lab strains are usually harmless, fungi may cause disease, so assume them to be pathogenic.	Wear lab coats, safety glasses and gloves; wash hands thoroughly at end. Decontaminate benches before and after activity. Flood spills with bleach.
Micro-organisms will grow on the agar plates.	Do not open plates once they are securely taped. Dispose of plates appropriately after autoclaving.
Disposable gloves may pose an allergy risk.	Use a type of glove that removes allergy risk and is suitable for the chemicals being used.

What to do

Note: To use aseptic technique, wipe your bench down with ethanol (or bleach) and keep your work near the Bunsen burner to take advantage of the updraught the flame will create to waft potential contaminants away from your materials.

Inoculation of exposure plates

1 Collect eight YED agar plates and label them as follows using a permanent marker.



Key:
UV = UV-sensitive yeast (mutated strain)
WT = wild-type yeast
Number = Time plate will be exposed to sunlight

2 Using a plastic pipette, add 1 mL of sterile water into a sterile culture tube.
3 Using a sterile inoculation loop, carefully scrape a single colony of the UV-sensitive yeast from the starter plate.
4 Select a large colony (>4 mm in diameter) or, if the colonies are small, scrape up two, or even three, on to the loop.
5 Place the loop in the water in the sterile tube and spin/swirl it to transfer the yeast into the sterile water.
6 Visually check that the cell mass has transferred from the loop to the sterile water.
7 Using a plastic pipette, immediately pump the liquid to distribute and suspend the yeast cells in the water. To do this, draw the liquid in and out of the pipette by squeezing and releasing the bulb of the pipette. Avoid introducing air bubbles or splashing the liquid up the sides of the tube. When finished, hold the tube up to the light to check that there are no visible lumps or particles in the water.
8 Dip a sterile swab into the yeast suspension and, as you withdraw it, press it against the sides of the tube to squeeze out excess water. It should come out moist but not dripping.
9 Using aseptic technique (see note above), 'swab' the surface of a YED plate in three directions to inoculate for a lawn culture, which is a culture that covers the entire plate as evenly as possible.
10 Immediately cover the plate to shield it from light and allow it to rest (right way up—that is, with the agar at the bottom) for a period of at least 15 minutes and up to one hour. This allows the moisture from the swab to be absorbed by the agar.
11 Repeat steps 8 to 10 for the remaining three UV-sensitive yeast plates. Then repeat this procedure for the four plates using the wild-type yeast.

Exposure to sunlight

1. State your hypothesis.
2. After the post-inoculation resting period, expose the one inoculated plate from each strain to direct sunlight for 5, 10, 15 and 20 minutes, respectively.
3. Immediately after exposure, incubate the plates in darkness for 48 hours at 30°C or 4 days at room temperature. For best results, follow these guidelines:
   - Keep the plate shielded from light until the last moment.
   - Use adhesive tape to attach the lid of the Petri dish to the base, but do not allow the tape to extend on to the surface of the lid where it will absorb UV light and shield the yeast from exposure.
   - Orient the plate so the lid is pointing directly at the sun. Aim to minimise the size of the shadow. If the sun's rays strike the lid at a glancing angle, most of the UV light will be reflected and the effectiveness of the exposure will be reduced.
   - Schedule the investigation at a time of year when you can be sure of bright, sunny conditions.
4. After the incubation period, observe and compare the level of coverage between the plates. Record your results in the table below.

Studying your results

1. Copy and complete the table below with the results of your experiment. Use the key below to indicate the level of coverage of the yeast on each agar plate.

+++	High coverage
++	Medium coverage
+	Low coverage
-	No coverage

UV exposure results

EXPOSURE TIME (MINUTES)	UV-SENSITIVE YEAST COVERAGE	WILD-TYPE YEAST COVERAGE
0		
5		
10		
15		
20		

2. Compare the results of your UV-sensitive yeast sample with the wild-type yeast sample. What differences do you observe? What conclusions can you draw from this data?
3. Graph your results.

Discussion

1. What is your independent variable?
2. What is the range of your independent variable?
3. What is your dependent variable?
4. What are your control variables and how did you control them?
5. What type of mutation does the UV-sensitive yeast portray?
6. Compare your results with others in your class. Were the results consistent?
7. Did your experiment support or refute your hypothesis, or were your results inconclusive?
8. Based on your findings, how does UV light impact the two different yeast strains? Do they differ? If they do, explain why.
9. Suggest how your findings might relate to evolution.

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Taking it further

To protect our skin from harmful UV rays, we apply different sunscreens with different sun protection factor (SPF) values. Do these values have any merit, and do commercially produced sunscreens offer better protection than alternatives such as coconut oil, clothing material and sunglass lenses?

ACTIVITY 9.2 Venusians: Investigating natural selection

Venusians are an imaginary group of people from the planet Venus. Because of the intense heat, their skin is jet-black; all individuals are homozygous for skin colour. If a mutation occurs resulting in a Venusian of a lighter skin colour, the individual usually dies before being able to reproduce. However, one such mutation created a brown-skinned individual who did survive and reproduced, passing the new allele to some of his children. The skin of the individuals affected by this mutant allele was extremely thick, providing them with added resistance to a lethal biting insect.

As time went by, the number of Venusians with the mutant allele increased. However, when two of these individuals produced children, homozygotes died in infancy.

In this activity, we will investigate how the mutant allele becomes distributed through the population over time. To simplify our activity, we will start with heterozygotes, and assume that all those who are homozygous for the mutant allele die before they can reproduce. We will also assume that one out of every three Venusians who are homozygous for the normal allele dies from a lethal insect bite.

You will need (for each pair)

Two containers: 2 L ice-cream containers work well; 20 black beads or counters to simulate the black skin allele (B) in each gamete; 20 white beads or counters to simulate the brown skin allele (b) in each gamete; felt pen; tally sheet; pencil

What to do

1. Label one container 'Male Parent' and the other 'Female Parent'.
2. Place 10 of the black beads in each container, then add 10 of the white beads.
3. Prepare a tally sheet similar to the one below using the symbol 'B' for the black skin allele and 'b' for the brown skin allele.

	GENOTYPES IN THE VENUSIAN OFFSPRING		
	BB	Bb	bb
1			
2			
3			
4			
5			
6			
7			
8			
9			
10			

4. Simulate reproduction by shaking the containers well and drawing out one bead from each. The beads represent gametes. Use the colour of the beads to determine the genotypes of the offspring. Place a tick in the relevant box on the tally sheet, then replace the beads.
5. When you have completed 10 draws, place the beads back into the containers. Your partner should repeat steps 1 to 4 above. Together you should have two completed tally sheets.

Studying your data

1. Because the first column contains individuals that are homozygous for black skin ('BB'), only two out of every three survive to adulthood. Tally the number of offspring that will survive to produce the next generation.
2. Individuals with the genotype 'bb' will all die in the first year. If you eliminate these individuals, how many surviving offspring do you now have?
3. What is the ratio of black skin to brown skin offspring that survive to adulthood?
4. Combine your data with the other groups in your class to obtain a bigger sample. What is the ratio now?

Interpreting your data

1. How has this activity shown that mutations that increase an individual's chances of survival and reproduction affect the proportions of particular characteristics in a population?
2. What has happened to the proportion of the allele 'b' in the population? Has it been entirely eliminated? Do you think it ever will be?
3. Summarise how this chance mutation has helped the survival of the Venusian population.

ACTIVITY 9.3 Modelling natural selection

In this activity, you will model the effects of natural selection on a hypothetical population of frogs. The frogs are naturally green, yellow or orange in colour, and are preyed upon by water birds. You will simulate the different predation rates on the three variations of frog colour over a number of generations by throwing a die.

You will need (for each group)

Coloured cards or counters – 30 green, 30 yellow and 30 orange; a die

What to do

1. Draw up a table similar to the following one to record your results.

Number of different coloured frogs in successive generations

GENERATION NUMBER	GREEN FROGS	YELLOW FROGS	ORANGE FROGS
1	10	10	10
2			
3			
4			
5			
6			
7			
8			
9			
10			

2. From the pool of coloured cards, select 10 of each colour. These will be your first generation of frogs. Shuffle these cards so that they are well sorted, and then deal them out in pairs. You should have 15 pairs of cards representing 15 pairs of frogs.

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3 We will assume that each pair of frogs consists of a male and a female, and that each pair produces only one offspring. The pairs produce offspring according to the following rules.
   - Two green frogs produce a green offspring.
   - Two yellow frogs produce a yellow offspring.
   - A green frog and a yellow frog produce an orange offspring.
   - Two orange frogs produce a colour that can be decided by the throw of a die:
     1 = a green offspring
     2 = a yellow offspring
     3 or 4 = an orange offspring
     5 or 6 – throw the die again until you get a 1, 2, 3 or 4.
   - A green frog and an orange frog produce a colour that can be decided by the throw of a die:
     1, 2 or 3 = a green offspring
     4, 5 or 6 = an orange offspring.
   - A yellow frog and an orange frog produce a colour that can be decided by the throw of a die:
     1, 2 or 3 = yellow
     4, 5 or 6 = orange.

4 Because their colours do not blend into the background so easily, yellow and orange frogs are more likely to be preyed upon by birds than are the green frogs. Simulate predation in your population of 45 frogs. Fifteen of the frogs are to be taken as prey. Throw a die 15 times and for each throw remove one frog according to the following rules.
   - If 1, 2, 3 is thrown, remove a yellow frog.
   - If 4 or 5, remove an orange frog.
   - If 6, remove a green frog.

5 You should have 30 cards remaining. This is your second generation of frogs. Count the cards and record the number of each colour in the table.

6 Shuffle the cards well and repeat steps 3, 4 and 5 to get the third generation. Record your results in the table.

7 Continue the process until all the frogs are the one colour, or until you have completed 10 generations.

Studying your results

1 Which colour frog became the most frequent in the population? Why do you think this was the case?

2 Which colour frog was eliminated first? Explain why this occurred.

3 Compare your results with other groups in the class. Have all groups obtained similar results? How much variation was there in the results between the different groups?

Interpreting your results

1 How has this activity modelled the process of natural selection? In your answer, describe what was creating the selection pressure on the population of frogs.

2 Explain why there was variability, if any, between the groups in your class.

3 What changes would you have to make to predation by the water birds to achieve a completely orange population of frogs? Repeat the activity with your changed parameters. Was your prediction correct?

4 Over several generations, what would happen to the composition of the frog population if water birds preyed equally on the three frog colours?

5 Write a summarising paragraph, using the principles of natural selection, to link the breeding patterns of the frogs and predation by water birds.

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ACTIVITY 9.4 Investigating sickle-cell haemoglobin

In an article published in the *British Medical Journal* in 1954, AC Allison first put forward the hypothesis that the possession of the sickle-cell allele may have a selective advantage in areas where malaria is prevalent. In investigating this proposition, Allison inoculated 30 African adult male volunteers with malaria and then observed them for 40 days. At the end of the period of observation, he treated all the participants with a prolonged course of antimalarial chemotherapy.

Allison's volunteers were of similar age and none had been in an area where malaria occurred for at least 18 months. They all appeared to be comparable except for the presence or absence of the sickle-cell allele. His results are shown in the following table.

Allison's results from the inoculation of Africans with malaria

	NUMBER OF PARTICIPANTS	DEVELOPED MALARIA	DID NOT DEVELOP MALARIA
With a sickle-cell allele	15	2	13
Lacking a sickle-cell allele	15	14	1

Interpreting the results

1. What was Allison's dependent variable? What was his independent variable?
2. What factors did Allison appear to control in his experiment?
3. Which group of subjects was the control group, and which the experimental?
4. Did Allison's results support his hypothesis? Explain why you think so.
5. Do the results Allison obtained suggest a reason why the sickle-cell allele has survived in Africa?
6. Refer to Figures 9.21 and 9.24. Does the information provided in these figures support your answer to Question 5? Give reasons for your answer.
7. Explain how the high incidence of the sickle-cell allele in parts of Africa could be considered an example of natural selection.
8. Would a university ethics committee today be likely to approve an experiment such as the one that Allison performed? Give reasons for your answer.

Allison's article describing his investigation

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CHAPTER 9 SUMMARY

- Evolution is the change in characteristics of a species over time due to changes in allele frequencies. It is influenced by mutations, migration, natural selection and genetic drift.
- A population is a group of organisms of the same species living in the same location at the same time. Geneticists study the frequency of alleles in the gene pools of populations. Variations in allele frequencies reflect differences in characteristics.
- Mutations are changes in the DNA that can occur spontaneously or due to exposure to a mutagen such as ionising radiation. They may affect a single gene (gene mutation) or more than one gene (chromosomal mutation).
- Mutations may be classified based on the following:
  - *Cause:* They can be spontaneous or induced.
  - *Heritability:* Mutations in somatic cells are not inherited (somatic mutations), but mutations in gametes are inherited (germline mutations).
  - *Effect:* Missense mutations change the protein produced, nonsense mutations produce a shorter protein that is not functional, neutral mutations change an amino acid but not the functioning of the protein, while a silent mutation does not change the amino acids and so the protein remains the same.
  - *Extent:* Gene mutations only affect one gene, while chromosomal mutations affect a number of genes or the whole chromosome.
  - *Change in DNA:* A nucleotide may be inserted, substituted or deleted; sections of DNA may be duplicated, deleted or translocated; chromosome pairs may not separate during meiosis.
- Insertion or deletions of nucleotides will result in a frameshift unless they are of a multiple of three nucleotides. This means that the base codes are read from a different base and, therefore, all the amino acids from that point on are affected.
- Examples of conditions due to mutations are: Duchenne muscular dystrophy and cystic fibrosis, due to gene mutations; Down syndrome, Patau syndrome and Klinefelter syndrome, due to trisomy; and Cri-du-chat syndrome and Turner syndrome, due to monosomy.
- Tay-Sachs disease is a recessive, autosomal lethal condition due to a mutation in the HEXA gene where the enzyme needed to break down GM2 ganglioside isn't produced. This results in a build-up of the toxic fatty substance that destroys neurons. Babies show intellectual and physical deterioration from age three to six months, and die early in childhood.
- Gene flow is the movement from one population to another. It is enabled by migration and stopped by some barriers such as geographical barriers, religion, language and sociocultural barriers.
- The theory of evolution by natural selection was proposed by Charles Darwin and Alfred Russel Wallace. It was based on the observation that there is variation within a species, the birth rate is greater than food supplies can sustain, and the species' numbers remained relatively constant. This led to the idea that species struggle for existence and to the theory of survival of the fittest. This means that favourable characteristics led to survival and were reproduced in the process of natural selection. The environmental factor that determines the survival is called the selective agent.
- Evolution occurs by natural selection, when the alleles for the favourable characteristics are passed on to future generations and therefore increase in frequency. This changes the characteristics of the species over a number of generations (evolution).

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- Sickle-cell anaemia is due to a point mutation on the HBB gene which changes the structure of haemoglobin, resulting in red blood cells with a distorted shape that become crescent shaped in low oxygen levels. The homozygous form is often fatal; however, the heterozygous form has milder symptoms and actually gives the individual protection from malaria. Therefore, in malaria-affected areas, the frequency of the sickle-cell allele is higher as it gives a survival advantage.
- Tay-Sachs disease is a fatal, recessive condition; therefore, affected individuals do not live long enough to reproduce. People who are heterozygotes have a selective advantage due to being protected from tuberculosis. Therefore, they will survive and reproduce, passing on the recessive and dominant alleles. This means that the incidence of Tay-Sachs disease is higher in areas affected by tuberculosis.
- Thalassemia is a gene mutation that results in changes to either the alpha globin or beta globin proteins in haemoglobin. Individuals with thalassemia have more red blood cells with less haemoglobin than unaffected individuals. These appear to give them some protection against malaria. Therefore, the incidence of thalassemia is greater in malaria-affected areas.
- Genetic drift is the random, non-directional change in allele frequency. It occurs in all populations; however, it usually only has a significant effect on small populations. It occurs when, by pure chance, more of one allele is passed on than others.
- The founder effect is an extreme example of genetic drift that occurs when a small group moves away from the original population. If, by chance, the allele frequency of the new group differs from the original population, then the frequency of features will also be different.
- The frequency of Tay-Sachs disease is greater in the population of Ashkenazi Jews than in other populations. This is due to a higher incidence in the common ancestor and to the fact that the Ashkenazi Jews are isolated by traditionally marrying within the religion.
- The bottleneck effect occurs when the population size is dramatically reduced by something like a natural disaster. The allele frequency of the remaining individuals may, by chance, be different from the original population.
- A species is a group of individuals that share many characteristics and are able to interbreed to produce fertile offspring. Evolution may lead to new species when the allele frequencies change so much that they are no longer able to interbreed. The process of producing new species is called speciation.
- Speciation occurs due to different selective pressures applied to different groups of a species. It goes through the process of variation, isolation, selection and speciation.

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CHAPTER 9 GLOSSARY

Achromatopsia An inherited form of total colour blindness

Albinism An inherited inability to produce pigment in hair, skin and eyes

Allele frequency How often each allele of a gene occurs in a population

Anaemia A condition in which there is a reduced amount of haemoglobin in the blood, or a reduced number of red blood cells

Aneuploidy A change in the chromosome number as a result of non-disjunction

Bottleneck effect An extreme form of genetic drift that occurs when the size of a population is severely reduced due to a sudden event such as a natural disaster. The allele frequency of survivors may not reflect that of the original population

Chromosomal mutation A change to the structure and/or number of chromosomes in an organism

Cri-du-chat syndrome A rare genetic disorder caused by a missing part of chromosome 5

Cystic fibrosis A disorder controlled by a recessive allele carried on an autosome that is incurable but can be detected during foetal development; mucus-secreting glands, particularly in the lungs and pancreas, become fibrous and produce abnormally thick mucus, resulting in, among other things, chest infections

Down syndrome *see* trisomy 21

Duchenne muscular dystrophy A genetic disease resulting in wasting of leg muscles and then arms, shoulders and chest

Evolution The gradual change in the characteristics of a species

Evolved Having gone through the process of evolution

Founder effect A type of genetic drift where a new population is formed by a small number of individuals; the small sample size can cause marked deviations in allele frequencies from the original population

Frameshift A mutation involving an insertion or a deletion that results in a change in the way that the sequence is read

Gene flow The transfer of alleles from one population to another through migration

Gene mutation An alteration to a single gene

Gene pool The sum of all the alleles carried by the members of a population

Genetic drift *see* random genetic drift

Geneticist A scientist who specialises in the study of genetics

Genotype The combination of alleles for a gene

Geographical barrier A feature of the landscape that prevents populations from interbreeding; includes oceans, mountain ranges, large lake systems, deserts and expansive ice sheets

Germinal mutation *see* germline mutation

Germline mutation A change in the hereditary material in the egg or sperm that becomes incorporated into the DNA of every cell in the body of the offspring

Heterozygote advantage A situation where a heterozygous genotype has a higher chance of survival than either homozygous genotype

Induced mutation A mutation caused by a mutagenic agent

Klinefelter syndrome A genetic disorder resulting from inheritance of two X chromosomes and one Y chromosome

Lethal recessive A recessive allele that, inherited in the homozygous condition, results in the death of the embryo, foetus or child

Migration The movement of people from one area to another with the intention of settling permanently

Missense mutation A mutation that causes a change in an amino acid resulting in a different protein being produced

Monosomy Where an individual has only one copy of a chromosome instead of two

Mutagen *see* mutagenic agent

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Mutagenic agent An environmental agent that increases the rate of mutation

Mutant An organism with a characteristic resulting from a mutation

Mutation A change in a gene or chromosome leading to new characteristics in an organism

Natural selection The process by which a species becomes better adapted to its environment; those individuals with favourable characteristics have a survival advantage and so pass those characteristics on to subsequent generations

Neutral mutation A mutation that causes a change in an amino acid; however, it does not cause an overall change in the protein

Nonsense mutation A mutation that results in a STOP codon, producing a shortened peptide chain

Partial monosomy Where part of a pair of chromosomes is missing

Patau syndrome A genetic disorder resulting from an extra copy of chromosome 13

Phenotype The observable characteristic due to the genotype

Phenylketonuria (PKU) An inherited disease resulting in damage to the growing brain and, thus, extreme intellectual deficiency, a tendency towards epileptic seizures, and failure to produce normal skin pigmentation

Point mutation A change in just one of the bases in a DNA molecule

Population A group of organisms of the same species living together in a particular place at a particular time

Random genetic drift The occurrence of characteristics in a population as a result of chance rather than natural selection; occurs only in small populations; also called genetic drift or Sewall Wright effect

Selective agent Any factor that causes the death of organisms with certain characteristics, but which has no effect on individuals without those characteristics

Sewall Wright effect *see* random genetic drift

Sickle-cell anaemia An inherited disease causing early death; results from the inheritance of two alleles for sickle-cell anaemia

Silent mutation A mutation that does not change the sequence of amino acids

Sociocultural barrier Barrier to interbreeding that is due to social or cultural factors

Somatic mutation A change occurring in a gene in a body cell (not in a gamete)

Special creation The belief that a god created all species

Speciation The process of new species developing

Species The basic unit of biological classification; members of a species are capable of interbreeding and producing fertile offspring

Spontaneous mutation A mutation that occurs due to an error in a natural biological process

Struggle for existence A principle where the number of organisms is greater than the resources in the environment can support; therefore, there is competition between the organisms for these resources

Survival of the fittest A principle whereby organisms with favourable characteristics survive, but organisms with unfavourable characteristics die before they have a chance to reproduce

Tay-Sachs disease (TSD) A genetic disorder caused by a missing enzyme that results in fatty substances accumulating in the nervous system

Trisomy 21 A genetic disorder resulting from an extra copy of chromosome 21 or an extra part of chromosome 21; also called Down syndrome

Turner syndrome A genetic disorder resulting from inheritance of one X chromosome and no other sex chromosome

Variation The differences that exist between individuals or populations of a species

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CHAPTER 9 REVIEW QUESTIONS

Recall

1 Define a 'population'.

2 What do scientists mean when they speak of a 'gene pool'?

3 a Define 'mutation'.
  b List the ways that the DNA may be changed in a mutation.
  c Distinguish between gene mutations and chromosomal mutations.
  d Give an example of a congenital disorder that can be caused by a gene mutation and one that can be caused by a chromosomal mutation.

4 a What are mutagens (or mutagenic agents)?
  b List five examples of mutagenic agents.

5 What is a lethal recessive?

6 a Distinguish between trisomy and monosomy.
  b Give an example of each condition.

7 Briefly describe the significance of the founder effect in human evolution.

8 a Define 'gene flow'.
  b List the common barriers that may lead to the isolation of one gene pool from another, and give examples of each type.
  c List five different kinds of sociocultural barriers to gene flow, and describe how each is thought to act.

9 Outline the main points of Darwin's theory of natural selection. Include an explanation of the terms 'struggle for existence' and 'survival of the fittest'.

Explain

10 Explain the difference between somatic and germline mutations.

11 Explain how mutations could change the proportion of certain alleles in a gene pool.

12 a Explain what random genetic drift is.
  b Select a modern population in which genetic drift is thought to have had an effect and describe why this might be the case.

13 Using the example of Tay-Sachs disease, explain how genetic diseases can lead to changes in allele frequencies in a population.

14 People of short stature tend to live in cold climates, and people with long limbs and short torsos tend to live in hot climates. Explain how these adaptations to cold and hot environments could have come about.

15 a What is sickle-cell anaemia?
  b Explain why sickle-cell anaemia is usually lethal.
  c List the advantages and disadvantages of having the sickle-cell trait in an area where malaria is prevalent.

16 How could isolation lead to selection and speciation?

Apply

17 Why does special care need to be taken when pregnant women require X-rays?

18 Summarise the pattern of inheritance that occurs in genetic disorders such as Duchenne muscular dystrophy. When there is no history of such disorders in a family, how are they thought to arise?

19 Discuss why mutations occurring in the reproductive cells are considered more important than those occurring in the body cells. In your discussion, describe the possible long-term effects of the two situations.

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20 The more often cells divide, the greater the risk of errors and mutations. For this reason, scientists have hypothesised that when a baby is born with a congenital disorder caused by an error in cell division, the father is the parent more likely to have contributed the gene with the mutation. Compare the number of eggs produced by a female with the number of sperm produced by a male and explain why scientists have proposed this hypothesis.

21 Lethal recessive alleles result in the death of an individual. How would this affect the allelic composition of the gene pool?

22 The risk of having a baby with Down syndrome increases as the mother gets older. The following table shows the relationship between Down syndrome and maternal age.

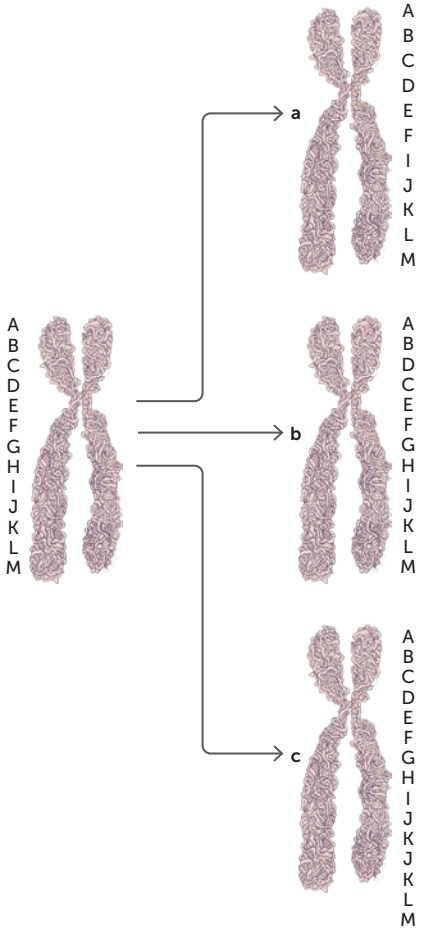
- **a** Draw an appropriate graph to display the data in the table.
- **b** The risk of a baby having any chromosome abnormality increases dramatically with increasing maternal age. Suggest reasons for this.

Mother's age and risk of having a baby with Down syndrome

AGE OF MOTHER (YEARS)	RISK OF DOWN SYNDROME
20	1 in 1667
23	1 in 1429
26	1 in 1176
29	1 in 1000
32	1 in 769
37	1 in 227
40	1 in 106
43	1 in 50
46	1 in 23
48	1 in 14
49	1 in 11

Source of data: Adapted from Dr Mark Hill 2020, UNSW Embryology

23 The following figure shows the sequence of the genes A to M on a chromosome. What type of chromosomal mutation is represented by each of a, b and c?



24 During the 14th century, plague epidemics drastically reduced the human population of Europe. Use this as an example to describe the way natural selection operates so that only the fittest tend to survive.

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25 According to a recent report, 13% of Scotland's population are redheads. Two out of every five Scots carry the allele for red hair. However, only 2% of the world's population are estimated to be natural redheads.

- **a** Suggest a reason for the high frequency of the allele for red hair in the gene pool of the Scots.
- **b** In the population of Scotland, what do you think will happen to the frequency of the allele for red hair over time? Give reasons for your answer.

26 A team of American scientists has been trying to develop a vaccine to give permanent immunity against malaria. What do you think will happen to the frequency of the sickle-cell gene within a population if this vaccine is effective? In writing your answer, ensure that you explain the adaptive value of the various genotypes and the selection pressures on each.

Extend

27 Western Australia has been a world leader in the application of carrier detection to reduce the incidence of Duchenne muscular dystrophy.

- **a** What does carrier detection involve?
- **b** What takes place following the detection of a carrier?
- **c** What is preventing the complete elimination of Duchenne muscular dystrophy?

28 Malthus claimed that species of organisms always produce more offspring than the existing resources can support. Is this true of the human species in the past or at present? Is it likely to be true of the human species in the future?

29 Describe the barriers to gene flow that exist for the following populations:

- **a** groups in South Africa
- **b** groups in the islands of Polynesia, such as New Zealand, Tahiti and Hawaii
- **c** Jewish people.

30 Using analysis of mitochondrial DNA, researchers have determined that all humans are descended from a woman who lived in Africa 200 000 years ago – the so-called mitochondrial Eve. If we are all descended from a common ancestor, how is it that there are so many different types of humans today? Describe the processes that must have taken place to produce the differences between present-day groups of humans.

31 Speculate on what might be the long-term effect on allele frequencies if a mutation suddenly produced a favourable allele that gave a natural resistance to all forms of heart disease.

32 One of the best-researched investigations into natural selection is the work of a British geneticist, Henry Bernard Davis Kettlewell, on the peppered moth, *Biston betularia*. The peppered moth gets its name from the scattered dark markings on its otherwise pale wings and body. The moth flies at night and rests by day on tree trunks. These trunks are usually encrusted with lichens, and the pale-coloured moth is practically invisible against this background.

However, in 1849, a coal-black mutant form of the moth was found near Manchester in England. Within a century, this black form had increased to 90% of the population in this region. The change in allele frequency that occurred in this example is a good model of how natural selection takes place.

Find out:

- **a** how the black form of the moth became the more prevalent variant
- **b** which form is the most prevalent today.