PEARSON

Science

STUDENT BOOK | VICTORIA

10

VICTORIAN CURRICULUM

TOPIC 1

Genetics and models of inheritance

Has anyone ever told you that you look like your mother or father, or maybe one of your siblings? How you look (physical traits) and many of your other bodily characteristics are mostly determined by your genetic material. The genetic material that is found in the nucleus of your cells is called deoxyribonucleic acid (DNA). DNA is packaged into organised structures called chromosomes. It is your chromosomes that contain the information to determine your characteristics.

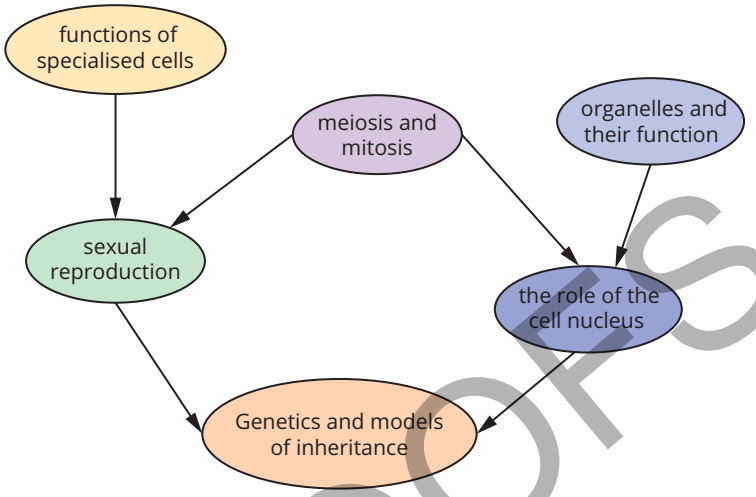
In this topic you will learn about DNA and chromosomes, which will be further explored to understand genotypes and phenotypes, including how you can predict these using Punnett squares. You will also examine inheritance patterns and observe how errors can cause mutations that may lead to a genetic disease.

Learning intentions

- ☐ To understand the role of DNA and chromosomes in passing genetic material to offspring **xx**
- ☐ To be able to conduct an experiment to extract an observable amount of DNA **xx**
- ☐ To be able to create a model to represent the structure of DNA **xx**
- ☐ To understand relationships between DNA, chromosomes, genes and alleles **xx**
- ☐ To understand the role of mitosis and meiosis **xx**
- ☐ To understand the genetic outcomes of meiosis **xx**
- ☐ To understand how genetic material is expressed in the characteristics of organisms and to be able to predict phenotypes **xx**
- ☐ To be able to model genetic variation **xx**
- ☐ To understand how patterns of inheritance can be represented using a pedigree diagram **xx**
- ☐ To understand the kinship laws, family structures and marriage restrictions of First Nations Australians **xx**
- ☐ To understand the causes and effects of changes in genetic material **xx**
- ☐ To understand the role of genetic changes that lead to cancer, haemochromatosis, sickle cell anaemia and cystic fibrosis **xx**

1 Genetics and models of inheritance

The key concepts that you will use in this topic:



The following key knowledge questions will help to support your learning in the topic and can be attempted before the first lesson.

The role of the cell nucleus

1 State the name of the material that is found within the nucleus of eukaryotic cells.

2 Describe the role of the nucleus within the cell.

Sexual reproduction

3 Sexual reproduction requires the union of male and female sex cells (gametes).

a Name the male and female gametes in humans.

b Name the male and female gametes in flowering plants.

4 Explain a benefit of sexual reproduction in comparison to asexual reproduction.

Genetics and models of inheritance

5 List three physical characteristics that all mammals share.

6 Strawberries can reproduce both sexually and asexually. The new plants produced by asexual reproduction are known as clones. What are some characteristics of organisms that are clones?

1.1 Introduction to chromosomes and DNA

Lesson overview

Cells form the basis of all living things, from a single-celled amoeba to a complex multicellular organism like you. Each cell, with a few exceptions, contain organelles and genetic information that allow it to function and survive. Genetic information is critical to life because it contains all the instructions for organisms to grow, repair and reproduce.

The genetic information of an organism is stored as deoxyribonucleic acid (DNA) molecules. The genetic material in prokaryotic cells is found within the cytoplasm, as these cells lack a nucleus. In comparison, eukaryotic organisms contain a nucleus and other organelles. All cells begin the process of growing, repairing or reproducing by weaving and coiling their DNA into chromosomes. To do this, cells undergo an amazing process: they replicate (make copies) and transfer their DNA so that they can create new cells.

In this lesson you will learn about how DNA is organised into chromosomes so that an organism can pass its genetic material to its offspring.

Learning intention

To understand the role of DNA and chromosomes in passing genetic material to offspring

Success criteria

SC 1: I can recall the role of genetic material in organisms.

SC 2: I can explain how genetic material is stored within different organisms.

SC 3: I can label a diagram of a chromosome.

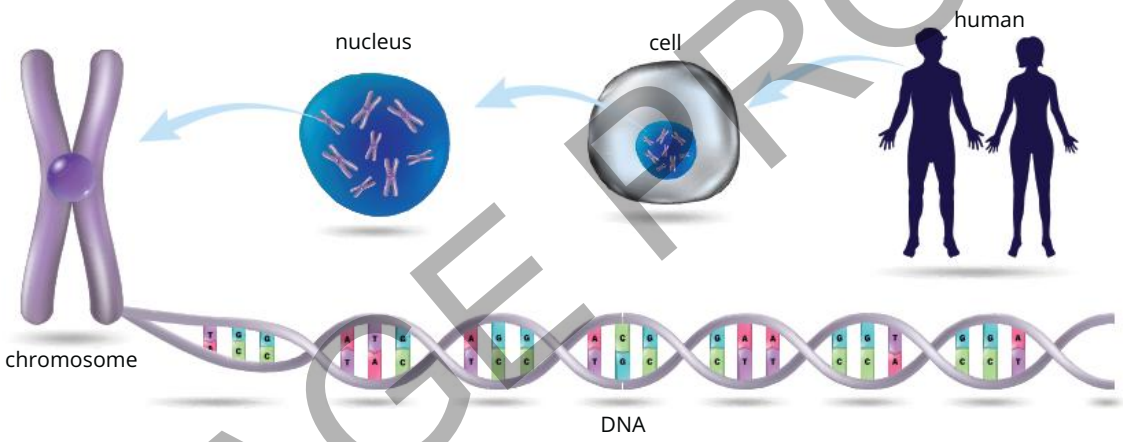


FIGURE 1.1.1 This image shows chromosomes located in the nucleus of a cell and unravels the structure of a chromosome so that the coiled DNA is visible.

SC 1 I can recall the role of genetic material in organisms

Your **deoxyribonucleic acid (DNA)** is the molecule that carries your genetic material – the complete set of instructions needed to make you uniquely you. This genetic material is stored in every cell and guides cell function, growth and **reproduction**. DNA also determines many of your characteristics, or **traits**. Because it is passed from parents to their children during reproduction, **offspring** often inherit traits from their parents.

Understanding cell structure and how genetic material determines specific traits has enabled scientists to explain the inheritance of characteristics from parent to offspring. There are several scientists that have helped to improve understanding of inheritance and genetics.

KEY TERMS

deoxyribonucleic acid (DNA) a double helix made of nucleotides; the molecule that determines the genetic characteristics of most living things

reproduction the process of parents producing new individuals, or offspring

trait inherited characteristic

offspring individuals produced by reproduction

KEY TERMS

genetics the study of inherited characteristics called traits
double helix shape like that of a twisted rope ladder

Gregor Mendel

Gregor Mendel, a monk who studied science, and known as the father of **genetics,** explored the concept of inheritance in plants in the 1850s.

Mendel investigated how pea plants of a certain colour produced offspring of the same and different colours in different generations (Figure 1.1.2). He concluded that each offspring received two factors (genes), one from each parent. Current knowledge of genetics stems from these important historical experiments completed by Mendel.

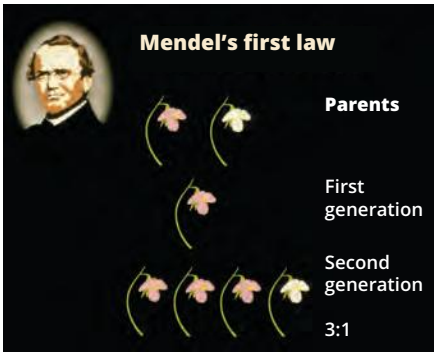


FIGURE 1.1.2 Mendel's experiments on pea plants showed how traits were passed down through generations.

James Watson and Francis Crick

In 1953, James Watson and Francis Crick proposed that DNA was made up of chains of nucleotide pairs and developed a two-stranded helical (spiral) model of DNA that could be used to represent genes (Figure 1.1.3).

Rosalind Franklin

Rosalind Franklin, a young female scientist, contributed to discovering the structure of DNA but was not acknowledged at the time. Her work with X-ray crystallography proved the existence of a **double helix** (spiral) (Figure 1.1.4) structure that was critical to the understanding and development of Watson and Crick's model.

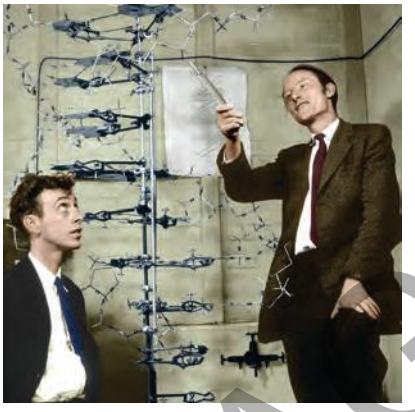


FIGURE 1.1.3 Watson and Crick standing with their physical DNA model.

FIGURE 1.1.4 Rosalind Franklin with her image of DNA, which she discovered using X-ray crystallography.

SC 1 CHECK YOUR UNDERSTANDING

Describe the purpose of genetic material in humans.

SC 2 I can explain how genetic material is stored within different organisms

What is DNA?

DNA is a long double-stranded helical (spiral) nucleic acid that contains the instructions for life. DNA is found in the **nucleus** of a **eukaryotic cell**, and the cytoplasm of prokaryotic cells.

DNA is packaged together in **chromosomes**.

In body cells (**somatic cells**), chromosomes come in pairs. For example, one of your skin cells has a total of 46 individual chromosomes organised into 23 pairs.

DNA is a nucleic acid made up of units called **nucleotides**. Figure 1.1.5 shows that nucleotides are made of three parts: a phosphate group, deoxyribose sugar and one of four nitrogen-rich **bases**.

The four nitrogenous bases are adenine (A), thymine (T), guanine (G) and cytosine (C). Each base has a different chemical structure and shape that complements its matching base. Adenine always pairs with thymine (A–T), and guanine always pairs with cytosine (G–C). This pairing is known as complementary base pairing. Complementary base pairing gives DNA its structure by creating the 'ladder-like' effect of the DNA double helix. The two strands of nitrogenous bases are like long chains that are held in place and spiral around each other. This is known as the double helix structure of DNA.

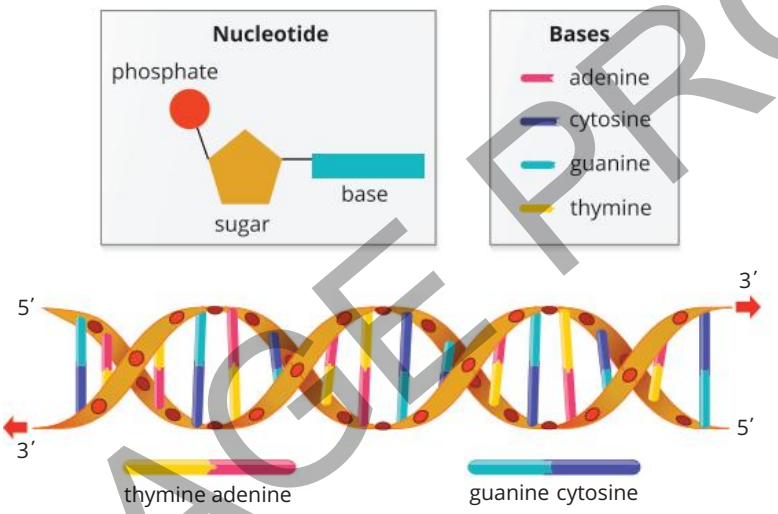


FIGURE 1.1.5 DNA is made up of nucleotides, which consist of three parts: sugar, phosphate and a nitrogenous base. There are four possible bases.

Scifile

Microscopic storage

The nucleus of a cell is usually 5–10 µm (micrometres) in diameter, which is roughly 0.005 to 0.01 mm. If you were to stretch out all the DNA from a single human cell end to end, it would be about 2 m long. That is an incredible amount of genetic material packed into an extremely small space!

KEY TERMS

nucleus organelle that contains the genetic information for the cell (plural nuclei)
eukaryotic cell cell that contains a nucleus
chromosome thread-like structure in the nucleus; composed of DNA and proteins; contains genetic information in the form of genes
somatic cell any cell except a cell that gives rise to gametes (eggs and sperm)
nucleotide a molecule consisting of a five-carbon sugar (ribose or deoxyribose), a nitrogenous base (purine or pyrimidine) and a phosphate group; the main building blocks of nucleic acids
base part of a nucleotide; the four types include adenine (A), guanine (G), cytosine (C) and thymine (T)

Chromosomes

A cell might be microscopic, but it contains billions of nucleotide pairs. To package DNA so that it fits inside a cell nucleus, it is wound tightly around proteins called histones to form chromosomes as seen in Figure 1.1.6.

KEY TERMS

autosome chromosome that is not a sex chromosome
sex chromosome chromosome that determines the biological sex of an individual; X and Y chromosomes in humans
homologous chromosome one of a matching pair of chromosomes in a diploid organism; homologous chromosomes carry the same genes in the same location
gamete sperm cell or egg cell
haploid number the number of chromosomes in gametes; one set or n

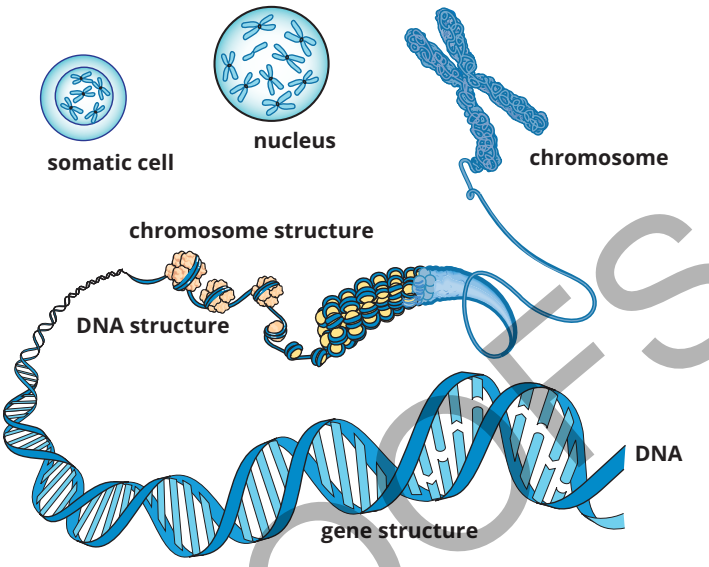


FIGURE 1.1.6 DNA is wound tightly around histones and organised into chromosomes, which are found inside the cell nucleus.

Most cells in the human body contain 46 chromosomes. Of these, 44 are known as **autosomes** that can found in pair. These contribute most of the genetic material to the cell. The remaining two chromosomes are known as **sex chromosomes**. These are the chromosomes that determine whether a person is biologically typically male or female. In males, the sex chromosomes are one X and one Y (XY). In females, the sex chromosomes are a pair of X chromosomes (XX).

Chromosomes that are found in pairs are known as **homologous chromosomes** (Figure 1.1.7) that contain the same position of genes.

FIGURE 1.1.7 A pair of number 4 homologous chromosomes viewed with a coloured scanning electron micrograph (SEM).

Gametes (sex cells) only contain half the amount of genetic material as somatic cells. They have 23 single (unpaired) chromosomes and are known as **haploid**.

Scifile

How many chromosomes?

Different organisms have different chromosome numbers that do not dictate the complexity of the organism. For example, tomato plants have 24 chromosomes in each cell, fruit flies have 8, and the *Agrodiaetus amandus* butterfly has 268.

Genes

Genes are segments of DNA that code for specific features and functions. Each gene has a particular sequence and number of base pairs (A, T, G and C). Genes may vary in size from a few hundred base pairs to more than a million base pairs. The order of the bases along the strand of DNA is known as the **genetic code**.

Each chromosome contains many genes.

KEY TERMS

gene a section of DNA that carries the genetic code for a particular characteristic

genetic code the sequence of three nucleotides in DNA that determines, or codes for, genes

Genetic material in prokaryotes

The structure of genetic material in prokaryotic organisms differs from eukaryotic organisms. It is far less organised because prokaryotic organisms lack membrane bound organelles such as a nucleus. Most prokaryotic cells have a singular round chromosome of centrally located circular DNA (sometimes known as nucleoid DNA) that contains genes (Figure 1.1.8) and plasmids, which are smaller circular shapes. The number of genes in each prokaryotic cell differs and can range from 140–7600 genes that assist with survival. This is considerably less than the estimated 20 000 coding genes in humans.

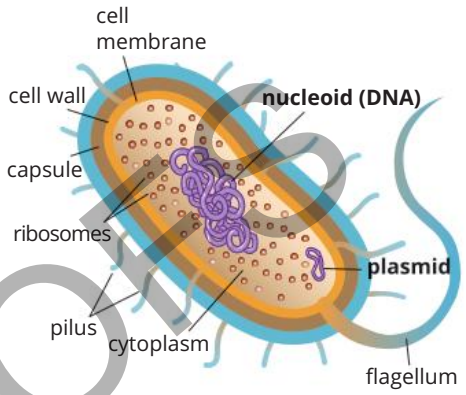


FIGURE 1.1.8 A prokaryotic cell showing genetic material.

SC 2 CHECK YOUR UNDERSTANDING

Describe the structure and location of DNA within a eukaryotic cell.

SC 3 I can label a diagram of a chromosome

Chromosomes

Chromosomes are threadlike structures located in the nucleus of a cell (Figure 1.1.9). They are made of long, double-stranded DNA molecules wrapped around histone proteins.

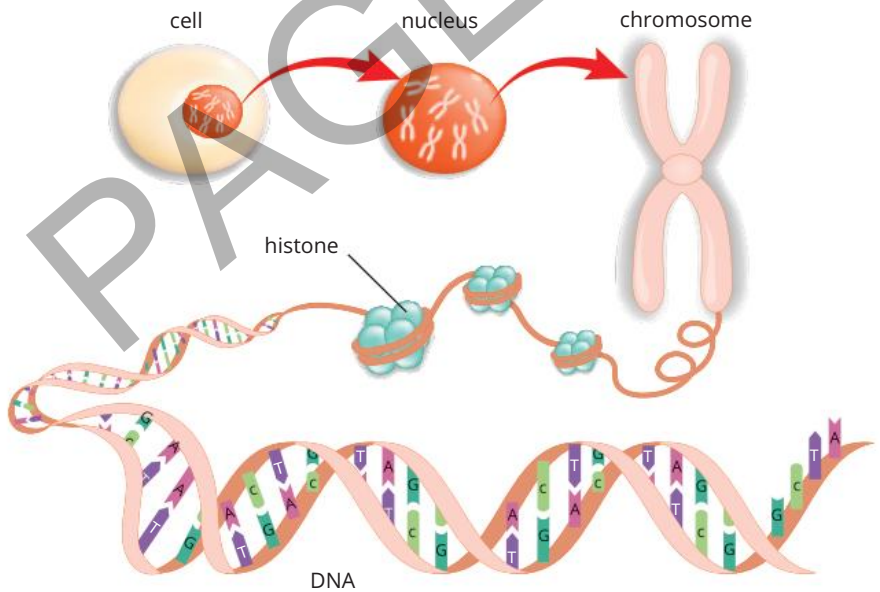


FIGURE 1.1.9 The hierarchy of DNA organisation within the nucleus of a cell.

Chromosomes come in different sizes, but each has the same basic structure: two short arms, two long arms and a **centromere** joining the two together.

KEY TERM

centromere the point on a chromosome where the two chromatids are joined together

KEY TERMS

chromatid one of the strands of a chromosome following replication

karyotype a visual representation of the number, size and shape of chromosomes in an individual

In body cells, a chromosome is made up of two sister **chromatids**. A chromatid is one of the two identical halves of a chromosome.

For each chromosome in a homologous pair, genes are found in the same location.

Karyotype

A **karyotype** is an image of an organism's complete set of chromosomes (Figure 1.1.10).

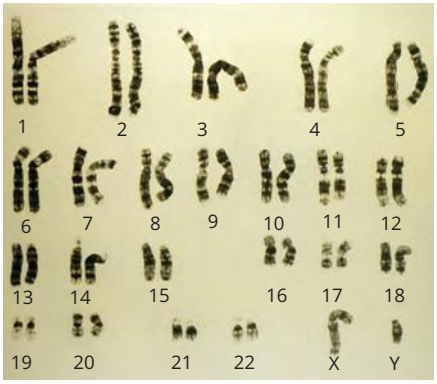


FIGURE 1.1.10 A male karyotype; notice the XY pair at chromosome 23.

In a human karyotype, the first 22 pairs are the autosomes and the 23rd pair are the sex chromosomes. In females, the pair of sex chromosomes are XX and in males they are XY. The first 22 pairs of chromosomes in a karyotype are arranged in order of size from largest to smallest.

Karyotypes can be used to look for genetic abnormalities, such as a trisomy. Trisomy means that there is an extra copy of a chromosome in each cell.

For example, trisomy 21 is when an individual has three copies of chromosome 21 as highlighted in Figure 1.1.12. This is commonly known as Down syndrome. People with Down syndrome usually have distinct facial features, short stature and low muscle tone (Figure 1.1.11).

FIGURE 1.1.11 A student with Down syndrome characteristics.

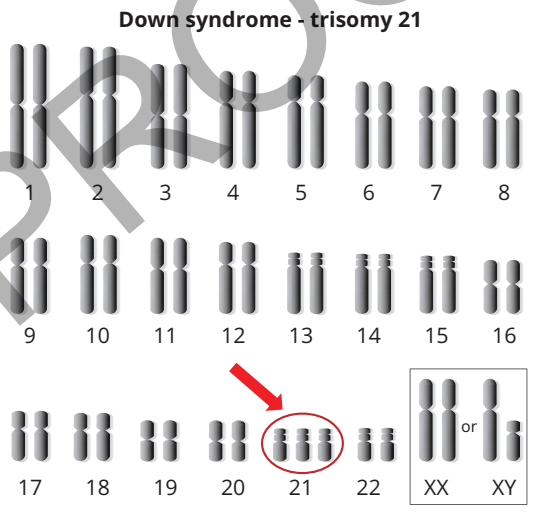


FIGURE 1.1.12 A karyotype showing three copies of chromosome 21, which is known as trisomy 21 and results in Down syndrome.

SC 3 CHECK YOUR UNDERSTANDING

Draw and label a diagram of a homologous pair of chromosomes.

Lesson review

Use these questions to check whether you have met the learning intention for this lesson.

1. List the three components of a nucleotide.
2. Order the following from smallest to largest: DNA, base, gene, nucleotide, chromosome.
3. Describe the contribution Rosalind Franklin had to the structure of DNA.
4. Compare the genetic material in eukaryotic cells with that of prokaryotic cells.
5. Explain how karyotypes can be used to identify the sex of an individual.

1.2 Extracting DNA

Introduction

Deoxyribonucleic acid (DNA) is a molecule found in the nucleus of plant and animal cells. It carries the genetic instructions for the development, growth and reproduction of the organism. DNA molecules are polymers, which means that they are made up of many nucleotides. Each DNA molecule is made up of two chains of nucleotides that spiral around each other, which gives DNA its double helix structure.

In this practical investigation you will follow a method to produce a solid sample of DNA from peas. You may be surprised with how much DNA you can extract.

Learning intention

To be able to conduct an experiment to extract an observable amount of DNA

Success criteria

SC 1: I can accurately perform an experiment ensuring that quantities are carefully controlled.

SC 2: I can use scientific knowledge of cells, chemical reactions and physical change to explain the processes used in DNA extraction.

SC 3: I can evaluate the effectiveness of the DNA extraction method.

Background

DNA can be extracted from cells by breaking down cell structures. In a cell, the DNA molecules are often connected to histone proteins. DNA is soluble (can dissolve) in water but insoluble (cannot dissolve) in alcohol, so when alcohol is added, DNA can be collected as solid material.

Aim

To extract and examine DNA from peas

Materials

- 125 mL $\left(\frac{1}{2}\text{ cup}\right)$ dried split peas (soaked overnight)
- 200 mL water
- 80 mL dishwashing detergent
- $\frac{1}{4}$ teaspoon meat tenderiser powder
- 10 mL alcohol (ethanol)
- large measuring cylinder
- blender
- fine-mesh kitchen strainer
- 500 mL beaker
- dropping pipette
- glass rod or skewer
- spatula
- 2 large test tubes and test-tube rack
- paper towel

SAFETY NOTES

- Wear eye protection throughout the practical investigation.
- Take care when using the blender to ensure there are no leaks.
- Do not touch or taste any substances.
- Discuss your safety notes and risk assessment with your teacher before proceeding.

Assessment of risk

Ensure you are aware of the risks of this practical investigation and have considered how safety can be improved before carrying out this activity.

Method

1. Process the peas and water in the blender for about 20 seconds. The mixture should be a thin, soupy consistency.
2. Pour the mixture through the strainer into the large beaker.
3. Add about 80 mL of dishwashing detergent to the strained mixture to help break down the cell membranes. Stir thoroughly with the glass rod.
4. Add a spatula-full of meat tenderiser, which contains an enzyme that breaks down proteins. Continue stirring for about 5 minutes.
5. Quarter-fill the large test tube with the pea mixture.
6. Holding the test tube at an angle, gently pour about the same quantity of alcohol (about a quarter of a test tube) down the side of the test tube so that the alcohol forms a layer on top of the pea mixture. Alcohol causes the DNA to come out of solution and form a solid.
7. Observe the mixture for a few minutes. A white, threadlike substance should rise from the pea mixture to lie above the alcohol layer. This is the DNA that you have extracted from the cells of the peas.
8. Half fill a clean test tube with alcohol.
9. Position the tip of the glass rod or skewer where you can see the threads of DNA. Slowly and steadily twist the rod or skewer as shown in Figure 1.2.1. You should be able to pull the strands of DNA out of the mixture.
10. Transfer the DNA into the clean test tube containing alcohol, as shown in Figure 1.2.2.

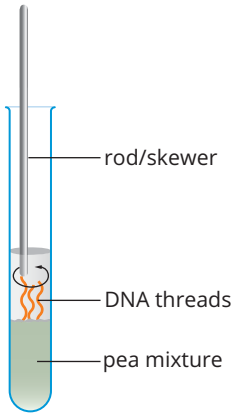


FIGURE 1.2.1 Experimental set-up of DNA extraction following the sequence in step 9.

FIGURE 1.2.2 Test tube with extracted DNA suspended in alcohol solution.

Results

Describe the appearance of the DNA.

Conclusion

With reference to cell structure, chemical reactions and physical change, explain the following stages of the extraction:

Step 1: processing in the blender

Step 2: straining

Step 3: adding detergent

Step 4: adding meat tenderiser

Step 6: adding alcohol

Step 9: extracting threads.

Evaluation

1. Use your observations to evaluate the success of your extraction experiment.
2. Describe one change to the method that might improve the results of the experiment.

1.3 Structure of DNA

Introduction

Deoxyribonucleic acid (DNA) is the molecule that carries the genetic information for all living organisms. This information is encoded in the structure of DNA, which is a polymer made up of repeating units called nucleotides. Each nucleotide consists of a phosphate group, a sugar molecule, and a nitrogen-rich base.

While all nucleotides share the same phosphate and sugar components, the nitrogen base can vary. There are four different bases, as shown in Figure 1.3.1. The specific sequence, or order, of these bases forms the genetic code. Each segment of this sequence contains instructions that direct various functions in the body.
In this practical investigation you will create a model of a strand of DNA and use it to demonstrate the huge number of possible combinations that can be created from just four bases, adenine (A), thymine (T), cytosine (C) and guanine (G).

Learning intention

To be able to create a model to represent the structure of DNA

Success criteria

SC 1: I can correctly label a diagram of DNA.

SC 2: I can create a two- or three-dimensional model to represent the structure of DNA.

SC 3: I can explain complementary base pairing using a model of DNA.

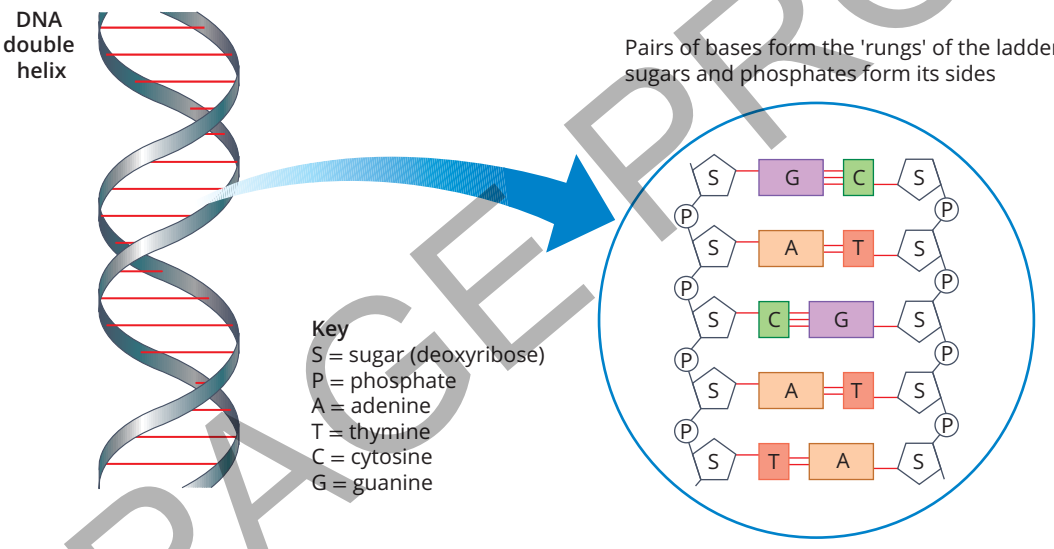


FIGURE 1.3.1 Close-up of a DNA molecule reveals a combination of the four bases that link each spiral of the double helix.

Background

The four bases in molecules of DNA are adenine (A), thymine (T), cytosine (C) and guanine (G). They are called nitrogenous bases (or nitrogen-rich bases) because they contain many nitrogen atoms.

In DNA, these bases form pairs that span between the two spiralling chains of the DNA molecule. However, due to the molecular structure of the bases, each type of base can only pair with one other, complementary (matching) base. These complementary pairs are called base pairs: adenine pairs with thymine to form an A–T (or T–A) pair, and guanine pairs with cytosine to form a G–C (or C–G) pair as seen in Figure 1.3.2.

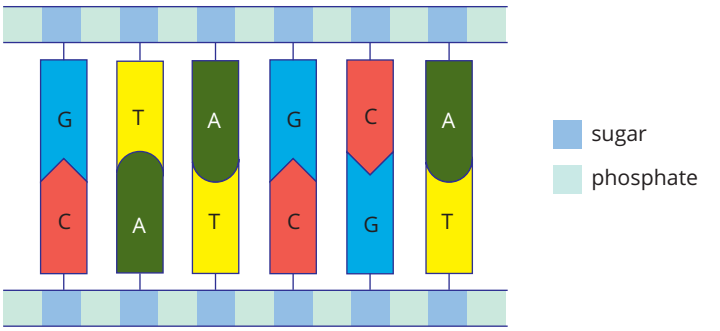


FIGURE 1.3.2 The molecular shape of adenine (A) fits with the shape of thymine (T) to form a base pair. In the same way, the shape of cytosine (C) fits with guanine (G).

Aim

To construct a model of DNA and use it to investigate the number of combinations of base pairs

SAFETY NOTES

- Write safety notes for your investigation.
- Discuss your safety notes and risk assessment with your teacher before proceeding.

Materials

- 20 coloured paperclips: 5 yellow, 5 green, 5 blue, 5 red
- 2 strips of paper: 1.5 cm × 30 cm
- coloured pencils

Assessment of risk

Ensure you are aware of the risks of this practical investigation and have considered how safety can be improved before carrying out this activity.

Method

1. Use paperclips to represent the bases in your DNA molecule. Choose a different colour for each type of base: adenine, guanine, cytosine and thymine. Make a note of which colour you have assigned to each type of base.
2. Mark the two strips of paper into 2 cm sections.
3. Shade each 2 cm section of the two strips of paper using two alternating colours to represent the sugar and phosphate molecules.
4. Attach 10 of your coloured paperclips randomly (in any sequence you like) to the 'sugar molecules' along one of the strips as shown in Figure 1.3.3.

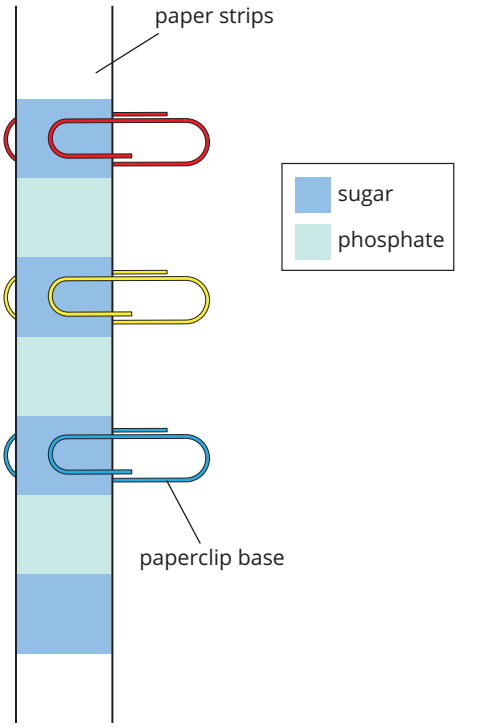


FIGURE 1.3.3 Step 4 of the method showing the paper clip (nucleotide) bases attached to the sugar part of the chain.

5 Use the base-pairing rules described in the background information to build the complementary strand for this DNA molecule. Attach the two strands to each other by joining each paperclip, as shown in Figure 1.3.4.

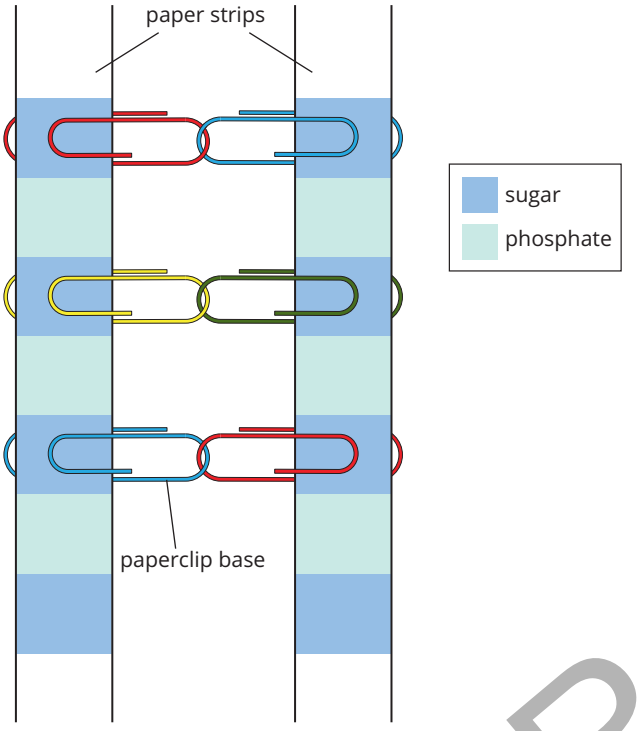


FIGURE 1.3.4 Step 5 of the method showing complementary pairing of the paper clip bases and joining of two DNA chains.

Results

Draw a labelled diagram of the model DNA molecule you have made.

Conclusion

1 Compare your model to some of the other models made in your class and comment on any differences and similarities that you can observe.

2 Estimate the number of variations of single DNA strands that can be made using only the 10 bases that you started with.

3 Comment on what would happen to the number of variations that could be made if the strand of DNA were thousands of bases in length.

4 Explain why, although you could choose any order of bases on the first strand, there is only one possible order of bases on the second strand.

Evaluation

Evaluate how well your model explains the structure of DNA.

1.4 The role of DNA in heredity

Learning intention

To understand the relationships between DNA, chromosomes, genes and alleles

Success criteria

SC 1: I can describe the structure of a gene and its role in determining alleles in an individual.

SC 2: I can explain the functional relationships between DNA, chromosomes, genes and alleles.

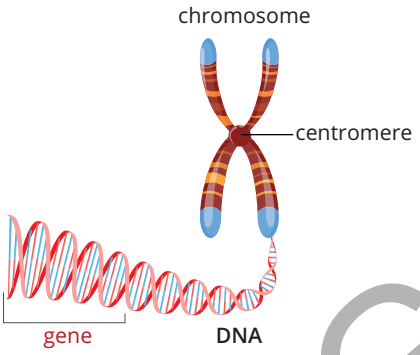


FIGURE 1.4.1 Genes are the basic units of inheritance.

KEY TERMS

protein natural polymer required for growth and repair in living organisms

allele one of two or more alternative forms of a gene that can occupy a specific position on a chromosome

Lesson overview

Will you have brown hair if your biological parents have brown hair? What about if one parent has brown hair and one parent has blonde hair? Gregor Mendel studied pea plants and found that offspring inherit two factors (genes), one from each parent. James Watson and Francis Crick, with the assistance of Rosalind Franklin, then went on to confirm the structure and existence of deoxyribonucleic acid (DNA).

In this lesson you will learn about how DNA is arranged into chromosomes and contains genes that determine an individual's alleles.

SC 1 I can describe the structure of a gene and its role in determining alleles in an individual

Genes

A gene is a section of DNA composed of a sequence of four different bases (nucleotides, Figure 1.4.1). DNA is tightly wrapped to form the shape of a chromosome.

DNA sequence determines a gene's function

Each gene will have a different order and number of bases (A, T, C and G). Genes code for specific **proteins**. These proteins create the structures and functions needed for cells and organisms to survive. Different chromosomes are made up of different genes, which means that they code for different proteins. Your 23 pairs of chromosomes code for all the proteins, structures and functions in your body. Your genes are what make you unique.

Alleles

You receive two copies of each gene, one from each biological parent. The genes for a particular characteristic are found at the same site for each of the two chromosomes in a homologous pair. Each copy of a gene that you inherit will be different. The different forms of a gene are called **alleles** (Figure 1.4.2).

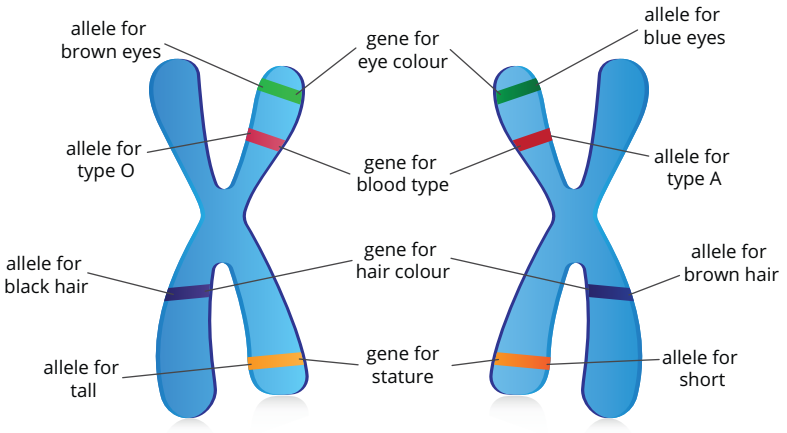


FIGURE 1.4.2 Genes exist as alleles on each of the two chromosomes in a homologous pair.

For example, you inherit one gene for hair colour from your biological mother and one from your biological father – these two genes are then referred to as alleles. It is important to note that your biological parents also have two alleles for each gene. However, in genetic inheritance through **sexual reproduction**, each parent contributes only one of their two alleles to the offspring. As a result, you and your biological siblings may inherit different alleles from each of your parents.

KEY TERM

sexual reproduction the process of a sperm and an egg joining together and then growing into a new individual

The DNA code that makes up a specific gene is different in different alleles. For example, the DNA code in the allele for blue eyes is different to the DNA code in the allele for brown eyes.

Similar processes also happen in the development, growth and reproduction of plants and other animals. Figure 1.4.3 shows two homologous chromosomes found in a flowering plant. The plant has two copies of the gene for flower colour, one for red flowers and one for white flowers.

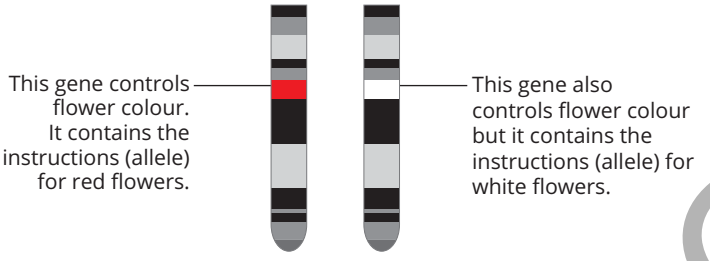


FIGURE 1.4.3 This homologous pair of pea plant chromosomes illustrates the genes that control the colour of the plant's flowers; in this example, one allele codes for red flowers and the other allele codes for white flowers.

SC 1 CHECK YOUR UNDERSTANDING

Define the term allele.

SC 2 I can explain the functional relationships between DNA, chromosomes, genes and alleles

DNA, chromosomes, genes and alleles

Chromosomes are made of DNA, which has different segments (sections) known as genes. A summary of the hierarchy of DNA to chromosomes can be seen in Figure 1.4.4. Each gene codes for something different. Each pair of homologous chromosomes will have the same genes at the same locations. You inherit two forms of each gene, known as alleles.

DNA	Gene	Chromosome
• double helix • sugar-phosphate backbone with nitrogenous bases • nitrogenous bases code for a gene	• the instructions for a particular trait • alternative forms of genes are known as alleles	• DNA containing genes is tightly wound up into a chromosome that is found within the nucleus of a eukaryotic cell

FIGURE 1.4.4 A summary of how genetic material is organised within eukaryotic cells.

Alleles control traits

Alleles are different versions of the same gene, each with a unique DNA sequence. In a homologous pair of chromosomes, the same gene is found at the same location on each chromosome. However, because the DNA sequence of each allele can differ, they may produce different variations of a trait; for example, different hair or eye colours.

KEY TERMS

genotype entire genetic information of an individual; refers to alleles present for each gene

phenotype observable characteristics of the individual; the way the genotype is expressed

Alleles are often represented using letters: a dominant allele is shown with a capital letter (such as *B* for brown eyes), while a recessive allele is shown with a lowercase letter (such as *b* for blue eyes). You will learn more about this later in the topic.

Genotype and phenotype

The two alleles that you have for each gene are your **genotype**. How the genotype physically expresses itself is the **phenotype** (Figure 1.4.5). You cannot always determine someone's genotype just by looking at them – but you can usually see their phenotype, unless it is microscopic.

FIGURE 1.4.5 These two people have a different phenotype for eye colour; one has brown eyes and the other has blue eyes so their alleles for eye colour are different.

SCIENCE IN SOCIETY

Genetic research in Australia

In Australia, genetic research is advancing the understanding of how DNA, chromosomes, genes, and alleles interact to influence traits and diseases. For example, researchers at the Garvan Institute of Medical Research are studying the genetic basis of diseases like cancer and diabetes. By understanding how specific genes and their alleles contribute to these conditions, scientists can develop targeted treatments (Figure 1.4.6).

In agriculture, understanding these relationships helps in breeding programs. Australian farmers use genetic information to select plants and animals with desirable traits, such as disease resistance or higher yields. This not only improves productivity but also ensures the sustainability of farming practices.

FIGURE 1.4.6 A scientist preparing DNA samples for analysis in the laboratory.

Switching genes on and off

Every somatic cell in an organism has the same set of chromosomes and, therefore, the same set of genes. However, not all genes are active in every cell.

Some genes must be switched on while others are switched off, either during specific cell stages or in different parts of the body. This allows cells in the body to have many different functions. For example, when a cell needs to create colour for a strand of hair, it will switch on the genes that code for hair colour and may switch off other genes that are not needed at the time.

SC 2 CHECK YOUR UNDERSTANDING

Explain how DNA sequences control the phenotype of an individual.

Lesson review

Use these questions to check whether you have met the learning intention for this lesson.

1. Define the term phenotype.
2. Describe the components that make up genes.
3. Outline an example of a phenotype that is controlled by alleles found in humans.
4. Explain the relationship between homologous chromosomes and alleles.
5. All humans have 99.9% of their DNA in common. You and your friends have only 0.1% of a difference in your DNA code. Explain why humans have different characteristics despite 99.9% of their DNA being the same.

1.5 Mitosis and meiosis

Learning intention

To understand the role of mitosis and meiosis

Success criteria

SC 1: I can describe the purpose and process of mitosis.

SC 2: I can describe the purpose and process of meiosis.

KEY TERMS

zygote the cell formed by fusion of two gametes, a sperm and egg

cell cycle the events in the life of a cell, from its formation by cell division through its growth and function until it divides again

mitosis the type of cell division that produces two daughter cells identical to the parent cell

cell division the process by which a single cell divides into two or more identical daughter cells

interphase the phase in the cell cycle when the cell is not undergoing mitosis

Lesson overview

Cells replicate (make copies of themselves) so that an organism can grow, regenerate and repair. Cells also reproduce and divide so that individuals can produce offspring. They do this by replicating the deoxyribonucleic acid (DNA) that they contain.

In this lesson you will learn about two ways that cells divide: mitosis and meiosis.

SC 1 I can describe the purpose and process of mitosis

Cell division

You started out as a **zygote** (a fertilised egg). Over time, you developed into a complex organism through a series of processes. Throughout your life, your body will continue to grow, repair and regenerate by making new cells from pre-existing cells through a process known as the **cell cycle**. Part of this cell cycle is **mitosis**, a type of **cell division**.

Cell cycle

The cell cycle is the process in which a cell divides and makes two new cells. To replicate in this way, a cell must go through a series of stages, including mitosis (Figure 1.5.1).

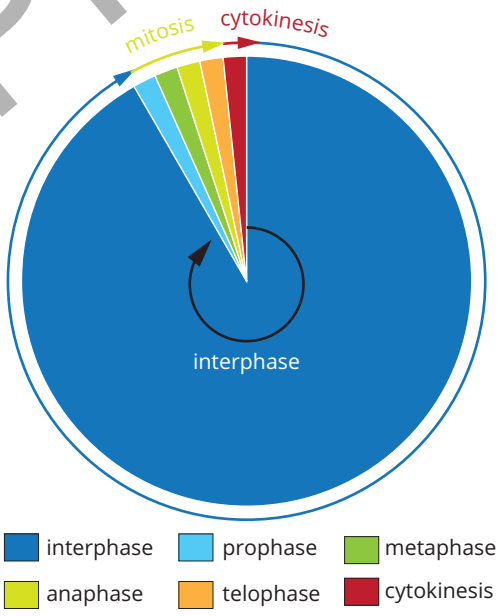


FIGURE 1.5.1 The cell cycle: interphase takes the longest time, followed by the four stages of mitosis, and then cytokinesis.

Interphase

Interphase is the stage when the cell is not dividing but is preparing for mitosis. During this time, the cell replicates its DNA, producing two identical chromatids. Interphase also regulates the rate of cell division. This is the longest phase of the cell cycle and is divided into three stages: G1 (preparation), S (DNA replication) and G2 (preparation).

M phase

The M phase (mitosis phase) is when the cell undergoes mitosis, dividing the two copies of the cell's genetic material into two new identical **daughter cells**.

KEY TERMS

daughter cell a new cell formed by cell division.
asexual reproduction a form of reproduction where only one parent cell produces offspring without joining of sperm and eggs
cytokinesis the separation of a cell following mitosis or meiosis, when the cytoplasm divides and the cell splits into daughter cells
centriole barrel-shaped structure made from microtubes that is part of the cytoskeleton in animal cells

Mitosis

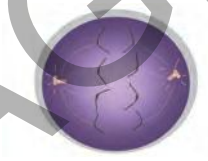
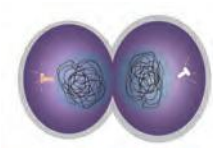
Mitosis is the next stage of the cell cycle after interphase. During mitosis, a cell separates its duplicated chromosomes to create two identical nuclei.

Mitosis allows cells to grow, repair tissue damage, regenerate tissue (in some organisms) and enable **asexual reproduction** (in some organisms). Mitosis results in two genetically identical daughter cells and can be used to replace cells that are old, dead or damaged.

Stages of mitosis

Scientists have identified four key stages of mitosis: prophase, metaphase, anaphase and telophase (Table 1.5.1). After mitosis comes **cytokinesis**, which is the physical process that finally splits the cell into two identical daughter cells. Each daughter cell contains one of the two nuclei created during mitosis.

TABLE 1.5.1 A summary of the stages of mitosis

Stage of mitosis	Diagram	Processes that occur in this stage
Prophase		• chromosomes and their replicated chromatids become visible • nuclear membrane breaks down • **centrioles** start to form spindle fibres
Metaphase		• replicated chromosomes line up along the equator (centre) of the cell • spindle fibres attach to the centromere of each chromosome
Anaphase		• chromatids separate and move to opposite poles (ends) of the cell • spindle fibres separate the sister chromatids (two identical halves of each chromosome) and move them to opposite ends of the cell
Telophase		• two nuclei form, each with the same number of chromosomes as the parent cell • after mitosis, cytokinesis completes the process of cell division by dividing the cytoplasm to make two identical daughter cells, each containing its own nucleus; note: cytokinesis (division of the cytoplasm) is a separate process to mitosis (division of the nucleus)

Chromosome number

In human somatic cells there are 46 chromosomes (23 pairs of chromosomes). An italic, lowercase letter '*n*' is used to indicate one full set of chromosomes. One chromosome in each pair is inherited from your biological mother; the other chromosome in each pair is inherited from your biological father.

There are 22 pairs of autosomes and one pair of sex chromosomes in every human somatic cell. These cells are said to be **diploid** (*2n*, indicating two full sets of chromosomes).

KEY TERMS

diploid the number of chromosomes in body cells; two sets or 2*n*

meiosis the type of cell division that produces gametes with half the number of chromosomes of the parent cell

crossing over the exchange of genetic material between homologous chromosomes during prophase I of meiosis

independent assortment the random allocation of homologous chromosomes to the spindle fibre during meiosis I of meiosis

genetic variation differences in the sequences of genes or alleles between individual organisms

SC 1 CHECK YOUR UNDERSTANDING

List the stages of the cell cycle in order. In your response, include the specific stages of mitosis.

SC 2 I can describe the purpose and process of meiosis

Meiosis

Meiosis is another type of cell division. Meiosis is the process that produces gametes (egg and sperm cells) in sexual reproduction.

Meiosis consists of two rounds of cell division, known as meiosis I and meiosis II.

Meiosis starts with one diploid (*2n*) cell. This diploid cell goes through the phases of meiosis I and meiosis II to produce four non-identical and genetically unique daughter cells (Figure 1.5.2).

Meiosis is a unique process in sexually reproducing organisms. During prophase I, a process known as **crossing over** occurs. This is where genetic material is exchanged between homologous chromosomes producing unique genetic combinations. This process is followed by **independent assortment** in metaphase I, where homologous chromosomes line up randomly. Together, these two processes create **genetic variation**, making the resulting daughter cells genetically unique.

In meiosis, as the daughter cells (gametes) are genetically different to the parent cell and contain only one copy of each chromosome, they are known as haploid (*n*, indicating one full set of chromosomes).

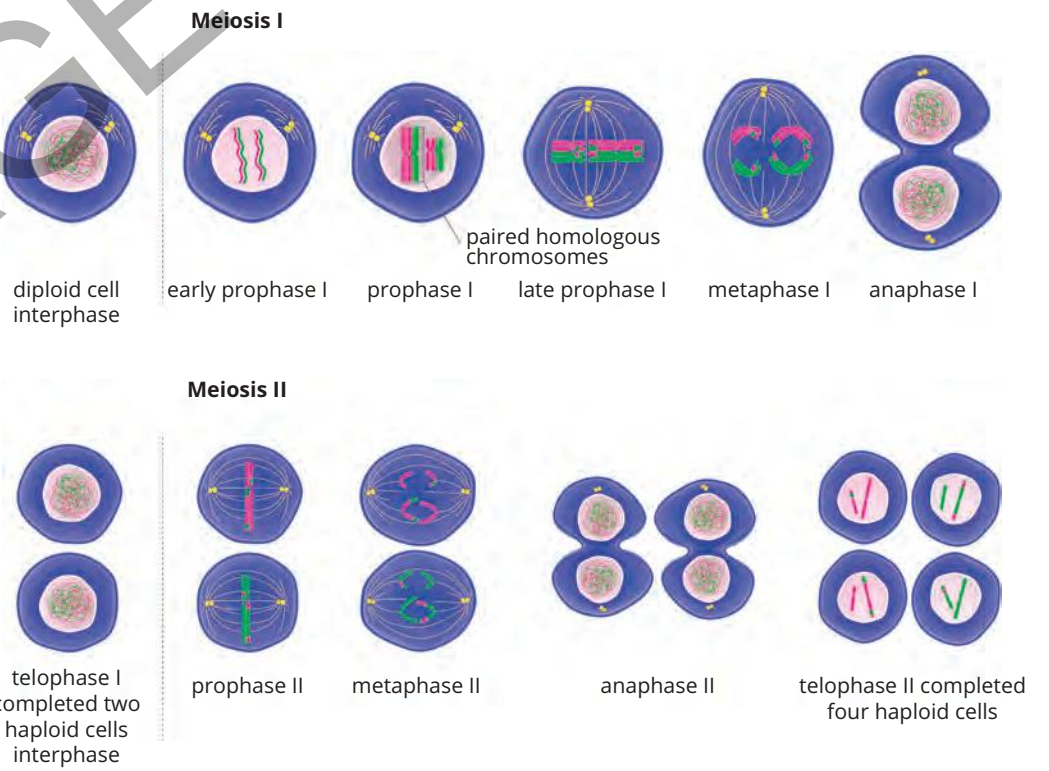


FIGURE 1.5.2 Through the process of meiosis, one diploid cell produces four genetically different haploid cells.

Comparing mitosis and meiosis

Table 1.5.2 compares the role of mitosis and meiosis in the human body.

TABLE 1.5.2 Comparison of mitosis and meiosis in the human body

	Mitosis	Meiosis
Reproduction type	asexual reproduction	sexual reproduction
Stages	a single stage process that keeps the amount of genetic material the same	a two-stage process that reduces the amount of genetic material
Daughter cells	two identically daughter cells	four genetically different daughter cells
Ploidy daughter cells	diploid (2*n*)	haploid (*n*)
Purpose in humans	to enable growth and to repair and replace cells	to produce egg and sperm cells that have genetic variation

Making gametes

Most animals produce offspring though sexual reproduction. In humans, sexual reproduction occurs through the **fertilisation** of a female ovum (egg cell) with a male sperm cell.

KEY TERM

fertilisation the joining of gametes

Making eggs

In females, meiosis takes place in the ovaries to produce haploid egg cells in a process known as oogenesis. Oogenesis produces one haploid gamete (ovum) and three haploid nuclei (known as polar bodies), which degenerate and disappear naturally. This process can be seen in Figure 1.5.3.

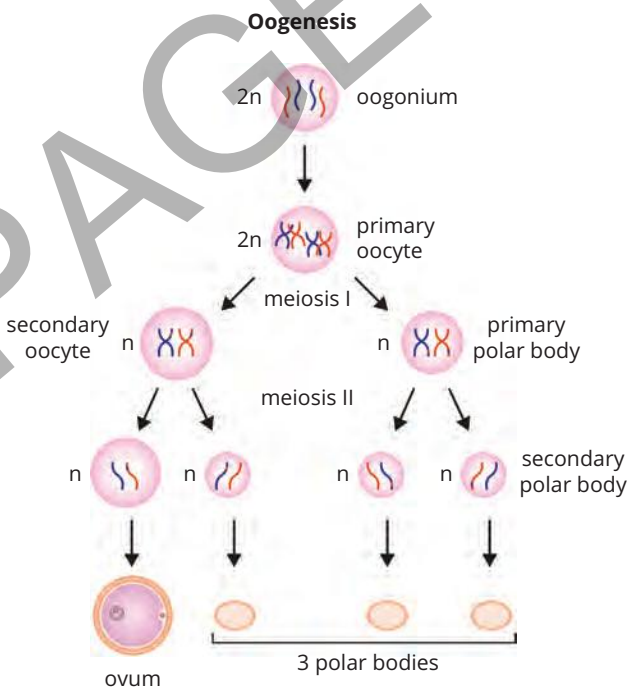


FIGURE 1.5.3 Two rounds of cell division take place during oogenesis, which occurs in the ovaries and produces ova (egg) cells.

Making sperm

In males, meiosis takes place in the testes to produce four haploid gametes (sperm cells) in a process known as spermatogenesis (Figure 1.5.4).

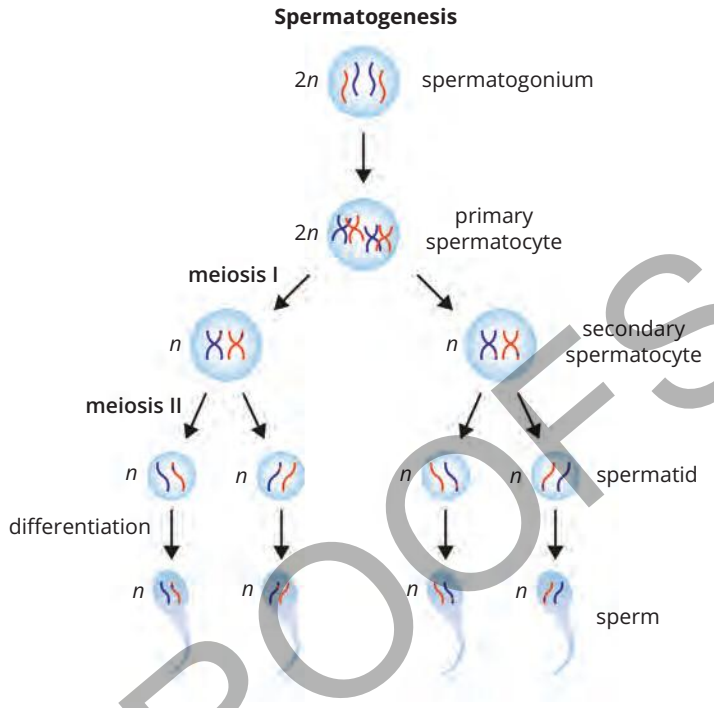


FIGURE 1.5.4 Two rounds of cell division take place during spermatogenesis, which occurs in the testes and produces sperm cells.

SC 2 CHECK YOUR UNDERSTANDING

Describe the number of cells produced by meiosis and their defining features.

Lesson review

Use these questions to check whether you have met the learning intention for this lesson.

1. Identify the three stages of interphase.
2. State the name of the process that occurs following mitosis that separates the cells.
3. Outline the main purpose of mitosis.
4. Meiosis is critical for sexual reproduction and genetic variation.
   a. List the stages of meiosis.
   b. Explain the purpose of meiosis in sexually reproducing organisms.
   c. Explain how meiosis contributes to genetic variation in offspring.
5. Compare the outcomes of mitosis and meiosis.

1.6 Outcomes of meiosis

Lesson overview

Meiosis is an important type of cell division. It ensures that gametes only contain half the amount of genetic material as the parent cells. Gametes are different from other cells in your body. Somatic (body) cells are diploid and contain 23 pairs of chromosomes. Gametes are haploid and contain only half the number of chromosomes.

In this lesson you will learn about the role that meiosis plays in genetic variation between generations – which is why you are not a clone of your parents (Figure 1.6.1)!

SC 1 I can explain allele variation in gametes as an outcome of meiosis

Learning intention

To understand the genetic outcomes of meiosis

Success criteria

SC 1: I can explain allele variation in gametes as an outcome of meiosis.

SC 2: I can define the terms homozygous and heterozygous.

SC 3: I can use a Punnett square to predict possible genotypes.

FIGURE 1.6.1 The two daughters in this family share some features from both of their parents.

Allele variation in gametes

As a result of meiosis, each new gamete is genetically unique to the original cell and contains only half of the genetic material as the parent cell. This allows a zygote (fertilised egg) to be diploid (*2n*, indicating two full sets of chromosomes). As seen in Figure 1.6.2 each parent passes on 22 autosomes and one sex chromosome, and the resulting zygote has 46 chromosomes in total.

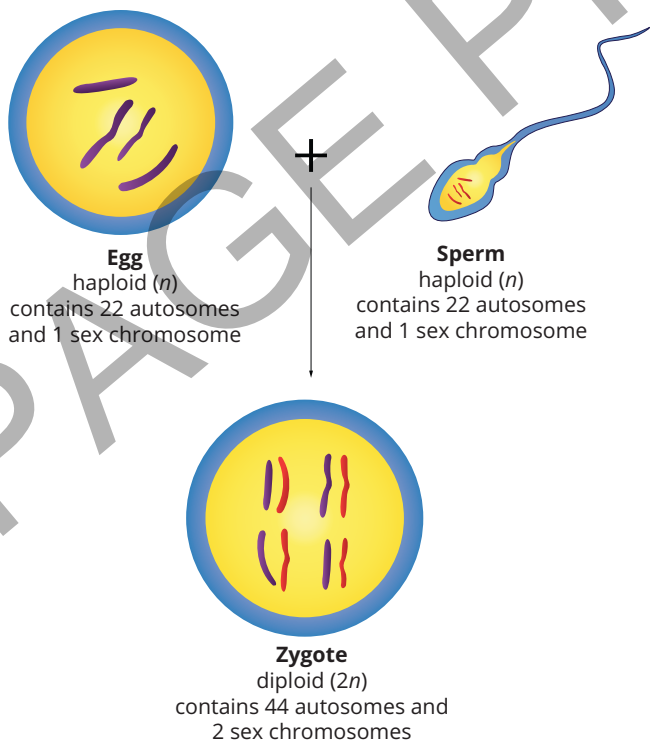


FIGURE 1.6.2 The union of two haploid (*n*) gametes (egg and sperm cells) produces a diploid (*2n*) zygote that can develop into an individual.

Mendel's law of segregation

KEY TERMS

law of segregation one of the two alleles from the parental genotype is present within the gamete cell

chiasma a point where two chromosomes exchange genetic material during meiosis

As chromosomes separate into different gametes during meiosis, the two different alleles that the parent has for a particular gene randomly segregate (separate) so that only one of the two alleles goes into each gamete. This is known as Mendel's **law of segregation**. The daughter cells that are produced in meiosis are not genetically identical, which creates genetic variation in individuals and in the species.

Crossing over

Crossing over is another process that occurs during meiosis and creates further genetic variation. During prophase I, homologous chromosomes exchange genetic material (cross over) at a point called a **chiasma**. At the chiasma, chromatids break and rejoin, exchanging some of their genes. Chromosomes may exchange (swap) entire genes or smaller segments of DNA when they cross over (Figure 1.6.3). This creates new combinations of genetic material (called recombinant chromatids) in the chromosomes of the daughter cells, which results in genetic variation of alleles in the gametes.

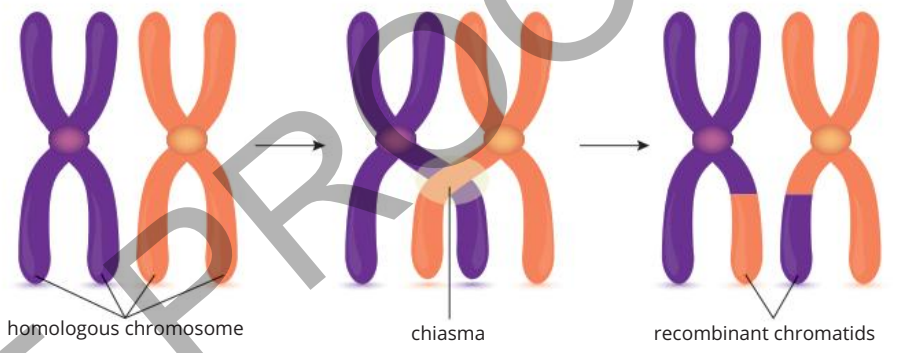


FIGURE 1.6.3 A pair of homologous chromosomes crossing over and the resulting genetic variation in the new chromosomes.

FIGURE 1.6.4 A light micrograph showing chromosomes crossing over during meiosis. Look for the figure-8-shaped structures: this is where the chromatids are linked together, which indicates that crossing over is occurring.

Allele variation in gametes

Meiosis makes genetically different cells from the original parent cells, because they will become gametes. When alleles are crossed over and independently assorted before they are segregated into gametes, the genes can be shuffled around. This crossing over of genetic material (Figure 1.6.4) is important as it ensures that there is a high chance of each individual within the next generation being different, and those individuals will also be different from the generation before.

SC 1 CHECK YOUR UNDERSTANDING

State the name of the point at which homologous chromosomes exchange genetic material.

SC 2 I can define the terms homozygous and heterozygous

During meiosis, gametes with different genetic combinations of alleles are created. When the egg and the sperm come together it creates a unique combination of alleles, and this creates the physical traits that you see.

If both your parents have brown hair, could you end up with red hair? What about if both parents have brown eyes; will you have brown eyes too? This depends on the genes you inherit, and more specifically the alleles of each gene. Because somatic cells contain two full sets of chromosomes, each gene appears twice in each cell.

From genetic code to physical expression

An individual's genetic code determines their physical traits. All of the genetic information within an individual is called their genotype.

Each allele is made up of a unique genetic sequence that produces a physical trait known as a phenotype.

Alleles for a particular trait may be either dominant or recessive (non-dominant). If an individual carries a dominant allele for a trait (that is, one or both of their alleles is the dominant allele), then the **dominant trait** will be expressed in their phenotype. The **recessive trait** is only expressed when the individual inherits two copies of the recessive allele (one from each parent).

Which allele is in control?

Alleles can be represented by letters to describe the different forms that a gene can take and the different genotype for certain traits:

- Dominant alleles are represented by uppercase letters. For example, *R* may be used to represent the allele for the presence of dimples, which is a dominant genetic trait (Figure 1.6.5).
- Recessive alleles are represented by the same letter in lowercase. For example, *r* may be used to represent the allele for not having dimples.

When an individual has two copies of the same allele in their genotype, they are **homozygous**. When they have two different versions of the allele, they are **heterozygous** (Figure 1.6.6). In genetics, the term genotype is often used when writing the possible allele combinations that an individual has.

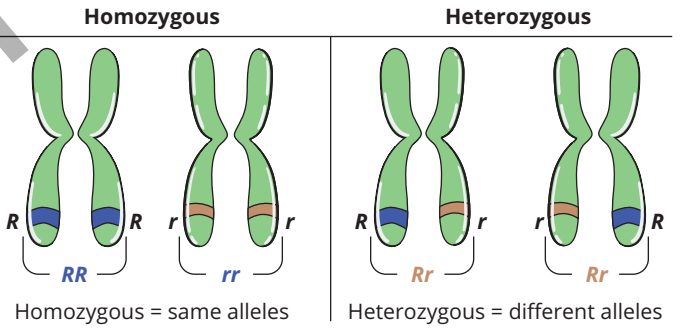


FIGURE 1.6.6 This diagram shows homozygous and heterozygous genotypes for dominant traits (*R*) and recessive traits (*r*).

KEY TERMS

dominant trait the trait that is observed in the outward appearance of a heterozygous individual

recessive trait the trait that remains hidden in the heterozygous condition and seen only in the homozygous condition

homozygous having two identical alleles on homologous chromosomes

heterozygous having two different alleles on homologous chromosomes

FIGURE 1.6.5 The presence of dimples is primarily a genetic trait; whether you have this depends on the alleles that you inherited from your biological parents.

Scifile

Inherited traits

Most human genetic traits are controlled by multiple genes and, in some cases, the environment. For example, tongue rolling is often described as a genetic trait that can be predicted from generation to generation. However, scientists now know that multiple factors are involved, and that simple patterns of inheritance may not be observed.

HINT

Terminology tip

Many biological words come from Greek and Latin languages. In genetics, the prefixes *homo-* and *hetero-* are from the Greek: *homo-* meaning 'the same' and *hetero-* meaning 'different'.

There are three possible genotypes for the autosomal genetic traits in Figure 1.6.6. Any of the following may occur:

- homozygous dominant (two dominant alleles, *RR*)
- heterozygous (one dominant allele and one recessive allele, *Rr*)
- homozygous recessive (two recessive alleles, *rr*).

Mendel's law of dominance states that the dominant allele will be expressed in the phenotype of a heterozygous individual. Having dimples is a dominant trait, which means that, if you have dimples, then at least one of your two alleles codes for dimples (*RR* or *Rr*). A person who does not have dimples can only have both recessive alleles (*rr*).

SC 2 CHECK YOUR UNDERSTANDING

Define the terms homozygous and heterozygous.

SC 3 I can use a Punnett square to predict possible genotypes

KEY TERMS

Punnett square a visual representation of the genotypes of a genetic cross and the possible genotypes of their offspring

genetic cross the breeding of two individuals to produce offspring that are representative of the parents' genetic material

Predicting genotypes with Punnett squares

In 1905, Reginald Punnett devised a diagram that could be used as a tool to predict the genotypes of the offspring of two parent individuals with known genotypes. The diagram was named a **Punnett square** in his honour.

What is shown in a Punnett square?

A Punnett square is a table that shows all the possible outcomes for a **genetic cross** (sexual reproduction) between two individuals with known genotypes.

The genotype of each parent is shown on the outside of the Punnett square (Figure 1.6.7). The two alleles for the female parent are written at the top of the two columns (top of the square), and the two alleles for the male parent are written to the left of the two rows (left of the square).

The possible outcomes for each genetic combination are shown in the middle squares.

The convention (rule) is that dominant alleles are written first (for example, you write *Tt* , not *tT*).

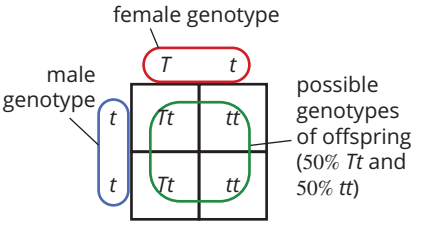


FIGURE 1.6.7 Punnett square showing the possible genotypes of offspring in a genetic cross.

SkillBuilder

Punnett squares

If two parents' genotypes are known, a Punnett square can be used to predict the genotypes of their offspring. Genotypes can be homozygous dominant, homozygous recessive or heterozygous.

In the Punnett square to the left (Figure 1.6.7), a heterozygous female for the allele *T* is crossed with a homozygous recessive male for the allele *t*:

- heterozygous female genotype: *Tt*
- homozygous recessive male genotype: *tt*

The possible genotypes for the offspring of the cross between these two individuals are:

- heterozygous (*Tt*), 50% chance, phenotype *T* (dominant phenotype)
- homozygous recessive (*tt*), 50% chance, phenotype *t* (recessive phenotype).

It is important to remember that the genotypes of any offspring produced are independent of each other. The probability of each genotype occurring in each cross between two individuals is independent of (not influenced by) other crosses between those individuals.

Worked example

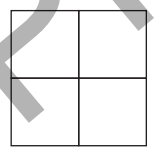
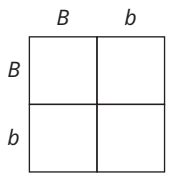
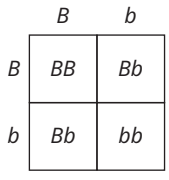
Punnett squares

In this worked example you will learn how to predict the genotypes of offspring using a Punnett square.

Problem

Broad lips (*B*) are a dominant genetic trait. Two heterozygous parents have offspring. Predict the probability of the offspring having broad lips.

Solution

Thinking	Working
Write the parents' genotypes.	Both parents are heterozygous, so they both have the genotype *Bb*.
Draw a Punnett square.	
Write each parent's genotype on the Punnett square.	
Complete the Punnett square.	
Write the ratio for each possible genotype in the offspring.	*BB*:*Bb*:*bb* = 1 (25%):2 (50%):1 (25%)

Try yourself

Punnett squares

Predict the genotypes for the dominant trait of brown hair (*B*) for the offspring of a heterozygous parent and a homozygous recessive parent.

SC 3 CHECK YOUR UNDERSTANDING

Freckles are a dominant trait. Draw a Punnett square to show a genetic cross between a homozygous dominant individual and a heterozygous individual. Use the letter *'A'* to show the gene variants.

Lesson review

Use these questions to check whether you have met the learning intention for this lesson.

1. State the genotype of each individual, using the letter *G*, for:
   - **a** a heterozygous individual
   - **b** a homozygous dominant individual
   - **c** a homozygous recessive individual.
2. State the steps taken to construct a Punnett square.
3. Describe the process of crossing over.
4. Earlobe attachment has two different variations. Amara has detached earlobes, and she has the genotype *AA*.
   - **a** Are detached earlobes a dominant or recessive condition?
   - **b** Determine the possible combination of genotypes that Amara's parents could be.
5. Compare the predicted genotypic ratios from a Punnett square of two homozygous individuals (*AA* × *aa*) versus two heterozygous individuals (*Aa* × *Aa*).

1.7 Inheritance: Genotypes and phenotypes

Lesson overview

Eye colour varies among individuals and is influenced by genetics. Individuals inherit eye colour alleles from their parents, which contribute to their own eye colour. Genes are responsible for the characteristics you inherit. Alleles can be dominant, recessive, codominant, incompletely dominant or sex-linked (located on one of the sex chromosomes).

In this lesson you will learn about dominant, recessive and sex-linked inheritance, and how to use Punnett squares to predict genotypic and phenotypic ratios of offspring.

SC 1 I can use dominant and recessive patterns of inheritance to predict the phenotype of an individual based on their genotype

Learning intention

To understand how genetic material is expressed in the characteristics of organisms and to be able to predict phenotypes

Success criteria

SC 1: I can use dominant and recessive patterns of inheritance to predict the phenotype of an individual based on their genotype.

SC 2: I can explain sex-linked inheritance.

SC 3: I can predict the genotypic and phenotypic ratios of offspring in a monohybrid cross.

KEY TERMS

codominance a genetic trait where both alleles in a pair are fully expressed in the phenotype, resulting in a combined trait rather than one allele being dominant over the other

incomplete dominance when neither allele in a gene pair is completely dominant over the other, resulting in a phenotype that is an intermediate between the two parental phenotypes

sex-linked related to genes that occur on the sex-chromosomes (X and Y in humans)

Types of inheritance

In a Punnett square, alleles from each parent's genotype are used to predict the possible genotype (and hence, phenotype) of any offspring.

Alleles can show patterns of inheritance such as dominance, **codominance**, **incomplete dominance**, or be **sex-linked** (located on one of the sex chromosomes).

This lesson will explore alleles that are dominant, recessive, and sex-linked.

Dominant and recessive alleles

Your genotype is a record of the alleles you have for each gene; your phenotype is how your genotype is expressed. Having freckles is the dominant phenotype (*F*) for facial skin, and non-freckles is recessive (*f*). If you have the freckled phenotype, you will be homozygous dominant (*FF*) or heterozygous (*Ff*). If you have non-freckled phenotype, your genotype must be homozygous recessive (*ff*).

If you know the genotypes of two parents, then it is possible to use a Punnett square to predict the genotypes of their offspring.

For example, George has no freckles (so he must have the genotype *ff*). Kristine has freckles with a heterozygous genotype (*Ff*). The Punnett square below (Figure 1.7.1) illustrates the possible genotypes of any of their offspring.

	F	*f*
f	*Ff*	*ff*
f	*Ff*	*ff*

FIGURE 1.7.1 Punnett square for a non-freckled male (*ff*) and freckled heterozygous female (*Ff*).

If two non-freckled people were to produce offspring, then genetically, all the offspring would also be non-freckled as seen in Figure 1.7.2.

	f	*f*
f	*ff*	*ff*
f	*ff*	*ff*

FIGURE 1.7.2 All offspring of this cross will be non-freckled (*ff*).

It is important to remember that the genotypes of any offspring produced are independent of each other. The probability of each genotype occurring in each cross between two individuals is independent of (not influenced by) other crosses between those individuals.

SC 1 CHECK YOUR UNDERSTANDING

Sidrah and Amir are expecting a new baby. Sidrah and Amir both have freckles and are both heterozygous. Predict the possible phenotypes of their new baby.

SC 2 I can explain sex-linked inheritance

Sex chromosomes

The sex chromosomes in females are XX, and in males they are XY. Sex chromosomes also contain a number of genes that are not associated with biological sex. Genes that code for any trait are called sex-linked if they are located on a sex chromosome (X or Y). The X chromosome is much larger than the Y chromosome and contains many more genes. Sex-linked conditions are usually found on the X chromosome and are said to be **X-linked**. Not many genes are said to be Y-linked because very few genes exist on the shorter Y chromosome. However, there are some genetic conditions that are Y-linked (the gene responsible for the condition is located on the Y chromosome), but they are much less common than X-linked conditions. Y-linked traits are only inherited by males because only males have a Y chromosome.

KEY TERM

X-linked related to genes on the X chromosome

X-linked traits

If a condition is X-linked, it means that the gene responsible for the condition is located on the X chromosome (Figure 1.7.3). These traits can either be X-linked recessive or X-linked dominant.

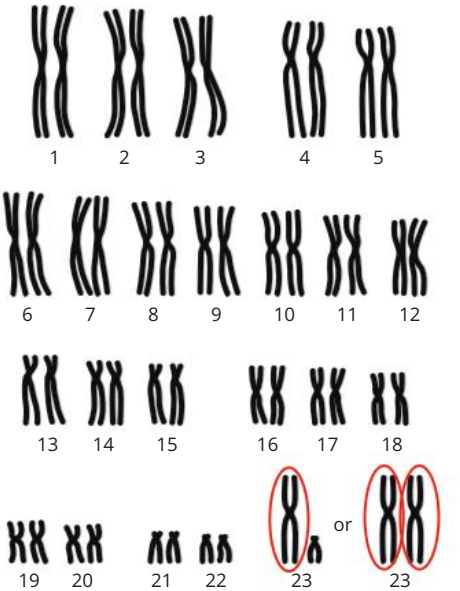


FIGURE 1.7.3 The X chromosome in the karyotype can be seen circled in red; this is the location of X-linked genes.

In X-linked dominant traits, the individual only needs one copy of the dominant allele to show the phenotype. X-linked recessive traits must have two copies of the recessive allele to show the phenotype.

Most X-linked traits are recessive. Males are more likely to be affected by X-linked recessive traits because males only need to inherit one copy of an X-linked recessive allele for it to be expressed in their phenotype. Females must inherit two copies of an X-linked recessive allele for the trait to be expressed in their phenotype.

	X	X
X	XX	XX
Y	XY	XY

FIGURE 1.7.4 Punnett square showing the likelihood of a zygote (fertilised egg) having an XX (female) or XY (male) pair of chromosomes.

X and Y chromosomes

If you think of an individual's sex chromosomes as a pair, you can use a Punnett square to consider the chance of offspring having either an XX or XY genotype. All ova (egg cells) contain an X chromosome. Sperm cells contain either an X or a Y chromosome. This means that each time a sperm fertilises an egg, known as a zygote, there is a one in two (50%) chance that the offspring is female (XX) and the same chance that the offspring is male (XY) (Figure 1.7.4).

Sex-linked alleles

Genetics focuses on the X chromosome as both males and females have this chromosome and it carries a wide range of genes that are not only used for sexual characteristics (for example, colour-blindness (Figure 1.7.5). X-linked genes can be either dominant or recessive.

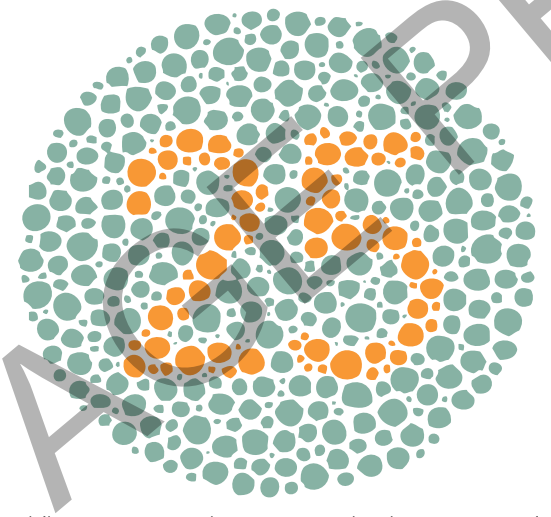


FIGURE 1.7.5 An Ishihara test, used to assess whether a person is colourblind (an X-linked trait).

You can show that a condition is X-linked by adding the allele as a superscript letter. For example, a male who has X-linked recessive colour blindness would have the genotype X^dY. A female will only be affected if she inherits two copies of the allele for colour-blindness, with the genotype X^dX^d.

Carriers

When a female has the genotype X^DX^d, she is said to be a carrier of colour blindness. This means that she has one allele for colour blindness (X^d) but does not have the condition herself. However, she can pass on the X^d allele to her offspring (Figure 1.7.6).

Scifile

X-linked muscular dystrophy

Muscular dystrophy is a group of genetic conditions that cause progressive weakness and loss of muscle mass. Duchenne muscular dystrophy (DMD) is a common form of muscular dystrophy that is X-linked recessive. Muscular dystrophy causes muscle weakness and wasting. People who have this condition often need to use a wheelchair.

	X^D	X^d
X^D	X^DX^D	X^DX^d
Y	X^DY	X^dY

FIGURE 1.7.6 Punnett square showing a cross between a female carrier of colour blindness X^DX^d and an unaffected male X^DY.

The possible outcomes of this cross between a female carrier and an unaffected male include unaffected female and male offspring (X^DX^D or X^DY), female carriers (X^DX^d) and males with the condition (X^dY). Note that if a trait was said to be X-linked dominant then affected individuals would have capital letters in superscript.

SC 2 CHECK YOUR UNDERSTANDING

Sex-linked traits are more common in males than females.

a Outline the defining feature that makes a trait sex-linked.

b Explain why males are more likely to be affected by X-linked traits than females.

SC 3 I can predict the genotypic and phenotypic ratios of offspring in a monohybrid cross

KEY TERMS

genotypic ratio expected frequency of genotypes in the offspring of a genetic cross
phenotypic ratio expected frequency of phenotypes in the offspring of a genetic cross
monohybrid cross a genetic cross between a homozygous dominant individual and a homozygous recessive individual

Predicting genotypic and phenotypic ratios

Punnett squares allow you to predict the potential genotypes, and therefore also the phenotypes, of offspring. **Genotypic ratios** and **phenotypic ratios** can be predicted using a Punnett square in a monohybrid cross. The ratios show the proportion of phenotypes or genotypes in the offspring. Monohybrid crosses are the most common type of cross used in genetics.

Monohybrid cross

A **monohybrid cross** is a cross between two individuals. Monohybrid crosses are used to examine one specific trait that is determined by one gene (*mono-* meaning one).

In a standard monohybrid cross, a homozygous dominant parent (e.g. *AA*) is crossed with a homozygous recessive parent (e.g. *aa*) to produce heterozygous offspring (e.g. *Aa*). This is called the first generation (F1). The heterozygous offspring are then crossed to produce the second generation (F2).

For example, in guinea pigs, black fur (*B*) is dominant to white fur (*b*). If a guinea pig that is homozygous for black fur (*BB*) is crossed with a guinea pig that is homozygous for white fur (*bb*), all the offspring in the F1 generation will be heterozygous with black fur (*Bb*) (Figure 1.7.7).

	B	*B*
b	*Bb*	*Bb*
b	*Bb*	*Bb*

FIGURE 1.7.7 Punnett square showing a monohybrid cross between a homozygous black fur guinea pig (*BB*) and a homozygous white fur guinea pig (*bb*).

F1 generation analysis:

- Parents – $BB \times bb$
- F1 genotypes – 100% *Bb*
- F1 phenotypes – 100% black fur

If the offspring of the F1 generation are crossed (*Bb*) to produce the F2 generation (Figure 1.7.8).

	B	*b*
B	*BB*	*Bb*
b	*Bb*	*bb*

FIGURE 1.7.8 Punnett square showing the outcomes of a cross between two F1 generation heterozygous parents.

F2 generation analysis:

- Parents – $Bb \times Bb$
- F2 genotypes – 25% *BB*, 50% *Bb*, 25% *bb*
- F2 phenotypes – 75% black fur, 25% white fur

Genotypic and phenotypic ratios

As genotypic and phenotypic ratios are the proportion of genotypes and phenotypes shown in the offspring, using Figure 1.7.8 it could be determined that:

The genotypic ratio of the F2 is shown by the percentage of each genotype. As the F2 genotypes are 25% *BB*, 50% *Bb*, 25% *b*, a common factor of 25 can be used to obtain a ratio of 1 : 2 : 1 (*BB* : *Bb* : *bb*).

The phenotypic ratio follows the same idea with 75% black fur, 25% white fur and a common factor of 25. The ratio obtained is 3 : 1 (black : white) phenotypic ratio (Figure 1.7.8).

SC 3 CHECK YOUR UNDERSTANDING

Describe the characteristics of a standard monohybrid cross.

Lesson review

Use these questions to check whether you have met the learning intention for this lesson.

1 State the name given to the diagram used to determine the chance of specific genotypes being produced by a genetic cross.

2 List the possible genotypes of a female and a male that are both affected with an X-linked recessive condition that is denoted with the superscript letter *D* or *d*.

3 Predict the phenotypic ratio of offspring from a monohybrid cross between a heterozygous tall pea plant and a homozygous recessive pea plant. Assume *T* is the allele for tall and *t* is the allele for short.

4 Rett syndrome is a dominant sex-linked dominant trait carried on the X chromosome.

- **a** List the possible genotypes of males and females with Rett syndrome.
- **b** Draw a diagram of a genetic cross between a female without Rett syndrome and a male with Rett syndrome.
- **c** Describe the possible offsprings produced from the cross in part b.

5 The offspring of two beetles produced two different colour variations, red and green. There were 50 red beetles and 155 green beetles counted.

- **a** Write the phenotypic ratio of the beetles produced.
- **b** Using the letters *B* and *b*, determine the most likely genotypes of the red and green beetles.
- **c** Predict the genotypes of the parent beetles.

1.8 Modelling genetic variation

Learning intention

To be able to model genetic variation

Success criteria

SC 1: I can construct and use appropriate representations to demonstrate genetic variation.

SC 2: I can analyse collected data to explain genetic variation within the data.

Introduction

The genetic makeup of offspring is determined by the genes in the chromosomes of their parents.

Genes, which are sections of deoxyribonucleic acid (DNA), control characteristics. Humans have two versions of each gene: one inherited from each parent. These are called alleles, and both parents randomly pass on one allele for each gene to their offspring. Therefore, which allele is passed on from each parent is a matter of chance. The DNA in a fertilised egg (zygote) will be present in every future cell of that organism (Figure 1.8.1).

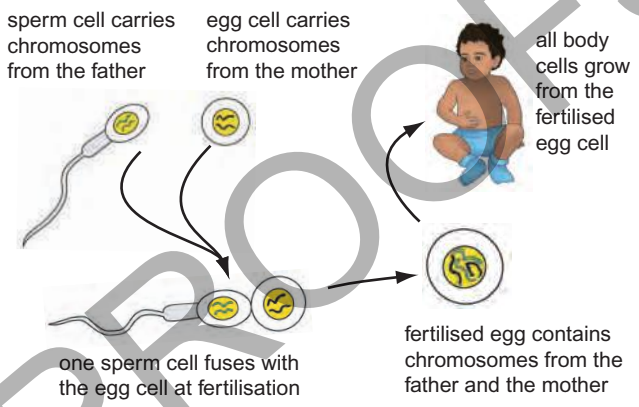


FIGURE 1.8.1 The combination of chromosomes from the parents in a child: the fertilised egg (zygote) contains two versions of each gene (alleles) to produce a unique individual.

In this practical investigation, you will simulate the random processes that govern genetic inheritance through a card game. You will create cards to represent the alleles for five traits in two individuals, and use a die to model meiosis (the process by which alleles are randomly selected to form gametes). These gametes will combine to form a zygote, which will represent the offspring. Finally, you will model fertilisation by combining the alleles to determine the potential genotypes and phenotypes of the offspring.

This game will allow you to explore how genetic variation occurs by analysing five traits: eye colour, the presence of freckles, finger shape, lip shape, and dimples.

Background

In sexually reproducing organisms, the individual begins life as a zygote — a single fertilised ovum (egg) cell. The zygote's nucleus contains DNA that influences the organism's traits, such as biological sex and eye colour, as well as other traits specific to humans, like hair colour and the presence of dimples.

A pair of homologous chromosomes carries two versions (alleles) of each gene. An individual inherits one allele from each parent. Dominant alleles are represented by capital letters, while recessive alleles for the same trait are written as lowercase letters. When gametes fuse during fertilisation, genetic material from both parents combines, introducing genetic variation.

Aim

To use cards to model the genetic variation in offspring

Materials

- dice
- 20 cards approximately the size of playing cards
- marker pen

Method

Part 1: Creating the cards

1 Use the information in this table to make 20 cards. Make two identical cards for each allele.

Trait	Allele 1	No. of cards	Allele 2	No. of cards
eye colour	brown eyes (*T*)	2	blue eyes (*t*)	2
freckles	freckles (*F*)	2	no freckles (*f*)	2
little finger	bent little finger (*B*)	2	straight little finger (*b*)	2
shape of lips	broad lips (*L*)	2	thin lips (*l*)	2
dimples	dimples (*D*)	2	no dimples (*d*)	2
	Total	10	**Total**	10

2 Divide the cards into two sets of 10. Each set should contain one copy of Allele 1 and one copy of Allele 2. Each set represents the genes of one parent. Label the cards in the first set **Parent 1** and label the cards in the second set **Parent 2**.

Note that this activity assumes that both parents are heterozygous for each of the genes that control these five traits, which means that they have one dominant allele and one recessive allele in each pair. Each set of 10 cards should contain five heterozygous pairs of alleles, as shown in the table below.

	Pair 1	Pair 2	Pair 3	Pair 4	Pair 5
Card 1	*T*	*F*	*B*	*L*	*D*
Card 2	*t*	*f*	*b*	*l*	*d*

3 In your results, list the phenotype that each parent has for each of the five traits.

Part 2: Creating gametes

4 Start with Trait 1 (Pair 1) for Parent 1.

- Roll the die.
- If you roll an even number, select the card for the dominant allele (uppercase letter).
- If you roll an odd number, select the card for the recessive allele (lowercase letter).
- Continue rolling and selecting cards until you have selected one allele for each of the five traits for the Parent 1 gamete.
- Place the cards that you have selected for this gamete in a pile and put them aside. This step represents meiosis.

5 Repeat Step 4 for Parent 2 to select the alleles for a second gamete. Place these cards in a separate pile.

Part 3: Creating offspring

6 Take the two piles of cards (gametes) that you have created and arrange the cards to pair the two alleles for each trait. These pairs represent the genotypes for each of the five traits in the first zygote (offspring). This step represents what happens during fertilisation of an ovum (egg) with a sperm.

7 In your results, record the genotypes and phenotypes of this offspring for each of the five traits.

8 Return all the cards to the two sets (Parent 1 and Parent 2). Repeat Steps 4 to 7 to generate genotypes and phenotypes for three more offspring.

Results

1 Complete a table like this in your notebook to record the phenotype of each parent. The first one has been completed for you.

Trait	Parent 1		Parent 2	
	Genotype	Phenotype	Genotype	Phenotype
1	*Tt*	brown eyes	*Tt*	brown eyes

2 Complete a table like this in your notebook to record the genotypes and phenotypes for the four offspring.

Trait	Offspring 1		Offspring 2		Offspring 3		Offspring 4	
	Genotype	Phenotype	Genotype	Phenotype	Genotype	Phenotype	Genotype	Phenotype
1								

Conclusion

1. How does the method in this practical investigation model the processes of meiosis and fertilisation?
2. In this model you considered only five traits. How many combinations of alleles (genotypes) are possible for these five traits? Predict the number of possible combinations that would result if twice as many traits were modelled.
3. In step 8 of the method, you returned all the cards to the two sets (Parent 1 and Parent 2) before rolling the die again to select the alleles for the next gamete. Why?

Evaluation

1. Select one of the five traits you explored in this practical investigation.
   - **a** Draw a Punnett square to show the possible genotypes of the offspring of a cross between two heterozygous parents.
   - **b** What are the phenotypes of the offspring?
   - **c** What is the phenotypic ratio of the offspring?
   - **d** Does this phenotypic ratio match the results you recorded for this trait when modelling the same cross (see question 2 in the Results section)?
2. Evaluate the modelling process used in this practical investigation by reflecting on your responses to parts a to d above.

1.9 Pedigree diagrams

Learning intention

To understand how patterns of inheritance can be represented using a pedigree diagram

Success criteria

SC 1: I can identify the pattern of inheritance for dominant and recessive traits using a pedigree diagram.

SC 2: I can identify the pattern of inheritance for sex-linked traits using a pedigree diagram.

SC 3: I can develop a pedigree diagram to model inheritance of genes through families.

Lesson overview

Genetic traits (characteristics or conditions) tend to run in families. A pedigree diagram is like a family tree, but individuals are identified using symbols (shapes) instead of words. A pedigree diagram also shows whether an individual has a particular genetic trait.

In this lesson you will learn about pedigree diagrams and how they are constructed. You will learn how to use a completed pedigree diagram to determine whether a particular genetic trait is dominant or recessive and whether it is sex-linked or autosomal (not sex-linked).

SC 1 I can identify the pattern of inheritance for dominant and recessive traits using a pedigree diagram

A pedigree diagram uses symbols to show the genetic history of a family over several generations. Biological males are represented as square symbols, and biological females are represented as circle symbols.

Pedigree diagrams and charts

Each genetic trait can be represented in its own pedigree diagram (sometimes called pedigree charts; see Figure 1.9.1).

The key features of a pedigree diagram are:

- Males are shown as squares, and females are shown as circles.
- If an individual has the trait being studied, they will be represented on the pedigree diagram using a symbol that is shaded or coloured in. They are known as affected.
- Mating lines join individuals horizontally.
- Lines of descent go vertically. These show the offspring of a mating line.
- Each generation is labelled.

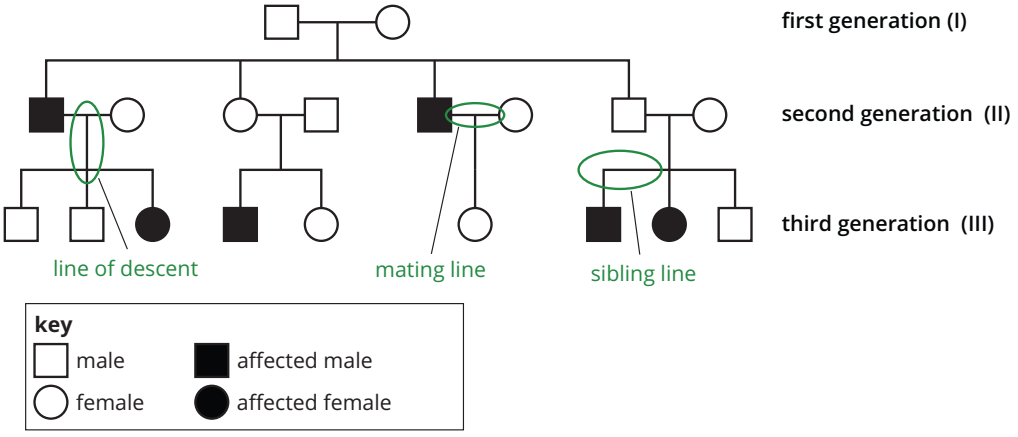


FIGURE 1.9.1 Pedigree diagram showing the pattern of inheritance of attached earlobes in a family.

Pedigree diagrams can be used to determine the pattern of inheritance of a certain trait. In the first generation shown in Figure 1.9.1, neither individual has the trait. However, there are two affected offspring in the second generation. This indicates that the trait is recessive and the individuals in the first generation are heterozygous. Heterozygous individuals do not have the trait themselves but carry one recessive allele for the trait and one dominant allele for the trait. Because two copies of the recessive allele are required for an individual to show a recessive trait, these individuals will not show the trait but can pass on the recessive allele to their offspring. If two individuals pass on their recessive alleles, the offspring will have two copies of the recessive allele (homozygous recessive) and will show the trait.

Punnett squares, genotypes and phenotypes

Punnett squares can be used to determine the genotype of each individual within a pedigree diagram. If you know the genotypes of two parents, you can use a Punnett square to determine the probability (likelihood) of each genotype occurring in their offspring.

Figure 1.9.2 is a Punnett square for two heterozygous (*Uu*) individuals with unattached earlobes (Figure 1.9.3). Unattached earlobes (Figure 1.9.4) is a dominant trait. The predicted genotypes and phenotypes of the offspring are 25% homozygous dominant, unattached lobes (*UU*), 50% heterozygous, unattached lobes (*Uu*) and 25% homozygous recessive attached lobes (*uu*) in a genotypic ratio of 1 : 2 : 1 (*UU* : *Uu* : *uu*) and a phenotypic ratio of 3 : 1 (unattached : attached).

This information can then be used to either make a pedigree or analyse a pedigree. Figure 1.9.3 shows the pedigree diagram for this cross.

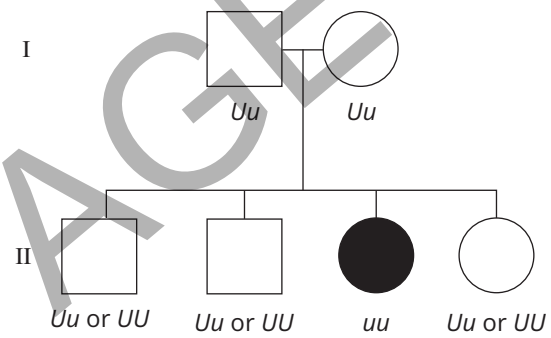
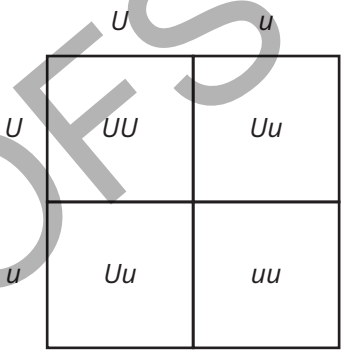


FIGURE 1.9.3 Pedigree diagram showing that attached earlobes is a recessive condition.



U – unattached earlobes
u – attached earlobes

FIGURE 1.9.2 Punnett square showing the possible genotypes of the offspring of two heterozygous individuals for unattached earlobes (*Uu*).

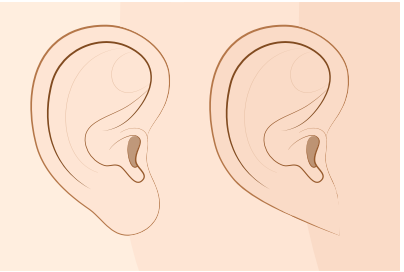


FIGURE 1.9.4 The ear on the left has unattached earlobes, and the ear on the right has attached earlobes.

Interpreting pedigree diagrams

In the scenario shown in Figure 1.9.3, the two individuals in the first generation both have unattached earlobes and they have offspring with both attached and unattached earlobes. It could be assumed that these individuals are both heterozygous.

It can be difficult to determine the genotype of every individual based on the information in a pedigree diagram. This can be seen in the two males of

generation 2 of Figure 1.9.3. These two could be either *Uu* or *UU*. The only way that the genotype of these individuals would be known is by examining any future offspring.

Studying the biological relationships between the individuals who are included in a pedigree diagram assists in determining if the trait is dominant or recessive. There are two key points to start with when conducting an analysis:

- If a trait is dominant and is expressed in an individual within the pedigree, then one of the parents must also have the trait. Figure 1.9.5 shows that normal pigmented fur colouring (*AA* or *Aa*) is a dominant trait as every individual with normal pigmented fur has a parent with normal pigmented fur.
- If the trait is recessive, then the trait may not be evident in the parents but can show up in offspring later in the pedigree. If two unaffected individuals produce offspring with a trait, then that trait must be recessive. In Figure 1.9.5 it can be seen that albino (non-pigmented) fur colouring (*aa*) is present in the second generation, but not in the first. This indicates that albino fur colouring is a recessive trait.

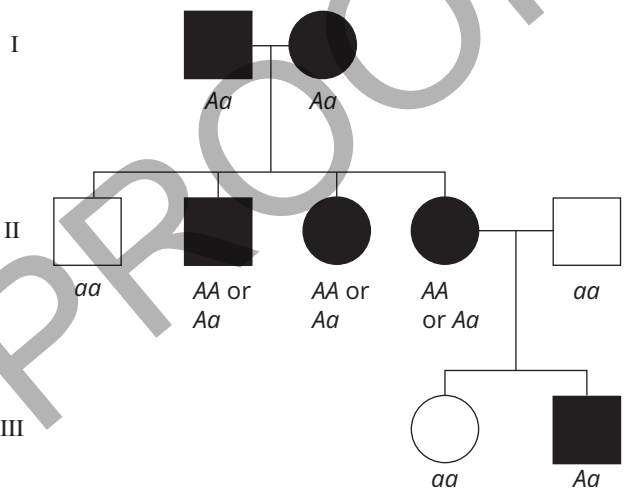


FIGURE 1.9.5 Pedigree diagram showing an autosomal dominant trait (normal pigmented fur colouring) through three generations.

The easiest way to start a pedigree diagram analysis is to:

- determine if the trait is dominant or recessive
- write the alleles of all recessive individuals
- assign one capital letter to all the dominant individuals
- determine if the dominant individuals are homozygous or heterozygous, by looking at the offspring that they produce
- complete the genotype for each individual.

SC 1 CHECK YOUR UNDERSTANDING

Identify the shape and shading for:

a a male that is affected by a condition

b a female that is affected by a condition

c a male that is unaffected by a condition

d a female that is unaffected by a condition.

SC 2 I can identify the pattern of inheritance for sex-linked traits using a pedigree diagram

Sex-linked traits

Remember that a sex-linked trait is a trait that is determined by a gene on one of the sex chromosomes. Traits that are determined by a gene on the X chromosome are known as X-linked. For example, haemophilia (Figure 1.9.6) is an X-linked trait.

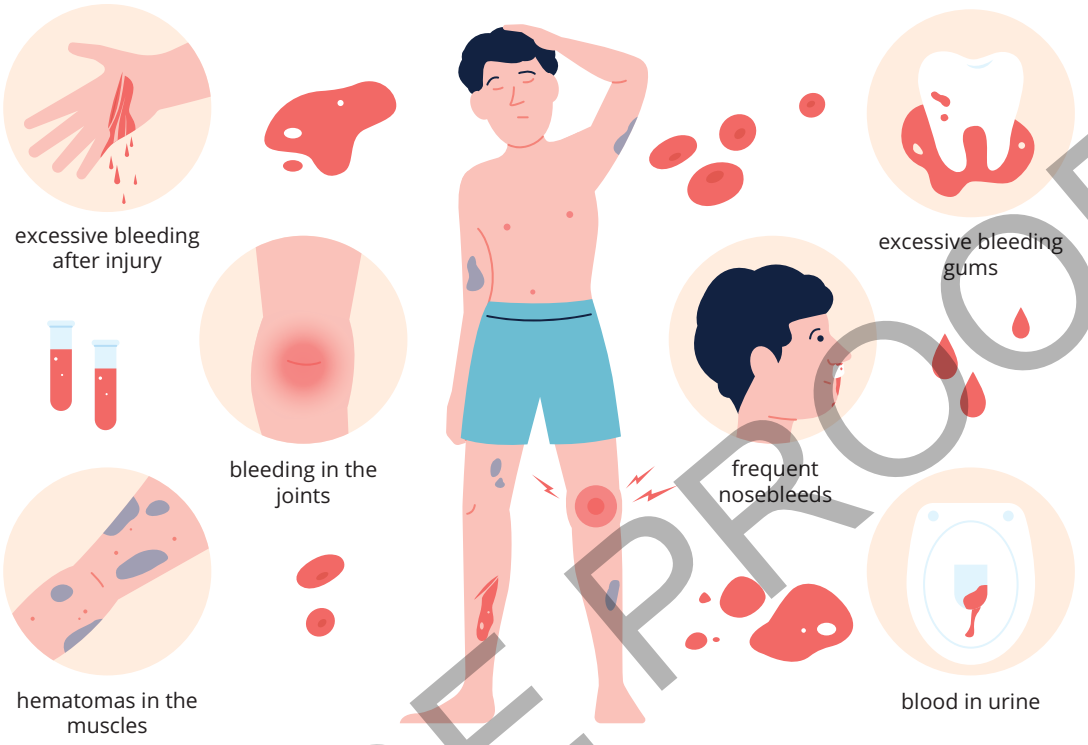


FIGURE 1.9.6 The symptoms of haemophilia, an X-linked recessive condition caused by a gene found on the X chromosome.

Like autosomal traits, sex-linked traits can either be dominant or recessive, and can be shown in a pedigree diagram.

Traits that are sex-linked affect the number of biological males and biological females differently. The following rules can be used to help you interpret sex-linked inheritance patterns in a pedigree diagram.

X-linked recessive

Mostly (or only) males are affected. Biological males are more likely to be affected by X-linked recessive traits because they only need to inherit one copy of an X-linked recessive allele for it to be expressed in their phenotype as they only have one X chromosome.

If a female is affected, her father must be affected as well.

X-linked dominant

Affected males have daughters who are all affected and sons who are not affected. This is because affected fathers pass on their only X chromosome, which carries the dominant allele, to all their daughters but pass on their Y chromosome to all their sons.

X-linked dominant traits are rare but, affect more females than males. This is because females inherit two X chromosomes so have twice the chance of inheriting an affected X chromosome compared to males who inherit only one.

Y-linked traits

Only males are affected, and they must always have an affected father, as the Y chromosome comes from the father.

Examples of X-linked traits

Colour blindness is an X-linked recessive trait. Figure 1.9.7 shows how colour blindness can be inherited throughout a family. The notation X^b indicates the allele for colour-blindness. To be affected (colourblind phenotype), a female must have the genotype X^bX^b (two copies of the allele), and a male must have the genotype X^bY (one copy of the allele). Unaffected females may have the genotype X^BX^b or X^BX^B, and unaffected males will have the genotype X^BY.

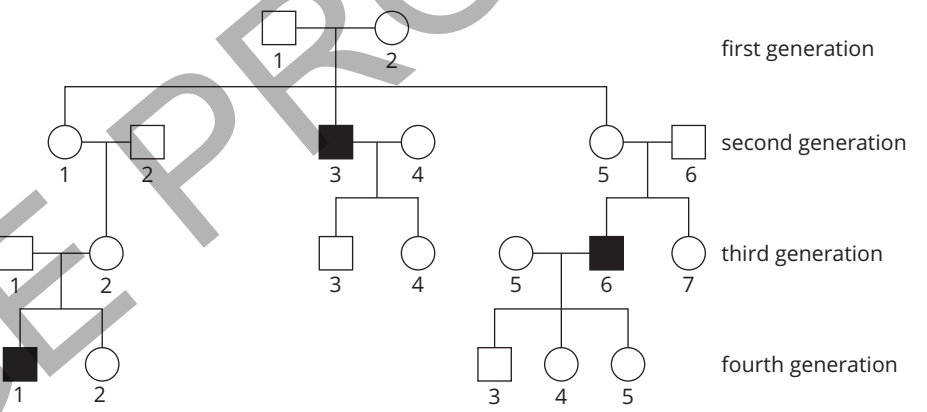


FIGURE 1.9.7 Pedigree diagram showing colour blindness in a family.

In Figure 1.9.7, there is no colour blindness in the first generation. However, the trait appears in the second generation (individuals 3 and 4), suggesting that colour blindness is recessive. The female (individual 2) in the first generation must therefore be a carrier (X^BX^b). No females in any generation are affected, which is a clue that this may be a sex-linked trait. While the pedigree diagram does not rule out the possibility that colour blindness is an autosomal recessive trait, it is in fact an X-linked recessive trait.

SC 2 CHECK YOUR UNDERSTANDING

State the rules for females that are affected in a X-linked recessive trait.

SC 3 I can develop a pedigree diagram to model inheritance of genes through families

Genetic inheritance

A pedigree diagram can be used to record when certain genetic traits appear in members of a family. Patterns in the pedigree diagram can then be used to predict the mode of inheritance for this particular trait.

Applied example: Earlobes

Kai wanted to develop a pedigree diagram for the trait of attached or unattached earlobes in his family on his mother's side. He had the following information:

- maternal grandmother: unattached earlobes
- maternal grandfather: unattached earlobes
- maternal aunt: attached earlobes
- maternal uncle: unattached earlobes
- mother: unattached earlobes
- father: unattached earlobes
- brother: unattached earlobes
- sister: attached earlobes
- Kai: attached earlobes.

Using this information, Kai was able to develop a pedigree diagram (Figure 1.9.8).

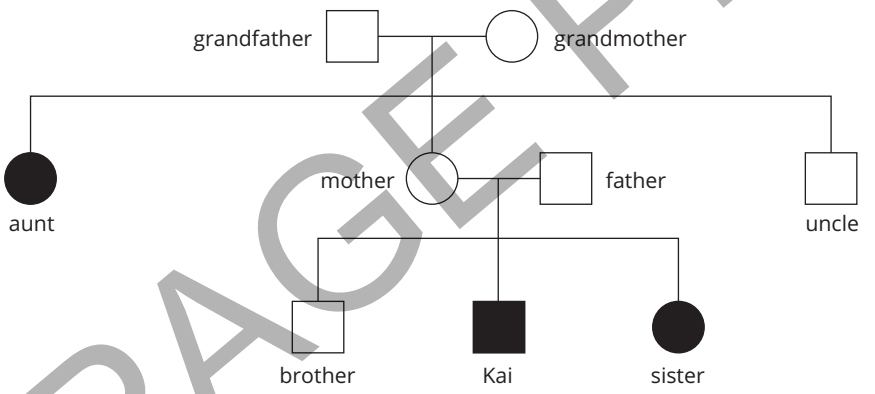


FIGURE 1.9.8 Pedigree diagram showing the trait of attached and unattached earlobes in Kai's family.

Kai noted the following key points while creating this pedigree diagram.

- Kai represented individuals with attached earlobes using shaded/coloured symbols.
- Neither of Kai's grandparents had attached earlobes, but attached earlobes appear in the next generation, which indicates that this genetic trait is recessive.
- This trait affects more females than males in Kai's family, so it is more likely to be autosomal than sex-linked.
- After considering all the information, Kai concludes that attached earlobes are an autosomal recessive trait.

Kai then assigned genotypes to this diagram to determine the genotype of each person. He started with assigning recessive alleles to himself, his sister and his aunt. Kai then established that:

- maternal grandmother: *Uu*
- maternal grandfather: *Uu*
- maternal aunt: *uu*
- maternal uncle: *Uu*
- mother: *Uu*
- father: *Uu*
- brother: *Uu* or *UU*
- sister: *uu*
- Kai: *uu*

SC 3 CHECK YOUR UNDERSTANDING

When developing a pedigree diagram, how are mating lines shown?

Lesson review

Use these questions to check whether you have met the learning intention for this lesson.

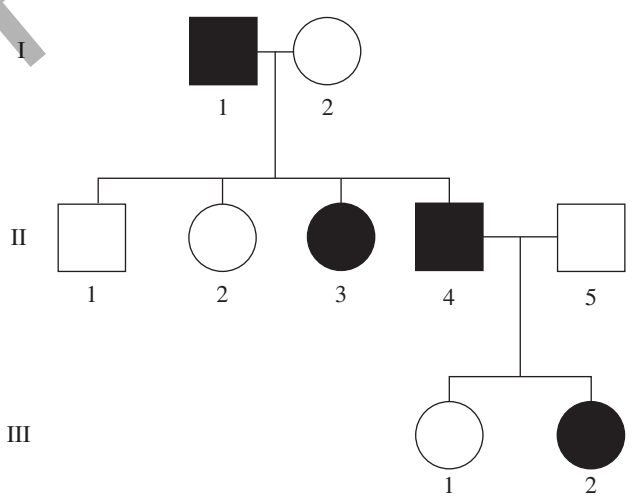
1 Outline what a pedigree diagram is.

2 Describe ways a pedigree diagram can be used to identify dominant and recessive traits.

3 Describe the steps to identify a sex-linked recessive trait in a pedigree diagram.

4 Sickle cell anaemia is an example of an autosomal recessive trait. Draw a pedigree diagram to show two unaffected parents with offspring that have sickle cell anaemia and assign genotypes to each of the individuals.

5 Analyse the following pedigree to determine the mode of inheritance in the affected individuals.



1.10 First Nations kinship laws

Lesson overview

Genetic disorders can occur randomly through mutations and can be passed along in families. There is an increased risk of genetic disorders appearing in populations that include many individuals who are homozygous for a gene, or populations that have limited genetic variation, which can happen when close relatives produce offspring. First Nations Australians have long held strict kinship laws that govern personal responsibility of individuals to their communities and towards the environment (Figure 1.10.1).

These laws also help to maintain genetic diversity in populations that are isolated, such as on an island or in a remote area.

In this lesson you will learn about First Nations Australian kinship laws, review the types of inheritance patterns for genetic disorders and consider the risks of sexual reproduction between relatives (people who are closely biologically related to each other).

SC 1 I can describe the kinship laws of First Nations Australians

Learning intention

To understand the kinship laws, family structures and marriage restrictions of First Nations Australians

Success criteria

SC 1: I can describe kinship laws of First Nations Australians.

SC 2: I can describe the potential outcomes of inheritance patterns of genetic defects.

SC 3: I can describe the genetic benefits of imposing certain rules through marriage restrictions.

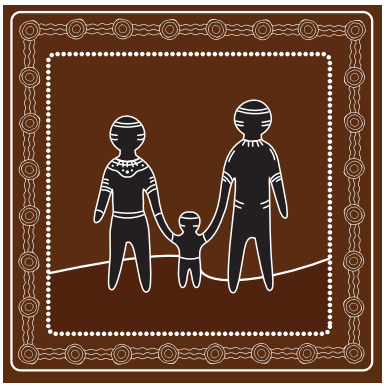


FIGURE 1.10.1 Australian First Nations peoples have strict kinship laws.

Kinship

Australia is home to hundreds of Aboriginal and Torres Strait Islander nations, each with its own languages and dialects. Some nations cover a large area, and some are much smaller.

These nations consist of clan groups (that share common **kinship** and language) and family groups. Connections between nations can be made through a system of moieties, totems and skin groups.

Kinship is an extremely important aspect of Australian Indigenous culture and governs all aspects of life. Generally, three levels of kinship exist between First Nations people.

Moiety

Moiety is the first level of kinship. In a moiety system, everything can be divided into two halves that are mirror images of each other.

The landscape, animals, plants and people can be divided in this way. All nations follow the moiety system, but nations will have different names for the two halves. For example, in the north of Australia in Arnhem land, the halves are referred to as Dhuwa and Yirritja, while in the Kulin nation, a person's moiety is either Waa the raven or Bunjil the wedge-tailed eagle (Figure 1.10.2). Individuals who share the same moiety are considered to be family and are mutually obligated to support and assist each other regardless of which nation they come from. Marriage must always occur with someone of the other moiety.

KEY TERMS

kinship family relationships; in Australian First Nations cultures, a system of laws governing social interactions and family relationships

moiety a Latin word meaning 'half'; in a moiety system, everything, including people and the environment, is split into two halves

FIGURE 1.10.2 The Kulin nation moiety: Waa the raven (left) or Bunjil the wedge-tailed eagle (right).

KEY TERMS

totem a specific animal, plant or natural feature that a person is spiritually linked to; determines relationships with others and rights

skin name a skin name is given to a First Nations person at birth and indicates their bloodline and their generation

Totems

Totems are the second level of kinship.

Each person has at least four totems: one each to identify your nation, clan and family group (bloodline), and one that you are given during your lifetime by your elders, which is chosen based on your individual strengths and weaknesses. Totems are used to help identify who you are and what your responsibilities are within the community and towards the environment.

Skin names

Skin names are the third level of kinship. Like a surname, your skin name identifies your family group (bloodline). It also identifies how generations of your society are linked and indicates how you should interact with others based on your skin group (others with the same skin name).

A husband, wife and child will all have different skin names as these names are generational, based on a sequential (ordered) system that cycles through eight levels (Figure 1.10.3). Each nation will have eight levels of skin names, but the exact skin names in each level will differ from nation to nation. The number of names in each level can also vary for each nation, from 16 to 32 names in each level. The skin names are also gendered by attaching a prefix or suffix (small word parts that are added to the skin name) to indicate male or female identity.

All the other people who have skin names from the same level are considered siblings (brothers and sisters). The people who have skin names from the level above are considered to be parents or aunties and uncles of the children in the generation below. The skin name system means that when a child is born, all the parents, aunties and uncles of that child are jointly responsible for parenting the child.

The skin name system is like having lots of brothers and sisters, mothers and fathers, aunties and uncles, grandmothers and grandfathers. It also means that all your first, second and third cousins (for example, your cousins' cousins) would share the same skin name level (skin group) and would be considered your siblings. Just as you cannot marry someone with the same moiety, you also cannot marry within the same skin group.

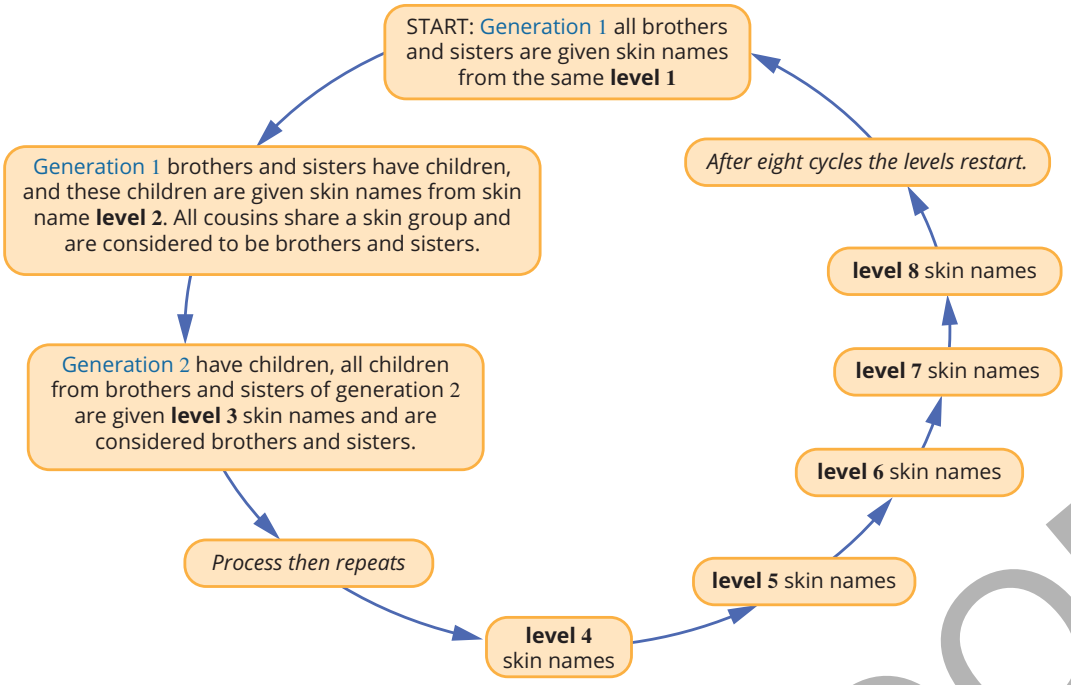


FIGURE 1.10.3 There are eight sequential levels of skin names, and each generation moves to the next level of names. When eight levels have been used for eight generations, the cycle begins again at level 1.

SC 1 CHECK YOUR UNDERSTANDING

Identify the three levels of kinship that exist between First Nations people.

SC 2 I can describe the potential outcomes of inheritance patterns of genetic defects

Patterns of genetic defects or disorders

Genetic material is the basis of all the characteristics of an individual. It is inherited from parent to offspring. If a genetic defect is present in an individual's genetic make-up, then this can be passed down through generations. Genetic defects can be found on both autosomes and sex chromosomes, and can be inherited as dominant or recessive conditions.

Autosomal inheritance of genetic defects

Autosomal dominant defects are inherited through a dominant allele. If an individual has at least one dominant allele in their genotype that is a genetic defect, they will express the trait in their phenotype. This means that both homozygous dominant and heterozygous individuals will express the dominant trait in their phenotype.

Some autosomal dominant conditions include achondroplastic dwarfism, Marfan syndrome, Neurofibromatosis type 1 (Figure 1.10.4) and Huntington's disease.

Scifile

Hemophilia in the royal family

The British royal family is known to be affected by haemophilia, which is a recessive blood disorder that affects the body's ability to create clots. Females are only affected by haemophilia when they inherit two recessive alleles for the condition. Males will be affected if they inherit just one recessive allele.

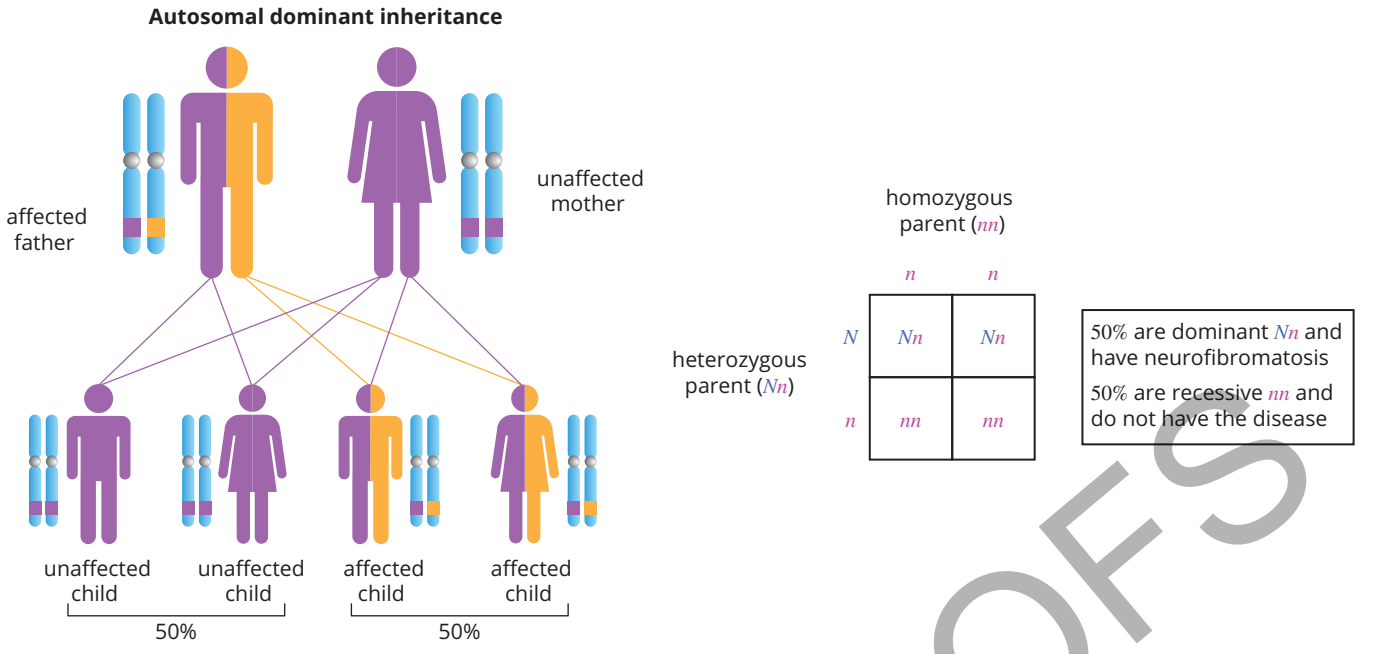


FIGURE 1.10.4 Diagram and Punnett square showing the inheritance pattern of autosomal dominant conditions such as Neurofibromatosis type 1; here, the dominant allele is shown as orange and the recessive allele is shown as purple.

Autosomal recessive inheritance of genetic defects

Autosomal recessive defects are inherited through recessive alleles. If an individual has two recessive alleles in their genotype for a genetic defect, they will express the trait in their phenotype. Two unaffected heterozygous parents may have a child that has a genetic defect.

Cystic fibrosis is an example of an autosomal recessive genetic disorder (Figure 1.10.5). An individual who has cystic fibrosis must have two recessive alleles for the condition. Other autosomal recessive illnesses include sickle cell anaemia (a blood disorder), Tay-Sachs disease (a fatal condition that affects the nervous system) and phenylketonuria (a disorder that affects how the body processes proteins in food).

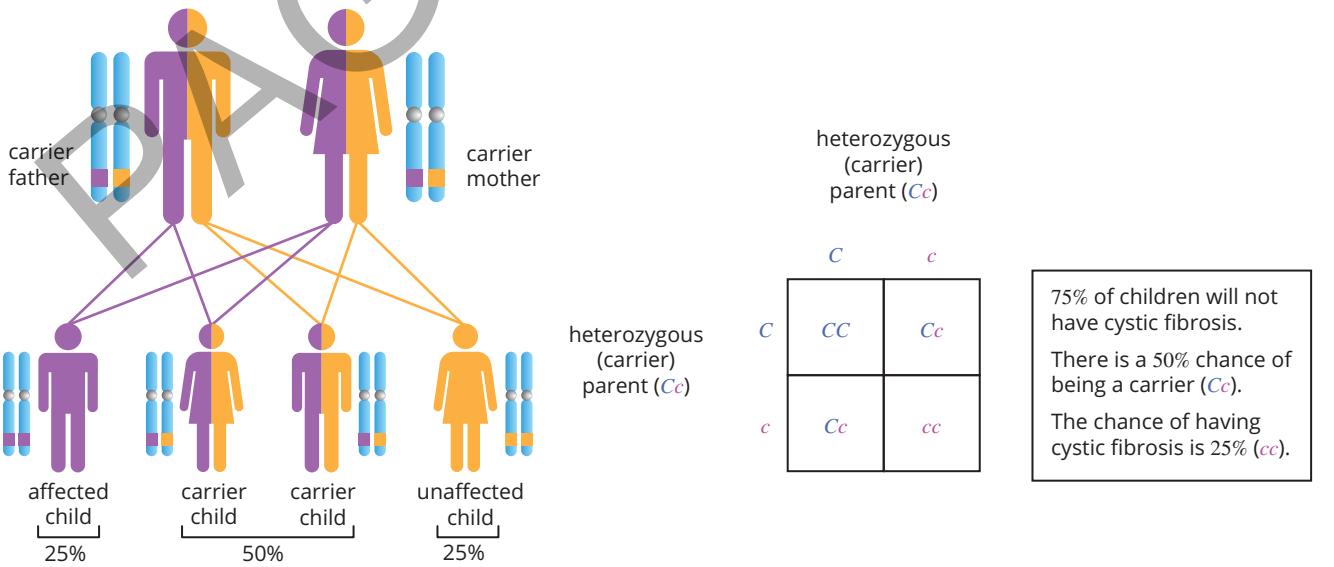


FIGURE 1.10.5 Diagram and Punnett square showing the inheritance pattern of autosomal recessive conditions such as cystic fibrosis.

Other sources of genetic defects or disorders

There are many other sources of genetic disorders that can lead to the inheritance of genetic conditions.

Lethal alleles

Lethal alleles are certain combinations of alleles that can be lethal (deadly) to the organism. Lethal alleles can be either recessive, where two copies of the defective gene are required, or dominant, when only one copy results in a lethal gene. Lethal alleles are more common in populations that have low genetic diversity or when higher rates of inbreeding occur. This is one way that kinship laws reduce the amount of genetic animalities in First Nations populations.

Tay-Sachs disease is an example of a recessive lethal allele. It is a fatal disorder of the nervous system. Tay-Sachs usually causes an affected individual to die by the age of five. Parents who have one copy of this faulty allele (heterozygous genotype) are carriers.

Sources of genetic defects

Mutations are permanent and random alterations (changes) that happen to a deoxyribonucleic acid (DNA) sequence, that can be a source of genetic defect. Mutations can occur while DNA is replicating or may be caused by environmental triggers such as radiation, certain viruses or exposure to toxic (poisonous) chemicals (Figure 1.10.8). These changes to the DNA sequence will not always be expressed in a person's phenotype. Whilst mutations that result in genetic disorders are rare, they can be the sources of inherited conditions.

KEY TERM

mutation a mistake that happens as DNA is copied, causing a change to the base sequence

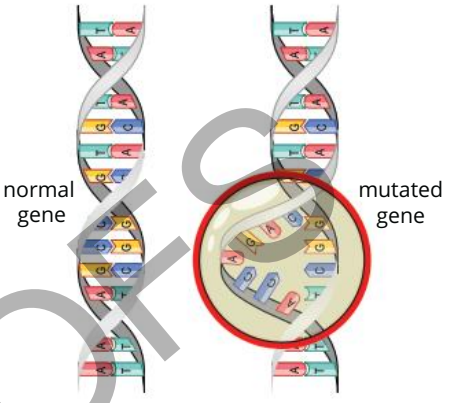


FIGURE 1.10.8 Mutations can occur during DNA replication, mitosis, or meiosis, or result from damage to DNA; these mutations can be caused by errors in the replication process or by exposure to environmental triggers (such as changes in the genetic code of the mutated gene).

SC 2 CHECK YOUR UNDERSTANDING

Describe how an individual could get a genetic disorder if neither of their parents have the defect.

SC 3 I can describe the genetic benefits of imposing certain rules through marriage restrictions

Australian marriage law

In Australia, the Marriage Act determines who can and cannot marry. Section 23.2 Australian Marriage Act (1961) states, 'Marriages of parties within a prohibited relationship: Between a person and an ancestor or descendant of the person; or between a brother and a sister (whether of the whole blood or half blood)'.

The Marriage Act prohibits (prevents or stops) marriage between siblings and between parent and child (or grandparent or grandchild), but it allows for other close relatives to marry (such as cousins). This means that modern marriage laws do not fully consider the risks of inheriting genetic conditions if sexual reproduction occurs between people who share a lot of genetic material. In this way these laws vary from, and are less sophisticated than, the kinship laws of First Nations Australians, which include individuals from the extended family.

This means in Australia you could legally marry your niece or nephew, your aunt or uncle, or even your cousin. However, these kinds of relationships come with greater risks of passing on genetic conditions to any offspring because there is more chance of inheriting recessive alleles. Sexual reproduction between people who are closely related also reduces genetic variation in offspring.

Genetic benefits of marriage restrictions

All humans share about 99.9% of their DNA (genetic material), which means that there is only a small amount of DNA (0.1% of the total) that varies between individuals who are not related. Families share even more than 99.9% of their DNA.

First cousins share about one eighth of the 0.1% that differs between non-related people. Although this sounds small, it means that there is an elevated risk of having a child with congenital abnormalities (present at birth) or inherited genetic disorders, particularly recessive disorders. The general risk of a child being born with a congenital genetic abnormality is approximately 3%, and this risk is roughly doubled for the children of first cousins.

Similarly, an uncle or aunt shares about one quarter of the 0.1% of DNA variability with a niece or nephew. Once you get past second-cousin marriages, the risks of genetic disorders are significantly reduced.

SC 3 CHECK YOUR UNDERSTANDING

Describe how Australian marriage law reduces the chance of inheriting genetic defects.

Lesson review

Use these questions to check whether you have met the learning intention for this lesson.

1. Define the term genetic defect.
2. Name two inherited genetic disorders and identify their mode of inheritance.
3. Explain the significance of kinship laws in First Nations Australian communities.
4. Predict the chance of obtaining a recessive genetic disorder when neither parent has the disorder themselves, but are both carriers.

1.11 Changes to DNA

Lesson overview

Your body is constantly replicating (copying) its deoxyribonucleic acid (DNA) as it makes new cells. In most cases, cells do this correctly and the copied DNA exactly matches the original DNA.

In this lesson you will learn about what happens when DNA is not replicated correctly. An error can happen when a small part of the nucleotide sequence of an individual gene is copied incorrectly resulting in mutations and errors. These errors can affect how cells function.

More significant mutations can also occur in chromosomes. Chromosomal mutations happen when the structure or number of chromosomes are altered, affecting larger sections of the DNA sequence.

SC 1 I can describe environmental conditions or other factors that can cause changes in DNA or chromosomes

Learning intention

To understand the causes and effects of changes in genetic material

Success criteria

SC 1: I can describe environmental conditions or other factors that can cause changes in DNA or chromosomes.

SC 2: I can describe different types of changes in DNA or chromosomes.

SC 3: I can explain potential changes in the body caused by changes in DNA or chromosomes.

DNA replication

DNA replication is continually happening in your body's cells. This occurs during the interphase of the cell cycle when new cells are being made. Specific nucleotide sequences in DNA (genes) code to produce proteins in the body. These proteins are used in other bodily structures or functions. It is important that cells accurately replicate the nucleotide sequences of the DNA that they contain to ensure that the correct proteins are produced.

Your body does a good job of replicating its DNA accurately most of the time, but sometimes there are factors that can interfere with this process. When DNA is replicated incorrectly, the new nucleotide sequence is called a mutation. If the exact sequence of nucleotide bases in the DNA is not replicated accurately then the correct protein may not be formed. Sometimes these changes have no effect or even a positive effect on the body.

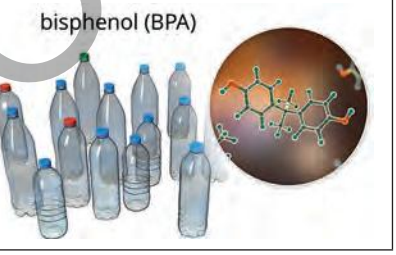
Environmental causes of changes to DNA

DNA mutations can happen in different ways. For example, some environmental triggers can cause changes to the nucleotide sequence in DNA. These environmental triggers are known as **mutagens** because they can change the DNA, leading to mutations. Common mutagens can be seen in Table 1.11.1.

KEY TERM

mutagen an agent that causes a mutation

TABLE 1.11.1 Common mutagens

Mutagen	Description and effects	
Ultraviolet (UV) radiation	• causes sunburn • interferes with the chemical bonding between nucleotides in DNA • can prevent cells from carrying out regular functions • can lead to skin cancer and premature aging of the skin	
Cigarette smoke	• contains carcinogens known to cause cancer • carcinogens bind to DNA • can cause mutations to the DNA sequence	
Plastics: Bisphenol A (BPA) and phthalates	• chemicals used in plastics and personal care products like shampoo • chronic (long-term) exposure can alter gene expression	

Scifile

Cancer genetics

Cancer is often caused by mutations in genes that control cell growth and division. These mutations can lead to uncontrolled cell growth, forming tumours. This is why Australia has packaging laws on cigarettes, which are known to cause cancer.

Any changes to DNA can be dangerous, and environmental triggers should be avoided where possible.

DNA can also be altered via spontaneous mutations, or mutations that occur during regular DNA replication where the source is unknown.

Sometimes mutations only occur in certain cells of the body, and these are known as acquired mutations. Some mutations happen when an embryo is forming and may then occur in every cell in the body.

SC 1 CHECK YOUR UNDERSTANDING

Define the term mutagen.

SC 2 I can describe different types of changes in DNA or chromosomes

Mutations in DNA

When DNA replicates, the replicate (copy) needs to be identical to the original. Sometimes mutations occur during DNA replication. Mutations can mean that sections of the DNA molecule (genes) are no longer viable (do not work properly). Genes with mutations may no longer code for the correct protein, which may mean that the structure or function in the body that the protein is involved in may not occur the way it should.

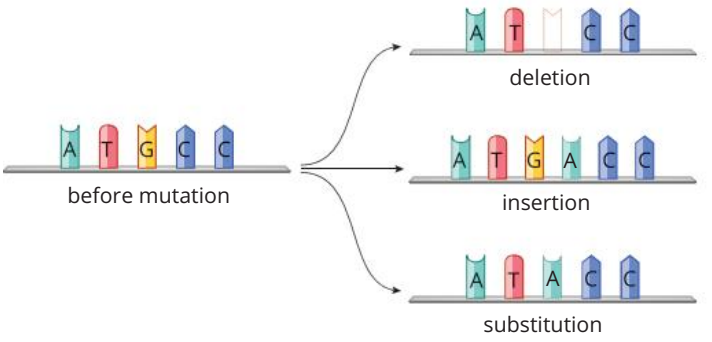
Types of DNA mutations

Coding mutations

DNA coding mutations include **point mutations**, which usually affect one or very few nucleotide base pairs. Several base pairs may be affected but not larger segments of the DNA sequence. Some, but not all mutations may affect the final protein made.

KEY TERM
point mutation a mutation that affects only one or very few nucleotide base pairs

Key types of coding mutations can be seen in Figure 1.11.1.



Deletion
One (or more) nucleotide base pairs is removed from the DNA sequence. The new sequence is slightly shorter than the original.

Insertion
One (or more) nucleotide base pairs is added to the DNA sequence. The new sequence is slightly longer than the original.

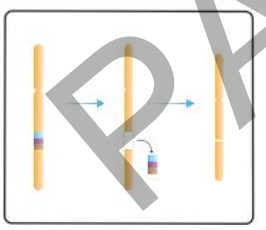
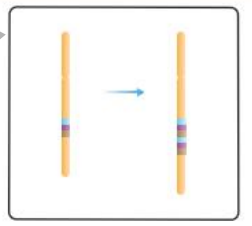
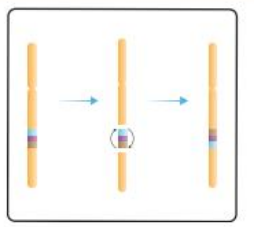
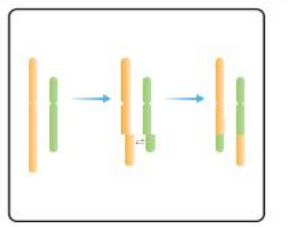
Substitution
One (or more) nucleotide base pairs is replaced by (swapped for) another one. The overall length of the new sequence remains the same as the original.

FIGURE 1.11.1 Type of DNA coding mutations.

Chromosomal mutations

Sometimes larger mutations occur that affect the chromosomes themselves, rather than just small sections of the DNA sequence. These mutations can result in chromosomal abnormalities such as changes to the chromosome structure (more commonly) or the number of chromosomes (less commonly). The different types of mutations can be seen in Table 1.11.2. Chromosomal mutations can have significant consequences for an individual's health and development.

TABLE 1.11.2 Types of chromosomal mutations

Deletion	Duplication	Inversion	Translocation
			
Loss of all or part of a chromosome.	Produces an extra copy of all or part of a chromosome.	A segment of a chromosome is inverted (reversed). Note the position of the blue band representing part of the chromosome.	Translocation means a change in location. This type of mutation occurs when part of a chromosome is transferred to another chromosome.

SC 2 CHECK YOUR UNDERSTANDING

List the different types of DNA coding mutations.

SC 3 I can explain potential changes in the body caused by changes in DNA or chromosomes

Point mutations (insertion, deletion and substitution) and chromosomal mutations (deletion, duplication, inversion and translocation) can both affect gene function. Mutations can result in diseases like cancer and genetic conditions such as Huntington's disease, sickle cell anaemia, Edwards syndrome, Down syndrome and cystic fibrosis.

Huntington's disease

Huntington's disease (Figure 1.11.2) is an autosomal dominant genetic condition that is caused by a mutation in the *HTT* gene on chromosome 4. This gene contains a CAG (cytosine, adenine, guanine) repeat sequence. Most people have fewer than 30 CAG repeats in this gene. People who have Huntington's disease have an abnormally large number of CAG repeats (more than 40 repeats). Huntington's disease is characterised by abnormal body movements and cognitive and behavioural problems. The mutation that causes Huntington's disease is a DNA insertion mutation.

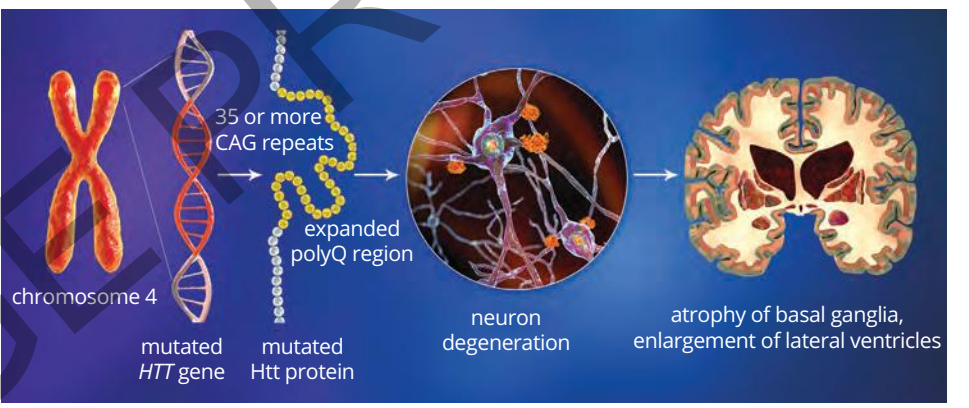


FIGURE 1.11.2 The genetic mutation responsible for Huntington's disease affects brain development.

Down syndrome

A person with Down syndrome has 47 chromosomes (instead of 46 in a diploid set). Down syndrome is also known as trisomy 21. Instead of a pair of chromosomes at position 21, there are three chromosomes. The extra chromosome 21 that causes Down syndrome is the result of a chromosomal mutation during meiosis. This mutation usually occurs when a pair of 21st chromosomes in a cell fails to separate. Both chromosomes from the 21st pair end up in a single daughter cell (gamete) and then go on to join with another gamete that also contains one chromosome in position 21, making a total of three in the zygote.

Some characteristics that are common (Figure 1.11.3) in Down syndrome include a flat bridge of the nose, eyes that slant up, a short neck, tiny white spots in the iris of the eye and small hands and feet. People with Down syndrome often experience problems with their heart and digestive system.

FIGURE 1.11.3 This young person has Down syndrome, also known as trisomy 21.

Genetic counselling

Mutations that affect the structure of chromosomes may impact many genes, therefore disrupting the production of many proteins. This means that all chromosomal mutations will result in adverse (harmful) effects for the individual.

Genetic counsellors help families to identify the risks of passing on genetic conditions to offspring. They can do this by analysing family history and inheritance patterns. They can also arrange for individuals to have genetic testing and then assist them to understand the results.

SC 3 CHECK YOUR UNDERSTANDING

Name one condition that results from DNA point mutations and one condition that results from chromosomal mutations.

Lesson review

Use these questions to check whether you have met the learning intention for this lesson.

1. List environmental factors that can cause changes in DNA or chromosomes.
2. Describe how UV radiation can cause changes in DNA.
3. Identify the type of mutation that has occurred when the following DNA code changed from GTACATCGATACA to GTACAATACA.
4. Compare the potential effects of a point mutation and a chromosomal deletion on an organism.
5. Describe the role of genetic counsellors.

1.12 Genetics and disease

Learning intention

To understand the role of genetic changes that lead to cancer, haemochromatosis, sickle cell anaemia and cystic fibrosis

Success criteria

SC 1: I can explain how changes in DNA can result in cancer, haemochromatosis, sickle cell anaemia and cystic fibrosis.

SC 2: I can identify different genetic technologies that can be used to assist in the diagnosis and treatment of cancer, haemochromatosis, sickle cell anaemia and cystic fibrosis.

SC 3: I can evaluate the benefits and risks of gene therapy or genetic engineering in the treatment of disease.

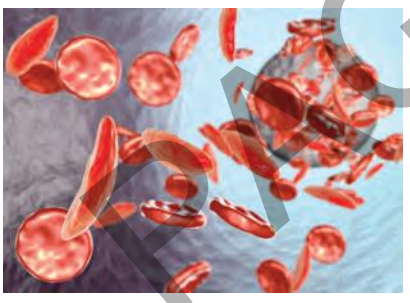


FIGURE 1.12.1 This illustration shows normal red blood cells (round) and red blood cells that are affected by sickle cell anaemia (crescent shaped).

Introduction

There are many health conditions that have genetic causes. Understanding the underlying causes of genetic conditions is important for individuals who are affected by these conditions and their families, and for healthcare professionals. Understanding the causes can also help to prevent genetic conditions arising in the first place and to inform diagnosis and treatment approaches for people who are affected.

Uncovering factors that contribute to genetic conditions enables better genetic screening (DNA testing to identify a person's risks of passing on genetic conditions to their offspring), therapies that target specific molecular pathways and proteins, better genetic counselling and personalised medicine approaches (which involve tailored treatments based on a person's predicted outcome).

In this inquiry activity you will explore the role of genetic changes that lead to cancer, haemochromatosis, sickle cell anaemia and cystic fibrosis, and participate in a panel discussion on the benefits and risks of gene therapy.

Background

Mutations are changes to genetic material that can result in defects and disorders. Sometimes mutations occur spontaneously, with no clear cause other than chance, and some occur as a result of exposure to a mutagen. Other mutations are inherited when a parent who has a faulty copy of a gene (allele) passes this allele to their offspring as an outcome of sexual reproduction.

Inherited mutations in specific genes can result in genetic conditions, such as hemochromatosis, sickle cell anaemia (Figure 1.12.1) and cystic fibrosis.

Cancer is a disorder that results from a mutation that can be caused by inherited factors (such as those seen in the *BRCA1* and *BRAC2* genes that cause breast cancer) or mutagens such as smoking or UV radiation, or even spontaneously.

Aim

To investigate how genetic changes contribute to disorders such as cancer, haemochromatosis, sickle cell anaemia, and cystic fibrosis, in order to support informed discussions about the potential use of gene therapy

Plan

Part A

The impact of mutations on human health

1 In groups of three to five, choose one genetic change to explore that can lead to one specific type of cancer, haemochromatosis, sickle cell anaemia or cystic fibrosis.

2 Research your chosen disorder for 15 minutes and prepare a short summary.

3 Include details such as the type of genetic change and where it occurs, the impacts of this genetic change, and any other factors, including environmental influences, inheritance patterns, and distribution or prevalence of the mutation in different populations.

4 Choose one person to read your summary to another group in the class.

Part B

The role of technology in genetic screening

5 With the same group, explore how different genetic technologies aid in the diagnosis and treatment of your chosen condition.

6 Research your chosen technologies for 15 minutes and prepare a short summary.

7 Choose one person to read your summary to another group in the class.

Part C

Panel discussion of benefits and risks of gene therapy

8 Each group in the class will be assigned 'for gene therapy' or 'against gene therapy'.

9 Research arguments for or against gene therapy—depending on which group you are in—for 10 minutes. Use evidence from your previous research to inform your ideas and points of view.

10 Prepare to participate in a panel discussion with another group that has an opposing viewpoint.

Design

- When selecting a condition, consider your group's existing knowledge or interest.
- Use concise notetaking to prepare for sharing.
- Ensure websites used are credible.
- For the gene therapy argument, each person should find one point, then combine them into a group perspective.

Conduct

- Ensure speakers have clear, collated notes before presenting.
- Presenters should:
  - face the group
  - speak clearly
  - be listened to by teammates.
- Take and answer questions at the end of each presentation.
- Share ideas point by point during the panel.
- Encourage respectful discussion—there are no 'wrong' ideas if they are researched and explained.

Evaluate

Through this inquiry, you learnt about the role of genetic changes leading to cancer, haemochromatosis, sickle cell anaemia and cystic fibrosis.

1. Comment on what you learned about the benefits and risks of gene therapy or genetic engineering in the treatment of disease, and describe any further learning or questions you have.
2. State the skills you used in this inquiry.

1 Genetics and models of inheritance

Topic summary

The key concepts included in this topic are:

- DNA is a double helix composed of nucleotide bases that is responsible for passing on traits to offspring.
- DNA including its genes, are tightly wound into chromosomes within the nucleus of eukaryotic cells.
- The role of mitosis is to make new genetically identical diploid daughter cells that are used in growth, repair and maintenance of an individual.
- The role of meiosis is to make new genetically unique haploid daughter cells that, when fertilisation occurs can be used to form an individual with genetic variation.
- Genes consist of a sequence of nucleotide bases that make up an individual's genotype and each gene can have different variations known as alleles.
- An individual's genotype is responsible for producing their physical characteristic known as their phenotype.
- Traits that are passed from parent to offspring can be analysed using Punnett squares and in larger families through pedigree diagrams.
- Traits are inherited in many ways including autosomal recessive inheritance, autosomal dominant inheritance and X-linked inheritance.
- Many diseases and disorders have a genetic basis that affects individual DNA sequences or chromosomes and this affects the phenotype of an individual.
- Australia has both the First Nation kinship laws and Australian Marriage law to reduce the chance of genetic disorders in populations.

Review questions

The following questions will assess your success in achieving the learning intentions for this topic.

Remember

1 Draw and label the structure of a nucleotide.

2 Name the four nitrogenous bases and specify their complementary base pairs.

3 Define the genetic term 'phenotype'.

4 Outline the purpose of using a buffer solution in DNA extraction.

Understand

5 Define the term gamete and explain how its chromosome number in humans leads to the formation of a zygote.

6 Explain the explain the need for ensuring kinship laws are followed in Australia.

Apply

7 Eye colour is an inherited trait that is determined by an individual's alleles.

a Explain the concept of alleles using eye colour as an example.

Two heterozygous brown-eyed individuals are wanting to start a family.

b Draw a Punnett square to show this genetic cross.

c Calculate the phenotypic and genotypic ratio of the predicted offspring for this cross.

d What is the chance that their child will have blue eyes?

8 Explain the significance of meiosis in maintaining genetic variation.

9 Genetic variation is important in maintaining populations. Identify three genetic crosses that produce offspring that have only one genotype.

10 Compare the roles of dominant and recessive alleles in determining traits.

Analyse

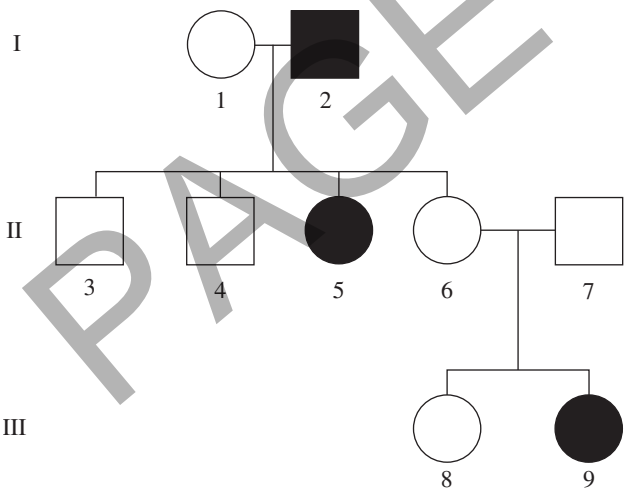
11 Polycystic kidney disease (PCD) is an inherited condition found on an autosome and follows a dominant pattern of inheritance.

- **a** Define the term autosome.
- **b** Calculate the chance that a child will have PCD if the father is unaffected, and the mother is heterozygous with PCD.
- **c** Draw a pedigree diagram to show the two parents and an affected child in this example.
- **d** Identify the potential genotypes of the father's parents.

12 Understanding cell division processes and the effects of genetic changes is crucial in genetics.

- **a** Compare the outcomes of mitosis and meiosis.
- **b** Compare the effects of a chromosomal deletion mutation to a chromosomal duplication mutation on an organism.

13 Inherited characteristics in families can be examined using a pedigree.



Use evidence to explain the mode of inheritance for the trait shown in the pedigree.

Extension: Research task

14 Non-invasive prenatal testing (NIPT) is a blood test that women can have in the early stages of pregnancy that can detect different types of chromosomal disorders.

Research NIPT and explain the types of genetic conditions that it is able to detect, including any conditions outlined in the topic.

Explore ethical, moral and social implications of NIPT and present ideas of potential arguments for and against this type of genetic analysis.

Topic reflection

The learning intentions for this topic are given in each lesson and at the beginning of the topic. Consider how well you have achieved them. Note down any particular areas that you are confident in, and others where you are not so sure.

1 Glossary

allele one of two or more alternative forms of a gene that can occupy a specific position on a chromosome

asexual reproduction a form of reproduction where only one parent cell produces offspring without joining of sperm and eggs

autosome chromosome that is not a sex chromosome

base part of a nucleotide; the four types include adenine (A), guanine (G), cytosine (C) and thymine (T)

cell cycle the events in the life of a cell, from its formation by cell division through its growth and function until it divides again

cell division process that results in two new cells each having the same genetic information as the parent cell; also called mitosis

centromere the point on a chromosome where the two chromatids are joined together

chiasma a point where two chromosomes exchange genetic material during meiosis

chromatid one of the strands of a chromosome following replication

chromosome thread-like structure in the nucleus; composed of DNA and proteins; contains genetic information in the form of genes

codominance a genetic trait where both alleles in a pair are fully expressed in the phenotype, resulting in a combined trait rather than one allele being dominant over the other

crossing over the exchange of genetic material between homologous chromosomes during prophase I of meiosis

cytokinesis the separation of a cell following mitosis or meiosis, when the cytoplasm divides and the cell splits into daughter cells

daughter cell a new cell formed by cell division.

deoxyribonucleic acid (DNA) a double helix made of nucleotides; the molecule that determines the genetic characteristics of most living things

diploid the number of chromosomes in body cells; two sets or *2n*

dominant trait the trait that is observed in the outward appearance of a heterozygous individual

double helix shape like that of a twisted rope ladder

eukaryotic cell cell that contains a nucleus

fertilisation the joining of gametes

gamete sperm cell or egg cell

gene a section of DNA that carries the genetic code for a particular characteristic

genetic code the sequence of three nucleotides in DNA that determines, or codes for, genes

genetic cross the breeding of two individuals to produce offspring that are representative of the parents' genetic material

genetic variation differences in the sequences of genes or alleles between individual organisms

genetics the study of inherited characteristics called traits

genotype entire genetic information of an individual; refers to alleles present for each gene

genotypic ratio expected frequency of genotypes in the offspring of a genetic cross

haploid the number of chromosomes in gametes; one set or *n*

heterozygous having two different alleles on homologous chromosomes

homologous chromosome one of a matching pair of chromosomes in a diploid organism; homologous chromosomes carry the same genes in the same location

homozygous having two identical alleles on homologous chromosomes

incomplete dominance when neither allele in a gene pair is completely dominant over the other, resulting in a phenotype that is an intermediate between the two parental phenotypes

independent assortment the random allocation of homologous chromosomes to the spindle fibre during meiosis I of meiosis

interphase the phase in the cell cycle when the cell is not undergoing mitosis

karyotype a visual representation of the number, size and shape of chromosomes in an individual

kinship family relationships; in Australian First Nations cultures, a system of laws governing social interactions and family relationships

law of segregation one of the two alleles from the parental genotype is present within the gamete cell

meiosis the process that forms sex cells, when a cell nucleus divides into four nuclei

mitosis the type of cell division that produces two daughter cells identical to the parent cell

moiety a Latin word meaning 'half'; in a moiety system, everything, including people and the environment, is split into two halves

monohybrid cross a genetic cross between a homozygous dominant individual and a homozygous recessive individual

mutagen an agent that causes a mutation

mutation a mistake that happens as DNA is copied, causing a change to the base sequence

nucleotide a molecule consisting of a five-carbon sugar (ribose or deoxyribose), a nitrogenous base (purine or pyrimidine) and a phosphate group; the main building blocks of nucleic acids

nucleus organelle that contains the genetic information for the cell (plural nuclei)

offspring individuals produced by reproduction

phenotype observable characteristics of the individual; the way the genotype is expressed

phenotypic ratio expected frequency of phenotypes in the offspring of a genetic cross

point mutation a mutation that affects only one or very few nucleotide base pairs

protein natural polymer required for growth and repair in living organisms

Punnett square a visual representation of the genotypes of a genetic cross and the possible genotypes of their offspring

recessive trait the trait that remains hidden in the heterozygous condition and seen only in the homozygous condition

reproduction the process of parents producing new individuals, or offspring

sex chromosome chromosome that determines the sex of an individual; X and Y chromosomes in humans

sex-linked related to genes that occur on the sex-chromosomes (X and Y in humans)

sexual reproduction the process of a sperm and an egg joining together and then growing into a new individual

skin name a skin name is given to a First Nations person at birth and indicates their bloodline and their generation

somatic cell any body cell except a cell that gives rise to gametes (eggs and sperm)

totem a specific animal, plant or natural feature that a person is spiritually linked to; determines relationships with others and rights

trait inherited characteristic

X-linked related to genes on the X chromosome

zygote the cell formed by fusion of two gametes, a sperm and egg

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