OUR SALIVARY GLANDS PRODUCE AROUND 1.5 LITRES OF SALIVA EACH DAY!

Fruit contains a sugar called FRUCTOSE.
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How are traits inherited?

Although children resemble their parents, grandparents and siblings, no two people are exactly alike, not even identical twins. You may have noticed that many personal features, such as height, hair colour, facial shape and athleticism seem to 'run in the family'. These features may be obvious, but family traits can also be hidden and subtle. Doctors may ask their patients about family members with conditions such as asthma, diabetes or cancer. This is because physical and biological features, as well as medical conditions, can be inherited through generations of a family.
SHOWCASE: CONFERENCE OF CHROMOSOMES AND PATTERNS OF HEREDITY

Your task is to investigate a genetic condition in humans caused by a DNA mutation. You will develop a suite of information about human genetics and present this information in a form of your choice to a classroom conference. In this showcase, you will complete these tasks.

1. Identify a genetic condition caused by changes in the DNA of a human chromosome. Use the weblink ‘Chromosome viewer’ to assist you in your choice.

2. Identify the human chromosome which carries this genetic condition.

3. Describe the change that occurs in the DNA of that chromosome to cause the condition. Identify some of the physical symptoms shown by people with the condition.

4. Discuss the following issues in relation to the genetic condition you selected.
   a. What is the pattern of inheritance for this condition – is it dominant or recessive? (Use a pedigree chart to show the pattern of inheritance.)
   b. Is the disease serious enough to warrant genetic testing? Consider whether there are any preventative actions that can be taken by a person who may inherit the genetic condition, and if the condition can be managed or treated if it is expressed.
   c. Where can a person with the genetic condition find advice and support, or make contact with others with the same condition?

Write up a report of no more than 800 words to present to the class, and include references.
Humans have been practising genetics from at least 15 000 BCE. The first attempts at selective breeding are evident in the sealed tombs of our ancestors. Ancient pollen from domesticated plants found in the tombs shows that farmers were selecting and breeding plants for desired characteristics. However, the farmers did not know how these characteristics were passed on from one generation to the next.

Genetics is the study of genes and inheritance, how characteristics are passed from one generation to the next. People working in many different fields must have a knowledge of genetics. For example, genetics is used in agriculture to increase crop and animal productivity, in medicine to identify people at risk of genetic conditions, and by the police to solve crimes.

To understand why we each have certain characteristics, we must look deep into the body’s cells. There is one molecule that contains instructions that control all of an individual’s characteristics, including the way the body functions. This molecule is found in the nucleus of most cells and is known as deoxyribonucleic acid (DNA).

The structure of DNA

There is approximately 3 m of DNA in the nucleus of each of our cells. The DNA molecule in cells is made up of two strands and is shaped like a ladder: it has two vertical ‘backbones’ (the sides of the ladder) made of alternating sugars and phosphates. The horizontal ‘rungs’ are made of units called nitrogenous bases. The DNA molecule is not flat like a real ladder; it is shaped like a ladder that is held at the top and bottom, and then twisted around. The resulting shape is known as a double helix (Figure 2.1).

A single strand of DNA is made up of many repeating units called nucleotides (Figure 2.2, page 00). Each nucleotide consists of a sugar, a phosphate and one of the four nitrogenous bases adenine (A), guanine (G), thymine (T) and cytosine (C). When two DNA strands pair to form a double helix, the bases are always found in complementary pairs. This means that the base on one strand of a DNA molecule will always pair with its complementary base on the other strand: A will always pair with T, and G will always pair with C. This is known as complementary base pairing. This structure plays an important role in how DNA is accurately replicated.
Modelling DNA

In this activity you will construct a DNA model that is 10 nucleotides long. This will represent the coding for a single gene. (In reality, genes are much longer than this.)

**Activity 2.1.1**

**What are the risks in doing this activity?**

<table>
<thead>
<tr>
<th>What are the risks in doing this activity?</th>
<th>How can you manage these risks to stay safe?</th>
</tr>
</thead>
<tbody>
<tr>
<td>Science laboratory desks may be contaminated with chemicals, which could be toxic.</td>
<td>Do not eat the lollies used in this experiment. Alternatively, conduct the experiment in a clean classroom or food technology kitchen.</td>
</tr>
</tbody>
</table>

**What you need**

- 40 gumdrops of any four different colours, such as blue, orange, pink and yellow
- 40 small marshmallows
- two thick, twisted liquorice sticks
- toothpicks
- a digital camera (optional)

**What to do**

1. Working in groups of three, collect your materials.
2. Construct your DNA model using the following key.

<table>
<thead>
<tr>
<th>Material</th>
<th>Represents</th>
</tr>
</thead>
<tbody>
<tr>
<td>Gumdrops</td>
<td>Bases, such as blue = A, orange = T, pink = C, yellow = G</td>
</tr>
<tr>
<td>Liquorice sticks</td>
<td>The sugar–phosphate backbone of the DNA</td>
</tr>
<tr>
<td>Small marshmallows</td>
<td>The part of the sugar that connects to the bases</td>
</tr>
<tr>
<td>Toothpicks</td>
<td>To bond backbone to sugar, sugar to phosphates</td>
</tr>
</tbody>
</table>

3. Write down the base pairs of your DNA model segment.
4. Take a photo or make a drawing of your DNA model and share it with your class.

**What do you think?**

Discuss the advantages and limitations of this model in helping you to understand the structure of DNA.
Extracting DNA

<table>
<thead>
<tr>
<th>What are the risks in doing this experiment?</th>
<th>How can you manage these risks to stay safe?</th>
</tr>
</thead>
<tbody>
<tr>
<td>Ethanol and methylated spirits are flammable and toxic.</td>
<td>Avoid handling ethanol or methylated spirits. Use latex gloves and/or a squeeze bottle. Do not use naked flames anywhere around ethanol or methylated spirits.</td>
</tr>
<tr>
<td>Laboratory desks and equipment may be contaminated with toxic chemicals.</td>
<td>Do not eat the fruit or fresh material provided. Use tongs to handle these items.</td>
</tr>
</tbody>
</table>

DNA is easily extracted from a variety of biological tissues, such as banana, kiwi fruit, onion, liver and wheat germ (a concentrated source).

Aim

To extract DNA.

Materials

- a source of DNA: 1 cm³ of banana, kiwi fruit, onion, liver or wheat germ
- mortar and pestle
- 1 teaspoon salt
- 1 tablespoon dishwashing detergent
- 10 mL buffer solution (8.8 g NaCl and 44 g sodium citrate per litre of water)
- 250 mL beaker
- funnel
- filter paper
- meat tenderiser
- ethanol or methylated spirits (ice-cold) in a squeeze bottle
- hooked Pasteur pipette or paperclip

Method

1. Place the DNA source into a mortar. Using the pestle, grind the material together with the salt, dishwashing detergent and the buffer solution. Note:
   - the physical grinding increases the surface area of cells in the sample that are in contact with the added substances
   - the detergent breaks down the oily cell membranes and proteins
   - the buffer helps the DNA come together.
2. Use the beaker, funnel and filter paper to filter the ground mixture. This removes the cell debris, but the cell nuclei will still pass through.
3. Add 1 tablespoon of meat tenderiser to the filtered material. This breaks down the nuclear membranes.
4. Add the ethanol by carefully allowing small amounts to run down the side of the beaker. Water is denser than alcohol, so the alcohol sits above it. After a few minutes the DNA, which is not soluble in alcohol, will precipitate on the interface of the two layers.
5. Lift the DNA out of the beaker using the hooked Pasteur pipette or paperclip.

Results

1. Identify the material from which you extracted DNA.
2. Using an annotated diagram, describe the appearance of the DNA that has been isolated.

Discussion

1. Construct a flowchart to show the steps in the method. Outline the purpose of each step.
2. Discuss whether you would expect the same results from different amounts of blending.
3. Explain the effect of the detergent on the cell membranes.
4. Explain why the alcohol remains in a layer on top of the liquid in the beaker.
5 Predict whether you would expect the results from different tissues to be similar. Compare your results with those of another group who used a different DNA source. Explain why the results are similar or different.

6 If the average plant cell contains 3 m of DNA and we consume 50 million cells in an average meal, calculate how many kilometres of DNA we eat in a week.

7 A person cannot see a single cotton thread 30 m away, but if you wound thousands of threads together into a rope, it would be visible from much further away. Explain how this statement is relevant to your procedure.

Conclusion

Compare the volume of strained DNA with the original amount of tissue you ground. Estimate the percentage difference.

Remembering

1 Define the following glossary terms: adenine, complementary base pairing, cytosine, deoxyribonucleic acid (DNA), double helix, genetics, guanine, nitrogenous bases, nucleotide, selective breeding and thymine.

2 Write out the full name of DNA.

3 Identify the basic subunit of a DNA molecule.

4 Identify the parts that make up a subunit of DNA.

5 Draw a labelled diagram to represent the structure of DNA.

6 Outline the meaning of complementary base pairing.

Understanding

7 Describe how DNA is like the alphabet.

8 Explain the relationship of DNA to genes.

9 Outline why a DNA molecule can be described as a ‘twisted ladder’.

Applying

10 Below is a sequence of DNA nucleotides.

   ATA TTG GGC GCC AAG ACT

Deduce the sequence of nucleotides on the complementary strand.

By the end of section 2.2 you will be able to:

- use models and diagrams to represent the relationship between DNA, genes and chromosomes.

2.2 Chromosomes and genes

Chromosomes

How does each microscopic cell nucleus fit 3 m of DNA? In cells, each DNA molecule is tightly coiled up with special proteins called histones to form chromosomes (Figure 2.3), much like cotton thread is coiled around plastic spools. Each chromosome contains one coiled molecule of double-stranded DNA and each DNA molecule contains many genes at specific locations along its length.
Figure 2.3 DNA is wound tightly around histones to form chromosomes. Chromosomes are often shown as they appear when they have replicated, because that is the stage at which they can be seen in cells. However, normally they are linear structures, not cross-shaped.

Activity 2.2.1

Looking at chromosomes
Using a microscope, you can see chromosomes in cells.

<table>
<thead>
<tr>
<th>What are the risks in doing this activity?</th>
<th>How can you manage these risks to stay safe?</th>
</tr>
</thead>
<tbody>
<tr>
<td>Microscopes are heavy and can injure you if dropped.</td>
<td>Use two hands to carry the microscope – one holding the ‘arm’ and one supporting the base.</td>
</tr>
<tr>
<td>Glass microscope slides can break easily and may cut you.</td>
<td>Handle prepared slides with care and clip them onto the microscope stage so they do not slip off while you are working.</td>
</tr>
</tbody>
</table>
**What you need**
- prepared slide of an onion root tip
- compound microscope

**What to do**
1. Set up your microscope according to your teacher’s instructions.
2. Place the prepared slide onto the stage and secure it with stage clips.
3. View on low power, moving the slide towards the tapered tip of the root until you have a clear view of the dividing cells.
4. Switch to high power.

**What did you discover?**
1. Draw and label one cell, clearly showing the chromosomes. Remember the rules for biological drawing (see Appendix 3).
2. State whether the chromosomes in different cells look alike or different. Explain why this is so.

**What do you think?**
Normally, you cannot see the chromosomes in a cell because they are too small to be seen by the naked eye. Propose two reasons why you can see the chromosomes in these cells.

**Chromosome numbers**
Different species have different numbers of chromosomes in their body cells (Table 2.1). Humans have 46 chromosomes: that is, 23 pairs of chromosomes. Twenty-two of these pairs are known as *autosomes*. They carry genes that determine many thousands of characteristics. The remaining pair determines the individual's sex and are known as the *sex chromosomes*. Females have two X chromosomes, whereas males have one X and one Y chromosome. The full set of human chromosomes is shown in Figure 2.4.

**Figure 2.4** Humans have 23 pairs of chromosomes, 46 in total. Shown here are the chromosomes of a male: there is one X chromosome and one Y chromosome.
### Table 2.1 The chromosome numbers of some species

<table>
<thead>
<tr>
<th>Species</th>
<th>Chromosome number</th>
</tr>
</thead>
<tbody>
<tr>
<td>Garden pea (<em>Pisum sativum</em>)</td>
<td>14</td>
</tr>
<tr>
<td>Plains lubber grasshopper (<em>Brachystola magna</em>)</td>
<td>24</td>
</tr>
<tr>
<td>Fire salamander (<em>Salamandra salamandra</em>)</td>
<td>24</td>
</tr>
<tr>
<td>European sea urchin (<em>Echinus esculentus</em>)</td>
<td>38</td>
</tr>
<tr>
<td>Human (<em>Homo sapiens</em>)</td>
<td>46</td>
</tr>
<tr>
<td>Dog (<em>Canis lupus familiaris</em>)</td>
<td>78</td>
</tr>
<tr>
<td>Black rhinoceros (<em>Diceros bicornis</em>)</td>
<td>84</td>
</tr>
</tbody>
</table>

### Genes

From the mid-20th century, scientists knew there was a relationship between the DNA in the cell’s nucleus and the many different proteins found in the cytoplasm. Proteins have many functions within a living organism. For example, they can act as enzymes (such as in digestion) or provide structure to cells (the cell membrane contains many different proteins). All proteins are made of many subunits called amino acids. Different proteins contain different amounts of amino acids in different orders.

Scientists discovered from experiments that DNA provides the plans for the production of all the proteins produced by cells, and that the order of base pairs in the DNA determines the order of amino acids in the protein. Along the length of each DNA molecule, particular regions code for different proteins. These regions are called genes (Figure 2.6). Genes are arranged in a line along the DNA molecule. Usually, each gene codes for one protein. Each DNA molecule has many genes, made up of many bases, located along its length.

![Figure 2.6](image)

**Figure 2.6** A gene is a section of DNA that codes for a particular protein. There can be different versions of the same gene: this is genetic variation.

### Activity 2.2.2

**A genetic city**

An analogy is a comparison of one thing with another. It can be used to illustrate the relationship between nucleotides, DNA molecules, chromosomes and genes. Create an analogy between these concepts and a city. Include a house, street, suburb and city in your analogy.

Try to think of another analogy to show the relationship between these concepts.
Questions 2.2

Remembering
1. Define the following glossary terms: amino acid, autosome, chromosome, gene, protein and sex chromosome.
2. Describe where genes are located in a cell.
3. Explain how very long DNA molecules can fit into the nuclei of cells.
4. Identify the components of a chromosome.

Understanding
5. Distinguish between autosomes and sex chromosomes.
6. Describe the difference in appearance between the human X and Y chromosomes.
7. Human chromosomes are numbered from 1 to 22 plus X and Y. Relate the number of the chromosome to its size.
8. Describe the relationship between genes, amino acids and proteins.

Applying
9. Two pieces of DNA have different sequences of nucleotides. Predict how this affects the proteins produced from them.
10. A dividing cell from an unknown source has been prepared and stained. Explain how you could tell if it was likely to be from a human and, if so, what sex the human was.

By the end of section 2.3 you will be able to:
- recognise that genetic information is passed to offspring through a gamete from both parents
- understand that gametes are produced by the process of meiosis and contain half the amount of DNA of other cells
- recall that the full amount of DNA is restored at fertilisation

Working on your showcase
You can use the knowledge and skills from section 2.2 to:
- identify a genetic condition and the human chromosome on which this genetic condition occurs.

2.3 Passing on genes

Sexual reproduction occurs when a male and a female parent produce offspring that are genetically different from their parents and from each other. It is one of the ways in which genetic variation is maintained in populations. This is how humans and many other species reproduce.

Both parents pass on genes

Sexual reproduction involves the fusion of male and female gametes (sex cells) to form a zygote (fertilised egg). Gametes carry the instructions for such family characteristics as eye colour, hair colour and height from one generation to the next. Up until the early 1900s, biologists were puzzled about how these microscopic cells could store so much information. The best explanation seemed to be that gametes carry information for the thousands of characteristics in a coded chemical form.

Improvements in technology and scientific procedures allowed scientists to conduct experiments to test this hypothesis. In 1902, biologist Theodor Boveri (1862–1915) showed that a fertilised egg contains a combination of genetic material from both parents, with 50% of chromosomes coming from the maternal (mother’s) side of the family and the other 50% from the paternal (father’s) side of the family.

How do the genes from our parents end up in our cells, in various combinations? The answers involve how chromosomes are sorted and passed on from one generation to another during a process called meiosis.

Maintaining chromosome number

How do gametes form so that the right amount of genetic material from each parent is passed on to the next generation? Before sexual reproduction, particular cells divide by meiosis. This produces sperm or ova that contain the correct number of chromosomes (and genes) to be passed on to the offspring.

In body cells, or somatic cells, each cell has two copies of every chromosome. These cells are described as diploid because they contain two full sets of chromosomes. Diploid number is often shown as 2n. Each pair of chromosomes is made up of a chromosome that
was inherited from the mother (a maternal chromosome) and one inherited from the father (a paternal chromosome). The pairs of chromosomes are called homologous chromosomes. Figure 2.4 (page 000) shows the homologous pairs of chromosomes in a human cell.

Belgian embryologist Edouard Van Beneden (1846–1910) discovered that gametes have half the number of chromosomes of other cells in the body. He observed eight chromosomes in the body cells of horse roundworm but found only four in its gametes.

The process of meiosis produces gametes with half the total number of chromosomes of normal body cells. Gametes only have one copy of each chromosome. They are described as haploid (and the haploid number is represented as $n$). So when the two gametes – the sperm and the ovum – fuse at fertilisation, the total complement of chromosomes for the organism is restored. The resulting fertilised egg or zygote has the diploid ($2n$) number.

$$n + n = 2n$$

**Making gametes: meiosis**

Species that reproduce sexually produce gametes by the process of meiosis. For example, humans produce cells that instead of the usual 46 chromosomes, contain only the haploid number of 23. Meiosis refers to the division of the cell nucleus (the division of the cell itself is called cytokinesis). In multicellular organisms such as humans, meiosis occurs in specialised gamete-producing structures called the gonads (the ovaries of females and testes of males).

Meiosis is preceded by interphase, during which DNA replicates. When meiosis begins, the chromosomes condense into the cross-shaped form we see when we view prepared cells under the microscope. Each vertical half of the replicated chromosome is known as a chromatid. Meiosis occurs in two stages. During meiosis I, one cell divides into two. During meiosis II, these two cells further divide, resulting in four cells. Each stage consists of a number of phases (Figure 2.8). Two crucial things happen during meiosis I. First, crossing over (during prophase I) occurs between homologous chromosomes: sections of DNA are exchanged and this results in the four chromatids each having a different combination of genes (Figure 2.9, page 000). Second, homologous chromosomes separate (during anaphase I). During meiosis II, the sister chromatids divide and haploid cells are formed.

**Interphase**

Most of a cell’s life takes place in interphase, during which chromosomes are not tightly coiled, and protein is being made using the instructions they contain. In this stage, the cell replicates the chromosomes. The replicated chromosomes are now made up of two identical chromatids, joined at the centromere (Figure 2.7).

**Figure 2.7** A model of a replicated chromosome; the section where the two sister chromatids are joined is called the centromere.
### MEIOSIS I

<table>
<thead>
<tr>
<th>Stage</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Prophase I</td>
<td>Chromosomes coil and become visible when stained. Nuclear membrane disappears. Spindle forms and attaches to the centromere. Homologous chromosomes move towards one another. Crossing over occurs (pieces of homologous chromosomes are exchanged).</td>
</tr>
<tr>
<td>Metaphase I</td>
<td>Homologous chromosomes align in the middle of the cell.</td>
</tr>
<tr>
<td>Anaphase I</td>
<td>Homologous chromosomes move apart. The side to which the maternal and paternal chromosomes go is random. Note that sister chromatids are still attached to each other at the centromere.</td>
</tr>
<tr>
<td>Telophase I</td>
<td>One chromosome of each pair reaches the opposite poles of the cell. In some organisms the nuclear membrane may re-form, and the chromosomes become less visible. Cell begins the reduction division, in which the number of chromosomes will be halved.</td>
</tr>
<tr>
<td>Daughter cells</td>
<td>The two daughter cells are haploid. Sister chromatids each have some material from the homologous pair.</td>
</tr>
</tbody>
</table>

### MEIOSIS II

<table>
<thead>
<tr>
<th>Stage</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Prophase II</td>
<td>Chromosomes condense. Spindle forms and attaches to the centromere. Nuclear membrane disappears.</td>
</tr>
<tr>
<td>Metaphase II</td>
<td>Chromosomes align in the middle of the cell.</td>
</tr>
<tr>
<td>Anaphase II</td>
<td>Sister chromatids separate at the centromere and move to opposite poles of the cell.</td>
</tr>
<tr>
<td>Telophase II</td>
<td>Chromatids reach the opposite poles of the cell. Cytokinesis (division of the cytoplasm) begins.</td>
</tr>
<tr>
<td>Daughter cells</td>
<td>After the second division there are four genetically unique haploid daughter cells.</td>
</tr>
</tbody>
</table>

Figure 2.8 The stages of meiosis
Meiosis I: shuffling and mixing DNA

Prophase I
A structure called a **spindle** starts to form. This is a network of thin protein fibres that stretches across the cell. Chromosomes attach to the spindle at the centromere and are moved along it. The spindle moves homologous chromosomes towards one another and starts to pair them at the **equator**. Crossing over occurs, resulting in different combinations of DNA in the four sister chromatids (Figure 2.9). Most homologous chromosomes undergo crossing over at least once. This is an important source of variation in sexually reproducing organisms, because it produces new combinations of traits.

Metaphase I
The homologous chromosomes line up along the equator of the cell in pairs so they face opposite sides of the cell. The spindle fibres are still attached to the centromere of each chromosome.

Anaphase I
During this phase of division, the spindle fibres contract and pull one chromosome of each homologous pair to either **pole** of the cell. Each pole receives a mixture of maternal and paternal chromosomes. This is called **independent assortment**.

Telophase I
During telophase I, nuclear membranes may form and **cytokinesis** occurs. Cytokinesis is the pinching of the cell membrane and the separation of cytoplasm and **organelles** into the two cells.

Meiosis II: separating sister chromatids
The two cells formed during meiosis I contain a chromosome from each homologous pair. These chromosomes still consist of chromatids joined by a centromere. During meiosis II, each chromosome splits at the centromere, allowing the two chromatids to move into separate cells. During telophase II, the nucleus re-forms and cytokinesis occurs, creating four haploid gametes. Each cell contains different combinations of genes and only half the original chromosome number.

The order of the phases in meiosis can be remembered by the acronym: IPMAT PMAT.

**Figure 2.9** A schematic diagram showing how crossing over during prophase I can produce four different chromatids

---

**Activity 2.3.1**

**The meiosis dance**
In a group of 8–10, stretch your legs and dance your way through the stages of meiosis. Each person should decide on a role to play. Use your textbook to follow the stages. Music is optional but will help you remember the stages.
Fertilisation
At the moment of fertilisation in humans, the haploid sperm and ovum fuse, bringing together the 23 chromosomes from the father and the 23 chromosomes from the mother. There are now 46 chromosomes in the zygote. This zygote grows and divides by the process of mitosis. Each time a cell divides, two identical daughter cells are produced. Each daughter cell has the same number and type of chromosomes as the initial cell (also called the parent cell). If the zygote inherits a Y chromosome from the sperm cell and an X chromosome from the egg cell, it will develop into a male. If it inherits two X chromosomes, one from the sperm and one from the egg, the zygote will develop into a female.

Activity 2.3.2
Determining sex with a game of ‘chance’
The image in Figure 2.10 is called a karyotype. A karyotype is made by taking an image of the chromosomes in the nucleus of a prepared cell and then arranging the chromosomes to show matching pairs. The paired chromosomes are ordered from largest to smallest.

Figure 2.10 A karyotype of chromosomes from a human

In humans, chromosome pair 23 are different from each other. As in most mammals, humans have one special pair of chromosomes that determines sex. Females have XX, two chromosomes of the same size. Males have XY, two chromosomes of very different sizes.

What you need
- coins or marked counters
- a human karyotype
What to do
When answering the questions, remember that only one half of the pair will go into any single gamete or sex cell.

1. Identify the sex chromosomes that females can pass to their gametes (ova).
2. Identify the sex chromosomes that males can pass to their gametes (sperm).
3. If a sperm cell carries a Y chromosome, deduce the sex of the foetus.
4. If a sperm cell carries an X chromosome, deduce the sex of the foetus.
5. Calculate the fraction of sperm cells that carry an X chromosome.
6. Calculate the fraction of sperm cells that carry a Y chromosome.
7. Toss a coin and use it to represent the kind of sperm that is produced – tails representing X, and heads representing Y – in three imaginary families with six children each. Record your results in the table.

<table>
<thead>
<tr>
<th>Family 1</th>
<th>Family 2</th>
<th>Family 3</th>
</tr>
</thead>
<tbody>
<tr>
<td>Toss</td>
<td>X or Y</td>
<td>Male (M) or female (F)</td>
</tr>
<tr>
<td>Toss</td>
<td>X or Y</td>
<td>Male (M) or female (F)</td>
</tr>
<tr>
<td>Toss</td>
<td>X or Y</td>
<td>Male (M) or female (F)</td>
</tr>
</tbody>
</table>

What did you discover?
1. Analyse the results to determine whether all of the imaginary families ended up with the same ratio of female and male children.
2. Assess whether the results of one coin toss changed the results of the next.
3. Calculate whether the resulting ratio approaches 50 : 50.
4. Explain why one family may have a different pattern of female and male offspring compared with the other families.

Remembering
1. Define the following glossary terms: anaphase, centromere, chromatid, crossing over, cytokinesis, diploid, diploid number ($2n$), equator, gamete, gonad, haploid, haploid number ($n$), homologous chromosomes, independent assortment, interphase, karyotype, maternal, meiosis, paternal, pole, prophase, somatic cell, spindle and zygote.
2. Distinguish between a gamete and a zygote.
3. Describe what happens in crossing over.

Understanding
4. Explain where the chromosomes in our own cells come from.
5. Explain why gametes only have one copy of each chromosome pair.
6. Relate the concept of a chromosome to a chromatid.
7. Explain how gametes, DNA and genes each play a part in passing on heritable characteristics during reproduction.

Applying
8. A kangaroo species has 12 chromosomes in each of its body cells.
   a. Explain what is meant by the diploid number ($2n$) of chromosomes.
   b. State how many autosomes there are in each cell.
   c. Propose how many sex chromosomes are in each sperm of a male kangaroo.
   d. Calculate the haploid number, $n$, for this kangaroo species.
9. A platypus has 52 chromosomes.
   a. Explain how many chromosomes came from its mother.
   b. State the diploid number of a platypus.
   c. Determine how many homologous pairs are present.
d State the number of chromosomes present in the sperm of a male platypus.
e Explain why the sperm must have this number of chromosomes.

10 A man and a woman plan to have a family. Calculate the probability that:

a their first child will be a son
b their second child will be a son
c they will have a daughter, a son and then another daughter
d they will have a son and a daughter, if they have two children.

2.4 Mutations

Mistakes or mutations can happen during DNA replication when cells are dividing for body growth (mitosis). They can also occur during DNA replication before meiosis, or during meiosis itself, giving rise to a change in the DNA code or chromosome structure.

The consequences of a mutation depend on whether it occurs in a body cell or in a gamete. A mutation in a body cell occurs only in the affected cell and the daughter cells produced from it by mitosis. All other cells of that individual lack the mutation. Mutations during cell division in body cells can cause tumours and cancer.

Mutations that occur in gametes have the potential to be inherited. If passed on to the next generation they are incorporated into every cell of the offspring. If the mutation results in developmental abnormalities the affected embryo or foetus may fail to develop and will often spontaneously abort. If carried through to birth, the mutation may result in a congenital disorder in the offspring. Only mutations that occur during meiosis can be passed on to offspring. Mutations are an important source of variation within a species.

There are many kinds of mutations. Some mutations are very harmful, but some can be beneficial. Occasionally, mutations produce a variation that is neither harmful nor beneficial to the organism. For example, harmless mutations can cause variations in hair colour and skin tone. They are not life-threatening and so they are not eliminated from the population.

Some mutations are ‘silent’ and do not produce a noticeable effect on the individual. For example, sometimes one nucleotide in a DNA sequence is replaced with a different one, but the protein that the gene codes for remains unchanged.

Causes of mutations

Mutations can happen spontaneously during DNA replication. However, cells have DNA repair mechanisms that are highly effective so spontaneous mutations are rare. In 1927, American biologist H. J. Muller found that many more mutations occurred in fruit fly (Drosophila melanogaster) exposed to X-rays. Since then it has been found that other factors in the environment (mutagens) can also increase the rate of mutation.

Physical mutagens

Physical mutagens include various types of radiation that cause DNA damage, such as ultraviolet (UV) light, X-rays and nuclear radiation. Exposure to excessive UV rays from the Sun can cause mutations in dividing skin cells. If the body is unable to repair this damage an affected cell can begin to divide and grow in an uncontrolled way. This growth can eventually form a tumour. Approximately 99% of non-melanoma skin cancers and 95% of melanoma are caused by UV radiation.

Chemical mutagens

Chemical mutagens change genetic material and so increase the rate of mutation. The way chemical mutagens affect genetic material varies. Some have similar structures to normal DNA nucleotides and can become incorporated into DNA during replication. This can lead to one base in the DNA sequence being replaced by another. Some chemical mutagens change the structure of bases and others slip between bases.
Cigarette smoke contains many chemicals which are mutagens and are known to cause cancer. Evidence shows clear links between tobacco smoking and lung cancer (and many other types of cancer). Polyaromatics are a type of chemical mutagen found in cigarettes. They can damage DNA, including genes that protect against cancer.

**Biological agents**

Genetic mutations sometimes arise because of the action of biological agents such as bacteria and viruses. Some viruses, for example, are capable of incorporating their genetic material into the genetic material of the cells they invade, causing mutations in subsequent generations.

The human papillomavirus (HPV) (Figure 2.13) can infect human skin cells and membranes and transfer its DNA to the nuclei of cells it infects (host cells). Here it is copied dozens of times and inserted into the host cell DNA. HPV genes shut down any DNA repair that the cell would normally carry out. The infected cells replicate at an increased rate, which means the virus is also replicated more. The cells divide out of control and any mutations that arise during cell divisions, even those not associated with HPV, are not repaired. An HPV infection often leads to cancer. HPV causes over 99% of cervical cancers, the second most common form of cancer in women worldwide.

### Changes to chromosome structure

Changes to chromosome structure most often happen when two or more double-strand breaks occur in chromosomes, and the broken segments are rearranged. Some of these breaks occur naturally during meiosis, when the chromosomes are entangled with one another, crossing over and moving apart. Others occur because of exposure to mutagens, which cause more double-strand breaks. The breaks are normally repaired, but sometimes the segments are mistakenly relocated in the repaired chromosomes. There are different types of chromosome rearrangements (Figure 2.15).

- **Deletion.** A segment of chromosome is missing (Figure 2.14).
- **Inversion.** A chromosome breaks and the segment in the middle rotates through 180° before being re-joined within the chromosome.
- **Translocation.** A section of one chromosome breaks off and attaches to another chromosome.
- **Duplication.** An extra copy is made of a section of chromosome and inserted either into the same chromosome or into another chromosome.

![Figure 2.13](https://example.com/figure2_13.png)

*Figure 2.13* The human papillomavirus can cause cervical cancer in humans. In 2006, a vaccine that protects against HPV, developed by Australian medical scientist Ian Frazer and Chinese virologist Jian Zhou, was first used.

![Figure 2.14](https://example.com/figure2_14.png)

*Figure 2.14* Canadian actor and singer Gabrielle Marion-Rivard has Williams syndrome: the condition arises because of a deletion of approximately 1.5 million nucleotide pairs from a copy of chromosome 7.

![Figure 2.15](https://example.com/figure2_15.png)

*Figure 2.15* Chromosomal mutations include a) deletion, b) inversion, c) translocation and d) duplication.
Questions

1. Define the following glossary terms: congenital disorder, mutagen and mutation.
2. Identify the processes during which mutations can occur.
3. Outline some of the changes that can result because of mutations in DNA.
4. Explain why some mutations are ‘silent’.
5. Identify three factors that can increase the rate of mutations.

Understanding

6. Describe the differences in the effects on an individual of mutations in body cells compared with mutations in gametes.
7. Classify the cause of a mutation (physical, chemical or biological) in the following situations:
   a. insertion into a cell of genes from a virus
   b. exposure to cosmic radiation during space flight
   c. a banned food preservative causing cancers in laboratory animals.
8. Describe the events that result in the following types of chromosomal mutations.
   a. Inversion
   b. Duplication
   c. Deletion
   d. Translocation
9. Explain why spontaneous mutations are relatively rare.

Applying

10. Many of the people exposed to nuclear radiation from the Chernobyl nuclear disaster in 1986 and from the flooding of the Fukushima nuclear plant in 2011 developed cancer. Some cancers are the result of chromosomal damage. Account for the high rate of cancer in these people using your knowledge of factors affecting mutation rates.

By the end of section 2.5 you will be able to:

- represent patterns of inheritance of a simple dominant/recessive characteristic through generations of a family.

2.5 Patterns of inheritance

If we understand inheritance patterns, we can predict how likely it is that an individual will pass on an inherited condition to the next generation. The first steps in understanding inheritance were made in the 19th century by Austrian Augustinian monk Gregor Mendel (1822–84).

Mendel’s studies of inheritance

Mendel performed breeding experiments with a species of self-fertilising garden pea, *Pisum sativum* (Figure 2.16). He noticed that certain varieties had the same characteristics appearing generation after generation. Mendel decided these plants were pure-breeding.

Mendel investigated what happened when two pure-breeding varieties of peas that differed in one characteristic were crossed; for example, when a round-seeded pea plant was crossed with a wrinkled-seeded pea plant. He removed the anthers of unopened flowers of a round-seeded pure-breeding plant (so that it could not pollinate itself) and brushed its stigma with pollen from a wrinkled-seeded pure-breeding plant. These first two plants were called the parental generation (P).

In these experiments, Mendel observed that the offspring plants, called the first filial (F₁) generation, all resembled only one of the parent plants. In this case, they all had round seeds. He called the feature they showed the dominant trait.

However, when the F₁ plants self-fertilised, in the next generation, or second filial (F₂) generation, most plants resembled the dominant parent (round seeds), but some resembled the non-dominant parent (wrinkled seeds). Mendel inferred that the non-dominant trait must have been carried, hidden, within the F₁ generation plants. He called the F₁ plants that carried the hidden trait hybrids (which means of mixed origin) and used the term recessive trait for the trait that could be carried or hidden.
Mendel’s results
Mendel investigated seven pairs of features of pea plant in thousands of P, F1, and F2 plants. Use the data from Table 2.2 to answer these questions.

1. Explain why Mendel used so many plants to perform crosses of the F1 generation.
2. Using the F2 results, calculate the ratio of each pair of features (round your results to the nearest whole number). Describe the pattern that emerges.

Table 2.2 Mendel’s original data from his experiments on pea plants.

<table>
<thead>
<tr>
<th>Feature</th>
<th>P Original parental characteristics</th>
<th>F1 Dominant trait revealed (number of plants crossed)</th>
<th>F2 Number of plants showing each parental trait</th>
</tr>
</thead>
<tbody>
<tr>
<td>1 Form of ripe seed</td>
<td>Round</td>
<td>Round (60 fertilisations on 15 plants)</td>
<td>5474 round</td>
</tr>
<tr>
<td></td>
<td>Wrinkled</td>
<td></td>
<td>1850 wrinkled</td>
</tr>
<tr>
<td>2 Colour of seed</td>
<td>Yellow/orange</td>
<td>Yellow/orange colour (58 fertilisations on 10 plants)</td>
<td>6022 yellow/orange</td>
</tr>
<tr>
<td></td>
<td>Bright green</td>
<td></td>
<td>2001 bright green</td>
</tr>
<tr>
<td>3 Colour of seed coat</td>
<td>Brown</td>
<td>Brown (35 fertilisations on 10 plants)</td>
<td>705 brown</td>
</tr>
<tr>
<td></td>
<td>White</td>
<td></td>
<td>224 white</td>
</tr>
<tr>
<td>4 Form of ripe pods</td>
<td>Inflated</td>
<td>Inflated (40 fertilisations on 10 plants)</td>
<td>882 inflated</td>
</tr>
<tr>
<td></td>
<td>Constricted</td>
<td></td>
<td>299 constricted</td>
</tr>
<tr>
<td>5 Colour of unripe pods</td>
<td>Dark green</td>
<td>Dark green (23 fertilisations on 5 plants)</td>
<td>428 dark green</td>
</tr>
<tr>
<td></td>
<td>Yellow</td>
<td></td>
<td>152 yellow</td>
</tr>
<tr>
<td>6 Position of flowers</td>
<td>Along stem</td>
<td>Along stem (34 fertilisations on 10 plants)</td>
<td>651 along stem</td>
</tr>
<tr>
<td></td>
<td>Top of stem</td>
<td></td>
<td>207 top of stem</td>
</tr>
<tr>
<td>7 Length of stem</td>
<td>Tall</td>
<td>Tall (even taller than parent) (37 fertilisations on 10 plants)</td>
<td>787 tall</td>
</tr>
<tr>
<td></td>
<td>Short</td>
<td></td>
<td>277 short</td>
</tr>
</tbody>
</table>

Mendel recognised that each F2 generation showed the dominant and recessive features in an approximate 3:1 ratio, respectively. This is now known as the Mendelian ratio. By mathematically analysing his results, Mendel concluded that the information for traits was carried in pairs. Only half of the information was passed on from each parent and fertilisation combined the halves back into a pair. The appearance of the offspring was determined by the information that was paired together. That he was able to reach these conclusions is amazing, because Mendel had no knowledge of the structure of DNA, the existence of genes or the process of meiosis.

With our understanding of chromosomes, genes and meiosis, we can see how Mendel’s discoveries match what actually happens in cells. For every gene (such as the one controlling seed shape) on one chromosome, there is a corresponding gene on the homologous chromosome. However, the word ‘gene’ had not been coined in Mendel’s time. He referred to ‘factors’. He discovered that in pea plants, there are alternative forms of these factors (genes), such as round or wrinkled seeds. In modern genetics, these different forms of the same gene are called alleles. Some alleles code for dominant characteristics, and some code for recessive characteristics (and some alleles have effects that are neither dominant nor recessive).

Mendel predicted and confirmed that when F1 hybrids are crossed, the resulting F2 plants showing the dominant trait do not all have the same combination of factors (alleles) (Figure 2.18, page 41). He suggested that some of the F2 plants expressing the dominant trait had two similar alleles for roundness. The pure-breeding plants are called homozygous (‘homo’ means same) because they produce sex cells (gametes) that all carry the same type of allele.
He suggested that the other two-thirds of the F2 plants expressing the dominant trait carry two different forms of the gene: one allele for the dominant trait (roundness) and one allele for the recessive trait (wrinkled). He named these types of plants *heterozygous* (hybrid) and claimed they produce sex cells (gametes) that carry either the allele for the dominant trait or the allele for the recessive trait. Finally, he concluded that if a recessive trait is to be expressed, the individual requires two copies of the allele for the recessive trait. Therefore, plants that express the recessive trait (wrinkled) are pure-breeding (homozygous).

### Activity 2.5.2

**Careers with plants and flowers**

Today, many careers that involve plants and flowers depend on understanding genetics.

1. List five possible careers.
2. Select one career from your list. Explain the career, the science involved, where people in this field work and anything else of interest. Provide a weblink where people can get more information about this career.

### Explaining and predicting the Mendelian ratio

The set of rules Mendel devised to explain his observations is still used. Using modern-day genetic terminology, Mendel’s rules are as follows.

1. Genes exist in a variety of forms (alleles). For example, the gene for seed type in a pea species has two different alleles, round and wrinkled.
2. Alleles occur in pairs in individuals – that is, one allele occurs on the maternal chromosome and the other occurs on the paternal chromosome of a pair of homologous chromosomes.
3. Alleles that code for dominant features are represented with a capital letter and alleles that code for recessive features are represented by the same letter but in lower case. For example, the allele for the dominant trait round seed is given the letter \( R \) and the allele for wrinkled seeds is denoted \( r \).
4. Pure-breeding (homozygous) varieties, such as peas that carry the allele for round seeds on both chromosomes (\( RR \)), will have the same allele on each chromosome in the homologous pair. Their gametes will all contain one chromosome with only an \( R \) allele. Similarly, the gametes of pure-breeding (homozygous) peas with wrinkled seeds (\( rr \)) have only the \( r \) allele. The representation of the alleles \( RR \) and \( rr \) for a characteristic is called the *genotype*.
5. Fertilisation combines two gametes. By convention, the capital letter for the dominant feature is shown first. For example, if the homozygous parents \( RR \) and \( rr \) are crossed, all \( F_1 \) plants will be heterozygous \( Rr \). These plants have round seeds because they have the dominant trait \( R \). The appearance of the plants is called their *phenotype*.
6. \( F_1 \) plants can produce gametes containing either allele; in our example, \( R \) or \( r \). Crossing \( F_1 \) plants produces \( F_2 \) plants in Mendelian ratios (see page 000). In this case, these will be:
   - \( RR \) homozygous for round seeds
   - \( Rr \) heterozygous (hybrids) – all these have round seeds
   - \( rr \) homozygous for wrinkled seeds (Figure 2.17).
Ratios in the F₁ generation

The genetic composition (the genotype) and the physical appearance (the phenotype) can be expressed as follows (Figure 2.18).

Genotypic ratio of $RR : Rr : rr$

1 $RR : 2 Rr : 1 rr$

Phenotypic ratio of round seeds to wrinkled seeds:

3 round seeds : 1 wrinkled seed.

Figure 2.18 Crossing $F_1$ pure-breeding round and wrinkled seed plants results in a 3:1 ratio in the $F_2$ generation.
**Activity 2.5.3**

**The Mendelian ratio**

*Drosophila melanogaster* (fruit flies) are a popular species to use to study Mendelian genetics (Figure 2.19). They have short generation times and many features that are each controlled by a single gene.

1. There is a gene for eye colour in fruit flies that has two alleles: red eyes (*R*) and white eyes (*r*). Work out the following crosses.
   - a. Show what would happen in a cross between a pure-breeding red-eyed fruit fly and a pure-breeding white-eyed fruit fly. Identify the ratio you would expect in the *F*₁.
   - b. Show what would happen in a cross between a fruit fly that is heterozygous for eye colour with one that is homozygous recessive. Identify the ratio you would expect in the *F*₁.

2. The gene for wing length in fruit flies has two alleles: long wings (*W*) and short wings (*w*). Show what would happen in a cross between two heterozygous fruit flies. Identify the ratio of wing length you would expect to see in the offspring.

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**Representing patterns of inheritance**

Inheritance patterns show how traits are passed to offspring over several generations. There are several ways of showing these inheritance patterns, including Punnett squares and pedigrees.

**The Punnett square**

English geneticist Reginald Punnett (1875–1967) is best known for his creation of the *Punnett square*. This is a table that allows geneticists to easily predict the probability that offspring from certain parents will inherit particular characteristics. A Punnett square summarises the combination of genes in gametes in genetic crosses. Punnett’s work helped to confirm and extend Mendel’s ideas.

For example, consider the characteristic of seed colour in Mendel’s pure-breeding plants, where yellow/orange is dominant (*Y*) and green is recessive (*y*). The parental cross between the two pure-breeding lines can be represented as follows.

\[ YY \times yy \]

During meiosis, the chromosomes containing these alleles are separated when gametes form. One parent will produce gametes that all contain the *Y* allele. The other parent will produce gametes that all contain the *y* allele. This can be shown as a Punnett square.

<table>
<thead>
<tr>
<th>Male gametes</th>
<th>All Y</th>
<th>Female gametes</th>
<th>All y</th>
<th>All Yy</th>
</tr>
</thead>
</table>

The possible *F*₁ genotypes are shown in the blue square. All of the *F*₁ will be *Yy*.

If we then cross the *F*₁ generation, we can use a Punnett square to predict the probability of obtaining a plant with bright-green seeds in the *F*₂. The *F*₁ cross is:

\[ Yy \times Yy \]

Each parent can produce two types of gametes: half the gametes will contain the *Y* allele and the other half will contain *y*.

Using the Punnett square, show the gametes from both parents and predict the probability of producing a plant expressing the recessive allele.
We can see from the Punnett square that there is a one-in-four, or 25%, chance that the cross between \textit{Yy} and \textit{Yy} will produce a plant with bright-green seeds, genotype \textit{yy}. What is the probability that they would produce a plant with yellow/orange seeds? Don't forget that this phenotype results from both \textit{Yy} and \textit{YY} genotypes.

### Activity 2.5.4

#### Punnett squares

1. Use Punnett squares to predict the ratio of possible offspring from the following crosses.
   - a. ‘Form of ripe seeds’ trait: homozygous \textit{rr} × heterozygous \textit{Rr}
   - b. ‘Colour of the unripe pods’ trait: heterozygous dark green \textit{Gg} × homozygous dark green \textit{GG}
   - c. ‘Colour of the seed’ trait: bright green seeds \textit{yy} × heterozygous yellow/orange \textit{Yy}
   - d. ‘Length of the stem’ trait: homozygous tall plants \( \times \) short plants

2. Explain the difference you would get in offspring in Question 1e if the parental tall plants are all heterozygous.

3. Explain why it is not necessary to say in Question 1e that the short plants are homozygous.

#### Pedigrees

Pedigrees are charts that organise information about a family and represent the family tree using symbols. They can help to determine both the mode of inheritance of certain genetic conditions and the genotype of particular individuals.

Some common symbols used to construct a pedigree are shown in Figure 2.20.

![Pedigree symbols](image)

**Figure 2.20** The symbols used in creating pedigrees

The Roman numerals on the left side of Figure 2.21 indicate there are three generations in the pedigree. Each individual is labelled from left to right; this is shown by the number given to each individual. This pedigree shows that the grandparents (I1 and I2) had four children: two girls (II2 and II3), a boy (II5) and then another girl (II7). The eldest daughter married individual III1, the second daughter married individual II4 and the son married individual II6. Together, these three couples had nine children in the third generation (III1–9).
The condition shown in the pedigree in Figure 2.21 – albinism – skipped a generation. No individuals in generation II show the condition. From this we can assume that the mode of inheritance is recessive. Because albinism appears to affect the males and females in similar frequency, we can also assume the gene is not carried on the X chromosome. Therefore, it is carried on an autosomal chromosome and is recessive. It is therefore known as an autosomal recessive condition.

The genotype of each individual can also be determined. We can use the letter $A$ to represent an individual with melanin (the dominant condition) and $a$ to represent an individual without melanin, or with albinism (the recessive condition). Individual II1 partners with an individual heterozygous for this condition (II2) and produces an albino son (III2). This means that individual II1 must be heterozygous. Individuals III1 and 3 can be either homozygous dominant or heterozygous – there is no way of telling which. Their genotypes are written as $A_-$, because the missing allele could be $A$ or $a$.

**Figure 2.21** A family pedigree for albinism

---

**Activity 2.5.5**

**Pedigrees**

Pedigrees are charts that show the inheritance of a gene through a family over several generations. They can be used to track a genetic disease and to predict the likely outcome of offspring inheriting that disease.

**Pedigree 1: Huntington’s disease**
Huntington's disease (dominant trait) is a degenerative disease of the nervous system. It leads to muscle decline, lack of coordination and dementia.

1. Identify the genotype of individual I2. Describe how you arrived at your answer.
2. Identify the genotype of individual II1. Describe how you arrived at your answer.
3. Individual II1 marries an unaffected man. Using a Punnett square, work out the likelihood of passing HD on to their three children.
4. Show the third generation by extending the pedigree.

**Pedigree 2: Sickle-cell anaemia**

Sickle-cell anaemia is a disease in which the red blood cells of homozygous sufferers change shape under conditions of low oxygen, blocking capillaries.

5. Work out the genotypes of each individual in the pedigree.
6. State if sickle-cell anaemia is dominant or recessive. Describe how you arrived at your answer.
7. Individual III2 marries a carrier of sickle-cell anaemia. Work out the likelihood that their firstborn will suffer from sickle-cell anaemia by:
   a. using a Punnett square
   b. extending the pedigree.

**Remembering**

1. Define the following glossary terms: allele, dominant trait, first filial (F₁) generation, genotype, heterozygous, homozygous, hybrids, Mendelian ratio, parental generation (P), pedigree, phenotype, Punnett square, pure-breeding, recessive trait and second filial (F₂) generation.
2. Distinguish between:
   a. gene and allele
   b. genotype and phenotype
   c. heterozygous and homozygous.
3. Explain the relationship between the P, F₁ and F₂ generations.
4. Identify two uses of pedigree charts.

**Understanding**

5. a. Identify three dominant traits that Mendel investigated.
   b. Explain how he determined whether a trait was dominant or recessive.
6. Explain who Reginald Punnett was and the contribution he made to genetics.
7. Explain how Mendel concluded, using mathematical analysis, that information for traits was carried in pairs.
2.6 Genetic testing

Symptoms of Huntington’s disease only appear when an individual is an adult. If a family is affected by Huntington’s disease – that is, one or more members of the family have a copy of the allele that causes the disease, \( H \) – early detection can be extremely useful. Scientists need to be able to test for the \( H \) allele in family members so they can inform them of their chance of developing the disease. The \( H \) allele and many other alleles can be detected by using genetic testing. Genetic testing identifies changes to genes (Figure 2.22).

Applying

8 A widow’s peak (a hairline shape) is dominant over the recessive form (a straight hair line). A woman homozygous for widow’s peak has a child with a heterozygous father. Calculate the probability that the child will carry the recessive allele. Use a Punnett square to show your working.

9 The neurodegenerative condition Huntington’s disease usually develops when the person is around the age of 40. It is a dominant condition.
   a Describe what is meant by ‘dominant condition’.
   b A heterozygous individual had a child with an unaffected individual. Use a Punnett square to determine the probability that the child will have Huntington’s disease.

10 The gene for hair curliness has two alleles: \( C \) and \( c \). Explain how two parents with curly hair (\( Cc \)) are able to have children with straight hair.

By the end of section 2.6 you will be able to:
• consider how information technology can be applied to bioinformatics and DNA sequencing
• consider the use of genetic testing.

Working on your showcase

You can use the knowledge and skills from section 2.5 to:
• identify the pattern of inheritance for the condition.

DNA sequencing

DNA sequencing is a technique which is used to find the exact sequence of nucleotides in a length of DNA. It can detect differences in DNA from different individuals and identify
individuals with mutations. In 2001, after 10 years of work and at a cost of US$400 million, the sequencing of human DNA, called the Human Genome Project, was completed. A draft sequence of the human genome, the sum of all DNA in a human cell, was published.

This significant branch of molecular biology has been made possible by advances in technologies, particularly robotics and bioinformatics. Sequencing DNA used to be slow and expensive. Automated processes in laboratories now make it possible to rapidly collect and store huge amounts of data, as well as enabling scientists to integrate, analyse and manipulate data at high speed. Individual genes can now be sequenced routinely. An entire genome can be sequenced for just US$1000 in 26 hours.

Comparisons between people’s DNA can yield an enormous amount of information about the role of inheritance in susceptibility to disease and how a person’s health could be affected by environmental influences. DNA sequencing can be helpful for diagnosing and treating conditions.

**Carrier testing and genetic counselling**

Genetic testing can be performed before birth or at any time during a person’s life, but is not available for all genes or all genetic conditions. Carrier testing is when genetic testing is used to identify people who carry one copy of a gene mutation that, when present in two copies, causes a genetic disorder. Testing is offered to individuals who have a family history of a genetic disorder and to people in certain ethnic groups with an increased risk of specific genetic conditions. If both parents are tested, the test can provide information about a couple’s risk of having a child with a genetic condition.

If a couple have already had a child with a congenital disorder, they will naturally be very anxious about the possibility of the same thing happening in later pregnancies. In other cases, one partner may have a genetic condition and the couple will be concerned that the problem could occur in their children. Other couples may have close relatives who have inherited diseases or have given birth to children with such disorders. In all these cases, the people concerned may seek advice about the risk of the inherited disorder occurring in their children. The advice given in these situations is called genetic counselling. Through genetic testing, the probability that a particular inherited condition will occur in their offspring can be determined.
Pre-natal genetic diagnosis
Medical scientists have developed ways of testing a foetus before birth for the presence of various genetic disorders. If a genetic disorder is diagnosed, the couple have the option of terminating the pregnancy.

If the pregnancy is being achieved by in-vitro fertilisation (IVF), a testing technique known as pre-implantation genetic diagnosis (PGD) can be used. This tests embryos for either a specific known genetic condition or a chromosome abnormality. PGD makes it possible to choose chromosomally normal embryos or those unaffected by a specific disorder for transfer during an IVF cycle, maximising the chance of a healthy baby.

Benefits and limitations of genetic testing
Whether the results come back positive or negative, genetic testing may benefit a person who is at risk of a genetic condition. They can feel a sense of relief from uncertainty, and knowing their status can help them make informed decisions about managing their health care.

Legal and medical authorities have raised concerns about genetic testing and the possibility of genetic discrimination in employment or insurance. Genetic discrimination is when people are treated unfairly because they have DNA sequences that increase their chance of getting a certain disease. For example, a health insurer might refuse to give coverage to a woman who has a gene that increases her chance of getting breast cancer. Employers could use DNA information to decide which workers to hire and fire.

Genetic tests often deliver probabilities rather than clear-cut predictions that an individual will develop a disease. Most health conditions are influenced by complex interactions between many different genes, and a person’s environment and lifestyle. Also, even in people with a definite genetic condition, the time of onset of the disease and how severe their symptoms are can vary widely.

Is more information always better?
Apolipoprotein E (APOE) is one of several proteins involved in blood transport of lipids. It occurs in three different versions. The e4 allele of the APOE gene increases an individual’s risk for developing Alzheimer’s disease. People who inherit one copy of the APOE e4 allele have an increased chance of developing the disease; those who inherit two copies of the allele are at even greater risk. It is important to note that people with the APOE e4 allele inherit an increased risk of developing Alzheimer’s disease, not the disease itself. Not all people with Alzheimer’s disease have the APOE e4 allele, and not all people who have this allele develop the disease.

Propose arguments for and against the following statements.
1 Insurance companies should have the right to ask about the results of genetic testing for the APOE e4 allele.
2 Doctors should be obliged to tell other family members the results of a patient’s genetic test for the APOE e4 allele.
Remembering
1 Define the following glossary terms: bioinformatics, carrier testing, DNA sequencing, genetic testing, genome, genetic counselling, in-vitro fertilisation (IVF) and pre-implantation genetic diagnosis (PGD).
2 Describe the role of a genetic counsellor.
3 Describe three circumstances in which genetic counselling is advised.
4 Compare the cost and time needed to sequence an entire genome when the first draft was published in 2001 with the cost and time now.

Understanding
5 Explain how advances in technology have allowed individual genes to be routinely sequenced.
6 Comparing gene sequences can be useful. Explain how.
7 Outline how carrier testing is used.
8 Identify at what stage of embryo development pre-implantation genetic diagnosis (PGD) takes place. Explain why it is done at this stage.

Applying
9 Commercial companies offer a service to sequence and analyse a consumer’s genome. Suggest why the results are expressed as probabilities of developing diseases rather than certainties.
10 A rare genetic disorder runs in someone’s family and a company is offering a genetic test for it. Identify two benefits and two concerns for the person if they take up the offer.

Questions 2.6
You can use the knowledge and skills from section 2.6 to:
- discuss whether the disease is serious enough to warrant genetic testing
- discuss where a person with the genetic condition could seek and find advice, support or others with the same condition.
CHAPTER REVIEW

REMEMBERING

1. List the base pairs in DNA.
2. Name and give an example of three causes of mutations.
3. Describe the difference in chromosome number between body cells and gametes.
4. Describe two ways of representing patterns of inheritance.

UNDERSTANDING

5. The choice of experimental organisms can make a difference to a scientist’s results. Discuss the advantage of using garden peas (Pisum sativum) for genetics experiments.
6. Draw a schematic diagram of a DNA molecule.
8. Contrast anaphase I and anaphase II.

APPLYING

9. Calculate the phenotypic ratio in the F₁ generation of Pisum sativum if two heterozygous round-seeded plants are crossed.
10. Some plants can have an extra copy, or two extra copies, of the whole chromosome set. This can produce larger fruits. The haploid number of a strawberry is \( n = 28 \). Calculate the diploid (\( 2n \)) and triploid (\( 3n \)) numbers for strawberries.
11. When two different species interbreed, their offspring are often infertile. A mule is the offspring of a female horse (\( 2n = 64 \)) and a male donkey (\( 2n = 62 \)). A hinny is the result of a male horse breeding with a female donkey. Explain why horse–donkey offspring are usually infertile. (Hint: Consider what happens in meiosis at metaphase I.)

12 a. Construct a pedigree to show the following information.
   - Two parents (generation I) had five children: two sons and three daughters, born in that order.
   - The eldest son (generation II) married a woman who suffered from the recessive condition sickle-cell anaemia. They had three children, all girls.
   - The eldest daughter (generation II) married a man with whom she had two children: a son and then a daughter.
   - The youngest daughter (generation II) married a man and they did not produce any children.

   b i. If the eldest son (generation II) was not a carrier of the recessive sickle-cell allele, calculate the chance that a fourth child for this couple would suffer from sickle-cell anaemia.
   ii. Calculate how many of the children of this couple would be carriers of the sickle-cell anaemia allele. State their genotype for this gene.
   iii. One daughter of this couple married a man who suffered from sickle-cell anaemia. Use a Punnett square to show the probability that their first child would also suffer from this disease.

13. Some herbicides, such as 2,4-D, work by stimulating cell division. This leads to uncontrolled growth. Explain how this makes these chemicals effective herbicides.
14. High levels of radiation cause damage to DNA. Explain why highly focused radiation therapy is used against some cancers.

ANALYSING

15. A yellow rat and a black rat mate and produce 12 offspring in the first mating and 9 in the second mating. Of these 21 offspring, 16 are yellow and 5 are black. From this information, determine the genotype of the parents in relation to coat colour (assuming only one gene is involved). Explain your logic.
16 Outline the benefits of combining information about a person’s DNA, medical history and family tree into a database. Discuss concerns people might have about this.

17 X-ray crystallography is a method of producing images that indicate the arrangement of the atoms in large molecules. British chemist Rosalind Franklin produced images that helped show DNA has a double helix structure and that the two outer strands consist of sugar and phosphate. Such images were used by Frances Crick and James Watson to deduce the structure of DNA. Rosalind Franklin died of ovarian cancer at the age of 37. Suggest any risks she may have been exposed to in her workplace and how these might have contributed to her early death.

18 Figure 2.24 shows a cell that is dividing. Explain why this cell cannot be undergoing the first stage of meiosis.

19 Using the pedigree in Figure 2.25, answer the following questions.
   a Who are the affected individuals?
   b Is the condition dominant or recessive? Provide evidence to justify your answer.
   c Write the likely genotypes for individuals II1, II4, II5, and III2 in the pedigree.
   d How many genotypes are possible for III3? List them.

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Figure 2.24 A dividing cell

Figure 2.25 A family pedigree for cystic fibrosis
EVALUATING

20 Mendel’s work on garden peas was advanced for the era in which he lived. Discuss what made Mendel’s investigations so successful.
21 ‘Genetic testing results will correctly predict if a person develops a disorder. They can usually foretell how severe the symptoms will be and when they will appear.’ Evaluate this statement.

CREATING

22 Choose 10 words from the glossary terms and create a concept map to show how the concepts are connected.
23 Imagine you are trying to explain the difference between chromosomes, genes and DNA to a brother or sister two years younger than you. Write down your explanation in such a way that they could understand it.

REFLECTING

24 Record your personal opinions about whether genetic testing should be widely available.