

2 Getting into genes

Can you roll your tongue? Did you know that your genes determine whether you can? Do you fit into your genes or do they fit into you? The characteristics of living things are determined by both the genetic information that they contain and the environment in which they live. New technologies have harnessed genetic machinery in order to change or create new organisms. What are the implications of manipulating the raw material of life?

Think about genes

- Do you have a Darwin's point?
- Why don't hairs grow on stomach linings?
- What have monks, peas, mathematics and genes got in common?
- What do Xs and Ys have to do with sex?
- Designer babies — should we or shouldn't we?
- Is the junk in your DNA actually a treasure?

In this chapter:

2.1 Patterns, order and organisation:
Nuclear matters 6

2.2 DNA — this is your life! 12

2.3 Unlocking the DNA code 15

2.4 Who do you think you are? 22

2.5 Dividing to multiply 24

2.6 The next generation 30

2.7 What are the chances? 37

2.8 Changing the code 43

2.9 Predicting with pedigree charts 46

2.10 **SCIENCE AS A HUMAN ENDEAVOUR**
Exposing your genes 51

2.11 Domesticating biotechnology 57

2.12 **SCIENCE AS A HUMAN ENDEAVOUR**
DNA — interwoven stories? 63

2.13 Thinking tools: SWOT analyses and priority grids

eBookplus
ONLINE ONLY

Study checklist/Digital resources 68

Looking back 69

ICT ACTIVITY **projectsplus**

The gene lab 73

YOUR QUEST

Genes

THINK, SHARE AND DISCUSS

In your team, look at the pictures of the individuals in the families shown below and share your observations.

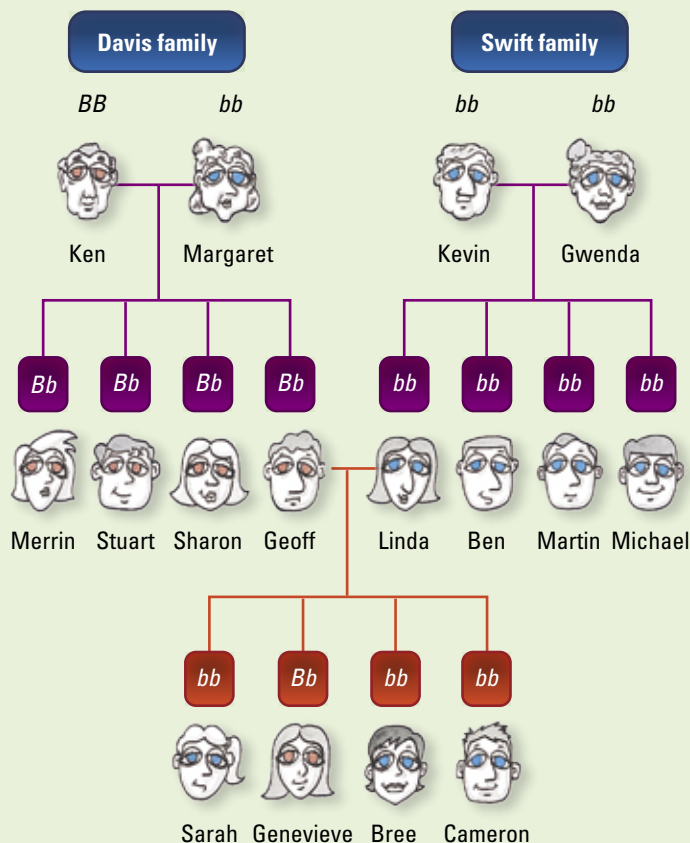
- Record any patterns that you notice.
- If the following couples had another child, suggest what their eye colour may be and give a reason for your suggestion.
 - Ken and Margaret Davis
 - Kevin and Gwenda Swift
 - Geoff and Linda Davis
- Suggest a reason why Geoff (brown eyes) and Linda (blue eyes) had brown-eyed and blue-eyed children, whereas Ken (brown eyes) and Margaret (blue eyes) had only children with brown eyes.
- Martin Swift's fiancée, Justine, has blue eyes, but both of her parents have brown eyes. If Justine and Martin have children together, what colour eyes do you think may be possible? Discuss reasons for your response.



- Bring to school a collection of photographs from as many members of your family as you can.
 - For at least one member of your team, carefully observe each photograph of the family members, looking for similarities.
 - Construct a table with the family features that you have observed and indicate which family members have these features.
 - Can you see any patterns or make any interesting suggestions on these observations? If so, discuss and record these.
 - Discuss which characteristics may be passed from parent to child and which may not.
 - Make a summary of your discussion to share with other teams. Add any other interesting points from these discussions to your team summary.

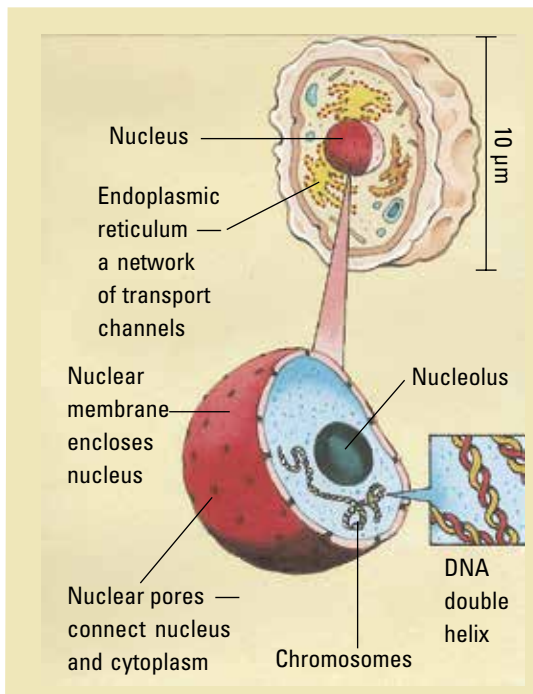
THINK AND DISCUSS

- Read and think about each of the following statements, then state whether you agree, disagree or don't know. Discuss your decisions with your team.
 - Because June and Frank have five sons, the chance of their next child being a daughter is increased.
 - People who have committed very violent crimes should be sterilised because their children will also be violent.
 - Parents should be allowed access to technologies that enable them to select the gender and specific characteristics of their children.
 - Technologies that alter the gametes (sperm and ova) should be illegal.



Patterns, order and organisation: Nuclear matters

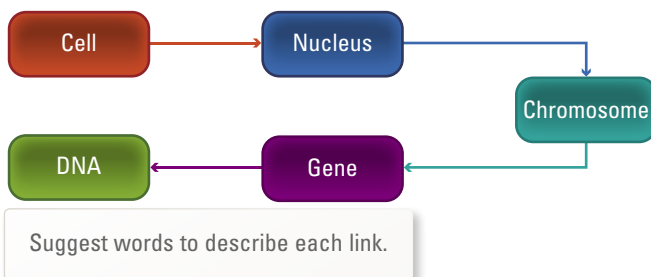
Where did you get those pointed ears, big nose and long toes from? Features or traits that are inherited are passed from one generation to the next in the form of a genetic code. This code is written in a molecule called **deoxyribonucleic acid (DNA)** and is located within the nucleus of your cells.



An animal cell

DNA: past, present and future

Most of your body cells contain all of the DNA instructions needed to make another you. Your DNA, however, is more than just a genetic blueprint of instructions; it is also an 'ID tag' and a very special ancient 'book' that holds secrets both from your ancestral past and for your possible futures.



Genes

Each genetic instruction that codes for a particular trait (for example, shape of ear lobe, blood group or eye colour) is called a **gene**. Genes are made up of DNA and are organised into larger structures called **chromosomes**, which are located within the **nucleus** of the cell.



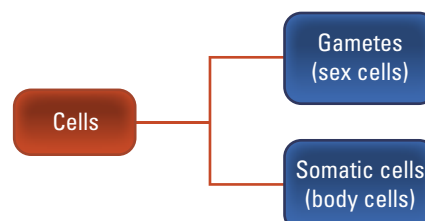
Scanning electron micrograph showing double-stranded chromosomes

Chromosomes

Your body is constantly making new cells for replacement, growth and repair. It achieves this by a process called **mitosis**, which is a type of **cell division**.

Prior to cell division your DNA replicates itself, and this long molecule (2–3 metres) bunches itself up into 46 little packages called chromosomes. They are called chromosomes (*chromo* = 'coloured' + *soma* = 'body') because scientists often stain them with various dyes so that they are easier to see.

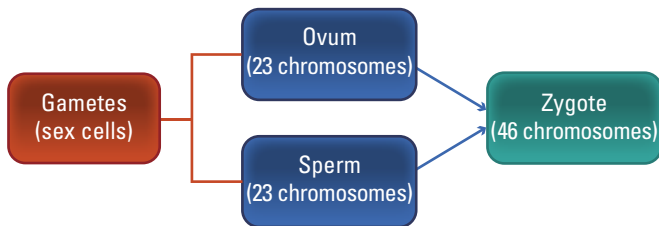
Chromosomes are only visible when a cell is about to divide or is in the process of dividing. When your cells are not dividing, chromosomes are not visible as the coils are unwound and the DNA is spread throughout the nucleus.



Suggest how these terms are linked.

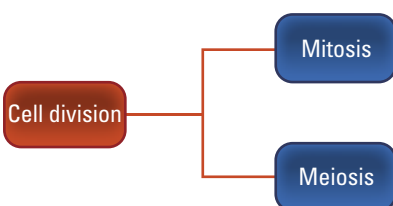
Sex cells

Another type of cell division called **meiosis** is used in the production of sex cells or **gametes**: **ova** (ovum is the singular form) and **sperm**. Meiosis results in the chromosome number being halved, so instead of pairs of chromosomes in each resulting cell, there is only one chromosome from each pair.



Which words could be used to describe each link?

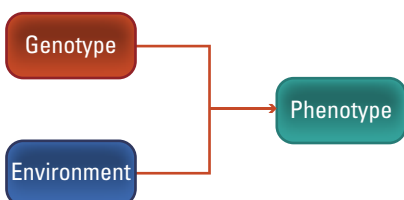
The genetic information that you received from your mother was packaged into 23 chromosomes in the nucleus of her egg cell (ovum), and the genetic information that you received from your father was packaged into 23 chromosomes in the nucleus of the sperm that fertilised your mother's egg cell. When these gametes fused together at **fertilisation**, the resulting **zygote** contained 23 pairs of chromosomes (one pair from each parent) — a total of 46 chromosomes.



Suggest words to describe each link.

Somatic cells

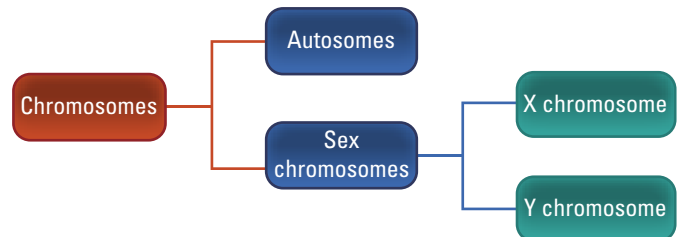
Cells of your body that are not your sex cells are often referred to as body cells or **somatic cells**. With the exception of your red blood cells (that lose their nucleus when mature so they can carry more oxygen), all of your somatic cells contain chromosomes in pairs within their nucleus. This double set of genetic instructions (one set from each parent) makes up your **genotype**. The visible expression of the genotype as a particular trait or feature is called the **phenotype**. The phenotype may also be influenced by your **environment**.



Suggest how these terms are linked.

Chromosomes — more than one type

Chromosomes can be divided into two main types: autosomes and sex chromosomes.



Which words could be used to describe each link?

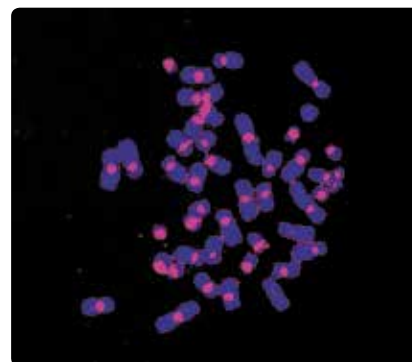
Autosomes

Of the 46 chromosomes in your somatic cells, 44 are present in both males and females and can be matched into 22 pairs on the basis of their relative size, position of centromere (refer to the following figure) and stained banding patterns. These are called **autosomes**. They are numbered from 1 to 22 on the basis of their size: chromosome 1 being the largest of the autosomes and chromosome 22 the smallest.

The members of each matching pair of chromosomes are described as being **homologous**. Those that don't match are called **non-homologous**. For example, two number 21 chromosomes would be referred to as homologous, and a number 21 chromosome and a number 11 chromosome would be non-homologous.



Homologous chromosomes have the same relative size, position of centromere and stained banding patterns.



Fluorescent-dyed chromosomes showing stained centromere

Sex chromosomes

The two remaining chromosomes are the **sex chromosomes**. In humans, these differ between males and females. Females possess a pair of X chromosomes (XX) and males have an X chromosome and a Y chromosome (XY). The sex chromosomes are important in determining an individual's gender (whether they are male or a female).

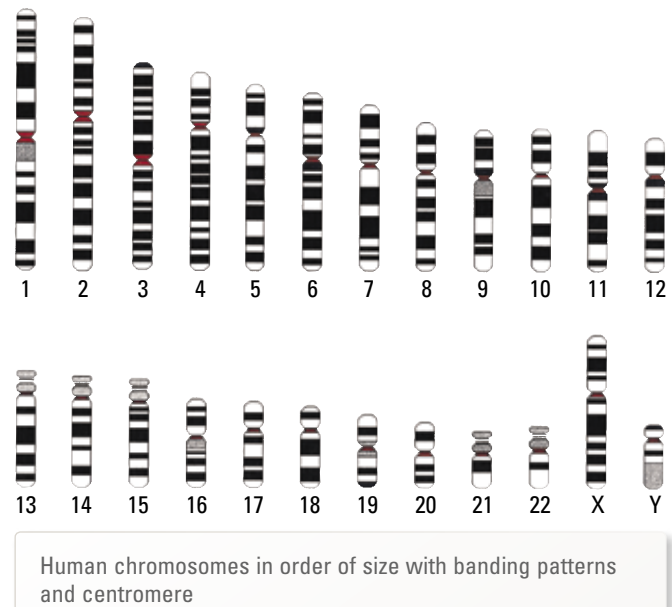
Too many or too few

Sometimes a genetic mistake or mutation can occur that results in more or less of a particular type of chromosome. Down syndrome is an example of a **trisomy** mutation in which there are three number 21 chromosomes instead of two. Turner syndrome is an example of a **monosomy** mutation that results in only one sex chromosome (XO).

Your kind of karyotype

Differences between the chromosome pair size, shape and banding can be used to distinguish them from each other. Scientists use these differences to construct a **karyotype**. Cells about to divide are treated and stained, mounted on slides for viewing, and photographed. These photographs are cut up and rearranged into pictures that show the chromosomes in matching pairs in order of size from largest to smallest. Karyotyping can reveal a variety of chromosomal disorders such as Down syndrome and Turner syndrome.

The gender of an individual can also be determined using karyotyping. In humans, females possess



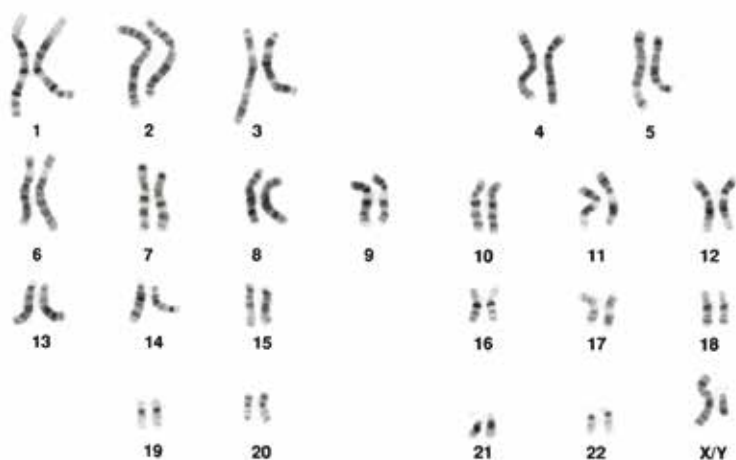
two similar-sized X sex chromosomes. In males, however, their sex chromosomes are not matching — they possess an X chromosome and a smaller Y chromosome.

Has the secret of age reversal been discovered?

In the 1970s, Tasmanian-born scientist Dr Elizabeth Blackburn made a discovery that contributed to our understanding of how cells age and die. She showed how the presence of a cap of DNA called a **telomere**

Some examples of chromosome changes and approximate incidence rates. Which syndrome is an example of a trisomy? A monosomy?

Chromosome change	Resulting syndrome	Approximate incidence rate
<i>Addition: whole chromosome</i>		
Extra number 21 (47, +21)	Down syndrome	1/700 live births
Extra number 18 (47, +18)	Edwards syndrome	1/3000 live births
Extra number 13 (47, +13)	Patau syndrome	1/5000 live births
Extra sex chromosome (47, XXY)	Klinefelter syndrome	1/1000 male births
Extra Y chromosome (47, XYY)	N/A	1/1000 male births
<i>Deletion: whole chromosome</i>		
Missing sex chromosome (46, XO)	Turner syndrome	1/5000 female births
<i>Deletion: part chromosome</i>		
Missing part of number 4	Wolf–Hirschhorn syndrome	1/50 000 live births
Missing part of number 5	Cri-du-chat syndrome	1/10 000 live births



A human karyotype



Dr Elizabeth Blackburn

on the tip of the chromosome enabled DNA to be replicated safely without losing valuable information. Each time the cell divides, however, these telomeres shorten. When the telomeres drop below a certain length, the cell stops dividing and dies. This is a normal part of ageing. Blackburn and her colleagues later discovered an enzyme, **telomerase**, that was involved in maintaining and repairing telomeres. In 2009, Blackburn and her colleagues were awarded the Nobel Prize in Physiology and Medicine for their work on how chromosomes are protected by telomeres and the enzyme telomerase.

Other scientists are now involved in finding out more about the exciting possibilities that our understanding of this process may open up. In 2010, for example, Mariela Jaskelioff and her colleagues in America genetically engineered mice with short telomeres and inactive telomerase to see what would happen when they turned this enzyme back on. Their results showed that after four weeks new brain cells were developing and tissue in several organs had regenerated — and the mice were living longer. If this happens in mice, what might future research suggest for humans?

INVESTIGATION 2.1

Extracting DNA

AIM To extract DNA from ground wheatgerm

Materials:

1 teaspoon of finely ground wheatgerm
 14 mL of isopropyl alcohol (or equivalent)
 1 mL of liquid detergent
 20 mL of hot tap water (50–60 °C)
 test tube
 measuring cylinders
 rubber stopper
 test-tube rack
 Pasteur pipette and bulb
 glass stirring rod

METHOD AND RESULTS

- 1 Draw a table in your book, allowing room for observations in the form of a diagram: immediately after adding the alcohol; at 3- and 15-minute intervals; and after you have collected and removed the DNA.

- ▶ Add the wheatgerm and hot water to a test tube. Twist the stopper in and shake for 3 minutes.
- ▶ Add 1 mL of detergent and mix gently with the glass rod for about 5 minutes. Do not create foam.
- ▶ If you do create foam, suck it out with the Pasteur pipette.
- ▶ Tilt the tube at an angle and slowly pour in the alcohol so that it sits at the bottom.

CAUTION Do not mix!

- 2 Note your observations in your table.
- 3 Fill in your observations after 3 minutes and again after 15 minutes.
- 4 Collect the DNA with the glass rod. Feel it with your fingers and make your final observations.

DISCUSS

- 5 What colour did you expect DNA to be? Why do you think it was the colour that you observed?
- 6 How could you confirm that it really was DNA?
- 7 Suggest improvements to the experimental design.

UNDERSTANDING AND INQUIRING

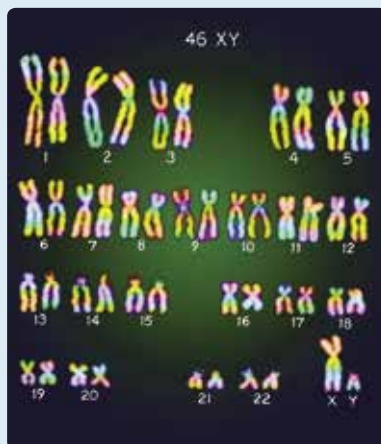
REMEMBER

- State the name of the:
 - molecule that DNA is an abbreviation for
 - location of DNA in a human cell
 - structures that genes are organised into
 - type of cell division used to produce gametes
 - male sex gamete
 - female sex gamete
 - process in which sex cells fuse together
 - cells of your body that are not sex cells
 - double set of genetic instructions
 - particular trait or feature that results from your genotype and environment
 - chromosomes that are not sex chromosomes
 - protective cap of DNA on the tip of chromosomes.
- Suggest why chromosomes are stained with dyes.
- Are chromosomes always visible in a cell? Explain.
- State how many chromosomes you would expect to find in a human:
 - somatic cell
 - gamete.
- Distinguish between the following pairs of terms.
 - Ovum and sperm
 - X chromosome and Y chromosome
 - Sex chromosomes and autosomes
 - Somatic cells and sex cells
 - Mitosis and meiosis
 - Homologous and non-homologous
 - Gene and DNA

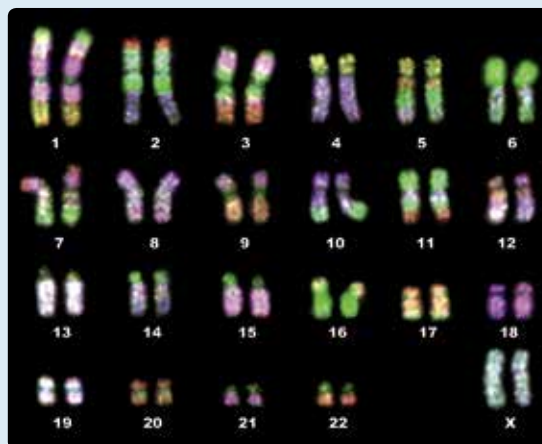
ANALYSE, INTERPRET AND DISCUSS

- The following questions refer to figures A and B below.
 - Carefully observe the figures and suggest features that are useful in pairing the chromosomes.
 - On the basis of information in the karyotype, suggest the gender of A and B. Justify your responses.
 - Suggest why karyotyping can be carried out only on cells that are about to divide.

A



B



- Each species has a particular number of chromosomes. The table below shows some examples of the number of chromosomes in the body cells of some organisms.
 - Using the data in the table, construct a column graph.
 - Identify the species with the:
 - highest total number of chromosomes
 - lowest total number of chromosomes.
 - Carefully observe your graph, looking for any patterns. Discuss possible reasons for these.
 - Do you think that the number of chromosomes reflects the intelligence of an organism? Provide reasons for your response.
 - Suggest the number of chromosomes in the sex cells of a:
 - housefly
 - sheep.

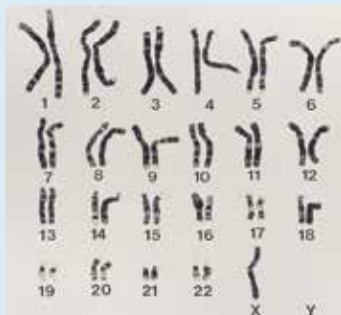
Number of chromosomes in body cells (non-sex cells) of some living things

Species of living thing	Number of chromosomes in each body cell	Species of living thing	Number of chromosomes in each body cell
Chimpanzee	48	Tomato	24
Euglena (unicellular organism)	90	Cabbage	18
Fruit fly	8	Frog	26
Human	46	Housefly	12
Koala	16	Pig	40
Onion	16	Platypus	52
Shrimp	254	Rice	24
Sugarcane	80	Sheep	54

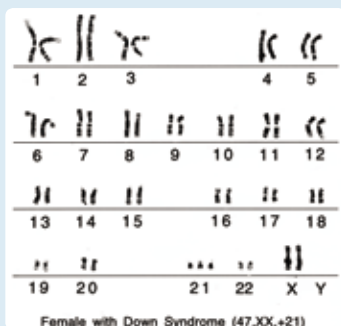
8 Carefully observe the karyotypes A and B below.

- Suggest the gender of the individual in A and in B. Justify your responses.
- Use your observations and the chromosome change table in this section to:
 - suggest which type of chromosome change is shown in each figure
 - suggest the name of the resulting genetic disorders.
- One of these disorders is also sometimes described as trisomy 21. Suggest a reason for this description.

A



B



ANALYSE, INTERPRET AND INVESTIGATE

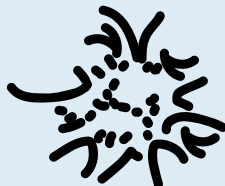
9 Observe the figures below that show the chromosomes belonging to four different types of organisms.



Kangaroo (6 pairs)



Human (23 pairs)

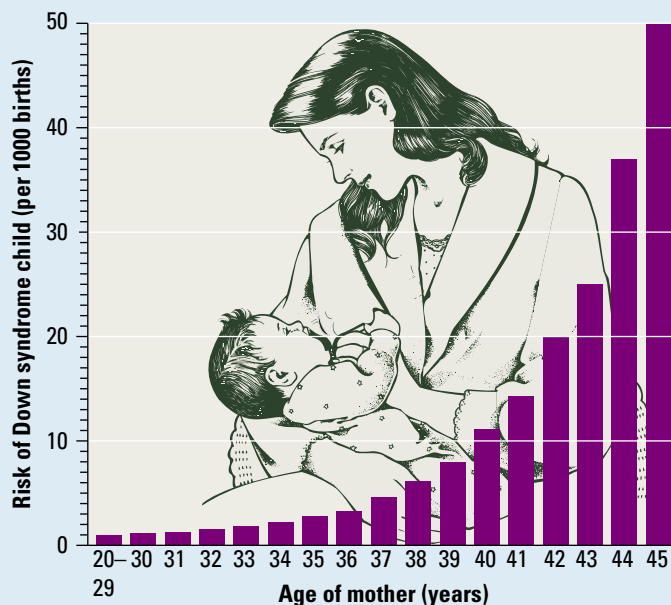


Domestic fowl (18 pairs)



Fruit fly (4 pairs)

- Suggest whether the chromosomes are from somatic cells or sex cells. Justify your response.
- Suggest which organisms possess chromosomes:
 - most like humans
 - least like humans.
 Justify your responses.
- Do any observations in (b) surprise you? Why?



10 The graph above shows the relationship between Down syndrome and maternal age.

- Observe the graph and describe any patterns.
- Suggest a hypothesis about Down syndrome and maternal age.
- Research and report on types, causes and symptoms of Down syndrome.

INVESTIGATE, DISCUSS AND REPORT

- Research and report on Elizabeth Blackburn's:
 - contribution to our understanding of DNA
 - stance on stem cell science that resulted in her losing her position on the President's Council on Bioethics.
 - What is bioethics?
 - What is the Presidential Commission for the Study of Bioethical Issues, and what does it have to do with science? How does it differ from the President's Council on Bioethics?
 - Use the **Bioethics** weblink in your eBookPLUS to find out more about the types of issues that have been considered by the Commission.

eBookplus

12 If each cell nucleus has about a metre of DNA, how does it all fit in? Use the internet to find animations that show how DNA is organised so that it can fit into cells.

CREATE

- Use the **Karyotype** weblink in your eBookPLUS. Follow the instructions provided to prepare and interpret karyotypes for some patients.

eBookplus

work
sheet

2.1 Genes and chromosomes

DNA — this is your life!

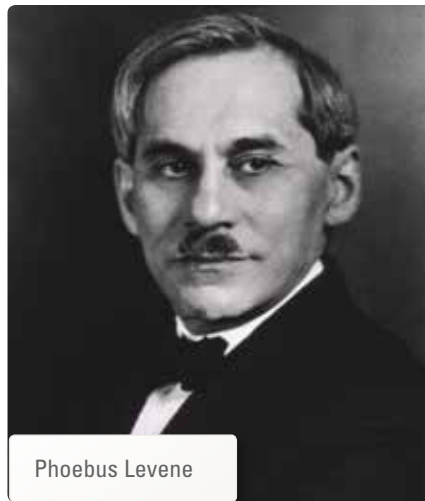
Even though DNA is as old as life itself, we have only recently been introduced to it. Like the story of the theory of evolution, DNA has its own story: a story of passion, imagination and determination that has involved the use of new technologies and the development of many more.

Greetings, DNA!

The abbreviation **DNA** is so well known, that it is often used as a word itself. DNA is the abbreviation for **deoxyribonucleic acid**. As the name suggests, it is a type of nucleic acid. It was not until 1869 that we were formally introduced to DNA, when it was discovered by Friedrich Miescher. Working in a laboratory located within a castle in Germany, Miescher — a young Swiss postgraduate student — isolated DNA from the nuclei of white blood cells from pus on bandages. Miescher named the compound he isolated from these cell nuclei 'nuclein'.



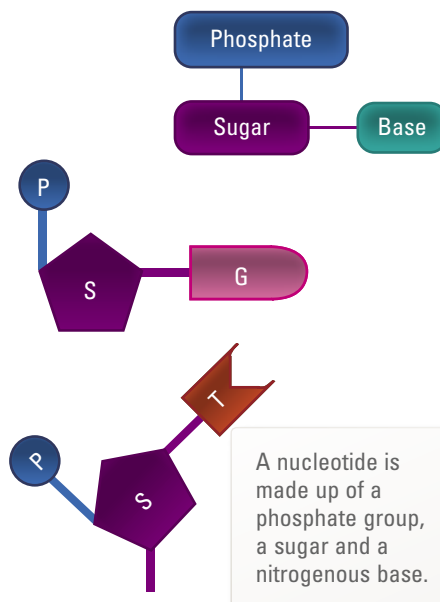
Friedrich Miescher



Phoebus Levene

In parts of three

In 1929, over 50 years after its discovery, Phoebus Levene showed that DNA was made up of repeating units called **nucleotides**. Each of these nucleotides consisted of a sugar, a phosphate group and a nitrogenous base. In the nucleotides that make up DNA, the sugar is **deoxyribose** and the nitrogenous base in each nucleotide is one of four different types: **adenine** (A), **thymine** (T), **guanine** (G) or **cytosine** (C).



Levene suggested that the nucleotides could be joined together to form chains. Although his theory was correct in terms of the chain formation, it was incorrect in other aspects of its structure. His tetranucleotide model, which proposed that DNA had equal amounts of all four nitrogenous bases, contributed to scientists of the time favouring proteins, rather than DNA, as the carrier of genetic information.

DNA carries messages from one generation to next

The experiments of Alfred Hershey and Martha Chase in 1953 supported those of Oswald Avery in 1943, suggesting that DNA was the molecule through which genetic information was carried between generations, not proteins.



Oswald Avery

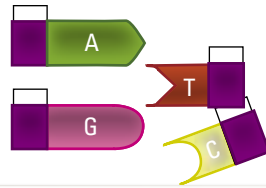


Erwin Chargaff

A with T and G with C

In 1950, Erwin Chargaff contributed to our understanding of the structure of DNA by his careful and thorough analysis of the four different types of nucleotides and their ratios in DNA. His research led to the concept of **base pairing**. This concept states that in DNA every adenine (A) binds to a

thymine (T), and every cytosine (C) binds to a guanine (G). This is now known as **Chargaff's rule**.



Examples of how base pairing using Chargaff's rule can be shown

A key piece of the puzzle

The next piece of the puzzle to solve the structure of DNA was contributed (some say without her knowledge) by Rosalind Franklin. Rosalind Franklin and Maurice Wilkins had decided to crystallise DNA so that they could make an X-ray pattern of it. They were specialised in making X-ray diffraction images of biological molecules so that they could be

analysed to find out information about their three-dimensional structures. Franklin's X-ray diffraction picture of a DNA molecule provided important clues about the shape of the molecule.

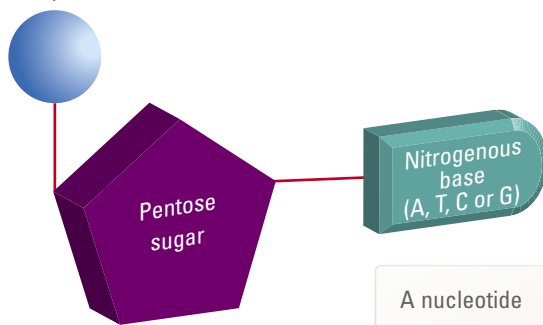


Rosalind Franklin provided a key clue to solve the structure of DNA.

Double helix

James Watson and Francis Crick were building a DNA model to try to solve its structure. They were shown Franklin's X-ray diffraction image of DNA, which strongly suggested that DNA had a helical shape. They used this information, as well as that from Chargaff and other researchers (such as their American colleague Linus Pauling), to successfully solve the structure of DNA. At last the structure was identified!

Phosphate



A nucleotide

eBookplus

eLesson

Rosalind Franklin and Watson and Crick

Watch a video from *The story of science* about the development of the DNA model.

eles-1782

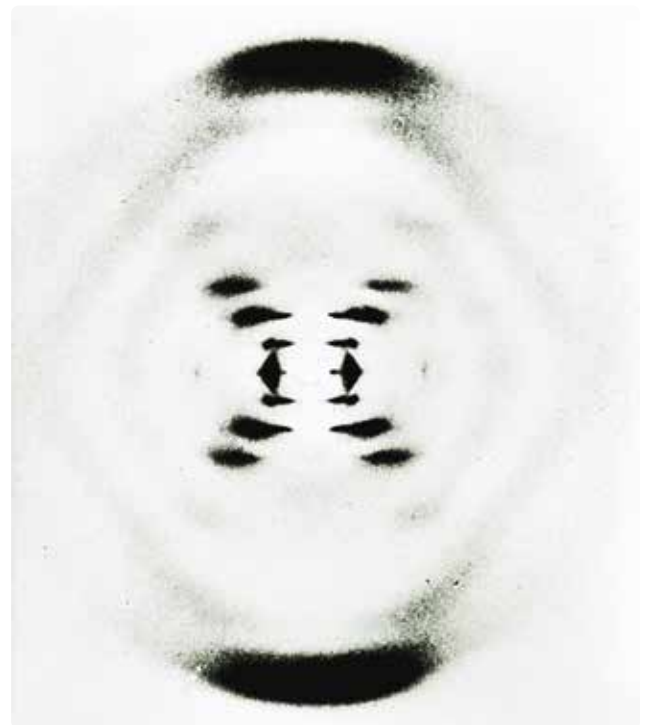
eBookplus

Interactivity

Complementary DNA

Construct a replicate DNA strand by dragging the correct complementary base into sequence.

int-0133



Rosalind Franklin's X-ray diffraction picture provided important clues about the shape of the DNA molecule.

UNDERSTANDING AND INQUIRING

REMEMBER

- 1 State what DNA is an abbreviation for.
- 2 Provide an example of a nucleic acid.
- 3 Identify the year and name of the scientist who first discovered DNA.
- 4 State the source of cells in which DNA was first isolated.
- 5 Use a diagram to show how the three sub-units that make up DNA are organised.
- 6 Outline what the research of Hershey, Chase and Avery suggested.
- 7 Describe what is meant by Chargaff's rule.
- 8 Describe Rosalind Franklin's contribution to the discovery of the structure of DNA.
- 9 Explain how Watson and Crick used information available to determine the structure of DNA.

THINK AND CREATE

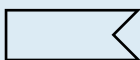
- 10 Use your own materials to construct a model of the double helix structure of DNA.
- 11 Use the information in this section and other sources to construct a timeline on the development of our understanding of DNA.
- 12 Construct a paper model of DNA. Some suggested shapes you could use to represent the parts that make up a DNA molecule are shown below.



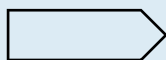
Sugar



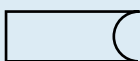
Phosphate group



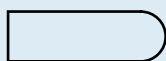
Thymine



Adenine



Guanine



Cytosine

- 13 Evaluate the model you made in question 12. Which aspects of the structure of DNA does your model show accurately? In what ways is your model different from an actual DNA molecule?

INVESTIGATE

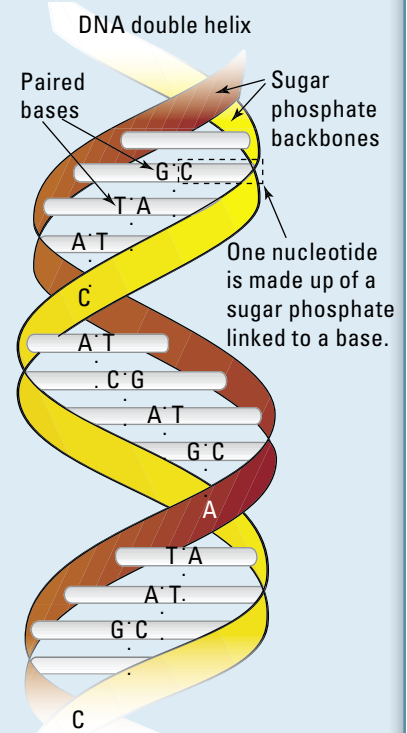
- 14 Select one of the scientists discussed in this section, research them and write a biography about their life and scientific contributions.
- 15 Find out more about DNA and how knowledge about its structure is being used in research and other applications. Present your findings as a documentary, animation or in a multimedia format.

- 16 Investigate more about the history of how we have obtained our genetic knowledge. Present your findings as a timeline.
- 17 Investigate the effect that our increased knowledge about the structure and function of DNA has had on:
 - (a) our species
 - (b) other species
 - (c) our planet.
- 18 Investigate and report on the development of the Watson and Crick double helix model of the structure of DNA.
- 19 Use internet research to help you to identify three questions that could be investigated about DNA. Collate these questions as a class, and then select one to investigate and report on.

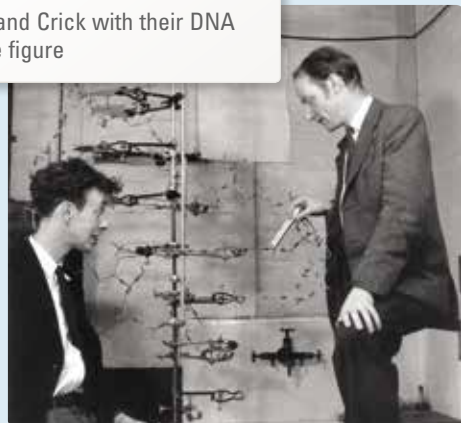
eBookplus

- 20 Discuss the following statement:
'Had Maurice Wilkins and Rosalind Franklin had a more harmonious working relationship it is likely that Franklin would have been involved in writing the scientific paper where the structure of DNA was first described, and that she would have been given the same credit for discovering the structure of DNA as Watson and Crick.'

Use the **Rosalind Franklin** weblinks in your eBookPLUS to research this question further.



Watson and Crick with their DNA molecule figure

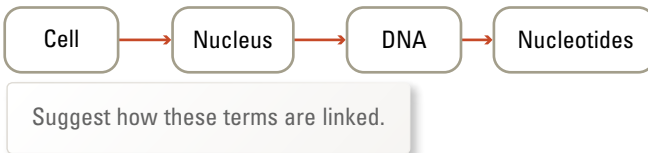


Unlocking the DNA code

Did you know that all living things share the same genetic letters? This universal genetic language provides strong evidence that all life on Earth evolved from one ancient cell line.

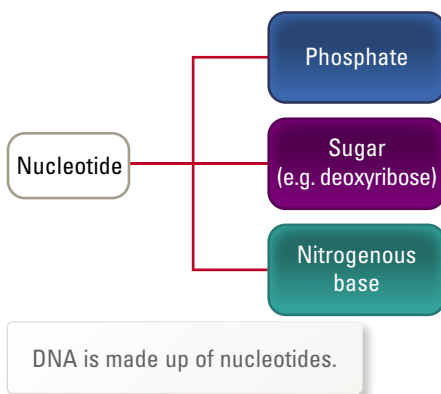
Key codes

Like other eukaryotic organisms, DNA is located within the nucleus and mitochondria of your cells. While both types of DNA (nuclear and mitochondrial) share a number of features in common, they also differ. These differences will be considered later. In this section, we will focus on your nuclear, rather than mitochondrial, DNA.

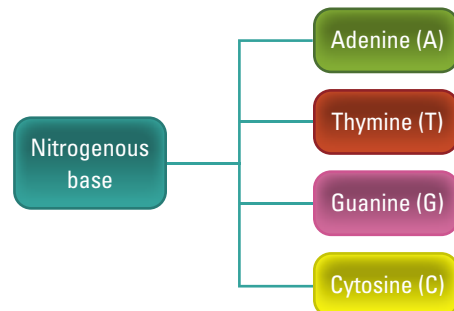
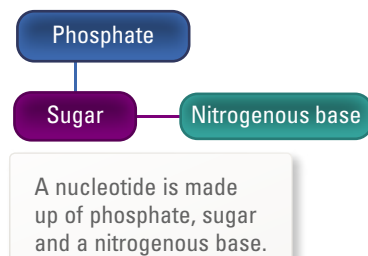


Stepping down the DNA ladder

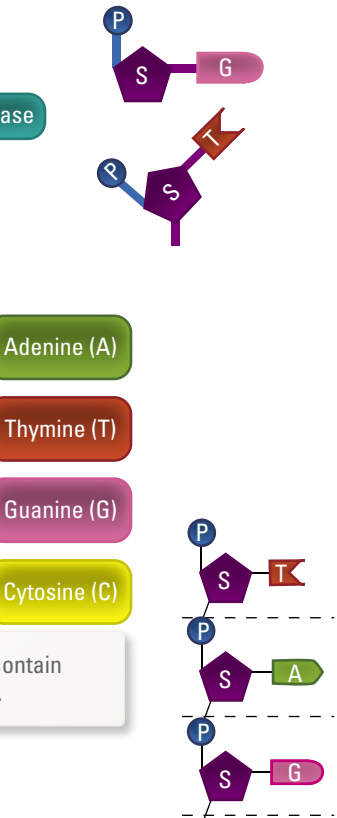
Like other **nucleic acids**, DNA molecules are made up of building blocks called nucleotides. Each nucleotide is made up of three parts: a sugar part, a phosphate part and a **nitrogenous base**. The figures below and above right show some ways in which the components of nucleotides may be drawn.



While the sugar (for example, deoxyribose) and phosphate are the same for each nucleotide in DNA, the nitrogenous base may vary. The four possible nucleotides in DNA are adenine (A), thymine (T), cytosine (C) and guanine (G).



The nucleotides are joined together in a chain. The sugar and phosphate parts make up the outside frame and the nitrogenous bases are joined to the sugar parts.



Nitrogenous bases are attached to the sugar part of the nucleotide.

Nitrogenous bases in pairs

A DNA molecule is made up of two chains of nucleotides. Hydrogen bonds join them at their complementary (or matching) nitrogenous base pairs. Adenine binds to thymine and cytosine to guanine. This matching of the nitrogenous bases is often referred to as the **base-pairing rule**.

For example, a segment of DNA that has one strand with the code GATTACA would have a complementary strand of CTAATGT. In its double-stranded view it looks like this:

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

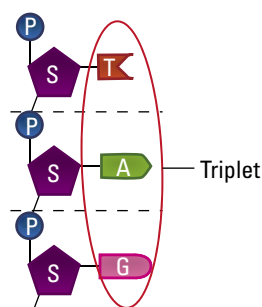
DNA molecules have the appearance of a **double helix** or spiral ladder. Using the spiral ladder metaphor, DNA could be considered as having a sugar-phosphate backbone or frame, and rungs or steps that are made

up of **complementary base pairs** of nitrogenous bases joined together by hydrogen bonds.

Unlocking DNA codes

The sequence of nucleotides in DNA is often described in terms of the nitrogenous bases that they contain. For example, if the first nucleotide contains guanine, the second contains adenine and the third thymine, then this sequence would be described as GAT. This sequence of three nucleotides in DNA is referred to as a **triplet**. Although some of these DNA triplets code for a start (e.g. TAC) or stop (e.g. ATT, ATC or ACT) instruction, most code for a particular **amino acid**. The triplet GAT, for example, codes for the amino acid aspartine.

The sequence of these triplets in DNA contains the genetic information to make your body's proteins. This includes all of your hormones, enzymes, antibodies and many other proteins that are essential for your survival. If one of these triplets (or its bases) is incorrect or missing, it may result in a protein not being coded for or produced — which could result in death.

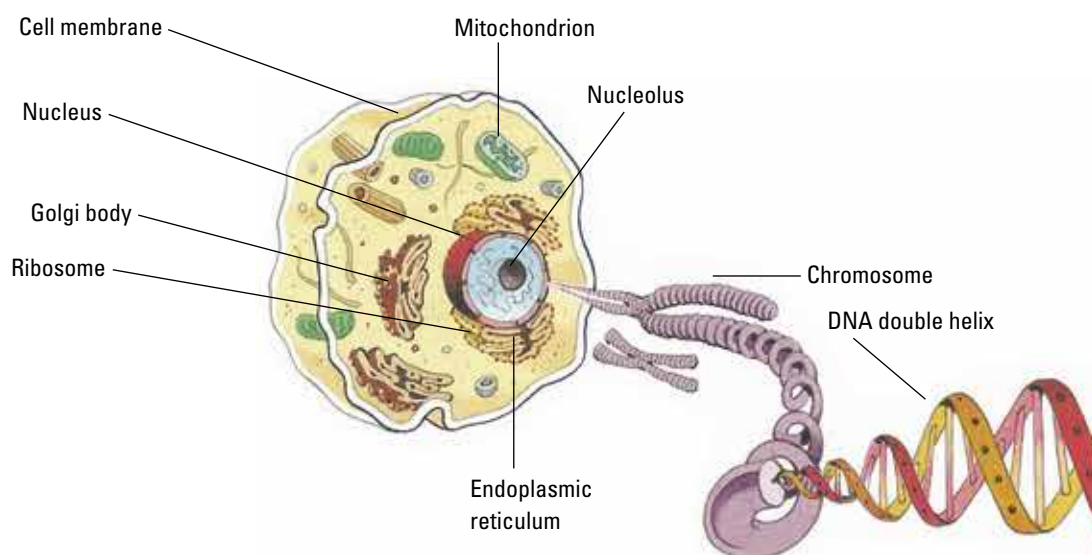


The DNA code is read three bases at a time.

Introducing RNA

Like DNA, RNA is a type of nucleic acid and is made up of nucleotides. Its nucleotides, however, are different from those of DNA. RNA contains the sugar **ribose** (instead of deoxyribose), and **uracil** (instead of thymine) is one of its nitrogenous bases. It is also shorter and single-stranded.

Another difference is that the triplet code in mRNA is referred to as a **codon**. The complementary mRNA codon for the start triplet TAC in DNA, for example, would be AUG.

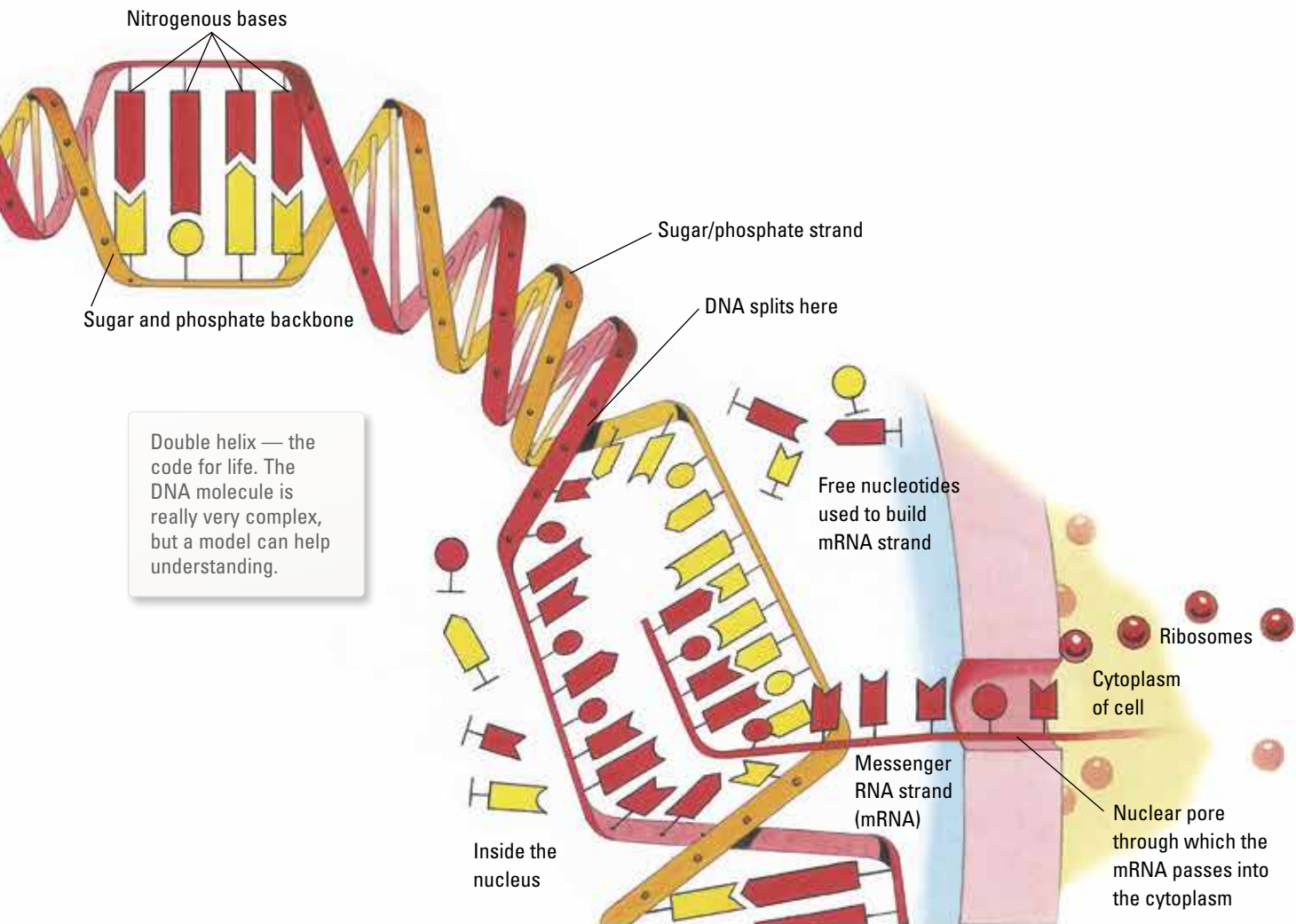
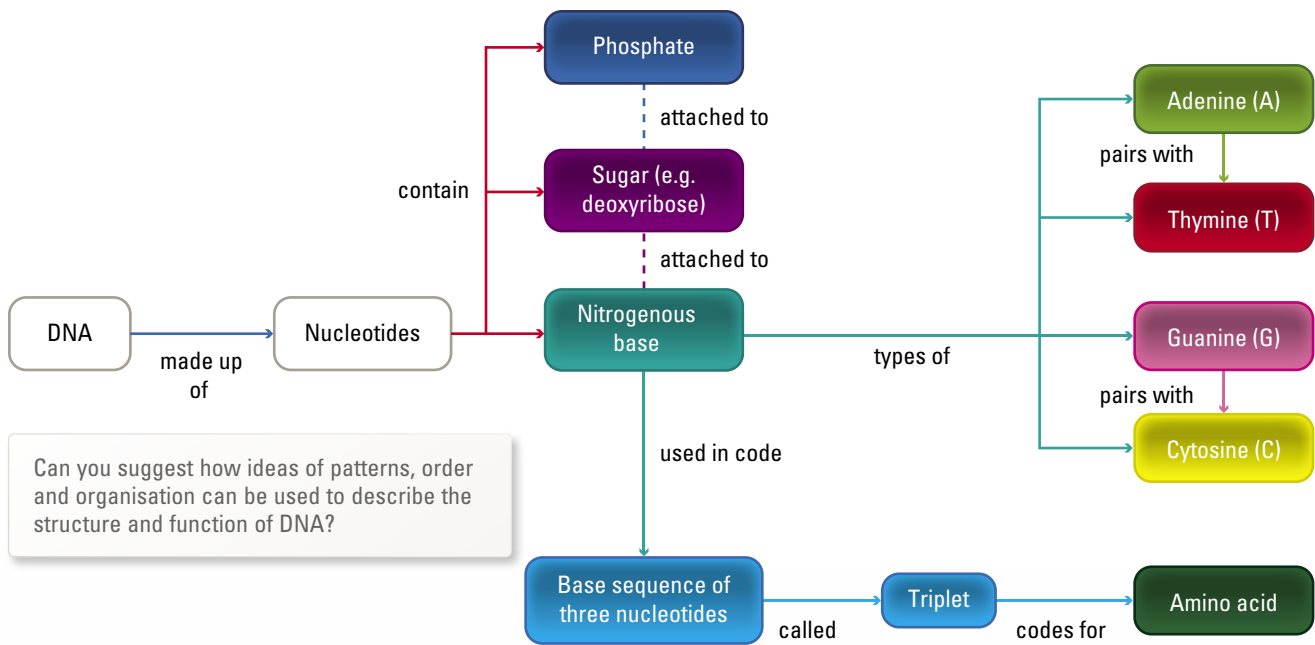


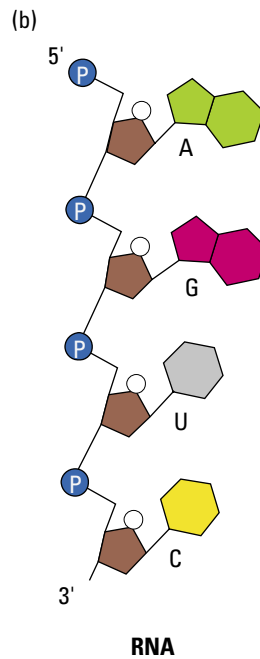
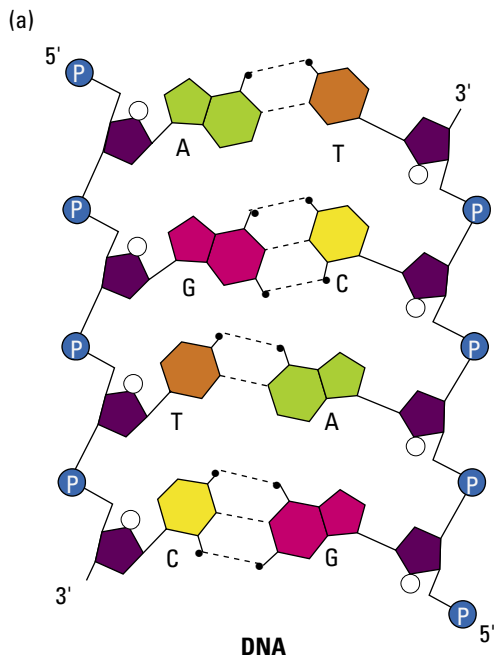
Protein synthesis: reading the code

DNA is in your genes, and it tells your cells how to make proteins. The instructions for making proteins are coded for in the sequence of nitrogenous bases in DNA. Within the nucleus, these instructions are transcribed into another type of nucleic acid called RNA in a process called transcription. This RNA copy then moves to a ribosome in the cytoplasm where the genetic message is translated into a protein.



The DNA message is transcribed into a mRNA message that is translated into a protein.



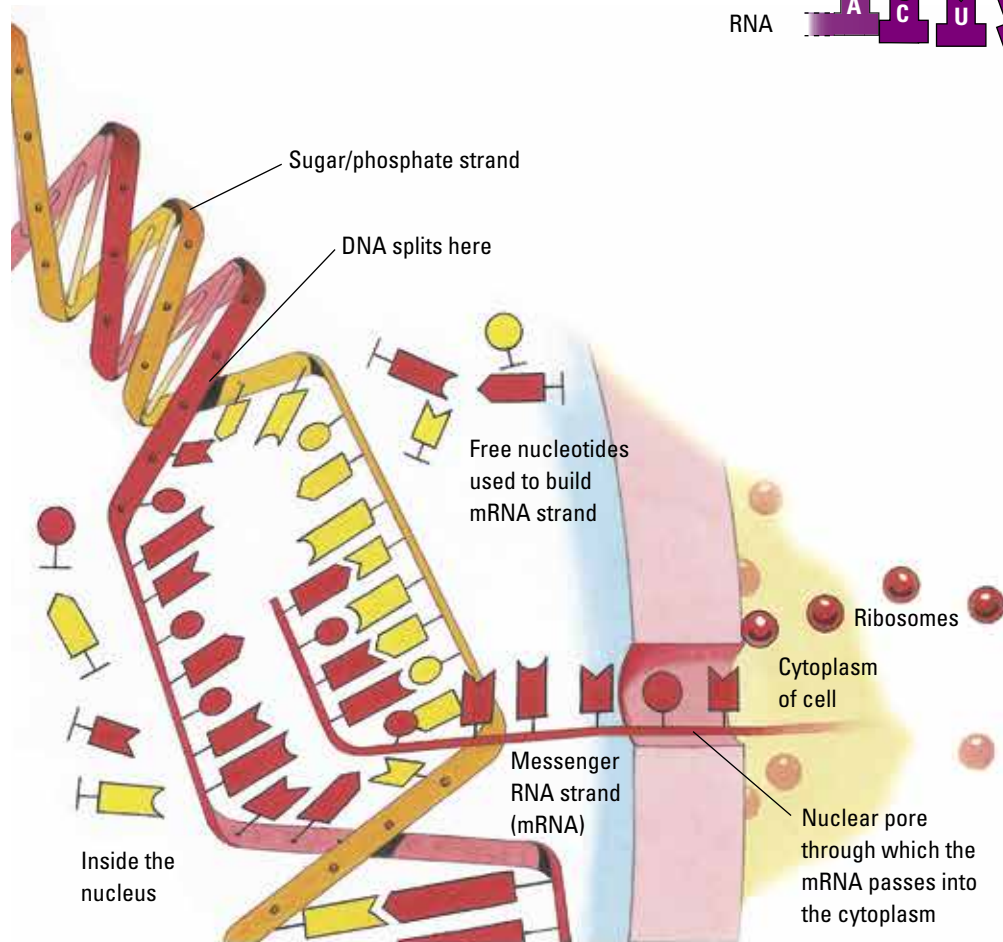


How many differences can you identify between DNA and RNA?

Transcription

The first step in making a protein involves the unzipping of the gene's DNA. When the relevant part of the DNA strand is exposed, a special copy of the sequence is produced in the form of **messenger RNA (mRNA)**. The process of making this complementary mRNA copy of the DNA message is called **transcription**.

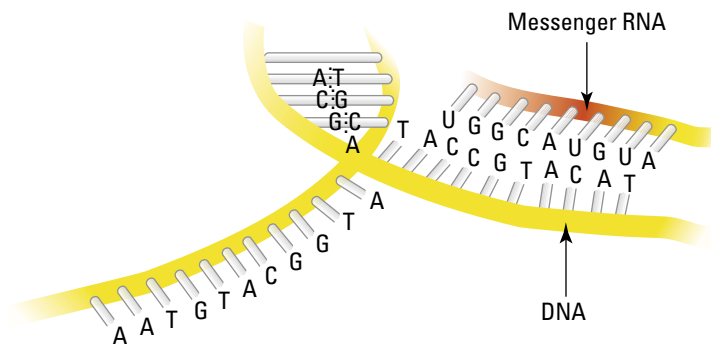
As its name suggests, messenger RNA (mRNA) passes through the pores of the nuclear membrane into the cytoplasm to take its genetic copy of the protein instruction message to **ribosomes**. These may be free floating in the cytosol or attached to the rough endoplasmic reticulum.



The complementary mRNA that had been transcribed from a segment of DNA such as TACATGCCA would be AUGUACGGU.



A section of the DNA unzips so that the mRNA copy can be made.



During transcription, an RNA molecule is formed with bases complementary to the DNA's base sequence.

Translation

Ipsa scientia potestas est. Unless you speak Latin, you will need some help to translate this sentence! Once it is translated, you can then do something with it. This is similar to the meaning of the sentence: Knowledge itself is power.

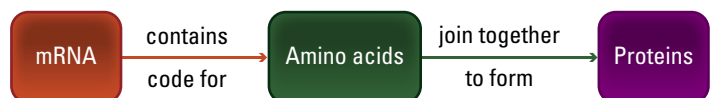
Once the mRNA has reached the ribosome, its message needs to be **translated** into a protein. The ribosome and another type of molecule called **transfer RNA (tRNA)** are involved in this process. tRNA already located in the surrounding cytosol collects and transfers the appropriate amino acid to its matching code on the mRNA. These amino acids are joined together by peptide bonds to make a protein.

DNA triplet and corresponding mRNA codon and amino acid

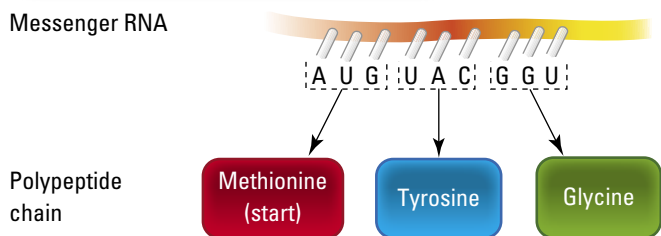
DNA triplet	mRNA codon	Amino acid
AAT	UUA	Leucine (leu)
ACG	UGC	Cysteine (cys)
TAC	AUG	Start/methionine (met)
ATT	UAA	Stop
CGG	GCC	Alanine (ala)
CAT	GUA	Valine (val)
ATG	UAC	Tyrosine (tyr)
CCA	GGU	Glycine (gly)

Precious proteins

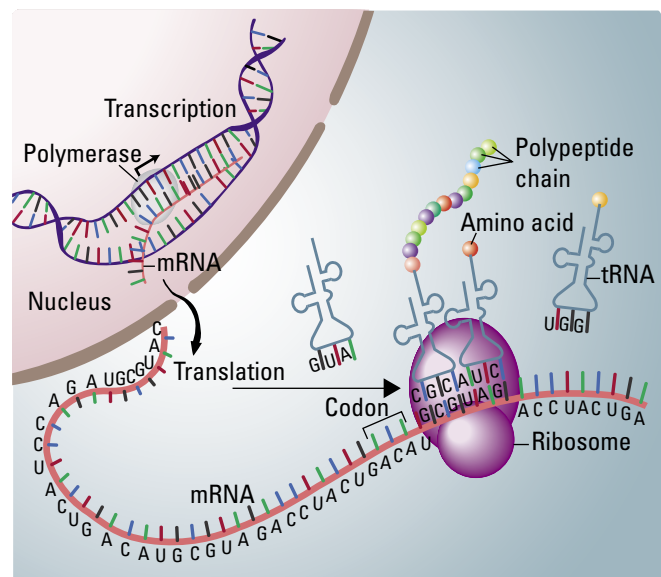
Why are proteins so important? Proteins form parts of cells, regulate many cell activities and even help defend against disease. Your heart muscle tissue contains special proteins that can contract, enabling blood containing haemoglobin and hormones to be pumped through your body. Haemoglobin is a protein that carries oxygen necessary for cellular respiration. Many hormones are proteins. Insulin, glucagon and adrenaline, for example, are hormones that influence activities of your cells. Enzymes are also made up of protein and can be involved in regulating metabolic activities such as those in chemical digestion and respiration. Antibodies are examples of proteins that play a key role in your immune system in its defence against disease.



Proteins are made up of amino acids.



mRNA codons code for particular amino acids.



Plants also rely on proteins for their survival. Their growth and many other essential activities are regulated by hormones (such as auxins) and enzymes.

Switched on or off?

Different genes are responsible for different characteristics, such as the colour of flower petals, the

markings on a snail shell, or a person's blood group or eye colour. Every body cell in an organism has the same set of genes called a genome, but not all genes are active. Some have to be switched on to act and some have to be switched off at different stages in the life of a cell. This is why hairs do not grow on the stomach lining and cheek cells do not grow on toenails.

UNDERSTANDING AND INQUIRING

REMEMBER

- Who am I? State the name(s) of the:
 - building blocks that make up DNA
 - three parts that make up nucleotides
 - four possible types of nitrogenous bases in DNA
 - four possible types of nitrogenous bases in RNA
 - complementary base that pairs with thymine in DNA
 - complementary base that pairs with adenine in RNA
 - sequence of three nucleotides in DNA that code for an amino acid
 - sequence of three nucleotides in mRNA that code for an amino acid
 - two steps in protein synthesis
 - site of protein synthesis
 - set of genes within a cell of an organism.
- Construct a Venn diagram or matrix table to summarise the similarities and differences between:
 - DNA and RNA
 - transcription and translation
 - nucleic acids and amino acids
 - codons and triplets.
- What is meant by the base-pairing rule? Use a diagram in your response.
- Explain the importance of protein synthesis.

ANALYSE, INTERPRET AND THINK

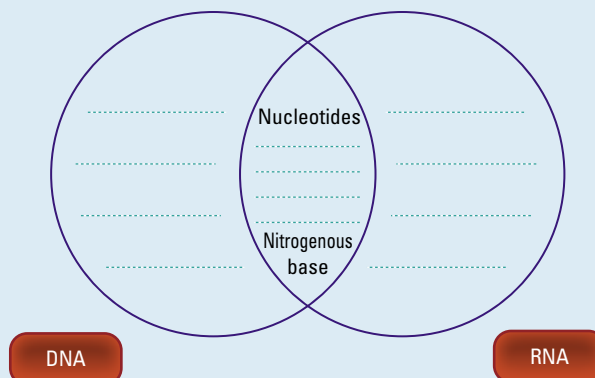
- Construct flowcharts, diagrams or concept maps to show connections or links between the following terms:
 - cells, DNA, nucleotides, nucleus
 - nitrogenous base, sugar, phosphate, nucleic acid, nucleotides
 - nitrogenous base, sugar, phosphate, deoxyribose, ribose, DNA, RNA, uracil, thymine, guanine, cytosine, adenine
 - DNA, mRNA, transcription, translation, amino acids, protein.

- For the DNA code below, suggest the (a) corresponding mRNA strand and (b) amino acids it codes for.
TAC CAT CGG CCA ATG ACG CGG CGG ATT

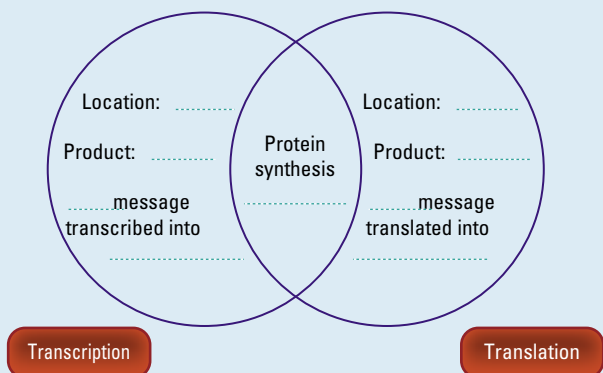
- All cells of a particular living thing, such as a spider, have the same sets of genetic instructions, but not all of that organism's cells have the same structure and function. Suggest what causes this and why cell specialisation is so important.

THINK AND DISCUSS

- Copy and complete the Venn diagram below using the following terms: thymine, uracil, deoxyribose, ribose, double-stranded, single-stranded, triplets, codons, adenine, guanine, cytosine, phosphate, nucleic acid.



- Copy and complete the Venn diagram below using the following terms: nucleus, protein, mRNA, ribosome, DNA, mRNA, protein, mRNA, mRNA.



CREATE

- 10 Design and make a model showing a simplified structure of DNA. Decide whether you wish to make a 3D or a 2D (flat) representation.
- What kinds of materials could you use in your construction?
 - Evaluate your model. What does it show or do well? What is it not able to show or do well?
- 11 Devise a role-play that demonstrates the way proteins are formed.
- 12 Rhymes such as the one below help us remember new information. Read or sing it, spelling out the triplets and codons with your fingers. Create your own rhyme about protein synthesis.

DNA is in my genes
Tells me how to make proteins
Got my genes from Mum and Dad
Mixed them up and made me glad
DNA is in my genes
Tells me how to make proteins.

DNA bases read times three
Always starting with TAC
mRNA codon would be AUG
DNA triplets tell the story of me
DNA bases read times three
Always starting with TAC.

ATT, ACT, ATC
Stop making proteins for me
mRNA codons for this would be
UAA, UGA, UAG
ATT, ACT, ATC
Stop making proteins for me.

INVESTIGATE

- 13 With increased knowledge and understanding, previous metaphors used to describe DNA are increasingly appearing to be less accurate in describing its complexities. The double helix, for example, describes its shape but not its function.
- Find out more about two of the following metaphors and suggest reasons why each is becoming less useful.

- Double helix
- Computer code of life
- Chemical building block
- Symphony of life
- Alphabet of life
- Blueprint
- Book of life

- In six words or less, suggest a metaphor that could be used to communicate what DNA is all about — especially to those who do not have a background in biology. Provide reasons to support the use of your metaphor.

- 14 James Watson (co-discoverer of the structure of DNA) and Craig Venter were both involved in investigating the human genome.

- Find out more about science as a human endeavour by following their two different stories of genome exploration, what they have in common, and how and why they clash. Start by clicking on the **James Watson** weblink in your eBookPLUS.

eBookplus

- Use the **DNA ownership** weblink in your eBookPLUS to watch an interview with James Watson in which he raises some interesting issues about the ownership of scientific discoveries that are worth reflecting on and discussing with other students.

- 15 Scientists have discovered a gene switch that has restored youthful vigour to ageing, failing brains in rats. Results from investigations suggest an 'on switch' for genes involved in learning. Injection of an enzyme flips the switch on and improves the learning and memory performance of older rats. Find out more about this type of research or other research that involves switching on genes.

- 16 Draw a timeline to show the rate of identification of human genes. A computer database called OMIM (On-line Mendelian Inheritance in Man) keeps a regular update. Use the **OMIM** weblink in your eBookPLUS to access the OMIM website.

eBookplus

- 17 Use this section of the textbook and other sources to research two scientists who contributed to the discovery of the double helix model of DNA. Write a brief account of their work.

- 18 Investigate further discoveries that have been made about DNA. Construct a timeline to share the who, what and when of your findings.

eBookplus

**work
sheet**

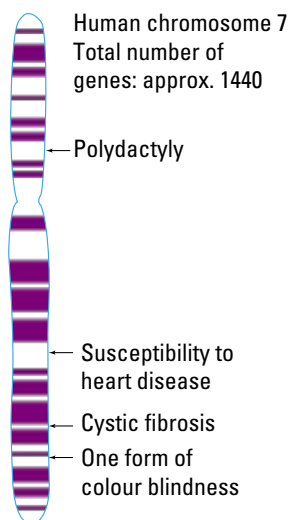
2.2 DNA

Who do you think you are?

You are very special. You have your very own unique DNA sequence. You have inherited this sequence from your ancestors. You are a human.

Where are your genes?

Much of who and what you are is determined by your genes. Genes determine many of the traits and characteristics that make you, you. A gene is a segment of double-stranded DNA that contains information that codes for the production of a particular protein or function. Located on specific chromosomes, humans possess around 20 000–24 000 genes. The position occupied by the gene on the chromosome is called its **locus**. Genes that are located on the same chromosome are described as being **linked**.



The locus for the cystic fibrosis gene is on chromosome 7. Polydactyly, cystic fibrosis and one form of colour blindness are linked genes — they are located on the same chromosome.

Genome maps

The total set of genes within an individual or cell is referred to as its **genome**. The study of genomes is called **genomics**.

Genome maps describe the

order of genes and the spacing between them on each chromosome. The genome size is often described in terms of the total number of base pairs (or bp). The genome size for organisms varies considerably: humans have about three billion base pairs, fruit flies about 160 million and brewer's yeast around 12 million.

The Human Genome Project

The Human Genome Project (HGP) was an international investigation to identify, sequence and study the genetic instructions within humans. Now that the information has been obtained, how can it be interpreted and further analysed? What are the potential benefits of this new knowledge? What ethical, social and political issues may arise?

Just the ingredients

It was anticipated that once we had the human genome sequenced, many mysteries would be unlocked and a new understanding of who we are would be unwrapped. Unfortunately, rather than an explosion of wonder and explanation, the sequencing only promoted more questions. Just like knowing the ingredients for a cake or the components of a car, we had the list, but not the delicious cake or speeding car.

The study of various genomes has revealed that the same genes that caused a fly to be a fly were also used to make a human a human. Parts of our genome are virtually interchangeable with those of our close primate 'cousins'. Rather than revealing the source of our diversity and uniqueness, our genome brought us closer to that of other life on Earth.

Epigenetics

While the HGP and its technologies provided us with information about the sequence of DNA, it is only part of the story. To understand more about its function, we may need to know more about the DNA of our ancestors. Maybe there are environmental triggers that switch on or off particular genes? If some of these involve lifestyle triggers, then could we be affected by the events our ancestors experienced? A new field called **epigenetics** suggests that this may be the case. This idea suggests that chemical changes can occur through environmental exposures and experiences that modify the DNA to a switched on or off form, and that these changes can be inherited.

This theory suggests that experiences of your great grandmother, for example, may have switched particular genes of hers on or off, and the modified gene(s) may have been passed on. Will you be involved in activities or events that change which of your genes are switched on, and then pass genes in this form to future generations?

HOW ABOUT THAT!

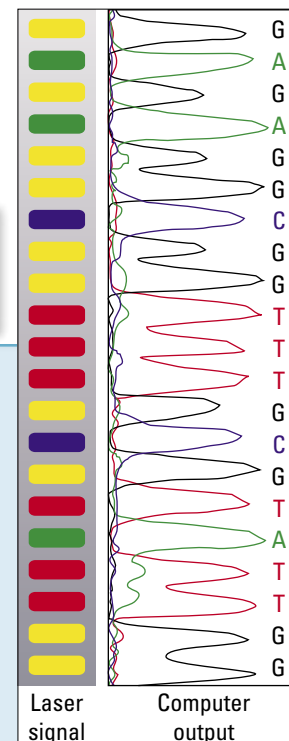
- Since mice and humans diverged from a common ancestor millions of years ago, most of the DNA that codes for functional genes has remained similar, whereas the 'junk' DNA has mutated and is now extremely different.
- Is this 'junk' DNA really junk? Could it have a purpose? What have we found out about it?

Gene sequencing

Gene sequencing identifies the order of nucleotides along a gene. DNA sequencers use four different-coloured fluorescent dyes (each binding to A, T, C or G in DNA) to identify the nucleotide sequence as it builds a complementary copy to the DNA template

sample provided. An example of the output of a DNA sequencer is shown at right.

DNA sequencers identify the base sequence of sections of a DNA fragment.



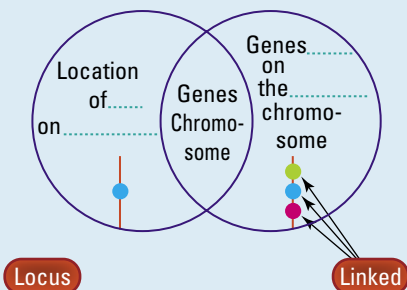
UNDERSTANDING AND INQUIRING

REMEMBER

- 1 Define the following terms: gene, locus, linked, genome, gene sequencing, gene map, genomics.
- 2 Describe the relationship between the following terms.
 - (a) Gene, chromosome, locus, linked
 - (b) Gene, genome, genomics, genome map
 - (c) Gene, gene sequencing, nucleotides, nitrogenous bases, DNA
- 3 Now that the human genome has been mapped, suggest three questions that could be asked.
- 4 Did the sequencing of the human genome answer our questions about why humans were unique? Explain.

THINK AND DISCUSS

- 5 Copy and complete the Venn diagram below using the following terms: gene, located, chromosome, same.



- 6 Different genetic instructions within and between species are due to different nucleotide sequences in their genes. The table below shows part of the sequences of different genes from various organisms.

Type of organism	Section of gene sequence
Duck	TAG GGG TTG CAA TTC AGC ATA GGG ATC
Human	TTG TGG TTG CTT TTC ACC ATT GGG TTC
Bacteria	AAT GAA TGT AAC AGG GTT GAA TTA AAA

- (a) Suggest how they are similar.
- (b) Suggest how they are different.
- (c) Suggest a reason why they all use the same letters in their genetic coding system.

- 7 Who do you think should know if you had a higher risk of dying from a genetic disorder in 15 years' time?

INVESTIGATE AND REPORT

- 8 Research and report on Craig Venter and Francis Collins and their research on the human genome.
- 9 Guidelines have been developed for companies in the US that supply 'custom DNA' or DNA sequences to order. These guidelines have been introduced to make it harder for bioterrorists to build dangerous viruses as potential bioweapons. There is concern, however, that as these rules are voluntary and most custom DNA is made outside the US, they may have limited value. Find out more about custom DNA, bioterrorism and bioweapons, and how these relate to gene sequencing.
- 10 Personal genome scans can provide a lot of information about your genetic disposition for particular diseases and disorders. They do not, however, always guarantee that you will show the disease. Find out more about the relationship between genotype, phenotype and environmental factors and how these relate to the use, accuracy and effectiveness of personal genome scans.
- 11 If you had a personal genome scan that suggested you have a 25 per cent chance of developing a disease, and if you were told that environmental factors such as diet and exercise were more important than possession of the genes, how would this affect your future lifestyle? Why?
- 12 Is *bio* the buzzword of the twenty-first century? Research and report on at least two of the following.
 - (a) Biotechnology
 - (b) Biomedicine
 - (c) Biomolecular scientist
 - (d) Biochemist
 - (e) Biophysicist

INVESTIGATE AND CREATE

- 13 Find out more about careers in genomics and genetic engineering, and research science fiction stories that include inheritance of interesting traits or genetic engineering. Based on your research, construct your own science fiction story about how knowledge of genetics may change our lives in the future. Share your work as a short story, animation or multimedia movie.

Dividing to multiply

All cells arise from pre-existing cells. That's pretty amazing when you really think about it! This means that all organisms living today originated from cells from the past. The cells you are made up of come from an unbroken line of cells. Where, when and who did your original cell come from?

Cell division in eukaryotes

Scientists are still grappling with many questions about the origin of life. Maybe you will be the one to shed new light on some possible answers in the future? What we do know, however, is there are two key types of cell division. Mitosis is the type of cell division involved in growth, development and repair of tissues. Some eukaryotic organisms also use mitosis for **asexual reproduction**. Organisms involved in **sexual reproduction** use another type of cell division in their reproductive process called meiosis.

Nucleus, chromosomes and DNA

All eukaryotic cells have a nucleus, which contains genetic information with instructions that are necessary to keep the cell (and organism) alive. This information is contained in structures called chromosomes, which are made up of a chemical called deoxyribonucleic acid (DNA).

Mitosis

What happens when skin wears away and damaged tissues need repairing? How do seedlings grow into giant trees? How did you get to be so big? Throughout the life of multicellular organisms, mitosis is the type

of cell division that is used for growth, development, repair and asexual reproduction.

The cells produced by mitosis are genetically identical to each other and to the original cell. They have the same number of chromosomes and DNA instructions. As they have identical genetic information, they are described as being **clones** of each other.

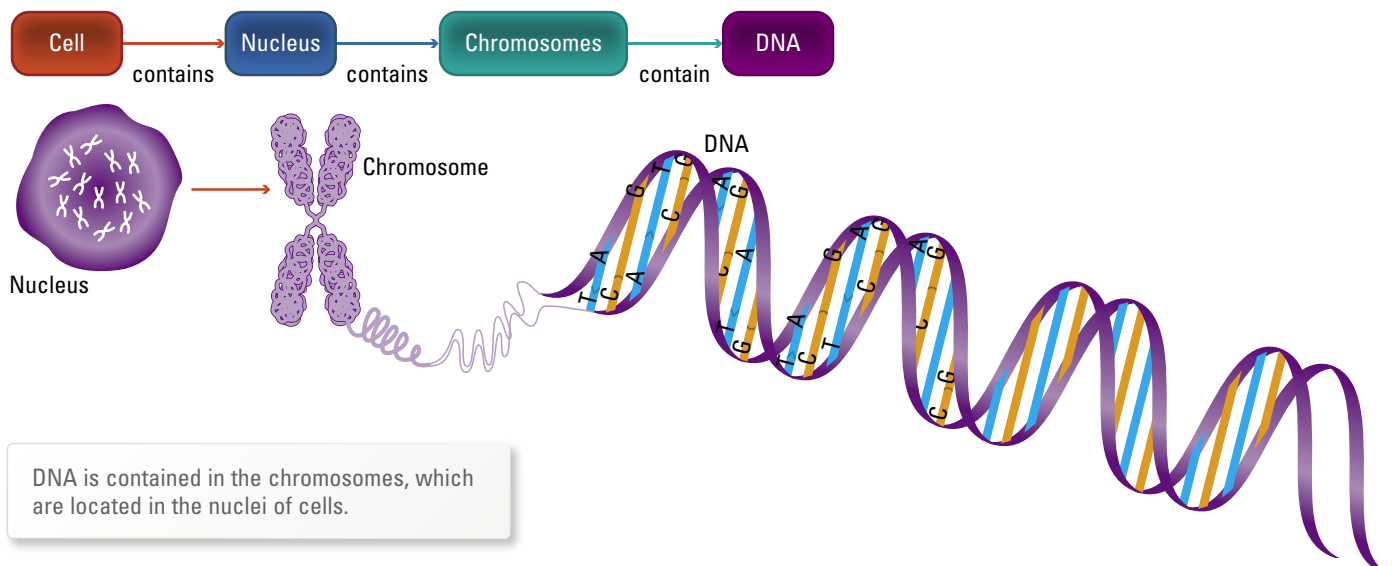
Cytokinesis

Mitosis is a process that involves division of the nucleus. Once a cell has undergone this process, the cell membrane pinches inwards so that a new membrane is formed, dividing the cell in two. The process of dividing the cytoplasm is called **cytokinesis**.

Counting chromosomes

Within the **somatic cells** (or body cells) of an organism, there is usually a particular number of chromosomes that is characteristic for their species. In humans, the total number of chromosomes in a somatic cell is 46. These chromosomes appear as 23 pairs in each body cell. The term used to describe chromosomes in pairs is **diploid**, because there are two sets of chromosomes.

Our gametes (or sex cells), however, contain only one set of chromosomes. They are referred to as being **haploid**. You may see the symbol n used to identify the haploid number. The diploid number would be identified as $2n$. How many sets of chromosomes do you think an organism would have if it was identified as $4n$ and **tetraploid**?



Meiosis

Why do gametes only have one set of chromosomes? If they didn't, then each time the egg and sperm nuclei combined during fertilisation, the number of chromosomes in the next generation of cells would double! For example, if each gamete had 46 chromosomes, the resulting cell after fertilisation would have 92 chromosomes.

Meiosis is the kind of nuclear division that prevents the doubling of chromosomes at fertilisation. It is a process in which the chromosome number is halved. In humans, that means the parent cell that is to undergo meiosis would initially be diploid ($2n$) and the resulting daughter cells or gametes produced by meiosis would be haploid.

A key source of variation

Variation within a species can provide some individuals with an increased chance of surviving over others. Depending on the environment and selection pressures at a particular time, different variations may be advantageous. Lots of different variations among individuals will mean that there is more chance that some will survive to reproduce. This improves the chances of the species surviving.

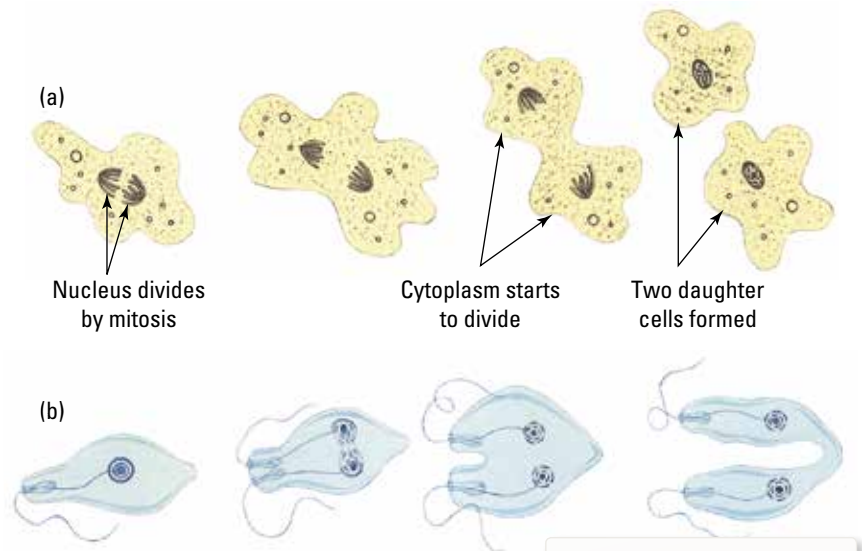
Meiosis mix-up

Each parent produces gametes by the process of meiosis. Within each gamete are chromosomes from each parent. Chromosomes carried in

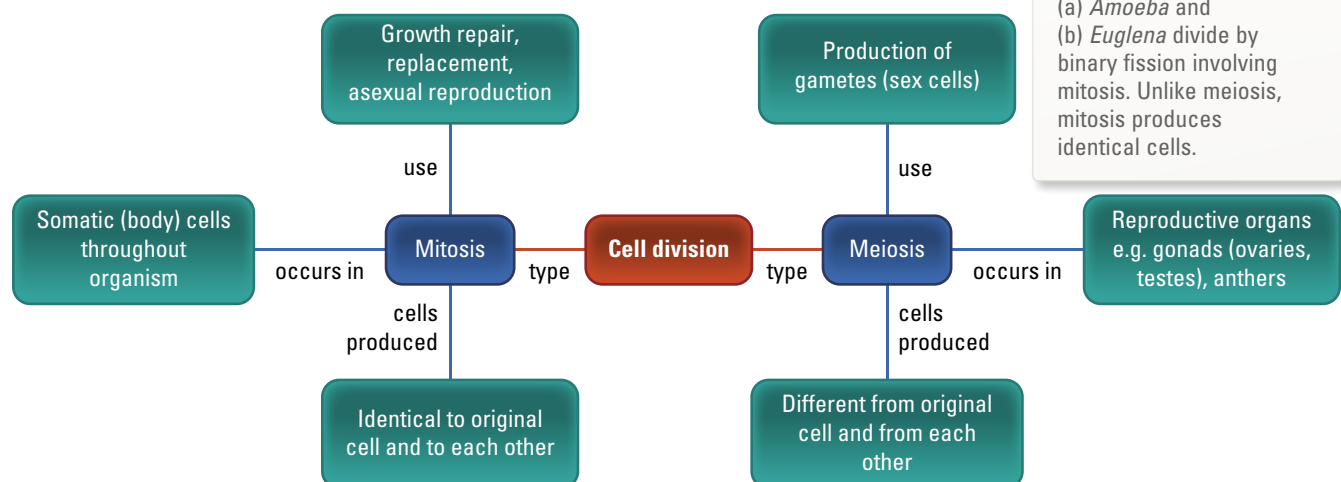
the sperm are referred to as **paternal chromosomes**, and chromosomes from the ovum are referred to as **maternal chromosomes**.

Meiosis provides sexually reproducing organisms with a source of variation. One way in which it increases variation is in terms of the number of combinations in which the chromosomes could be divided up into the gametes. For example, given that humans have 23 pairs of chromosomes, there are around 8 388 608 (2^{23}) different possible ways to divide up these chromosomes into each type of gamete.

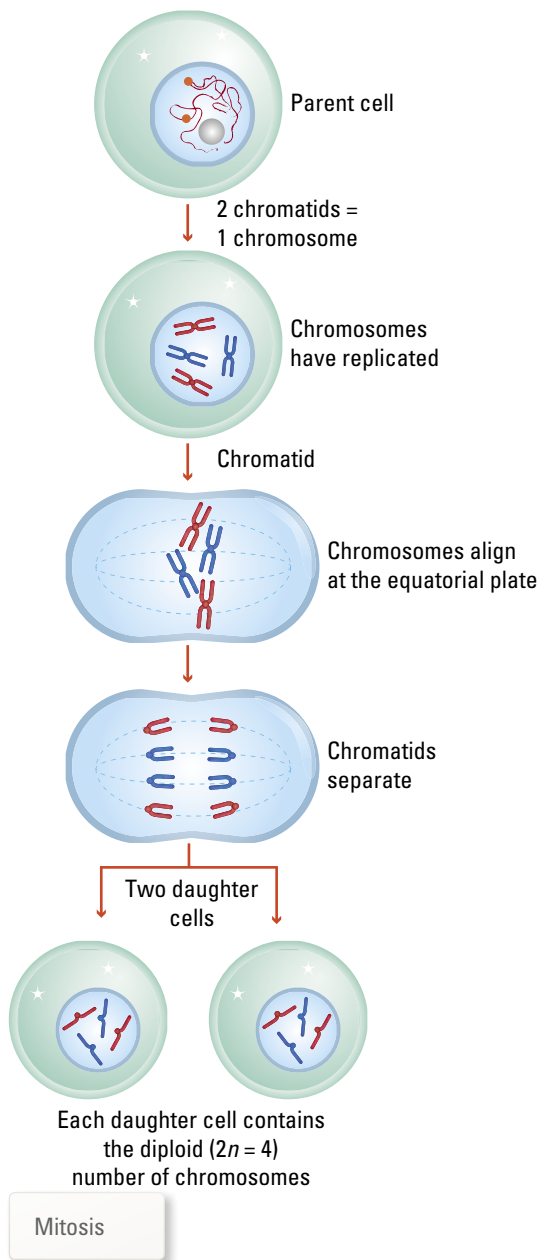
Another source of variation in meiosis is **crossing over** between chromosomes of each pair. This results in a section of one chromosome swapping its genetic information with another. For example, genes that were once on a paternal chromosome can be transferred or crossed over onto a maternal chromosome and vice versa.



Eukaryotic unicellular organisms such as (a) *Amoeba* and (b) *Euglena* divide by binary fission involving mitosis. Unlike meiosis, mitosis produces identical cells.



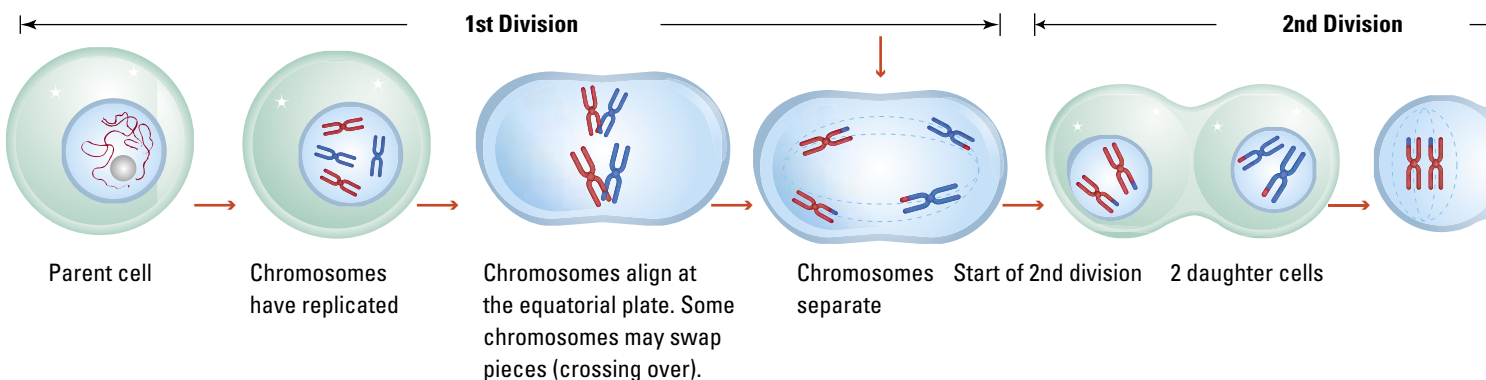
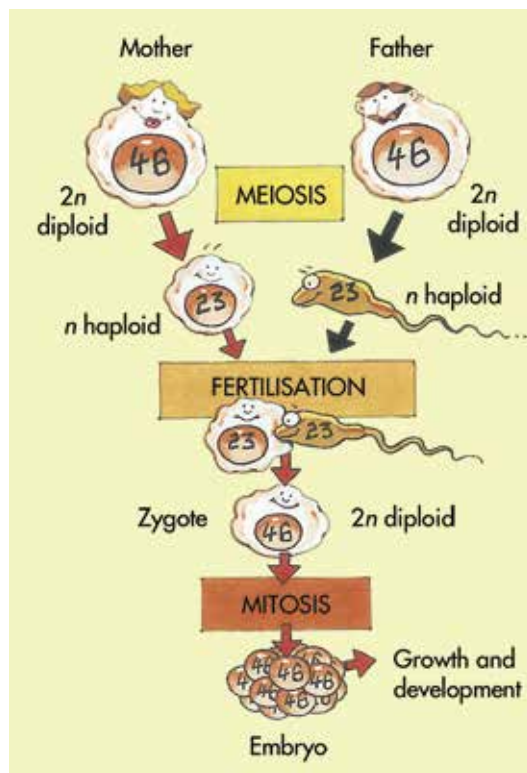
Mitosis and meiosis are two types of cell division.



Fertilisation

In humans, fertilisation occurs when a **haploid gamete** from each parent fuse together to form a **diploid zygote**. But which sperm will fertilise the ovum? The identity of the lucky sperm that will contribute its genetic information to the next generation depends largely on chance. Depending on which sperm fertilises the egg, there are many different genetic combinations possible. This is another source of genetic variation that can give sexually reproducing organisms an increased chance of survival.

The zygote contains 23 paternal chromosomes from its father and 23 maternal chromosomes from its mother. Each pair of chromosomes will consist of a chromosome from each parent. The zygote divides rapidly by mitosis to form an embryo that will also use this type of cell division to develop and grow. Each time



Meiosis: Crossing over of genetic information between each pair of chromosomes is a source of variation in a species.

this process occurs, cells with this complete new set of chromosomes are produced.

Boy or girl?

When a friend or family member is expecting a baby, one of the first questions people wonder or ask is whether it will be a boy or a girl. Probability suggests that the answer is that there is a 50 per cent chance either way. This can be predicted because it is determined by the sex chromosome combination that the child receives when the gametes from each parent fuse together at fertilisation.

Human somatic cells contain 22 pairs of autosomes and a pair of sex chromosomes. The sex chromosomes in the body cells of males and females differ. While females contain a pair of X sex chromosomes, males contain one X and one Y sex chromosome. Often this gender sex chromosome difference is abbreviated, so that females are described as being XX and males as being XY.

As a result of meiosis, gametes will contain only one sex chromosome. Human females (XX) can only produce gametes that contain an X chromosome. Human males (XY), however, will produce half of their gametes with an X chromosome and the other half with a Y chromosome. So, if a

gamete containing a Y chromosome fuses with the ovum (which contains an X chromosome), the resulting zygote will be male (XY). Likewise, if the ovum is fertilised by an X-carrying gamete, then a female (XX) will result.

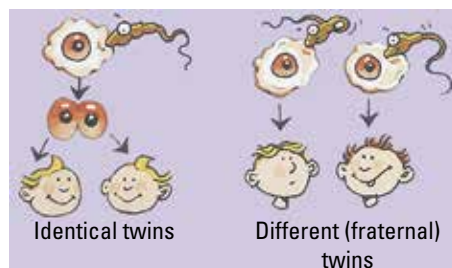
HOW ABOUT THAT!

The gender-determining factors of other animals can be quite different from those of humans. In birds, for example, it is the female that has different sex chromosomes, Z and W, and the male has two Z chromosomes. In some reptiles, gender is determined by the temperature at which the egg is kept rather than chromosomes. The temperature of the sand in which some crocodiles and turtles bury their eggs can determine whether the offspring will be male or female. The gender of brushturkey chicks is also determined partly by temperature.

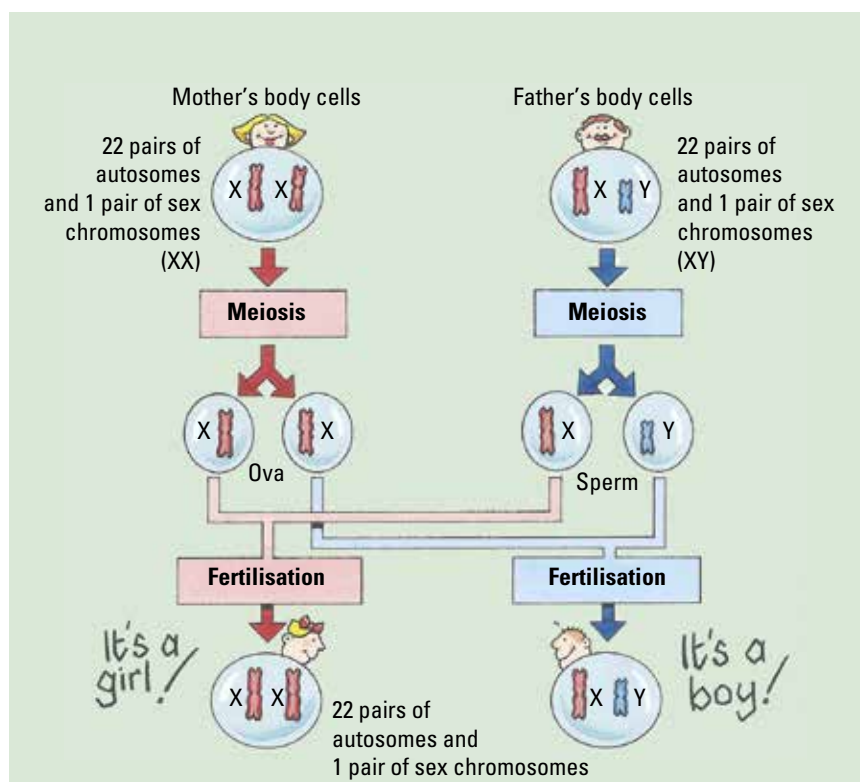
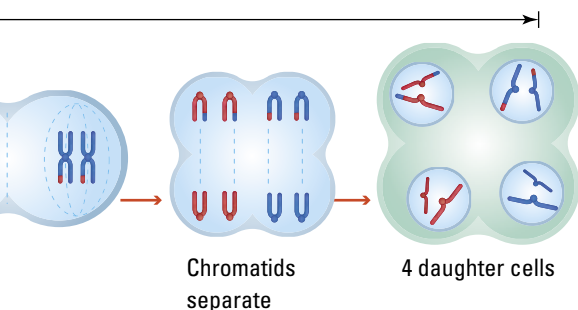
Twins — or more!

Sometimes in the very early stages of division following fertilisation, clusters of a few cells develop into two separate individuals. If this happens, identical twins result as each cluster has the same genetic make-up as the other.

Usually, only one ovum is released at a time. However, if several are released, twins can result from fertilisation by different sperm. In this case, the babies are not identical because they have different genetic make-ups.



Identical or fraternal twins — one sperm or more?



Is the mother or father the key determiner of the gender of the child?

INVESTIGATION 2.2

What's the chance?

AIM To simulate the chance of a male or female being conceived at fertilisation

Materials:

20-cent coin

METHOD AND RESULTS

- After reading the instructions and before you carry out the experiment, predict the number of times you will toss heads and the number of times you will toss tails. Give a reason for your prediction.
- Toss a coin 50 times. Count the number of heads and tails and record the data in a table like the one below.

	Number of heads	Percentage of heads	Number of tails	Percentage of tails
Individual tosses				
Combined class result				

- Calculate the percentage chance of obtaining heads and the percentage chance of obtaining tails.
- Combine the results of the whole class and calculate the percentage chance of obtaining heads and tails.

DISCUSS AND EXPLAIN

- Draw a graph of your results.
- Analyse your data.
 - Was your prediction supported or not?
 - Were the percentage results obtained for 50 tosses the same as or different from the total class results? Suggest reasons for the similarities or differences.
- If you tossed a coin 1000 times, would you obtain similar results?
- What is the chance of obtaining heads each time you toss the coin?
- If heads represented a sperm carrying an X chromosome and tails represented a sperm carrying a Y chromosome, suggest how this activity could link to the chances of a male or female baby being conceived.
- Suggest a strength, a limitation and an improvement for this investigation.

UNDERSTANDING AND INQUIRING

REMEMBER

- Where does the cell theory suggest that cells come from?
- State the names of the two main types of cell division.
- List three functions of mitosis.
- What is DNA an abbreviation of?
- Use a flowchart to show the link between the terms: DNA, cell, nucleus and chromosome.
- Describe the features of the offspring cells produced by mitosis.
- Distinguish between the following pairs of terms.
 - Cytokinesis and mitosis
 - Mitosis and meiosis
 - Diploid and haploid
 - Ovum and sperm
 - Maternal chromosomes and paternal chromosomes
 - Gamete and zygote
 - Fertilisation and meiosis
 - Autosomes and sex chromosomes
 - XY and XX
 - Somatic cells and gametes
 - Identical twins and fraternal twins

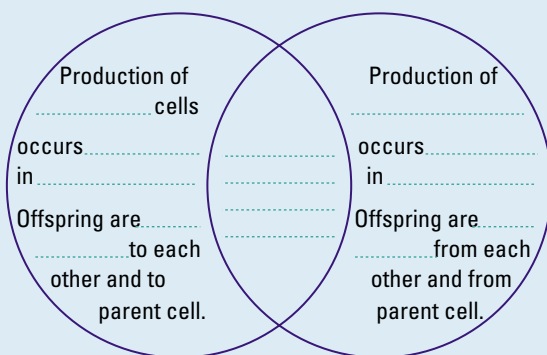
THINK AND DISCUSS

- Who am I?
 - My other name is sex cell.
 - I reduce the number of chromosomes in the daughter cells by half that of the parent cell.
 - I describe the number of chromosomes in normal human somatic cells.
 - I describe the fusion of gametes.
 - I describe the number of chromosomes in human gametes.
 - I am a type of cell division important for growth, repair and replacement.
- Copy and complete the table below.

Type of cell division	Why use it?	Where does it occur?	Features of cells produced
Mitosis			
Meiosis			

- With the use of a diagram, explain how the sex of a human baby is determined.

- 11 If a woman has already given birth to three boys, what are the chances of her next child being a girl?
- 12 In many cultures throughout history, a woman has been blamed for not producing sons and has been divorced. From a biological point of view, could this be justified? Explain your answer.
- 13 A few genetic traits, such as hairiness in ears, are due to genes carried on the Y chromosome. Would males and females have the same chance of having the trait?
- 14 Copy and complete the Venn diagram below, choosing from the following terms: somatic, only, body, gonads, gametes, anywhere, different, identical, chromosomes, cell division, eukaryotes.



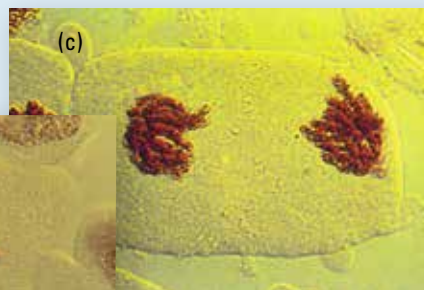
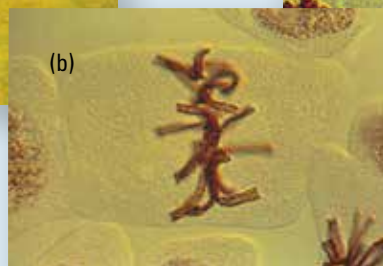
Mitosis

Meiosis

- 15 The Y chromosomes of human males are shorter than the X chromosomes. Would the same number of genes be carried by both chromosomes? Discuss your response.

THINK, ANALYSE AND DISCUSS

- 16 Figures (a)–(d) below show bluebell cells in various stages of mitosis. Suggest which order they should be placed in.



- 17 Using the table below, suggest the possible effect of increasing global temperatures on turtles, crocodiles and lizards.

Temperature control of sex in some reptiles

Reptile	Cold 20–27 °C	Warm 28–29 °C	Hot > 30 °C
Turtle	Male	Male or female	Female
Crocodile	Female	Male	Female
Lizard	Female	Male or female	Male

INVESTIGATE

- 18 The kind of job a man does can affect whether he produces more or less Y sperm or any sperm at all. Chemicals and hormones washed into waterways or used in producing food can affect fertility. Research an example of an environmental impact on fertility and report your findings. Make sure you quote the sources of your information.

REMEMBER

- 19 Complete the **Mitosis and meiosis** interactivity in your eBookPLUS to test your knowledge of the different processes of cell division, and challenge yourself to see if you can differentiate between mitosis and meiosis. **int-0680**

eBookplus

- 20 To find out more about the different types of cell division use the **Mitosis** and **Meiosis** weblinks in your eBookPLUS.

eBookplus

work sheets 2.3 Mitosis
2.4 Meiosis

The next generation

Have you ever browsed through the family photo album and looked at family members at different ages? Did any look like you? Which features do you share with them? Do certain characteristics seem to appear and disappear from one generation to the next? How could this happen?

From one generation to the next

The passing on of characteristics from one generation to the next is called **inheritance**. The study of inheritance involves a branch of science called **genetics**. These characteristics or features are examples of your **phenotype**. Your phenotype is determined by both your **genotype** and your environment. Your genotype is determined by genetic information in the chromosomes that you received in the gametes of your parents.

It's not all about your genes!

Environmental factors contribute to characteristics that make up your phenotype. Your weight, for example, although influenced by genetic factors, is also

influenced by what you eat and how active you are. Exposure to and use of chemicals in your environment (such as pollution, hair dyes, tanning lotions and make-up), stress, intensity of sunlight and temperature ranges are other examples of environmental factors that can contribute to your phenotype.

A product of chance

The similarities and differences in how you look compared to your relatives are partly due to chance. Chance was involved in which of the many sperm produced by your father fertilised your mother's ovum.

When fertilisation takes place, the zygote receives a pair of each set of chromosomes, the maternal and paternal chromosomes. Located within these chromosomes are the genes for particular characteristics. In the family generations diagram on the next page, the inheritance of the gene for eye colour is illustrated. There are two different eye colours shown. These alternative forms or expressions of a gene are called **alleles**.

INVESTIGATION 2.3

How does the environment affect phenotype?

AIM

- 1 Write an aim for this experiment.

Materials:

10 seedlings grown from cuttings of the same plant
potting mix in two small pots

METHOD AND RESULTS

- 2 Copy and complete the following table:

	Pot A	Pot B
Number of seedlings that are still alive		
Colour of leaves		
Average height of seedlings		
Average number of leaves per seedling		

- Plant five of the seedlings in pot A and five in pot B.

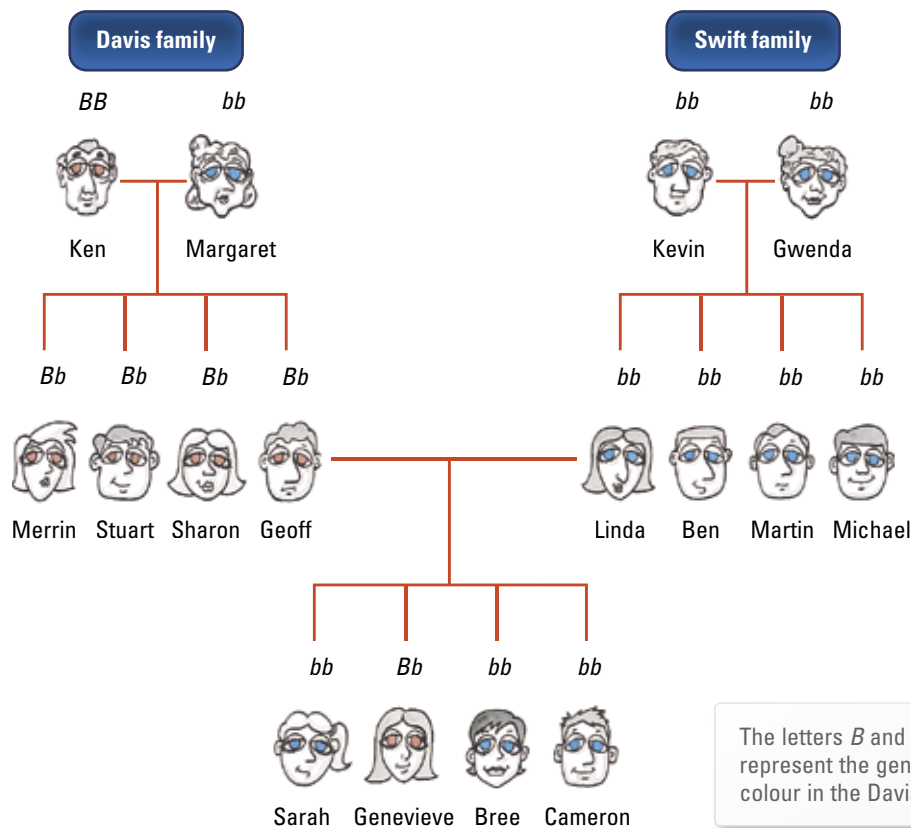
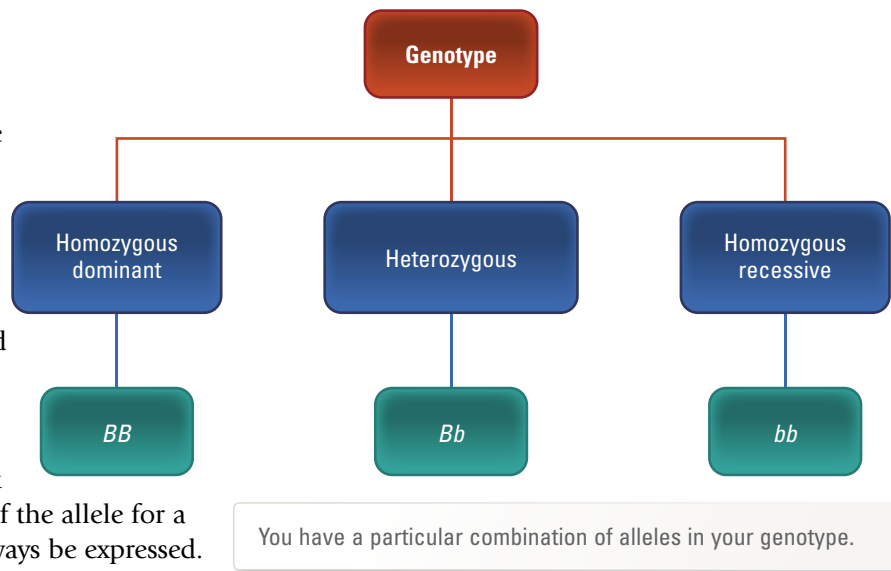
- Place pot A in a dark cupboard and pot B near a window.
- Leave the plants undisturbed for two weeks. Water both pots when necessary. Ensure you use the same amount of water for both plants. After two weeks compare the plants in both pots.

DISCUSS

- 3 Explain how you calculated the average number of leaves and the average height of the seedlings.
- 4 In this experiment:
 - (a) what is the independent variable
 - (b) what is the dependent variable
 - (c) which environmental factors were controlled?
- 5 Why is it important to use seedlings grown from cuttings of the same plant for this experiment?
- 6 Why were five seedlings planted in each pot?
- 7 Construct graphs of your data.
- 8 Comment on observed patterns in your data.
- 9 Explain why this experiment demonstrates that environmental factors play a part in determining the phenotype of an organism.

Hide and seek

The mixing of your parents' chromosomes at fertilisation resulted in two alleles for each gene coming together. Each of these alleles can be described using a letter. In the family generations diagram below, the expression of the gene for eye colour is shown. The allele for brown eyes is denoted as a capital letter *B*, because it is the **dominant** trait. The allele for blue eyes has been denoted by a lower case letter *b*, because this trait is **recessive** to the brown eye trait. If the allele for a dominant trait is present, it will always be expressed. The recessive trait is hidden in the presence of the dominant trait and can be expressed only if the allele for the dominant trait is not present.



Mix and match

The combination of the alleles that you have for a particular gene is called your genotype. If your alleles for that gene are the same (e.g. *BB* or *bb*), then you are described as **homozygous** (or **pure breeding**) and if they are different (e.g. *Bb*) then you are **heterozygous**

(or **hybrid**) for that trait. A genotype of *BB* can be described as **homozygous dominant**, while a genotype of *bb* can be described as **homozygous recessive**. So the genotype has to do with the combination of alleles present; the phenotype describes the expression of the trait (e.g. brown or blue eyes).

INVESTIGATION 2.4

Genetics database

AIM To increase awareness of a variety of inherited traits

METHOD AND RESULTS

- Copy and complete the table below. Enter data for 10 students in the table. You may need to refer to the pictures below to work out what each characteristic means.

Name of student				
Widow's peak?				
Can roll tongue?				
Right thumb over left when clasp hands?				
Cleft chin?				
Right handed?				
Ear lobes attached?				
Freckles?				
Gap between front teeth?				
Hair naturally straight?				
Colour blind?				



Do you have a widow's peak (left) or a straight hairline (right)?



Do you have a gap between your front teeth?

- Use the instructions provided in your eBookPLUS to create an **Access** database where you will enter the data you collected and run a query on the database.

eBookplus

DISCUSS AND EXPLAIN

- The database you created contains only a small amount of data so using a query to search for particular data did not save time. (It probably took you more time to set up the query than it would have taken to look through the data manually!) Can you think of examples of databases that contain so much information that it would take days to search the data manually?
- Does your school keep a computerised database of student details? What type of information is kept in the database?



When you clasp your hands, is your right or left thumb on top?



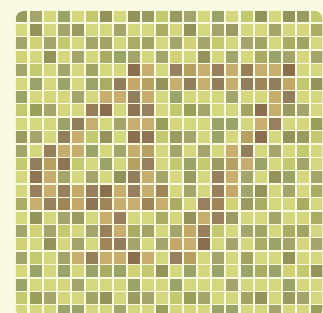
Do you have a smooth or cleft chin (shown above)?



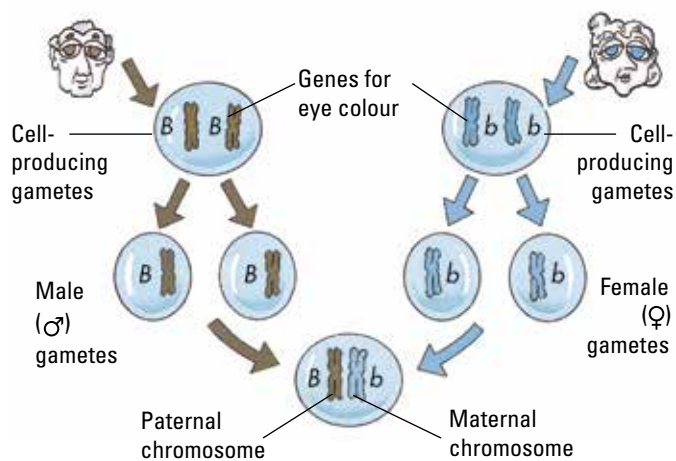
Are your ear lobes detached (left) or attached (right)?



Can you roll your tongue?



If you cannot see the number 47 in the picture above, you could be colour blind.



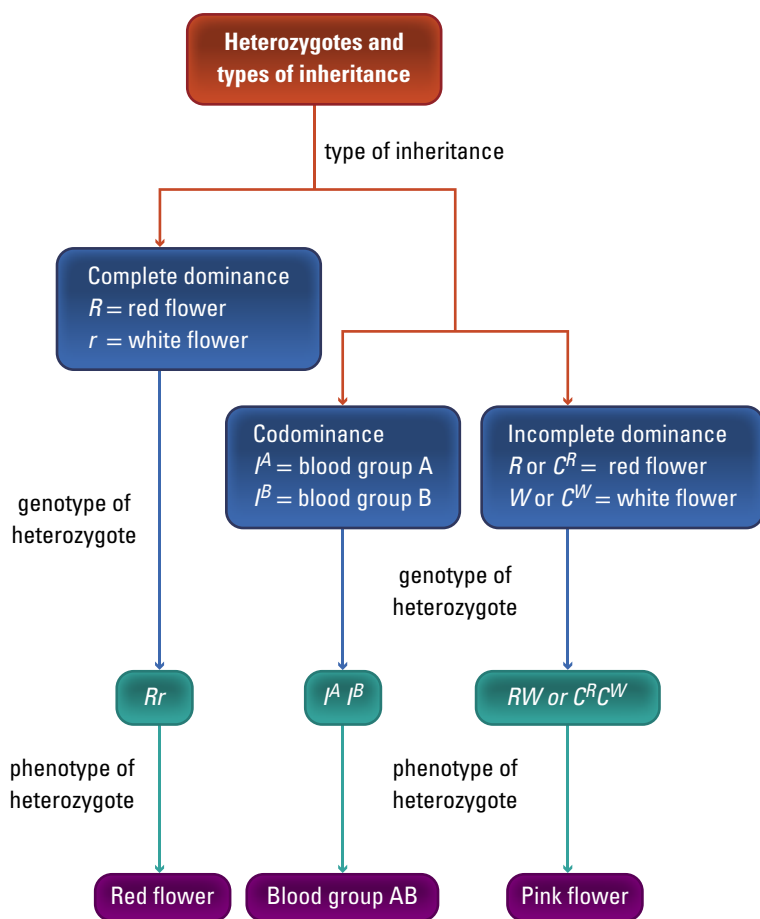
Alleles on chromosomes inherited from each of your parents contribute to your genotype.

Are you a carrier?

The term **carrier** refers to someone who is heterozygous for a particular trait and carries the allele for the recessive trait (such as the alleles for blue eyes or red hair). Generally people are not aware of being a carrier because it does not show in their phenotype. They may, however, have children that show the recessive trait. Can you suggest how two brown-eyed parents (dominant trait) could have a child who has blue eyes (recessive trait)?

Degrees of dominance

In **complete dominance**, the expression of one trait is dominant over the other. This results in both the homozygous dominant and heterozygous genotypes being expressed as the same phenotype. There are two other types of inheritance in which neither allele is dominant over the other. In **codominance**, the heterozygote has the characteristics of both parents. An example of this type of inheritance is seen in the human blood groups. In **incomplete dominance**, the heterozygotes show a phenotype that is intermediate between the phenotypes of the homozygotes. An example of this type of inheritance is seen in the flower colour of snapdragons.



The phenotype of the heterozygote can indicate the type of inheritance.

	Donor's blood			
	O	A	B	AB
Patient's blood	O			
	A			
	B			
	AB			

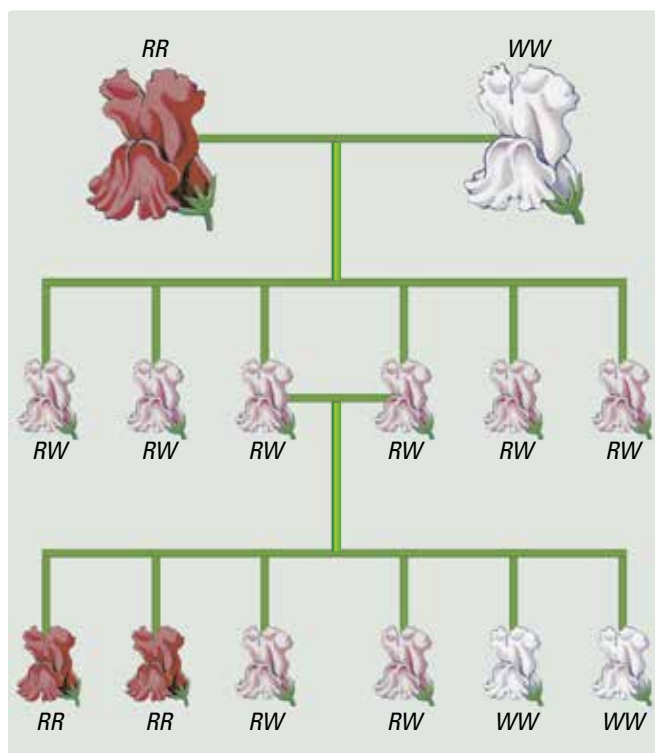
The inheritance of the human ABO blood groups is by codominance. The type of blood group you have determines who you can donate to or receive blood from. Which blood type are you? Are you the same blood type as either of your parents?

Incomplete confusion

You will find that there may be variations in the definitions of the terms recessive, dominant, codominance and incomplete dominance in various resources. New technologies and new knowledge can modify how we see, understand and communicate our knowledge. This eventually results in the creation, modification or replacement of terminology and theories that are used by a majority or enforced by those with the highest authority or persuasion.

Both codominance and incomplete dominance can be considered examples of **partial dominance**. The common feature of these types of inheritance is that the **heterozygote** will show or express a phenotype that is different from the phenotype of an individual with either homozygous genotype.

Another area of confusion is with the terms recessive and dominant. Some resources abbreviate 'the allele for the recessive and dominant trait or phenotype' as 'recessive allele and dominant allele'. In terms of biology, it is increasingly accepted that the expression of the genotype as a particular phenotype is what is dominant or recessive, rather than the allele itself. If you continue on with senior Biology, it is important for you to check which of these definitions your authorities assess by.



Suggest how red-, white- and pink-flowered offspring can result from pink-flowered parents.

Mendel's memos

Gregor Mendel (1822–1884), an Austrian monk, carried out experiments on pea plants in a monastery garden for 17 years. His work was unknown for about 35 years. When it was discovered in 1900, he became known as the 'father of genetics'.

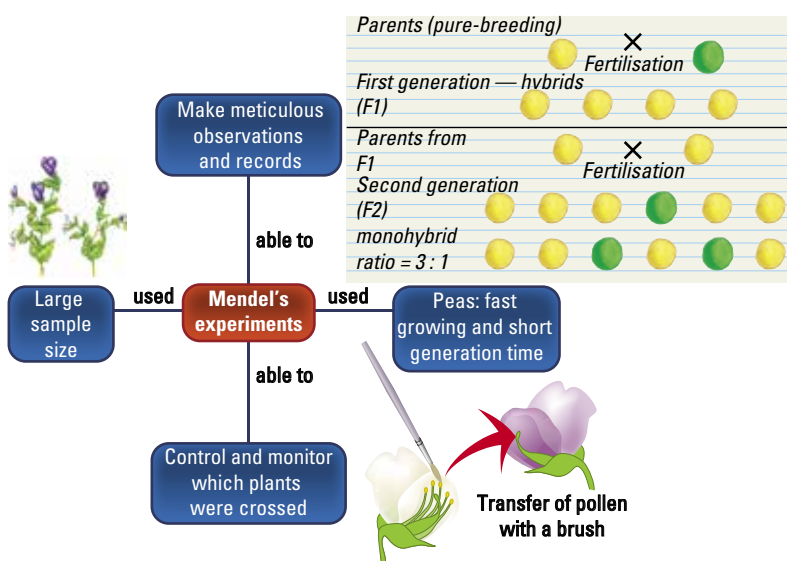


Gregor Mendel

From his experiments, Mendel was able to explain patterns of inheritance of certain characteristics. Why did Mendel use pea plants and not cabbages? Pea plants are easily grown in large numbers and have easily identifiable characteristics that have either/or alternatives. Mendel could control their breeding by taking pollen from a particular pea plant and putting it on the stigma of another. Pea plants can also be self-pollinated.

Mendel crossed a pure-breeding tall plant with a pure-breeding short plant. A plant is pure breeding for a characteristic if it has not shown the alternative characteristic for many generations.

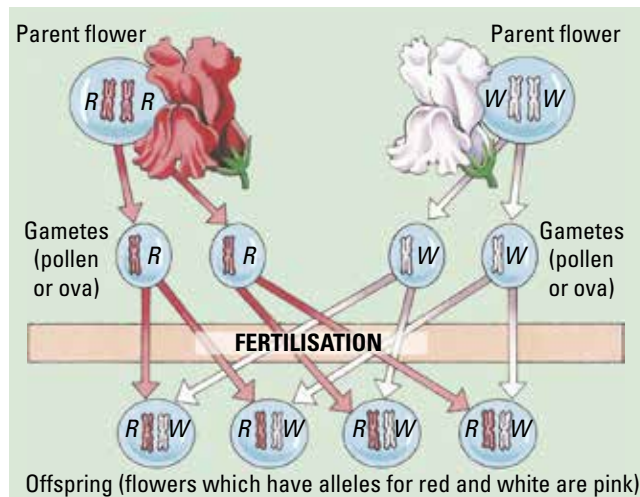
Mendel showed the factor for shortness had not disappeared because when he crossed the tall offspring (called the F_1 generation) with each other, about a quarter of those offspring (called the F_2 generation) were short. He called shortness a recessive factor because it was hidden or masked in the F_1 generation.



Mendel's experiments were well designed and his record-keeping was meticulous.

	Dominant trait	Recessive trait
Stem length		
Seed (cotyledon) colour	Tall  Yellow	Short  Green
Seed (cotyledon) shape	 Round	 Wrinkled
Seed coat colour	 Grey	 White
Pod texture	 Inflated	 Constricted
Pod colour	 Green	 Yellow
Flower position	 Flowers along length of stem	 Flowers at top of stem

Pea plants showing the characteristics Mendel used in his experiments



Incomplete dominance can result in offspring that express a phenotype not observed in either parent.

A plant is a hybrid if it has parents with both alternatives, such as tallness and shortness, for a characteristic. We now know that Mendel's 'factors' are genes. The alternative forms of the factors are alleles.

Mendel bred plants for single characteristics such as height. He worked out that if many pure-breeding tall and short plants were crossed and then the first generation (F_1 generation) was also crossed, the ratio of tall to short plants would be about 3 : 1. He repeated these experiments many times using the other characteristics of the pea plants and came up with similar ratios. This is called the **monohybrid ratio**.

UNDERSTANDING AND INQUIRING

REMEMBER

- 1 Distinguish between the following pairs of terms. You may wish to use symbols, visual thinking tools or diagrams in your responses.
 - (a) Inheritance and genetics
 - (b) Genotype and phenotype
 - (c) Chromosomes and gametes
 - (d) Fertilisation and zygote
 - (e) Genes and alleles
 - (f) Dominant trait and recessive trait
 - (g) Homozygous and heterozygous
 - (h) Homozygous dominant and homozygous recessive
 - (i) Carrier and recessive trait
 - (j) Dominance and codominance
 - (k) Maternal and paternal chromosomes
 - (l) Pure breeding and hybrid

- 2 Describe the difference between the phenotypes of a heterozygous individual for a trait that shows complete dominance and for a trait that shows codominance.
- 3 State how many alleles there are on a homologous pair of chromosomes for a particular trait. Provide a reason for your response.
- 4 Suggest four strengths in the design of Mendel's experiments.

THINK AND DISCUSS

- 5 Suggest why the ability to self-pollinate and cross-pollinate was an advantage for the pea plants in Mendel's experiments.
- 6 (a) Suggest the phenotype of a pea plant that showed:
 - (i) dominant traits for seed colour, shape and coat colour

- (ii) recessive traits for stem length and flower position
- (iii) a dominant trait for pod texture, but a recessive trait for pod colour.

(b) Devise a simple table to include the phenotype and genotype for the trait of each plant. Use an appropriate letter to match each of the characteristics Mendel studied.

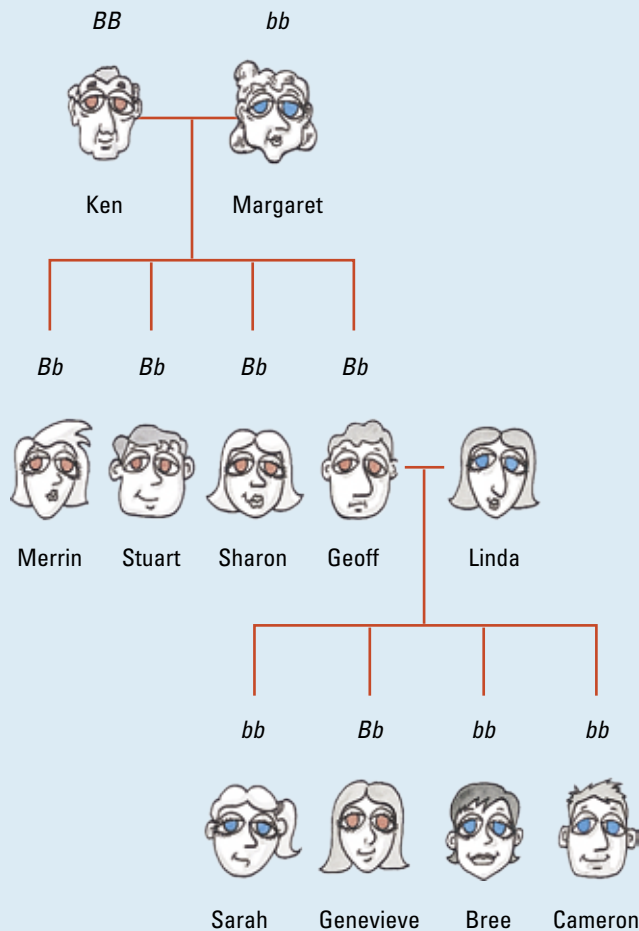
7 Mendel obtained a ratio of 3 tall to 1 short plant in the offspring when he crossed pure-breeding tall and short plants. Convert this monohybrid ratio of 3 : 1 into a:

- (a) fraction
- (b) percentage.

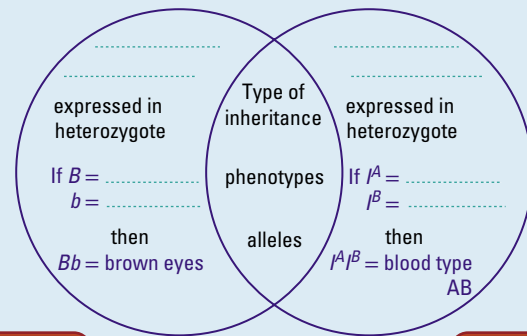
8 Suggest the colour(s) of snapdragon flowers that you would expect in the offspring of a red-flowered plant and a pink-flowered plant.

9 Refer to the Davis family tree diagram below to answer the following questions.

- (a) Could the parents of the Davis family, Ken and Margaret, ever have offspring with blue eyes? Explain your answer.
- (b) Suggest why all of Geoff and Linda's children do not have blue eyes.



10 Copy and complete the Venn diagram below using the following terms: dominant phenotype, brown eyes, both phenotypes, blood type A, blood type B, blue eyes.



Complete dominance

Codominance

11 Construct a Venn diagram with the headings 'Determined by genetics' and 'Determined by environmental factors', and then place the following terms in the most appropriate category: eye colour, tattoo, skin colour, cleft chin, freckles, colour blind, hair colour, scar, widow's peak.

THINK, DISCUSS AND CREATE

12 On your own, in pairs or in teams, create a rhyme, song or poem that effectively uses as many of the key terms in this section as possible. An example is shown below. Add movements or actions for each line and share it with your class.

Alleles are alternative forms of genes
Sometimes showing, sometimes behind the scenes
Genotypes are made up of two of them
Homozygotes have two the same
Heterozygotes have one of each kind
From each parent, alleles you will find.

INVESTIGATE

- 13 Investigate one of these genetic conditions: Huntington's disease, Tay-Sachs, cystic fibrosis, fragile X syndrome, PKU. Briefly describe the disease. Is it dominant or recessive?
- 14 Complete the **Making families** interactivity in your eBookPLUS. Challenge yourself to complete the family — mother, father and offspring — that demonstrates each dominance type as it appears on-screen. **int-0681**

eBookplus

work sheets

2.5 Dominant and recessive
2.6 Mendel's experiments

What are the chances?

Selecting a mate can be one of the most crucial decisions in your life. This selection process involves both conscious and unconscious choices. Next time you look at that special person, take a *really* good look. One day you might be mixing your genes together! What might the result be?

Predicting possibilities

Reginald Punnett (1875–1967) was a geneticist who supported Mendel's ideas. He repeated Mendel's experiments with peas and also did his own genetic experiments on poultry. Punnett is responsible for designing a special type of diagram, which is named after him. A **Punnett square** is a diagram that is used to predict the outcome of a genetic cross.

A Punnett square shows which alleles for a particular trait are present in the gametes of each parent. It then shows possible ways in which these can be combined. The alleles in each of the parent's genotypes for that trait are put in the outside squares and then multiplied together to show the possible genotypes of the offspring.

Punnett rules

When using a Punnett square for a dominant/recessive inheritance, you use a capital letter for the allele of the dominant trait (e.g. *B*) and a lower-case version of the same letter for the allele for the recessive trait (e.g. *b*). If the type of inheritance is incomplete or codominant, then different letters are used to represent them

Punnett square for $Bb \times Bb$

B = allele for brown eyes
b = allele for blue eyes

		Father	
		<i>B</i>	<i>b</i>
Mother	Possible gametes	<i>B</i>	<i>b</i>
	<i>B</i>	<i>BB</i>	<i>Bb</i>
	<i>b</i>	<i>Bb</i>	<i>bb</i>

Offspring probabilities

Genotype: $\frac{1}{4} BB$; $\frac{1}{2} Bb$; $\frac{1}{4} bb$

Phenotype: $\frac{3}{4}$ brown eyes; $\frac{1}{4}$ blue eyes

In a Punnett square, alleles from each parent's genotype are used to determine the possible genotypes and phenotypes of the offspring.

(e.g. *R* and *W* or I^A and I^B). The sex chromosomes are included when an **X-linked trait** is involved (e.g. $X^B X^b$ and $X^B Y$).

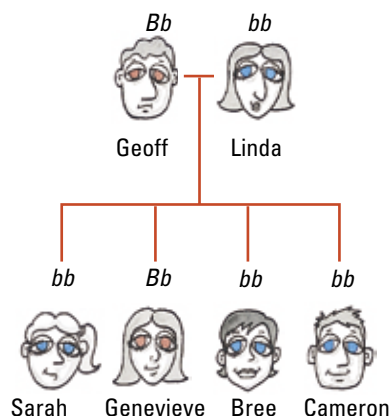
What is the chance?

The chances of having offspring that show a particular trait depends on the trait's type of inheritance; that is, whether it is inherited by complete dominance, codominance, incomplete dominance or **sex-linked inheritance**.

Complete dominance

Remember Linda Swift and Geoff Davis from section 2.6? The inheritance of eye colour was shown in their family. Inheritance of brown eyes was dominant to the inheritance of blue eyes. The diagrams below show that Geoff has brown eyes (*Bb*), Linda has blue eyes (*bb*) and their children have either brown or blue eyes.

But how were the alleles from each parent inherited? The mix of alleles that Linda and Geoff contributed to Genevieve and Cameron's genetic make-up can also be shown in the format at the top left of the next page.



	Linda	Geoff	Genevieve	Cameron
Chromosomes and genes for eye colour				
Genotype	<i>bb</i>	<i>Bb</i>	<i>Bb</i>	<i>bb</i>
Phenotype	Blue eyes	Brown eyes	Brown eyes	Blue eyes

The inheritance of blue eyes is recessive to the inheritance of brown eyes.

Linda's eggs			
Geoff's sperm		$\frac{1}{2}$ b	$\frac{1}{2}$ b
	$\frac{1}{2}$ B	$\frac{1}{4}$ Bb brown	$\frac{1}{4}$ Bb brown
	$\frac{1}{2}$ b	$\frac{1}{4}$ bb blue	$\frac{1}{4}$ bb blue

Each parent contributes alleles to the genotype of their offspring.

This can be more simply written as a Punnett square.

Punnett square $Bb \times bb$
 B = allele for brown eyes
 b = allele for blue eyes

		Geoff	
		Possible gametes	
Linda	b	Bb	bb
	b	Bb	bb

Offspring probabilities
 Genotype: $\frac{1}{2} Bb$: $\frac{1}{2} bb$
 Phenotype: $\frac{1}{2}$ brown eyes: $\frac{1}{2}$ blue eyes

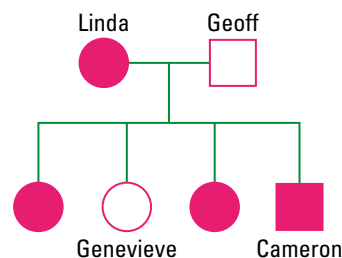
Punnett squares show us the chance of offspring inheriting particular combinations.

This tells us that the chance of producing a child with the combination Bb (heterozygote) is 2 out of 4 or $\frac{1}{2}$, and the chance of producing the combination bb (homozygous recessive) is 2 out of 4 or $\frac{1}{2}$. Hence, each of Linda and Geoff's children have a 50 per cent chance of having blue eyes and a 50 per cent chance of having brown eyes. All of their children could have had blue eyes or all brown eyes. It is important to note that the chance of inheritance calculated for one child is not dependent on the inheritance of another.

Pedigree charts

A diagram that shows a family's relationships and how characteristics are passed on from one generation

to the next is a **pedigree chart**. A pedigree chart for Linda and Geoff's family is shown below. Instructions on how to draw your own pedigree chart are on the next page.



Key

= Female with blue eyes	= Female without trait
= Male with blue eyes	= Male without trait

What is your blood type?

Do you know which type of blood you have flowing through your capillaries? The inheritance of blood types A, B, AB and O are determined by the ABO gene.

Multiple alleles

There are three different alleles for the ABO gene. Two of these carry instructions to make a particular type of protein called an **antigen**; the other does not. The types of antigens coded for by the alleles are different. One allele codes for antigen A and the other codes for antigen B. If you possess both of these alleles, then you have the instructions to produce both antigen A and antigen B. This is an example of **codominant inheritance** because both blood types are expressed in the heterozygote.



There is an element of chance in why you are you!

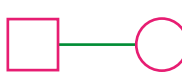
SOME RULES FOR DRAWING PEDIGREE CHARTS

1. To show the gender of an individual:

 a square is used to represent a male

 a circle is used to represent a female.

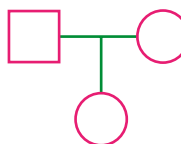
2. To show the marriage or breeding relationship between individuals:

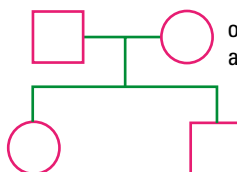
 a line connecting the male and female is used to represent a breeding couple or marriage.

3. To show the offspring relationships:

 a line from the breeding couple/marriage line indicates children.

For example,

 an only child (in this case, a daughter)

 or two children (in this case, a daughter and son).

4. To show carriers of traits, the symbol may have a dot.

 Female carrier  Male carrier

It is important to note, however, that carriers' symbols are not always dotted and may appear blank.

5. To show which individuals show a particular trait, an individual's symbol is shaded and this information is shown in a key next to the pedigree chart.

 Female with trait  Male with trait
 Female without trait  Male without trait

If you refer to the ABO gene as I , then the allele that codes for:

- antigen A could be referred to as I^A
- antigen B could be referred to as I^B
- neither antigen could be referred to as i .

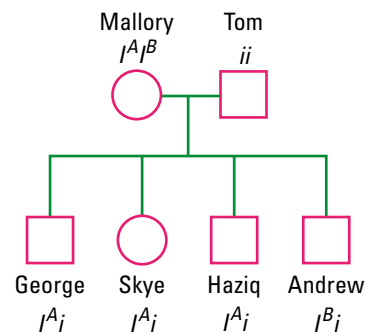
The ability to make antigen A or B is shown as a capital letter because it is dominant to the inability to make either antigen (which is recessive and shown as a lowercase letter).

Genotype and phenotype of blood groups

Genotype	Phenotype
$I^A I^A$	Blood type A
$I^A i$	Blood type A
$I^B I^B$	Blood type B
$I^B i$	Blood type B
$I^A I^B$	Blood type AB
ii	Blood type O

Family blood

Can you have a blood type different from both of your parents? The answer is yes! The pedigree chart below shows Tom (blood type O) and his wife Mallory (blood type AB) and their four children. A Punnett square can be used to predict the blood types that are possible for their children. This calculates that each child has a 50 per cent chance of inheriting blood type A or blood type B — blood types that neither parent possesses. What blood types do their children show in the pedigree chart below? Can you suggest why $\frac{3}{4}$ of the children have blood type A, when their chance of inheriting it was $\frac{1}{2}$?



Punnett square for $I^A I^B \times ii$

I^A = allele for blood type A

I^B = allele for blood type B

i = allele for blood type O

Possible gametes	I^A	I^B
i	$I^A i$	$I^B i$
i	$I^A i$	$I^B i$

Offspring probabilities

Genotype: $\frac{1}{2} I^A i : \frac{1}{2} I^B i$

Phenotype: $\frac{1}{2}$ blood type A: $\frac{1}{2}$ blood type B

Depending on their inheritance of particular alleles, children can have different blood types from their parents.

Sex-linked inheritance

The genes that have been considered so far have been those on autosomes. These types of genes show **autosomal inheritance**. The genes for some traits, however, are located on sex chromosomes. These traits are referred to as being sex-linked, and the type of inheritance is called X-linked if they are located on the X chromosome and Y-linked if they are located on the Y chromosome. Because of the small size of the Y chromosome it does not contain many genes, and most examples of sex-linkage that you will come across will be those of X-linkage.

The X-files

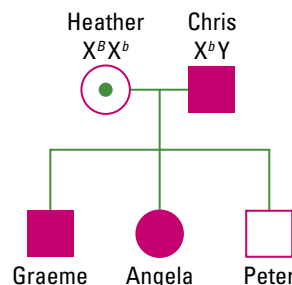
Haemophilia and some forms of colour blindness are examples of X-linked recessive traits. This means that females need to receive two alleles for the recessive trait, whereas males need to receive only one. This is why there is a greater chance of males showing these traits than females.

The genotype for X-linked traits includes the sex chromosomes in its description. For example, females may be heterozygous, $X^B X^b$, or homozygous, $X^b X^b$ or $X^B X^B$. Males, who possess only one X chromosome, are hemizygous and would have the genotypes $X^B Y$ or $X^b Y$. When stating the phenotypes for X-linked traits it is important to also specify the person's gender (e.g. colour blind male).

Colour blindness

In the pedigree chart above right, Chris is colour blind and, although Heather carries the colour blindness allele, she also has the allele for normal vision and so does not show this X-linked recessive trait. The Punnett square shows the probabilities of their children

inheriting colour blindness. Which children are colour blind? What were their chances of inheriting colour blindness?



Key



= Colour blind female



= Colour blind male

Being colour blind is an X-linked recessive trait.

Punnett square $X^B X^b \times X^b Y$

X^B = allele for normal vision

X^b = allele for colour blindness

Possible gametes	X^B	X^b
X^b	$X^B X^b$	$X^b X^b$
Y	$X^B Y$	$X^b Y$

Offspring probabilities

Genotype: $\frac{1}{4} X^B X^b$: $\frac{1}{4} X^b X^b$: $\frac{1}{4} X^B Y$: $\frac{1}{4} X^b Y$

Phenotype: $\frac{1}{4}$ normal vision female: $\frac{1}{4}$ colour blind female:
 $\frac{1}{4}$ normal vision male: $\frac{1}{4}$ colour blind male

When writing out the phenotype of an X-linked trait, it is important to also show the gender of the individual.

UNDERSTANDING AND INQUIRING

REMEMBER

- Describe the function of a Punnett square.
- Provide an example of a Punnett square.
- Outline the differences between the symbols used to identify the alleles in dominant/recessive inheritance, codominance and sex-linked inheritance.
- Describe the function of a pedigree chart.
- In a pedigree chart, what do the circles and squares represent?
- With regard to human blood type inheritance, identify:
 - the gene involved

- four possible blood types
- three possible alleles
- the type of inheritance
- the genotype of an individual with blood type O
- the genotype of an individual with blood type AB
- the phenotype of an individual with genotype $I^A I^A$
- the phenotype of an individual with genotype $I^B i$.

7 Distinguish between:

- autosomal inheritance and sex-linked inheritance
- X-linked and Y-linked inheritance
- X-linked recessive and X-linked dominant traits.

8 Suggest why males have a greater chance of showing an X-linked recessive trait than females.

ANALYSE, THINK AND DISCUSS

- 9 Predict the probabilities of the phenotypes and genotypes of the offspring of:
- a homozygous brown-eyed parent and a blue-eyed parent
 - two parents heterozygous for brown eyes.

Punnett square for $BB \times bb$

B = allele for brown eyes
 b = allele for blue eyes

	B	B
b		
b		

Offspring probabilities

Genotype:

Phenotype:

Punnett square for $Bb \times Bb$

B = allele for brown eyes
 b = allele for blue eyes

	B	b
B		
b		

Offspring probabilities

Genotype:

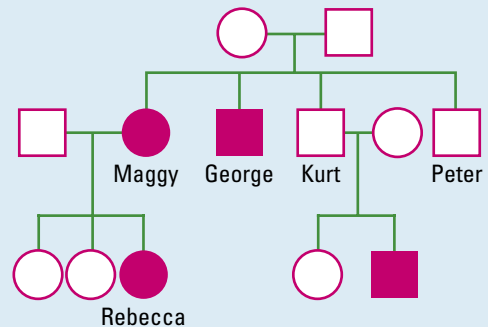
Phenotype:

- 10 Refer to Chris and Heather's family pedigree chart and information on their inheritance of colour blindness on page 40.
- State the genotype for:
 - Chris
 - Heather.
 - State the phenotype for:
 - Chris
 - Heather.
 - What is the chance of:
 - Graeme having colour blindness
 - Peter having colour blindness
 - Angela having colour blindness?
 - Is it possible for Peter to have a child who is colour blind? Explain.
- 11 State the genotype of the following individuals.
- Heterozygous for blood type A
 - Homozygous for blood type B
 - Blood type O
 - Blood type AB
- 12 If a man who was homozygous for blood type A ($I^A I^A$) had a child with a woman who had blood type O (ii), what is the chance that the child would have:
- blood type A
 - blood type B
 - blood type O?
- 13 If a child had blood type AB, suggest the possible combinations of genotypes of the parents.
- 14 Determine the chance of a couple with blood types AB and A having a child with:
- blood type A
 - blood type B
 - blood type AB
 - blood type O.

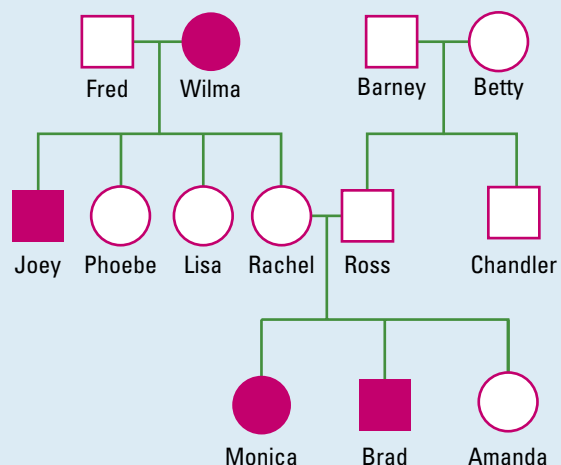
- 15 Can a father with blood type A and a mother with blood type B have a child with blood type O? Explain.

- 16 What is the chance of Linda and Geoff, described on page 37, producing a child with the homozygous dominant combination (BB)?

- 17 Refer to the pedigree of the family in the diagram below. The inheritance of broad lips (B ; unshaded individuals) is dominant to the inheritance of thin lips (b ; shaded individuals).
- How many females are shown in the pedigree chart?
 - How many males are shown in the pedigree chart?
 - How many females have the thin lips trait?
 - Suggest the genotype of Maggy's parents.
 - Suggest how Maggy inherited thin lips, when her parents did not.
 - Suggest the genotypes of (i) Peter, (ii) Kurt, (iii) George and (iv) Rebecca.



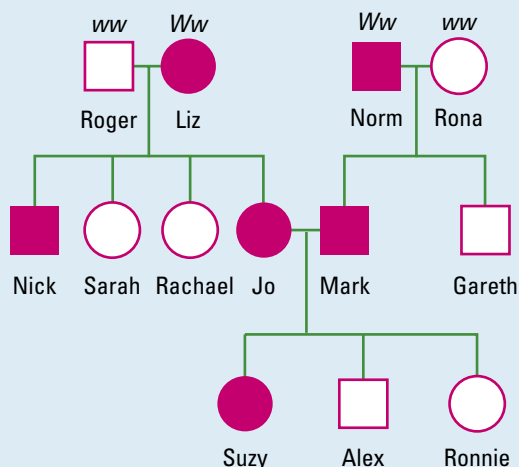
- 18 The pedigree below traces the recessive trait of albinism in a family. The shaded individuals lack pigmentation and are described as being albinos.



- List any observations from the pedigree that support albinism being a recessive trait.

- (b) If the albinism allele was represented as n and normal skin pigmentation as N , state the possible genotypes for each of the individuals in the pedigree.

19 The pedigree below traces the dominant trait, a widow's peak, in a family.



- List any observations from the pedigree that support the widow's peak being a dominant trait.
- If the widow's peak allele is represented as W and the straight hairline as w , state the possible genotypes for each of the individuals in the pedigree.
- If Jo and Mark were to have another child, what is the chance of it having a widow's peak?
- If Ronnie were to have a child with a man who did not have a widow's peak, what is the probability that their child would have a widow's peak?
- If Norm and Rona were to have another child, what is the probability that they would have a child without a widow's peak?

20 Use the dominant and recessive table below and Punnett squares to assist you in answering the following questions.

Dominant trait	Recessive trait
Free ear lobes	Attached ear lobes
Mid-digital hair present	Mid-digital hair absent
Normal skin pigmentation	Pigmentation lacking (albinism)
Non-red hair	Red hair
Rhesus-positive (Rh +ve) blood	Rhesus-negative (Rh -ve) blood
Dwarf stature (achondroplasia)	Average stature
Widow's peak	Straight hairline

- Find the probability of Sally (who is homozygous for dwarf stature) and Tom (who has average stature) having a child with dwarf stature.
- Find the probability of Fred (who is heterozygous for dwarf stature) and Susy (who has average stature) having a child with dwarf stature.
- What is the chance of two parents who are both heterozygous for free ear lobes having a child with attached ear lobes?
- Michael is heterozygous for mid-digital hair, whereas Debbie does not have mid-digital hair. What is the chance of their children having mid-digital hair?

INVESTIGATE, THINK AND DISCUSS

- What are some physical attributes of males that suggest sexual potency and good genes?
- Suggest what the major histocompatibility complex has to do with mate selection.
- What is sexual selection? Give two examples.
 - How is sexual selection different from natural selection?
 - Suggest implications of sexual selection for our species.
 - Suggest the possible impact of sexual selection on your future reproductive life.
- While the science of love is still in its infancy, advances in molecular biology and technology have increasingly allowed us to peer through its window.
 - Find out examples of research on the chemistry of love or love potions.
 - Do you believe that this research should be continued? Give reasons.
 - Suggest possible issues that may arise with the knowledge obtained and its possible applications.
 - Discuss if, how and who should regulate or control this type of research.
- Increasing numbers of people are finding love and their partners on the internet.
 - What is your opinion on this?
 - Discuss your opinion with others in your team.
 - Discuss the use of internet dating from biological, cultural, social and ethical viewpoints, and construct a PMI chart to summarise your discussion.
- Use the **Careers** weblink in your eBookPLUS and choose one of the scientists profiled. Assess whether you would like to do this person's job when you are older. In your answer include a brief description of the type of work involved and some reasons why you may or may not want to do this job.

eBook plus

work
sheet

2.7

Pedigrees

Changing the code

Errors or changes in DNA, genes or chromosomes can have a variety of consequences. These genetic mistakes are called **mutations**.



Polydactyly (having more than 10 fingers and toes) is usually due to a DNA mutation.

DNA replication

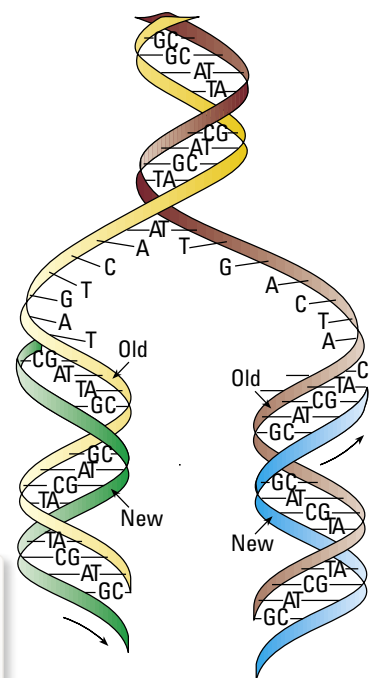
DNA is very stable and can be replicated into exact copies of itself. This process is called **DNA replication** and enables genetic material to be passed on unchanged from one generation to the next. DNA replication begins with the 'unzipping' of the paired strands. A new complementary DNA strand is made for each original DNA strand. This results in the formation of two new double-stranded DNA molecules, each containing one new DNA strand and one original DNA strand. This model of DNA replication is called the **semi-conservative model** because it has conserved one of the old DNA strands in each new double-stranded DNA molecule.

The process of DNA replication has a number of checkpoints to test for any mistakes that may be made, so that they can be corrected or destroyed. Sometimes, however, the mistakes get through this screening process. When this happens, we say that a mutation has occurred.

Mutagenic agents

Mutations can happen by chance or have a particular cause. When the cause of the mutation cannot be identified it is called a **spontaneous mutation**, and when the cause can be identified it is referred to as an **induced mutation**.

A factor that triggers mutations in cells is called a **mutagen** or **mutagenic agent**. Examples of mutagenic



Arrows denote direction of synthesis.

DNA replication is semi-conservative. The new DNA has one old and one new DNA strand.

agents include radiation (e.g. ultraviolet radiation, nuclear radiation and X-rays) and some chemical substances such as formalin and benzene (which used to be common in pesticides).

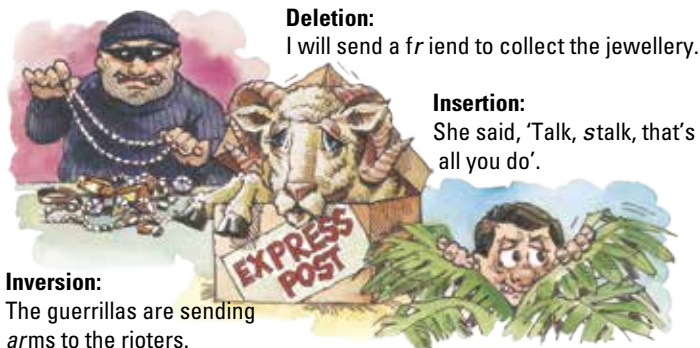
As a result of the thinning of the ozone layer in the atmosphere, we are exposed to increasing amounts of **UVB radiation** that can damage (or mutate) our DNA. This can lead to the development of skin cancers. Protective clothing and sunscreens can help reduce our exposure to this dangerous, potentially mutagenic environmental radiation.

Errors in the code

Many important hormones and enzymes are made up of protein. Changes in the genetic code due to mutations may result in a particular protein not being made or a faulty version being produced. In some cases, the production of an essential enzyme may be impaired, disrupting chemical reactions and resulting in the deficiencies or accumulation of other substances. This may cause the death of the cell and, eventually, the organism.

Point mutations

Occasionally, errors can occur during DNA replication as DNA is being copied. This means that the instructions carried by the code are not followed exactly. This may be the result of an incorrect pairing of bases; the **substitution** of a different nucleotide; or the **deletion** or **insertion** of a nucleotide. Such a **point mutation** can change the genetic message by



Deletion:

I will send a friend to collect the jewellery.

Insertion:

She said, 'Talk, stalk, that's all you do'.

Inversion:

The guerrillas are sending arms to the rioters.

Just like changing letters in a word can change its meaning, changes in the DNA sequence can change the meaning of the genetic code.

coding for a different amino acid, leading to the production of a different or non-functional protein. This can have consequences to the phenotype of the organism.

Sickle-cell anaemia

Sickle-cell anaemia is a disease that is usually associated with a mutation in the gene that codes for one of the polypeptides that make up **haemoglobin** in red blood cells. In this mutation, an adenine base is substituted by a thymine base. The result is a phenotype of misshapen red blood cells that can clump together and block blood vessels.

Chromosome mutations

Point mutations relate to changes in the genetic information in genes; however, mutations can also involve chromosomes. These may involve the addition or deletion of entire chromosomes, or the deletion, addition or mixing of genetic information from segments of chromosomes. Some examples of disorders that result from chromosome mutations are shown in the table at the top of the following page.

eBookplus

eLesson

DNA and Hiroshima

Watch a video from *The story of science* about DNA.

eles-1781

Mutants unite!

Not all mutations are harmful. Some mutations can increase the survival chances of individuals within a population, and hence the survival of their species.

Spray resistance

Pesticides kill the majority of insects sprayed. Some insects within the population, however, may survive because they possess slight variations or mutations in their genes that give them **resistance** to the pesticide. The mutated gene in the surviving insects is passed on to their offspring, who gain that resistance too. While the insects without the resistance die out, those with resistance increase in numbers.

Good for you, but not for me

When we look at natural selection as a mechanism for evolution in chapter 3, we see how mutations can be a very important source of new genetic material. While such mutations can be beneficial for the survival of the species under threat, they are not necessarily beneficial to humans. The resistance of bacteria to antibiotics, for example, has resulted in selection for antibiotic-resistant bacteria. This means we are unable to use these antibiotics to treat diseases caused by these resistant bacteria, as the drugs are no longer effective.

Malaria and sickle-cell mutation

Malaria is a disease that is very common in many parts of Africa, Asia and South America. It is caused by a parasite that uses a species of mosquito as a vector and grows in red blood cells of its human host. This

	Normal red blood cell	Sickle-cell red blood cell
DNA sequence	GAC TGA GGA CTC	GAC TGA GGA CAC
Complementary RNA sequence	CUG ACU CCU GAG	CUG ACU CCU GUG
Amino acid sequence	leu — thr — pro — glu	leu — thr — pro — val
Phenotype of red blood cell	Normal doughnut-shaped blood cell 	Sickle-shaped blood cell 

Examples of human chromosome abnormalities (mutations)

Chromosome abnormality	Resulting disorder	Incidence (per live births)
Extra chromosome number 21	Down syndrome	1 in 700; risk rises with increase in maternal age
Missing sex chromosome (XO)	Turner syndrome	1 in 5000 (90% of these conceptions are aborted)
Extra sex chromosome (XXY)	Klinefelter syndrome	1 in 1000; risk rises with increase in maternal age

disease is one of the main global causes of human disease-related deaths.

The mutation that results in sickle-cell anaemia can increase your resistance to malaria. If you are heterozygous for this trait (you have one copy of the sickle-cell allele), the parasite cannot grow as effectively in your red blood cells; hence you are less likely to die from malaria than people in the population without the allele. This is an example of what is known as **heterozygote advantage**.

Not all mutations are inherited

Only mutations that have occurred in the germline cells such as the sex cells or gametes (sperm and ova) are inherited. In sexually reproducing organisms,

mutations that occur in somatic cells are not passed on to the next generation.



Mutations can be caused by chemicals in your environment and may result in cancerous growths (tumours) within your body show as foggy areas here.

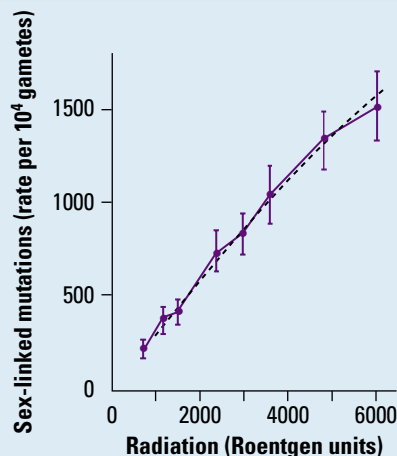
UNDERSTANDING AND INQUIRING

REMEMBER

- 1 Name the process by which DNA makes copies of itself.
- 2 Explain why the model used to describe the process identified in question 1 is called semi-conservative. Include a diagram in your response.
- 3 Describe what is meant by the term *mutagenic agent*. Provide an example.
- 4 Distinguish between the terms *spontaneous mutation* and *induced mutation*.
- 5 Suggest the relationship between the thinning of the ozone layer and the increased incidence of skin cancer.
- 6 Outline the relationship between sickle-cell anaemia and mutated DNA.
- 7 Identify two disorders associated with chromosome mutations.
- 8 Are mutations always detrimental? Provide an example to justify your response.

INVESTIGATE, THINK AND DISCUSS

- 9 Suggest why radiographers wear special protective clothing and use remote controls for taking X-rays.
- 10 Suggest mutations that increase chances of survival.
- 11 Examine the graph above right.
 - (a) Describe the pattern or trend. Incorporate the axis labels in your description.
 - (b) Suggest an interpretation of the data.



ANALYSE, INTERPRET AND THINK

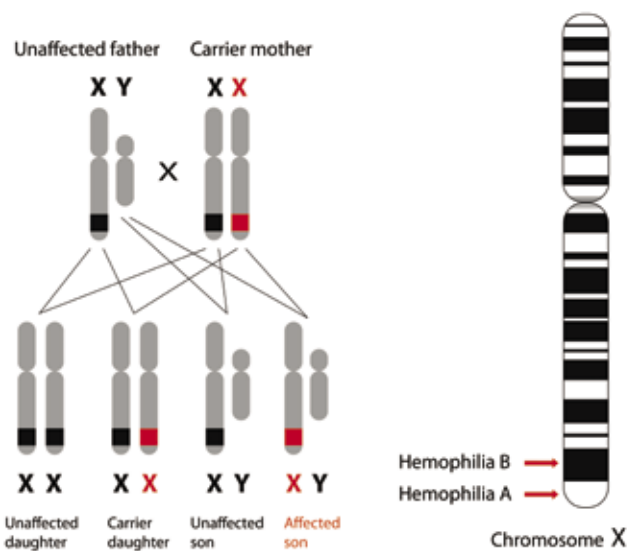
- 12 Use the **Down syndrome** weblink in your eBookPLUS to read an article about Down syndrome research. Use your own knowledge and information in the article to answer the following questions.
 - (a) How many chromosome 21 copies are in the somatic cells of a person with Down syndrome?
 - (b) Is this the same number of chromosome 21 copies that are in the somatic cells of a person who doesn't have Down syndrome? Explain.
 - (c) Suggest why the *DSCR1* gene is of importance.
 - (d) On which chromosome is the *DSCR1* gene located?
 - (e) Outline the advantage suggested by the research of possessing an extra copy of the *DSCR1* gene.

eBookplus

Predicting with pedigree charts

You are the combined result of your parents' gametes and your environment. If someone's sperm or ovum carries a DNA abnormality, there is a chance that their child will be affected. Inherited gene and chromosome abnormalities may result in genetic disorders. These can be slight, such as red-green colour blindness, or more severe, such as haemophilia, a disorder in which the blood does not clot.

The image below shows a potential inheritance pattern for haemophilia. Haemophilia is a rare genetic disorder characterised by an inability for blood to clot, meaning even minor cuts and bruises can be life threatening. It is passed on by recessive X-linked inheritance, which means that a mother may carry the disease without knowing and may pass it on to some of her sons.

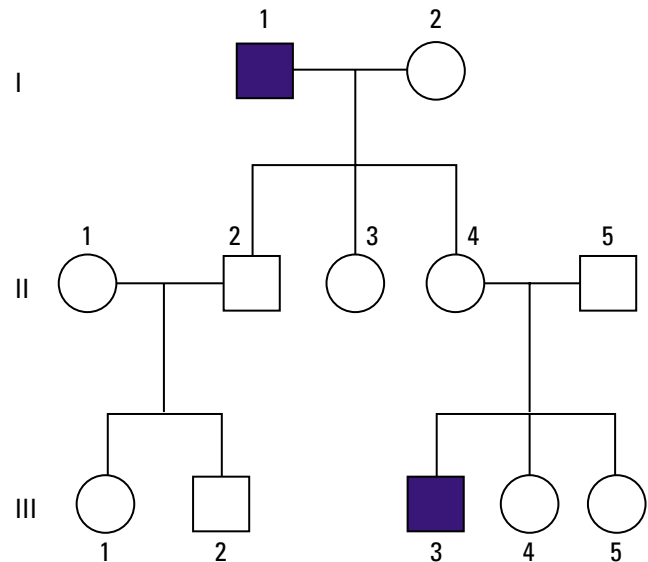


Inheritance of haemophilia

Who's who in a pedigree chart?

Pedigree charts can be used to observe patterns and to predict the inheritance of traits within families. Patterns in the inheritance of these traits can also show whether the trait is dominant or recessive and whether it is carried on an autosome or sex chromosome.

The pedigree chart above right shows how individuals and generations can be identified so that interpretation of patterns can be more effectively communicated. The shaded individual at the top of the chart is identified as I-1 (individual 1 in the first generation) and the shaded individual in the



Pedigree charts can show patterns of inheritance in families and enable identification of individuals within the family. Which individual do you think could be described as II-3?

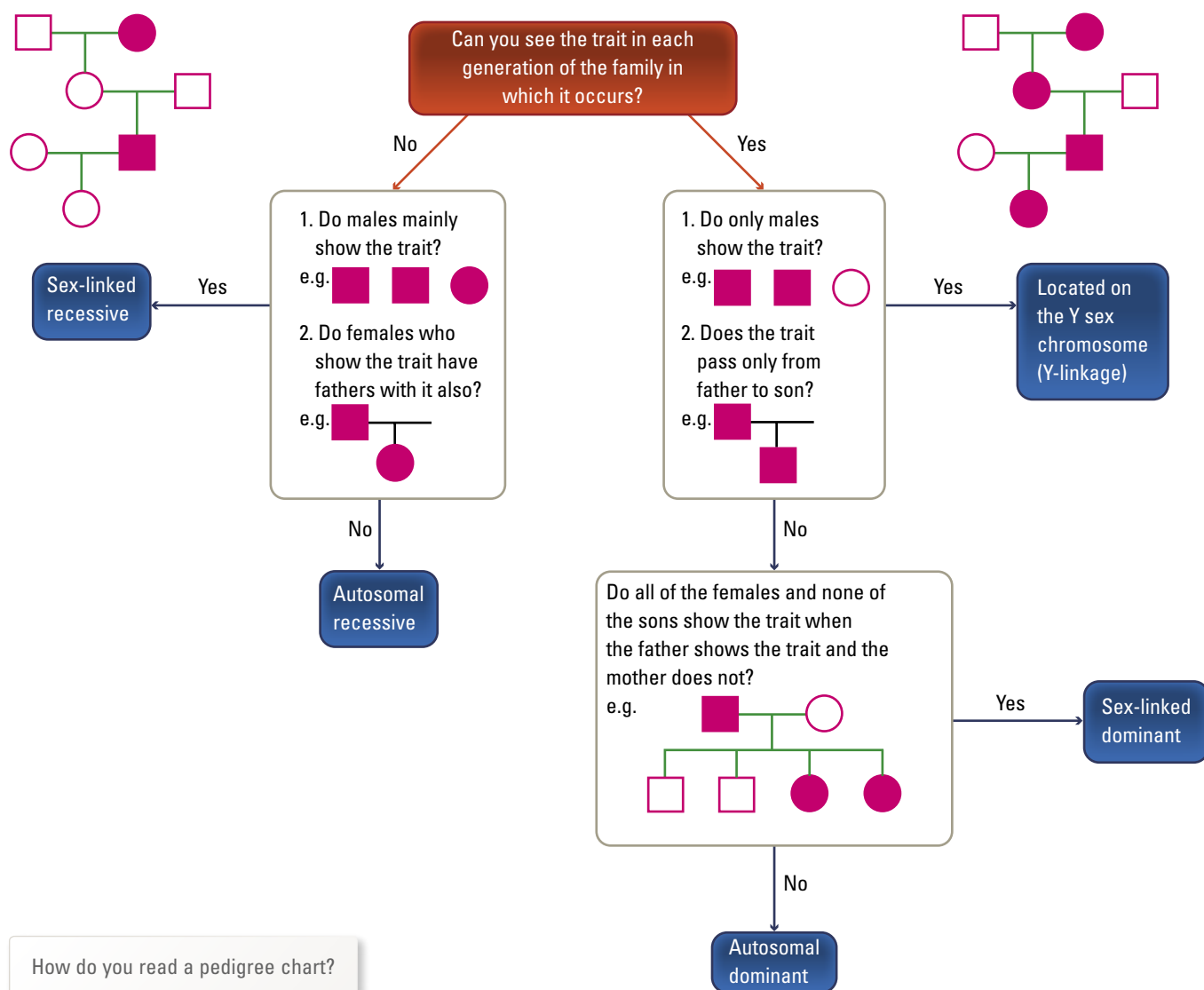
bottom row is identified as III-3 (individual 3 in the third generation). The daughters of individual I-1 are identified as II-3 and II-4.

Naming inheritance

Within the nucleus of each human somatic (body) cell are 46 chromosomes. There are two sex chromosomes (either XX or XY) and 22 pairs of autosomes. The autosomes are numbered, based on their size and shape, from 1 to 22.

The inheritance of various traits can be described in terms of the location of the gene responsible and whether the inheritance is dominant or recessive. For traits located on the sex chromosomes, the trait is considered to be sex-linked. For traits located on the autosomes, the trait is considered to be autosomal.

A trait that is inherited recessively and caused by a gene on an autosome (e.g. chromosome 21) is described as **autosomal recessive**. Likewise, a trait located on the X chromosome and inherited recessively is described as **X-linked recessive**. The table at the bottom of the following page provides examples of some inherited diseases and how they can be inherited.



Some diseases that can be inherited

Inherited disorder	Type of inheritance	Symptoms of disorder
Fragile X syndrome (FRAX)	Sex-linked	Leading cause of inherited mental retardation
Haemophilia A and B (HEMA, HEMB)	Sex-linked recessive	Bleeding disorders
Huntington's disease (HD)	Autosomal dominant	Usually mid-life onset; progressive, lethal degenerative neurological disease
Intestinal polyposis	Autosomal dominant	Many small bulges in the colon form; may lead to colon cancer
Dwarfism	Autosomal dominant	Inhibited growth
Sickle-cell disease	Autosomal recessive	Red blood cells become deformed into a sickle shape when oxygen levels are low, which can lead to impaired mental function, paralysis and organ damage
Thalassaemia (THAL)	Autosomal recessive	Reduced red blood cell levels

Cystic fibrosis

About 1 in 2500 people suffer from an autosomal recessive genetic disorder called cystic fibrosis (CF). The CF allele is located on chromosome number 7. One amino acid in a chain of 1480 amino acids is not produced, causing a faulty protein to be synthesised. This results in the production of large amounts of thick mucus by cells lining the lungs and in the pancreas where digestive juices are secreted. The mucus interferes with the working of the respiratory and digestive systems. Infection readily occurs and sufferers tend to have a shortened life span. Pedigree analysis can show the likelihood of a child suffering from cystic fibrosis.

Checking to see if you are a CF carrier

Since the identification of the defective allele in 1989, the DNA of parents-to-be can be analysed to find out if they are one of the 1 in 25 people who carry the allele. This is useful information because, although they may not have cystic fibrosis themselves, they may be a carrier. This means that there is a chance they will have a child with cystic fibrosis. For example, if both parents are carriers, there is a 1 in 4 chance that they will have a child with cystic fibrosis.

A chance event?

Genetic counselling can help parents-to-be who are both carriers of the CF allele with their decision about whether to have a child. If they decide to go ahead, genetic tests can be used to determine the genotype

of the embryo. If both parents are heterozygous for cystic fibrosis, each child they have has a 25 per cent chance of having two CF alleles, and hence having cystic fibrosis, and a 75 per cent chance of not having the disease. It is important to note that the chance is independent for each child. If the parents already had one child with cystic fibrosis, the next child would still have a 25 per cent chance of also having it. There is a 75 per cent chance that the child will not have the disorder, but only a third of these children will not have the CF allele. There is a 50 per cent chance that the child will not have the disorder but will be a carrier with one CF allele. You can see how these chances are calculated in the Punnett square below.

If the genetic test shows that the child will have cystic fibrosis, the parents need to make an important decision — will they keep the baby? Genetic counselling may also help the parents with this very difficult decision. What would you do? If it was a more severe genetic disease, would that change your response?

Punnett square for $Nn \times Nn$

N = normal allele

n = cystic fibrosis allele

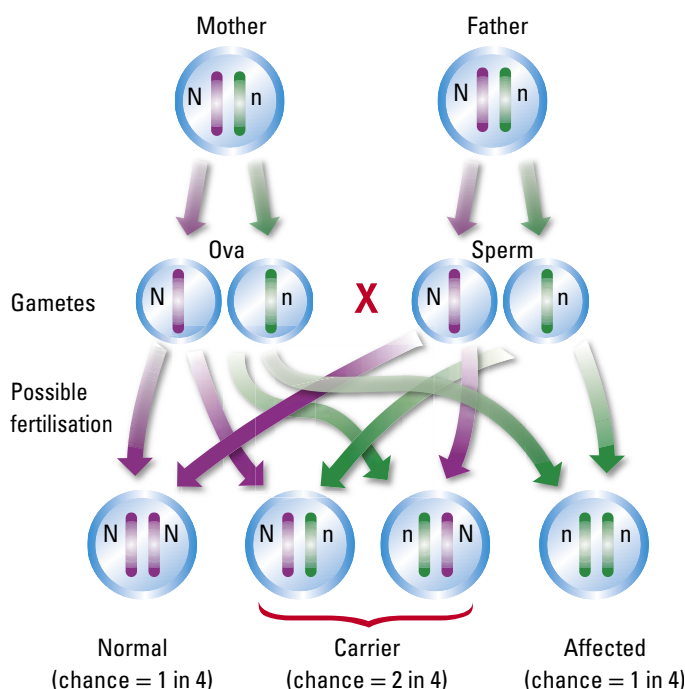
	N	n
N	NN	Nn
n	Nn	nn

Offspring probabilities

Genotype: $\frac{1}{4} NN$: $\frac{1}{2} Nn$: $\frac{1}{4} nn$

Phenotype: $\frac{3}{4}$ normal: $\frac{1}{4}$ cystic fibrosis

If these two heterozygous parents had five children, would it be possible for none of them to have the disease?



HOW ABOUT THAT!

Many genes, such as those controlling the production of enzymes necessary for respiration, are active throughout the life span of a person. Some are switched on only at particular times and in specific tissues. This regulates development. Late onset genetic disorders, such as Huntington's disease and a form of Alzheimer's disease, result from particular defective genes becoming activated later in life. Duchenne muscular dystrophy or muscle deterioration is another disease that gradually develops from late childhood.

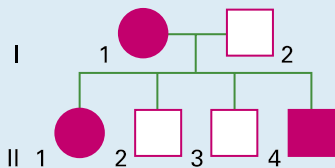
UNDERSTANDING AND INQUIRING

REMEMBER

- State the type of inheritance by which haemophilia is passed between generations.
- Outline two uses of pedigree charts.
- Outline how you could describe the identity of individuals with the following notation in a pedigree chart.
(a) I-1 (b) III-3 (c) II-4
- State the key difference between autosomal inheritance and sex-linked inheritance.
- Identify an inherited disorder that is:
(a) X-linked recessive
(b) X-linked dominant
(c) autosomal dominant
(d) autosomal recessive.
- On which chromosome is the cystic fibrosis allele located?
- What are the symptoms of cystic fibrosis and what causes these symptoms?
- Suggest why people may be tested for the cystic fibrosis allele.
- Suggest how genetic counselling can be helpful in decision making.
- What are the chances of two CF carrier parents having a child:
(a) who has cystic fibrosis
(b) who does not have cystic fibrosis
(c) who is a carrier for cystic fibrosis
(d) who does not have cystic fibrosis and is not a carrier?

ANALYSE, INTERPRET AND THINK

- Jacob has hypertrichosis or werewolf syndrome, as does his mother. His father, however, does not. Hypertrichosis is an X-linked dominant trait that is characterised by increased hair growth on the face and upper body. Jacob is shown in the pedigree chart below as individual II-4.

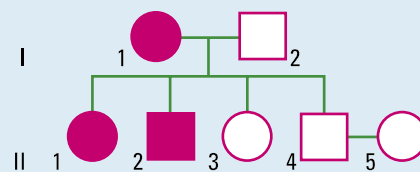


For the following questions, assume that X^H = hypertrichosis and X^h = normal hair growth.

- Use the Punnett square above right to determine the chances of Jacob's parents having children with the syndrome and state the chance of their having a:
(i) daughter with hypertrichosis
(ii) son with hypertrichosis
(iii) child with hypertrichosis.

	X^H	X^h
X^h	$X^H X^h$	$X^h X^h$
Y	$X^H Y$	$X^h Y$

- If Jacob had children with Bella (who does not have hypertrichosis), use a Punnett square to determine the chances of their child being:
(i) a daughter who has hypertrichosis
(ii) a son who has hypertrichosis
(iii) a daughter who does not have hypertrichosis
(iv) a son who does not have hypertrichosis.
 - Since Jacob is affected with hypertrichosis, do his sons and daughters have the same chance of inheriting the condition? Explain.
 - How does this compare to a father who has an autosomal recessive trait? If he shows the trait and his wife does not, what are the chances that his daughters will show the trait? Explain.
 - How does this compare to a father who has an autosomal dominant trait? If he shows the trait and his wife does not, what are the chances that his daughters will show the trait? Explain.
- Huntington's disease is an autosomal dominant condition. Refer to the diagram below to answer the following.
(a) If H = Huntington's disease and h = normal, state the genotype(s) and phenotypes of:
(i) I-1
(ii) I-2
(iii) II-1
(iv) II-4
(v) II-5.
(b) Use a Punnett square to predict the chances of:
(i) I-1 and I-2 having a child with Huntington's disease
(ii) II-4 and I-5 having a child with Huntington's disease.



- Queen Victoria was a carrier of the X-linked recessive trait haemophilia. This trait affects blood clotting. Use the figures at the bottom of the next page to answer the following.
(a) If X^H = normal trait and X^h = haemophilia, state the genotype of:
(i) Queen Victoria
(ii) her husband
(iii) her daughter Beatrice
(iv) her son Leopold (Duke of Albany)
(v) her granddaughter Alexandra
(vi) her great grandson Alexis.

- (b) If Queen Victoria and her husband had had another child, what was the chance that their child would have:
- had haemophilia
 - been a carrier for haemophilia?
- (c) If Alexandra and her husband, Tsar Nikolas II of Russia, had had another child, would they have had the same chance of having a haemophilic son as her mother and father? Explain.
- (d) Suggest why our current Queen Elizabeth doesn't have haemophilia and why none of her children are haemophiliacs.



Queen Victoria carried the allele for haemophilia on one of her X chromosomes. This germline mutation was passed on to other members in her family.

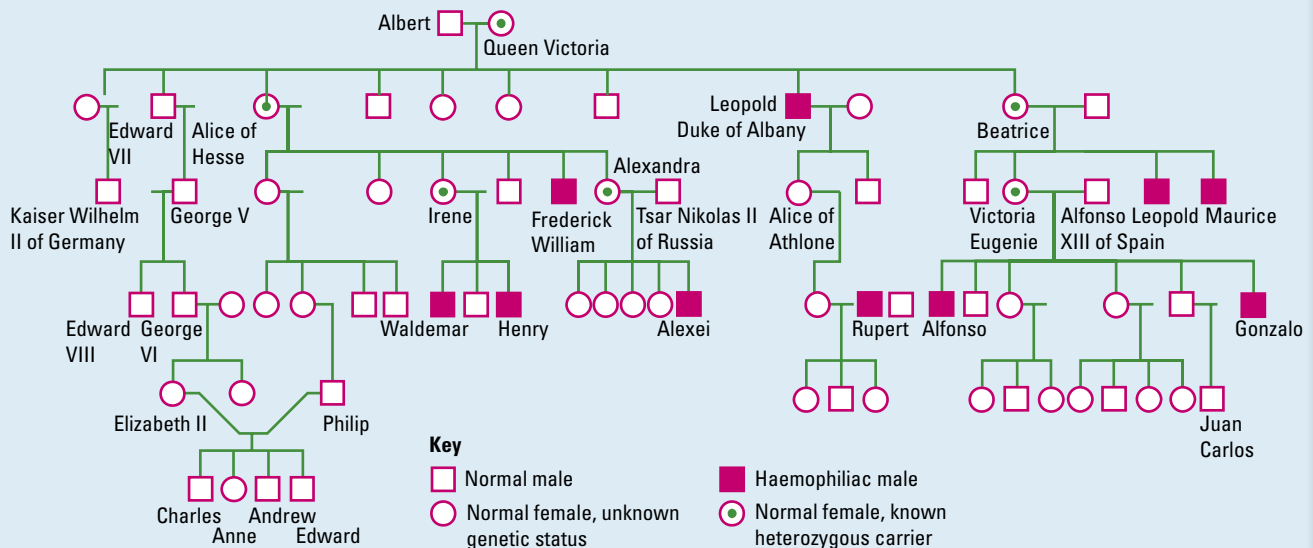
THINK, INVESTIGATE AND DISCUSS

- 14 (a) Find out what types of genetic testing occur in Australia.
- (b) Are there any laws, rules or regulations associated with genetic testing? If so, what are they?
- (c) List examples of different views and perspectives on genetic testing.
- (d) Suggest why there are differing views.
- (e) Construct a PMI chart on genetic testing.
- (f) What is your opinion on genetic testing?
- 15 In a group, make a list of human genetic disorders. Each person is to write a report on one. Your report could take the form of a poster or brochure.
- Include which gene or chromosomal abnormality is responsible for the disorder and some of the characteristics the affected person would show.
 - Find out whether there are organisations available to support people who have the disorder and their families.

CREATE

- 16 Create a rhyme, song or poem about pedigree analysis or the types of inheritance that effectively uses as many of the key terms in this section as possible. An example is given below.

Dominant traits always with an affected parent
While in recessive sometimes there aren't
X-linked dominant — dad to his daughters
No skipping seen, always loiters
X-linked recessive — mum to her sons
Can skip generations and hide in carrier ones.



Exposing your genes

Do you think that you can hide your identity? Maybe today you can, but that probably won't be the case in the future. DNA technology is rapidly providing techniques that will bring your deepest secrets out into the open.

How much do you want to know about your genes? How much do you need to know about the genes of a potential partner or members of your own family? Who should have access to your genetic information, and what should they be allowed to do with it? Who owns your genes?

examination of the DNA molecule itself. Other tests include biochemical tests for various gene products (for example, enzymes and other proteins) or the microscopic examination of stained or fluorescent chromosomes.

Over 6000 single-gene disorders have been identified. Many other inherited diseases are considered multifactorial because they may be caused by a combined effect of the interaction of a number of different genes with each other and the environment (for example, Alzheimer's disease, diabetes and asthma).

Why use genetic tests?

Genetic tests can provide information that can be used in gender determination; carrier screening for genetic mutations; or in the diagnosis, prediction or predisposition to particular genetic diseases or other inherited traits. These tests may be performed prenatally or on newborns, children or adults. Trying to control the characteristics of human populations by selective breeding or by genetic engineering is called **eugenics**.

Testing at birth

Australian state screening laboratories carry out a series of tests on a newborn baby's blood (see the table below). Early testing allows doctors to start any necessary treatment.

Screening tests

Genetic disorder	Symptoms	Incidence
Cystic fibrosis	Respiratory and digestive problems; shortened life expectancy	1 in 2500
Phenylketonuria (PKU)	Brain damage due to excessive levels of an amino acid in the blood	1 in 12 000
Hypothyroidism	Slowed growth and mental development owing to a poorly developed thyroid gland	1 in 3400

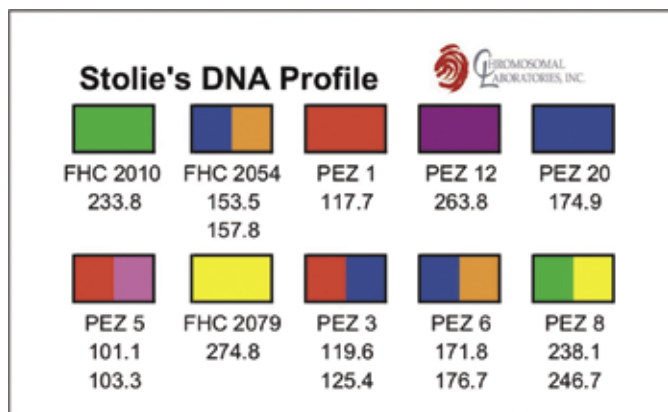
Doggie Licence

Stolie Bolin
4556 N. Canine Ln.
Wagtail, VA 12896

Breed St. Bernard
Color White/Brown/Black
Sex Female
Weight 118

Birthdate 9/16/03



Already companies around the world are offering DNA profiling. Will you soon be required to carry your DNA profile around with you — and show it on request?

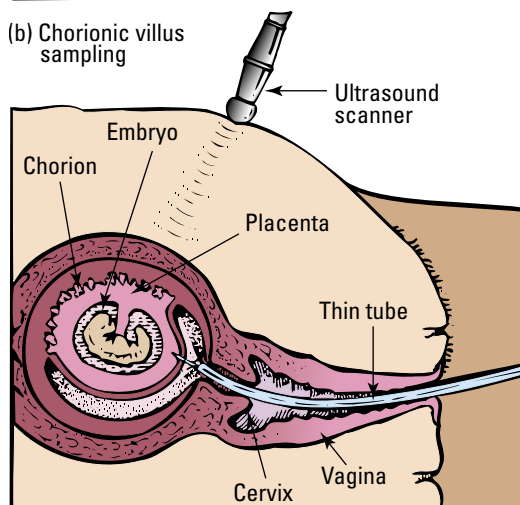
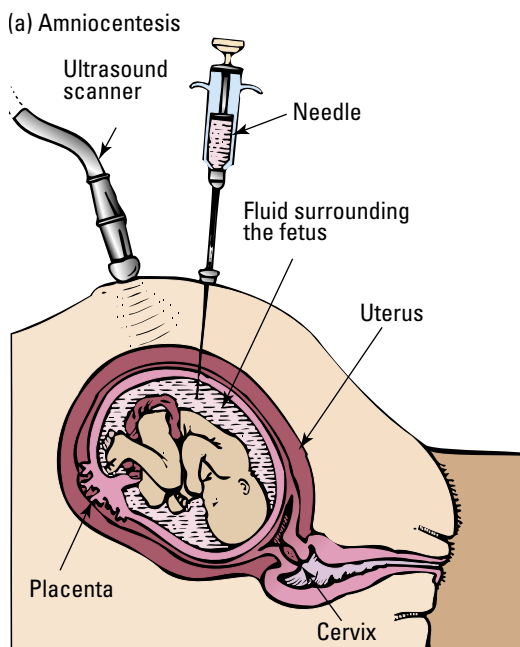
Genetic tests

There are various tests that can be used to find out about your genetic information. Among these are gene tests and DNA-based tests that involve direct

Counting chromosomes

The presence of chromosomal abnormalities such as Down syndrome can be determined by analysing cells of the developing fetus. These cells can be obtained by a technique called chorionic villus sampling (CVS), which collects actual cells of a fetus that is 10–12 weeks old. Another technique called amniocentesis can be used to collect samples of fluid from the uterus that contains cells shed by a fetus that is 14–16 weeks old.

These techniques can also be used to obtain cells that can be analysed for the presence of particular alleles. However, both of these techniques of cell sample collection are accompanied by some risk of miscarriage or damage to the fetus.



Amniocentesis and chorionic villus sampling can be used in the identification of chromosomal abnormalities.

Counting DNA stutters

Did you know that the function of a large percentage of your DNA is still unknown? These supposedly non-functional or non-coding parts vary in length and can consist of patterns of repetitive base sequences called **microsatellites**.

DNA fingerprints

Patterns of variations in these repeated base sequences form the basis for **DNA fingerprinting**. This technique produces a kind of barcode of the natural variations found in every person's DNA. It is this barcode or DNA fingerprint that enables the identification of an individual to be made.



DNA fingerprinting is based on variations in the patterns of repeating base sequences in DNA between individuals.

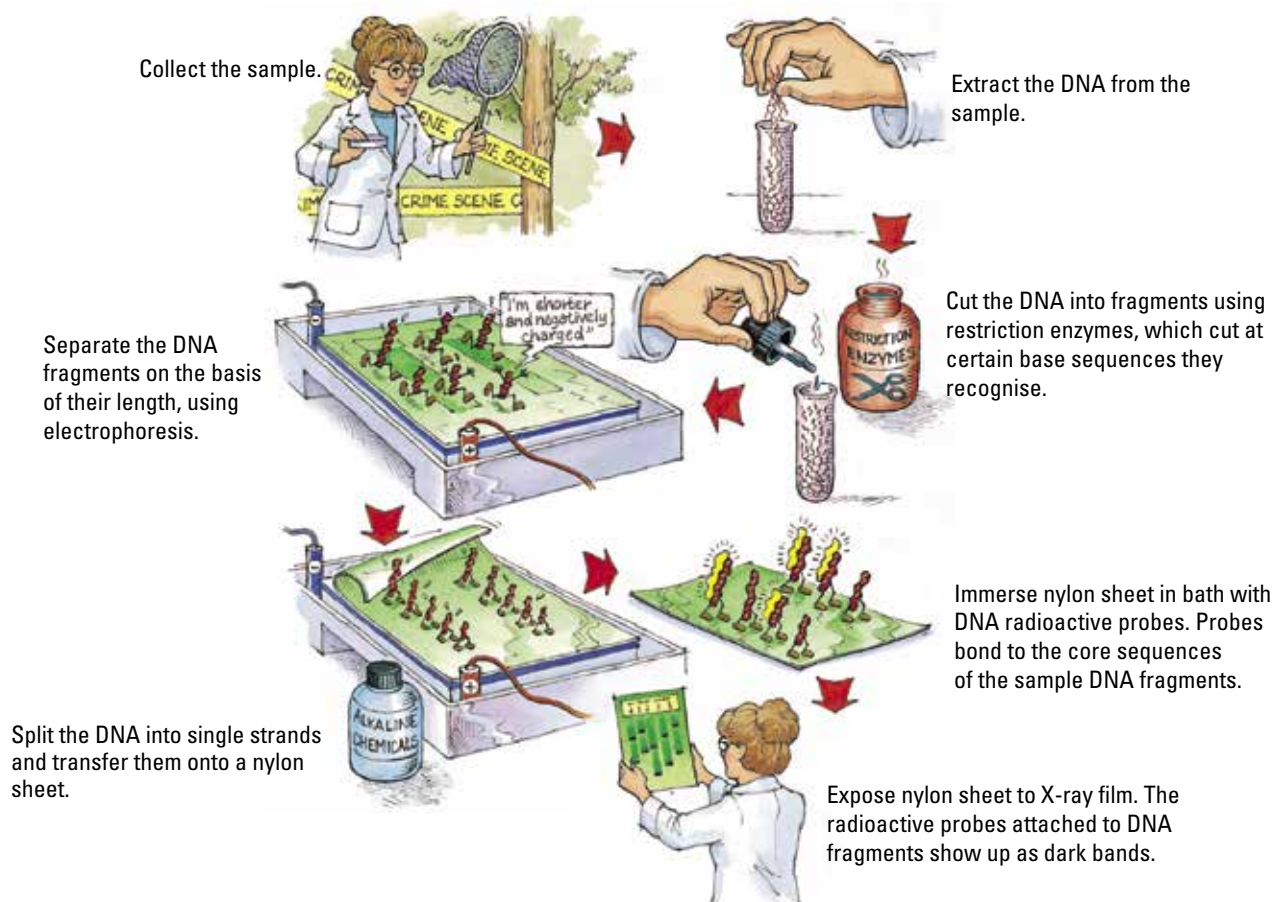
DNA fingerprinting involves analysing DNA fragments. After the extraction of DNA fragments from biological material, **restriction enzymes** are used to cut the DNA into specific fragment lengths. The technique of **electrophoresis** is used to separate the fragments on the basis of their size and charge. DNA probes are then used so that the DNA patterns can be observed.

What's the use of DNA fingerprints?

DNA fingerprints can be useful in forensic investigations, paternity tests and evolutionary studies (to determine the relatedness of different organisms), and to search for the presence of a particular gene.

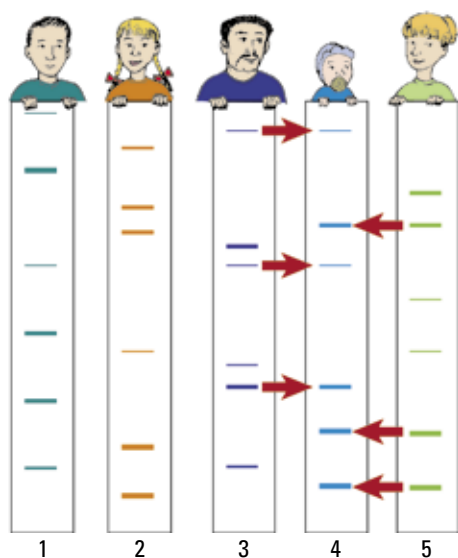
Fast computers and statistical genetics

Statistical methods have been used to establish gene linkage and estimate recombination fractions (due to crossing over in meiosis) since the 1930s. British scientists Bell and Haldane were the first to establish linkage between haemophilia and colour



DNA fingerprinting uses a variety of different genetic engineering tools and techniques.

blindness with X-linked genes in 1937, and Mohr found linkage between blood group types on an autosome in 1954.



Who are the parents of individual 4? Are persons 1 and 2 related?

It was not until around 1980 that DNA sequence differences were used as **molecular markers**. The combination of these new markers with the use of **restriction fragment length polymorphisms (RFLPs)**, new multilocus mapping methods, suitable algorithms, and the affordability and availability of fast computers revolutionised human genetic mapping. In the late 1980s, the **polymerase chain reaction (PCR)** technique was beginning to revolutionise **molecular genetics**. This technique enabled amplification of small amounts of DNA, increasing the amount and hence the depth to which it could be studied. During this time, new types of genetic markers (such as RAPD, STRP and SSCP) were developed using PCR. The increase in the number of markers available enabled genome-wide scans to be performed. These scans could search the entire genome for linkage between a trait and markers, identifying the location of genes that contributed to a wide range of phenotypes — including those associated with inherited diseases.

More recent research has focused on **single nucleotide polymorphisms (SNPs)** in the human genome. A current map of these in our genome contains more than

10 million SNPs. These SNP markers can be used in **genotyping** — a process that determines the alleles at various SNP markers within the human genome. Current technologies enable the genotyping of around 1 million SNPs per person within 24 hours for a cost of around \$1000!

Linkage analysis

A team of scientists at the Walter and Eliza Hall Institute in Melbourne are using statistical models and fast computers to identify possible locations of particular genes within genomes. Information from families in the investigation is collected so that pedigrees can be constructed. They then use markers to scan the genome and perform a **linkage analysis** in their attempt to map the gene.

The team analyse the pedigree, trait and genotyping information using probability models that measure the significance of the linkage. Linkage analysis has already proved successful in mapping the genes for Huntington's disease and muscular dystrophy, and the breast cancer genes *BRCA1* and *BRCA2*.



Australian scientists are involved in increasing our knowledge about our genes and inheritance.

Bioinformatics

Bioinformatics involves the use of computer technology to manage and analyse biological data. It has implications for a variety of fields, one of which is biotechnology.

DNA chips

DNA chips are made up of probes consisting of short fragments of selected genes attached to a wafer. Adding a sample of an individual's DNA to a particular type of DNA chip will result in a 'light up' response in the

presence of one of the genes on the chip. This positive response is caused by the matching DNA locking onto the relevant probe on the chip. For example, a chip that carries probes for cystic fibrosis will search for the cystic fibrosis gene (allele), ignoring all other genes.

Implications of genetic testing

DNA chips are critical for making some sense of the enormous amount of information that research has supplied us with about the human genome. Although the most obvious use of these chips (and similar future technologies) is for diagnosis, there are important implications related to the ease with which genetic information can be accessed. With increasing knowledge about the human genome and inheritance, probes for genes associated with particular phenotypes (both favourable and unfavourable) may be incorporated into these DNA chips. Which gene probes should be used? Could they be used without your permission (or awareness)?

The construction of DNA databases, applications of bioinformatics and increased availability of other types of DNA profiling techniques will open up a database of new questions, problems and issues. Who owns the resulting genetic information and decides what is done with it? Who should make these decisions?

No room for error

The beady eye of DNA regulators needs to fall on paternity testing. The genetics revolution has progressed at breakneck speed since the discovery of the structure of DNA, and regulators have often struggled to keep up. It has been a few years since the 'personal genomics' industry took off, and the US Food and Drug Administration is only now warning firms that genome scans are 'medical devices' that require approval.

New Scientist

Drop of blood reveals age of perpetrator

Blood left at a crime scene could be used to estimate the age of a perpetrator, thanks to a new DNA test. The test could narrow down the range of possible suspects.

New Scientist

Genetic rights

After more than a decade, the US Senate has finally passed the *Genetic Information Nondiscrimination Act* (GINA).

New Scientist

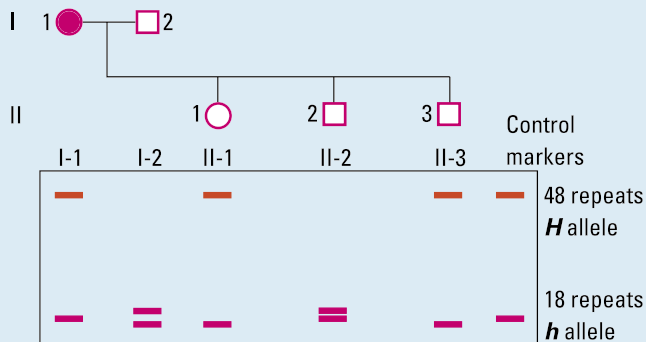
UNDERSTANDING AND INQUIRING

REMEMBER

- 1 Suggest six reasons for using genetic tests.
- 2 State three genetic disorder screening tests performed on newborn babies.
- 3 Outline similarities and differences between chorionic villus sampling and amniocentesis.
- 4 Identify the function of:
 - (a) restriction enzymes
 - (b) electrophoresis
 - (c) DNA probes
 - (d) PCR (polymerase chain reaction).
- 5 List four reasons for using DNA fingerprinting.
- 6 Describe how the use of computers and technology has increased our knowledge about genetics.

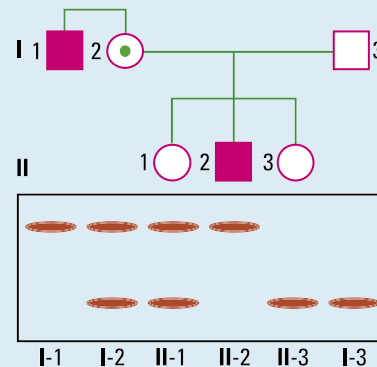
THINK, ANALYSE AND INVESTIGATE

- 7 The symptoms of the autosomal dominant inherited disorder Huntington's disease (HD) don't usually appear until the affected person is over 30 years old. Predictive testing can be carried out to determine an individual's genetic status. In the figure below, *H* represents the faulty HD allele and *h* the normal allele. The mother, I-1, is currently showing the symptoms of HD and has the genotype *Hh*.

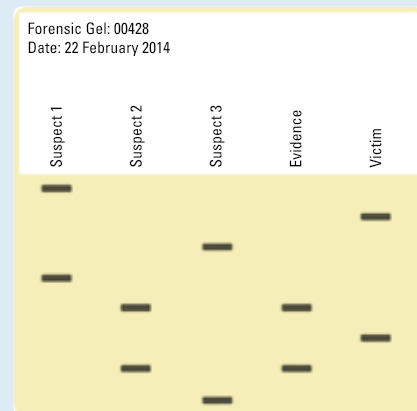


- (a) Suggest the genotype of:
 - (i) individual II-3
 - (ii) individual I-2.
 - (b) Suggest whether the children are likely to develop HD. Justify your response.
 - (c) Find out more about HD and current related research and issues.
- 8 Duchenne muscular dystrophy (DMD) is an inherited X-linked recessive disorder. The figure above right shows a pedigree and RFLP patterns that were obtained using a direct gene probe. Those individuals affected are shaded.
- (a) State the individuals with DMD.
 - (b) Describe the RFLP pattern of those individuals with DMD.
 - (c) If the mother, I-2, is a carrier of the DMD gene, which of her daughters is also a carrier?

- (d) Is the father, I-3, a carrier of the DMD gene? Explain.
- (e) Find out more about DMD and current related research.



- 9 The diagram below shows the DNA fingerprint of a victim, the DNA fingerprint from evidence taken from her body after an attack, and the DNA fingerprints of three suspects.



- (a) Using the information in the DNA fingerprints, which of the three suspects is most likely to be guilty of the crime against the victim?
 - (b) Give reasons for your response to part (a).
 - (c) Suggest why a sample was taken from the victim as well as the foreign DNA sample being collected from her body.
 - (d) (i) State some other forensic diagnostic tools that exist to identify those guilty of crimes.
(ii) How do these compare with DNA fingerprinting?
- 10 (a) Consider and answer each of the following questions, justifying your responses.
 - Should the creation of a child from two different genetic mothers be encouraged?
 - Should a killer's jail sentence be reduced because they have a genetic disposition towards violence?
 - Can your genes absolve you of responsibility for a particular crime?
 - If you could 'engineer' your own child, would you?
 - (b) Propose three of your own genetic issue questions for class discussion.

INVESTIGATE, THINK AND DISCUSS

- 11 Select two of the article headlines from page 54 in this section and find out more about the topics. Write your own article on one of the topics for your class science magazine.
- 12 Find out more about the Australian Genome Research Facility (AGRF) and its involvement in genotyping many markers for many individuals in a single day.

eBookplus

- 13 What does a museum have to do with genetics? Use the **Bioinformatics** weblink in your eBookPLUS to find out about bioinformatics at a museum.
- 14 In 1993, American scientist Kary Mullis won the Nobel Prize in Chemistry for investigating PCR. Find out more about the discovery and applications of PCR.
- 15 What are bioethics and how do they relate to genetic testing? Research the use of a particular type of genetic testing (such as embryo selection, personal genomes, carrier status, predictive testing) and consider relevant bioethics issues.
- 16 Find out any implications of having a genetic disease for obtaining life and health insurance in Australia.

1 What is the issue?

2 Who will be affected by the issue?

3 What are the positive points of view?

(Who or what benefits, and why and how?)

4 What are the negative points of view?

(Who or what is disadvantaged, and why and how?)

5 What are some of the possible alternatives?

6 How may these alternatives affect those involved?

7 What is a possible solution that may be acceptable to those involved?

8 What is your opinion on the issue?

When involved in a bioethical discussion, you need to consider these questions.

- 17 Suggest ways in which information from genetic tests may be used by organisations such as insurance companies, medical facilities and workplaces.
- 18 Suggest implications of patenting any of the following.
 - (a) Genes
 - (b) Gene products
 - (c) Specific drugs that target a gene or gene products
- 19 Various countries and organisations are already developing DNA databases.
 - (a) What is a DNA database?
 - (b) Use a PMI chart to categorise possible applications of DNA databases.
 - (c) Provide your own personal opinion on DNA databases. Include reasons for your opinion.
- 20 Research one of the following genetic careers: genetic statistician, genetic engineer, bioethicist, genetics counsellor, genetic researcher, genetic pathologist, molecular biologist, forensic scientist, sequencing specialist, bioinformatics/functional genomics officer.
- 21 Research one of the following Australian research institutes and find out more about their genetic research.
 - Ludwig Institute for Cancer Research
 - Howard Florey Institute of Experimental Physiology and Medicine
 - Walter and Eliza Hall Institute
- 22 What is thalassaemia? Find out more about screening and diagnostic tests for this genetic disorder. Find out more about the Thalassaemia Society and its involvement in genetic counselling.

eBookplus

- 23 Find out more about:
 - (a) the Genetic Support Network in your state
 - (b) companies that provide gene testing and screening. Use the **GeneScreen** weblink in your eBookPLUS to read about one example.

CREATE

eBookplus

- 24 Click on the **PCR** weblink in your eBookPLUS to listen to and read the lyrics of the PCR song about how to amplify DNA.
 - (a) Do you consider this effective advertising? Justify your response.
 - (b) Design your own advertisement to promote scientific equipment, products or services.
- 25 Find out about jobs in the field of genetics by clicking on the **Genetic careers** weblink in your eBookPLUS.

Domesticating biotechnology

Human genes in bacteria? Insect genes in plants? Cotton plants producing granules of plastic for ultra-warm fibre? These sound bizarre but are not in the realms of fantasy — they are happening now! What new creations will tomorrow bring?

Tomorrow, today?

You are living in the midst of a biological and technological revolution. Advances in biotechnology are gathering momentum so fast that your life will never be the same. We are speeding towards a future in which domesticated biotechnology may be a way of life.

Tools of the trade

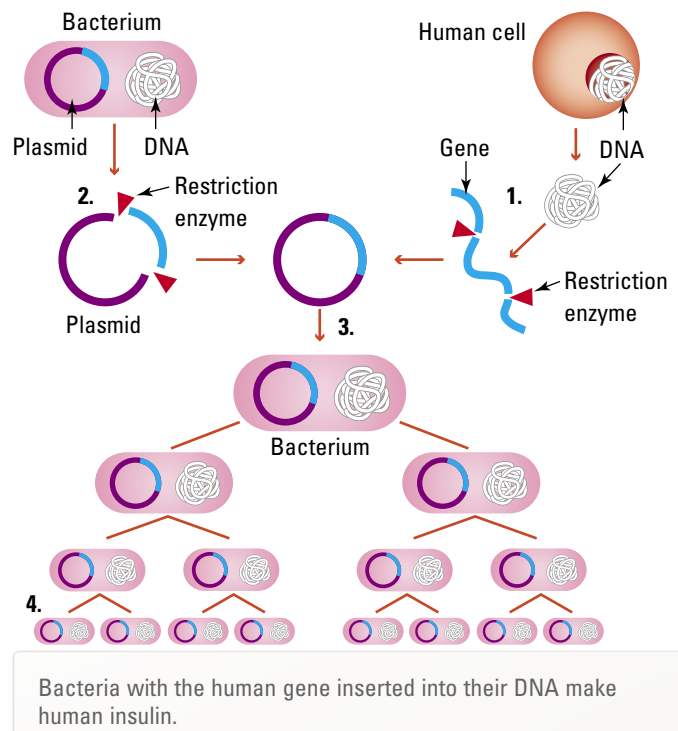
Genetic engineering is one type of biotechnology that involves working with DNA, the genetic material located within cells. **Genetic engineers** use special tools to cut, join, copy and separate DNA.

Transferring the code

Genes from Arctic fish can be added to the genome of tomato plants so that they become frost resistant. This is an example of **recombinant DNA technology**. This technology uses specific enzymes called restriction enzymes to cut the DNA at specific points, so that a particular gene is removed. This DNA can then be pasted into the DNA of another organism using **DNA ligase**. If the organism belongs to another species, it is described as being **transgenic**. The feature coded for by the foreign gene is then expressed by its new host.

Cloning

If you saw the movie *Jurassic Park*, you may recall the scene in which scientists extract dinosaur DNA from mosquitoes that had been trapped in amber. They placed this prehistoric DNA (with a mix from some other living organisms to fill the gaps) into surrogate



eggs. While the science in *Jurassic Park* has more than a few holes in it, we do have (and are still developing) technologies to clone single genes, some types of tissues and organs, and entire organisms.

Gene cloning involves the insertion of a specific gene into bacteria, so that the bacteria will act as microfactories and produce considerable quantities of desired proteins. This type of cloning has been used to produce insulin for diabetics and missing clotting factors required by haemophiliacs.

Therapeutic cloning and nuclear transfer cloning both involve the insertion of a nucleus from a somatic cell into a fertilised egg cell (which has had its own nucleus removed or destroyed) to create **totipotent stem cells**. The cells in therapeutic cloning are treated so that they will grow and divide into cells of a particular type or produce a specific type of tissue or organ. The cells in nuclear transfer cloning are transplanted into a surrogate host animal and result in the production of identical copies of the organism that supplied the donor DNA.

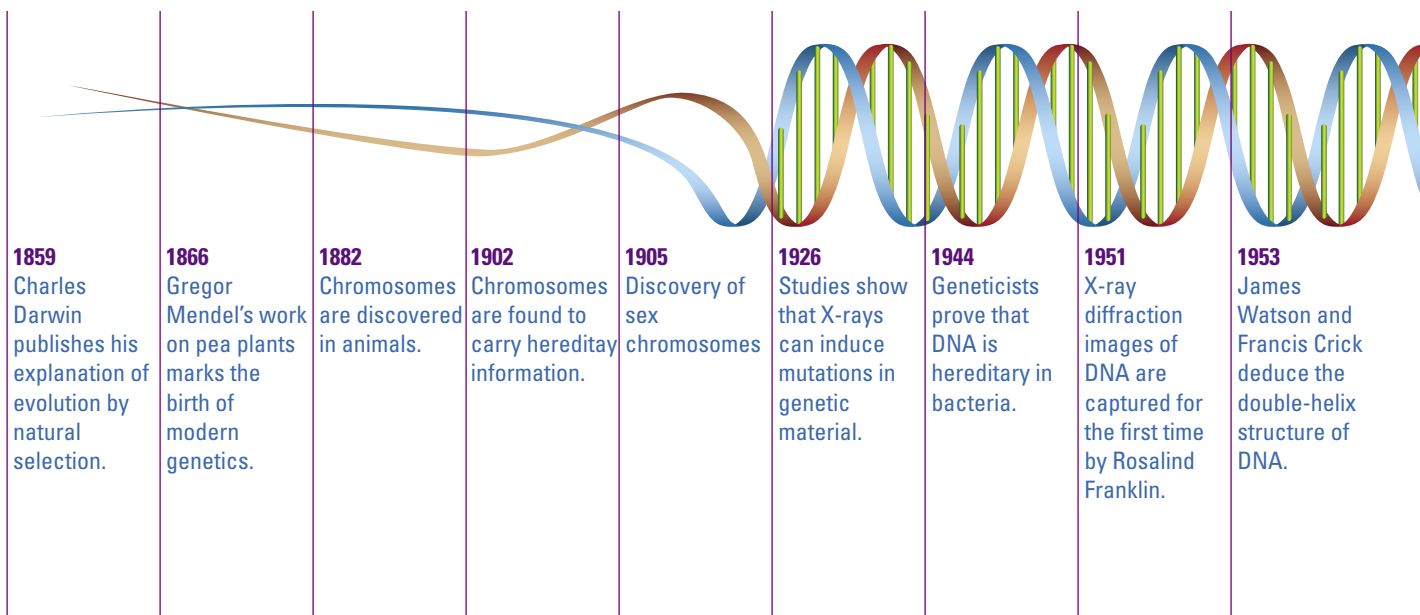
Reproductive cloning involves separating the cells of the developing embryo and implanting them into different surrogate mothers. The offspring from these surrogates will be identical to each other.

eBook plus eLesson

Ancient resurrection

Do we have the right to resurrect ancient species? Watch an ABC *Catalyst* video to find out more.

eles-1070

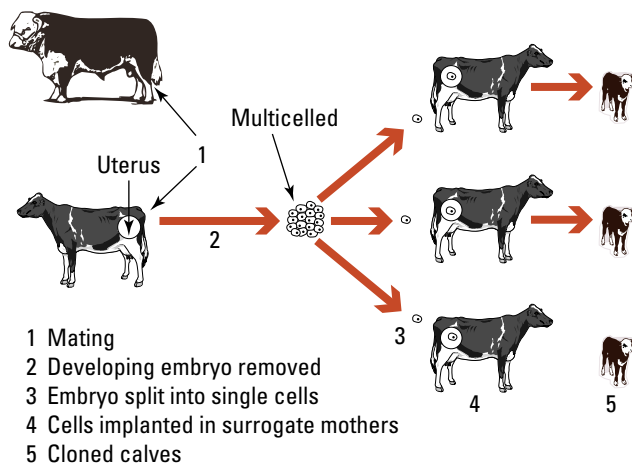


Are we in the midst of a molecular biology and biotechnology revolution? What new discoveries will the future bring and what implications will they have on our lives?

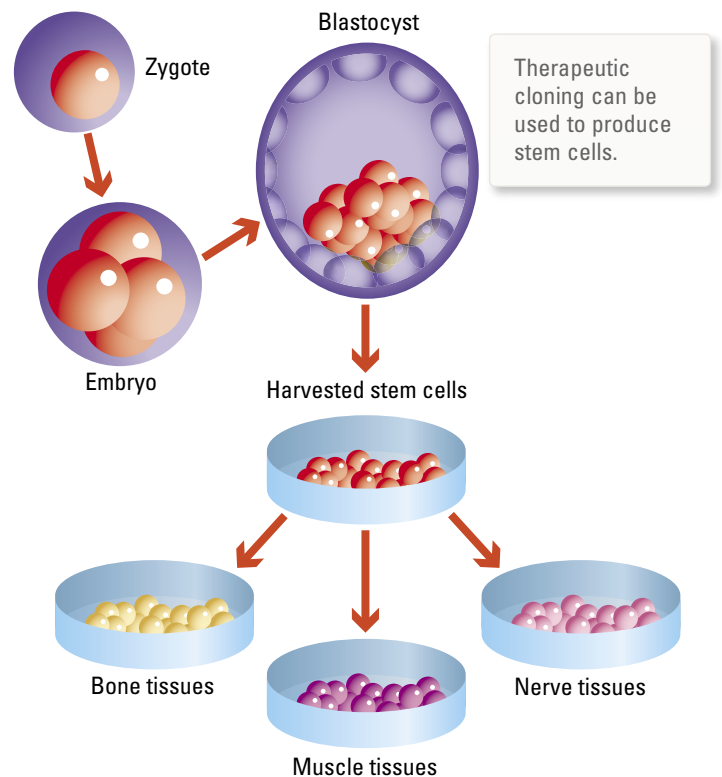
Human cloning

It can be argued that Australian scientists are leaders in the field of molecular biology and genetics. Around Australia science researchers are involved in cutting-edge investigations in genetics and molecular biology.

Australia made headlines in December 2006 when a ban on research on therapeutic cloning or **somatic cell nuclear transfer** was lifted in a national parliament vote, and Australia issued the first licence to clone human embryos.

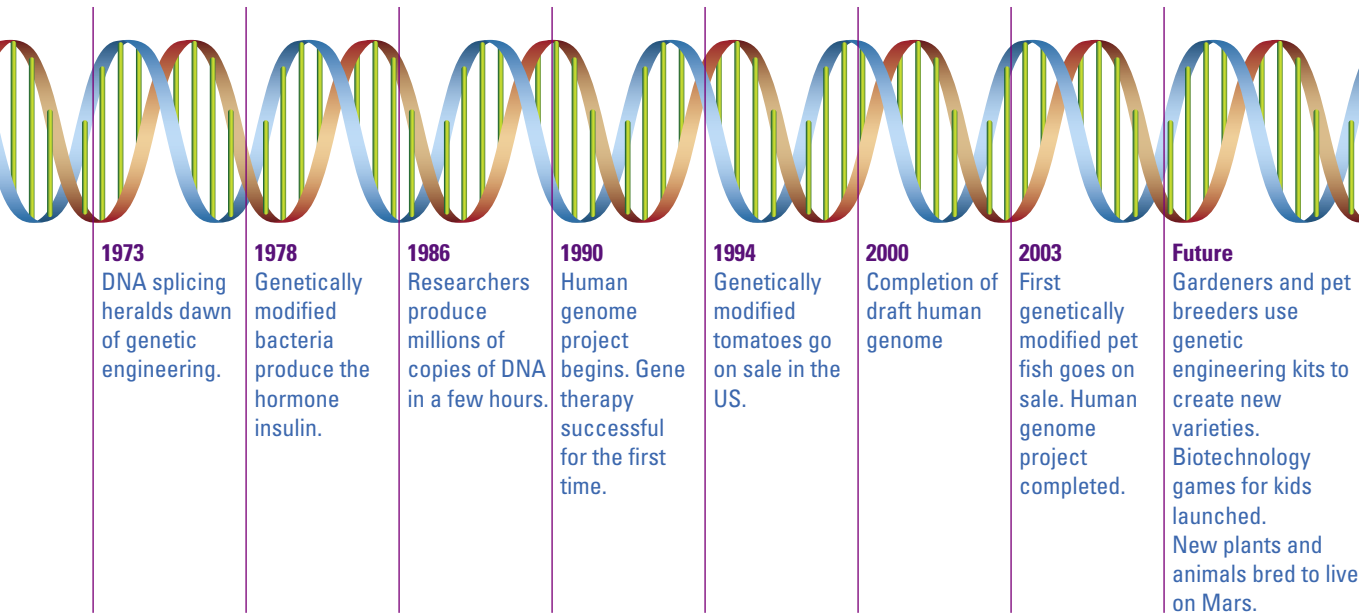


Suggest why these surrogate mothers produce offspring that are identical to one another.



The plant invader

Agrobacterium tumefaciens is a soil bacterium. It is able to get inside and infect many plants such as vines and fruit trees. In doing so, it transfers a tiny piece of its DNA into the host cell. This programs the host cell to



make chemical compounds for the sneaky bacterium to feed on. Genetic engineers saw the possibility of using this bacterium as a **vector** to carry the genes they wanted from one plant into another.

Other kinds of bacteria and viruses act as vectors and carry genetic information from one organism

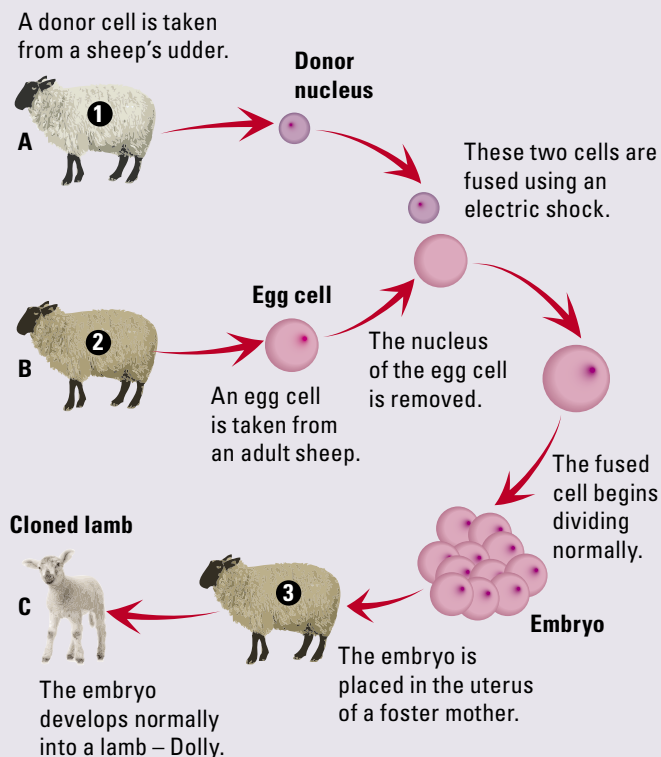
(or synthesised to be like that organism) to another organism. Vectors can be used to carry genes for producing protein in soybean and sunflower plants, enzymes to control chemical processes, or compounds that keep insects or pathogenic viruses at bay.

HOW ABOUT THAT!

The story of Dolly the sheep began at the Roslin Institute in Scotland or, more specifically, as a single cell from the udder of a ewe. Follow her story in the diagram at right.

Dolly made history as the first mammal to be cloned from a single adult cell. Until then, biologists did not believe that once a cell had developed and become specialised, it could be reprogrammed to become different.

A group of cells that come from a single cell by repeated mitosis have the same genetic coding as each other. They are **clones** of each other. All of Dolly's cells came from the original fusion of an unfertilised egg and DNA from an udder cell. As there was no genetic input from another sheep, Dolly was a clone of the parent ewe from which the udder cell came.



How Dolly the sheep was created

Take aim ... fire!

Recent developments have enabled foreign genes to be inserted into plant tissues by shooting them in with gas guns. Fine particles of gold are coated with the DNA and shot into the cells. Some cells are killed in the process but some survive, carry out mitosis and develop into complete plants with an altered genotype. Many plant crops such as maize and soybean have had favourable genes added to them in this way.

Biotechnology involving gene technology is a rapidly expanding branch of science. Already there have been trials of viral-vector nasal sprays to help treat people affected by cystic fibrosis. Some of the viruses carried by these vectors penetrate cells lining the respiratory tract and insert the normal gene into those cells. Vaccines against a number of diseases in humans and other animals are being investigated. Biotechnologists are trying to genetically engineer bananas to produce a vaccine against the hepatitis B virus. Will genetically engineered vaccines eventually prevent diseases such as AIDS?

Do-it-yourself creation kits

Imagine if everyone was able to access genetic engineering tools to create and develop new forms of life. Imagine being able to create new plants for your garden, and pets that you could only dream about. Will designing genomes become a personal thing — a type of art form or expression of creativity?

What sorts of biotechnology games will be designed and produced? What sorts of lessons and learning may kindergarten children get from creating their own organisms to watch grow and interact with? Should there be rules and regulations for this new biotechnology? What sort of rules and regulations should there be? Who should make them? How can they be enforced?



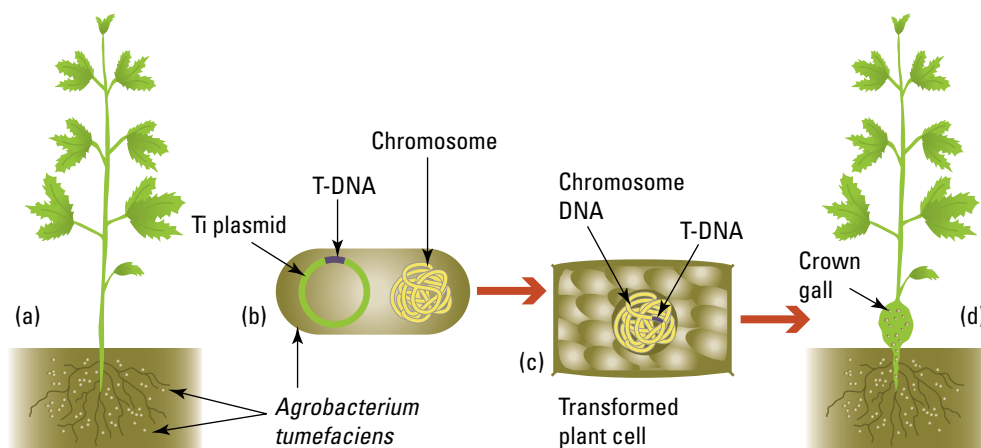
Gene guns can be used to insert DNA into cells.

Should we or shouldn't we?

Is transferring genes a wise use of gene technology? Some people argue that not enough is known about the way genes can jump the species barrier. Maybe they will end up where they shouldn't, such as in food chains. What could be the effect on other species in the environment? Could foreign genes interact with host genes and cause problems? Could viral vectors and genes mutate so that they would infect not only the target species but others too?

Gene therapy

Gene therapy is currently an experimental discipline and there is still considerable research required before it reaches its full potential. Gene therapy has a specific goal of targeting the gene that is responsible for a genetic disease. This type of therapy can be used to replace a faulty gene with a healthy version or insert a new gene that may cure or reduce the effects of a genetic fault.



The bacteria *Agrobacterium tumefaciens* has the ability to infect plants by inserting some of its DNA into the DNA of the plant. It has been used by geneticists to insert specific genes into plant DNA.

A delivery problem

There has been extensive research on the use of gene technology to treat or cure a variety of inherited diseases. If this is to be a viable alternative, the gene that causes the disease and the location of the affected cells need to be found. It also requires the availability of a healthy version of the gene and a way for it to be delivered to the cell. The delivery of the new genetic material has been one of the major stumbling blocks so far.

Viral transport

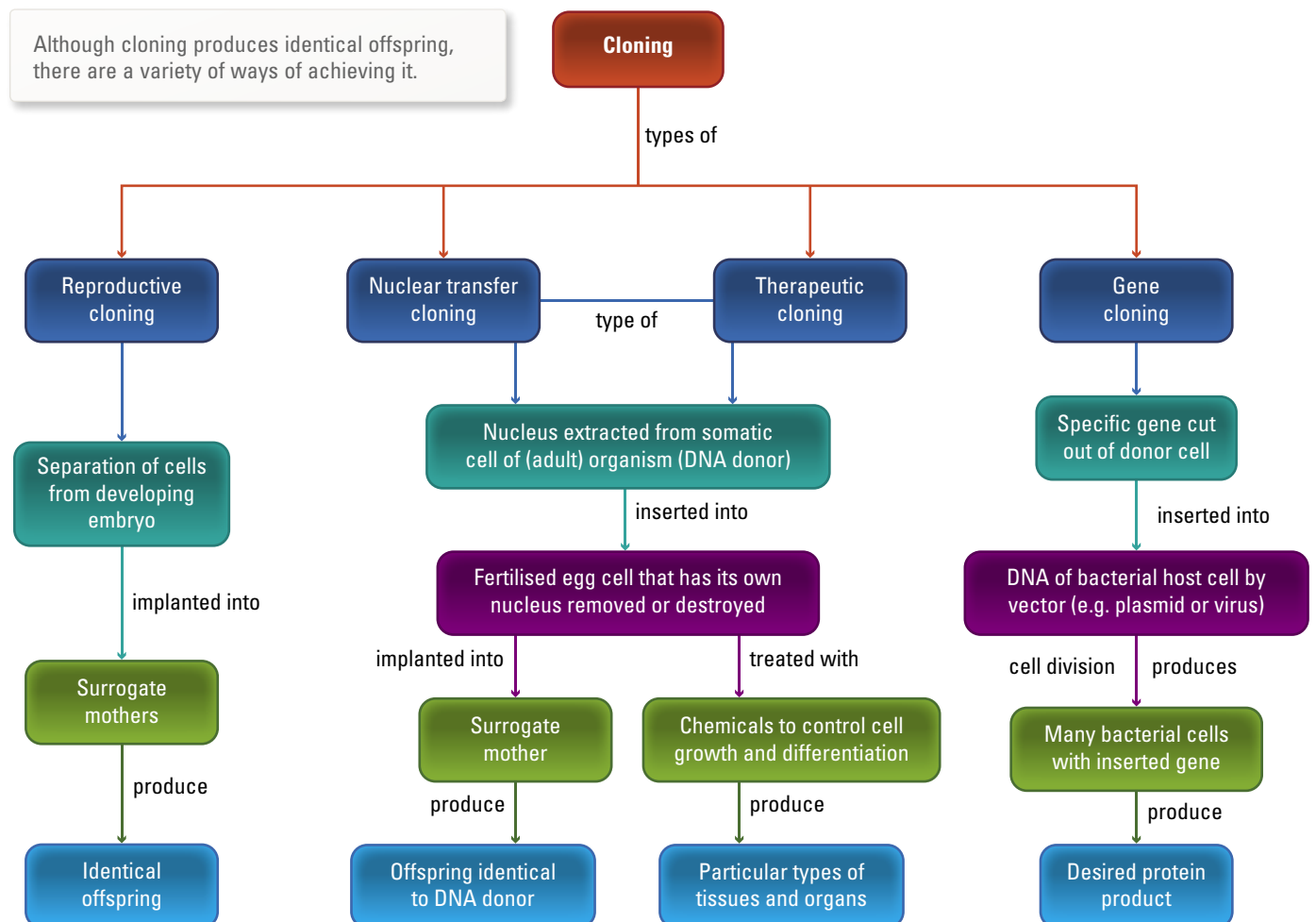
Early trials used a type of adenovirus with a healthy version of the cystic fibrosis gene inserted into its DNA. It was anticipated that this altered adenovirus would infect cells in the respiratory system, take over the cell's genetic machinery and make viruses that would make the required protein. While there was some success, there were also complications. This led to the development of different types of vectors that were less likely to mutate or cause adverse reactions within the hosts of these genetically engineered delivery vehicles.

Risks

While there are considerable potential benefits from the use of gene therapy, there are also risks. Some of these include the host's immune response to the foreign genetic material, the incorrect insertion of the new genes into the DNA or into an unintended cell, and the production of too much of the missing enzyme or protein. If viruses are used as vectors, the deactivated virus may target unintended cells or may be contagious, spreading to other organisms. The consequences of many of these risks remain unknown.

Issues

If and when these stumbling blocks in the delivery of the new genetic information are overcome, some new ethical and moral issues may arise in their place. Will gene therapy have the potential to create a more elite human being? Will there be attempts to alter characteristics such as height, intelligence or whatever the fashionable traits are at the time? Who will have access to this technology? Will gene therapy be available only to the rich and those in power? Who should be able to own and profit from this technology?



UNDERSTANDING AND INQUIRING

REMEMBER

- 1 What are clones?
- 2 Why is Dolly famous?
- 3 Refer to the diagram on page 59 showing Dolly's creation.
 - (a) What had to be done to the unfertilised egg taken from ewe number 1?
 - (b) How did the donor cell DNA (from ewe 2) get inside the empty cell from ewe 1?
 - (c) What was ewe 3's role?
- 4 Summarise the tools of genetic engineering into a mind, concept or cluster map. Include diagrams or figures to help you remember what each of the tools does.
- 5 What is gene technology? What are its potential benefits?
- 6 Draw a simple flow diagram to summarise how bacteria are used to produce insulin that people with diabetes can use.
- 7 Explain what transgenic organisms are and give an example.
- 8 What is a vector?
- 9 What methods are used to transfer foreign genes into other organisms?

INVESTIGATE, THINK AND DISCUSS

- 10 Why is Dolly called a clone?
- 11 Which ewe was the original Dolly?
- 12 Does Dolly have any genetic material from a ram?
- 13 There is now no theoretical scientific reason why cloning of humans, even dead ones, is not possible. Some people have suggested that cloning of humans should be permitted, while some countries, such as the United States, have outlawed it.
 - (a) What reasons would people give for wanting to clone humans?
 - (b) Research media stories that raise issues related to cloning, particularly of humans. Draw an issue map to identify some of the implications. Keep a record of your references.
- 14 Since Dolly, a number of other animals have also been cloned. Find out more about the cloning of at least one other animal. Report on:
 - (a) the reasons for cloning that particular animal
 - (b) how the animal was cloned
 - (c) the advantages and disadvantages associated with the cloning of the animal.
- 15 Use the timeline in this section to answer the following questions.
 - (a) In which year were sex chromosomes discovered?
 - (b) Find out when Dolly was born and add the date to the timeline.
 - (c) Find two other biotechnological events to add to the timeline.
 - (d) Research one of the events on the timeline in more detail and share your findings with your class.
- 16 (a) Outline how gene therapy could be used to treat and cure genetic diseases.
 - (b) Suggest reasons why gene therapy is not currently being extensively used to treat genetic diseases.
 - (c) Identify issues related to gene therapy.
 - (d) Use a SWOT analysis to categorise points made in a discussion on gene therapy.
 - (e) Identify different possible uses of gene therapy.
 - (f) Construct a priority grid to map out different potential uses of gene therapy.
- 17 Design a biotechnology game of the future.
- 18 Find out the environmental conditions on Mars and think about what sort of life form could survive them. Design a plant or animal that you think would have a good chance of surviving on Mars.
- 19 What are some of the advantages and disadvantages of gene technology? Use specific examples to support your views.
- 20 Would you eat genetically manipulated food? Why or why not?
- 21 In a group, compile a folio of about 10 journal and other media articles relating to gene technology. Make sure you note the date and source of each one. Each person will choose an article to analyse. In your analysis include:
 - (a) the kind of gene technology being reported on
 - (b) a simple description or explanation of the particular example of technology
 - (c) any issues relating to the example.

DNA — interwoven stories?

Do you think you are special? Of course you are! Your DNA contains a wonderful tapestry of not just your human ancestry but also that of other genomes.

Hologenome theory of evolution

Although we see ourselves as being purely human, we are not. Like other plants and animals, we contain DNA from a variety of sources. The **hologenome theory of evolution** emphasises the role that micro-organisms have within our evolution.

Micro-organisms (microbiota) within their hosts (such as plants or animals) can be described as being **symbionts**. The term *holobiont* is used to describe the host and all of their symbiont microbiota collectively. Some scientists have suggested this as a unit of selection in evolution.

The hologenome is made up of the combined genomes of the host and the microbes within it. Genetic variation within the holobiont may occur in both the host and the microbial symbiont genomes, and the variation can be inherited by the offspring. Some of this variation may occur within existing microbes; other variation may be due to microbes newly acquired from the environment. Some scientists consider this view as Lamarckian, as the inheritance of variation via microbes follows Lamarck's idea that traits acquired within the lifetime of the parent can be transmitted to the next generation.

From 'junk' to treasure?

Did you know that our genome contains viral DNA? Our DNA is made up of coding and non-coding sections. It was long thought that the non-coding DNA served no purpose. As this DNA contained highly repetitive sections and did not code for amino acids, it was often referred to as 'junk' DNA. New technologies and discoveries, however, are fast changing our views of this. Scientists have discovered that some of these non-coding regions contain genes that regulate many activities, and without them, protein synthesis using the coding DNA sections would not occur.

Identified by your 'junk'?

Scientists have found that the complexity of an organism is not matched to the total amount of DNA that it contains. What they have discovered is that there

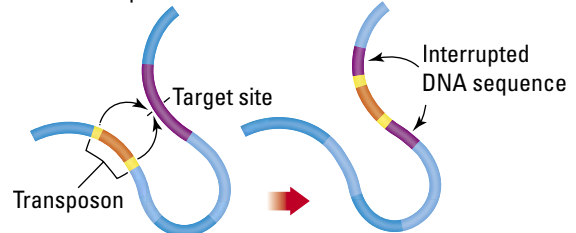
is a relationship between non-coding and coding DNA. When testing people's tissue types, Malcolm Simons, a New Zealand-born immunologist, discovered that the pattern of the 'junk' DNA surrounding MHC genes was a very good predictor of tissue type. Some scientists suggest that this 'junk' DNA has played a key role in making us human, as it distinguishes primates from other mammals. It has been suggested that this 'junk' DNA is due to the invasion of a million copies of jumping genes!

Jumping genes

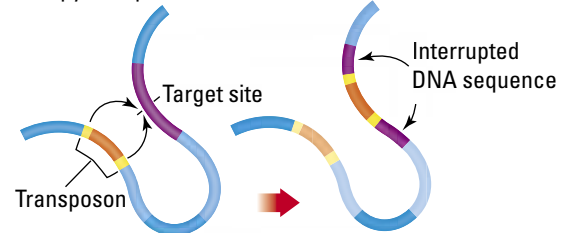
Barbara McClintock (1902–1992) was a cytogeneticist whose scientific theories (and possibly her gender) clashed with the scientific community of her early research years. McClintock investigated how chromosomes change during reproduction in maize (*Zea mays*). Maize proved to be the perfect organism for the study of transposable elements (TE) or 'jumping genes'. She contributed to our understanding of the mechanism of crossing over during meiosis and produced the first genetic map for maize. She linked regions of the chromosome with physical traits and demonstrated the roles of telomeres and centromeres. She also discovered **transposition** and outlined how

Two methods of transposition:

1. Cut-and-paste mechanism



2. Copy-and-paste mechanism



DNA sequences known as transposons or 'jumping genes' can copy themselves into other sections of the genome.

it could be involved in turning on and off the expression of genes.

McClintock pioneered the study of **cytogenetics** in the 1930s. Before the structure of DNA was even discovered, McClintock was the first scientist to outline the basic concept of **epigenetics**, recognising that genes could be expressed and silenced. McClintock's theories were revolutionary because they suggested that an organism's genome was not a stationary entity, but something that could be altered and rearranged. This view was highly criticised by the scientific community at the time. She was eventually awarded the Nobel Prize in Physiology or Medicine in 1983, when she was over 80, for research she had done many years before.

Your jumping genes

Some of the repeating sequences within our non-coding DNA are known as **transposons** or 'jumping genes'. They may have originated from invading viruses. These sections have the ability to copy themselves independently of the rest of the genome and then randomly insert themselves in other sections of the genome. There have been suggestions that our evolution has been shaped by these transposons.

Genetic invasion

The human immunodeficiency virus (HIV) is an example of a retrovirus. Retroviruses convert their RNA genome into DNA before implanting it into host chromosomes. This process is called **endogenisation**. If the viral genome is incorporated into the chromosomes in the host's germline, it can become a part of the genome of future generations.

Are you aware that such germline endogenisation has happened repeatedly in our own lineage? This mechanism may help explain the varied sources of the DNA in your own genome. It provides an explanation as to why our genome may contain thousands of **human endogenous retroviruses (HERVs)**. Are these the legacies of viral invasions throughout our evolutionary history?

Viruses have an amazing ability to unite, genome to genome, with their hosts. This has a powerful evolutionary significance; it supplies the host with a new source of variation for evolution. If a virus happens to introduce a useful gene, natural selection will act on it. Like any other beneficial mutation, it will spread through that population.

If the human genome has evolved as a holobiontic union of vertebrate and virus, could plagues be considered a vital evolutionary survival tool for our descendants? What are the implications of this theory in terms of how we treat diseases and think about

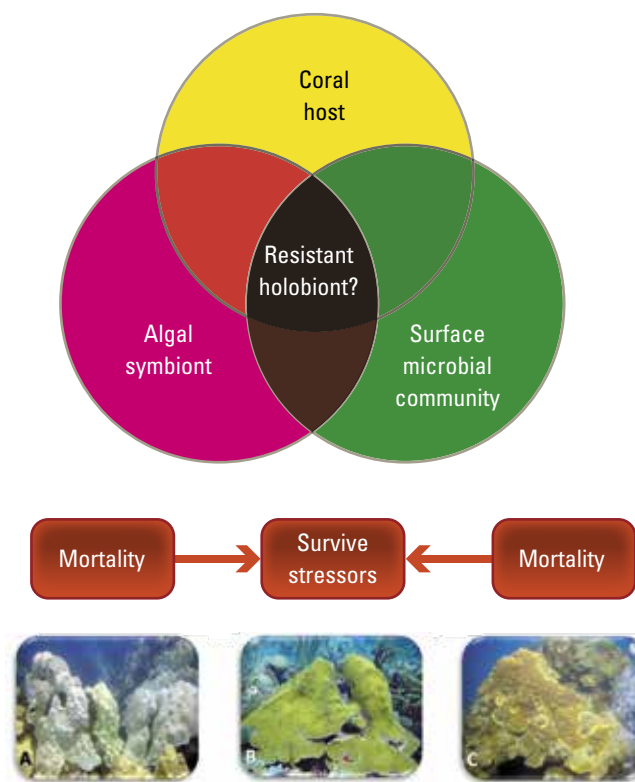
viral infections? Do viruses have a place not just in our present, but in shaping our future evolution?

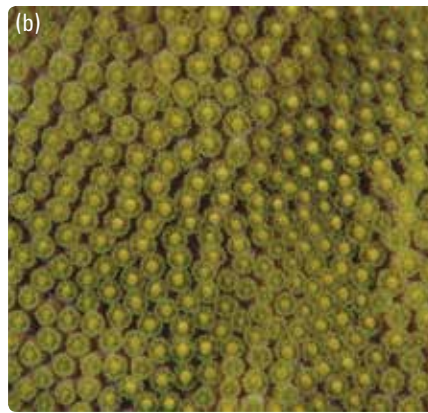
Coping with climate change

Some scientists have suggested that reef corals and possibly some other multicellular organisms may alter the microbial communities within their bodies to cope with environmental stresses such as those caused by climate change. More studies on this may provide some strategies we can use to help reduce the loss of biodiversity in ecosystems threatened by environmental changes related to climate change.

A career in Caribbean coral holobionts

Emmanuel Buschiazzi began his academic career as a scientist studying applied marine biology in Edinburgh, Scotland. He worked at the Marine Environment Laboratory in Monaco in marine ecotoxicology, then did his PhD in New Zealand studying the evolution of microsatellite DNA. His next adventure sent him to Canada for a postdoctoral fellowship in conifer genomics. He is currently involved in blending genomic approaches and population genetics to unravel the biology of two Caribbean coral holobionts, *Montastraea faveolata* and *Acropora palmata*.

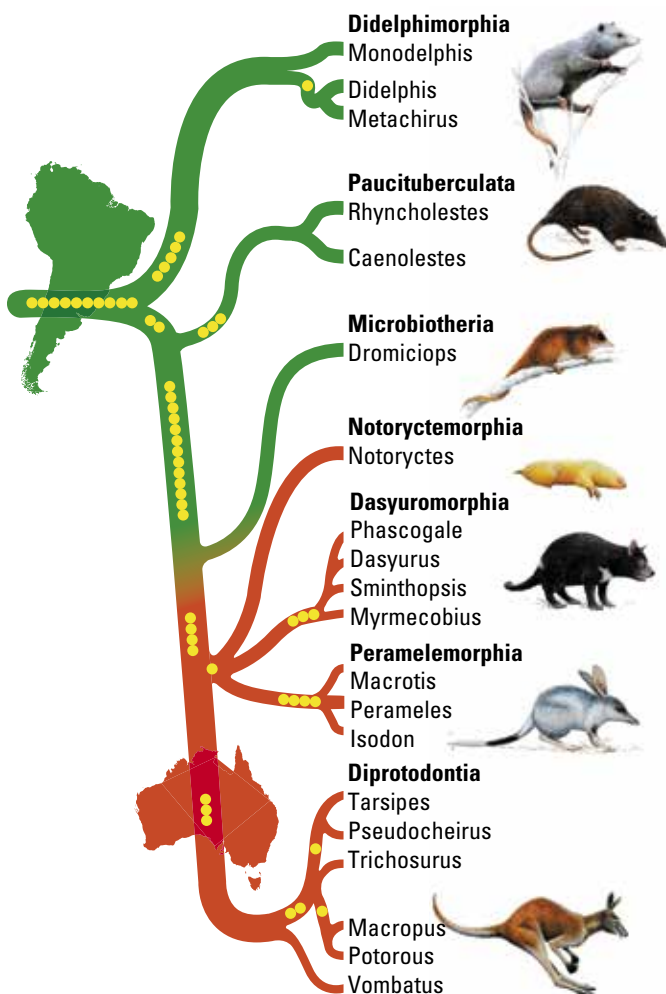




(a) Dr Emmanuel Buschiazzo (b) *Montastraea faveolata* coral (c) *Acropora palmata* coral

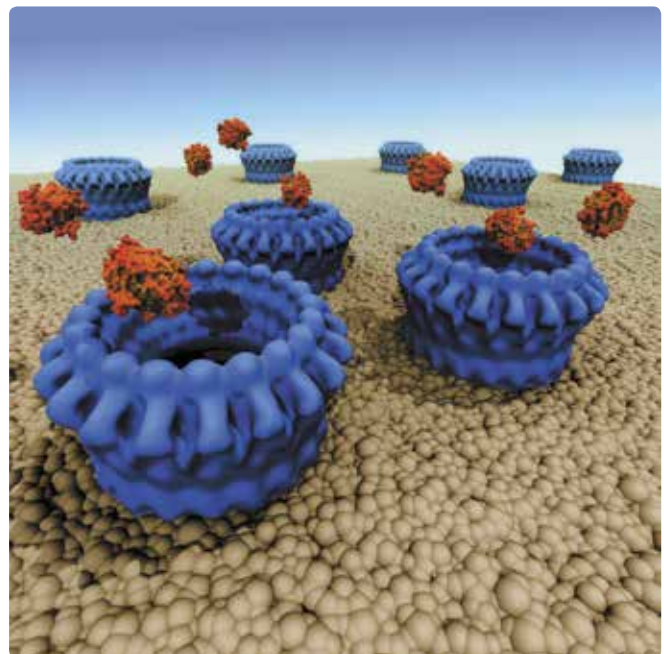
Australian marsupials and jumping genes

Scientists have argued about how marsupials such as kangaroos, opossums and Tasmanian devils evolved in South America and Australia. DNA sequencing and the fossil record tell two different stories. Do jumping genes hold the answer to the mystery?



A German evolutionary biologist, Maria Nilsson, has been investigating this mystery by looking at strange bits of DNA called **retroposons**. Retroposons have the ability to break off chromosomal DNA and then copy and paste themselves elsewhere in the genome. Once they copy and paste themselves their locations are stable, making them a reliable marker for determining evolutionary relationships.

Nilsson's retroposon data suggest that the Australian and South American marsupials could be divided into two distinct groups that had little contact as they evolved. This supports the DNA sequencing data that they share a single South American ancestor that travelled to Australia before the continents drifted apart, and that they evolved separately afterwards.



Where did perforin, our protein assassin, come from?

Our protein assassin

Australian and British scientists have identified the process through which our natural killer cells (part of our immune system) puncture and destroy virus-infected or cancerous cells. A protein called **perforin** is responsible for forming a pore in the diseased cell. The natural killer cells can then inject toxins into the diseased cell to kill it from within.

By using powerful technologies such as the Australian Synchrotron and cryo-electron microscopy, scientists have determined the structure of perforin and how it creates pores. This protein resembles cellular weaponry used by bacteria, and it is possible that our immune system may have incorporated the genetic information from bacteria within our evolutionary past.

UNDERSTANDING AND INQUIRING

REMEMBER

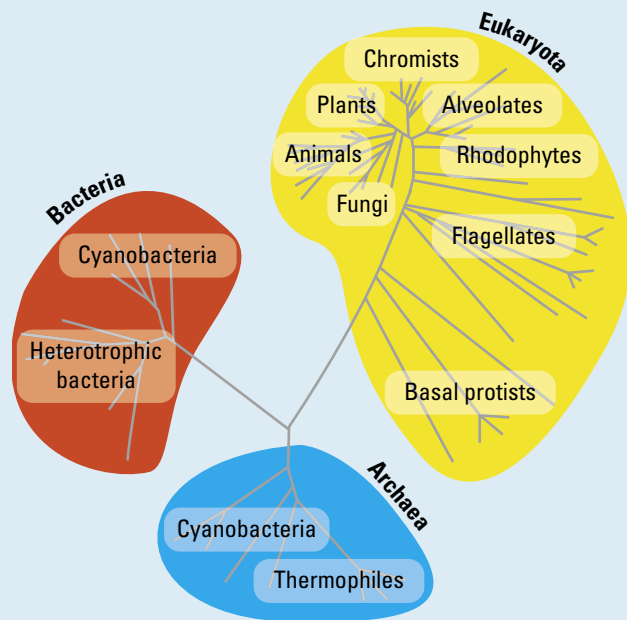
- 1 Suggest why we should not think of ourselves as purely human.
- 2 Suggest what the hologenome theory of evolution emphasises.
- 3 Suggest what is meant by the following terms.
 - (a) Symbiont
 - (b) Holobiont
 - (c) Hologenome
 - (d) Endogenisation
 - (e) Transposon
 - (f) Retroposon
- 4 Suggest why we no longer call regions in our DNA that do not code for proteins 'junk' DNA.
- 5 Who was Barbara McClintock? List examples of ways in which she contributed to our understanding of genetics.
- 6 State another name for 'jumping genes'.
- 7 Provide an example of a retrovirus.
- 8 Suggest how we know that germline endogenisation has occurred within our own lineage.
- 9 Suggest how research on retroposons has contributed to our knowledge about the evolution of Australian marsupials.

INVESTIGATE, THINK, DISCUSS AND REPORT

- 10 Research and report on one of the following.
 - (a) The hologenome theory of evolution
 - (b) Symbionts
 - (c) Holobionts
 - (d) Hologenome
 - (e) Barbara McClintock
 - (f) Transposons (or 'jumping genes')
 - (g) Endogenisation
 - (h) Microbiota and climate change
 - (i) Caribbean coral holobionts
 - (j) Australian marsupials and jumping genes
 - (k) Retroposons
 - (l) Epigenetics
 - (m) Perforin
 - (n) Astrobiologists
- 11 If viruses kill about 20 per cent of all living material in the oceans every day, releasing their contents for other organisms to grow, does that mean that they drive ocean ecosystems?
- 12 Yellowstone Park in America is a place that attracts scientists. Suggest why many scientists view this park as a voyage back in time.
- 13 Discuss the following statement: If it weren't for viruses, ocean ecosystems would stop.
- 14 How do you feel about the possibility of having viral DNA in your DNA? Construct a PMI chart for a discussion on this question with your peers.
- 15 If you had a question to ask about the human genome, what would it be?
- 16 If you could investigate an aspect of human evolution, what would your research question or hypothesis be?
- 17 Some people suggest that there was a second genesis on Earth and that it is living among us undetected. It could even be extraterrestrial in its origin. Discuss this possibility and propose a number of questions that could be investigated.
- 18 Do you think that life (as we know it) could have occurred on planets other than Earth? Justify your response and discuss it with your peers.
- 19 Do you think that all life on Earth descended from a common origin? Discuss this and justify your opinion.
- 20 Suggest how astrobiologists could detect a life form on Mars.
- 21 The genetic code consists of 64 possible triplet DNA combinations that code for one or more of the 20 different natural amino acids; all species on Earth use the same code. Suggest why this might be used as evidence that there has only been one genesis on Earth.
- 22 Due to research by Carl Woese in the 1970s, many scientists now accept that prokaryotes can be divided into two distinct groups and that those in the archaea group are older than bacteria. These ancient ancestors of life on our planet were riddled with viruses. Does that

mean that life on Earth originated from viruses? Discuss this question in groups and record comments made during your discussion.

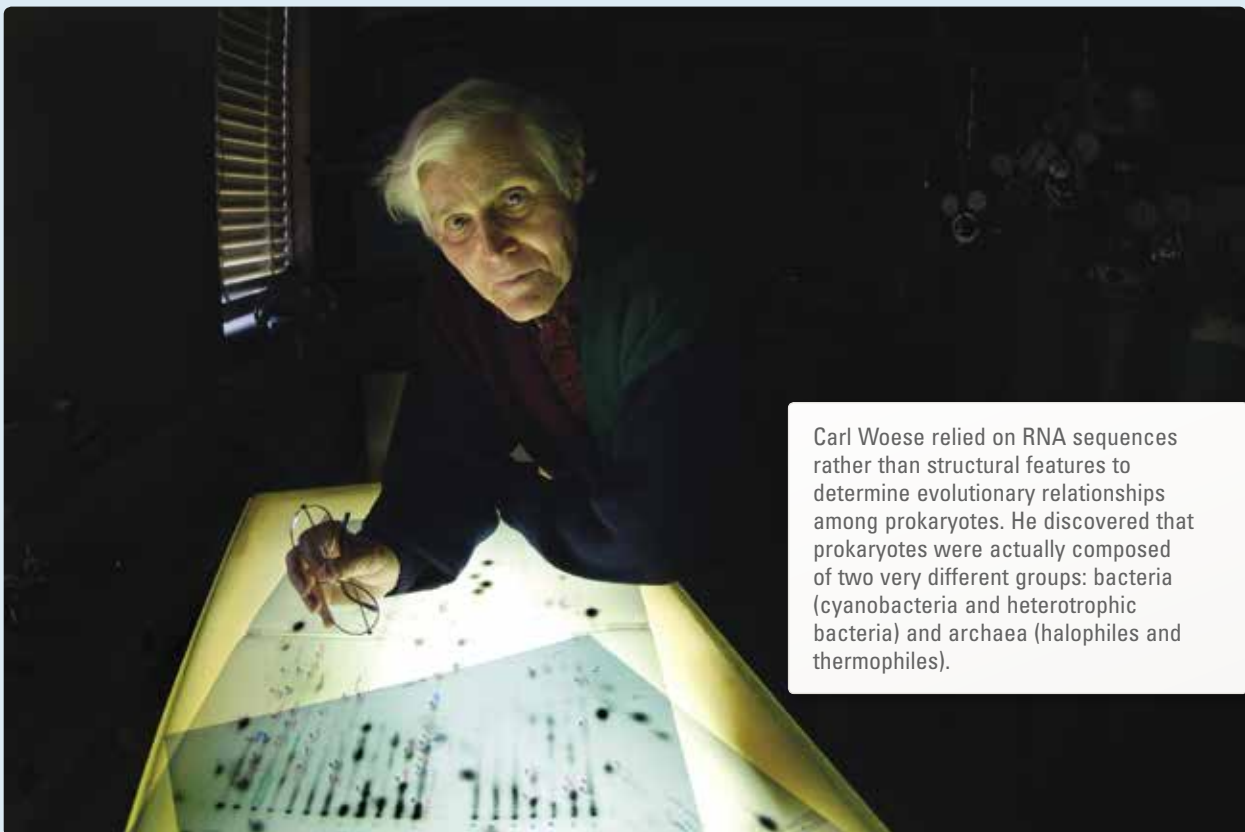
- 23 If living things did not share an ancestor that shared ribosomes, ATP and the triplet code, then why are these found universally among all living things? Discuss and justify your reasons.
- 24 Suggest how we might identify life as we don't know it.
- 25 Given that chance plays a large part in the evolution of life, it is unlikely that life from a very separate origin would have the same biochemistry. Discuss this statement. Do you agree with it? Justify your response.
- 26 If you were an astrobiologist, you might refer to known organisms as 'standard' life and alternative forms as 'weird' life. You might not know what you are looking for, or where to look to find it. But how weird is 'weird'? Which criteria do you assign to life, and how will you know when you have found it?
- 27 *Deinococcus radiodurans* is a halophile and an example of an extremophile that can survive high doses of radiation. It can be found living in waste pools of nuclear reactors. Find out more about this microbe and others that can survive high levels of radiation.
- 28 The unusual Murchison meteorite fell north of Melbourne just two months after the lunar landing in 1969. It belonged to a rare class of carbonaceous chondrites. Find out more about this meteorite and its evolutionary implications.



eBookplus

work
sheet

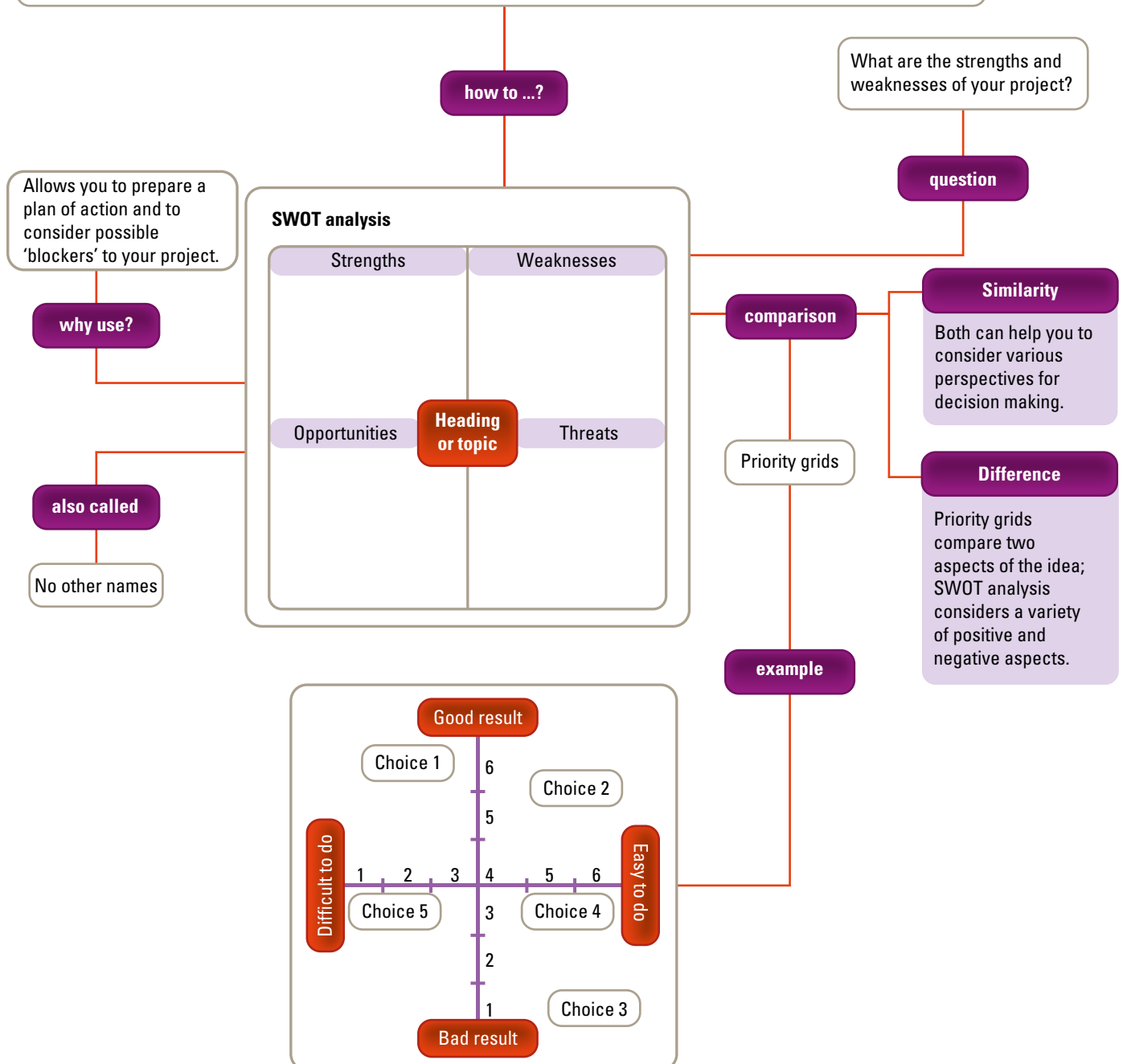
2.8 Big picture science



Carl Woese relied on RNA sequences rather than structural features to determine evolutionary relationships among prokaryotes. He discovered that prokaryotes were actually composed of two very different groups: bacteria (cyanobacteria and heterotrophic bacteria) and archaea (halophiles and thermophiles).

SWOT analyses and priority grids

1. Draw up a square and divide it into four quarters. In the centre of the diagram write down the topic or issue that you are going to analyse.
2. Think about or brainstorm the positive features and behaviours, and record them in the Strengths section.
3. Think about or brainstorm the negative features and behaviours, and record them in the Weaknesses section.
4. Think about or brainstorm possible opportunities and record them in the Opportunities section.
5. Think about or brainstorm possible threats and record them in the Threats section.



STUDY CHECKLIST

DNA, GENES AND CHROMOSOMES

- ☐ describe the structure of DNA
- ☐ outline the process and importance of DNA replication
- ☐ relate the structure of DNA to its function
- ☐ distinguish between alleles and genes
- ☐ state the relationship between DNA, genes and proteins
- ☐ define 'karyotype' and describe its use
- ☐ explain how the gender of a baby is determined
- ☐ compare processes of mitosis and meiosis
- ☐ define the terms 'mutation' and 'mutagen'
- ☐ explain how mutations can reduce an organism's chance of survival
- ☐ identify examples of how mutations can be beneficial
- ☐ identify three different types of mutation

INHERITANCE

- ☐ outline the role that DNA plays in inheritance
- ☐ distinguish between genotypes and phenotypes
- ☐ define the following terms: dominant, recessive, heterozygous, homozygous
- ☐ explain how both genetic and environmental factors determine phenotypes

- ☐ predict the outcome of genetic crosses using Punnett squares
- ☐ interpret pedigree diagrams

GENETIC APPLICATIONS

- ☐ identify applications of DNA fingerprinting
- ☐ outline the procedure to create transgenic species
- ☐ discuss issues relating to the positive and negative impacts of gene technology and cloning

SCIENCE AS A HUMAN ENDEAVOUR

- ☐ investigate the history and impact of developments in genetic knowledge
- ☐ suggest how values and needs of contemporary society can influence the focus of scientific research
- ☐ describe how science is used in the media to justify people's actions
- ☐ use knowledge of science to evaluate claims, explanations and predictions
- ☐ recognise that financial backing from governments or commercial organisations is required for scientific developments and that this can determine what research is carried out

eBookplus

DIGITAL RESOURCES



ANSWERS for this chapter can be found online in your eBookPLUS.

Online section

This section of the chapter can be found online in your eBookPLUS.

2.13 Thinking tools: SWOT analyses and priority grids

eBookplus
ONLINE ONLY

Individual pathways

Activity 2.1

Revising genetics

doc-8464

Activity 2.2

Investigating genetics

doc-8465

Activity 2.3

Investigating genetics further

doc-8466

FOCUS activity

Access more details about focus activities for this chapter in your eBookPLUS.

doc-10670

eLessons

Ancient resurrection

Do we have the right to resurrect ancient species? Watch an ABC *Catalyst* video to find out more.

Searchlight ID: eles-1070

Rosalind Franklind and Watson and Crick

Watch a video from *The story of science* about the development of the DNA model.

Searchlight ID: eles-1782

DNA and Hiroshima

Watch a video from *The story of science* about DNA.

Searchlight ID: eles-1781

Interactivities

Mitosis and meiosis

Use this interactivity to test your knowledge of the different processes of cell division and challenge yourself to see if you can differentiate between mitosis and meiosis.

Searchlight ID: int-0680

Making families

Challenge yourself to complete the family — mother, father and offspring — that demonstrates each inheritance as it appears on screen.

Searchlight ID: int-0681

Complementary DNA

Construct a replicate DNA strand by dragging the correct complementary base into sequence.

Searchlight ID: int-0133



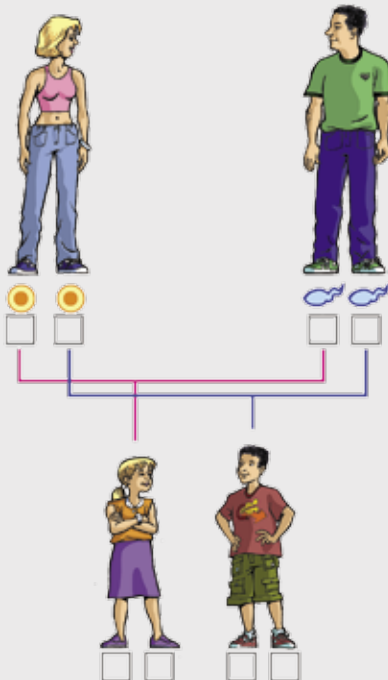
LOOKING BACK

- 1 Arrange the sentence fragments below to complete the sentence that has been started for you.

is made up of	which are made up of
cells	chromosomes
DNA	which contain
which contain in the nucleus	genes

A living organism _____

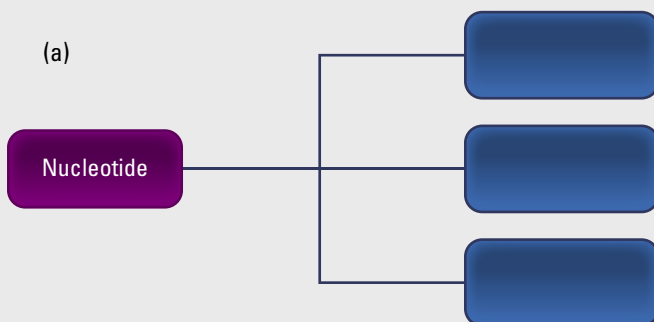
- 2 Suggest the missing sex chromosome labels for the figure below.



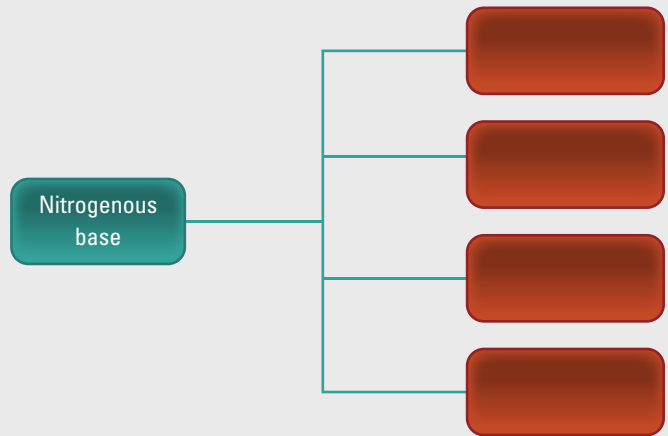
- 3 Copy and complete the following linked figures using the terms in the box below.

homozygous dominant	phosphate
cytosine	heterozygous
sugar	thymine
nitrogenous base	homozygous recessive
adenine	guanine

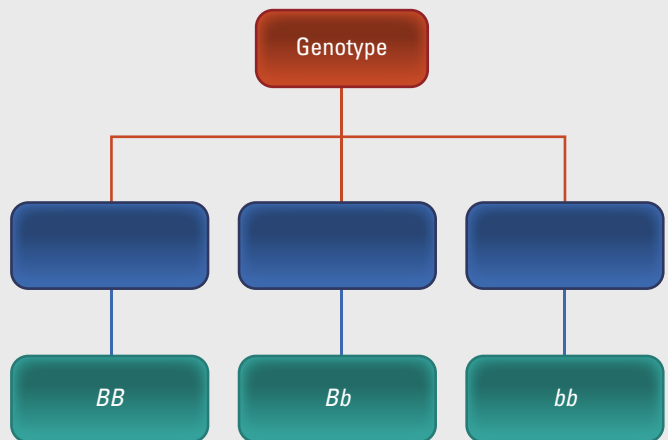
(a)



(b)

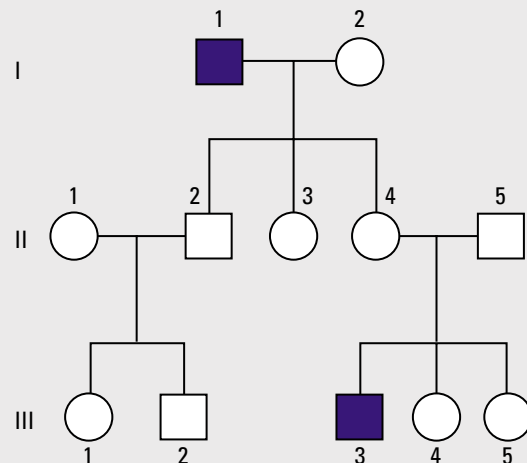


(c)

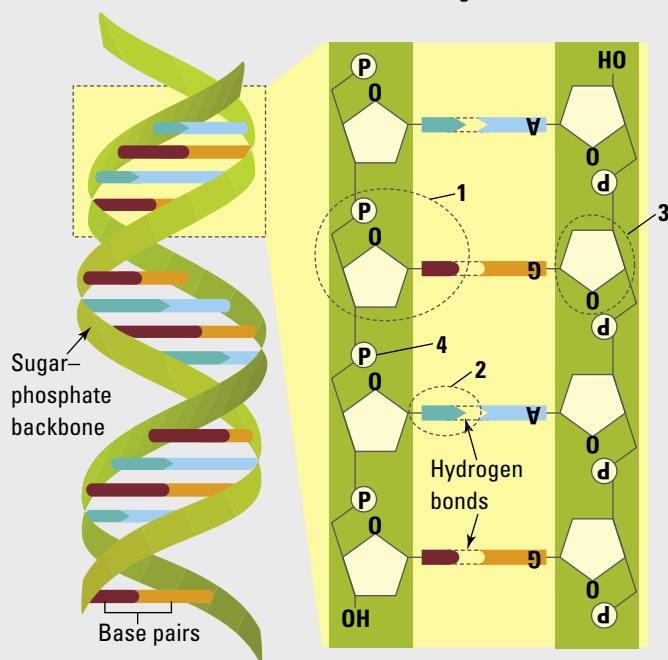


- 4 Use the *Some rules for drawing pedigree charts* diagram in section 2.7 and the cystic fibrosis pedigree chart below to determine:

- which type of inheritance is responsible for cystic fibrosis
- if individual II-3 is a male or a female
- for each individual I-1, I-2, II-4, III-3 in the CF pedigree, whether you think they have cystic fibrosis or are carriers of cystic fibrosis. Give a reason for your suggestion.



- 5 A diagram representing a DNA molecule is shown below. Which row in the following table shows the correct names for the structures labelled 1–4 in the diagram?



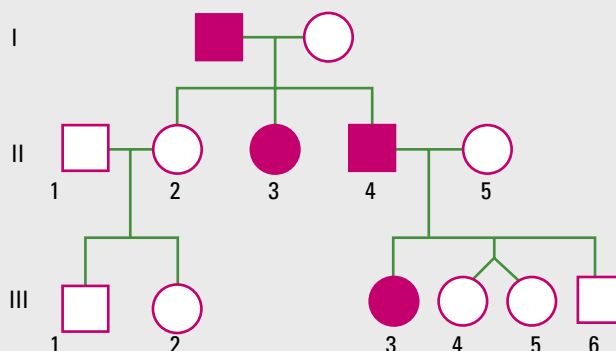
	Part 1	Part 2	Part 3	Part 4
A	Nucleotide	Nitrogenous base	Phosphate group	Sugar
B	Chromosome	Nucleotide	Adenine	Cytosine
C	Nitrogenous base	Sugar	Phosphate group	Polypeptide
D	Nucleotide	Nitrogenous base	Sugar	Phosphate group

- 6 (a) Using the letters *B* or *b* to represent the gene for coat colour, predict the results of the following crosses. Draw diagrams or Punnett squares to show your predictions.
- A pure-breeding (homozygous) black mouse with a hybrid (heterozygous) black mouse.
 - A pure-breeding black mouse with a pure-breeding white mouse.
- (b) Is black dominant to white or white to black? Support your answer.
- 7 The pedigree chart above right shows the inheritance pattern of Huntington's disease. This disease is due to a dominant HD gene on chromosome 4.
- How many generations are shown?
 - How many females are in the pedigree?
 - How many males are in the second generation?
 - Identify three individuals who have Huntington's disease.
 - If *H* represents the allele for Huntington's disease, state the genotypes of:
 - individuals 1 and 2 in the first generation
 - individuals 2 and 4 in the second generation.

- (f) How would the pattern in the pedigree be different for a recessively inherited trait?

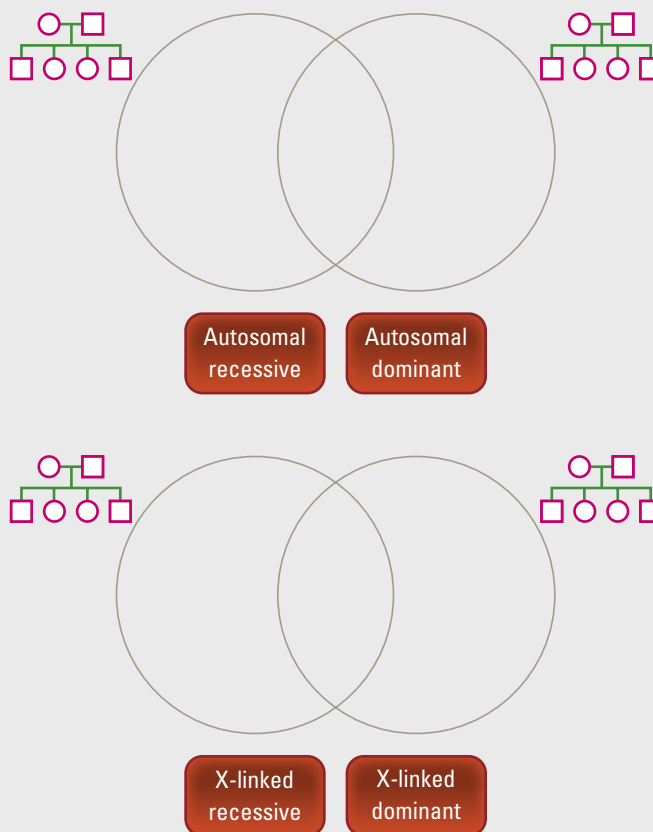
Key

	Normal male		Affected female
	Affected male		Normal female
	Identical twin	1, 2, 3, etc.	Sequence of individuals

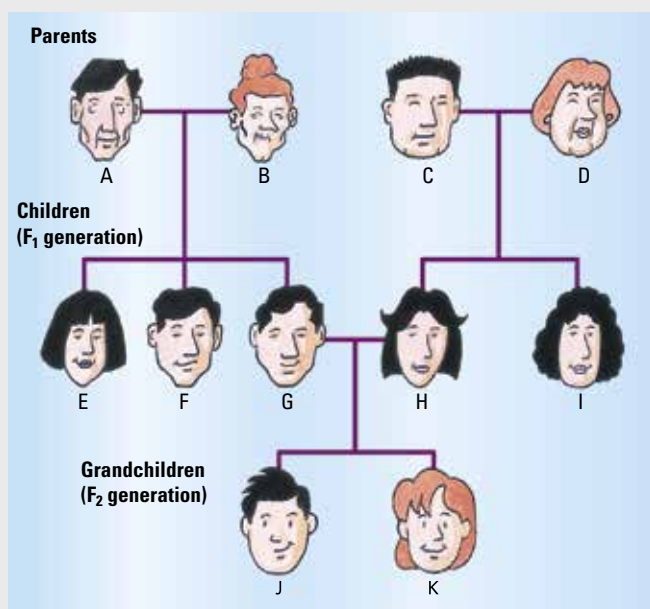


Pedigree chart showing the inheritance pattern of Huntington's disease

- 8 Construct Venn diagrams and add shading to pedigrees like those shown in the figures below to illustrate the similarities and differences between each of the following types of inheritance.



- 9 Examine the pedigree chart below. Let the dark hair allele be represented by B and the red hair allele by b .



- Write the genotypes for the individuals B, G, H and K.
- Write the phenotypes for the individuals F and D.
- If individuals G and H had another child, what is the chance that it would have red hair?
- If individuals F and I had a child, do you think it might be possible for it to have red hair? Explain your reasoning.

- 10 (a) For each of the following statements, rate your opinion on the scale shown below:

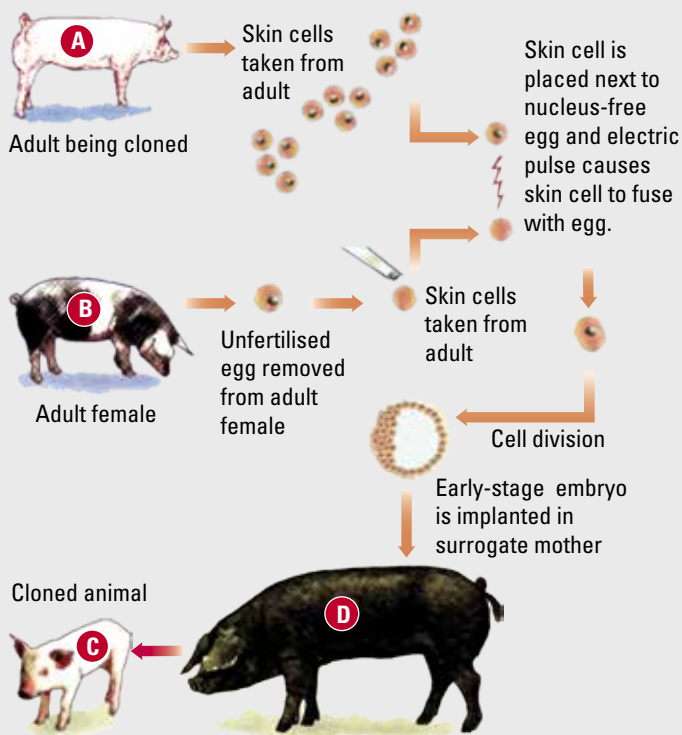
0	1	2	3
(Strongly disagree)	(Disagree)	(Agree)	(Strongly agree)

- Insurance companies and employers should have access to your genetic information.
 - Once the tools for genetic engineering are available, everyone should have access to them.
 - You should be able to use gene technology to select specific characteristics of your future children.
 - Before dating, you should exchange all of your genetic information with your potential partner.
- Select and discuss one of the statements with a partner, constructing a PMI chart to summarise the key points.
 - Share your PMI chart with that of another pair or team. Add any points that you consider to be worthwhile and appropriate.
 - On the basis of the discussions with your peers, would you change your initial scaled opinion or is it exactly the same? Comment.

- 11 (a) Create a concept map that uses as many of the terms below as you can.
- (b) Select at least five terms and create a poem or song to help teach others how the terms are linked or connected. Share your creative piece with others in your class.

autosome *sex linked* sex chromosomes allele
 dominant trait gene *autosomal recessive*
 pedigree Punnett square genotype DNA
 heterozygous homozygous chromosomes phenotype
 cell prokaryotic eukaryotic ova
 nucleus nucleic acids RNA transcription
 translation protein ribosome messenger RNA
 transfer RNA gametes sperm pure breeding haploid
 mitosis diploid genetic engineering
 fertilisation restriction enzymes electrophoresis
 zygote hybrid DNA probe vector transgenic
 ligase enzymes

- 12 Use the following diagram to decide which of the statements below is correct.



- Pig A and pig C are genetically identical.
- Pig D and pig C are genetically identical.
- Pig B is the surrogate mother of pig C.
- None of the pigs are genetically identical as the environment in which the pigs grow can affect their genotype.

- 13 Use the **New genetic test** weblink in your eBookPLUS to read the article *New genetic testing technology for IVF embryos* and answer questions 13–16.

eBookplus

- What does PGD stand for?
- What does the PGD test identify in embryos? Include four specific examples in your response.
- Outline the opportunity that this test offers families with histories of genetic disorders.
- Outline a negative aspect of the PGD test.
- At which stage is the embryo when a single cell is removed from it?
- Are you aware of any bias in the article? How many different perspectives were included? If you were to write the article, what other information or details might you include?

- 14 (a) In a team, construct a SWOT analysis on pre-implantation genetic diagnosis.
 (b) Share your SWOT with another team and discuss any similarities and differences between your SWOTs.
 (c) On the basis of your discussions, construct your own SWOT on the article.
- 15 In your team, brainstorm statements or choices related to genetic testing of embryos. Select five of these statements and position them on a priority grid with the following labels.

Horizontal

Left-hand side — difficult decision
 Right-hand side — easy decision

Vertical

Top — good result
 Bottom — bad result

- 16 Find out more about two of the genetic disorders listed in the article. Share and discuss your findings with your team or class members.
- 17 Genetic tests can be ordered on the internet!
 (a) Find out about examples of tests that are available.
 (b) Suggest some implications of being able to buy genetic tests in this way.
 (c) Is the unregulated sale of genetic tests potentially misleading or unethical? How can the accuracy and privacy of these tests be ensured?
 (d) Who owns the information of genetic tests ordered on the internet? Comment on your findings.
 (e) Construct your own SWOT analysis of your findings, and then share it with others in your class.
- 18 Use the **Clone licence** weblink in your eBookPLUS to read the article *Australia issues first licence to clone human embryos*, and then answer the following questions.
 (a) State the number of human eggs that Sydney IVF was granted licence to access.
 (b) Is human cloning for reproductive purposes allowed? Explain.
 (c) State the stage of development to which the embryos are allowed to develop.

eBookplus



A single cell being extracted from an eight-cell embryo

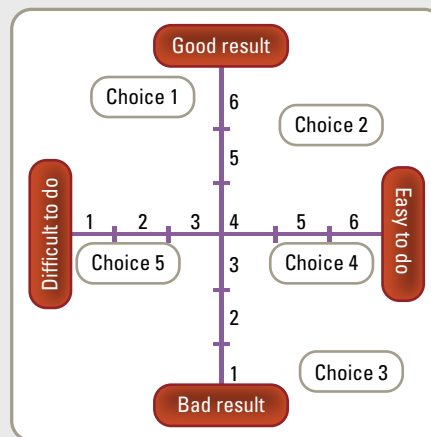
- Find out more about therapeutic cloning or somatic cell nuclear transfer.
- Identify issues related to therapeutic cloning.
- Use a SWOT analysis to categorise points made on a discussion on therapeutic cloning.
- Identify possible uses of different types of cloning.
- Construct a priority grid to map out different potential uses of different types of cloning.

- 19 Use the **Religious views on cloning** weblink in your eBookPLUS to answer the following questions.

eBookplus

- Which religions have the most favourable views towards cloning? Explain why.
- Which religions are most against cloning? Explain why.

SWOT analysis



eBookplus

work sheets

- 2.9 Getting into genes: Puzzle
 2.10 Getting into genes: Summary

projectsplus

The gene lab

SEARCHLIGHT ID: PRO-0111

Scenario

Think of how different dog breeds such as chihuahuas, great danes, dachshunds, blue cattle dogs and dobermans are from each other. Yet all of our pet dog breeds — regardless of size, colour, coat and intelligence — are still members of the same species. All dogs are descended from a long-gone species of wolf. Over the thousands of years that they have been humankind's companions, we have selectively bred dogs together so that particular characteristics became more pronounced while others faded out. For example, greyhounds with their long graceful legs were bred for speed while bull-mastiffs were bred for their size and strength. Over time, these characteristics became fixed in that breed. The breeding process is continuous, with new breeds being registered with the International Federation of Dog Breeders every few years. Dog breeders try to produce dogs that are the ideal examples of their breed, and do so by carefully selecting which dogs to mate. Unfortunately, in their quest to establish these perfect examples, the dogs produced may inherit genetic disorders as a result of unfortunate genetic combinations or inbreeding. Purebred labradors, for example, may develop hip dysplasia, knee problems and eye problems such as progressive retinal atrophy which — as well as preventing the dog from being

shown in competitions — have serious effects on the dog's quality and length of life. Now that genetics and DNA are more fully understood, it is not uncommon for dog breeders to consult with genetic scientists to ensure that the puppies they produce have the smallest risk of developing these disorders.

Your task

You are part of a team of vets that works for the Dog Breeders Association of Australia as genetic counsellors. Your client has a labrador bitch that has a family history of progressive retinal atrophy — a condition that causes gradual blindness. The client would like to breed her to produce for show as many puppies as possible that do not carry the gene for the disorder. There are three available stud dogs that the bitch can be mated with. Given the pedigree of each of these dogs, you must determine which of them should be selected to sire the litter. You will give your recommendations to the client in the form of a genetic report explaining your decision — including family trees, phenotype and genotype identification, and final breeding recommendations.

Process

Open the ProjectsPLUS application for this chapter located in your eBookPLUS. Watch the introductory video lesson and then click the 'Start Project' button to set up your project group.

