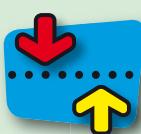


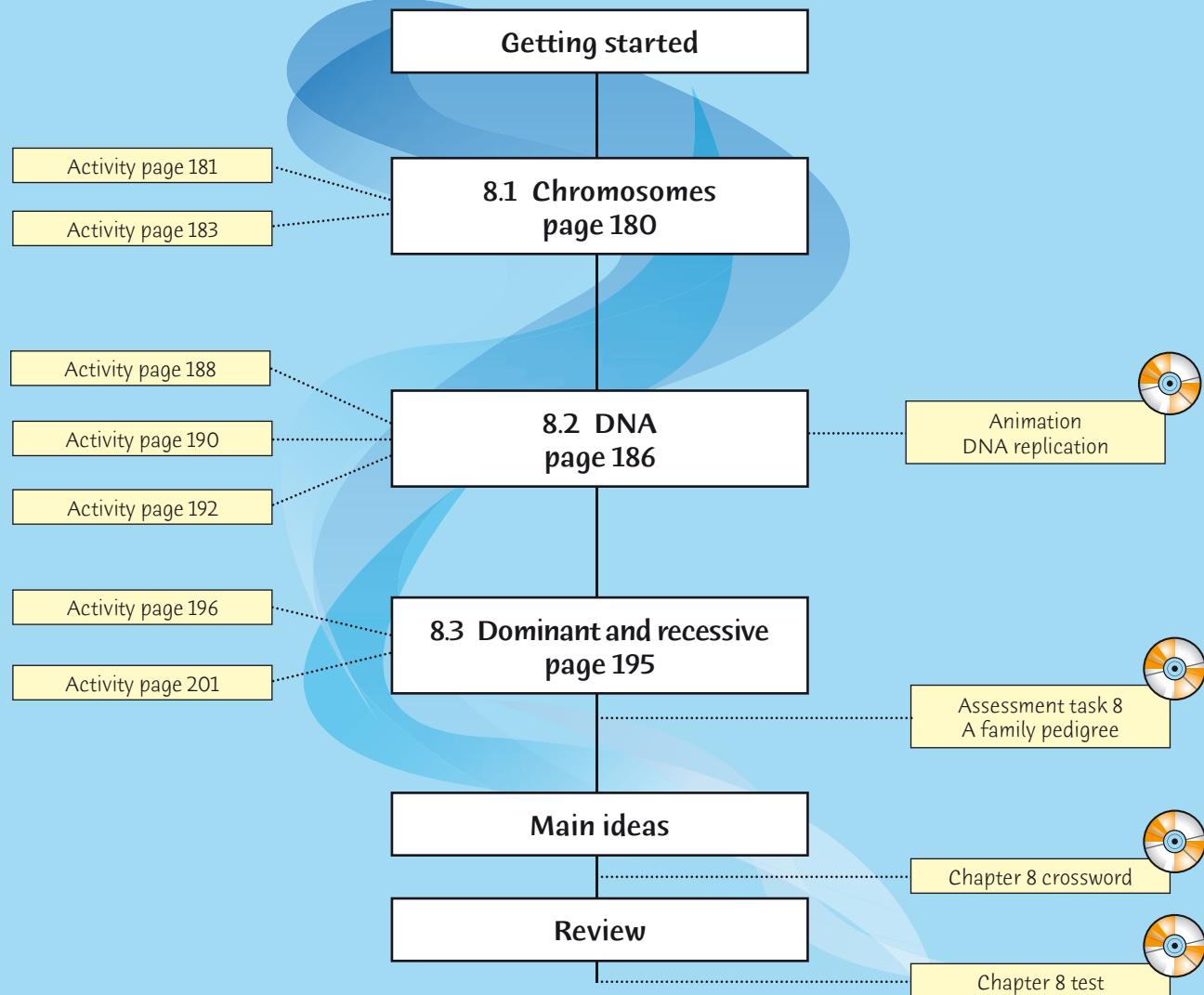
8



Inheritance



Planning page



Essential Learnings for Chapter 8

| Essential Learnings | References | | |
|--|--|---------------------------|---|
| | Student book (page number) | Workbook (page number) | Teacher Edition CD (Assessment task) |
| Knowledge and understanding <i>Life and living</i> All the information required for life is a result of genetic information being passed from parent to offspring | pp. 180–204 | pp. 61–66 | Assessment task 8 A family pedigree |
| Science as a human endeavour Responsible, ethical and informed decisions about social priorities often require the application of scientific understanding | Activity p. 192 Science in action pp. 193, 204 | | |
| Ways of working Communicate scientific ideas, explanations, conclusions, decisions and data, using scientific argument and terminology, in appropriate formats | Punnett squares pp. 197, 200 Pedigrees pp. 198, 202–203 | pp. 64–65 | Assessment task 8 A family pedigree |
| Reflect on learning, apply new understandings and justify future applications | Activity p. 192 | | |
| Research and analyse data, information and evidence | | pp. 59–60 p. 66 | Assessment task 8 A family pedigree |

QSA Science Essential Learnings by the end of Year 9

Vocabulary

albino
alleles
amniocentesis
chromosomes
deoxyribonucleic acid (DNA)
dominant
foetus
genes
genome
genotype
heterozygous
homozygous
meiosis
melanin
mitosis
mutation
nucleotides
pedigree
phenotype
Punnett square
recessive
replication

Focus for learning

Discuss what DNA is and how it can be used in crime scene investigations (page 179).

Equipment and chemicals (per group)

- Activity page 181 pieces of pipe cleaners (1 long white, 1 long green, 1 medium white, 1 medium red, 1 short white, 1 short blue)
- Activity page 183 11 small plastic discs, felt pen
- Activity page 188 18 paperclips of various colours, piece of cardboard (25 cm × 12 mm)
- Activity page 190 model strand of DNA from previous activity
- Activity page 196 20 black discs and 20 red discs (or 40 coins, half of them marked)

Special preparations

- Activity page 181 To colour the pipe cleaners, soak them in food colouring overnight, rinse, and allow them to dry. PVC-coated electrical wire can be used instead of pipe cleaners.



8

Inheritance



Getting Started

Work in a group of 3 or 4 and discuss the following questions. Keep your answers for later in the chapter.

The forensic scientists in the photo are investigating a crime scene. They are looking for



sources of DNA from the victim and also from the suspect.

- 1 What is DNA and where is it in your body?
- 2 What would be good sources of DNA at this crime scene?
- 3 Suggest how a suspect could be convicted using DNA.
- 4 If you were a forensic scientist what other evidence would you gather to help convict the suspect?

Starting point

- 1 Before starting this chapter, it is a good idea to check school records to see if any student in the class is adopted, fostered or experiencing a family trauma. Be aware of and sensitive to their feelings and needs. If students are aware of the circumstances of their fellow student(s), encourage acceptance and sensitivity.
- 2 A fun way to start the chapter is to set up the room as a mock crime scene. Have strewn on a bench items such as a broken glass bottle, some cloth stained reddish-brown and splatters of red liquid, two drinking glasses with lipstick or fingerprints on them, strands of hair and so on. Around the bench you could have a few muddy footprints. In teams, students could generate a set of questions that a forensic scientist might need to ask to help solve the crime. What methods could forensic scientists use to answer the questions? Lead the class into discussing the Getting Started questions. Make sure students try to explain what they think DNA is.
- 3 Students could skim through this chapter and jot down about 10 questions related to inheritance. Allow them to use the section headings as pointers to help generate their questions. At the

completion of the chapter, students could see how many of these questions they can answer. They will probably be delighted to discover they know most of the answers (if not all of them).

- 4 Compile a class list of words relating to inheritance, and develop a concept map or glossary. For a concept map, students could choose whether to draw pictures or write linking sentences. A glossary could be in the form of an audio file so that it can be played with an MP3 or MP4 player. A possible word list is: *inheritance, cell, chromosome, sperm, ovum, meiosis, mitosis, DNA, bases, gene, mutation, genome, dominant, recessive, alleles, pedigree, genotype, phenotype*. Students should have prior knowledge of some of the words in the list as they were covered in *ScienceWorld 2*, Chapter 6. They may like to use the glossary in the back of the textbook to help them. It will be helpful for you to see how many correct links students can make. This activity could form the basis of a pre-test.

Hints and tips

Reinforce that chromosomes are usually only seen when a cell is dividing, and the two new cells formed each get one of every chromosome. Because the chromosomes duplicate during cell division, each new cell (daughter cell) contains exactly the same number of chromosomes as the original cell (parent cell). The process is repeated billions of times over, with each of the cells containing an identical set of chromosomes. This type of cell division is called mitosis and may be worth revising (*ScienceWorld 2* page 121).

Learning experience

Generate a class list of questions to be investigated individually.

Although teamwork is an important part of learning, working individually is just as valuable.

Consider starting the question list with the following:

- Do all organisms have the same number of chromosomes?
- Do individuals of the same family have the same number of chromosomes? For example, do all cats (leopards, lions, domestic cats, etc) have the same number?
- If an organism has a higher number of chromosomes than another organism, does this mean it is more intelligent?

If all humans have 23 pairs of chromosomes, what makes each person unique? That is, why are we not identical replicas? What is going on scientifically? Turn this into a higher order thinking task and allow students enough time to work through their ideas by brainstorming and inquiring/investigating.

Students may discover through their investigation that some humans, in fact, have more or less than 46 chromosomes. What happens here? Are they still human?

Give students the initial question at the beginning of the lesson, and say that you will give them more time at the end of class to review their response if new concepts have been covered. Students could use the jigsaw method, or some other cooperative learning tool, to help with their investigation.

8.1 Chromosomes

Long ago the Ancient Egyptians recognised that the racing characteristics of greyhounds were inherited. It is believed that they selected parent dogs that were strong and fast runners, and bred them to produce young dogs with these characteristics. By selectively breeding they produced dogs that were highly valued in Egyptian society.

Greyhounds reproduce sexually. That is, the male produces sperm and the female produces eggs. A new individual forms when the nuclei of the sperm and egg join together. After fertilisation, the egg divides and the new organism gradually grows in size.

It is reasonable to assume that the sperm and the egg must contain all the instructions for the cells to make the new organism.

Chromosomes and inheritance

At the turn of the 20th century, many scientists wanted to find out what part of an organism's cell was responsible for passing on characteristics from one generation to another. An American biologist, Walter Sutton, provided one of the earliest clues to the problem. He wondered about the importance of sausage-shaped objects that had been observed in cells undergoing division. He noticed that the rounded nucleus in a cell seemed to change into these sausage-shaped objects just prior to cell division, and each of the new cells contained these objects. These objects we now call chromosomes.

Sutton also observed that a particular species of organism always had the same number of chromosomes, and that the number of chromosomes was different for different organisms.

Sutton chose fruit flies for his experiments because they had only 8 chromosomes in their cells and they bred very quickly. On the basis of his experiments, Sutton inferred that the chromosomes carry the inherited characteristics and that each organism has a particular number of chromosomes.

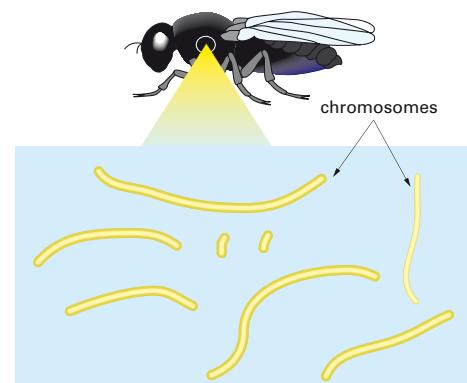


Fig 2 The chromosomes in the cells of a fruit fly carry all of its inherited characteristics.

Chromosomes in cells

In *ScienceWorld 2* you learnt that chromosomes can be seen only during cell division, and the two new cells that are formed each get one of every chromosome. This means that the new cells contain *exactly* the same number of chromosomes as the original cell.

Chromosomes are usually in pairs. This is why the numbers in the table below are even numbers. For example, humans have 23 pairs, a fruit fly has 4 pairs and corn has 10 pairs.

In the diagram on the next page, the 46 chromosomes in a human have been placed in pairs from largest to smallest (except the X chromosomes). Notice that each chromosome is roughly H shape. This is because each chromosome strand is duplicated and the strands are joined at one place.

| Organism | Number of chromosomes |
|-----------|-----------------------|
| human | 46 |
| mouse | 40 |
| fox | 34 |
| fruit fly | 8 |
| pea | 14 |
| corn | 20 |
| chicken | 78 |
| algae | 148 |

Learning experience

Storyboards show a sequence of ideas, procedures or events in a pictorial format. They are similar to flow diagrams but use pictures rather than written information. Ask students to make a storyboard about chromosomes, particularly relating to humans. The board could be added to progressively to include new information about DNA, genes, genomes and so on.

Research

All living things have chromosomes. The chromosomes in each organism are unique and different in number. Ask students to research and list the number of chromosomes some organisms have. Can they find out any interesting information about chromosome numbers?

It is important to remind students to check if the researched number indicates how many pairs and, if so, the number has to be doubled to give the total number of chromosomes.

Hints and tips

- Humans normally have 46 chromosomes (23 pairs) in all cells except the gametes (sex cells). The process of cell division of sex cells is called meiosis. When the sperm and ovum join during fertilisation, a single cell (zygote) is formed, with 46 chromosomes (23 pairs). Because this zygote has the full number of chromosomes, it has the ability to develop into a new organism. Half of each chromosomal pair is inherited from the mother's egg (ovum) and the other half from the father's sperm.
- The arrangement of the set of chromosomes in the cell of an organism is called its karyotype. The chromosome pairs are usually ordered according to their size, leaving the pair containing the sex-determining gene last. Generally, the largest chromosome is referred to as chromosome 1, the next largest as chromosome 2, and so on.
- The karyotype of the human female contains 23 pairs of homologous chromosomes (22 pairs of autosomes and one pair of X chromosomes). See Fig 3. A male human has the same 22 pairs of autosomes but one X chromosome and one Y chromosome. See Fig 7 on page 183.

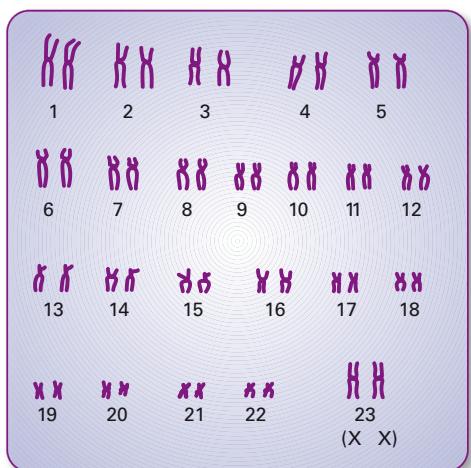
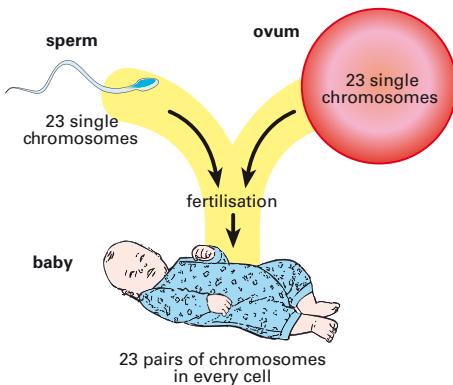


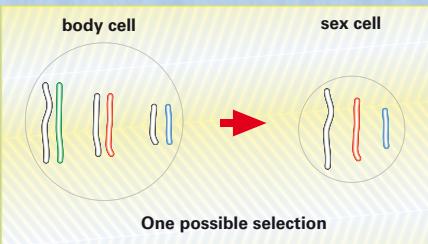
Fig 3 The 46 chromosomes in a human female are arranged in pairs based on size. The pair numbered 23 contains the sex-determining genes.

The nuclei of sex cells (sperm and eggs) contain only *half* as many chromosomes as the nuclei of all other cells. Why is this? The formation of sperm and ova occurs by a special type of cell division during which the chromosome pairs separate. This means that each sex cell receives 23 *single* chromosomes. When the nuclei of the sperm and egg join during fertilisation, the new cell then contains 23 *pairs* of chromosomes.

**Activity****Chromosomes in sex cells**

In this activity pipe cleaners will represent chromosomes. You will need 3 pairs of pipe cleaners, each pair a different length. One of each pair is white and the other is coloured.

- Place the pipe cleaners in pairs so that you have a white one and a coloured one in each of the pairs.
- Draw a circle at the top of the sheet of paper and lie the 3 pairs of chromosomes in it. This represents a body cell containing 3 pairs of chromosomes.
- Your task is to make sex cells. For this you select one long chromosome, one medium one and one short one. One selection has been done for you.



- How many different sex cells can you make?
Use coloured pencils to draw the three single chromosomes in each of the sex cells.

Learning experience

With the exception of sex cells (sperm and ovum), all human cells—skin, nerve, muscle, fat cells, and so on—have 23 pairs of chromosomes, made up of 23 different chromosomes and 23 copies. Ask students to explain why sex cells have only 23 chromosomes and no copies. They should respond to the question using the appropriate language outlined in this chapter and be allowed to choose how to present their information.

Learning experience

Give students a word list and ask them to find each word's meaning. They should then write a simple definition using suitable language so that anyone learning genetics for the first time will understand the word's meaning. Encourage students to draw diagrams to help with their explanations. Use a combination of words from this chapter and some new words to look up, such as *gamete*, *diploid cell*, *haploid cell*, *zygote*, *homologous pair*, *karyotype*, *centromere*.

Activity notes

- Pipe cleaners can be bought from an art supplies company or retail art supplies outlet.
- A plastic petri dish makes a good representation of a cell, or could be used to trace around for their circles.

Learning experience

What is the chance of there being another person just like you? Extend students' thinking and reasoning, especially those who are gifted or mathematically able. Ask them to work out what chance there is of the same two parents having identical offspring (not including those from multiple births). They could draw/write a sequence of diagrams/steps showing how they arrived at their conclusion. (Answer: For a human, the chance is 1 in over 70 million million: 2^{46})

Hints and tips

There are many commercial DVDs on genetics that can be shown during the chapter. Make sure to preview any before you show them to determine how appropriate they are, and prepare worksheets for the students to fill in. There are also some great YouTube videos showing meiosis. Go to <www.youtube.com> and search for ‘meiosis’, then preview the material before showing the class.

Learning experiences

- 1 Make sure students can clearly differentiate between mitosis and meiosis. They could come up with their own mnemonic, jingle, poem or limerick which explains meiosis.
- 2 Students could make mobiles or bookmarks showing the process of meiosis. Each diagram should be neatly labelled, explaining each step of the process. Hang the mobiles around the room. Bookmarks would provide students with a revision tool, especially for this chapter.



Formation of sex cells

When your body cells divide, each new cell has the same number of chromosome pairs as the parent cells. In humans this is 23 pairs. These cells are referred to as *diploid* cells: *di* means two and *ploid* refers to the chromosome number. So in your body most of the cells are diploid and have two copies of each chromosome. In *ScienceWorld 2* you learnt that this type of cell division is called *mitosis*.

When sex cells are formed they have only one chromosome of each pair. These cells are called *haploid* cells. This means the cell division process has to be different from mitosis. The process of sex cell formation is called *meiosis* (my-OH-sis). Meiosis occurs only in special cells in the testes and ovaries in animals, and in the anthers and ovaries in plants.

In mitosis two daughter cells are produced in each division. However, in meiosis four different sex cells are produced, as shown the diagram on the right and the animation.

Questions

- 1 Meiosis can be thought of as having two cell divisions. Describe how the first division is different from the second division.
- 2 There is another way the chromosomes can be arranged in line 3 of the diagram. Using red and blue pens, draw this arrangement. Draw the sex cells this arrangement will produce. How is it different from those in the diagram?
- 3 Imagine the blue chromosomes originally came from the organism's mother. What is the chance that a sex cell could carry all of the mother's chromosomes? Justify your answer.



To see an animation of this, open the animation *Making sex cells* on the CD.

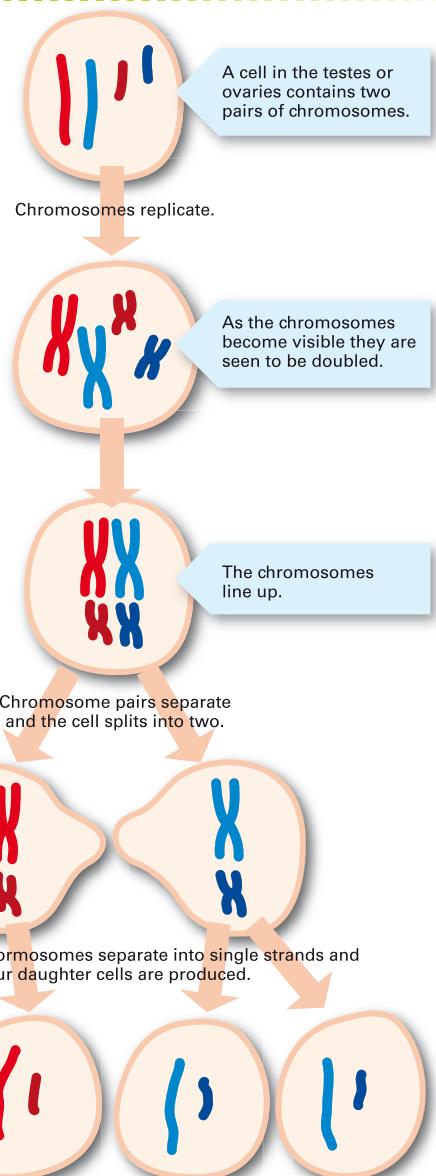


Fig 6

The process of meiosis produces sex cells with half the number of chromosomes of a body cell.

Learning experience

Ask students what they think could happen next after meiosis. Do the four daughter cells divide further? If so, how? If fertilisation takes place between the ovum and sperm to form a zygote, what kind of cell division takes place after this?

Develop a set of critical thinking questions about meiosis. Students could work in pairs to problem-solve and then exchange ideas with the rest of the class. Can students successfully think critically and apply concepts in a wider context? Questions could be asked using the following critical thinking/Socratic thinking model.

| Clarification | Assumptions | Reasons and evidence |
|--|--|--|
| What is meiosis? Do all living things undergo meiosis? How does this relate to our body? What happens in the process of meiosis? Where does meiosis occur in the body? Why do cells undergo meiosis? What might happen next after meiosis? | Have any assumptions been made? Why would someone make that assumption? | What would be an example of meiosis? How do you know when the process has occurred? |

| Viewpoint or perspective | Implications | Questions about the questions |
|--|---|--|
| What effect does this kind of cell division have on the body? Does the body have an alternative to meiosis? | If cell division goes wrong, what implications are there? Are there any ethical implications of meiosis? | This section is for further questions that might be asked. |

Determining sex

The chromosomes on the right are from a human male. How are they different from the chromosomes in a female in Fig 3 on page 181?

Notice that chromosome pair 23 in the male is different from that in the female—the male has two quite different chromosomes. The larger one is called the X chromosome and the smaller one is the Y chromosome. These chromosomes are called *sex chromosomes* because their genes determine the sex of the offspring.

Sperm cells, which are made in the testes, contain either an X chromosome or a Y chromosome, and 22 other chromosomes. On the other hand, ova, which are made in the ovaries, contain an X chromosome and 22 others.

If you are a boy, you would have received a Y chromosome from your father and an X chromosome from your mother. And if you are a girl, you would have received an X chromosome from each of your parents.

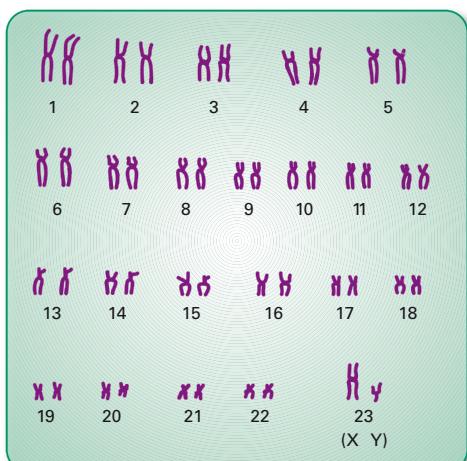


Fig 7 The chromosomes from a human male



Activity

If a couple are having a baby, what are the chances of having a boy? A girl? You can use a model to find out.

You will need eleven small plastic disks for this activity.

- 1 Use a felt pen to write X on five disks and Y on five others. These disks represent sperm.
- 2 Place all ten disks in a box or bag.
- 3 Mark the eleventh disk with an X. This represents an ovum.
- 4 Place the X disk on a piece of paper on the table.
- 5 Then, without looking, select a sperm disk and place it on the table next to the ovum.
- 6 Record whether the result is a boy or a girl.

- 6 Replace the disk and repeat for a total of 10 draws.

7 Calculate the percentage of boys and girls.

- 7 Repeat the procedure for a total of 100 draws.

8 Calculate the percentages of boys and girls. Suggest why these results are different from the result in Step 6.

9 Pool all the class results. Are the results the same as your results?

10 In 1 mL of male semen there are about 40 million sperm. How many would be carrying the Y chromosome? What is the chance that a sperm carrying an X chromosome will be the first to fertilise the ovum?

11 A couple have five daughters. What is the chance that their next child will be a son?

Activity notes

- Explain to students why only one X (female) disk is needed.
- If disks are not available, tossing a coin would work. Assign heads for females and tails for males.

Learning experience

Some human mutations are caused by errors during the formation of zygotes or at some stage in the process of meiosis. Sometimes extra chromosomes are copied, partially deleted or deleted altogether. These errors mean that after fertilisation, the organism has an abnormal number of chromosomes and often spontaneously aborts long before birth. However, sometimes organisms do continue to develop and offspring are born. These offspring may have physical, mental or physiological disorders.

What are some possible chromosomal abnormalities? Students can use the following activity to investigate some karyotypes of chromosomal disorders and then further research the disorder.

Teacher preparation

- Photocopy enlargements (A5 size) of Fig 3 and Fig 7 in the textbook.
- Carefully draw/copy any additional chromosomes or delete some according to the table.
- Delete the chromosome number and photocopy the karyotypes again to make a class set (five copies for each disorder).
- For each karyotype, cut the sheet into single chromosomes and place them into an envelope.
- Alternative examples of human karyotypes with more distinguishable chromosome pairs can be found on the internet. For this activity it is important that every pair is different from every other pair.

Student activity

Give each student an envelope containing a karyotype set and the chromosome disorder table (left). Get them to spread out their chromosomes, pair them up and number them according to Fig 3 and Fig 7. Using the information in the table, they can then identify what type of disorder the karyotype represents. Students could further research the probability of being born with each disorder, if it affects males or females, symptoms of the disorder and treatment.

| Chromosomal disorder | Affected chromosome pair | Common name |
|---|--------------------------|------------------------|
| Trisomy 21 (47XX+21 female, 47XY+21 male) | 21 (one more) | Down syndrome |
| Trisomy 18 (47XX+18 female, 47XY+18 male) | 18 (one more) | Edwards syndrome |
| Trisomy 13 (47XX+18 female, 47XY+18 male) | 13 (one more) | Patau syndrome |
| XXY (47XXY male) | 23 (extra X) | Klinefelter's syndrome |
| XYY (47XYY male) | 23 (extra Y) | XYY syndrome |
| XXX (47XXX female) | 23 (extra X) | Triple X syndrome |
| X (45X female) | 23 (one less) | Turner syndrome |

Issues

Some chromosomal disorders can be tested for during pregnancy. Ask students to imagine they are doctors who have to tell a pregnant woman and her partner that their baby will be born with a chromosomal disorder. How do they think the couple might feel about receiving such news? What would they advise the couple to do? Ask them to explain their answers. If they were the parent, what would they do?

Students could then construct a PowerPoint or another type of multimedia presentation from the perspective of a medical professional, pointing out issues relating to chromosomal disorders.

Check! solutions

- Chromosomes are very long, thin strands that carry information which helps the cell to function properly. They are normally found in the nucleus of the cell. You can see them when the cell is dividing because they become shorter and thicker.
- Sutton used fruit flies for several reasons, such as: they had only 8 chromosomes, they bred quickly, they were relatively easy to keep and observe, and using them did not present any ethical issues.
- Human sperm cells contain either an X or a Y chromosome, whereas in ova the cells all contain an X chromosome.
- You would find only 23 chromosomes in the sex cells. In females these are the ova in the ovaries and in males they are the sperm in the testicles.
- You can assume that Cosmo has red hair but neither his mother nor father does. Probably, the only other person in his family to have red hair is his grandmother. What he says is true because he got the gene for red hair from his grandmother via his parents.
- They are both wrong. The combination of a sperm and ova is a random event and each child has an equal chance of being a boy or a girl. It is just like tossing a coin and watching it come down heads or tails.
- a The ovum of a fox contains 17 chromosomes.
b The pollen of a corn plant contains 10 chromosomes.

Check!

- What are chromosomes? When can you see them?
- Suggest why fruit flies were used in Sutton's experiments and not humans.
- How do the chromosomes in human sperm cells differ from those in ova?
- In humans most cells contain 23 pairs of chromosomes. In which parts of the body would you find cells with 23 single chromosomes?
- Cosmo said to his friend, 'I inherited my red hair from my grandmother'. What does he mean by this?

- The Smiths have three sons. Mrs Smith is pregnant and believes she is having another son. Mr Smith disagrees and says that because they have three sons the next child has a greater chance of being a daughter. Who is correct? Explain your answer.
- Use the table on page 180 to work out how many chromosomes there are in:
 - a fox's ovum.
 - the pollen of a corn plant.
- The father's sperm determines the sex of the child. Is this statement true or false? Give a reason for your answer.
- Look at the table of the number of chromosomes in different organisms on page 180. Notice that they are all even numbers. What is the reason for this?



challenge

- The diagram below shows the chromosomes of a human female. The chromosomes circled are a pair.

Could both of these chromosomes have come from only one of the parents? Explain.



- Is it true to say that the most complex organisms have the greatest number of chromosomes? Use the table on page 180 to justify your answer.

- There are two types of twins in humans—identical twins and fraternal twins.



Fig 8 Identical twins (top) and fraternal twins (bottom)

- This statement is true because each sperm cell will contain either an X or a Y chromosome. Because all ova cells contain an X, it is the sperm which determines whether the new cell (called a zygote, pronounced 'ZIE-goat') is male or female.
- The reason that they are all even numbers is that they are all present as pairs.

Challenge solutions

- No, all organisms get one of each pair from each parent. This is because each sperm and ova cell contains one of each pair of chromosomes.
- No, it is not generally true to say this. Humans are the most complex of organisms and have 46 chromosomes, whereas chickens have 78 and algae have 148.

Identical twins form when an ovum splits into two just after a sperm fertilises it. Fraternal twins form when two ova are fertilised by two different sperm.

- Explain in terms of chromosomes why identical twins are always the same sex, yet fraternal twins can be the same or different sex.
 - Explain why one member of fraternal twins can have blue eyes while the other has brown eyes, yet identical twins both have the same eye colour.
- 4 In the formation of sex cells in the testes or ovaries (meiosis), the pairs of chromosomes line up in the middle of the cell. Then each member of the pair separates and moves apart. A new cell membrane forms down the middle and separates the two daughter cells.

Sometimes a pair of chromosomes may not separate, resulting in one sex cell with too many chromosomes and the other with too few. Using X and Y chromosomes only, explain, using diagrams, how a person's cells could contain three sex chromosomes, XXY.

- 5 The person in the photo below has Down syndrome, a genetic disorder affecting about one in every 600 births. Children born with this disorder often have a lower than normal immunity to disease. Before antibiotics and other treatments became available, many of these children died when they were very young. People with Down syndrome have one extra chromosome in their cells.



Use the diagram in Fig 11 at the top of the next column and the ones on pages 181 and 183 to work out:

- which chromosome pair is affected.
- whether the person whose chromosomes are shown in Fig 11 is a male or a female.

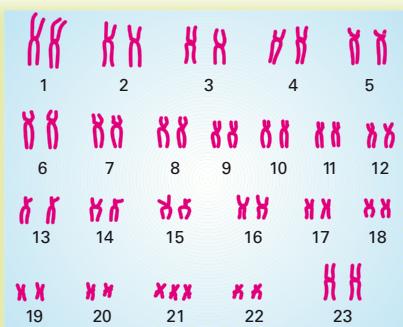


Fig 11 Chromosome map for Challenge 5

6 Use the information above and in Challenge 5 to suggest how a person could be born with Down syndrome.

7 Not all the features of organisms are controlled by genes. Some are influenced by factors in the environment experienced when the young organism is growing.

- What environmental factors might affect the features of humans?
- During which part of a human's life would these environmental factors have most influence?
- The blue flowers in the photo below were from a hydrangea plant grown in acidic soil. The red flowers were produced from a cutting of the same plant grown in basic soil.

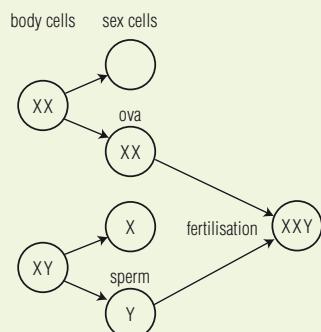
What other environmental factors might affect the features of a plant?



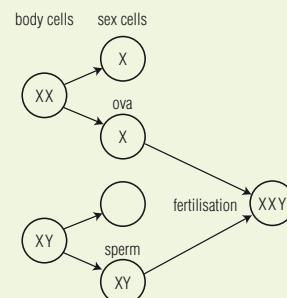
Fig 12 Hydrangea flowers grown in acidic soil (blue) and basic soil (red)

- 3 a Identical twins are always the same sex because they develop from the same fertilised ovum and will both have identical chromosomes. If they are boys they will both have XY chromosomes, and if they are girls they will have XX chromosomes. Fraternal twins are just like normal brothers and sisters except they are born on the same day.
- b Identical twins come from the same ovum and sperm and therefore have identical genes. This means that their eye colour is the same. Fraternal twins come from different ova and sperm and will have different characteristics.

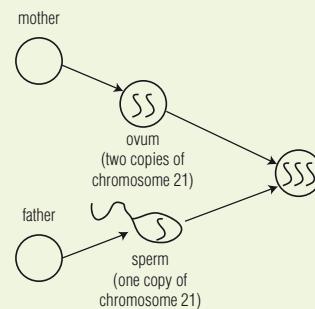
- 4 There are two possibilities, as shown below. It may be that there are two X chromosomes in a single ovum, which then combine with a normal sperm.



It is also possible that there are both an X and a Y chromosome in a sperm, which then combines with a normal ovum.



- 5 a The chromosome pair which is affected is number 21.
b The person whose chromosomes are shown is a female because there are two X chromosomes.
6 A person could be born with Down syndrome if they receive two copies of chromosome 21 from their mother. If they receive one copy from their father, then the baby will have three, as shown in this diagram:



- 7 a The main environmental factors that influence the features of humans are diet, lifestyle, diseases and injuries.
b These factors are likely to have a greater influence in younger or 'formative' years.
c Plants will be affected by factors such as the availability of light, water, pH and nutrients. Some plants will also be affected by physical damage, such as pruning.

Hints and tips

- Chromosomes are thread-like strands of protein and deoxyribonucleic acid (DNA) that carry genes and transmit heredity information. Different chromosomes contain different genes, and a particular gene is always found on the same chromosome. This means that each chromosome contains a specific portion of the genome. (Genomes are discussed on page 191.)

For example, in humans the gene for alpha globin, part of the haemoglobin protein that carries oxygen in red blood cells, is located on chromosome 16, while the gene for beta globin, the other part of the haemoglobin protein, is found on chromosome 11. Muscular dystrophy is a disease caused by a defective gene on the X chromosome. (Further information can be found on page 204.)

- Give students a list of interesting facts, or, better still, ask students to come up with their own list about human genes and DNA. Some starting points could include:
 - There are around 10^{14} cells in the human body, with almost all containing 46 chromosomes in their nucleus.
 - Just about every part of your body contains DNA. Interestingly, red blood cells don't, but white blood cells do.
 - DNA in a cell forms a double helix (spiral). The DNA in a cell contains 3 billion base pairs (A, T, C, G), and is divided into 23 separate pieces (chromosomes). The DNA's double-stranded fibre extends unbroken the entire length of each chromosome and is between 1.7 and 8.5 cm long. If the 23 pieces of DNA were added together it would total about 1 metre.
 - If the entire DNA in a human was lined up end to end it would be

8.2 DNA

Chromosomes contain a substance called deoxyribonucleic acid or DNA for short. It is the DNA in the chromosomes that determines your characteristics.

The chemical composition of DNA was first investigated in 1869 when the German chemist Friedrich Miescher found that a substance from cell nuclei was acidic and contained the element phosphorus. Because it was found in the nucleus, this substance was initially called *nuclein* and later called deoxyribonucleic acid.

In the 1950s, Watson, Crick and Wilkins proposed that the DNA molecule is shaped like a *double helix*—something like the lookout in King's Park, Perth (see the photo below).

The DNA contains sugars (deoxyribose), phosphates and nitrogen-containing substances called *bases*. There are four types of bases: adenine (A), guanine (G), thymine (T) and cytosine (C). In the DNA molecule, base A on one strand will bond only with T, and C will bond only with G. For this reason A – T, and C – G are called *base pairs*.

The DNA molecule is double stranded with each base on one strand weakly bonded to its base pair on the other strand. This bonding makes the two strands lock together to form the double helix shape.

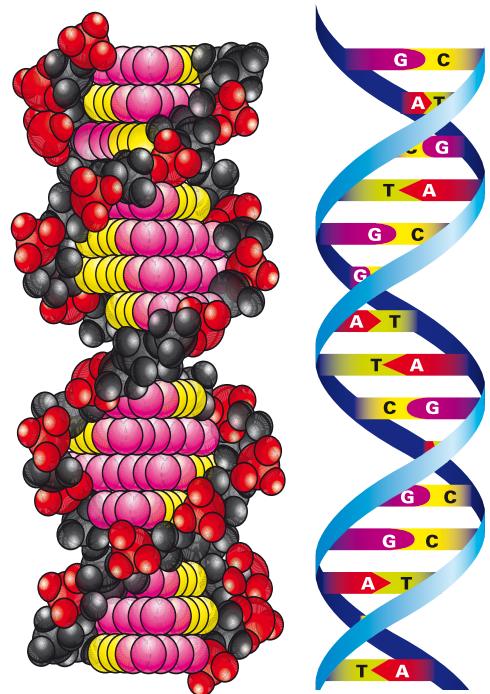


Fig 14

A model of DNA and a simplified structure on the right. The weak bonds between the matching base pairs on each strand hold the DNA molecule in its helical shape.



about 1000 times the distance from the Earth to the Sun.

- In each cell there are around 20 000 to 25 000 protein-coding genes and the rest is termed 'DNA junk' (only because scientists don't yet know its function). In fact, only about 6% of DNA codes for proteins.
- Remind students that enzymes don't actually take part in reactions, but act as catalysts.

Genes

Organisms are different because the proteins in their cells are different. It is the DNA that provides information about the types of amino acids that make up proteins. The arrangement of the four different bases along a DNA strand will determine what type of protein will eventually be made. These sections on the DNA containing the bases are called *genes*. A gene is the portion of a chromosome that tells the body what type of protein to make.

Many of the substances in cells are made with the aid of enzymes, and all enzymes are proteins. So by determining the types of enzymes that are produced, the DNA code determines the organism's characteristics.

Learning experience

Students would probably have found from earlier research that organisms of the same species have the same number of chromosomal pairs. For example, leopards and domestic cats have 19 pairs (38 chromosomes). However, so too do lungfish. Chickens and dogs also share the same number of chromosomes, both having 39 pairs. What makes each species different? Turn this question into a Round Table activity, or ask students to form small groups to brainstorm and research an answer. This activity should lead students into what DNA and genes are.



Science bits

DNA replication

During cell division the DNA in the chromosomes copies itself. This process is called **replication** because the DNA copy is an exact **replica** of the original DNA.

The DNA is made up of building blocks called **nucleotides** (see Fig 15). The nucleotides are made of three molecules—sugar, phosphate and a base molecule. The nucleotides are linked together in the DNA to form strands.

The two twisted strands of DNA in the double helix are held together by weak bonds between the base pairs on each strand. When replication begins the weak bonds break and the DNA ‘untwists’, as shown below.

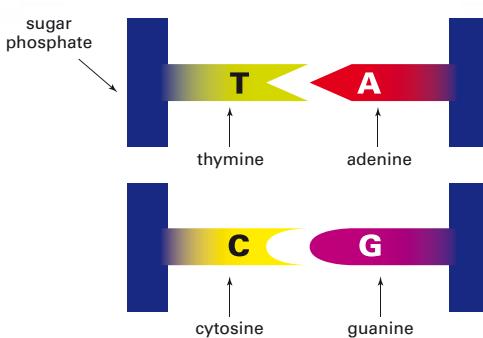
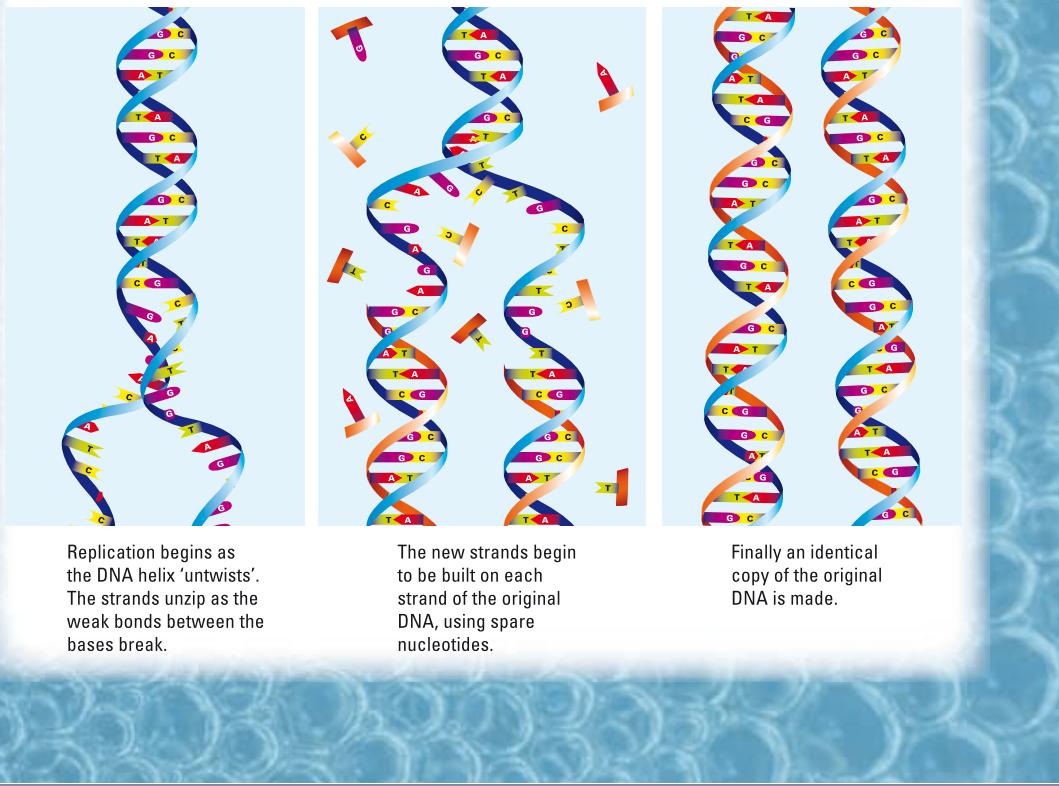


Fig 15 Nucleotides are the building blocks of DNA. They consist of sugar, phosphate and base molecules.

To see an animation of DNA replication, open **DNA replication** on the CD.



Learning experience

Have the class develop a flow diagram of concepts relating to chromosomes, DNA and genes. Students should be able to describe the relationship between these. The concepts could be a series of pictures or sentences. Consider starting the flow diagram with the following:

Chromosomes are made of DNA and proteins, which store genetic information → Differing amounts of proteins in the cells is what makes organisms different → DNA determines the types of proteins made, so DNA determines your characteristics → ...

Learning experience

Have students develop a flow diagram about DNA or DNA replication. Again, concepts could be as a series of pictures or sentences. Consider starting the diagram with the following:

DNA is deoxyribonucleic acid → DNA is made up of sugars, proteins and bases → ...

or

DNA is deoxyribonucleic acid → During cell division, the DNA in chromosomes copies itself → This process is called replication → ...

Learning experience

Students could make their own model of DNA. Encourage creativity but accuracy. Their models need to show the basic features of DNA and be able to demonstrate the process of replication. You might like to have a combined class effort to construct a model that can be hung from the ceiling, like a paper-chain decoration.

Hints and tips

DNA copies itself whenever new cells are made. A portion of DNA containing a sequence of up to 1000 bases forms a single gene. It is the difference in the ordering of the bases which makes each gene different—its **genetic code**.

Research

Ask students to research a genetic scientist and find out more about their contributions to genetics, then present their information as a profile of the geneticist. They could answer questions such as:

- How has their contribution helped with the understanding of genetics?
- When did the geneticist publish their findings?
- What other information can be researched about the geneticist? Geneticists that could be researched include Francis Crick, James Watson, Rosalind Franklin, Gregor Mendel, Barbara McClintock and Linus Pauling.

To avoid students plagiarising information from unknown sources, you may prefer to print out sets of information sheets about some geneticists for them to use. This way they will know that you will be able to check easily for copying. Also, insist on students including a bibliography (in the correct format). Another way to avoid plagiarism is to give creative options for students to present their work. Consider getting them to write or perform an interview with the geneticist for a science program, write an article for a science textbook, develop a quiz with answers about the geneticist, or choose another creative approach.

Homework

Have students add to their word list and write a simple definition of each word using suitable language, so that anyone learning genetics for the first time will understand the meaning of the word. Students whose preferred learning style is visual could add diagrams to help with their explanations. Students with an auditory preference could make a podcast to play on an MP4 player. Students with a kinaesthetic learning preference might like to construct some plasticine models with labels. To assist ESL students, get them to write each word and its meaning in their native language. Suggested words are: *DNA, gene, codons, nucleotides, amino acids, double helix, base pairs, proteins.*

Hints and tips

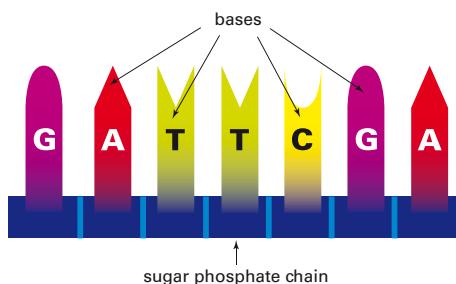
Students could make a set of summary cards for this chapter. On one side of the card they put a diagram or word, and on the back its definition/explanation. Get the students to place the cards on the table, with the definition/explanation side down. Working in pairs, students take it in turns to select a card and try to explain its meaning. If they explain it correctly, they keep the card. The winner is the person with the most correct cards.

Activity notes

- Students will find it easier if they divide their strip into sections of three paper clips and use a pen to mark the divisions on the cardboard.
- Remind students to keep their model for the next activity.
- Note that Fig 19 on page 189 shows the DNA triplets and not the RNA codons which are complementary.

The DNA code

If the two strands in the molecule of DNA separated, part of a single strand would look like the structure in the diagram below. It is the sequence of the bases along the DNA that forms the code.



Any *three* of these bases form a triplet code for one amino acid. For example, GAT will code for one type of amino acid and TCG will code for another. However, with four types of bases you can make $4 \times 4 \times 4 = 64$ different triplets. This is many more than the 20 amino acids, so some amino acids have two or more alternative codes (see the table on the next page).

Different sequences of bases code for different amino acids, which make up different proteins; and the number of amino acids in a protein molecule can vary between 50 and 50 000 or more. DNA molecules contain millions of bases. Hence, a DNA molecule can code for thousands of proteins.

Mutations

The fawn (baby deer) in the photo on the next page was born without any skin colouring or hair colouring. This characteristic is called *albinism* and the animal is called an *albino*.

This fawn has a gene in its cells that has stopped the production of any pigment. There has been an alteration in the original gene that codes for normal colouring. Alterations to genes are called **mutations**. (See Science Bits on page 190 to learn more about the chemical basis of albinism.)

Some mutations can be detrimental to the organism, and most are fatal causing death. Other

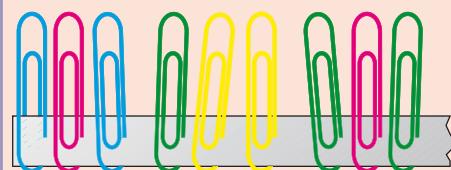


Activity

Model DNA

You will need some coloured paper clips and a small sheet of cardboard.

- 1 Choose four colours from the pile of paper clips, and assign a base to each colour. For example, blue = A, red = T, etc.
- 2 Cut a 12 mm wide strip from the sheet of cardboard. Make it between 20 and 30 cm long. The strip represents the sugar phosphate chain of the DNA molecule.
- 3 Place 18 paper clips (in any order) along the strip of cardboard. For convenience, group them into threes with a small gap in between.



The table on the next page shows the code for the 20 amino acids that are found in the body. Notice that most amino acids have two or more alternative triplet codes. Notice there are also codes for STOP and START.

- 4 Use the table on the next page to find the code for STOP. This indicates the end of the amino acid chain of the protein that is to be made. Add this triplet to the end of your chain.

Use the table to work out the amino acid sequence on your model DNA.

Swap strips with your partner and work out their amino acid sequence.

Keep your model for the next activity.

mutations can be beneficial. For example, humans have benefited from a mutation in seeded grapes which produced seedless grapes.

How do mutations cause changes to characteristics? Suppose a small part of a DNA

Learning experience

Consider getting the class to do an experiment to extract DNA from fresh wheatgerm, kiwifruit or ground split peas. Most senior biology books will list the materials and method. When doing the experiment it is important to maintain the correct temperature and to stir the mixture very gently. In the final stages of the experiment, when the DNA has precipitated into the ethanol in the test tube, use a stirring rod to draw out a strand of DNA and view it under a microscope.

| DNA code | amino acid | DNA code | amino acid | DNA code | amino acid | DNA code | amino acid |
|----------|---------------|----------|------------|----------|----------------------|----------|---------------|
| AAA | phenylalanine | GAA | leucine | TAA | isoleucine | CAA | valine |
| AAG | | GAG | | TAG | | CAG | |
| AAT | leucine | GAT | | TAT | | CAT | |
| AAC | | GAC | | TAC | methionine/ START | CAC | |
| AGA | serine | GGA | proline | TGA | threonine | CGA | alanine |
| AGG | | GGG | | TGG | | CGG | |
| AGT | | GGT | | TGT | | CGT | |
| AGC | | GGC | | TGC | | CGC | |
| ATA | tyrosine | GTA | histidine | TTA | asparagine | CTA | aspartic acid |
| ATG | | GTG | | TTG | | CTG | |
| ATT | STOP | GTT | glutamine | TTT | lysine | CTT | glutamic acid |
| ATC | | GTC | | TTC | | CTC | |
| ACT | | GCA | arginine | TCA | serine | CCA | glycine |
| ACA | cysteine | GCG | | TCG | | CCG | |
| ACG | | GCT | | TCT | arginine | CCT | |
| ACC | tryptophan | GCC | | TCC | | CCC | |

Fig 19 The triplet codes on DNA and the amino acids they code for—notice that there are alternative triplet codes for almost all of the 20 amino acids, as well as codes for STOP and START.

strand had the following sequence of bases:

AATCAACCTTCA

For convenience, let's separate the triplets.

AAT CAA CCT TCA

Using the table above this would code for the following amino acids:

leucine valine glycine serine

If there was a change to one of the bases:

AAT CCA CCT TCA

the new sequence of amino acids would be:

leucine glycine glycine serine

This change could produce quite a different protein from the one that was made originally.

Mutations occur naturally, and it has been estimated that 1 in 1 000 000 cells contains a mutation in its DNA. However, the rate of mutations can be increased by exposure to



Fig 20 An albino fawn (baby deer)

Hints and tips

- Make sure students realise that the order of the DNA triplet code is important. For example, AAG (phenylalanine) is not the same as GAA (leucine).
- Get students to share with each other what they have learned so far in the chapter using the 'Inside/Outside Circles' method. (See Learning experience, page 94, for instructions.) As this method can be quite noisy, spread students as much as possible around the room.

Learning experience

Get students to investigate the difference between a mutation and a mutagen. What are some examples of mutations and some examples of mutagens?

Hints and tips

Albino animals occur in almost every animal species. Because albino animals have no natural protection from UV radiation, they are prone to skin and eye cancer.

Activity notes

As an extension to this activity, consider designing some questions that allow the students to reflect on the material covered and their involvement in group activities. Here are some possible questions:

- What did you learn from the work you did today?
- Why is being able to solve a problem an important science skill?
- Why do you think understanding DNA is important?
- Has our understanding of DNA improved the quality of our lives?
- What is an example of a problem solved or being solved in this chapter?
- How did you feel when you had to share your ideas in a group?
- What did you learn about yourself during the activity?
- What role did you play in your group, and how did you assist the group?

high energy radiation from X-rays and nuclear reactors, as well as exposure to chemicals such as formalin and certain pesticides.

The UV part of sunlight can also increase mutations in organisms. These mutations can lead to cancer, particularly in skin cells. This is why over-exposure to the Sun is dangerous, especially for people with light coloured skin.



Activity

DNA and mutations

You will need the model strand of DNA that you made in the activity on page 188.

Simulate a mutation by selecting one paper clip at random and replacing it with another colour.

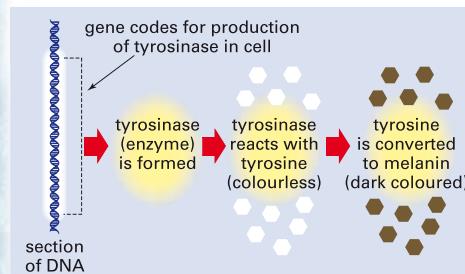
- Find the new sequence of amino acids.
- Sometimes a mutation will not change an amino acid in a protein. Work out how you can change a base in your sequence but not change the original amino acid sequence.

science bits

Why there are albinos

The colour of your skin, eyes and hair is due mainly to a chemical compound called *melanin*. Melanin is made in special cells called *melanocytes* found at the bottom of the epidermis in your skin. The colour of your skin and hair depend upon the size of the melanin granules in the melanocytes. In black skins, the granules are larger and in white skins they are less obvious.

Melanin is made from tyrosine, a colourless amino acid. Tyrosine is converted to melanin by an enzyme called *tyrosinase*. If a mutation occurs in the gene that codes for the production



of tyrosinase, the enzyme is not produced and melanin is not made. This results in an albino with whitish-pink skin, white hair and red eyes.



Fig 23 An albino Papuan boy

WEBwatch

Use your internet browser to find more information about albinism. Search using the following words: *melanin*, *melanocytes*, *albino*, *skin colour*.

Find out why albinos cannot tan in the sun and why an albino's eyes are red. Also try to find out why albino wild budgerigars are yellow.

Learning experience

Students could find out if their local zoo or wildlife centre has an albino animal and, if so, investigate what 'care plan' is needed to look after the animal. How is the animal protected from sun exposure? What sort of life expectancy would the animal have if it didn't live in captivity? What are the chances of survival of albino animals? Ask students to explain their answers.

Learning experience

We are physically and emotionally what we are because of a combination of genetics and our environment. Investigate how our environment can change our genetic make-up. Survey the class to find out who has had an X-ray or some other nuclear medical test. Discuss how exposure to radiation can cause changes to a person's DNA. Link this section to Chapter 6 and the effects of radiation exposure, particularly the Chernobyl disaster. Students could draw a cartoon strip showing the possible effects of radiation exposure.

Learning experience

Do the Webwatch if you have access to computers in class, or give it as a homework exercise.

Genomes

In 1916, after years of experimenting, Walter Sutton inferred that the inherited characteristics of fruit flies were carried on the chromosomes in the cell. In March 2004, scientists who had been experimenting collaboratively for five years found there were 250 million bases in the DNA in each fruit fly cell. They also determined the order in which the bases are arranged on the DNA.

The whole of the fruit fly's genetic information is found in these 250 million bases, and is called its **genome**.

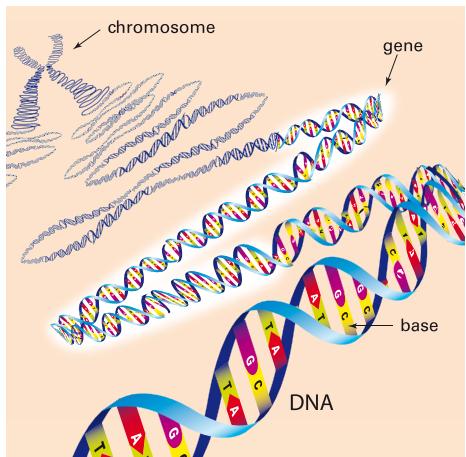


Fig 24 A gene is a section of DNA containing a particular sequence of bases

What makes up a genome?

The researchers working on the fruit fly genome have also identified 13 601 genes on the DNA. These genes are made up of thousands of bases. The average gene contains about 2000 bases. If you multiply the number of genes by the number of bases an average gene contains, you find that the genes make up less than 20% of the genome.

The larger part of the genome contains sections that control how genes are turned on and off. There are also long sections of bases that are called 'junk' or nonsense DNA because scientists are uncertain of their function.

The Human Genome Project

The Human Genome Project (HGP) which ran from 1990 to 2003 mapped the position of the genes in the human genome. This massive task was co-ordinated by the US National Institutes of Health and involved hundreds of scientists from at least 18 countries, including Australia. By the time the HGP had finished it had:

- identified 20,000 protein-coding genes in human DNA, and another 2000 DNA segments which are predicted to be genes
- determined the sequence of the 3 billion bases on human DNA
- stored the information on databases
- addressed ethical, legal and social issues that arose from the project.

The expected benefits of the Human Genome Project are:

- improved diagnosis of disease
- treatment of genetic diseases such as cystic fibrosis
- manufacture of custom drugs
- replacement of defective genes
- reduced risk of inheritable mutations
- improved techniques for DNA identification in legal and criminal cases.

HGP—the future?

Although the HGP was coordinated by government departments, one of the project's aims was to transfer all the technologies to private companies. Now almost all of the HGP follow-up research is being done by private commercial companies, particularly multi-national US-based companies, who expect to earn billions of dollars through sales of new drugs, equipment, technologies and information.

However, will the knowledge of the human genome benefit all humankind? Here are some problems that might arise:

- Will treatment and diagnosis be so expensive that only the rich will benefit?
- Will an individual's genetic file be private and secure, or will it be available to banks, health funds, insurance companies etc?
- Will the large companies patent their drugs so that they control certain treatments?

Hints and tips

There are many big ideas related to gene technology that can be explored. Consider giving gifted and talented students a task that involves them presenting/teaching information to the class on a topic that is currently in the media. Students need to collect media articles about their chosen topic, review them and present a balanced scientific viewpoint.

Research

Gene technology and its uses are debatable topics. Most negative issues surrounding this technology are to do with ethics. Have students research the use of gene technology in a particular area. They should spend time brainstorming the issue and could use tools such as an issues map, PMI chart or an advantages/disadvantages chart. Get students to construct a flow diagram of how the technology works or is used. It is important that students explore not only the science involved in their topic but also any ethical considerations. Possible topics are:

- therapeutic cloning/stem cells
- stem cell research
- genetic testing for diseases/disorders.

Their aim should be to inform the general public about how the technology works or is used, pros and cons, and ethical considerations. Students should use appropriate language outlined in this chapter, and present their work as an information poster, booklet, pamphlet, multimedia presentation, podcast or vodcast. Encourage them to choose a format that they have not used previously. (There is more on gene technology in Chapter 9.)

Learning experience

Ethics is the study of what is right and wrong. Get students to infer what bioethics is. Although we all differ in our viewpoints, we still have a conscience and moral obligation to do what is ethically right. However, what is right? This is the real dilemma faced not only by scientists but by us as well. Having the power to change or manipulate things doesn't always mean it is right. Have students find out if there is an organisation or group of people that regulates ethical issues in science.

Activity notes

Because this activity requires a lot of reading, make sure you are monitoring students who experience language difficulties. Where appropriate, suggest their note taking is in point form, with material clearly referenced.

Hints and tips

- Consider discussing electrophoresis. It is a process similar to chromatography but instead separates DNA. The DNA molecule is cut into pieces, samples are placed in a gel (agarose), and an electric current is applied. The current causes pieces of DNA to move quickly or slowly depending on their size.
- After reading this section in the book, revise the work students did at the beginning of the chapter where they investigated a mock crime scene. Have students re-examine their questions and conclusions. Would they change any of their ideas now? Ask them to explain.

Homework

Red blood cells don't contain DNA. How and where are they made? Find out if there are any other human body cells that do not contain DNA. Investigate how a blood sample can still be tested for DNA. (White blood cells contain DNA, and a blood sample has both red and white cells.)

Activity



Work in a group of 3 or 4 people and discuss some of the issues in the statements and questions below. Use the internet, or find material from library books, magazines and newspapers.

- Getting a man on the moon in 1969 was a massive project involving many scientists. How was this different from the scientific collaboration in the Human Genome Project?
- Collect as much information as you can on new discoveries, techniques or inventions resulting from the Human Genome Project. Compile a scrapbook or information file and share this with the other groups in the class.
- Suppose you have no family history of genetic diseases, and are thinking about having children. Should you ask your partner to have a DNA test done to make sure that their genome is disease-free?
- Some life insurance companies are suggesting that a person's genetic file

be submitted when they apply for life insurance, so that the company can assess whether that person is a high-risk case. What do you think? Give reasons for your answer.

WEBwatch

Go to www.scienceworld.com.au and follow the links to the websites below.

Student Guide to the Human Genome Project

Extensive and easy-to-read information on HGP and links to a large number of other sites.

Frequently asked questions—Human Genome Project

Answers to questions about the HGP.

Educational Resources (HGP)

The education section of the National Human Genome Project Institute has links to facts sheets and information about the HGP.

DNA detective work

With the completion of the Human Genome Project and advances in computer technology, large segments of DNA or even the whole genome will allow precise identification of an individual. This is important in the following situations:

- matching donor organs with recipients in transplant operations
- identifying suspects whose DNA matches evidence left at a crime scene
- exonerating people who are wrongly accused
- identifying victims in major accidents, catastrophes or natural disasters
- establishing paternity of a child.

How is DNA used to identify people?

Detectives and forensic scientists have to gather a number of pieces of evidence together in order to convict a person of a crime. For example,

a suspect's blood type and fingerprints are compared with those found at the crime scene, as are pieces of hair or fabric from their clothes and the tread of their shoes. All these pieces of evidence have to prove 'beyond reasonable doubt' that the suspect is the guilty person.

When using DNA to convict people, forensic scientists have to match the sequence of bases in a number of regions of the DNA of the suspect with those of the DNA samples found at the crime scene. Since only 0.1% of your DNA is different from anybody else's, scientists have to match base sequences of the DNA in those regions which are different.

A court will not convict a person when only one or two of the DNA base sequences match. This is too little evidence. However, when at least five DNA base sequences match, a jury can be confident 'beyond reasonable doubt' that the suspect is the guilty person.

Learning experience

Solving crimes by using DNA might not be the perfect solution. It is suggested that a criminal could take someone else's DNA and use a polymerase chain reaction (PCR) to replicate it, and then place it around the crime scene. Have students investigate how PCR can be used to replicate DNA.

Learning experience

Do the Webwatch if you have access to computers in class, or give it as a homework exercise.



Fig 25 A forensic scientist examines the sequence of bases in sections of DNA.

Solving the French royal mystery

In 1795 Louis-Charles, the young son of King Louis XVI and Marie-Antoinette, supposedly died in a Paris prison. But many people believed he had escaped the brutality and executions of the French revolution and fled to England.

In April 2000, scientists used DNA matching technology to try to solve one of history's greatest mysteries. They used some tissue from the young prince's suspected remains and compared the DNA in it with the DNA in hair from his mother, as well as other samples from living and dead members of the royal family.

The DNA matching tests showed that the tissues were from the young prince, but many people are still not convinced. Go to www.scienceworld.net.au and follow the links to **Louis-Charles** for more information about this mysterious case.



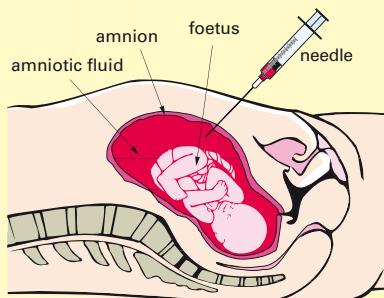
Science in action

Testing the foetus

If parents-to-be know that one or both of them has a family history of genetic disorders, doctors may suggest that the pregnant woman have one or more tests. Doctors also know that children born to women more than 37 years old have a greater chance than normal of having genetic disorders such as Down syndrome.

Doctors will first suggest an *ultrasound* test when the woman is about 11–13 weeks pregnant. In this test reflected sound waves generate a picture of the foetus. If doctors suspect an abnormality, they may recommend other tests.

In a test called *genetic amniocentesis* (AM-nee-oh-sen-TEE-sus), a fine hollow needle is passed through the abdomen of the pregnant woman and into the amnion. This is a fluid-filled sac surrounding and protecting the foetus. A tiny amount of amniotic fluid is withdrawn and tested.



High levels of protein in the fluid may indicate diseases such as spina bifida. DNA in the foetal cells that are found in the fluid are tested to check for the presence of Down syndrome and other genetic abnormalities. However, because amniocentesis is an invasive test there are certain risks. About one in every 200 tests results in a miscarriage.

If the woman is found to be carrying an abnormal foetus she may be given choices of action. She may continue with the pregnancy or she may terminate the pregnancy before the legal limit (28 weeks in most states of Australia).

Hints and tips

As students enter the room, hand them a card containing a word or a phrase related to genetics. Words from the glossary or vocabulary list that they started at the beginning of the chapter could be used. When students are seated, randomly ask students to read out the word and, in one minute, describe what it is, its function, how it relates to genetics and any other useful information they know about it.

Learning experience

Have students find out about the process of making a DNA fingerprint on X-ray film. They could draw a flow diagram of the process with accompanying pictures. Fig 25 is an example of a DNA fingerprint on X-ray film.

Learning experience

With advances in gene technology it will become easier to manipulate human embryos. Already it is possible to screen embryos for some genetic disorders and select healthy ones. It is also possible to choose the sex of the embryo using advanced reproductive techniques during IVF. In pairs, get students to discuss and write a series of questions they would like to ask a scientist about this information. Then have them make a prediction about how they think things might have changed 30 years from now.

Check! solutions

- 1 DNA is the abbreviation for deoxyribonucleic acid, which is found in the nucleus of all cells in the body.
- 2 A base pair consists of one base on one strand of DNA and its complementary base on the other strand, eg A-T or C-G.
- 3 a The genetic code is a sequence of bases on DNA that is found in chromosomes.
b The genetic code eventually leads to the production of proteins.
c The cells and tissues in your body contain proteins. Proteins are needed for the growth and repair of cells. As enzymes, they also help to regulate the activity of cells and body movement.
- 4 The matching (complementary) bases are CCTATCGAACATCGC.
- 5 Each amino acid needs a triplet of bases, so the total will be $51 \times 3 = 153$ bases. There will also usually need to be a STOP codon so the total will be 156.
- 6 The sequence of amino acids will be: cysteine – proline – isoleucine – valine – glycine – serine – proline.
- 7 If a mutation occurs to the first base of the first triplet and it changes from 'A' to 'T', the base will be changed from cysteine to serine. If amino acids are changed, the protein will have a different structure and may not be able to perform its particular function. However, this will not happen in every case. For example, if the third base in the second triplet is changed from 'T' to 'A', there will be no change to the base; it will still be proline.
- 8 Some environmental factors that are known to increase the rate of mutations are chemicals (eg formalin and DDT) and radiation (eg ultraviolet light and X-rays).
- 9 The Human Genome Project has the potential to be of benefit in the following ways:
 - diagnosis and treatment of genetic diseases
 - replacement of faulty genes in plants and animals and, possibly humans
 - manufacture of special drugs
 - improved methods for using DNA to solve crimes and catch criminals.
- 10 Here are some issues. No doubt you will have more.
 - Who should have access to personal genetic information, and how should



- 1 What is DNA, and where is it found in the body?
- 2 What is a base pair? How do the bases help form the double helix shape?
- 3 a What is the genetic code?
b What materials does the genetic code eventually produce?
c Why are these materials so important in organisms?
- 4 A small part of a DNA strand contains the following bases: GGATAGCTTAGCG
What are the matching bases on the other strand of DNA?
- 5 Insulin is a relatively small protein, having a total of 51 amino acids in its structure. What is the smallest number of bases on a DNA strand needed to code for insulin?

- 6 A small segment of DNA has the following sequence of bases:

ACA GGT TAA CAA CCT TCA GGG

Use the table on page 189 to work out the amino acid sequence.

- 7 Explain, by using the base sequence in the question above and the table on page 189, how mutations alter proteins.
- 8 Which environmental factors can increase the rate of mutations in organisms?
- 9 Make a list of the benefits that might be gained and the problems that might arise from the Human Genome Project.
- 10 The Human Genome Organisation has allocated 3–5% of its budget for the study of the project's ethical, legal and social issues. Make a list of some of these issues. You may want to discuss this with others.



- 1 Biologists estimate that 99.9% of all your genes are similar to the genes of other people. The 0.1% makes you different. However, about 80% of your genes are the same as those in a cat or dog, and about 60% are the same as in an earthworm. What can you infer from this information?
- 2 The following amino acids are from a small section of a protein.
leucine–glycine–tyrosine–lysine–lysine–glycine
a Work out one base sequence on the DNA strand that would code for this section of protein.
b Compare your base sequence with others in the class. How many different sequences did the class make for this section of protein?
c What is the base sequence on the other strand of the double-stranded DNA?
- 3 Suppose a mutation occurs in a sperm cell of an animal and a gene is altered. This sex cell fertilises an ovum and an offspring is produced. However, many mutations are fatal and the offspring dies.
Use your knowledge of the materials genes make to suggest why this occurs.

- 4 A forensic scientist matches the DNA from the suspect with a sample from the crime scene. Explain to someone who doesn't know what DNA or forensic science is how this works. Use the terms *base sequence*, *DNA regions* and *beyond reasonable doubt* in your answer.
- 5 Man A believes he is the father of a child. The mother believes that man B is the father. How would you use DNA technology to go about solving this problem?
- 6 Use the internet to find out about other ways to test the health of a foetus. Try looking for Chorionic Villus Sampling (CVS) and the Maternal Serum Test.



Watson and Crick were awarded the Nobel Prize in Physiology and Medicine in 1962 for their work on DNA structure.

Use the internet to find out more about their work. Also find out about the work of Rosalind Franklin and why she might have shared the Nobel Prize with Watson and Crick.

Then write a feature story about the discovery of the structure of DNA.

Challenge solutions

- 1 From this information you can infer that organisms that have more DNA in common are likely to look more similar. You can also infer that approximately half of the DNA present in human cells performs very basic functions, such as those in an earthworm.
- 2 a One possible base sequence is: AAT CCA ATA TTT TTT CCA.
b Because most amino acids have more than one base triplet, there are actually 256 possibilities. So it is quite possible that you all had different sequences.
c The complementary strand would be TTAGGTTATAAAAAAGGT.

8.3 Dominant and recessive

Suppose you have set up an aquarium with two black fish—one male and the other a female. One day, a few months later, you notice 8 baby fish. Your two parent fish have bred and had offspring. However, the puzzling thing about the baby fish is there are six black fish and two red ones!



The colour of the fish is controlled by a particular gene. This gene comes in two forms—one that codes for black colour and the other that codes for red colour. Different versions of the same gene are called **alleles** (a-LEELs).

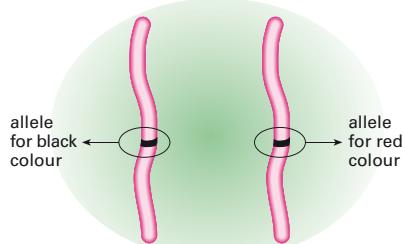


Fig 28 The colour of the fish's skin is controlled by two alleles of the same gene. Each allele is found at the same location on each of the chromosomes in the pair.

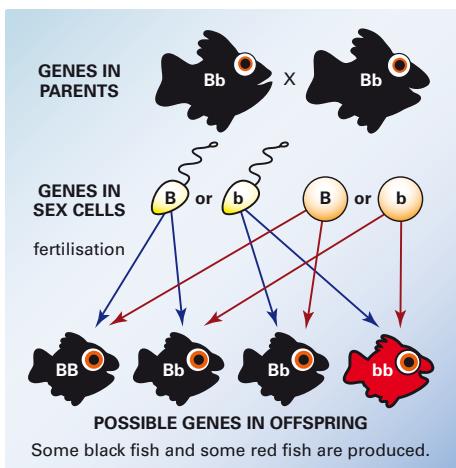
However, if the parents have the genes for black colour as well as red colour, why don't you see any red colour in the parents? This is because the gene for black colour completely masks the gene for red colour. The gene for black colour is said to be the **dominant gene**. The gene for red colour, which is masked by the dominant gene, is called the **recessive gene**.

It is usual to represent the genes by the first letter of the dominant gene. A capital letter is used for the dominant gene, and the lower case of that letter for the recessive gene. In this case, the allele for black colour is **B**, and the allele for red colour is **b** (not **r**).

Fish carrying the genes **BB** will be black, and those carrying the genes **bb** will be red. Those fish carrying **Bb** will also be black because **B** is the dominant gene. (The dominant gene is usually written first, ie **Bb** and not **bB**.)

If the parents have a gene for black colour and one for red colour, they can produce some baby fish with black colour and some with red colour.

BB = black colour
Bb = black colour
bb = red colour



Hints and tips

- In *ScienceWorld 2 Chapter 6*, students would have investigated inherited features and pedigrees. See how much they can remember by giving them a quick quiz. This way you will be able to gauge if work needs to be revised.
- Reinforce that the dominant gene is written with a capital letter while the recessive gene has the same letter but in lower case.

Learning experience

Have students draw a Venn diagram with two overlapping circles. Label one circle 'Mum', the other circle 'Dad' and the common region 'Me'. In the circles, get students to list characteristics that each parent has. In the overlapping part, they should list what they seem to have inherited from their parents. Explain that not all characteristics are inherited because of single dominant genes. The usual set of characteristics can be used for the Venn diagram: tongue rolling, attached ear lobes, widow's peak, right-handedness, left thumb crossing over right, fifth finger bent inwards, and skin pigment are all characteristics controlled by a single dominant gene. The complementary characteristics are recessive.

You could do a class survey to see how many students have inherited each dominant characteristic listed. Are the results what the class would expect? Explain. Be sensitive towards those students from a split or single-parent family.

Learning experience

If the school has an aquarium, students may find it fun to breed fish, although this can be slow and sometimes tricky. Alternatively, if you have an animal room, they could breed mice. Make sure the students take responsibility for the care of their mice, and know the ethical procedures for handling them. Ask the lab technician to choose two coloured mice (not white) so the class can, hopefully, see variation in the offspring. (Usually a local wildlife centre that has snakes or birds of prey will accept any unwanted mice.) Check with your lab technician to ensure the school has an Animal Handling Certificate.

- Mutations alter the bases in the triplets on the DNA. This leads to changes in the amino acids and therefore the proteins produced by the animal. This will affect the way the animal functions, but it may still survive. However, during reproduction the genes determine the structure and function of the offspring. So if the genes are defective the offspring will not function properly and will die.

- Your explanation would go something like this. All living things carry information about how to grow and develop as *base sequences* on DNA in chromosomes in their cells. Scientists are able to compare certain *DNA regions* of samples found at the scene of a crime and compare them to

suspects to see whether there is a match. If so, this is strong evidence that the person committed the crime and puts the person's guilt *beyond reasonable doubt*.

- Cells are taken from both possible fathers, the mother and the child, and certain sections of DNA are compared. Obviously, all of the child's DNA comes from its genetic parents so scientists can decide who the real father is.
- You will find that both of these techniques are now available and, in fact, encouraged in cases in which there is increased risk. This includes situations in which the mother is over the age of 35 or there is a family history of a genetic disease.

Hints and tips

Consider showing the class the results of Mendel's experiments with pea plants. Explain why the first and second generation of pea pod colours are different. Using the results, further explain the differences between genotype, phenotype, homozygous and heterozygous. This exercise will lead nicely into the next section of work. See if students can work out the possible combinations for a third generation of peas. Alternatively, you could be more creative and make up your own example, or get students to do this.

Activity notes

- If you use coins they must all be the same type, eg all 10 cent coins, otherwise students will be able to tell them apart by feeling them. To differentiate between the coins, mark them with a felt-tipped marker or coloured stickers.
- The disks could be placed in brown paper bags.

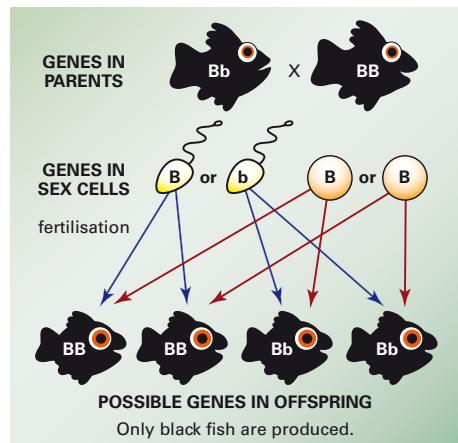
Learning experience

Have students write a self-reflection of what they have learned so far from this chapter. They could begin with what they have learned, how they have learned this, and how they have helped others. Students should write at least 10 sentences. Next lesson, call on students to select one thing they have learned and share it with the class. They will soon realise just how much they have learned.

Learning experience

Do the Webwatch if you have access to computers in class, or give it as a homework exercise.

If only one of the parents has the gene for red colour then none of the offspring will be red, as shown in the diagram below.



WEB watch

Gregor Mendel was a monk and science teacher in a monastery in Austria. In 1866 he published his results of breeding experiments with peas. His meticulous and accurate records showed how characteristics could be passed from generation to generation. Mendel is often called the father of genetics.

Use the internet to answer the following questions. Remember to record the websites where you found the information.

- What characteristics in peas did Mendel first study? What were his conclusions?
- Why is Gregor Mendel called the father of genetics?
- How did Mendel try to explain how characteristics were passed on from generation to generation?
- It is said that Mendel's work was poorly understood and largely forgotten until after his death. Suggest a reason for this.

Activity



Fish genes model

In the example on page 195, you found that if both parent fish have genes Bb , they will have some black offspring and some red offspring. Let's investigate how many of each colour are produced.

You will need 20 black disks and 20 red disks for this activity. (You can use other colours, or coins, if you wish.)

- Place 10 black disks and 10 red disks in a small container. These disks represent the ova carrying the colour genes. Label this container Female.
- Place the remaining disks in another container labelled Male. These disks represent the sperm carrying the colour genes.
- Without looking, select a disk from the first container and one from the second.

Record the genes and the colour of the offspring. (Remember the gene for black colour is the dominant gene.)

- Replace the disks and make another selection. Do this 10 times.

Calculate the ratio of black fish to red fish.

- Make another 10 draws.

Add these results to the first results and again find the ratio of black fish to red fish.

Which results give a more accurate indication of the ratio of black fish to red fish? Why?

Collect the class results and find the ratio. Give your answer as the nearest whole number.

- Repeat this activity to confirm that parent fish with genes Bb and BB will produce only black fish.

Learning experience

Have students make their own bilby baby. Students will need two coins to represent the ova and sperm. Assume the bilby parents are heterozygous for each of the possible characteristics of the baby. Tossing the coin will determine which characteristics the bilby will inherit. Students can then make a model of what their bilby baby will look like. The bilby can be made from women's stockings (three different shades of brown); soft-toy stuffing; black, green and pink sticky

dots (for eyes); cotton thread; heavy-duty string; and pipe cleaners.

To determine the sex, only one coin needs to be tossed (heads for female). Assign one coin for the female and the other the male. For each characteristic listed, students toss each coin one at a time and record 'H' for heads and 'h' for tails. This is the bilby's genotype. Then they work out the phenotype and put it into Table 1, opposite. To make the model of the bilby, students use the information in Table 2.

Some genetics terms

The two fish below look the same but have different genes. Biologists use special terms to describe this situation.



black fish
genes = BB



black fish
genes = Bb

The type of genes in an organism is called its **genotype** (JEE-no-type). What the organism looks like or its physical characteristics is called its **phenotype** (FEE-no-type).

In this example, both fish have the same phenotype but different genotypes.

The fish with genotype BB is said to be **homozygous** (HO-mo-ZYE-gus), or a pure breeder, because both the alleles for the skin colour gene are the same. The other fish is said to be **heterozygous** (HET-er-o-ZYE-gus), or **hybrid**, because its two alleles for skin colour gene are different.

So the first fish in the diagram could be described as being a homozygous black fish, while the other one is heterozygous black.

Hints and tips

Often students confuse genotype and phenotype, and also homozygous and heterozygous. Reinforce the differences and continually revise the meanings of each. Using examples works best.

Predicting crosses

In the previous activity you should have found that the ratio of black fish to red fish was about 3:1. It is possible to predict the type of offspring produced when two organisms mate. This mating is called a *cross*. One of the easiest ways of predicting crosses is to use a *Punnett square*.

In the Punnett square below two heterozygous black fish have been crossed.

Parents = Bb x Bb

| | | Parent 2 (Bb) | |
|---------------|-----|---------------|----|
| | | Sperm | b |
| Parent 1 (Bb) | Ova | B | b |
| | B | BB | Bb |
| b | Bb | Bb | bb |

Expected ratio of phenotypes = 3 black : 1 red

Parents = BB x Bb

| | | Parent 2 (Bb) | |
|---------------|-----|---------------|----|
| | | Sperm | b |
| Parent 1 (BB) | Ova | B | b |
| | B | BB | Bb |
| B | Bb | Bb | Bb |

Expected ratio of phenotypes = all black

Learning experience

Take the class outside and make a giant interactive Punnett square. Mark out a large square with a 'girls' group on one side (Parent 1) and a 'boys' group on the adjacent side (Parent 2). Use the examples on this page as a guide. Make some cards labelled B and b so that there are enough for all combinations. Try BB, Bb and bb as possible 'Parents'. Give them four cards according to the type of parent they are. So for parent Bb they will have two Bs and two bs. Now model the combinations. Make sure each student has a turn in the Punnett square.

Table 1

| Characteristic | Female | Male | Genotype | Phenotype |
|----------------|--------|------|----------|-----------|
| Body shape | | | | |
| Ear length | | | | |
| Tail | | | | |
| Fur colour | | | | |
| Eye colour | | | | |
| Nose shape | | | | |
| Whiskers | | | | |
| Sex | | | | |

Table 2

| Characteristic | HH | Hh | hh |
|----------------|------------------|------------------|-------------------|
| Body shape | Oval | Oval | Round |
| Ear length | Long | Long | Short |
| Tail | Long | Long | Very long |
| Fur colour | Mid-brown | Dark brown | Light brown |
| Eye colour | Black | Green | Pink |
| Nose shape | Long and pointed | Long and pointed | Short and rounded |
| Whiskers | Straight | Straight | Curly |
| Sex | | | |

Hints and tips

At the start of a lesson it is important to re-examine material covered from previous lessons. Giving the class a quiz based on the material they have already learned helps you get an indication of whether any concepts need revising, and helps the students consolidate their ideas. Ask them to write the answers only (no need for the questions).

Homework

Get students to make their own pedigree for a particular genetic characteristic and show the sequence of genotypes and phenotypes. Have them use the list of characteristics listed in the Learning experience on page 195. They should use at least one set of grandparents. Students will find it easier to work backwards, starting with their generation.

Pedigrees

Earlobe attachment is an inherited characteristic. The gene for attached earlobes is dominant over the gene for unattached earlobes.

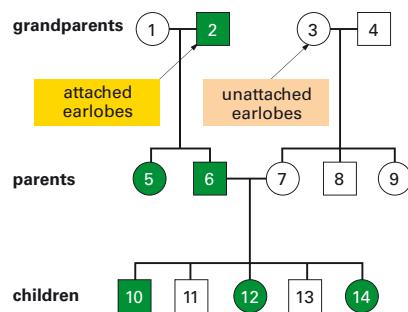


Fig 34 Attached earlobes (left) and unattached earlobes (right)

By studying family histories, biologists can build up a pattern of inherited characteristics. This pattern can be seen on diagrams called family trees or **pedigrees**. Pedigrees can show the phenotypes of related individuals over a number of generations.



Look at the pedigree at the top of the page. It traces the history of earlobe attachment through three generations. The circles represent females and the squares represent males. The shaded circles represent females with attached earlobes, and the shaded squares represent males with attached earlobes.



Pedigrees can be used to work out the genotypes of the individuals. For example, biologists know that the gene for attached earlobes is dominant over the gene for unattached earlobes. Let's call the gene for attached earlobes **A**, and the gene for unattached earlobes **a**. Using these symbols you can deduce that:

- grandparents 1, 3 and 4 will be **aa**
- individuals 7, 8, 9, 11 and 13 will be **aa**
- grandparent 2 could be **AA** or **Aa**
- individuals 5, 6, 10, 12 and 14 will be **Aa**.

The pedigree can be rewritten to show the phenotypes and genotypes of the individuals.

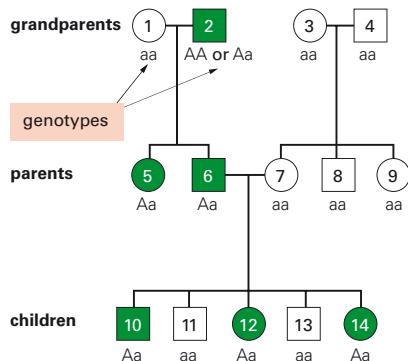


Fig 37 Pedigrees can show the genotypes as well as the phenotypes of individuals

Learning experience

Students could look up different breeding pedigrees for animals such as cats, dogs and horses. Which characteristics in some breeds are desired and which do breeders attempt to get rid of? Explain. Students could make posters showing the pedigrees.

Learning experience

Have students investigate disorders such as haemophilia, and construct a pedigree showing its inheritance. Why does it mainly affect boys? Again, a good format to present work is as a poster for a doctor's surgery. Other disorders such as Huntington's disease or Duchenne muscular dystrophy could be investigated.

Not so simple inheritance

Gregor Mendel delivered his scientific paper detailing the results of his breeding experiments with garden peas in 1866. His experiments were well designed and well recorded, and he tested more than 28 000 plants to reduce experimental errors. His success was largely due to the great care he took with his experimental methods. His knowledge of mathematics also helped him interpret his results.

However, Mendel was also very lucky. The seven characteristics he studied in peas were each determined by a single gene. For example, the gene that determines seed pod colour has two alleles. The dominant one codes for green pods, while the recessive one codes for yellow pods.

Single gene inheritance

There are very few characteristics in organisms that are controlled by a single gene. The following are two human characteristics determined by a single gene:

- Earlobe attachment is determined by the presence of the dominant allele.
- *Cystic fibrosis* is a severe disease of the lungs and intestines caused by a recessive gene. A person inheriting the two affected alleles would develop cystic fibrosis. A person with only one affected allele would not develop cystic fibrosis, but would be a 'carrier' for the disease.

Human eye colour

For many years eye colour in humans was thought to have been controlled by a single gene. The allele for brown eyes was dominant over the allele for blue eyes. However, this idea did not explain green eyes or grey eyes or the variation of colours in between.



Biologists currently think that eye colour is controlled by at least three genes and probably others that may control how the coloured pigment is distributed over the iris causing flecks and rings. Eye colour is thought to be controlled by:

- a gene found on chromosome pair 15 which controls brown-blue colours,
- a gene found on chromosome pair 19 which controls green-blue colours, and
- another gene located on chromosome pair 15 that may affect the other two genes.

Eye colour

The colour of eyes is primarily determined by the amount of melanin in them. This dark brown pigment is deposited in the cells on the front surface of the iris. If a lot of melanin is present, your eyes will be brown. If very little or no melanin is present your eyes will be blue.

There is also a brownish-yellow pigment found in people with green eyes. It may also determine whether you have dark brown or light brown eyes.

There is no blue pigment in humans. Blue eyes is the absence of melanin. The cells in the front of the iris scatter the blue light in sunlight more than the red light. So your eyes appear blue just like the sky appears blue.

Hints and tips

Asking students to read this article aloud not only keeps them focused but assists those who have a preference for auditory learning.

Learning experience

Have students create a presentation about inheritance and eye colour. Allow them free choice of format for their presentation. Some suggested formats are a booklet, multimedia presentation, podcast, conceptual cartoon or cartoon strip.

Hints and tips

At various points throughout the lesson you could use reflective questioning. Questions should be focused on students' understanding and thinking, interpersonal development and personal learning. Questions could be 'What did you learn?', 'How do you know that you have learned it?' and 'How will you use that learning again?'

Research

Research more about human blood to find out what the Rhesus factor is. Construct a Punnett square that shows the combinations if both parents are heterozygous Rh positive. The Rhesus factor is controlled by two alleles. Having the Rhesus antigen (R) is dominant over not having it (r). A person who is heterozygous Rh positive is Rr.

Blended genes

Budgerigars are native birds that live naturally in woodlands and grasslands throughout inland Australia. There are two purebred forms—yellow birds and blue birds.



Fig 40 The green budgerigar (right) is the offspring of one parent with blue feathers and the other with yellow feathers.

When a homozygous blue bird mates with a homozygous yellow bird, the offspring are green, not blue or yellow. In this case, the two alleles for feather colour are not dominant or recessive over each other, but instead result in a mixture or blend of characteristics. This is said to be **incomplete dominance**.

There are many examples of incomplete dominance. A red shorthorn bull mates with a white shorthorn cow to produce red and white calves, and red carnations cross with white ones to produce plants with pink flowers.

Suppose a blue male budgerigar mates with a yellow female. All the offspring have green feathers. Let's call the allele for blue feathers **B**. The allele for yellow colour cannot be **b** because it is not recessive. In this case we will call it **Y**, because offspring with alleles **BY** will be green.

The Punnett square at the top of the page shows the results of the cross between a blue budgerigar and a yellow one. When the green offspring are crossed, birds with blue, yellow and green feathers are produced.

Parents = **BB** x **YY**

| | | | |
|---------------------------|------------|------------------------|-----------|
| | | Parent 2 (YY) | |
| | | Sperm | |
| | | Y | Y |
| Parent 1 (BB) | Ova | BY | BY |
| | B | BY | BY |

Expected ratio of phenotypes = all green

Human blood types

There are four main blood types in humans—A, B, AB and O type. ABO blood type is controlled by a gene found on chromosome number 1, and biologists have found that there are three alleles of this gene—**A**, **B** and **o**. A person can have only two of the three alleles, one on one chromosome and the other on its pair. Allele **o** is recessive, while both alleles **A** and **B** are dominant. Therefore, if a person inherits allele **A** from one parent and allele **o** from the other, they will carry **AO** alleles and will have A type blood.

But if a person inherits allele **A** from one parent and allele **B** from the other, the resulting blood type is **AB**. This blood type has features of both A type blood and B type blood, and not a blend of the two as you would get with incomplete dominance. This type of gene action is called **co-dominance**. The table below shows the relationship between the phenotypes and the genotypes.

| Alleles (genotype) | Blood type (phenotype) |
|--------------------|------------------------|
| AA or Ao | A |
| BB or Bo | B |
| AB | AB |
| oo | O |

Learning experience

Revise past work by having students write a personal reflection about one of the following topics.

- Everyone is unique. What does it mean to you to know that there is only one you in the world?
- How would you feel if you found out you had a genetic disorder that could have been tested for and prevented?

- If you were cloned for 'spare parts' for another sibling, how would you feel?
- If you could genetically modify something in the world, what would it be? Why?
- If you inherited a gene that protects you from a certain disease, would you be prepared to donate this gene to other people?
- How do you think genetic engineering will change our lives by 2020?



Activity

There are two parts to this activity. For each part, work in a group of 3 or 4 people.

Part A: Determining blood groups

Because there are three different alleles for blood type it is possible for children of a couple to:

- all have one blood type
- have two different blood types
- have three different blood types
- have four different blood types.

Use the table on the previous page to help you work out the genotypes of the parents and the children in each of the four situations above.

A woman of blood type B claims that a man of blood type A is the father of her two children, who have blood types AB and O. Explain whether her claim is true or false. Does your explanation prove that he is the father of the children?

Part B: Blood transfusions

Carefully read the following extract.

A person requiring a blood transfusion has to be carefully matched to the donor's blood.

The structure of certain molecules on the surface of the cell membrane of a red blood cell determines blood type. There are two types of surface molecules, called A and B, which are controlled by genes. For example, a person with allele A will have red blood cells with A type molecules. A person with allele B produces B type molecules, while alleles A and B together produce both A and B type

molecules, and gene o produces no surface molecules. Thus, there are four main blood types in humans.

Now if a person with A type blood is given B type blood, a reaction occurs resulting in a blood clot. These blood clots can block blood vessels in the heart, brain and other organs, and can be fatal.

The reason the clot forms is due to molecules called antibodies in the blood plasma (the clear part). Antibodies are produced when the body recognises that the surface molecules on cells are foreign. They destroy them by joining to them. This also happens to bacteria and 'foreign' blood.

People with A type blood produce anti-B antibodies. If this person is given B type blood, anti-B antibodies will be produced and will react with the type B blood cells forming a clot.

The table below shows the types of red blood cells and the antibodies that can be produced in the four different blood types.

What are antibodies? Why are they important in the human body?

Draw up a table showing a patient's four possible blood types. Then in another column show the possible blood types that could be given to each patient in a transfusion.

Explain why blood type O can be given to people with any blood type.

Which blood type can receive all the other types of blood?

| Blood type | Type of surface molecules on red blood cells | Antibodies found in plasma |
|------------|--|----------------------------|
| A | A | anti-B |
| B | B | anti-A |
| AB | A and B | neither |
| O | neither | both anti-A and anti-B |

Activity notes

- Obviously there are serious health and safety issues involved with students testing their own or others' blood, and regulations do not permit this. However, screened blood samples from the different blood groups are available from the Australian Red Cross Blood Service for use in practical classes. Arrangements and availability vary slightly from state to state, and some service charges may apply. For further enquiries, you can email <non-clinicalsupply@arcbs.redcross.org.au>.
- Further information on blood donation and blood products is available at <www.transfusion.com.au> and <www.donateblood.com.au>.
- Approval and supply of the blood samples may take one to two weeks.

Hints and tips

If students did a concept map pre-test, now is a good time to get them to do it as a post-test. Also, now is a good time for students to take out the set of questions they developed at the beginning of the chapter and see how many they can answer now. Most students will be delighted to discover how much they have learned.

Check! solutions

- 1 a A dominant gene is able to mask or hide the alternative gene, whereas a recessive gene will be hidden or masked by a dominant gene (and may 'skip' generations).
b An organism that is heterozygous for a characteristic has two genes that are different, whereas one that is homozygous has two genes that are the same.
c The genotype of an organism is the type of genes it has, whereas the phenotype is its physical appearance.
- 2 a The recessive gene is **g**.
b Two genotypes that produce the same phenotype are **GG** and **Gg**.
- 3 On average, this is true but there are many thousands of genes; some will be the same and some will be different, and they combine at random. Sometimes, a child will show more of its mother's characteristics and sometimes more of its father's.
- 4 You know that the genotype of the white mouse is **bb**. If the brown mouse is **Bb** then you would expect half of the litter to be white (**bb**). However, they are all brown so it is most likely that both of the mice are homozygous. The brown mouse would be **BB** and the white mouse **bb**.
- 5 When genes show incomplete dominance there is a third, intermediate phenotype. A good example is feather colour in budgies. When the features of each gene are expressed in the phenotype, the genes are said to be co-dominant. A good example is human blood types, in which one 'A' gene and one 'B' gene produces an 'AB' phenotype.

X-linked genes

In humans, there are a number of genes on the X chromosome that have no equivalent on the smaller Y chromosome. This is because the X is much larger than the Y chromosome. These genes are said to be **X-linked** and the characteristic is said to be sex-linked. Colour vision and blood clotting are two examples.

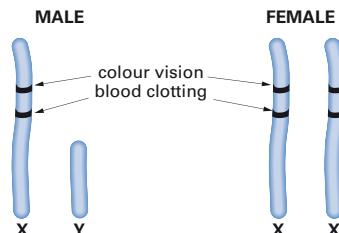
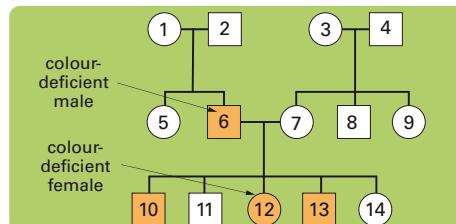


Fig 42

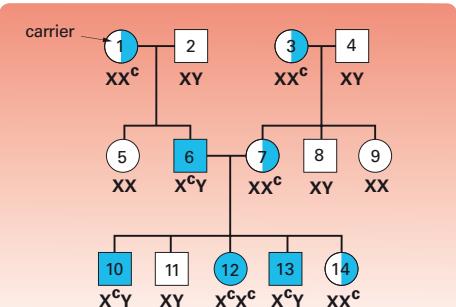
The genes for colour vision and blood clotting are found on the X chromosome. Females have two alleles for each characteristic but males have only one because there are none on the Y chromosome.

The allele for normal colour vision is dominant in humans (as you would expect). Now suppose a male is colour deficient (incorrectly called colour blind). He has an affected gene for colour vision on the X chromosome, but there is no matching allele on the Y chromosome. On the other hand, a female who has an allele for colour-deficient vision on one X chromosome and an allele for normal colour vision on the other, will have normal colour vision. However, she is called a *carrier* because she carries the affected gene for colour-deficient vision.

The pedigree below shows the inheritance of the gene for colour vision.



Male 6 is colour deficient and must have inherited the gene on the X chromosome from female 1 who is a carrier. Males 10 and 13 inherited their genes from female 7, while their colour-deficient sister, 12, got her genes from both her mother, 7, and her father, 6.



X^c = X chromosome with gene for colour deficiency

Check!

- 1 Explain the differences between these terms:
a dominant and recessive genes
b heterozygous and homozygous
c genotype and phenotype.
- 2 Suppose a characteristic in humans is represented by the alleles **G** and **g**.
a Which one is recessive?
b Show two genotypes that produce the same phenotype.
- 3 If you get half your genes from your mother and half from your father, why don't you have half your mother's features and half your father's?
- 4 Toby crossed a brown mouse with a white mouse. He discovered that all of the baby mice were brown. What can you infer about the genotypes of the parent mice?
- 5 Explain the difference between genes which show incomplete dominance and those that are co-dominant.

Learning experience

Have students add to their word and definition list by either writing a simple definition of each new word, drawing explanatory diagrams, adding to their audio file or making more labelled models.

Challenge solutions

- 1 You can represent the dominant gene for right-handedness by the letter 'R' and the recessive gene for left-handedness with the letter 'r'.
a Sperm R r
Ova all r Rr rr
- b So you expect a ratio of 2:2
The combination of genes is a random event and the more times it happens the closer the observed ratio will be to the expected ratio. The fact that in this case you expect 2:2 but you observe 3:1 is not unusual.

- 6 A farmer breeds a black rooster with a white hen. She finds that all the chickens have grey feathers.
- Use appropriate symbols to show how this happens.
 - What colour feathers should the offspring have if two of the grey-feathered chickens are crossed?

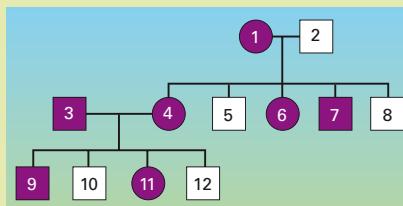


challenge

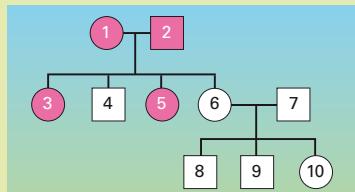
- Suppose that the allele that controls right-handedness is dominant over the one for left-handedness. A heterozygous right-handed male and a left-handed female have four children, three who are left-handed and one right-handed.
 - If the allele for right-handedness is called R, use a Punnett square to calculate the proportion of children that should be right-handed.
 - Suggest a reason for the difference between your result in a and the actual proportion of children with this characteristic in this family.
- When a homozygous hen with black eyes mates with a rooster with red eyes, all the chickens have black eyes.
 - Which allele for eye colour is dominant?
 - What is the genotype of the chickens?
 - Suppose two of the chickens mated when they matured. Use a Punnett square to find the genotypes and phenotypes of the chickens in the next generation.
- In guinea pigs short hair is dominant over long hair. Explain how you could get some long-haired baby guinea pigs if you had a short-haired female and you had to buy a male guinea pig.
- Muscular dystrophy is an inherited disease caused by an X-linked recessive gene. A man and a woman without the disease have a son with muscular dystrophy.
 - What are the genotypes of the parents?
 - What are the chances that their next son will have the disease?
 - What are the chances that their next female child will have the disease?
- In humans, the allele for long eyelashes is dominant over the allele for short eyelashes. In the pedigree in the next column the shaded circles and squares show people with long eyelashes.

- 7 Explain in simple language what the following statements mean:
- The gene for blood clotting is an X-linked gene.
 - Females can be carriers for X-linked genes but males cannot.
 - What would happen if a person with A type blood was injected with B type blood?

- Use appropriate symbols to work out the genotypes of the members in the pedigree.
- List the individuals who are definitely heterozygous and those who are definitely homozygous. Which ones are in doubt?



- 6 The ability to roll your tongue into a tube is an inherited characteristic in humans. In the pedigree below, the shaded individuals can roll their tongues. Work out if tongue-rolling is dominant or recessive. Then deduce the genotypes of the individuals.



- 7 You are a genetic counsellor advising a couple who wish to have a baby. The man tells you that his only sister has had a son and a daughter; the daughter died from Gaucher's disease, a non-X-linked recessive disease of the blood. The woman has no family history of the disease.
- Use a pedigree to find the chance of the man being a carrier for the disease.
 - What are the chances that the man's children will have the disease?

- 2 You can represent the gene for black eyes with the letter 'B' and the gene for red eyes by 'b'.
- The gene for black eyes is dominant.
 - The genotype of all of the chickens is Bb.

| | B | b |
|-----|----|----|
| Ova | BB | Bb |
| b | Bb | bb |

Approximately $\frac{3}{4}$ of the chickens will be black-eyed and approximately $\frac{1}{4}$ will be red-eyed.

Check! solutions

- 6 This is a good example of incomplete dominance because there is a third, intermediate phenotype. You can represent the gene for black feathers with 'B' and the gene for white feathers with 'W'.
- Parents
Black rooster \times White hen
Genotypes BB \times WW
Sperm and ova all B, all W
Chickens' genotype all BW
Chickens' phenotype all grey
 - Parents
Grey rooster \times Grey hen
Genotypes BW \times BW
Sperm and ova $\frac{1}{2}$ B, $\frac{1}{2}$ W \times $\frac{1}{2}$ B, $\frac{1}{2}$ W
Chickens' genotype $\frac{1}{4}$ BB, $\frac{1}{2}$ BW, $\frac{1}{4}$ WW
Chickens' phenotype $\frac{1}{4}$ black, $\frac{1}{2}$ grey, $\frac{1}{4}$ white
 - a The gene for blood clotting is on the X chromosome. Human females have two X chromosomes and, therefore, two genes for blood clotting, whereas human males have only one.
b Because females have two genes for this characteristic they can carry one recessive gene and still have the normal phenotype.
 - A person with blood type A will produce antibodies to blood type B. These will combine with the injected B type blood to form clots, which can block blood vessels and threaten life.

- 3 You should buy a long-haired male guinea pig. If the female is homozygous, however, none of the first generation will be long-haired. In this case, the offspring will have to interbreed to produce some with long hair.
- 4 You can represent the gene for muscular dystrophy as X^d and the normal gene as X^D . There is no gene for muscular dystrophy on the Y chromosome.
- The genotypes of the parents are: father $X^D Y$ and the mother $X^D X^d$.
 - The chance that their next son will also have muscular dystrophy is 1 in 2 because there is a 50% chance that he

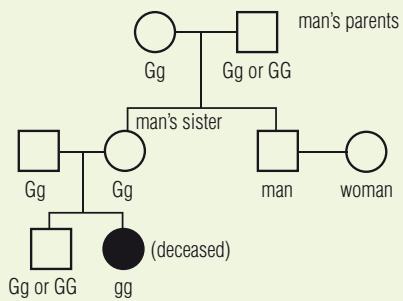
- will get the defective X chromosome from his mother.
- c The chance that the next female child will have the disease is zero because she will always get a normal dominant gene from her father.

- 5 You can represent the gene for long eyelashes with the letter 'L' and that for short eyelashes with the letter 'l'.
-

| Individuals | Genotype |
|-----------------|------------------------|
| 2, 5, 8, 10, 12 | must all be ll |
| 1, 3, 4, 6, 7 | must all be Ll |
| 9, 11 | can be either LL or Ll |

- b** As shown in the table in **a**, the individuals who must be heterozygous are 1, 3, 4, 6 and 7. The individuals who must be homozygous are 2, 5, 8, 10 and 12. Those who could be either are individuals 9 and 11.
- 6** The gene for tongue rolling must be dominant (T) because it cannot be recessive. If it was recessive all of the children of 1 and 2 would be able to roll their tongues. You can deduce that:
- Individuals 4, 6, 7, 8, 9 and 10 are all tt.
 - Individuals 1 and 2 must be Tt.
 - Individuals 3 and 5 can be either TT or Tt.

- 7** Assume that the man's sister has a son and a daughter with Gaucher's disease. Using a pedigree such as the one below, and using the letters 'G' and 'g' to represent the genes:



- a** If both of the man's parents are Gg, the chance that the man is heterozygous is 67%. If one parent is GG and the other Gg, the chance that the man is heterozygous is 50%.
- b** The woman has no family history of the disease and is very unlikely to be heterozygous. Therefore, the chance of them having children with the disease is zero.



Science in action

Genetic counselling

Mrs Van Tronk has just given birth to a baby boy. A few days later she and her husband are asked whether they will permit their baby boy to be tested for the presence of Duchenne muscular dystrophy, or DMD, an X-linked disease which develops mainly in boys and occasionally in girls. It causes the muscles of the lower limbs to weaken and waste away, and eventually control of all muscle movement is lost.



Fig 45

Duchenne muscular dystrophy is an inherited disease caused by a defective gene on the X chromosome.

After careful consideration, the Van Tronks give permission for the test to be done. Doctors first test the blood for the presence of a particular enzyme which indicates muscle damage, but this is not a conclusive test for DMD. They find the enzyme in the baby's blood, so a DNA test is done to see whether the baby has the DMD gene.

The results of the DNA test show that baby Van Tronk has the gene for DMD. The parents are then counselled by medical and social experts and are made aware of the difficulties of raising a DMD child, and also of the help that is available.

Two years later, Mrs Van Tronk becomes pregnant once more. The Van Tronks know that there is a chance that this baby might also develop DMD. Because the baby is an 'at risk' baby, they make an appointment to see a genetic counsellor. The genetic counsellor explains to them about chromosomes and X-linked genes. They are told that the disease is caused by a gene on one of Mrs Van Tronk's X chromosomes, so that if the developing baby is a boy, he has a 50:50 chance of suffering DMD. The counsellors also discuss how X-linked genetic diseases run in families, and trace her family history with the aid of a pedigree. The counsellors suggest two possible courses of action.

- Test the developing baby's chromosomes to determine the sex of the child and whether it carries the DMD gene. The mother can continue with the pregnancy or terminate it within 28 weeks.
- Have no tests, but be aware that the child may have DMD if it is a boy, and be prepared to care for him.

Questions

- 1 Work with two or three other people and draw up a list of options that are available to the Van Tronks.
 - a Make a decision on what you would do if you were a Van Tronk. Give reasons for your decision.
 - b Suggest what help and support may be necessary for the Van Tronks with each of the options.
- 2 Should it be compulsory to test all pregnant women to find out if their offspring have genetic abnormalities? Give reasons for your opinion.
- 3 Under United States law, discrimination based on genetics is banned. At present, this is not so in Australia. How do you think Mrs Van Tronk could be discriminated against if her gene file was known?
- 4 Go to www.scienceworld.net.au and follow the links to **Gene Testing**. You will find information about gene testing, the pros and cons of the procedures and the current regulations.

Learning experience

In small groups, have students construct a Y chart on a large sheet of paper (butcher's paper). They should label the three sections *Medical specialist*, *Parents* and *Child*. Each group is to brainstorm thoughts, questions, viewpoints and feelings surrounding genetic counselling— from their perspective. To get students

started, have them imagine they are the medical specialist having to explain to the parents that their unborn baby has a genetic disorder, then they are the parents receiving the news, then the child. Once finished, each group could walk around the room and view the responses given by others. How are they different from their own?

Learning experience

An interesting book which students might like to read is *My sister's keeper* by Jodi Picoult. It is about the ethical dilemma faced when a couple genetically engineer a baby to create a bone marrow match for their other daughter, who is terminally ill.



Copy and complete these statements to make a summary of this chapter. The missing words are on the right.

- 1 ____ are found in the nuclei of cells. They carry ____ which determines what an organism looks like and how it functions.
- 2 Body cells contain pairs of chromosomes while ____ contain only single chromosomes. When fertilisation occurs, the single chromosomes form pairs in the cells of the new organism.
- 3 Sex in humans is determined by sex chromosomes—females have a pair of ____ and males have an X and a ____.
- 4 Chromosomes are made of _____. The sequence of the ____ on the DNA determines which types of proteins will be made.
- 5 ____ alter the sequence of bases in cells, and occur spontaneously or from exposure to ____ or certain chemicals.
- 6 The whole of an organism's genetic information found in its DNA is called its _____.
- 7 Different versions of a gene, called _____, are found at the same location on a pair of chromosomes.
- 8 For an inherited characteristic, the _____ form of the gene masks the recessive one.
- 9 The _____ of an organism is the types of genes it contains, whereas its physical characteristics are called its _____.

alleles
bases
chromosomes
DNA
dominant
genes
genome
genotype
mutations
phenotype
radiation
sex cells
X chromosomes
Y chromosome

Try doing the Chapter 8 crossword on the CD.



- 1 Chromosomes are found in:
 A sex cells only
 B all cells
 C fertilised eggs only
 D animal cells only
- 2 A gene is:
 A a chromosome
 B a molecule of DNA
 C part of a chromosome that carries a single instruction
 D one base on a molecule of DNA
- 3 Tongue rolling in humans is controlled by a single gene. Which one of the following statements is correct?
 A Both genes for tongue rolling came from the male parent.

- B** Both genes came from the female parent.
C One gene came from each parent.
D Two genes came from each parent.
- 4 Horses have a total of 64 chromosomes in each of their body cells. Male horses have an X and a Y chromosome in their cells.
 - a** How many pairs of chromosomes are found in the body cells of horses?
 - b** How many chromosomes are found in the sperm of a horse?
 - c** Which chromosomes do sperm carry?
- 5 A strand of DNA contains the bases AAGTC.
 - a** What is the sequence of bases on the other matching strand of DNA?
 - b** How are the two strands of DNA held together in the double helix?

Main ideas solutions

- 1 chromosomes, DNA
- 2 sex cells
- 3 X chromosomes, Y chromosome
- 4 genes, bases
- 5 mutations, radiation
- 6 genome
- 7 alleles
- 8 dominant
- 9 genotype, phenotype

Review solutions

- 1 B
- 2 C
- 3 C
- 4 **a** 32 pairs
b 32 single chromosomes
c An X chromosome and 31 others, or a Y chromosome and 31 others.
- 5 **a** TTCAG
b The two strands of DNA are held together by weak bonds between the base pairs on each strand.

- 6** **a** 7 amino acids
b asparagine-serine-glutamic acid-phenylalanine-proline-arginine-serine
c The mutation will change the phenylalanine in the sequence to leucine.
d The change in the amino acid sequence might stop the action of the gene and hence your blood would not clot when your skin was cut or damaged.
- 7** **a** The gene for blood type O is recessive, therefore Mrs Sloan with blood type O is definitely homozygous.
b The daughter's genotype is oo. One of these genes came from her father. Therefore, Mr Sloan's genotype must be Ao.
c Baby Sloan has a 50:50 chance of having A type blood (as shown below).

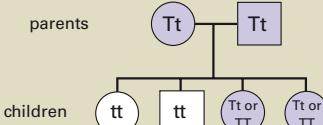
| father | | mother | |
|--------|---|--------|------------|
| A | o | o | Ao oo |
| o | | | Ao oo |
| | | | |

- 8** **a** SS
b The plant would have smooth seeds because the gene S is dominant.
c ss
d The gene for wrinkled seeds (s) is recessive. Therefore for a plant to have wrinkled seeds its cells would have to contain two genes for wrinkled seeds (ss).
- 9** **a** The genotypes of the parent plants are TT and Tt. They are both tall.

| | |
|---|------------|
| T | T |
| t | Tt Tt |
| T | TT TT |

- b** All the plants are tall.
10 Suppose the gene for tongue-rolling is T and the gene for non-tongue-rolling is t. Then the phenotype for the daughter and son who cannot roll their tongues is tt. The father and mother must have genotypes Tt because they can both roll their tongues and have children who cannot.

The purple circles and squares indicate those who can roll their tongues.



REVIEW

- 6** A small section of DNA has the following sequence of bases.

TTAAGACTCAAGGGGTCTCA

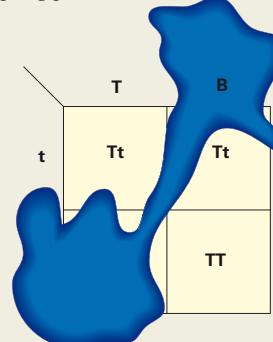
- a** How many amino acids does this section code for?
b Use the table on page 189 to work out the sequence of amino acids in the section of DNA.
c A mutation changes the triplet AAG to GAG. How will this affect your answer to **b**?
d Suppose the section of DNA is part of the gene in humans that makes your blood clot when blood vessels have been damaged. Suggest what might happen if this mutation did occur.
- 7** The Sloans have just had a baby boy. They also have another son and a daughter. Mrs Sloan is blood type O while her husband is blood type A.

- a** Which parent is definitely homozygous for blood type?
b If the daughter has O type blood, what blood genotype does Mr Sloan have?
c What are the chances that baby Sloan will have A type blood?

- 8** In corn plants, there are two types of seeds. The gene for smooth seeds (S) is dominant over the gene for wrinkled seeds (s).
- a** If a plant is homozygous and has smooth seeds, what are its alleles?
b What is the phenotype of a plant with the alleles Ss? Explain.
c What is the genotype of a plant with wrinkled seeds?
d Why would a plant with wrinkled seeds have to be homozygous?

- 9** The gene for height in pea plants has two alleles. The allele for tallness is dominant over the one for shortness. Joshua used a Punnett square to predict the results of a cross between two pea plants. Unfortunately, he spilt some ink over his notebook. From his ink-stained calculations:

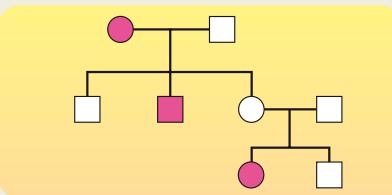
- a** work out the genotypes and phenotypes of the parent plants.
b calculate the ratio of phenotypes in the offspring plants.



- 10** A couple have four children—three girls and a boy. The father, mother and two of the daughters can roll their tongue, while the other daughter and son cannot. Tongue-rolling is controlled by a single gene. If the allele for tongue-rolling is dominant, use a pedigree to work out the possible genotypes and phenotypes of all the members in the family.

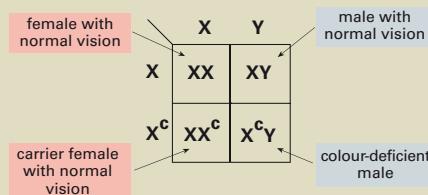
- 11** Colour-deficiency is an X-linked characteristic. Use pedigrees and the symbols X, X^c and Y to explain why males are much more likely to be colour deficient than females.

- 12** The people in the pink-shaded shapes in the pedigree below have a particular characteristic. Use the information in the pedigree to work out whether the allele for the characteristic is dominant or recessive.



Check your answers on pages 335–336.

- 11** Suppose a woman with a gene for colour deficiency on one X chromosome (XX^c) has children with a man with normal colour vision (XY). The Punnett square below shows (in theory) that the female children will have normal vision but half of them will carry the colour-deficiency gene. In the male children, 50% of them will be colour deficient.



- 12** The characteristic is definitely recessive. If you represent the alleles as A and a, then the people with aa genes have the characteristic. You can see from the pedigree that this characteristic has 'skipped' a generation. The child in the bottom row has it, while her parents in the line above do not.

