

1.2

Practical activities

1 Observing mitosis

Purpose

To observe mitosis in plant roots.

Materials

- prepared slides of onion root tips
- microscope

Procedure

- 1 Using a prepared slide and the low power on the microscope, focus on cells just behind the tip of the root (Figure 1.2.17).
- 2 Search for nuclei that appear to contain threads instead of appearing as dark circles. These are the cells that will be undergoing mitosis.
- 3 Focus on these cells and then switch to high power and focus on a cell where the chromosomes are clearly visible.
- 4 Find other cells that seem to be in different stages. For example, look for evidence of two newly formed cells.

Results

- 1 Draw diagrams of the cells you have found.
- 2 Organise your diagrams so that they represent the process of mitosis.
- 3 Draw a diagram showing where in the root mitosis is taking place.

Discussion

- 1 **Discuss** whether or not you would expect all cells in a root to be undergoing mitosis.
- 2 **Use** your observation to **assess** whether all of the cells in the area of the root tip you looked at were undergoing mitosis.
- 3 Growth due to mitosis occurs near the tip of the root rather than right on the tip or further back. **Propose** the benefits to the plant of this arrangement.

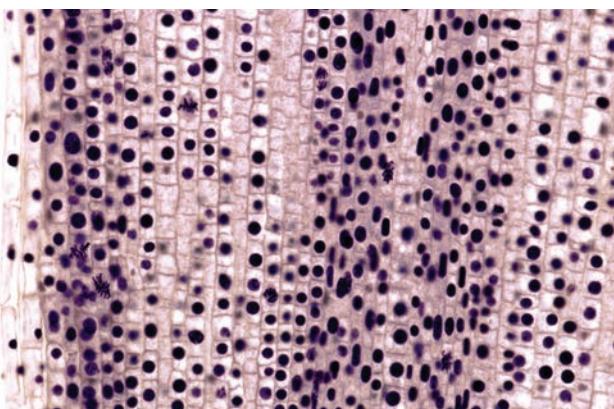


Figure
1.2.17

Stained onion root tip cells

2 Observing meiosis

Purpose

To observe meiosis in the anther of a flower.

Materials

- prepared slide of an anther
- microscope

Procedure

- 1 Using a prepared slide and the low power on the microscope, focus on cells inside the anther.
- 2 Search for nuclei that appear to contain threads instead of appearing as dark circles. These are the cells that will be undergoing meiosis.
- 3 Focus on these cells and then switch to high power and focus on a cell where the chromosomes are clearly visible.
- 4 Find other cells that seem to be in different stages. For example, look for evidence of two or four newly formed cells.

Results

Draw diagrams of the cells you have found.

Discussion

- 1 **Compare** your drawings and then place a number beside each diagram to represent the order they would appear in the process of meiosis.
- 2 **Explain** why meiosis would be occurring in the anther of a flower.
- 3 **Explain** how many chromosomes the gametes will have compared with the cell that divided to form them.
- 4 a **Propose** where else in a flower you could look for meiosis taking place.
b **Justify** your proposal.

1.3

Characteristics and inheritance



INQUIRY science 4 fun

Family resemblances

What are your family's traits?



Collect this ...

- photograph of close family members who are your relatives, such as your parents, grandparents, siblings, aunts and uncles (It is often easier to see resemblances in a photograph than by looking at the real person.)
- If you do not have photographs of close family, then find a suitable photograph of another family by searching the internet.

Do this ...

Observe the photographs to see where there are similarities.

Record this ...

Describe the resemblances you found.

Explain the relationship between the people displaying these characteristics.

At some time you have probably been told that you look like other members of your extended family. Relatives make comments such as 'He's got his father's ears' or 'Her hair is the same colour as great Aunt Madge's was when she was young'. Humans are fascinated by inheritance and the ways that characteristics pass from one generation to the next.

Discovering genetics

Genetics is the study of inherited characteristics called traits. In Austria in 1856, a monk called Gregor Mendel (1822–84) (Figure 1.3.1) carried out experiments on pea plants. The results of these experiments led him to construct theories that became the basis for the study of modern genetics, and are still recognised and used today.



Gregor Mendel was not a world-renowned scientist. He was the only son of farmers who lived in the area now known as Austria.

Figure 1.3.1

Dominant/recessive inheritance

In one series of experiments, Mendel worked with pure-breeding red-flowered pea plants and pure-breeding white-flowered pea plants. In **pure-breeding** lines, all the individuals have the same genetic information. Therefore, only red-flowered offspring could be produced from red-flowered parents and white-flowered offspring from white-flowered parents.

When pollen from red-flowered plants was used to cross-pollinate the white-flowered plants, all the plants in the next generation produced red flowers. Mendel called the red characteristic the **dominant** characteristic. The dominant characteristic is the characteristic that can be observed in the appearance of the individual. The other characteristic he called the **recessive** characteristic—the one that remained hidden.

Mendel then cross-pollinated these red flowers of the first generation, or F_1 generation. Each set of crosses that he performed produced both white and red flowers in the next generation—the F_2 generation. As shown in Figure 1.3.2, the proportions were roughly $\frac{3}{4}$ red flowers and $\frac{1}{4}$ white flowers.

The conclusion from these results is that genes work in pairs to determine which characteristic is shown or expressed. The gene that was studied in this situation was the gene for flower colour and this gene came in two varieties. Variations of genes are known as **alleles**. In this instance, the gene for flower colour had an allele for the red flower trait and an allele for the white flower trait.

When studying crosses and potential characteristics of offspring, geneticists use shorthand conventions. The dominant allele is represented by an upper-case letter related to the name. In this case, the red flower allele could be represented by the letter R (pronounced as 'big r'). The recessive characteristic (white flower) is then represented by the lower-case of the same letter—r (pronounced as 'little r'). By using R and r, it shows that a particular gene is being discussed.

An RR ('big r big r') combination of alleles will produce a red flower. Rr ('big r little r') will also produce a red flower because red is dominant to white. Only rr ('little r little r') will produce a white flower.

When an individual has two alleles the same, such as the RR and rr combinations shown in Figure 1.3.3, then the individual is said to be **homozygous** for that allele. The individual is described as a homozygote. Individuals with the Rr combination of alleles are **heterozygous**. These individuals are heterozygotes.

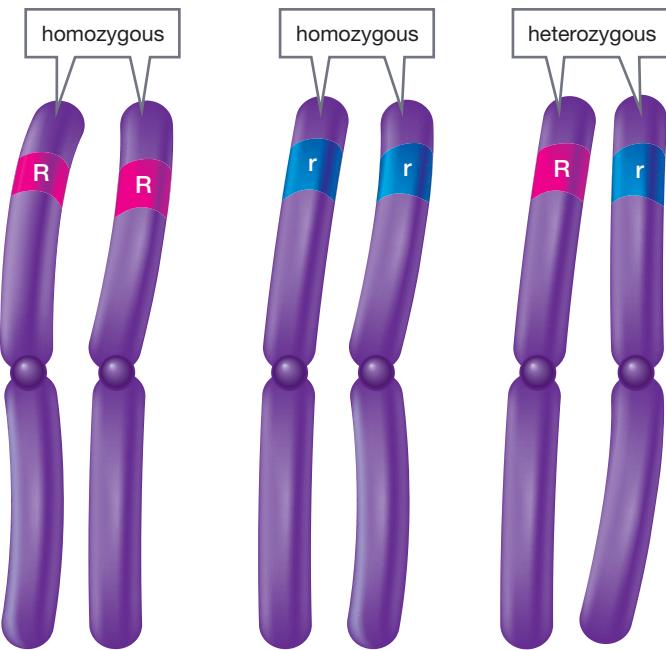
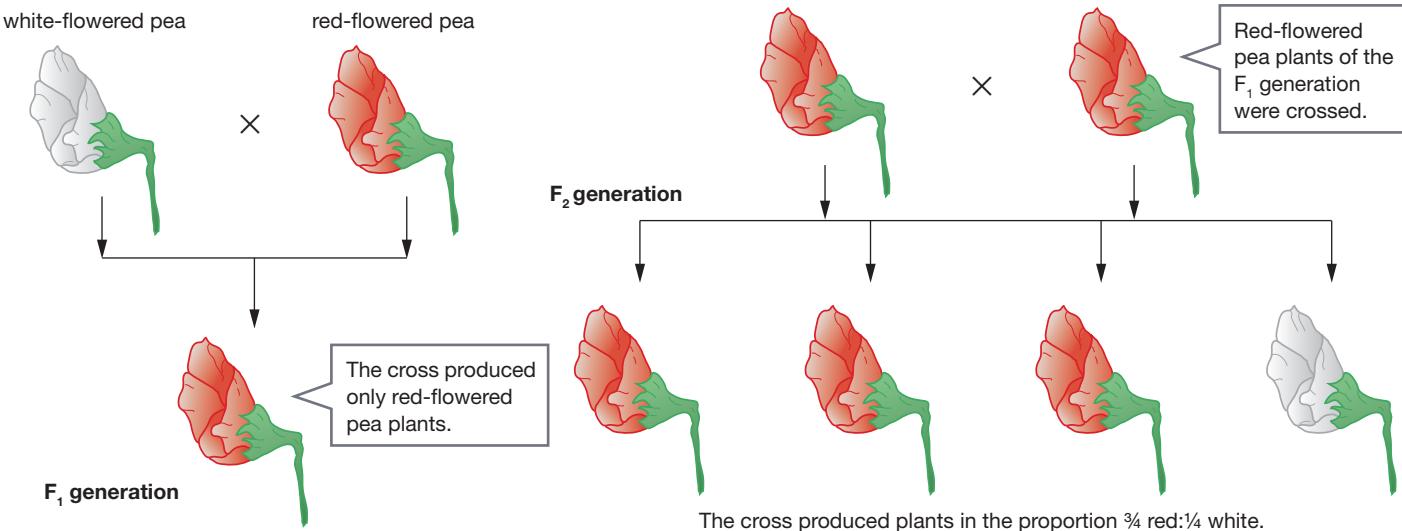


Figure 1.3.3

Individuals with the pairs of homologous chromosomes on the left and centre are homozygous. One is homozygous for the dominant allele (RR). The other is homozygous for the recessive allele (rr). The chromosomes on the right belong to an individual that is heterozygous (Rr).

Figure 1.3.2

The results of some of Mendel's experiments showing the flower colours appearing in the first (F_1) and second (F_2) generations of offspring.



The cross produced plants in the proportion $\frac{3}{4}$ red: $\frac{1}{4}$ white.

When working out the possible characteristics of offspring, it is important to refer back to meiosis and the movement of the genes as gametes are produced. Figure 1.3.4 demonstrates how gametes of a heterozygote end up with the different alleles of the flower colour gene.

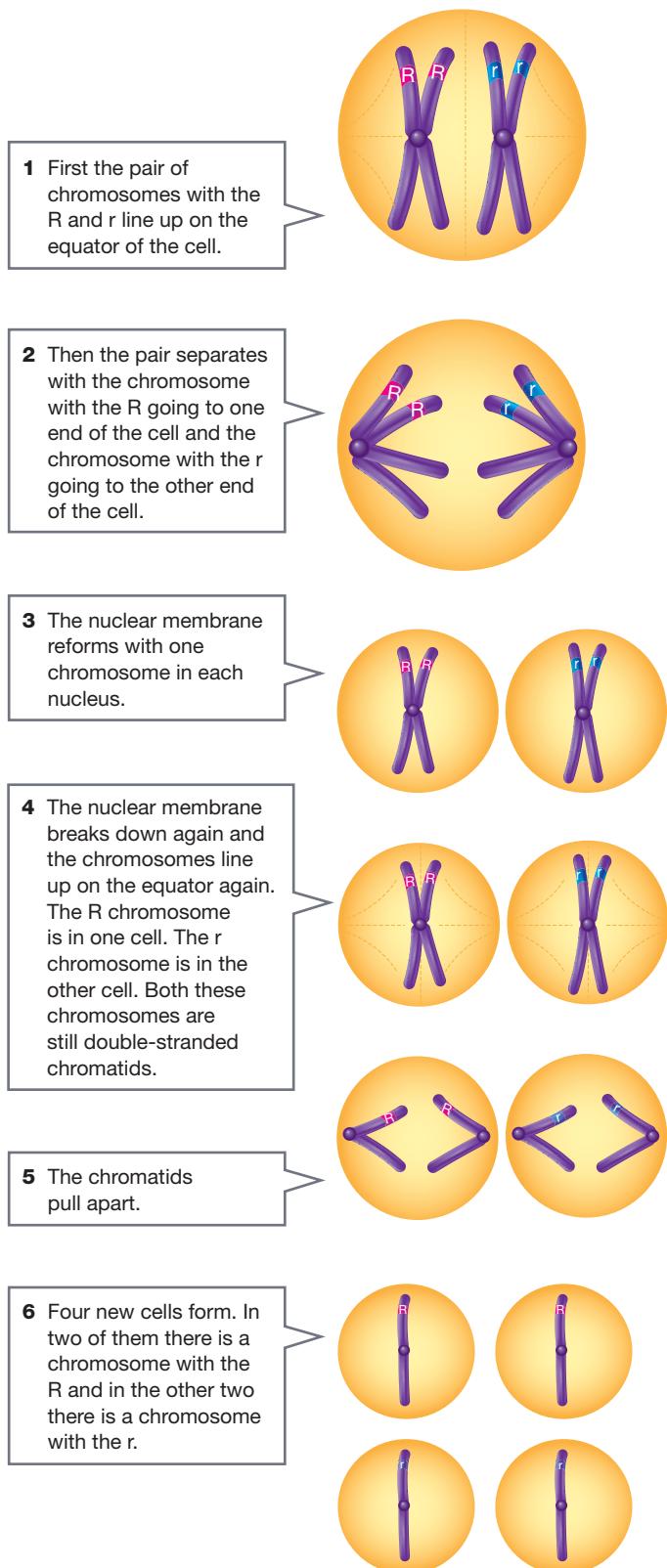


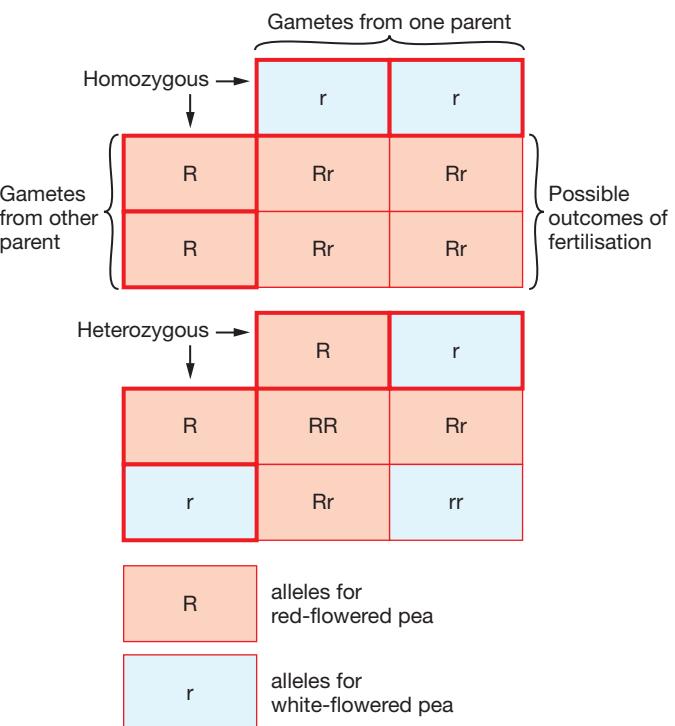
Figure 1.3.4

The behaviour of chromosomes during meiosis determines the alleles that end up in each of the gametes.

Fertilisation then determines which characteristics are present in each of the offspring.

Punnett squares—like those shown in Figure 1.3.5—are one way of showing all the possible types of offspring that could result from a cross. You cannot assume that the offspring will appear in exactly this order and in this exact ratio. It represents a probability.

In a Punnett square the possible gametes produced by one parent are shown across the top. The gametes from the other parent are shown down the side. In each square is a possible outcome of fertilisation.



The top Punnett square shows the possible outcomes of a cross between two homozygous individuals. The bottom Punnett squares shows the possible outcomes of a cross between two heterozygous individuals.

RR, rr and Rr represent the pea plants' **genotypes**—the actual genetic information carried by an individual.

The red or white colour of the flower is the **phenotype**—the observable characteristics of the individual.

In the example in Figure 1.3.5, RR and Rr have different genotypes but they both have the same phenotype—they both have red flowers.

Shared genes

All the variation between humans is caused by 0.1% of their DNA. The other 99.9% is identical no matter where you come from or what you look like.

We also share about 98% of our genes with chimpanzees and 90% with mice.





Predicting the results of a cross

It is possible to predict the results of a cross if you know the:

- genotypes of the parents
- relationship between the alleles of the trait (characteristic) of interest.

Consider two parents, P_1 and P_2 , who are both heterozygous for the gene F. Gene F has two alleles F and f. F is dominant to f.

What are the expected outcomes of a cross between P_1 and P_2 ?

As both parents are heterozygous their genotypes are both Ff and each can produce gametes containing an F allele or an f allele

A Punnett square can help you predict the outcomes.

		F	f
P ₁ gametes	F	FF	Ff
	f	Ff	ff

From the table the expected genotypes are:

$\frac{1}{4}$ FF, $\frac{1}{2}$ Ff and $\frac{1}{4}$ ff

Because F is dominant to f the expected phenotypes are:

$\frac{3}{4}$ trait F and $\frac{1}{4}$ trait f

If P_1 and P_2 had 12 offspring, you would expect about 8 trait F and 4 trait f.

WORKED EXAMPLE

Predicting the results of a cross

Problem

In the peas that Mendel studied, yellow colour is dominant to green colour. A pea plant heterozygous for the gene for colour is crossed with another pea plant heterozygous for the gene for colour and 100 offspring are produced.

Predict how many of the offspring will be yellow and how many will be green.

Solution

Step 1 Decide what letters you will give to the alleles for yellow and green colour.

As yellow is dominant—Y, green is recessive—y.

Step 2 Deduce the genotypes of the parent plants.

As both are heterozygous, they will have one of each allele and hence be Yy.

Step 3 Set up your cross in a Punnett square.

		P ₂ gametes	
		Y	y
P ₁ gametes	Y	YY	Yy
	y	Yy	yy

Step 4 Write down the expected genotypes.

$\frac{1}{4}$ YY, $\frac{1}{2}$ Yy, $\frac{1}{4}$ yy

Step 5 Work out the expected phenotypes.

$\frac{1}{4}$ YY + $\frac{1}{2}$ Yy = $\frac{3}{4}$ yellow

$\frac{1}{4}$ yy = $\frac{1}{4}$ green

Step 6 Work out how many of the 100 offspring will be yellow and how many will be green.

$\frac{3}{4} \times 100 = 75$ green

$\frac{1}{4} \times 100 = 25$ yellow



Incomplete dominance

Some genes do not have dominant and recessive alleles. Some alleles show **incomplete dominance**, and the appearance of a heterozygous individual results from a 'blending' of two such alleles. A heterozygote will look different from both its homozygous parents.

One example is in domestic chickens in which black feathers are incompletely dominant to white feathers. The heterozygous chicken has blue-grey feathers. The blue-grey colouring of cockatiels (Figure 1.3.6) is also the result of incomplete dominance.

The shorthand convention in this situation is to use uppercase letters related to the two different alleles such as B for black and W for white feathers. This is a reminder that neither allele is dominant to the other.

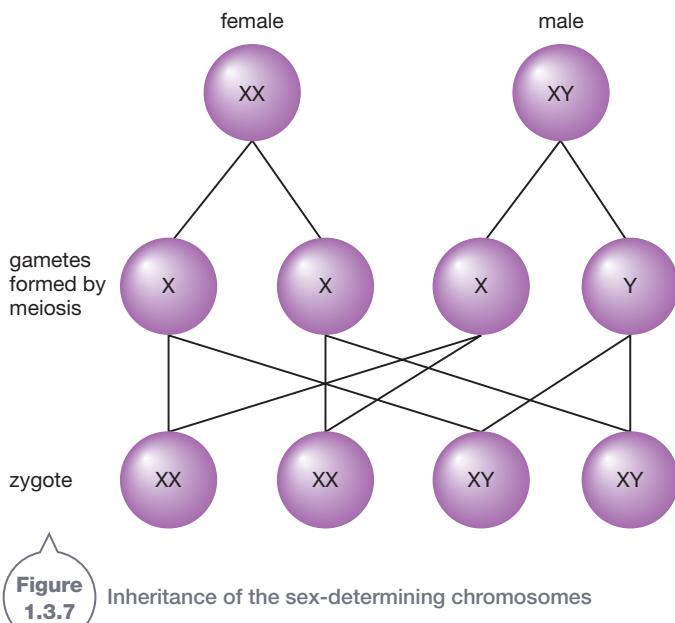


Figure 1.3.6

Feather colour in cockatiels is inherited in the same way as in chickens. The blue-grey colour of the feathers is the result of incomplete dominance.

Sex determination

Your two sex chromosomes determine which sex you are. Inheritance of these chromosomes can be seen clearly in Figure 1.3.7. All of the eggs produced by a female will have one X chromosome. Half the male's sperm will carry an X chromosome and the other half will have a Y chromosome. If a sperm containing an X chromosome fertilises an egg, then the offspring will be a female (XX). If a sperm carrying a Y chromosome fertilises an egg, then the offspring will be male (XY).



Sex linkage

Some genes are found on the X chromosome and not on the Y chromosome. These are called **sex-linked genes** because they are present on one of the chromosomes that are also responsible for the determination of sex.

The red-green colour-blindness gene is carried on the X chromosome. Normal vision (N) is dominant to red-green colour-blindness (n). Females who are heterozygous ($X^N X^n$) for colour-blindness will still have normal vision because the dominant allele masks the effect of the recessive allele. However, they are carriers of the allele. Carriers are able to pass the trait on to their children.

The Y chromosome does not carry a colour-blindness gene. Therefore, the only possible genotypes for a male are $X^N Y$ and $X^n Y$. In the genotype $X^n Y$, the recessive allele is the one that is expressed in the phenotype and the male is colour blind. Figure 1.3.8 demonstrates inheritance of the colour-blindness gene.

The daughter of a colour-blind male and a carrier female has a 50% chance of being colour blind. Therefore, colour-blind females are not common. If a normal female and a colour blind male have only male children, then the boys will not be colour blind and the disorder will not appear in subsequent generations unless it is re-introduced.

- Male normal vision
- Male colour blind
- Female carrier
- Female normal vision

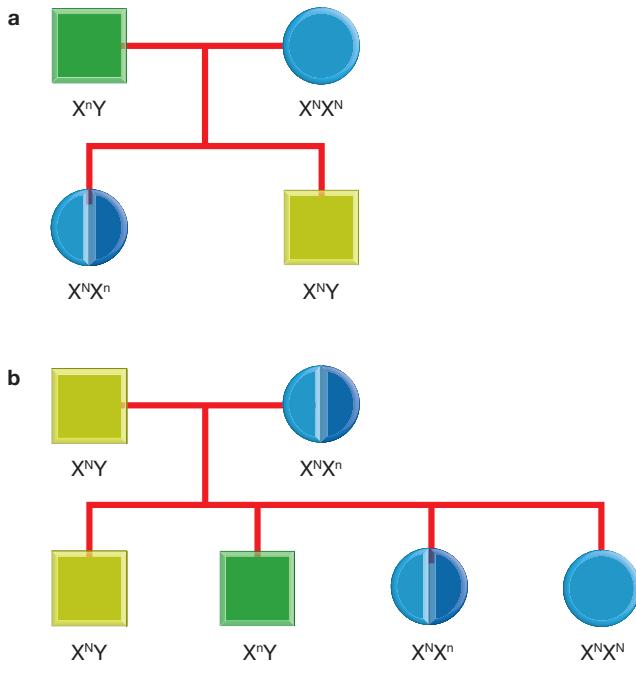


Figure 1.3.8

a The daughters are all carriers because they have inherited the colour-blindness allele from their father. However, their vision will be normal. The sons have normal vision.

b Half the sons of a carrier mother will have normal vision. The other half will be colour-blind. All the daughters will have normal vision, but half will be carriers of the disease.

Haemophilia and Duchenne's muscular dystrophy are two other examples of sex-linked characteristics.



Chromosomal abnormalities

Sometimes mistakes happen during meiosis when the sex cells are being produced and the information passed on to the next generation is changed.

If the chromatids fail to separate during meiosis, the child will be born with an extra chromosome or part of a chromosome. This is called chromosomal abnormality. Examples include Down syndrome and Klinefelter's syndrome.

Mistakes can happen as DNA is copied. The base sequence is changed and mistakes occur in the manufacture of proteins. This type of change is called a **mutation**. Mutations may arise spontaneously (by chance) or result from damage to a strand of DNA. UV radiation, nuclear radiation and certain chemicals such as nicotine and asbestos can cause mutations.

If the mutation occurs in the eggs or sperm, then there is a chance that they will be passed on to the next generation.

1.3

Unit review

Remembering

- 1 State what is meant by a pure-breeding line of plants.
- 2 Recall the term used to describe:
 - a alternative forms of gene
 - b an organism with different alleles for a particular gene
 - c the observable characteristics of an individual
 - d an individual with two copies of the same allele of a gene.

Understanding

- 3 Predict the number of chromosomes that would be found in the following human cells.

a ovum	b muscle cell
c skin cell	d sperm cell
- 4 Explain how sex is determined in humans.
- 5 Explain what is meant by sex linkage.

Applying

- 6 Use an example to explain the relationship between genes and alleles.
 - 7 Identify the option a–g that matches each description i–vii in the table below.
- | Symbol/Name | Description |
|--------------------|--|
| a Mm | i A dominant allele |
| b XY | ii A phenotype |
| c X ⁿ Y | iii Genotype of a homozygous individual |
| d M | iv Genotype of an individual heterozygous for dominant/recessive alleles |
| e PP | v A recessive sex-linked characteristic |
| f Red flower | vi Genotype of a male individual |
- 8 In guinea pigs, black coat is dominant to brown coat colour.
 - a Use an appropriate symbol to represent the alleles for coat colour.
 - b Use a Punnett square to demonstrate the genotype and phenotype of the offspring you would expect from a cross between heterozygous black and a homozygous brown guinea pig.
 - 9 Use genotypes to demonstrate how a human female could inherit the trait for red-green colour-blindness.

Analysing

- 10 Contrast:
 - a homozygous and heterozygous
 - b phenotype and genotype
 - c autosome and sex chromosome.

- 11 Analyse the following Punnett square where R is red and r is white. Identify the:

- a type of inheritance
- b ratio of genotypes in the offspring
- c ratio of the phenotypes and appearance in the offspring.

	R	r
r	Rr	rr
r	Rr	rr

Evaluating

- 12 Deduce the genotype of the parents, given the characteristics of the offspring.
 - a All the offspring for three generations had red flowers.
 - b All the plants in the study were tall but when the next generation came along about one-quarter of them were short.

Creating

- 13 a Design an experiment that you could carry out to determine the dominance or recessiveness of black coat and white coat in mice.
 
 - b Explain why this experiment would provide the evidence you need to make your decision.
- 14 Construct Punnett squares to show the F₁ and F₂ generations of a cross between a pure-breeding wild rabbit (AA) and an albino (aa) rabbit. Show both the genotype and phenotype of the offspring.
- 15 Construct a drawing of an imaginary animal or plant. Decide on a characteristic for which there will be two alleles. It could be something like flower colour or hair colour. Decide on letters to represent these alleles. Now make diagrams to represent the phenotype of your creature in each of the following situations. Include the genotype with each drawing.
 - One allele is dominant to the other and the creature is heterozygous.
 - The creature is homozygous for the recessive characteristic.
 - The gene is carried on the X chromosome.

Inquiring

Research the symptoms and effects on the individual of the following genetic diseases: Down syndrome, Klinefelter's syndrome, haemophilia, Duchenne's muscular dystrophy, cystic fibrosis, Huntington's disease and phenylketonuria. Present the results of your research in a table.

1.3

Practical activities

1 Chance variation

Purpose

To model the variation in potential offspring.

Materials

- die
- 20 cards about the size of playing cards
- marker pen
- the information in Table 1.3.1

Table 1.3.1

Each gene is on a different chromosome.		
Gene	Allele 1	Allele 2
1	Can roll tongue (T)	Cannot roll tongue (t)
2	Freckles (F)	No freckles (f)
3	Bent little finger (B)	Straight little finger (b)
4	Broad lips (L)	Thin lips (l)
5	Dimples (D)	No dimples (d)
Evens		Odds

Procedure

- Use the information in Table 1.3.1 to make two identical sets of cards. Each card represents an allele on one of the five pairs of homologous chromosomes. For example, there should be two cards saying ‘Can roll tongue (T)’, two saying ‘Cannot roll tongue (t)’. Follow this model until there are two cards for every allele. You will have 20 cards.
- The 20 cards represent five pairs of chromosomes from two individuals. Each pair of cards represents a gene for a characteristic and there are two alleles for each gene. Divide the cards into two sets of five pairs. These are the parents—P₁ and P₂. They are both heterozygous for each of the alleles.
- Copy Table 1.3.2 into your workbook and record the genotype and phenotype for each parent. Look carefully at the symbols used for the alleles to identify the type of inheritance.

Table 1.3.2

Gene	Parent 1		Parent 2	
	Genotype	Phenotype	Genotype	Phenotype
1				
2				
3				
4				
5				

- Now create the gametes. Start with gene 1 for parent 1. Toss the die. If an even number is tossed, then select the ‘Can roll tongue’ card from the evens list in Table 1.3.1. If an odd number is tossed, then select the ‘Cannot roll tongue’ card from the odds list in Table 1.3.1. Continue in this way until one allele for each gene has been selected for the P₁ gamete. Place the cards for the gamete to one side. They will be used in step 6.
- Create the P₂ gamete by repeating step 4 with the other set of cards.
- Take the two gametes you have created and arrange the cards into the pairs. They represent the genotype of the first zygote. Copy Table 1.3.3 into your notebook and record the genotype and phenotype of this zygote.
- Sort the cards back into the original piles for P₁ and P₂ and repeat the process of creating gametes and a zygote four more times.

Results

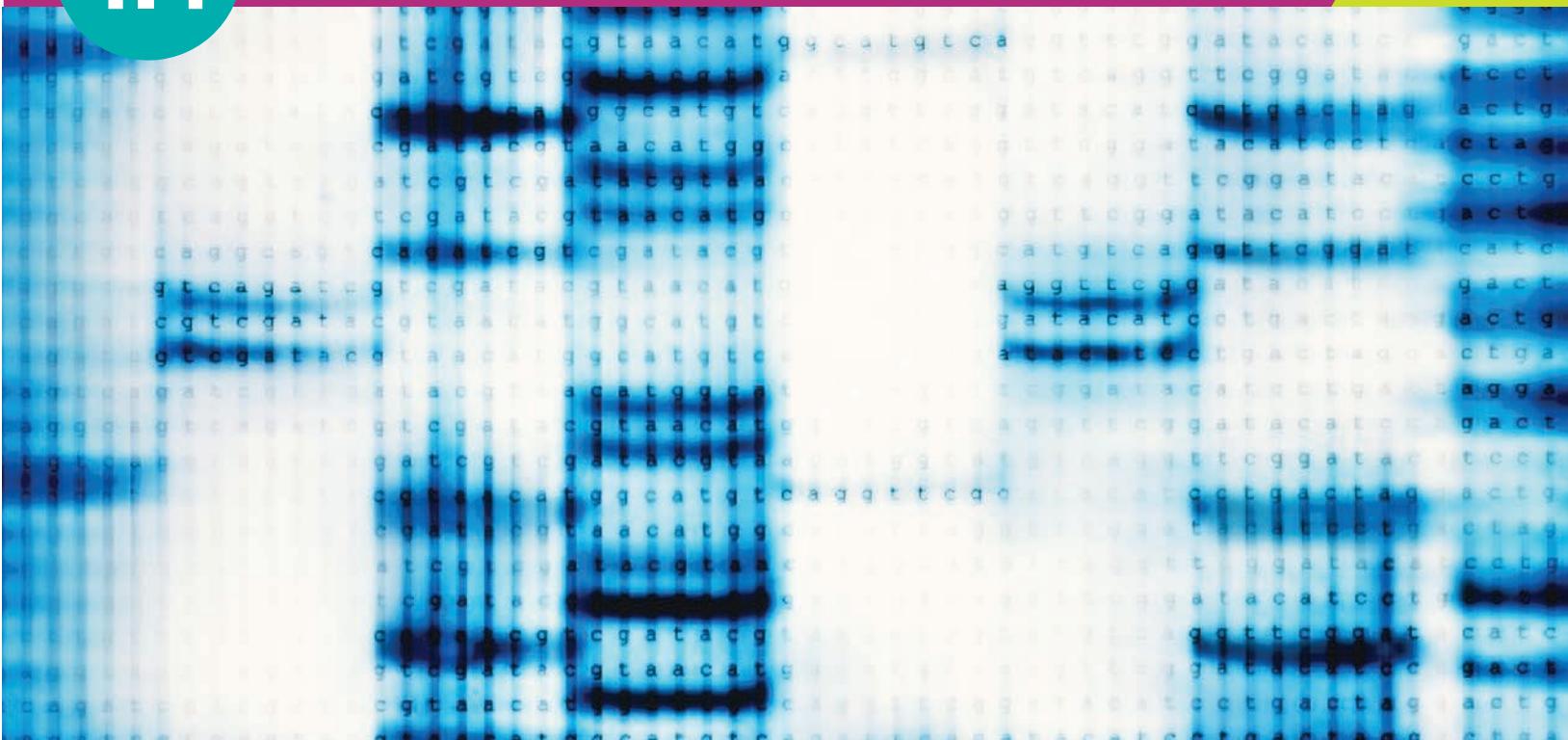
Copy and complete Table 1.3.3 by recording the genotype and phenotype of the zygote.

Table 1.3.3

Gene	Zygote 1		Zygote 2		Zygote 3		Zygote 4		Zygote 5	
	Genotype	Phenotype								
1										
2										
3										
4										
5										

Discussion

- Compare the zygotes you created.
- Explain why the zygotes were different when the genotypes of the parents were identical.
- In this model, you were looking at only five genes. Predict the amount of variation that would result if twice as many genes were modelled.
- Each chromosome in your body carries more than one gene. Deduce how that would affect variability in offspring.



As a young child, you probably played with building blocks like Lego®. Once you had worked out how the blocks fitted together, you were able to move them around to construct objects that were different from the pictures on the outside of the packaging. In a similar way, once scientists knew how the genetic information in living things was constructed, they experimented with ways of modifying it that were beneficial to humans; for example, to increase food production and improve human health.

Genetic modification

Scientists have developed gene technologies that enable plant cells to be **genetically modified**. In genetically modified (GM) organisms, the genetic information is changed by inserting new genes. The new genes are then copied to all the daughter cells when the parent cell divides by mitosis. These modified cells will mature (grow up) into a completely new strain of plant.

Using genetic modification, desirable traits such as insect resistance and increased nutrient value are added to plants. This technology has benefits but it also causes controversy.

Canola

Canola, shown in Figure 1.4.1, is a crop that produces edible oil. Western Australia is a major canola producer with exports valued at \$535 million in 2008–2009. By 2010, Western Australia, Victoria and New South Wales allowed farmers to grow GM canola. GM canola is resistant to herbicides that are commonly used to control weeds. Farmers can spray herbicide on the crop and kill the weeds but leave the canola unaffected. Production costs are reduced, enabling growers to compete in international markets.



Figure 1.4.1

Canola is used to produce cooking oil and is an important crop in Australia. Three Australian states allow cultivation of GM varieties of canola.

Rice

Rice is the main food source for more than half the world's population. White rice lacks essential minerals and vitamins, including vitamin A. Vitamin A deficiency is a cause of childhood blindness that affects up to 500 000 children worldwide each year. Golden rice-2 (Figure 1.4.2), is genetically modified using genes from daffodils, corn and bacteria. The rice contains beta-carotene, the chemical that gives carrots their orange colour, and which the body converts into vitamin A.



Figure 1.4.2

The beta-carotene in Golden rice-2 makes it appear yellow when compared with white rice.

About 225 grams of cooked Golden rice-2 would provide 50–60% of the recommended adult dietary allowance of vitamin A.

Golden rice-2 was developed to help people in developing countries where blindness due to vitamin A deficiency is a problem. However, there has been significant opposition from environmental and anti-globalisation groups to the commercial production of Golden rice-2. At present, this GM product is still grown for research but is not grown for human consumption.



More food needed

On World Food Day 2009, the Federal Government warned Australians that world food production will have to increase by 70% by the year 2050, with the population expected to boom to 9.1 billion.

SCIENCE

Spliced vegetables

What happens when genes are spliced?



Collect this ...

- carrot
- parsnip or small potato with approximately the same diameter as the carrot
- sharp knife

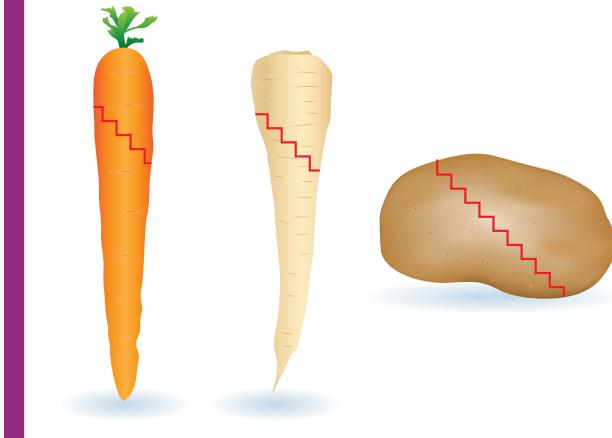
Do this ...

- 1 Use a step-like cut to cut the carrot into two parts across the middle.
- 2 Using the same pattern, halve the parsnip or potato.
- 3 Join one half of the carrot to one half of the parsnip or potato.
- 4 You have spliced the two vegetables together in a similar way to the way genetic engineers splice genes into chromosomes.

Record this ...

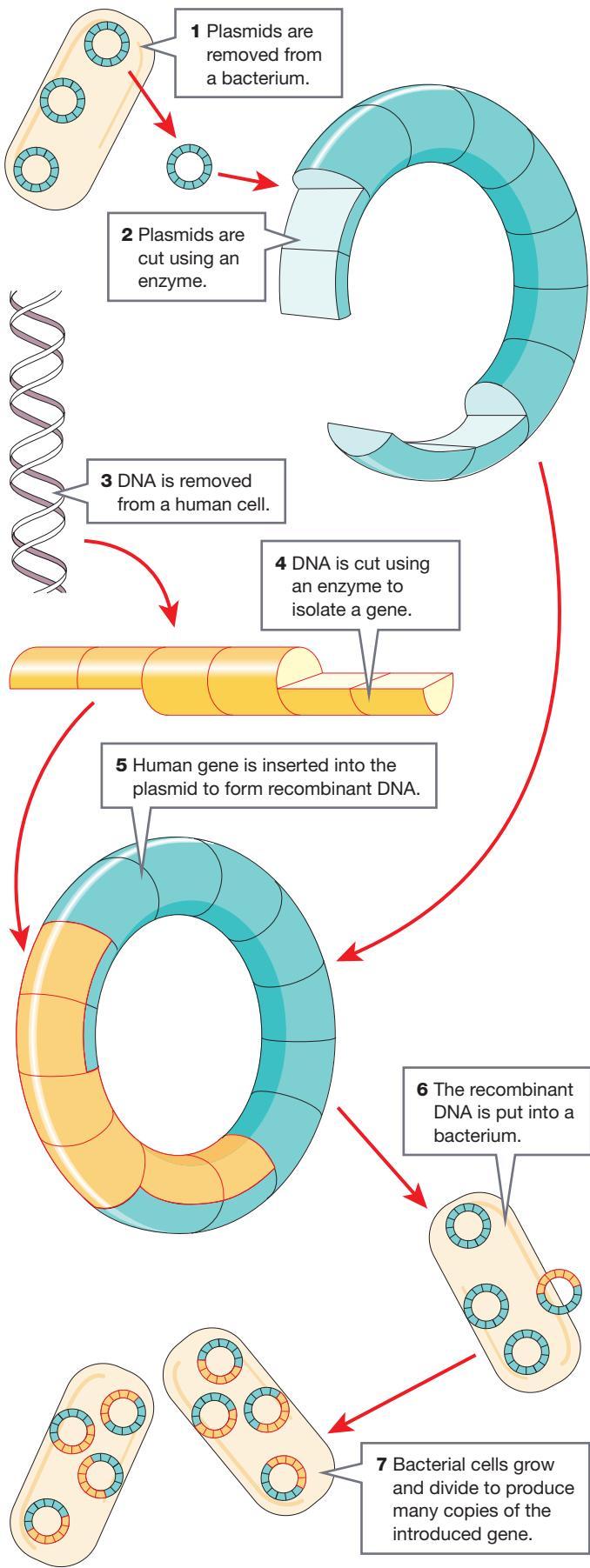
Describe what the final 'vegetable' looked like.

Explain why it was important to have the cuts on both vegetable a similar shape.



Gene splicing

Bacteria have DNA in chromosomes but they also have separate rings of DNA called **plasmids**. Using enzymes, scientists can cut these plasmids open and splice (insert) desirable genes into the plasmid. This process is called **gene splicing**. The technology of combining DNA from different genes is called **recombinant DNA technology** and the process is shown in Figure 1.4.3. Recombinant DNA technology has been used to splice the human gene that codes for insulin production into bacteria. These bacteria are stored in vats where they manufacture large quantities of human insulin for use by people with diabetes.



Human Genome Project

A **genome** is the genetic information carried by a haploid set of chromosomes. The **Human Genome Project** was an international project. It aimed to:

- identify all 20 000–25 000 genes in the human genome
- determine the sequence of the 3 billion base pairs that make up human chromosomes (Figure 1.4.4).

This information would be made available for further study and analysis. After working on the project for 13 years using very fast computers, a rough draft of the human genome was published in 2003. A more refined version followed in 2006. Although the project is finished, analysis of the data will continue for years.

All humans share about 99.9% of their DNA. Figure 1.4.5 on page 28 shows variation caused by the other 0.1%. Scientists have identified millions of locations that differ by only one base from one human to another. These differences, known as **single nucleotide polymorphisms (SNPs)**, may be associated with common diseases such as cardiovascular disease, diabetes, arthritis and cancers.



Figure 1.4.4

The genetic code appears as dark and light areas, indicating the genes.



Figure
1.4.5

Although people from different parts of the world may look quite different, only 0.1% of their DNA causes the differences.

Gene testing

Once the function of a gene is known, scientists are able to test for the gene. Over 400 genetic tests are available in Australia and people are using these tests for a variety of reasons.

Knowledge of your genetic make-up could help you avoid diseases that are controlled by lifestyle as well as genetics. For example, genetic testing could show that you were at risk of heart disease or type-2 diabetes. You could make lifestyle choices that may reduce your chances of developing these diseases.

Genetic testing can tell if people are carrying specific disease-causing genes that could be passed on to their children. Cystic fibrosis, thalassemia and Huntington's disease (Figure 1.4.6) are examples of genetic diseases. Knowledge of their genetic make-up can allow people to make decisions about whether or not to have children.

Genetic testing detects a particular problem gene. However, it cannot predict how severely the person carrying that gene

Gene technology has led to the situation where biotechnology firms are trying to patent genes and gene sequences. These are used for medical research or to develop drugs and other therapies. Patents are a way of recovering the costs of developing the treatments. These patented genes may be yours!



Figure 1.4.6

Huntington's disease is a genetic disorder that causes degeneration in the brain. The symptoms do not usually appear until the person is 35–50 years old. Before genetic testing was available, an affected person could have had children (possibly passing on the disorder) before realising they had the disease.

will be affected. For example, some people with the gene for cystic fibrosis have mild problems with their lungs; others may have severe lung, pancreatic and intestinal problems that have major effects on their quality of life and life expectancy.

Genetic testing can be used to diagnose genetic disorders in fetuses. Examination of whole chromosomes can indicate disease. For example:

- Down syndrome is the result of having an extra chromosome 21
 - Turner's syndrome is caused when a female only has one X chromosome
 - Fragile X syndrome is the most common inherited cause of mental retardation (Figure 1.4.7).

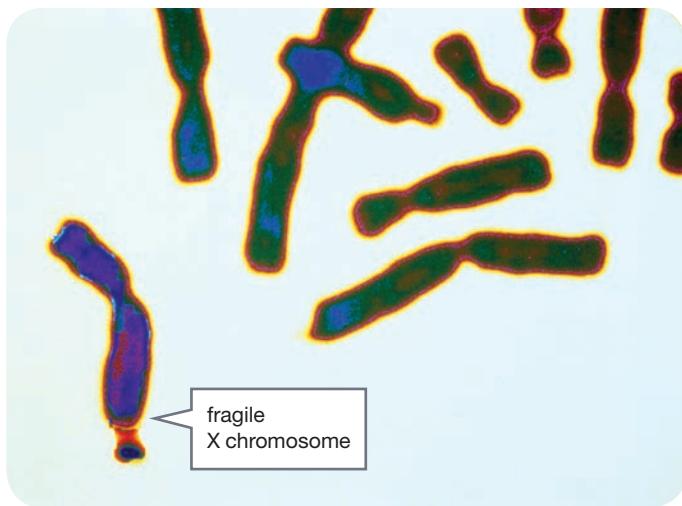


Figure 1.4.7

The defective end section of the fragile X chromosome is the fragile area. The body cannot use information in this area of the chromosome to manufacture a protein essential for normal brain development.

With knowledge of genetic disease in the fetus, parents can decide whether or not to continue with the pregnancy.

Other uses of genetic testing include:

- identification of a suspect in a criminal investigation by comparing their DNA with DNA found at a crime scene
- testing to identify the biological parent of a child in cases of adoption or disputed paternity (where the identity of the father is not known)
- analysis of the DNA of both the donor and the recipient (tissue typing) to reduce the chance of rejection in the case of bone marrow or organ transplantation.

Drawbacks

There are some drawbacks to genetic testing. For example, knowledge of genes that have the potential to cause disease may affect a person's ability to get life insurance cover. This does not only affect the individual. It affects the whole family.

If someone has disease-causing genes, other members of the family may carry the same genes. Family members then have to decide whether to be tested and what they will do with the knowledge once they get it.

Before getting tested and after receiving the results, individuals and families are offered information on the nature of the tests and counselling. This may help them to understand and cope with the results.

Gene therapy

Gene therapy has the potential to cure genetic diseases. In **gene therapy**, the defective gene is replaced with a normal gene. The idea is simple, but gene therapy is still in the experimental stage for most genetic diseases.

Cystic fibrosis is the most common genetic disease in Australia. A person with cystic fibrosis is homozygous for a recessive allele of a gene called CFTR. Cystic fibrosis has many

effects in the body. The main effect is on the lungs where thick mucus clogs the airways, making gas exchange difficult. Since 1989 when the CFTR gene was identified, scientists have researched ways of transferring normal CFTR genes into human lungs. So far they have not been successful.

Treating cancer

Cancerous tumours are fed by uncontrolled growth of abnormal blood vessels. In 2008, researchers at the Western Australian Institute for Medical Research discovered a gene that can be switched off. This reverses the growth of blood vessels inside tumours, making the blood vessels more normal in size. This is shown in Figure 1.4.8. The hope is that being able to control the blood vessels will eventually lead to control of the tumour itself.

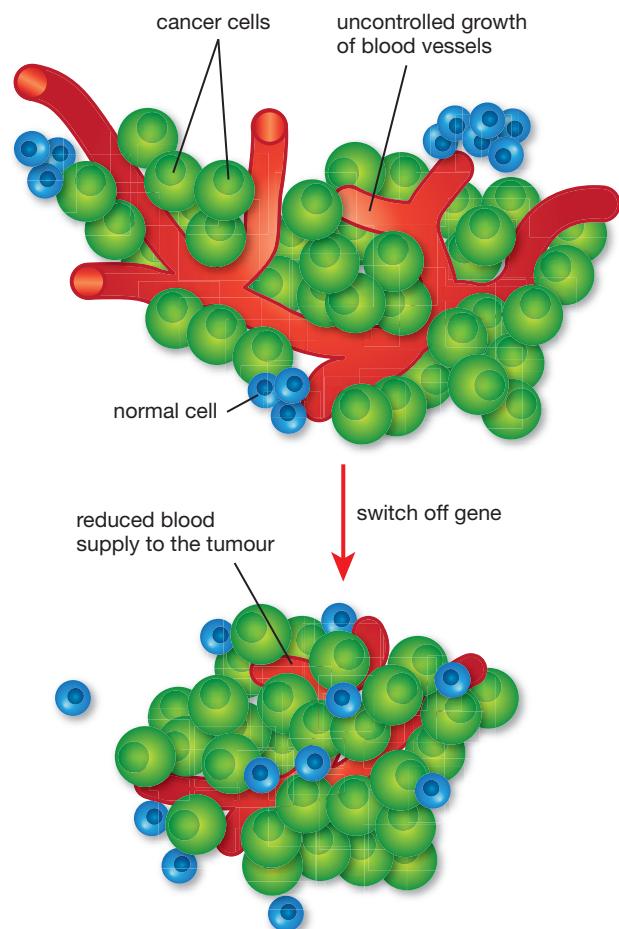


Figure 1.4.8

By switching off the RGS5 gene, the blood supply to a cancerous tumour is reduced.

Cancer

Cancer is not a single disease. It is many diseases all of which have similar characteristics. Cancers are uncontrolled cell growth that produces tumours. The tumours grow into and destroy nearby tissues. Cells break away from the tumours and may spread (metastasise) to other parts of the body through the lymphatic system.

SCIENCE AS A HUMAN ENDEAVOUR

Nature and development of science

Stem cells

Figure 1.4.9

Stem cells can become many different types of cells.



When an embryo is a few days old, it contains cells that are **pluripotent**. Pluripotent cells are capable of becoming any one of the 220 different cell types found in the human body. These cells are known as **embryonic stem cells**.

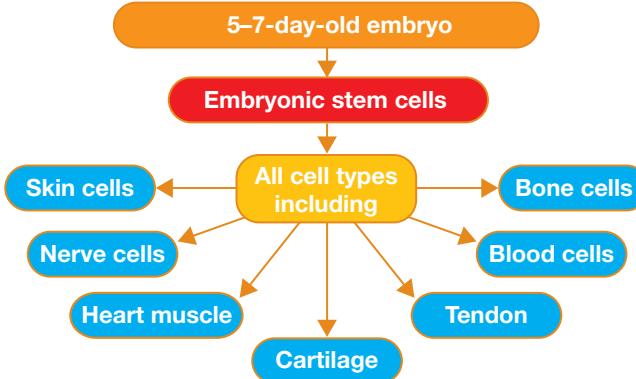


Figure 1.4.10

Embryonic stem cells can differentiate to become any of the types of cells found in the human body.

In a late stage embryo, the cells have **differentiated** (changed) and become fixed as skin cells, cardiac muscle cells or nerve cells in the brain (Figure 1.4.10). However, some cells in your body remain capable of dividing to make new cells so that you can heal wounds or replace worn-out cells.

Scientists experimenting with bone marrow for use in the treatment of leukaemia discovered **adult stem cells**. These cells allow you to regenerate and repair your tissues. For example, these are the cells that help your bones repair after being broken in an accident.

Adult stem cells lie deep within organs that need a constant supply of new cells, such as the skin. They are surrounded by millions of ordinary cells. Adult stem cells are specialised to some extent. This means that they are only able to make certain types of cells as shown in Figure 1.4.11.

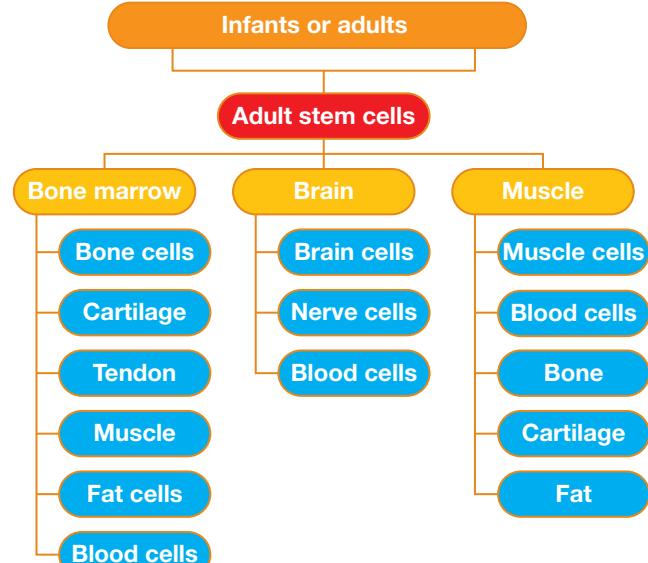


Figure 1.4.11

Adult stem cells can only differentiate to become certain types of cells.

Adult stem cells are found in the skin, the lining of the gut and the brain. There is a limit to the types of cells that they can differentiate into. For example, blood stem cells give rise to the different types of blood cells and stem cells in the skin regrow skin and hair.

Scientists believe that stem cells have potential to treat and possibly cure diseases such as cancer, diabetes and heart disease and spinal-cord injuries where cells have been damaged. However, adult stem cells are not always suitable and experimentation with embryonic stem cells is not accepted by many sectors of the community.

In 2006, Shinya Yamanaka of the University of Kyoto in Japan discovered a way of returning mature skin cells from a mouse to their pluripotent state. An outline of the process is shown in Figure 1.4.12. Yamanaka called these cells **induced pluripotent skin cells (iPSCs)**. Over the last few years, other scientists have successfully repeated Yamanaka's experiments.

More research is needed to make sure that these iPSCs behave in exactly the same way as embryonic stem cells. There have been times when the iPSCs have not functioned correctly.

Once these problems are solved, scientists believe that these cells have the potential to produce replacement parts of cells or organs damaged by disease. For example, they could replace nerve cells damaged through accidents (Figure 1.4.13), by Parkinson's disease and multiple sclerosis, or replace cardiac muscle damaged by heart attack. There would be no problems with rejection of the replacement tissue because it could be made from the patient's own cells.

Scientists have demonstrated that iPSCs can cure the genetic disease sickle cell anaemia in animals but there are many safety issues to deal with before there can be any human trials.



Figure 1.4.13

Many people who have suffered spinal injuries hope that stem cell research will find a way of repairing the damage.

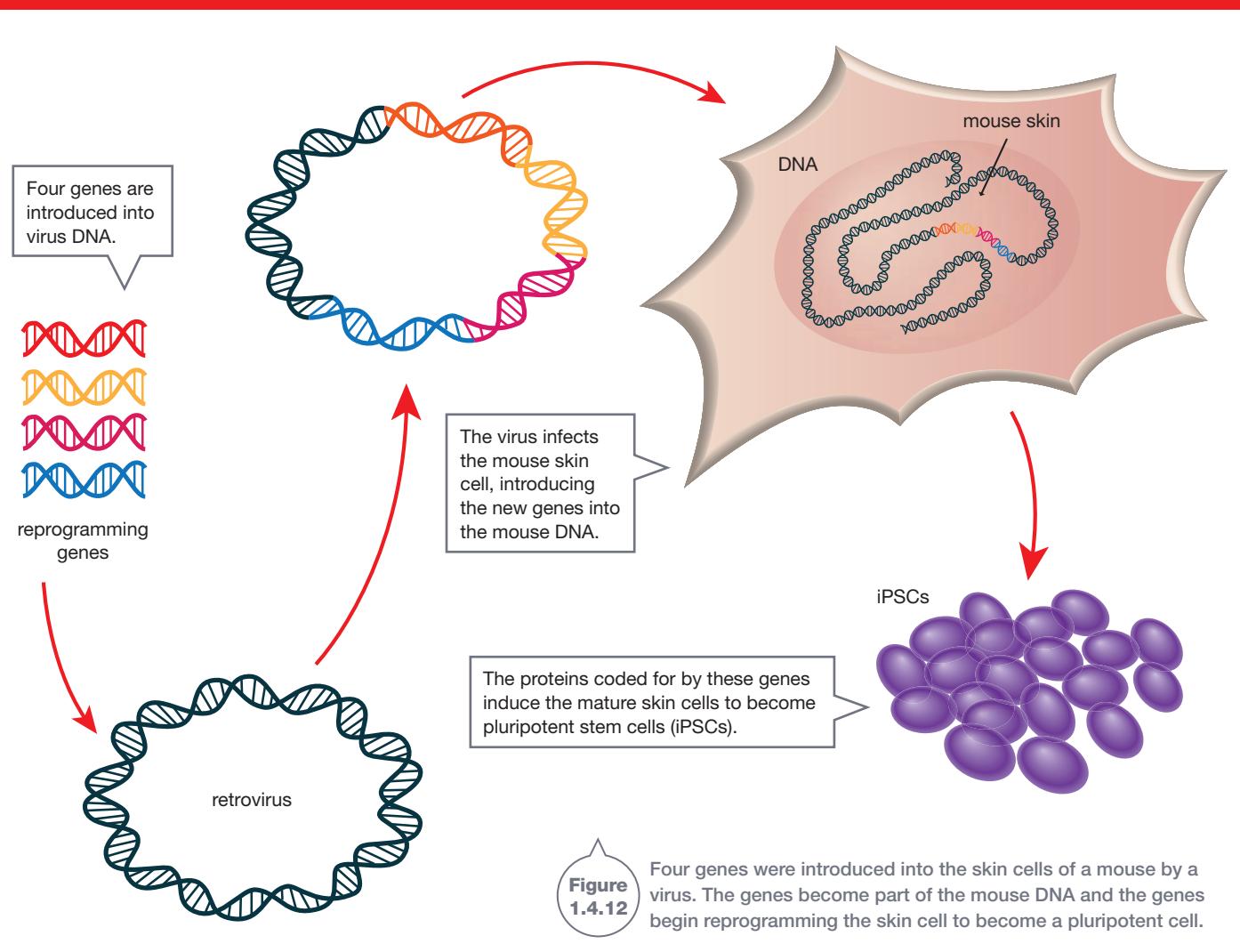


Figure 1.4.12

Four genes were introduced into the skin cells of a mouse by a virus. The genes become part of the mouse DNA and the genes begin reprogramming the skin cell to become a pluripotent cell.

1.4

Unit review

Remembering

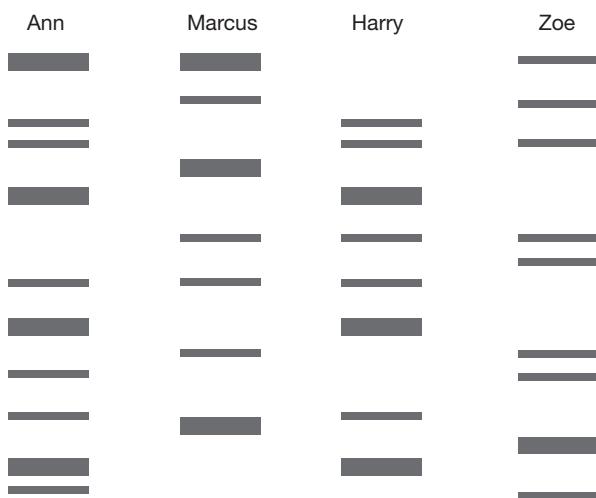
- 1 **Name** a genetically modified crop grown in Australia.
- 2 **Name** the circular DNA structure found in many bacteria and used in gene splicing.
- 3 **State** how long it took for the complete human genome to be mapped.
- 4 **List** three reasons why individuals would be given genetic tests.
- 5 **State** in simple terms the concept of gene therapy.

Understanding

- 6 **Define** the term *recombinant DNA*.
- 7 **Describe** the advantages that genetically modified plants have over other varieties of:
 - a rice
 - b canola.
- 8 a **Explain** what a single nucleotide polymorphism is.
b **Propose** ways that single nucleotide polymorphisms can occur.
c **Explain** why single nucleotide polymorphisms may be linked to human disease.
- 9 **Describe** four difficulties associated with successful gene therapy.

Applying

- 10 a The DNA profile of an individual can be shown as a series of bands. **Use** the DNA profiles in Figure 1.4.14 to **identify** the two people who are most closely related.
b **Explain** your choice.



Analysing

- 11 The human genome is mapped, but is the project finished?

Discuss

- 12 **Compare** the possible effect on an individual of knowing that they have the genes predisposing them (making it more likely) to type-2 diabetes and the dominant allele that causes Huntington's disease.



Inquiring

- 1 Investigate the arguments used against the introduction of genetically modified plants such as Golden rice-2.

- 2 Research and present a report on the advances made in genetic modification of animals for use in human organ transplants.

1.4

Practical activities

1 Genetic modification—public opinion

Purpose

To increase knowledge and understanding of issues surrounding GM technology and people's ideas on these issues.

Materials

- resources such as textbooks, encyclopedias and the internet
- questionnaire you have devised



Procedure

- In your group, discuss what you know about genetic modification of plants and animals and identify questions as a starting point for your research.
- Research genetic modification and through your research identify the issues that appear to be most controversial.

- Devise a questionnaire as a research tool for gathering information from other community groups on this topic. Your teacher can provide guidance on how to do this from Pearson Reader

Results

Prepare a report on the topic of genetic modification. In the report, present the arguments supporting both sides of any issue and the results of the questionnaire.

Discussion

- Analyse** your own opinions of genetic modification and write a short summary of them.
- Justify** your points of view.

2 Genetic technologies

Purpose

To discuss some genetic technologies and to enable you to develop your own opinion on the ethics of each.



Materials

- access to the internet and other reference materials

Procedure

- You are going to discuss four aspects of gene technology shown in the results table. You will be using a discussion strategy called Jigsaw. The class forms into four groups of approximately equal size. These groups are your Home groups. Each person in the Home group is given a number from 1 to 4. (There may have to be two students with the same number.) Your teacher will then assign a number to each of the technologies to be studied.
- Students with the same number move into a group together. With this group, you will study the technology with the same number. This is the Expert group.
- The job of the Expert group is to research and discuss the technology. Identify the positives and negatives of the technology. Record the number of people in the Expert group who were in favour of the technology.
- Report back to the Home group.

Results

Each person in the Home group records what is said by the Expert groups. The following table could be used to record comments.

Technology	Positives	Negatives	Decision: for/against
Genetic modification of food crops (plants)			
Gene therapy			
Gene testing			
Stem cell research			

Discussion

- a** **Identify** the technologies supported by the majority of the class.
b **Propose** reasons for this technology receiving the support.
- a** **Identify** the technology that had least support.
b **Propose** reasons for the lack of support.
- In a paragraph, **discuss** the technology that you are least willing to support.

Remembering

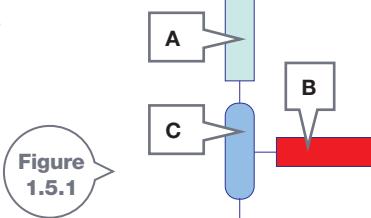
- 1 List the bases found in DNA.
- 2 Name the sugar found in DNA.
- 3 Name the following processes:
 - a adding genes into or removing genes from DNA
 - b replacing a defective gene with a normal gene
 - c selective breeding where offspring that show the most positive traits and fewest negative traits are selected and then crossed with one of the original parents.

Understanding

- 4 Explain why the 46 chromosomes in the human body are often described as 23 pairs.
- 5 Explain what makes one gene different from all other genes.
- 6 Predict the base sequence in the complementary strand of DNA that has the base sequence ATGTTCCAGCGAAATG.
- 7 Predict what would happen if gametes were produced by mitosis.
- 8 Explain why the research that created iPSCs is so exciting for scientists.

Applying

- 9 Identify the parts of the nucleotide labelled A, B and C in Figure 1.5.1.



- 10 Demonstrate how the number of cytosine molecules in a DNA molecule can be used to predict the number of guanine molecules.
- 11 Demonstrate how two homozygous parents could have a heterozygous child.
- 12 Identify the correct definition in column B for each of the terms listed in column A.

A	B
Genome	The chromosomes that are not sex chromosomes
Meiosis	Circle of DNA found in bacteria
Autosomes	The type of cell division that produces gametes with half the number of chromosomes of the parent cell
Plasmid	The genetic information carried by a haploid set of chromosomes

Analysing

- 13 Discuss the necessity of having two types of cell division—mitosis and meiosis.
- 14 Compare haploid and diploid cells.
- 15 Compare embryonic stem cells and adult stem cells.

Evaluating

- 16 Mules (male) and hinnies (female) are bred by crossing a horse and a donkey. Horses have a diploid number of 62 chromosomes and donkeys have 64 chromosomes. Propose why mules and hinnies do not produce gametes and are sterile.

Creating

- 17 a Use the information in Table 1.5.1 to construct graphs showing the change in amount of land planted with three GM crops.

Table 1.5.1 Area of land planted with GM crops

GM crop	Area of land planted (millions of hectares)						
	1997	1999	2001	2003	2005	2007	2009
Sweet corn	3	11	9	16	25	39	45
Canola	1	13	11.5	15.5	18.5	23.5	26.5
Soy	5	26	46	50	64	72	82

- b Use the information in Table 1.5.1 and the graphs you constructed to calculate the:
 - i crop that had the largest percentage increase in area of land planted in the years 1997–2009
 - ii years that showed the greatest increase in land planted with GM canola
 - iii years that showed the smallest increase in land planted with GM soy.
- c Identify the crop that has not experienced a decrease in the area of land planted.

- 18 Use the following ten key terms to construct a visual summary of the information presented in this chapter.

nucleotides
bases
chromosome
DNA
meiosis
mitosis
replication
plasmid
gene splicing
recombinant DNA

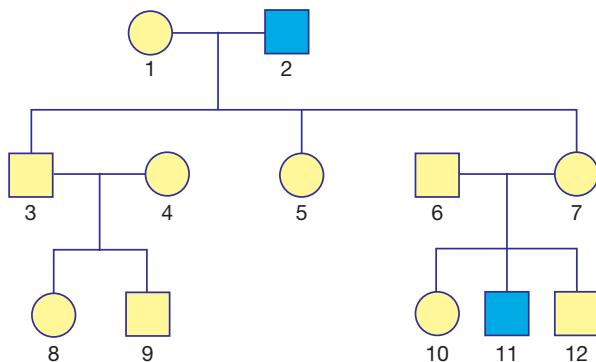


Thinking scientifically

Q1 In budgerigars, green feather colour (G) is dominant to blue feather colour (g). A blue budgerigar is mated with a heterozygous budgerigar. Identify the most probable genotypes of the offspring.

- A** All the offspring will be blue.
- B** All offspring will be green.
- C** $\frac{1}{2}$ Gg: $\frac{1}{2}$ gg
- D** $\frac{1}{2}$ GG: $\frac{1}{2}$ gg

Q2 The following diagram is called a pedigree. The individuals shaded blue have thalassemia, a disease that is inherited according to the rules of dominant-recessive inheritance. Based on the information in the family pedigree, identify the option in which both statements are true.



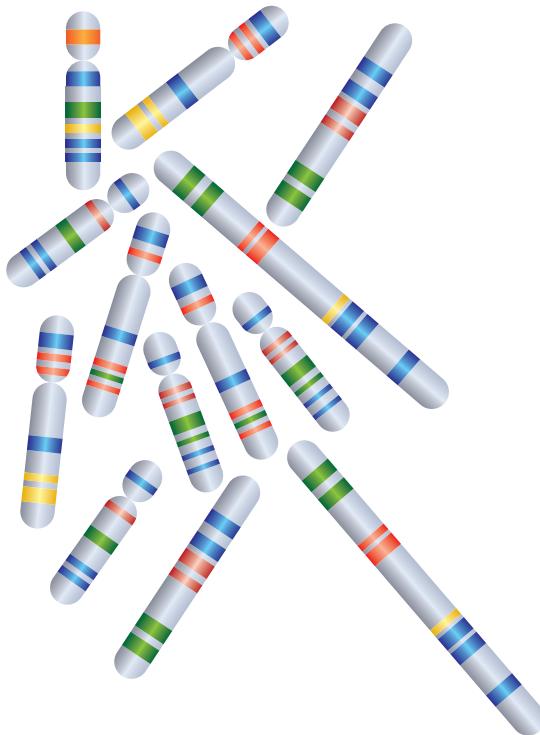
- A** 3 and 7 are both carriers of the disease. 8 or 9 must be a carrier.
- B** 6 and 7 are both heterozygous for thalassemia. 3 and 5 are carriers of thalassemia.
- C** 5 and 6 are heterozygous for the condition. 10 and 12 must be carriers of the disease.
- D** 2 is homozygous for thalassemia. 3 and 4 must be carriers of the disease.

Q3 In snapdragons, there are two alleles for the gene for flower colour—red and white. Red flower colour is incompletely dominant to white.

A snapdragon homozygous for the red allele is crossed with a snapdragon that is heterozygous for flower colour. Which one is the following is unlikely to be correct?

- A** Half the offspring would be red and half the offspring would be pink.
- B** Half the offspring would be red, one-quarter would be pink and one-quarter would be white.
- C** The heterozygous parent had pink flowers.
- D** The homozygous parent had red flowers.

Q4 Identify the number of pairs of homologous chromosomes in the following diagram.



- A** 7
- B** 6
- C** 5
- D** 4

Q5 Identify the small section of DNA that could be part of the longer DNA strand:

TAGTAGTCATACCGAATTGCCCGAATACTAGTAGGATC
ATCATCAGTATGGCTAACGGCCTATGATCATCCTAG

- A** TACCGAATCCCGGAATTC
ATGGCTTAGGGCCTTAAG
- B** TACCGAATTGCCCGGAATAC
ATGGCTTAACGGCCTTATG
- C** TACCGAATGCCCGGAATAC
ATGGCTTACGGCCTTATG
- D** TACCGATGCCGCAATAC
ATGGCTACGGCGTTATC

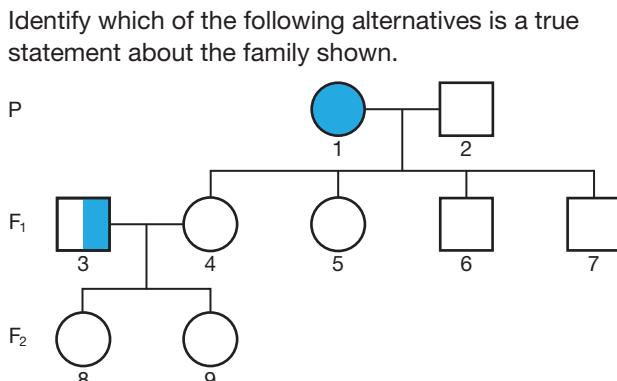
Thinking scientifically

- Q6** The data in the table provides information on the costs to farmers from four different states in India of growing genetically modified cotton.

State in India	Performance advantage of GM cotton over non-GM varieties (percentage)				
	Yield	Income	Cost of chemicals	Total cost	Profit
Maharastra	32	29	-44	15	56
Karnataka	73	67	-49	19	172
Tamil Nadu	43	44	-73	5	229
Andhra Pradesh	-3	-3	-19	13	-40
National average	34	33	-41	17	69

Analyse the data and decide which of the following statements is true.

- A** The state that made the greatest savings on chemicals also had the highest yield and the greatest profit.
 - B** The states of Maharastra and Kamataka both saved more than the national average on chemical costs and had a yield and profit above the national average.
 - C** The state that had the greatest advantage in terms of total income also had the greatest advantage in terms of total cost and yield.
 - D** Andhra Pradesh made a loss because the farmers in that state had to spend more on chemicals.
- Q7** Hair curliness is an example of incomplete dominance. The diagram below illustrates a pedigree showing three generations of a family. The mother is homozygous for curly hair. The father is homozygous for straight hair. Individual 3 marries into the family and his genotype is shown to be heterozygous. The remaining phenotypes of the F₁ and F₂ generations are not shown.



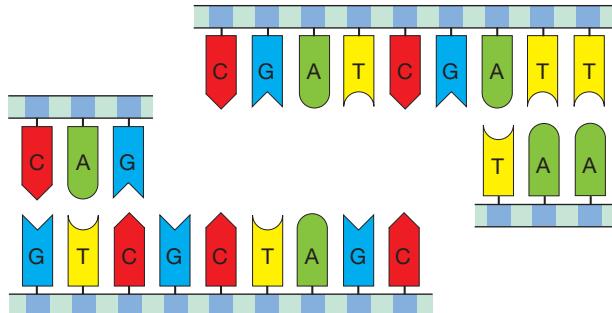
- A** Some of the F₁ generation will have curly hair and some will have straight hair.

- B** It is not possible for individual 9 to have straight hair.

- C** All of the F₁ generation will be different from their parents and have wavy hair.

- D** Individual 8 must have wavy hair.

- Q8** Scientists involved with research into genetic modification cut a piece of DNA using a particular restriction enzyme. The ends of DNA exposed are shown below.



From the four genes below, identify the gene that could be spliced into that piece of DNA.

- A** Gene A: C, G, A, T, C, G. Gene: C, G, T, A, C, G.
- B** Gene B: C, G, A, T, G, C. Gene: C, G, T, A, G, C.
- C** Gene C: C, G, A, T, C, G. Gene: G, C, T, A, G, C.
- D** Gene D: C, G, T, A, C, G. Gene: G, C, A, T, G, C.

Unit 1.1

Chromosomes: thread-like structures in the nucleus.

Composed of DNA and proteins; contains the genetic information in the form of genes

Complementary base pairs: a pair of bases that can join to make the rungs of the DNA ladder—adenine and thymine, guanine and cytosine

Deoxyribonucleic acid (DNA): a nucleic acid with deoxyribose sugar and phosphate as the backbone; the molecule that determines the genetic characteristics of most living things

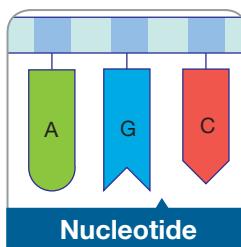
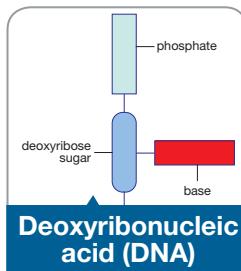
Deoxyribose sugar: one of the parts that make up a nucleotide

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

Nitrogen-rich base: part of a nucleotide; the four types are adenine (A), guanine (G), cytosine (C) and thymine (T)

Nucleotides: the building blocks of DNA

Phosphate group: one of the parts that make up a nucleotide



Unit 1.2

Autosomes: all the chromosomes in a cell other than the sex chromosomes

Centromere: the point on a chromosome where the two chromatids are joined together

Chromatid: one of the strands of a chromosome following replication

Diploid number: the number of chromosomes in body cells; two sets or $2N$

Haploid number: the number of chromosomes in gametes; one set or N

Homologous chromosomes: chromosomes with genes for particular characteristics at the same location

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

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

Replication: the process of making copies of DNA

Sex chromosomes: the chromosomes that determine the sex of an individual; in humans they are the X and Y chromosomes



Unit 1.3

Alleles: different forms of the same gene

Dominant: the characteristic that is expressed in the homozygous condition

Genotype: genetic information carried by an individual

Heterozygous: having two different alleles on homologous chromosomes

Homozygous: having two identical alleles on homologous chromosomes

Incomplete dominance: where the appearance of a heterozygous individual results from a 'blending' of the two alleles because one allele is not completely dominant over the other

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

Phenotype: observable characteristics of the individual; the way the genotype is expressed

Pure breeding: where all individuals have the same genetic information for a characteristic generation after generation

Recessive: the characteristic that remains hidden in the homozygous condition

Sex-linked genes: genes present on the sex chromosomes

Unit 1.4

Adult stem cells: cells that can make certain types of body cells

Differentiate: become different from others

Embryonic stem cells: cells found in the embryo that are capable of becoming any cell type found in the body of a complex organism

Gene splicing: the process used to add a gene into or remove genes from DNA

Gene therapy: the process of replacing a defective gene with a normal gene

Genetically modified: having the genes changed

Genome: the genetic information carried by a haploid set of chromosomes

Human Genome Project: an international project that aims to identify all the human genes and determine the sequence of the base pairs that make up human chromosomes

Induced pluripotent skin cells (iPSCs): mature cells that have been induced to revert to their pluripotent (capable of becoming any type of human cell) state

Plasmid: ring of DNA found in bacteria

Pluripotent: capable of becoming any one of the 220 different cell types found in the human body

Recombinant DNA technology: technology that allows DNA to be recombined with other genes

Single nucleotide polymorphisms (SNPs): differences of only one base between one human and another

