

Getting into genes 8

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

Key outcomes

Describe the genetic basis of inheritance.

Relate concepts of adaptation, biodiversity and evolution to the survival of species.

Explain how the action of micro-organisms can be both beneficial and detrimental to society.

Explain the role of DNA and genes in cell division and genetic inheritance.

Think about these

What is odd about cold sores?

Which has more chromosomes in its cells, a shrimp or a chimp?

What have ladders got to do with life?

Why don't hairs grow on stomach linings?

What has being paternal or maternal to do with cells?

What have monks, peas, mathematics and genes got in common?

Do you have Darwin's point?

What have Xs and Ys got to do with sex?

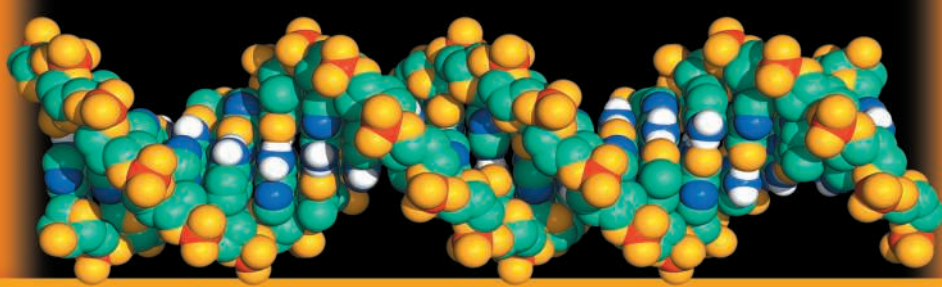
How do heat and cold affect a crocodile's sex?

How may the mystery of who Tutankhamen's dad was be solved?

What is the chance of suffering from cystic fibrosis?

Designer babies — should or shouldn't we?

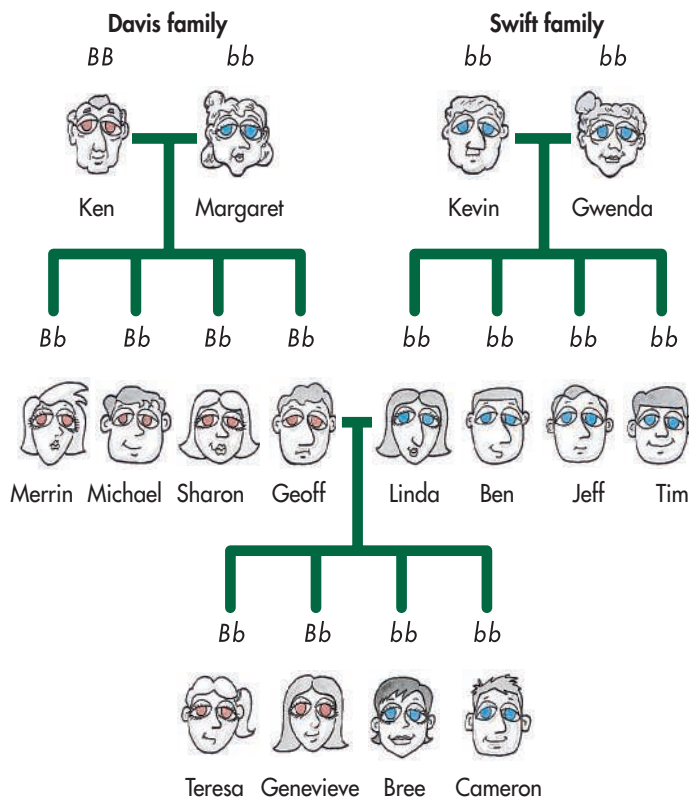
Life without males — is it possible?



YourQuest

Gene eyes?

THINK, SHARE AND DISCUSS



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

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

Just genes?

THINK, SHARE AND DISCUSS



Just genes — or do other factors contribute to what you look like?

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

Gene views

THINK AND DISCUSS

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

Cell stuff

The amazing thing about living things is that while they can be so different, they are all made of the same basic units — cells!

The same but different

Advances in microscopes and in staining techniques have enabled scientists to discover much about cells and the ‘stuff’ inside them. These discoveries led to the development of the **cell theory**:

- All living things are composed of cells.
- All cells are produced from pre-existing cells.

Cells are different in size, shape and in their **organelles**, but they have similar basic structures. Organelles are very small structures in the cytoplasm, each surrounded by a membrane and having a particular function. The organelles work together to maintain the **metabolic** or life processes of the cell.

All of the different parts of the cell have specific jobs to do (see

the table on the opposite page) so that the cell works properly and the organism is healthy. If a cell part does not do its job properly, cell function may become abnormal and the result is often disease.

Animals, plants, fungi and protoctists (e.g. protists and algae) are composed of **eukaryotic cells**. These cells possess a membrane-bound nucleus and other kinds of organelles. **Prokaryotic cells**, such as those of cyanobacteria (blue-green algae) and bacteria, do not have a nucleus or other organelles held in membranes. In these cells, the nucleic material is coiled up in part of the cell and the metabolic processes take place throughout the cytoplasm.

Living to die

Some cells change during their **life spans**. Where these cells are located in an organism in their early stages of development affects what kind of cells they become. These cells become **differentiated** as they mature, enabling



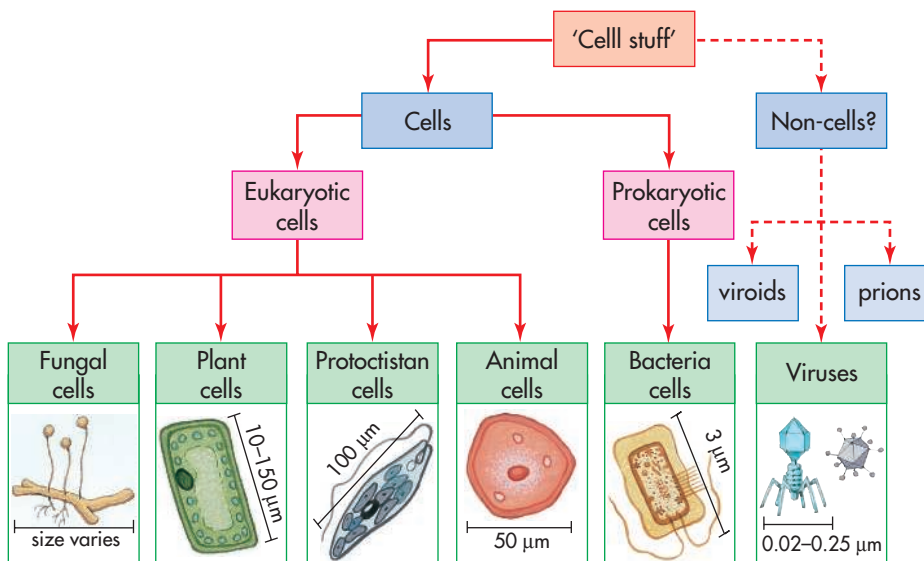
Electron micrograph of mitochondria

them to carry out a particular function. Some cells, such as those that make bone in mammals, or those that carry water in the stems and roots of plants, even die during this process.

The odd ones out

Are **viruses** living or not? Even scientists cannot agree. Viruses do not have organelles or carry out processes characteristic of living things. They penetrate the cells of organisms and inject their core of nucleic material into them. The host cells are made to produce more virus particles. The cell membrane ruptures as the particles are released. This is what happens if you suffer from a cold sore. The viruses lie dormant in the host cells until stress, fever or the ultraviolet radiation from the sun activates them. Outside the host, the virus exists only as an inactive particle rather like a crystal.

Viroids and **prions** are infectious agents that are even simpler than viruses. Viroids are tiny molecules of nucleic acid (RNA) that infect plants. Prions lack detectable amounts of nucleic acid. They are thought to be made up of protein and are hundreds of times smaller than a virus. ‘Mad cow’ disease and Creutzfeldt Jakob disease appear to be caused by prions.



The basic building units of living things — cells!

The 'stuff' in cells

Part	Job or function	Feature/location
cell membrane	controls movement of substances into and out of the cell	forms the boundary layer surrounding the cell
cytoplasm	thick fluid carrying organelles and materials in solution	contents of the cell
nucleus*	control centre of the cell; contains coding instructions for production of different enzymes which control metabolic processes; controls cell division and growth	large, dense, spherical structure in cytoplasm
nucleolus*	synthesis of nucleic acid involved in cell division	within the nucleus
mitochondria (singular: mitochondrion)	mini 'powerhouses' where aerobic respiration supplies energy for cell activities	organelles in cytoplasm
Golgi bodies	wrap up and package substances for transport out of cells	layers of membranes in the cytoplasm
endoplasmic reticulum (ER)	transports substances within the cell	network of channels in the cytoplasm
ribosomes	manufacture proteins including enzymes and hormones	attached to the ER or free in the cytoplasm
lysosomes	'digest' cell debris for reuse; contribute to cell self-destruction	sacs in cytoplasm containing enzymes
cell wall	supports, strengthens and protects cells; varies in composition between plants, bacteria and fungi	forms a layer outside cell membranes of prokaryotic, fungal plant cells
vacuoles*	large 'bubbles' of cell sap (stored dissolved sugars and salts)	common in cytoplasm of plant cells (may occupy nearly all the space in the cell)
centriole	involved in cell division	paired rod-like structures in cytoplasm of animal cells
chloroplasts*	contain chlorophyll which absorbs light energy	organelles in cytoplasm of photosynthetic cells
plastids	contain substances such as chlorophyll and starch	present in cytoplasm of plant cells

*visible using the light microscope

sciFacts

Is a giant virus part of your ancestry?

The discovery of a giant virus has raised a number of questions, not only about our current classification of viruses, but also about the origin and evolution of life on our planet. Some scientists believe that the Mimivirus breaks the boundary between viruses and cellular life forms. Measuring about 500 nm in diameter, it is more like the size of a bacterium. While most viruses have less than 10 genes, this giant virus contains over 900 genes in its double-stranded DNA, with about half of these genes being new to science. While the Mimivirus is getting some scientists to rethink what a virus is, other scientists are suggesting the need to rethink our current classification system and create a new grouping. There are even some scientists who suggest that ancestors of these giant 20-sided viruses may have become the nuclei in eukaryotic cells. If this theory is supported, what impact will it have on our future research and technologies?

Source: *New Scientist* 25 March 2006, pages 37–41

activities

ANALYSE AND EVALUATE

Use the data in the table above to answer the following questions.

- 1 Which organelle controls the activities of the cell? Explain briefly how it does this.
- 2 Enzymes control the rate of chemical processes throughout the cell. Where in the cell are these enzymes made and how do they reach the parts of the cell in which they are needed?

REMEMBER

- 3 State the two parts of the cell theory.
- 4 What is an organelle?
- 5 What is the main difference between a prokaryotic and a eukaryotic cell?

- 6 Why do some cells change during their life span?

- 7 Draw a flowchart to outline how viruses reproduce.

THINK AND DISCUSS

- 8 Construct a mind, concept or cluster map to summarise the key points in 'Cell stuff'.
- 9 When examined under the electron microscope, a muscle cell is seen to have many mitochondria. How do you account for this?
- 10 Give arguments for and against a virus being a living thing.

CREATE

- 11 Make models of a prokaryotic cell and a eukaryotic cell.
- 12 Create a rhyme or song about the 'stuff of cells'.

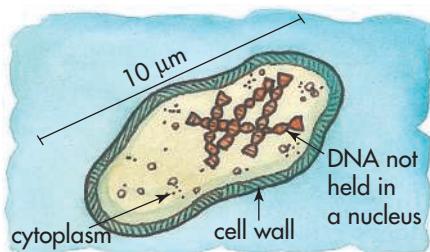
Nuclear matter

The nucleus is a tiny organelle in the cytoplasm of eukaryotic cells which contains the blueprint of life and determines how cells will develop. All eukaryotic cells have a nucleus — at least at the start of their lives. In some cells, such as red blood cells, the nucleus deteriorates and is broken down.

Carrying the message

The nucleus is usually difficult to see and needs to be stained to become visible under a light microscope. Only by using an electron microscope do the details of the cell and the nucleus become clear. The nucleus is separated from the rest of the cell by the nuclear membrane, which has many tiny pores or openings that connect at various places with the endoplasmic reticulum. The pores allow the transportation of materials into and out of the nucleus.

The nucleus contains chemical compounds called **nucleic acids**. These are deoxyribonucleic acid (**DNA**), which is able to 'store' coded instructions, and ribonucleic acid (**RNA**), which allows the DNA to read the coded instructions. There is about one metre of DNA in each cell. Imagine how long the stretch of DNA would be in the millions of cells in your body!



In prokaryotes, the DNA is more loosely located in an area of the cell and is not contained in a membrane.

The DNA molecule looks like a double helix. Its structure was discovered by Watson and Crick in 1953. It carries the code for making all the parts of all the different kinds of cells. The code also programs the way cells behave and how they carry out their functions, such as transmitting nerve impulses, supplying energy, contracting or carrying out photosynthesis. As it contains DNA, the nucleus is the control centre of the cell.

Captivating chromosomes

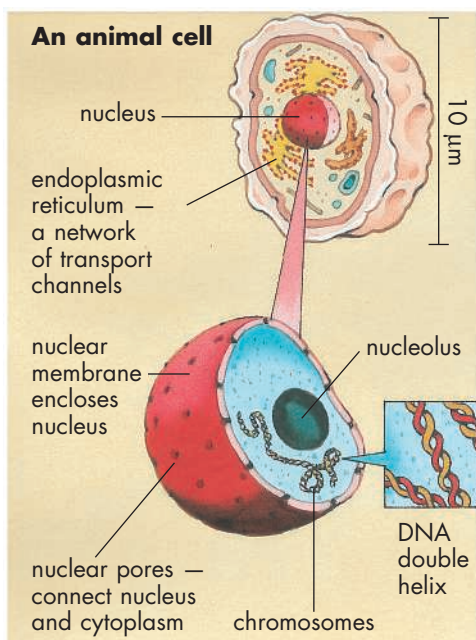
When the cell is about to divide, the lengths of DNA shorten and coil to form **chromosomes**. With suitable staining, the chromosomes become visible. At other times in the life cycle of the cell, the chromosomes are not visible as the coils are unwound and



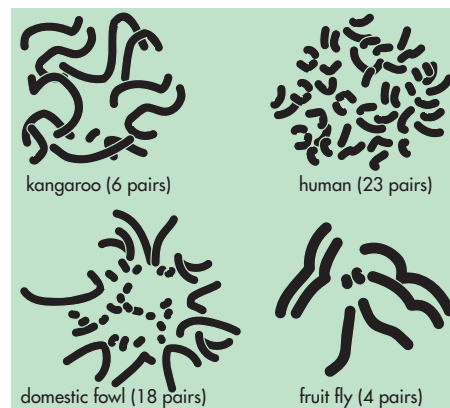
Scanning electron micrograph showing double-stranded chromosomes

the DNA is spread throughout the nucleus. Each chromosome appears to be made of two strands, or **chromatids**, held together by a **centromere**. During cell division, the chromosomes divide into single strands. One chromatid from each pair becomes part of the new cell formed by the cell division. Later, when the DNA molecule replicates itself, the chromatids become double stranded again. Exact replication of the DNA ensures that instructions (characteristics) are passed on unchanged.

Every kind of living thing has its own particular number of chromosomes in each body cell, normally occurring in matched pairs. A human cell, whether it is a white blood cell, or a cell from the cheek lining or the kidney, will have 23 pairs or 46 chromosomes in total. One of the pairs, known as the **sex chromosomes** (which contain information for sexual characteristics), does not match in males. There is a longer X chromosome and a shorter Y chromosome. In females, there are two X chromosomes. The non-sex chromosomes are called **autosomes**.



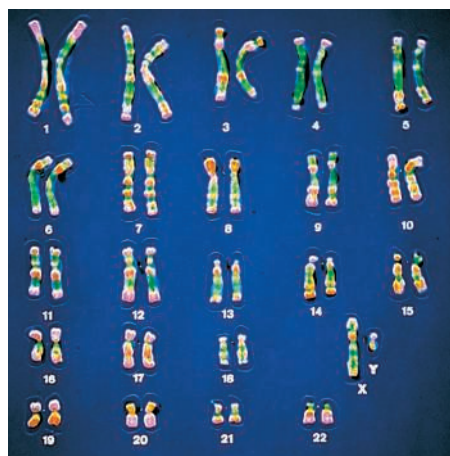
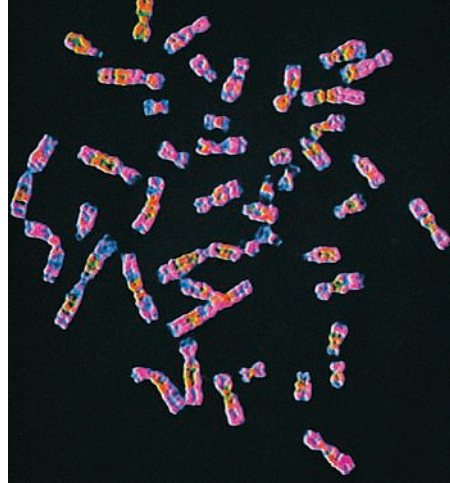
Knowing about the nucleus



Chromosomes of different kinds of living things. Can you match the pairs?

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

Species of living thing	Number of chromosomes in each body cell	Species of living thing	Number of chromosomes in each body cell
chimpanzee	48	tomato	24
Euglena (unicellular organism)	90	cabbage	18
fruit fly	8	frog	26
human	46	housefly	12
koala	16	pig	40
onion	16	platypus	52
shrimp	254	rice	24
sugarcane	80	sheep	54



Chromosomes (top) and karyotyped chromosomes (bottom). Can you tell if the karyotyped chromosomes are from a male or female?

Your kind of karyotype?

Chromosome pairs differ from each other in various ways, such as in size, shape and banding. This helps them to be distinguished from each other. **Karyotyping** chromosomes is the process of sorting chromosomes into their matched pairs. Cells about to divide are treated and stained, mounted on slides for viewing and photographed. The photographs are cut up and rearranged. Karyotyping is an important technique used in investigating chromosomal disorders.

activities

REMEMBER

- 1 Why is the nucleus often referred to as the control centre of a cell?
- 2 Where is the nucleic material located in prokaryotic cells such as bacteria?
- 3 Suggest a function for the many tiny pores in the nuclear membrane.
- 4 Why are cells stained?
- 5 What is a chromosome?
- 6 Are chromosomes always visible in a cell? Explain.
- 7 How many pairs of chromosomes does each body cell of a human contain?
- 8 What is the difference between sex chromosomes and autosomes?
- 9 What is the process of karyotyping and why is it important?

ANALYSE AND EVALUATE

- 10 Convert the information given in the table above into a bar graph showing the number of pairs of chromosomes in eight different organisms.

- 11 Which species' body cells have:
 - (a) the highest total number of chromosomes?
 - (b) the smallest total number of chromosomes?

- 12 Do you think that the number of chromosomes reflects the intelligence of an organism? Explain your answer.

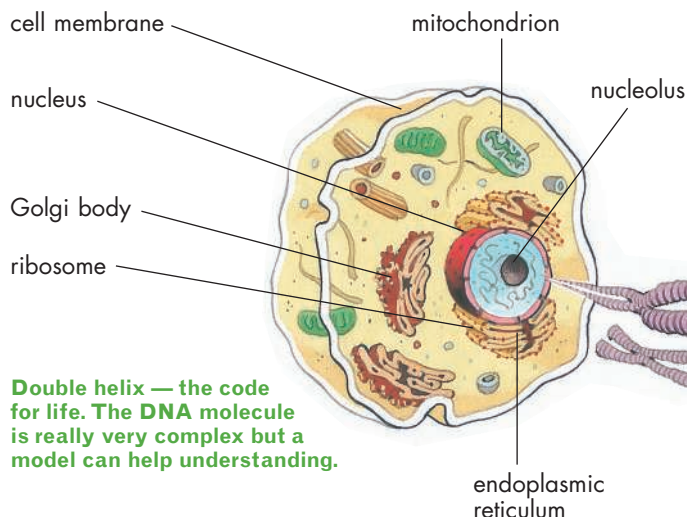
THINK AND DISCUSS

- 13 If each cell nucleus has about one metre of DNA, how does the DNA fit in?
- 14 Why can karyotyping be carried out only on cells about to divide?
- 15 How can karyotyping determine whether the cell is from a male or a female person?

CREATE

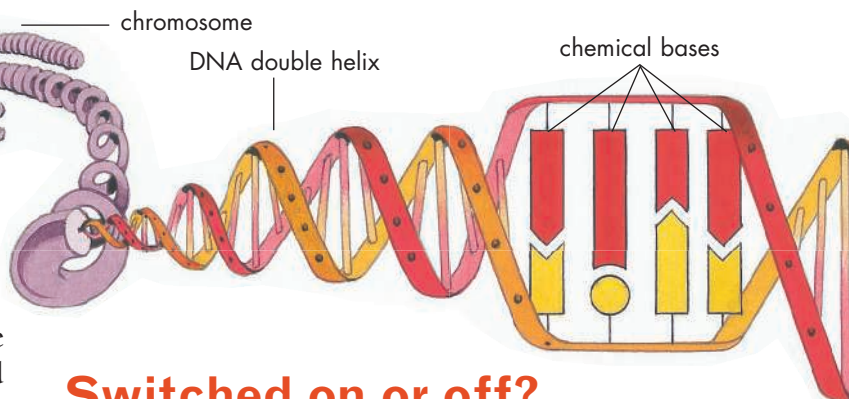
- 16 Using a variety of materials, including pipe cleaners of different colours and lengths, make a model of a nucleus with a set of four pairs of chromosomes.
 - (a) How many chromosomes are there altogether?
 - (b) Are they all alike?
 - (c) What structure will each chromosome have?
 - (d) Devise a 'container' representing the cell for your nucleus.

Unravelling the code



Locating genes

Segments of the DNA molecule contain particular sets of instructions called **genes**, which are often separated by 'junk' DNA, the function of which is not yet known. The position occupied by the gene on the chromosome is called the **locus**.



How is it that the DNA tucked inside the nucleus of a cell carries the coded instructions for the way that cells develop?

How living things appear and function is known as their **phenotype**. This is caused by the combined effect of their coding (or **genotype**) and their environment. For example, a person's cells may carry coded instructions to have freckles but without exposure to the sun, the freckles would not appear.

Key codes

Most DNA is found in the chromosomes of the nucleus and the molecule looks like a double helix or spiral ladder.

Just as the letters of the alphabet are the basis for written language, the basic units making up the DNA of chromosomes are arranged to form a genetic language or code. These building blocks are called **nucleotides**. Nucleotides are made up of three parts: a sugar part, a phosphate part and a nitrogenous base. While the sugar and phosphate found in all nucleotides are the same, the nitrogenous base in each nucleotide varies. The four different types of bases found in nucleotides of DNA are: adenine (A), thymine (T), cytosine (C) and guanine (G).

Unless something odd happens, the nitrogenous bases always pair according to the **base-pairing rule**: A is chemically attracted to T and C to G. These form the 'rungs' of the ladder of DNA.

A sequence of three nucleotides is called a **triplet**, which provides the coding for a particular amino acid.

Switched on or off?

Different genes are responsible for different characteristics, such as the colour of flower petals, the markings on a snail shell, or a person's blood group or eye colour. Every body cell of an organism has the same set of genes called a **genome**, but not all genes are active. Some have to be 'switched on' to act and some have to be 'switched off' at different stages in the life of a cell. That is why hairs do not grow on stomach linings and cheek cells do not produce acid!

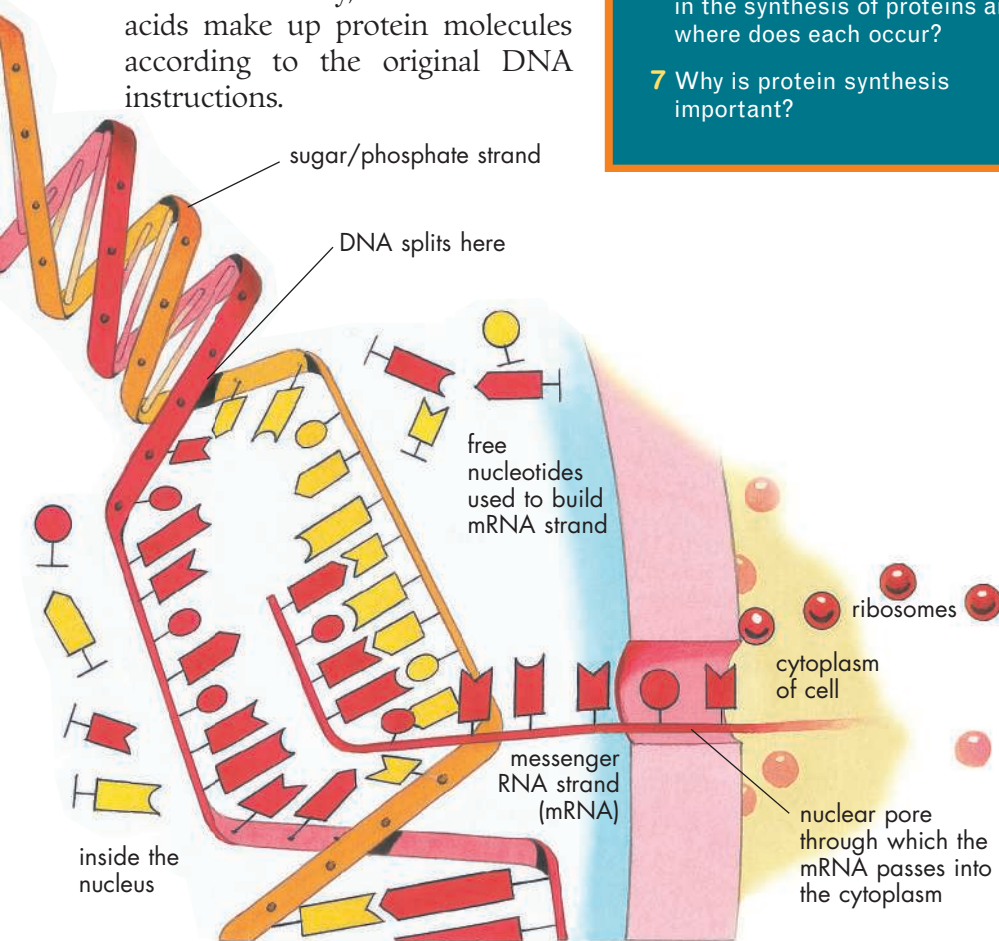
Protein synthesis

Inside the nucleus are instructions for every part of the organism. These instructions are like a genetic master plan. Before an instruction can be carried out, that particular part of the genetic master plan must be copied so that the master plan remains inside the nucleus.

These instructions code for the production of proteins, which are essential to the cell. Most genes code for this. To produce one of these proteins, certain enzymes 'unzip' the segment of DNA coding for that particular protein, exposing the two sides of the 'ladder'. The nucleotides on each side attract other free nucleotides in the nucleus to make a copy of the DNA sequence for the production of that protein. This process is called **transcription**. The copy of the DNA sequence is called **messenger RNA** (mRNA) because it passes through the pores

of the nuclear membrane into the cytoplasm to the ribosomes, where proteins are manufactured. The code or message for the production of the protein is carried with messenger RNA. The RNA strand is not quite the same as DNA because the base thymine is replaced by the base uracil (U).

At the ribosomes, the message is **translated**. Amino acids are collected from the cytoplasm and linked in sequence, like beads on a necklace, according to the triplet code carried by messenger RNA. In this way, chains of amino acids make up protein molecules according to the original DNA instructions.



Precious proteins

Why are proteins so important? Proteins form parts of cells. Special proteins of heart muscle and arteries contract, enabling blood containing haemoglobin and hormones to be pumped through the body. Haemoglobin is a protein that carries oxygen necessary for respiration. Many hormones are proteins. Hormones, such as insulin or adrenaline, influence our cells' activities. Proteins also make up enzymes, which control metabolic activities such as chemical digestion and respiration. Hormones also influence plant growth, and photosynthesis depends on enzymes.

activities

REMEMBER

- 1 What is the difference between the terms 'genotype' and 'phenotype'?
- 2 What is the base-pairing rule? Illustrate your answer by means of a simple labelled diagram.
- 3 What is the difference between DNA and RNA?
- 4 What are genes and what do they code for?
- 5 What does 'genome' mean?
- 6 What are the two main stages in the synthesis of proteins and where does each occur?
- 7 Why is protein synthesis important?

THINK AND DISCUSS

- 8 All cells of a particular living thing, such as a spider, have the same sets of genetic instructions but not all the cells have the same structure and function. What causes this? Why is cell specialisation important?
- 9 If the bases making up a strand of DNA are in the following sequence, what would be the corresponding sequence for the messenger RNA?

DNA:

ATT GTC CCA TGA CAG TGG

mRNA:

CREATE

- 10 Design and make a model showing a simplified structure of DNA. Decide if you wish to make a 3-D or a 2-D (flat) representation.
 - (a) What kinds of materials could you use in your construction?
 - (b) Evaluate your model. What does it show or do well? What is it not able to show or do well?
- 11 Devise a role-play that demonstrates the way proteins are formed.

INVESTIGATE

- 12 Draw a timeline to show the rate of identification of human genes. A computer database called OMIM (On-line Mendelian Inheritance in Man) keeps a regular update. You can access OMIM by going to the website for this book and clicking on the OMIM link (see 'Weblinks', page 305).
- 13 Research two scientists who contributed to the discovery of the double helix model of DNA. Write a brief account of their work.
- 14 Investigate further discoveries that have been made about DNA. Construct a timeline to share the 'who', 'what' and 'when' of your findings.

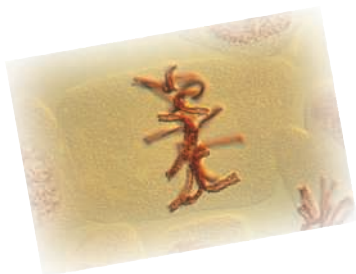
Passing on instructions

You were once a single cell — and now look at you! How did you get to be so big?



From one to many

What happens when skin wears away or damaged tissues need repairing? How do seedlings grow into gigantic trees? Throughout the life of multicellular organisms, cell division takes place for the growth, development, repair and replacement of tissues.



Mitosis in bluebell cells. What is the sequence of stages shown?

Reviewing reproduction

Cell division also has an important role to play in reproduction. In asexual reproduction, an organism grows a new living thing identical to itself as a result of **mitosis**, where the DNA in a cell replicates and two cells identical to the original cell are formed. The new cells are called daughter cells.

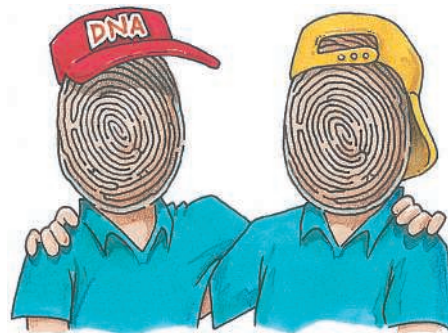
In sexually reproducing organisms, such as flowering plants and humans, **gametes** are produced in the sex organs. During fertilisation, the male gamete (sperm) fuses with the female gamete (ovum) to form the new individual or **zygote**. Because genetic instructions have come from two parents, the zygote will not be identical to either parent. By repeated mitosis, the zygote grows into an embryo and develops to become the new individual.

Each kind of cell division involves the chromosomes. What happens to the chromosomes in cell division determines how instructions are passed on from one generation to the next.



sciFacts

Although some parts of your DNA sequence may be similar to those of other people, no-one has exactly the same DNA — unless you have an identical twin! Each person has his or her own particular DNA profile, a fact used in forensic science to eliminate or identify suspects who have left some kind of tissue or body fluid at the scene of a crime.



Mitosis

New body cells resulting from cell division have to have the same number of chromosomes as their parent cell if they are to possess all the necessary coding instructions. The process of mitosis (see the diagram on the opposite page) ensures this.

The number of chromosomes in each body cell is called the **diploid** number. In normal body cells, chromosomes occur in pairs. The diploid number of these cells is represented as $2n$. Each species contains a fixed number of chromosomes. A human skin cell, for example, has 23 pairs. Its diploid number is therefore:

$$\begin{aligned} 2n &= 2 \times 23 \\ &= 46. \end{aligned}$$

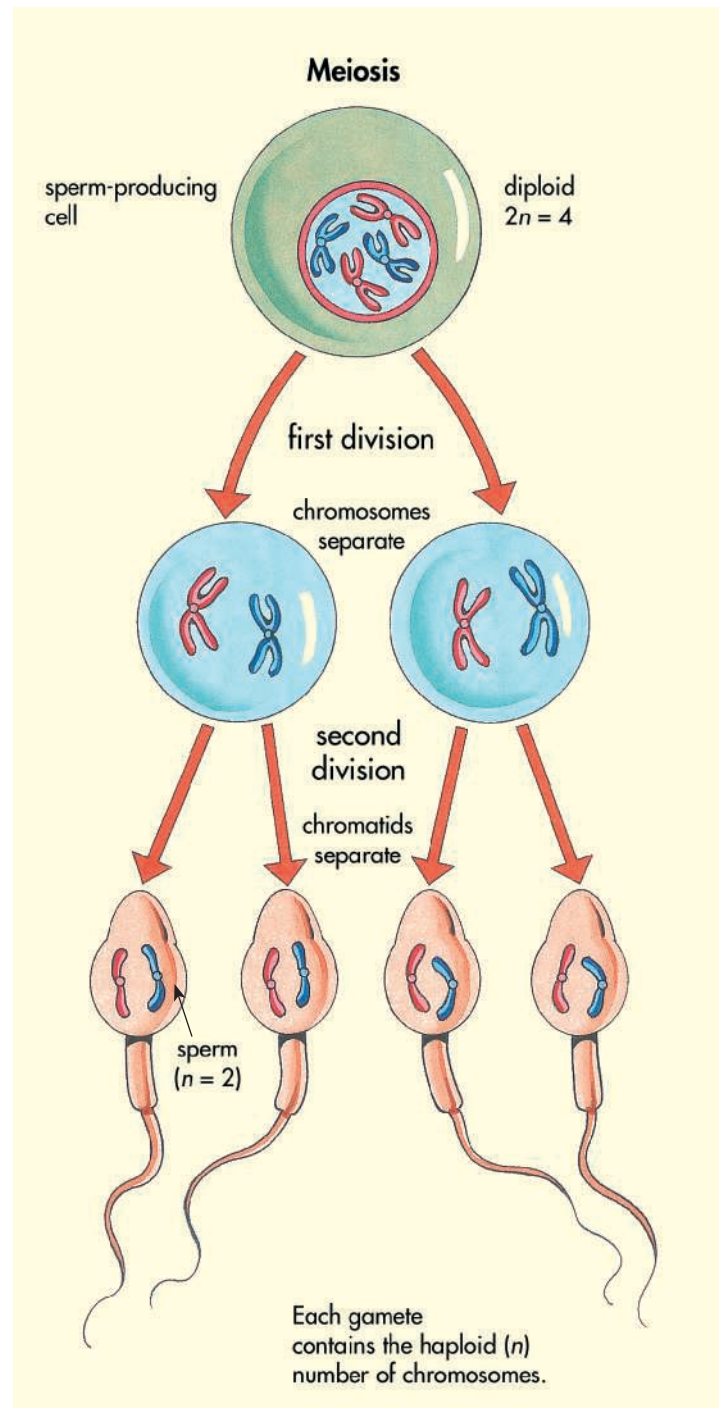
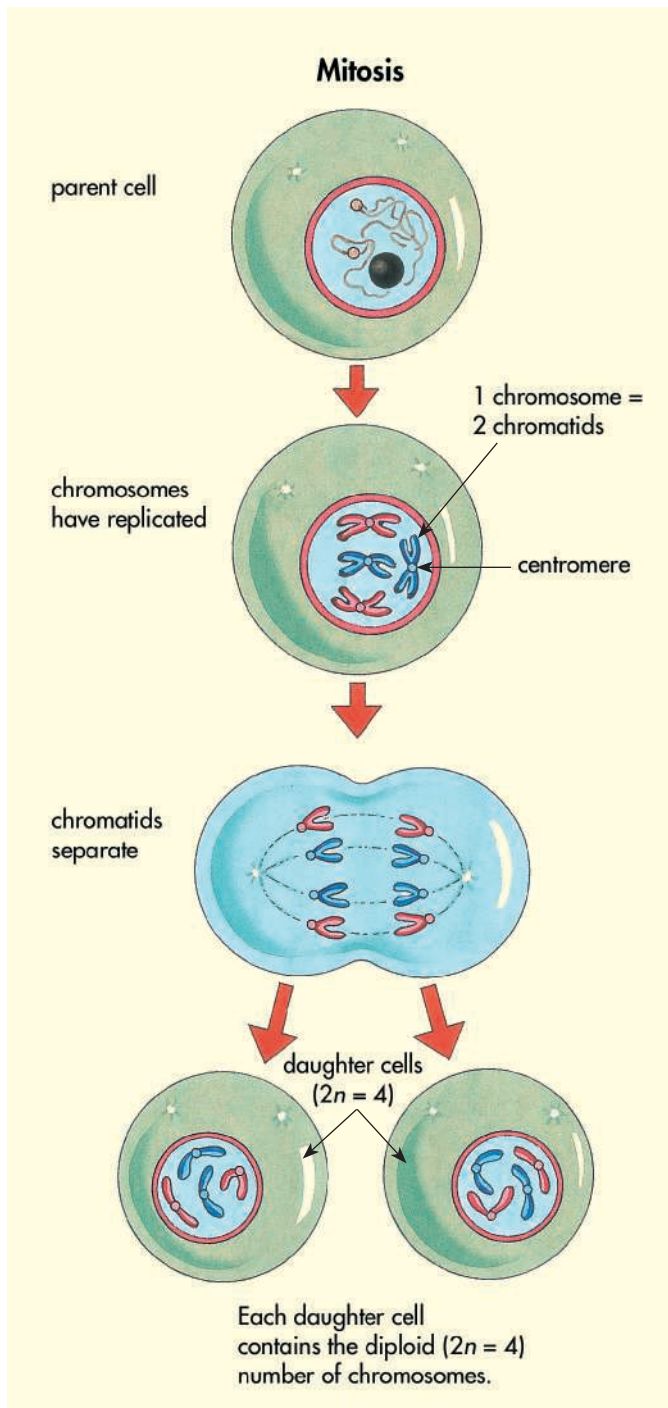


Meiosis

The chromosomes are not in pairs in gametes. If gametes were produced in the same way as other cells, what would happen to the chromosome number of the new cell at fertilisation? **Meiosis** is the kind of nuclear division that prevents the doubling of chromosomes at fertilisation. If each gamete had 46 chromosomes, a cell consisting of 92 chromosomes would be produced by the egg nucleus and sperm nucleus during fertilisation. In

meiosis, four gametes, each carrying half the number of chromosomes in body cells, called the **haploid** number (n), are produced. (See the diagram below.)

Chromosomes carried in the sperm from the father are called **paternal**. The chromosomes in the ovum from the mother are called **maternal**. As a result of fertilisation, the zygote is diploid. It then divides rapidly by mitosis to form the embryo, each cell carrying a full set of chromosomes.



The processes of mitosis and meiosis compared. The parent cell has four chromosomes ($2n = 4$).

Mitosis

process used in growth, repair and replacement of tissues

daughter cells contain diploid number of chromosomes ($2n$)

examples of mitosis: cuttings, runners, bulbs

occurs in the nucleus of body cells, e.g. skin, bone marrow, buds, capsules of moss plants



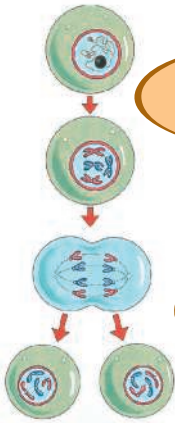
one cell division completes the process

two cells, which are identical to each other, are formed

no variation in new cells of offspring (unless environment influences); cells are genetically identical, i.e. clones of each other



used in tissue culture for skin grafts, cloning plants, i.e. narrowing variation and diversity



Meiosis

process used in producing sex cells (gametes)

daughter cells (gametes) contain the haploid number of chromosomes (n)

occurs in the gonads (reproductive organs) of living things, e.g. ovaries and testes of animals, ovaries and anthers of flowering plants



occurs in sexually reproducing organisms that reproduce by fusion of gametes, e.g. vertebrates, flowering plants

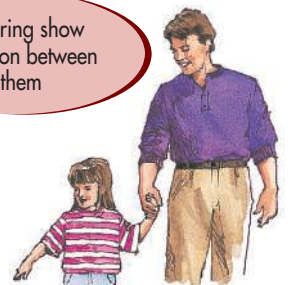
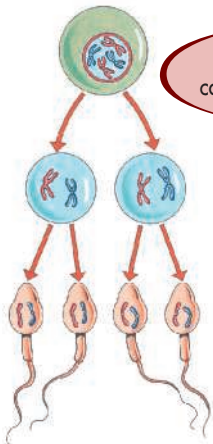


two cell divisions complete the process

four cells (gametes), which differ from each other, are formed

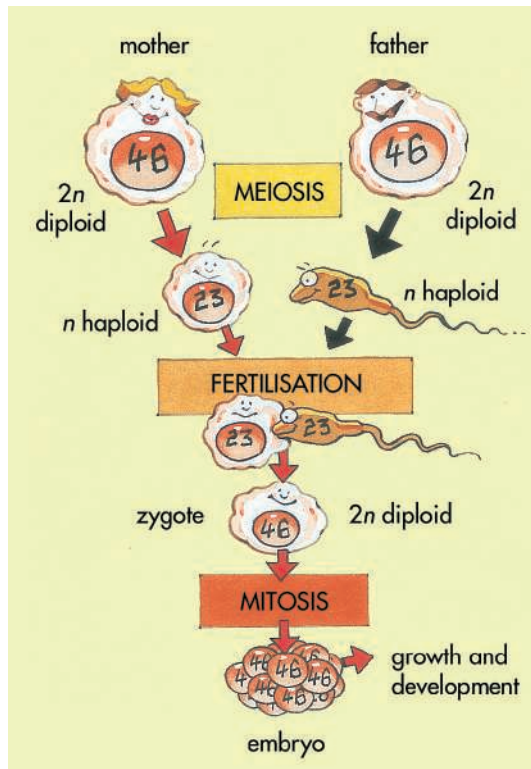
offspring show variation between them

produces new varieties of animals and plants, i.e. increasing variation and diversity



Varying the results

Offspring of sexually reproducing organisms can vary from each other because chance is involved as to which gamete will fertilise an ovum. The mixing of maternal and paternal chromosomes means that a large assortment of coding instructions is possible.



Chromosomes coming together: fertilisation in humans

try This

Mitotic dance

- Examine a prepared microscope slide showing stages of mitosis in plant cells, such as those of an onion. Use the photograph and diagram on pages 202 and 203 to help you identify the sequence of stages.
- Construct a table describing each stage in your own words to help you remember the key features. Include drawings of the chromosomes at these different stages.

sciFacts

Imagine burns destroying nearly 80 per cent of the skin on your body. Advances in tissue culturing now mean there is hope of recovery. Surgeons cut a 4 cm² piece of a burn victim's skin with an electric knife. The skin is washed in salt solution and finely cut into clusters of cells. These are specially cultured, separated and cultured again. From a sample the size of a postage stamp, scientists can grow 2 m² of skin in two weeks.

activities

REMEMBER

- 1 Why does cell division take place?
- 2 What are the two main types of cell division? What is the main purpose of each?
- 3 What are human male and female gametes called and where are they formed?
- 4 What is the difference between the terms 'diploid' and 'haploid'?
- 5 How many chromosomes are in:
 - (a) a human body cell?
 - (b) a human sperm or ovum?

THINK AND DISCUSS

- 6 Different kinds of cells do not grow at the same rate. Where in an animal, such as a mouse, or a plant, such as a bean, would cell growth take place most rapidly? Suggest why this would be the case.
- 7 If n is the number of pairs of chromosomes in a leaf cell of a eucalyptus tree, what would be the total number of chromosomes in the cell?
- 8 Explain why horticulturalists will often grow plants, such as carnations, from cuttings rather than from seed.
- 9 Explain how the body cells of a wallaby joey have the diploid number of chromosomes but the ovum does not.

CREATE

- 10 Work with a partner to make a game to help you learn the stages of either mitosis or meiosis. It could be a mix 'n' match set of cards. Swap your game with someone else. Suggest what aspects of your game could be assessed.

INVESTIGATE

- 11 In *Science Quest 2*, you were introduced to Bruno Annetta, a scientist who communicates scientific concepts using animated cartoons. His cartoon, *The Meiosis Square Dance*, provides a creative way of helping you to learn about the stages of meiosis. Find out more about this or other cell division animations and then create your own cartoon to illustrate the differences between mitosis and meiosis.
- 12 How does the nucleus of a pollen grain of a flowering plant reach the nucleus of the female ovum? Draw a clearly labelled diagram to show your findings. Research an example of a plant which can both self- and cross-pollinate. What is the advantage of this?
- 13 Some kinds of plants, such as mosses and ferns, and animals, such as water fleas and aphids, have two stages in their life cycle. One is a sexual stage and the other an asexual stage. Make a simple, labelled series of illustrations to describe one example.

The next generation

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

The passing on of characteristics from one generation to the next is called **inheritance**. The study of inheritance involves a branch of science called **genetics**.



Some characteristics are passed from generation to generation.

One plus one



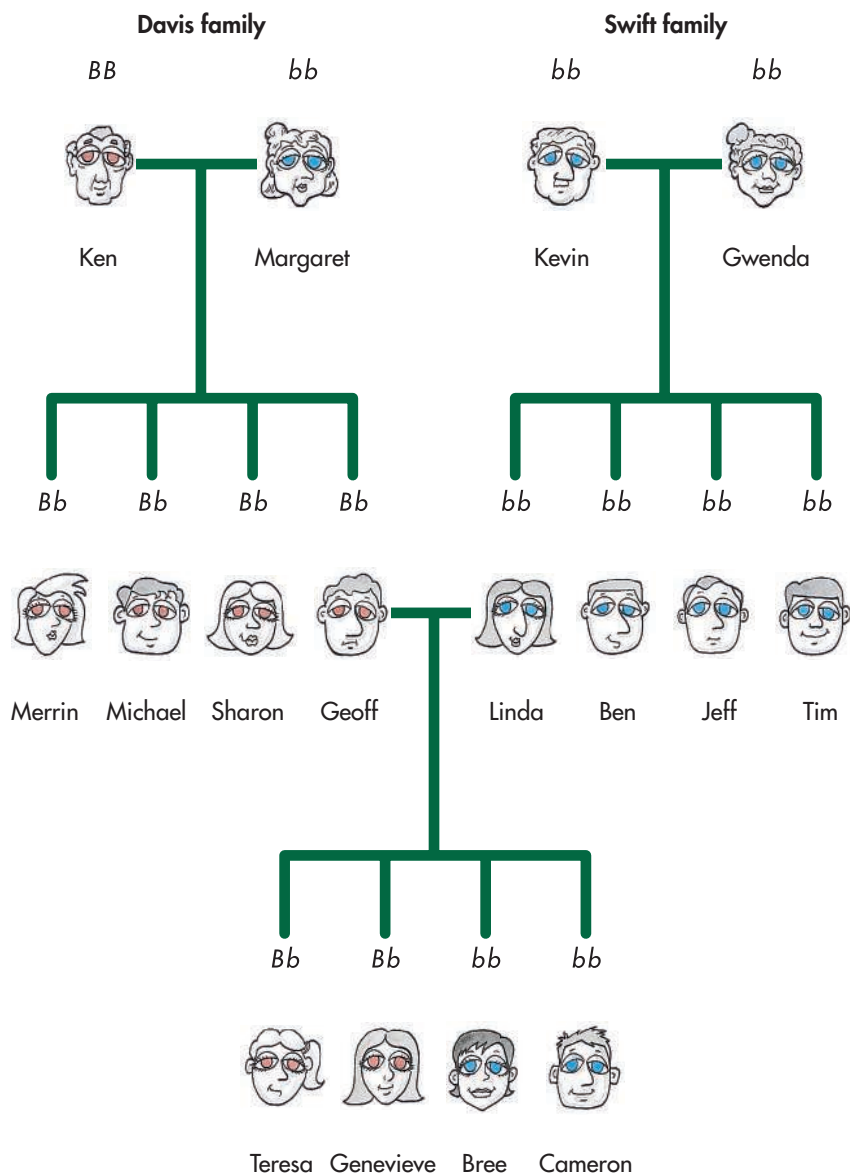
The similarities and differences in how you look compared to your relatives are partly due to chance. Chance was involved as to which of the many sperm produced by your father fertilised your mother's ovum.

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

Hide and seek

The mixing of your parents' chromosomes at fertilisation resulted in two alleles for each gene coming together. Each of these alleles can be described using a letter. In the family generations diagram, the expression of the gene for eye colour is shown. The allele for brown eyes is denoted as a capital letter *B*, because it is

the **dominant** trait. The allele for blue eyes has been denoted by a lower case letter *b*, because this trait is **recessive** to the brown eye trait. If the allele for a dominant trait is present, it will always be expressed. The recessive trait is 'hidden' in the presence of the dominant trait and can be expressed only if the allele for the dominant trait is not present.



The letters *B* and *b* can be used to represent the genetic code for eye colour in the Davis and Swift families.

Mix and match

The combination of the alleles that you have for a particular gene is called your **genotype**. If your alleles for that gene are the same (e.g. BB or bb), then you are described as being **homozygous** (or **pure breeding**) and if they are different (e.g. Bb) then you are **heterozygous** (or **hybrid**) for that trait. A genotype of BB can be described as being **homozygous dominant** while a genotype of bb can be described as **homozygous recessive**. So the genotype has to do with the combination of alleles present; the term **phenotype** describes the expression of the trait (e.g. brown or blue eyes).



Mix up mix up little alleles

1. Sing the song below to the tune of *Twinkle twinkle little star* (or as a rap) and create actions for each line of the song.

Alleles are alternative forms of **genes**

Sometimes showing, sometimes behind the scenes

Genotypes are made up of two of them

Homozygotes have two the same

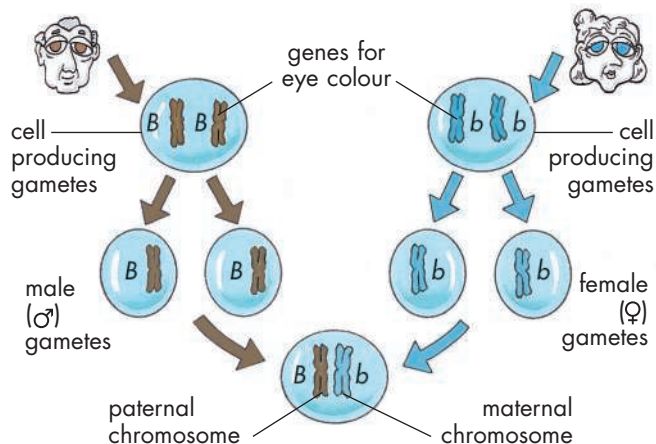
Heterozygotes have one of each kind

From each parent, **alleles** you will find.

2. On your own, in pairs or teams, create a rhyme, song or poem that effectively uses as many of the key terms in this spread as you can. Add movements or actions for each line and share it with your class.

What are you carrying?

The term **carrier** refers to someone who is heterozygous and carries the recessive allele for a trait such as red hair. Generally people are not aware of being a carrier because it is not shown in their phenotype, only in some of their children. How can you explain this?



Genes are carried on chromosomes from parent cells to gametes. What genotype and phenotype would the individual resulting from this fertilisation have?

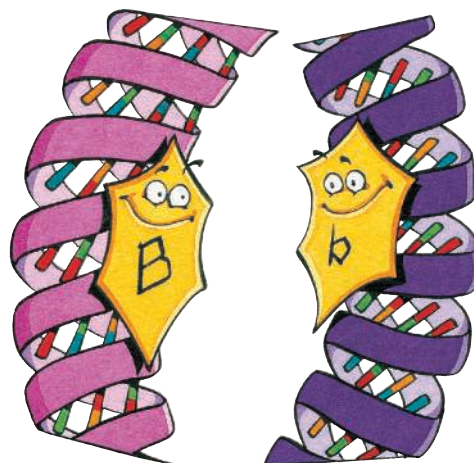


It's all in the family

- Refer to the table below and carry out a survey on your family (or someone else's if you ask first) to find out who has any of the characteristics listed. Draw up another table to record your data. What would you include in it? Make a rough draft first.
 - Analyse your observations by answering the following questions.
1. List any characteristics that seem to be dominant in the family you are studying.
 2. If any recessive characteristics appear, are they more or less frequent than the dominant traits or characteristics?
 3. Are there any characteristics that seem to occur from one generation to another?
 4. Do any seem to miss a generation and then reappear?
 5. What conclusions can you make about your findings?

Some human characteristics due to dominant or recessive alleles

Characteristic (trait)	Dominant trait	Recessive trait
hair	dark and light 	red 
ear	Darwin's point 	no point 
tongue	can roll tongue 	can not roll tongue 
hairline	Widow's peak 	straight 

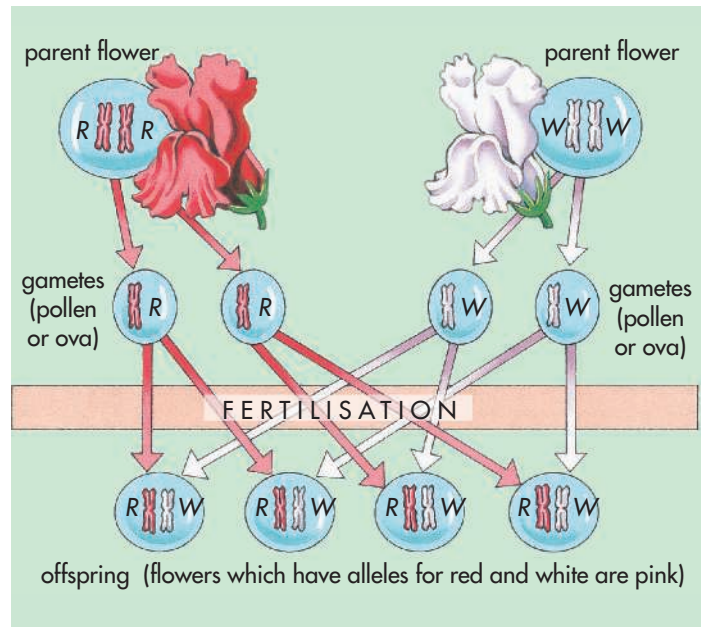


Neither one nor the other

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

sciFacts

For several thousand years, people have cultivated crops such as wheat, and bred animals for meat, wool or for being docile. More recently, horse and greyhound breeding have been popular, with fortunes lost and won on the outcomes. Selective breeding is practised in a whole range of occupations and hobbies, such as horticulture, agriculture, horse racing and dog breeding. By selecting parents with a desirable characteristic or trait and mating or crossing them, the chance of their offspring having that trait is increased.



The genetic code of these snapdragons is an example of incomplete dominance, where neither the red nor the white colouring is dominant.

Mendel's memos

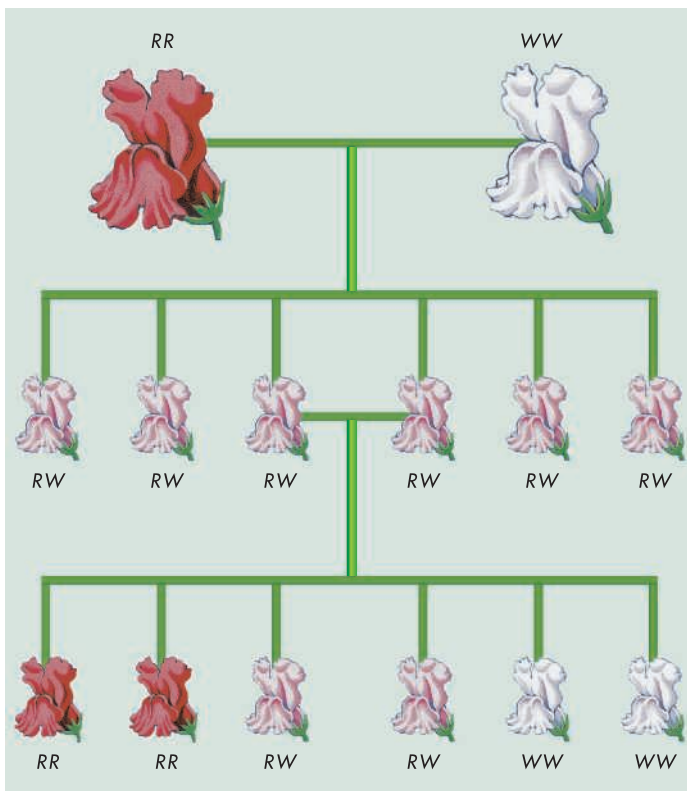
Gregor Mendel (1822–1884), an Austrian monk, carried out experiments on pea plants in a monastery garden for 17 years. His work was unknown for about 35 years. When it was discovered in 1900, he became known as the 'father of genetics'. From his experiments, Mendel was able to explain patterns of inheritance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

Why did Mendel use pea plants and not cabbages? Pea plants are easily grown in large numbers and they have easily identifiable characteristics that have 'either/or' alternatives. Mendel could control their breeding by taking pollen from a particular pea plant and putting it on the stigma of another. Pea plants can also be self-pollinated.

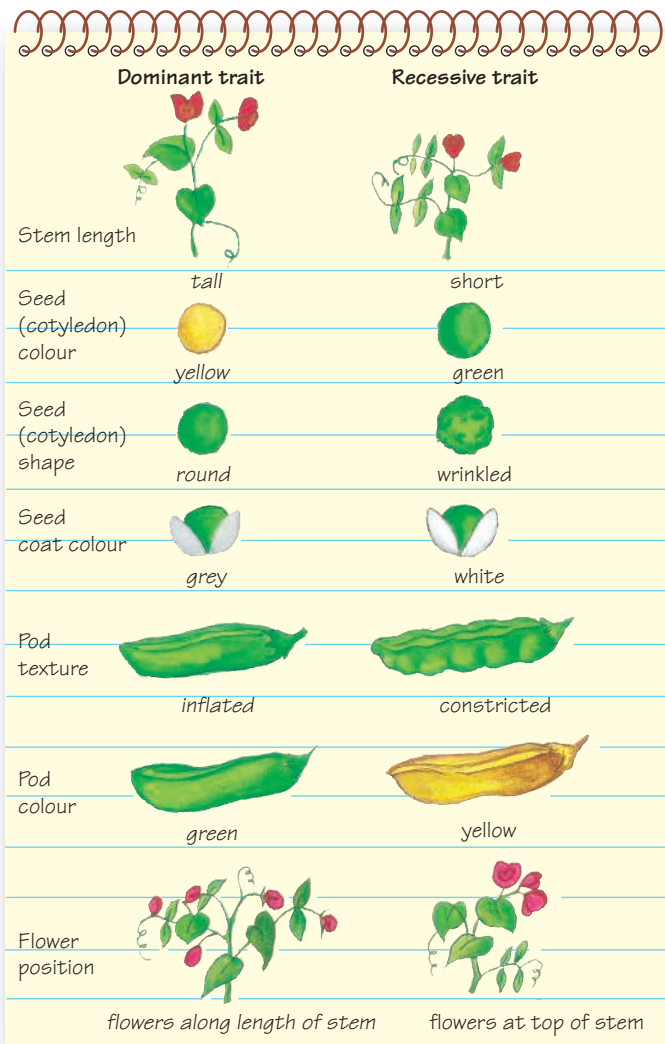
Mendel crossed a pure-breeding tall plant with a pure-breeding short plant. A plant is pure breeding for a characteristic if it has not shown the alternative characteristic for many generations.

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

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



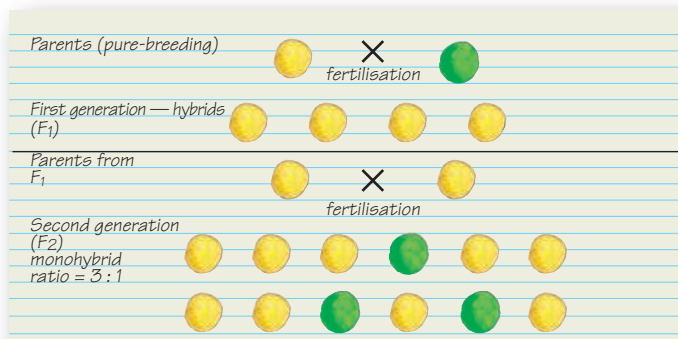
White- and red-flower 'parents'. Why are their offspring pink?



Pea plants showing the characteristics Mendel used in his experiments

A monohybrid cross

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



Monohybrid ratio for yellow and green seed colour

activities

REMEMBER

- 1 Describe selective breeding. Give an example.
- 2 What are Mendel's 'factors'?
- 3 What does F_1 mean?
- 4 Explain the difference between the following pairs. Use symbols and other kinds of illustrations to help with explanations.
 - (a) pure breeding and hybrid
 - (b) gene and allele
 - (c) dominant and recessive
 - (d) homozygous and heterozygous
 - (e) maternal and paternal chromosomes
- 5 State the difference between the phenotypes of a heterozygous individual for a trait that shows complete dominance compared to one that shows codominance.
- 6 How many alleles are there in a cell for a particular gene?
- 7 State the dominant trait out of the following:
 - (a) widow's peak in hairline, straight hairline
 - (b) ability to roll tongue, inability to roll tongue
 - (c) red hair, brown hair
 - (d) no point in ear, Darwin's point in ear.

THINK AND DISCUSS

- 8 Why was the ability to self-pollinate and cross-pollinate an advantage for the pea plants with which Mendel experimented?
- 9 Refer to the pea plant characteristics diagram. Devise a simple table to include the phenotype and genotype for the trait of each plant. Use an appropriate letter to match each of the characteristics Mendel studied.
- 10 Refer to the family tree diagram on page 206. Could the parents of the Davis family, Ken and Margaret, ever have any offspring with blue eyes? Explain your answer.
- 11 Why don't all of Geoff and Linda's children have blue eyes?
- 12 If Genevieve had children with a brown-eyed man, could she have blue-eyed children? Explain.
- 13 Suggest the colour(s) of snapdragon flowers that you would expect in the offspring of a red-flowered plant and a pink-flowered plant.

CREATE

- 14 Make up your own song, rhyme or poem to help you remember the definitions or links between the key terms in this spread.

Getting it together

Thinking about going out on a date? Got the 'hots' for someone special? Next time you look at that special person, take a *really* good look. One day you could be mixing genes together! What might the result be?

Blood type?

Do you know which type of blood you have flowing through your capillaries? The inheritance of blood type A, B, O or AB is determined by partial inheritance and by the ABO gene. This gene has *three* different alleles. Two of these alleles carry the instructions to make a particular type of protein called an antigen; the other allele does not. The types of antigens coded for by the alleles are different. One allele codes for antigen A and the other codes for antigen B. If you possess both of these alleles, then you have the instructions to produce both antigen A and antigen B.

If you refer to the ABO gene as I , then the allele which codes for antigen A could be referred to as I^A , the allele coding for antigen B as I^B and the allele which codes for neither antigen could be i . The ability to make the antigen A or B is dominant to the inability to make either antigen. For this reason, the I^A and I^B alleles are dominant to the i allele. The table below summaries the genotypes and phenotypes for the different blood types.

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

What's the chance?

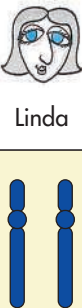
Possible combinations of gametes with particular alleles can be shown in a grid called a Punnett square (see opposite page). It is named after Reginald Punnett (1875–1967), the geneticist who invented it.

The chances of the possible combinations can be calculated using this grid.

The chances of having offspring that show particular traits will depend on their type of inheritance; that is, whether they are inherited by complete dominance, partial dominance (codominance) or incomplete dominance. In complete dominance, there are dominant and recessive traits. Some examples of these are shown in the table below.

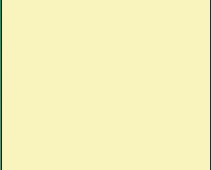
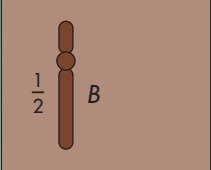
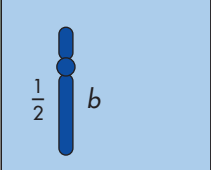
Dominant trait	Recessive trait
free ear lobes	attached ear lobes
mid-digital hair present	mid-digital hair absent
normal skin pigmentation	pigmentation lacking (albinism)
non-red hair	red hair
Rhesus positive (Rh +ve) blood	Rhesus negative (Rh -ve) blood
dwarf stature (achondroplasia)	average stature
widow's peak	'straight' hairline

Remember Linda Swift and Geoff Davis from the previous section? The diagram below shows how two of their children have inherited their eye colour.

	Linda	Geoff	Genevieve	Cameron
chromosomes and genes for eye colour				
genotype	bb	Bb	Bb	bb
phenotype	blue eyes	brown eyes	brown eyes	blue eyes

Why don't Genevieve and Cameron have the same eye colour?

The mix of alleles that Linda and Geoff contributed to Genevieve and Cameron's genetic make-up can also be shown in the format of the diagram below.

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

Another way to illustrate the different alleles in Linda and Geoff's gametes

This can be more simply written as a Punnett square.

		bb parent (female gametes)	
Bb parent (male gametes)		b	b
	B	Bb	Bb
	b	bb	bb

This tells us that the chance of producing the combination Bb (heterozygote) is 2 out of 4 or $\frac{1}{2}$ and that the chance of producing the combination bb (homozygous recessive) is 2 out of 4 or $\frac{1}{2}$. Hence, the couple have a 50 per cent chance of having a blue-eyed child and a 50 per cent chance of having a brown-eyed child.

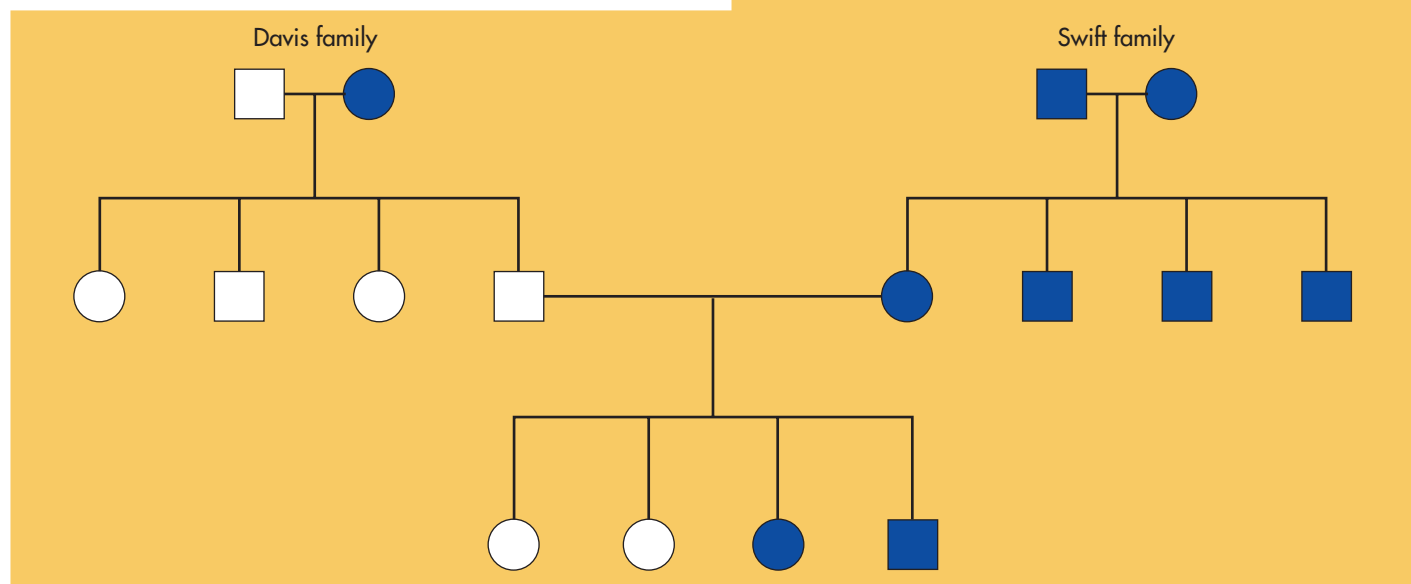
The diagram of the different generations of the Davis and Swift families on page 206 can be described as a **pedigree chart**. Pedigree charts show a family's relationships and how characteristics are passed on from one generation to the next. Another type of pedigree chart for the Davis and Swift families is shown below. Instructions on how to draw your own pedigree chart are on the following page.

sciFacts

A mutation of a gene on chromosome number 8 can result in alopecia universalis, a rare form of baldness. Two copies of the mutated gene cause an individual to have no body hair, eyelashes or eyebrows. Knowledge of the location of this gene provides scientists with information that may result in future therapies for other forms of hair loss.

Key

-  female with trait (blue eyes)
-  male with trait (blue eyes)
-  female without trait
-  male without trait



Pedigree chart for the Davis and Swift families

SOME RULES FOR DRAWING PEDIGREE CHARTS

1. To show the gender of an individual:

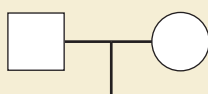
 a square is used to represent a male

 a circle is used to represent a female.

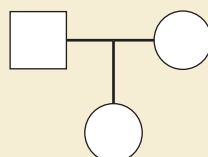
2. To show the marriage or breeding relationship between individuals:

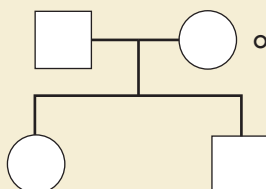
 a line connecting the male and female is used to represent a breeding couple or marriage.

3. To show the offspring relationships:

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

For example,

 an only child (in this case, a daughter)

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

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

 female carrier  male carrier

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

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

 female with trait  male with trait

 female without trait  male without trait

THINK AND DISCUSS

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

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

2 State the genotype of the following individuals:

- heterozygous for blood type A
- homozygous for blood type B
- blood type O
- blood type AB.

3 If a man who was homozygous for blood type A ($I^A I^A$) had a child with a woman who had blood type O ($i i$), what would be the chance that the child would have:

- blood type A?
- blood type B?
- blood type O?

4 If a child had blood type AB, suggest the possible combinations of genotypes of the parents.

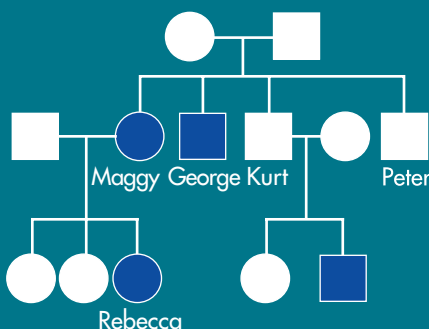
5 Determine the chance of a couple with blood types AB and A having a child with:

- blood type A
- blood type B
- blood type AB
- blood type O.

6 Can a father with blood type A and the mother with blood type B, have a child with blood type O? Explain.

7 What is the chance of Linda and Geoff producing a child with the homozygous dominant combination (BB)?

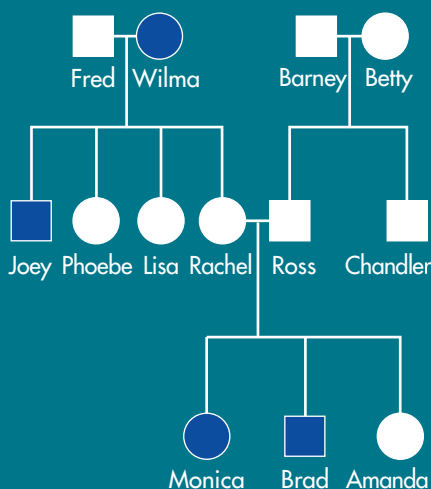
8 Refer to the pedigree of the 'Boofhead' family, in the diagram below. The inheritance of a 'boofhead' (B) is dominant to the inheritance of a 'pinhead' (b).



Shaded individuals show the recessive pinhead trait.
Unshaded individuals show the dominant boofhead trait.

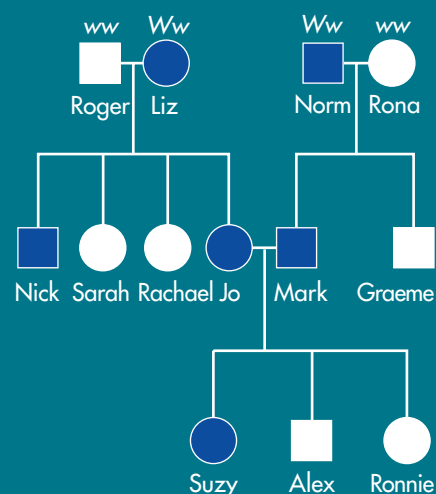
- How many females are shown in the pedigree chart?
- How many males are shown in the pedigree chart?
- How many females have the 'pinhead' trait?
- Suggest the genotype of Maggie's parents.
- Suggest how Maggie inherited a pinhead, when her parents did not.
- Suggest the genotypes of (i) Peter, (ii) Kurt, (iii) George and (iv) Rebecca.

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



- List any observations from the pedigree that support albinism being a recessive trait.
- If the albinism allele was represented as 'n' and normal skin pigmentation as 'N', state the possible genotypes for each of the individuals in the pedigree.

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



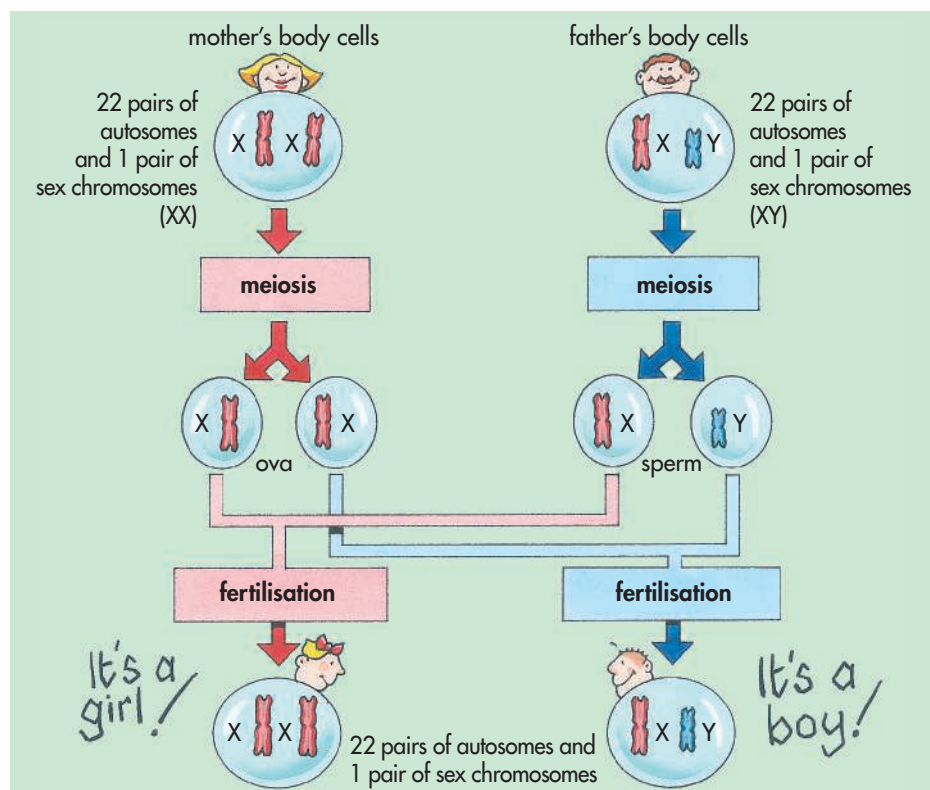
- List any observations from the pedigree that support the widow's peak being a dominant trait.
- If the widow's peak allele was represented as 'W' and the 'straight' hairline as 'w', state the possible genotypes for each of the individuals in the pedigree.
- If Jo and Mark were to have another child,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

Calculating sex

When a friend or family member is expecting a baby, everyone wonders 'Will it be a boy or a girl?'

Boy or girl?

Each human body cell normally carries 46 chromosomes as 23 pairs. Twenty-two of those pairs, called autosomes, are matched, leaving one pair that is not. The chromosomes of this last pair are called the sex chromosomes because they determine whether an individual is male or female. In each of a male's body cells, the pair of sex chromosomes consists of an X and a Y chromosome. The Y chromosome is smaller than the X chromosome and carries fewer genes. A female has 22 pairs plus two X chromosomes.



Boy or girl? Sex chromosomes determine the sex of an embryo.

Doing it differently

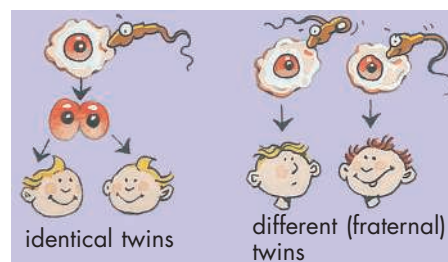
For other animals, the sex of their offspring is determined differently. In birds, it is the female that has different sex chromosomes, Z and W, and the male that has two Z chromosomes.

The temperature of the sand in which the eggs of some crocodiles and turtles are buried affects whether the hatchlings will be male or female. Deeper down at temperatures of 30°C, female hatchlings will develop. Closer to the surface, at 31°C, about half of the hatchlings will be female and half male. Near the warmer surface, at about 32°C, mostly males emerge.

Twins — or more!

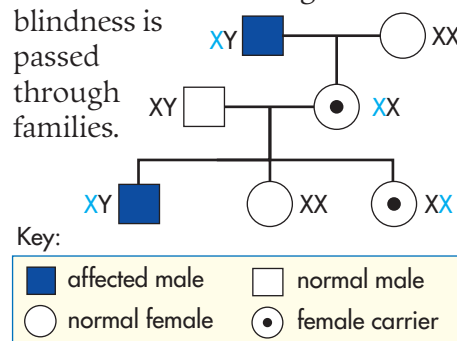
Sometimes in the very early stages of division following fertilisation, clusters of a few cells develop into two separate individuals. If this happens, identical twins result as each cluster has the same genetic make-up as the other.

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



The X-files

Certain types of disorder such as haemophilia and red-green colour blindness are called X-linked characteristics. This means the recessive genes responsible for each are carried on the X chromosome. As there is a greater chance of the disorders appearing in males, they are also referred to as sex-linked disorders. The pedigree chart below shows how red-green colour blindness is passed through families.



Inheritance of red-green colour blindness

What's the chance?

You will need:

20 cent coin

- After reading the instructions and **before** you carry out the experiment, predict what you think the outcomes will be. Give a reason for your prediction.
 - Toss a coin 50 times. Count the number of heads and tails and record the data in a table like the one below.
 - Calculate the percentage chance of obtaining heads and the percentage chance of obtaining tails.
 - Combine the results of the whole class and calculate the percentage chance of obtaining heads and tails.
1. Analyse your data.
 - (a) Was your prediction supported or not?
 - (b) Were the percentage results obtained for 50 tosses the same or different from the total class results? Suggest reasons for the similarities or differences.
 2. If you tossed a coin a thousand times, would you obtain similar results?
 3. What is the chance of obtaining heads each time you toss the coin?
 4. If heads represented a sperm carrying an X chromosome and tails represented a sperm carrying a Y chromosome, suggest how this activity could link to the chances of a male or female baby being conceived.

	Number of heads	Percentage of heads	Number of tails	Percentage of tails
individual tosses				
combined class result				

activities

REMEMBER

- 1 What is the difference between autosomes and sex chromosomes? How many autosomes and sex chromosomes are in each of your body cells? How many are in your gametes?
- 2 What combinations of sex chromosomes determine whether a baby will be a boy or a girl?
- 3 What determines the sex of a bird?
- 4 By what process do the chromosomes of a cell separate to form gametes?
- 5 What is meant by an X-linked characteristic?
- 6 How are identical twins formed?

THINK AND DISCUSS

- 7 With the use of a diagram, explain how the sex of a human baby is determined.
- 8 If a woman has already given birth to three boys, what are her chances of having a girl?
- 9 In many cultures throughout history, a woman has been blamed for not producing sons and has been divorced. From a biological point of view, could this be justified? Explain your answer.
- 10 A few genetic traits, such as hairiness in ears, are due to genes carried on the Y chromosome.

Would males and females have the same chance of having the trait?

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

INVESTIGATE

- 12 (a) Use the Internet to find out if there are similar numbers or considerably more of one gender (boy or girl) born in:
 - (i) Australia over recent years.
 - (ii) China over recent years.
 (b) Suggest reasons for any similarities or differences.
- 13 The kind of job a man does can affect whether he produces more or less 'Y' sperm or any sperm at all. Chemicals and hormones washed into waterways or used in producing food can affect fertility. Research an example of an environmental impact on fertility and report your findings. Make sure you quote the sources of your information.

CREATE

- 14 There are many 'old wives tales' about increasing the chances of having a boy or a girl. Try to find out some by asking older people in your family and by other research. Present your findings in either a PowerPoint presentation, poster, newspaper article, visual thinking tool or poem.

Science at work:

Disobeying orders

Not feeling too good? Sometimes our cells are not able to function as well as they should. It could be the effect of our lifestyle, such as poor diet, lack of exercise or rest, or disease or injury. It could be because of our genes.

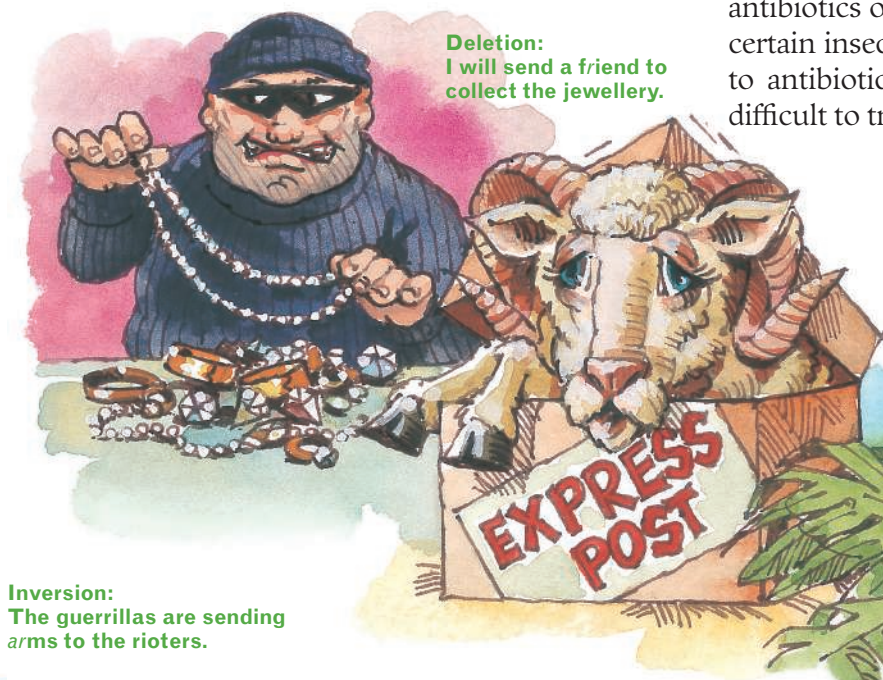
Errors in the code

DNA is very stable and can be replicated into exact copies of itself. This enables genetic material to be passed on unchanged from one generation to the next. Occasionally, however, errors can occur as the DNA is being copied. This means the instructions originally carried by the code are not followed exactly.

Uneven sharing of chromosomes and other chromosomal errors can occur during cell division. Such changes to genes and chromosomes in living things are called **mutations**.

Mutations

Mutations can happen by pure chance or be due to a particular cause such as ultraviolet radiation or too much exposure to X-rays. Any factor that triggers mutations in cells is called a **mutagen**. The chemicals formalin and benzene (which used to be common in pesticides) are examples of mutagens.



Inversion:
The guerrillas are sending arms to the rioters.

Changes in the letters can change a word's entire meaning. Read the sentences in the diagram below as they are, then delete, insert, or invert (turn around) the letters in italics and read them again.

Changes like these in the DNA sequence change the 'meaning' of the code. Minor mutations in body cells seem to have no adverse effect. The body's immune system usually recognises the changed cells and destroys them quite readily.

However, in some cases the mutation has a more severe effect. A necessary enzyme, for example, is not made or a faulty version is produced. Too much or too little of a particular protein could be the problem. A diabetic is not able to produce insulin because of a defective gene. Consequently, blood glucose levels can not be regulated without treatment.

Certain mutations result in uncontrolled cell division, which can result in cancerous **tumours**. Plants can suffer from tumours too.

Not all mutations are harmful. Some species of organisms, such as insects, depend on them for survival. Pesticides may kill the majority of insects sprayed, but there will be some that have slight variations or mutations in their genes, which give them resistance. The mutated genes are passed on to the offspring, who will gain that resistance too. The insects without the resistance will die out. Resistance by bacteria to antibiotics occurs in a similar way. While resistance in certain insects may be good news for them, resistance to antibiotics in bacteria means it can become very difficult to treat diseases caused by these bacteria.

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

Passing them on

If someone's sperm or ovum carries a DNA abnormality, there is a chance of their child being affected. Inherited gene and chromosome abnormalities result in genetic disorders. These can be slight, such as being red-green colour blind, or more severe, such as haemophilia, a disorder in which blood does not clot.

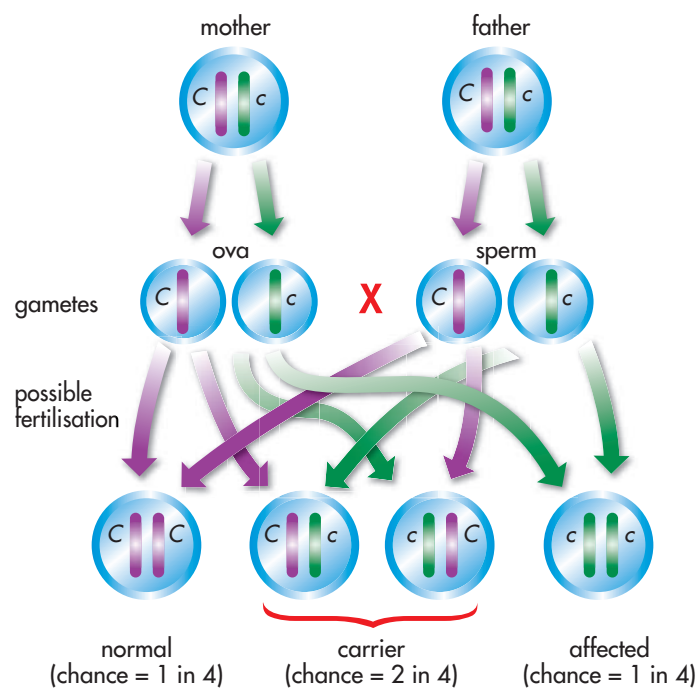
A chance event

About 1 in 2500 people suffer from a genetic disorder called cystic fibrosis (CF). What is the chance of a particular person getting it and why is it important to know?

The faulty recessive *CF* allele is on chromosome number 7. One amino acid in a chain of 1480 amino acids is not produced, causing large amounts of thick mucus to be produced by cells lining the lungs and in the pancreas where digestive juices are secreted. The mucus interferes with the working of the respiratory and digestive systems. Infection readily occurs and sufferers tend to have a shortened life span. Pedigree analysis can show the likelihood of whether a child will suffer from CF.

Since the identification of the defective gene in 1989, the DNA of parents can be analysed to find out if they are one of the one in 25 people that carry the defective gene.

Why is knowing whether you are a carrier so important? For parents who are carriers for CF, the chance of each child suffering from the disorder is at least one in four.

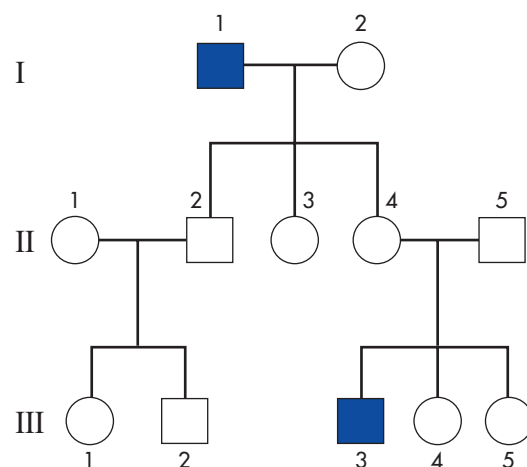


Predicting the chance

Counselling can help carrier parents decide whether to take the chance and have a child. If they choose to go ahead, genetic testing can determine the genotype of the embryo and therefore whether it does not have the condition, is a carrier, or has the condition. What would you choose to do if the embryo had the condition?

Predicting with pedigrees

If there is a history of a particular disorder in the family, how can we find out if the child has the chance of inheriting it? Analysing pedigree charts can help show whether the disorder is due to a dominant or recessive allele and whether the gene is carried on an autosome or a sex chromosome. Pedigrees for normal inherited characteristics, such as hair colour, can be worked out as well as pedigrees for disorders.

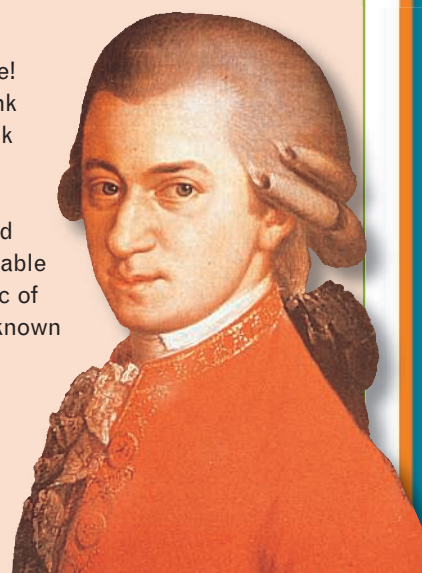


Pedigree chart for a recessive trait such as cystic fibrosis. Which individual do you think could be described as II-3?

sciFacts

Rude, crude, uncontrollable! Hardly what you would think of Mozart. Geneticists think he may have suffered from Tourette's syndrome, Swearing, pulling faces and short periods of uncontrollable twitching are characteristic of this disorder. Mozart was known for all of these.

Mozart may have suffered from Tourette's syndrome.



Naming inheritance

Humans have 46 chromosomes, which include two sex chromosomes (X or Y) and 22 pairs of autosomes. These chromosomes are numbered, based on their size and shape, between 1 and 22. In the nucleus of each body cell are a pair of sex chromosomes (either XX or XY) and one of each of the numbered pairs of chromosomes.

The inheritance of various traits can be described in terms of the location of the gene responsible and whether the inheritance is dominant or recessive. For traits located on the sex chromosomes, the trait is considered to be **sex-linked**. For traits located on the autosomes, the trait is considered to be **autosomal**. A trait that is inherited recessively and caused by a gene on an autosome (e.g. chromosome 21) would be described as being **autosomal recessive**. Likewise, a trait located on the X chromosome and inherited

recessively would be described as being **sex-linked recessive**. The table on the opposite page lists some inherited disorders and whether they are sex-linked or autosomal.



Inheritance song

Sing the lines below to the tune of *Twinkle twinkle little star* (or as a rap) and create actions for each line of the song.

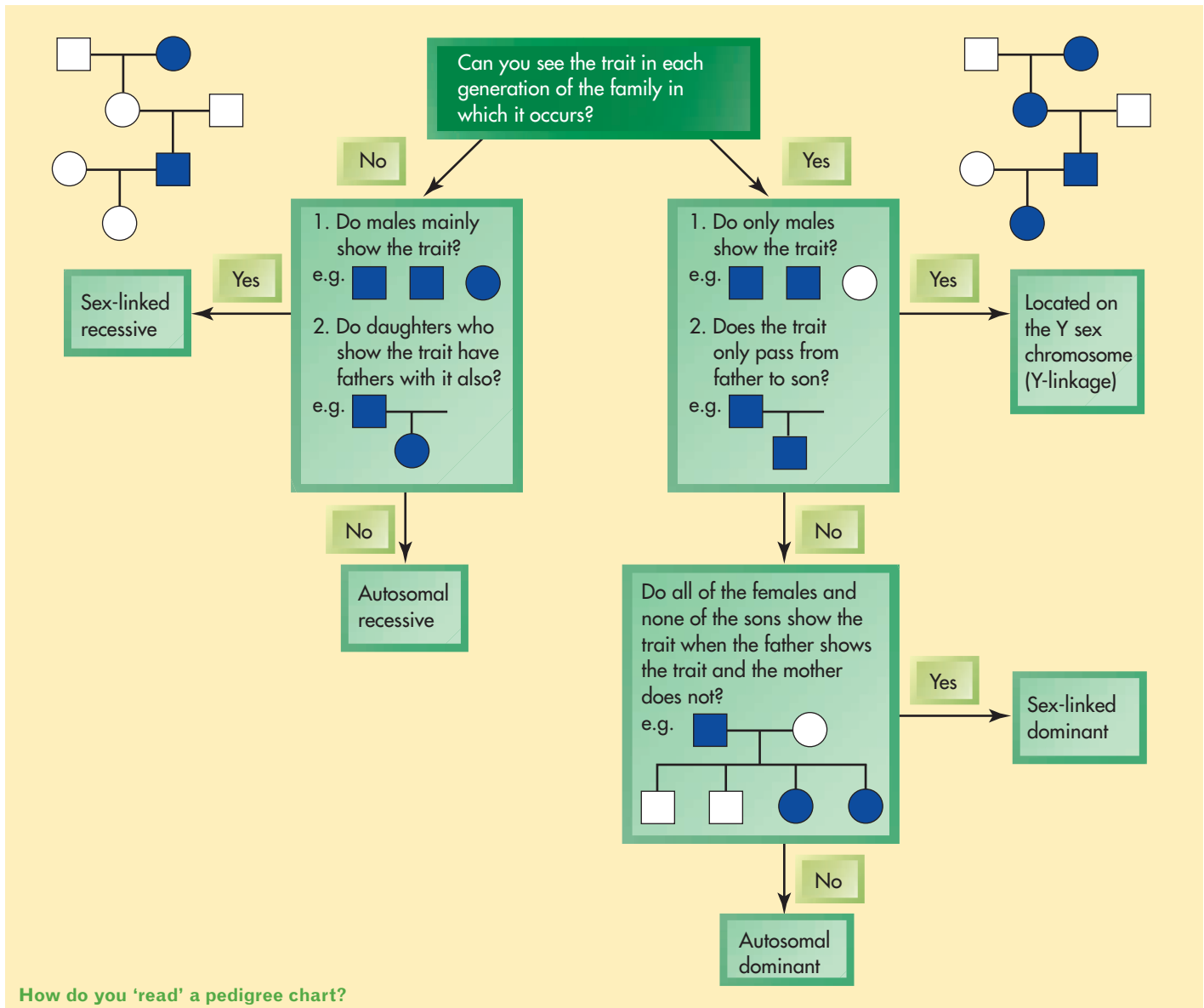
Dominant traits always with an affected parent
While in **recessive** sometimes there aren't

X-linked dominant — dad to his daughters

No skipping seen, always loiters

X-linked recessive — mum to her sons

Can skip generations and hide in **carrier** ones.



How do you 'read' a pedigree chart?

Some diseases that can be inherited

Inherited disorder	Type of inheritance	Symptoms of disorder
fragile X syndrome (FRAX)	sex-linked	leading cause of inherited mental retardation
haemophilia A and B (HEMA, HEMB)	sex-linked recessive	bleeding disorders
Huntington's disease (HD)	autosomal dominant	usually mid-life onset; progressive, lethal degenerative neurological disease
intestinal polyposis	autosomal dominant	many small bulges in the colon form; may lead to colon cancer
dwarfism	autosomal dominant	inhibited growth
sickle cell disease	autosomal recessive	red blood cells become deformed into a sickle shape when oxygen levels are low, which can lead to impaired mental function, paralysis and organ damage
thalassaemia (THAL)	autosomal recessive	reduced red blood cell levels

sciFacts

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

activities

REMEMBER

- 1 What is the difference between a mutation and a mutagen?
- 2 How is a tumour formed?
- 3 (a) How common is cystic fibrosis?
(b) Describe the symptoms of cystic fibrosis.
(c) On which chromosome is the allele for CF located?
- 4 What can pedigree analysis be used for?
- 5 What information does genetic testing provide?
- 6 What is the difference between a disease that is described as being autosomal recessive and one that is described as being sex-linked recessive?
- 7 State the type of inheritance for each of the following diseases:
(a) haemophilia
(b) Huntington's disease
(c) thalassaemia.

THINK, INVESTIGATE AND DISCUSS

- 8 Why do radiographers wear special protective clothing and use remote controls for taking X-rays?
- 9 Suggest examples of mutations that increase chances of survival.
- 10 (a) Find out what types of genetic testing occur in Australia.
(b) Are there any laws, rules or regulations associated with genetic testing? If so, what are they?
(c) List examples of different views and perspectives on genetic testing.
- (d) Suggest reasons for why there are differing views.
(e) Construct a PMI on genetic testing.
(f) What is your opinion on genetic testing?
- 11 In a group, make a list of examples of human genetic disorders. Each person is to write a report on one. Your report could take the form of a poster or information brochure.
(a) Include which gene or chromosomal abnormality is responsible for the disorder and some of the characteristics the affected person would show.
(b) Find out if there are organisations available to support people who have the disorder and their families.
- 12 What does the ozone hole have to do with mutations in plants and animals?
- 13 Describe the cystic fibrosis pedigree on page 217 by answering these questions.
(a) How many generations are shown?
(b) How many females are there in the second generation?
(c) Does the pedigree show any carriers of the trait? If so, which individuals?
(d) How many males show the trait?
- 14 Use the 'How to read a pedigree chart' diagram and the CF pedigree chart to determine:
(a) which type of inheritance is responsible for cystic fibrosis
(b) if individual II-3 is a male or a female
(c) for each individual I-1, I-2, II-4, III-3 in the CF pedigree, whether you think they have cystic fibrosis or are carriers of cystic fibrosis. Give a reason for your suggestions.

Science at work:

A calculated risk

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

There are various tests that can be used to find out about your genetic information. Among these are gene tests (or DNA-based tests), which involve the direct examination of the DNA molecule itself. Other tests include biochemical tests for various gene products (e.g. enzymes and other proteins) or the microscopic examination of stained or fluorescent chromosomes.

Some of the reasons for using genetic tests include:

- carrier screening
- prenatal diagnosis
- newborn screening
- pre-symptomatic testing for predicting adult-onset disorders
- confirmation of a diagnosis of an affected individual
- forensic or identity testing.

Testing at birth

Australian State Screening Laboratories carry out a series of tests on a newborn baby's blood (see the table below). Early testing allows doctors to start any necessary treatment and control. Trying to control the characteristics of human populations by selective breeding or by genetic engineering is called **eugenics**.

Screening tests

Genetic disorder	Symptoms	Incidence
cystic fibrosis	respiratory and digestive problems; early death	1 in 2500
phenylketonuria (PKU)	brain damage due to excessive levels of an amino acid in the blood	1 in 12 000
hypothyroidism	slowed growth and mental development owing to a poorly developed thyroid gland	1 in 3400

The Human Genome Project

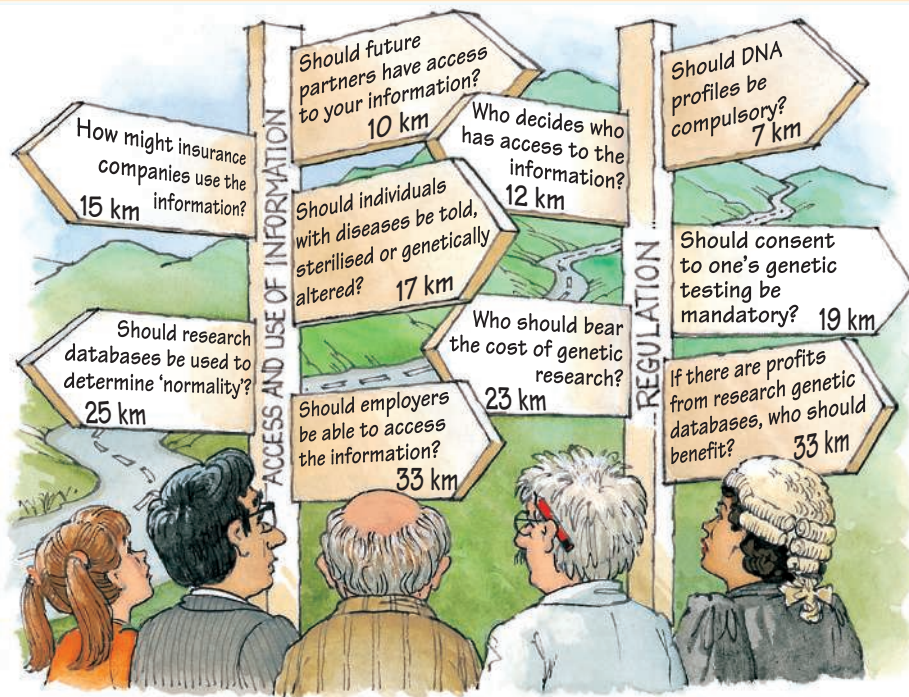
Broadly speaking, the **Human Genome Project (HGP)** is an international investigation aiming to identify, sequence and study the genetic instructions within humans. The genome is the complete set of genetic instructions for making an organism and is made up of genes. The genome size is often described in terms of the total number of base pairs (or bp). The human genome has been estimated

to contain about three billion bp arranged into about 20 000 to 25 000 genes located on chromosomes. Once this information is obtained, how can it be interpreted or analysed?

The HGP aims to produce descriptive maps called **genome maps** of each human chromosome. These maps describe the order of genes and the spacing between them on each chromosome. The total genome size for organisms varies considerably: the fruit fly has 160 million base pairs, while brewer's yeast has 12 million. Once this information is sequenced, it can be used in many different ways. Some of these applications may be beneficial, while others are linked to a variety of ethical, social and political issues.

- 1 What is the issue?
- 2 Who will be affected by the issue?
- 3 What are the positive points of view?
(Who or what benefits and why and how?)
- 4 What are the negative points of view?
(Who or what is disadvantaged and why and how?)
- 5 What are some of the possible alternatives?
- 6 How may these alternatives affect those involved?
- 7 What is a possible solution that may be acceptable to those involved?
- 8 What is your opinion on the issue?

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



The Human Genome Project is both an exciting and dangerous scientific journey.

Too many or too few?

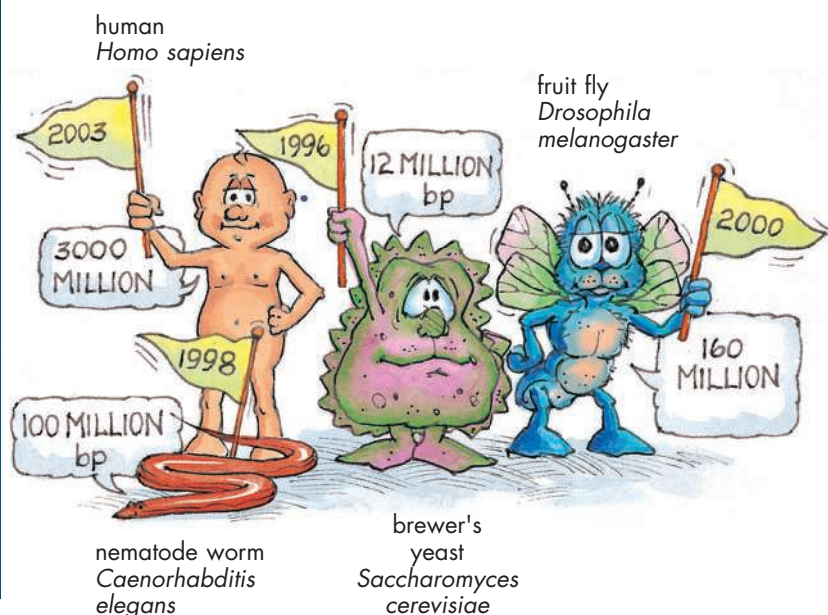
Karyotyping can indicate some kinds of chromosomal abnormalities resulting in disorders. Some examples of such abnormalities are shown in the table below.

Examples of human chromosome abnormalities (mutations)

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



Down syndrome karyotype. What kind of mutation does this show?



When was your genome map sequenced?

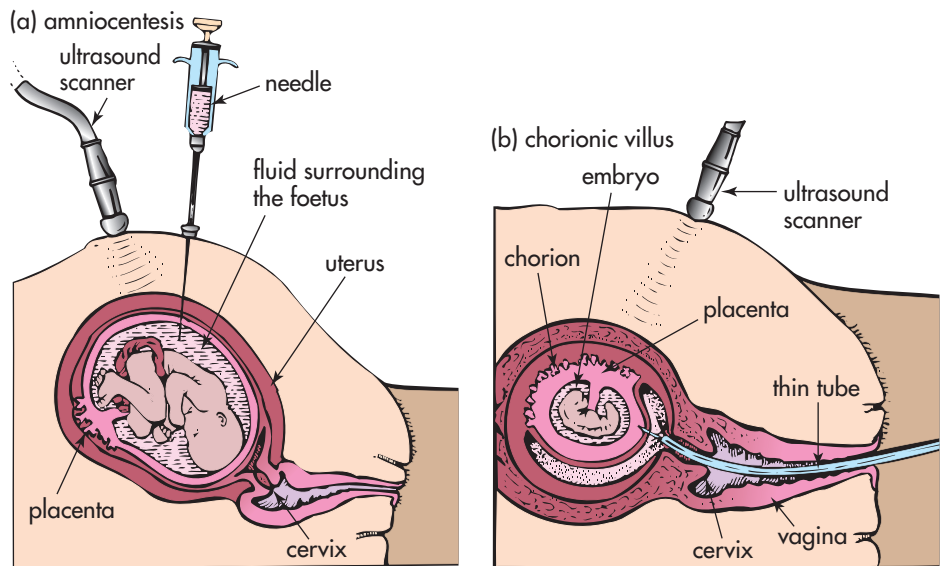
sciFacts

Did you know that:

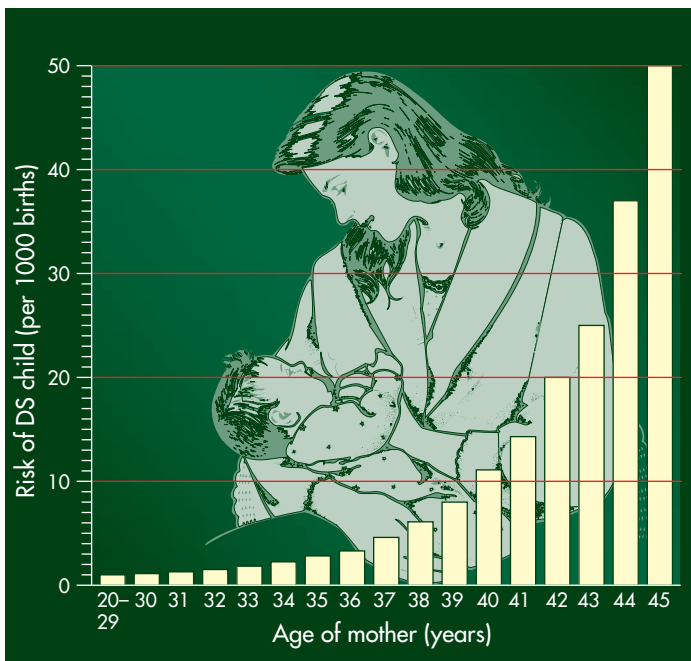
- mouse and human genomes both have about three billion bases, of which only three per cent code for functional genes? The rest is considered to be 'junk DNA'.
- since mice and humans diverged from a common ancestor millions of years ago, most of the DNA that codes for functional genes has remained similar, whereas the junk DNA has mutated and is now extremely different?
- our genome is almost 20 times the size of the fruit fly's and contains far more junk DNA?
- of the 289 human disease genes that researchers have looked for in the fruit fly's DNA sequence, they have located close matches for 60 per cent of them?

Finding out through genetic testing

Using the technique of **amniocentesis**, samples of fluid containing cells shed by the embryo can be extracted from the uterus during weeks 14 to 16 of the pregnancy. In **chorionic villus sampling (CVS)**, actual cells of the foetus are removed at about week 10 to 12 of the pregnancy. The cells are analysed to determine whether the defective allele is present. Both techniques have some risk of miscarriage or damage to the foetus.



Towards finding the answer — some medical techniques



What happens to the incidence of Down syndrome with the increase in maternal age?

Quest for a cure

Could 'normal' genes be inserted into the defective lung cells of a CF sufferer? **Gene therapy** such as this is an active area of research. Before human trials of any therapy can begin, techniques have to be developed, tried and tested in cultures and on animals. There are risks. Will cells with genes inserted be affected in an unpredictable way? Would DNA be damaged during the process of insertion and become cancerous? If the therapy works in non-human animals, does that mean it will work in humans?

sciFacts

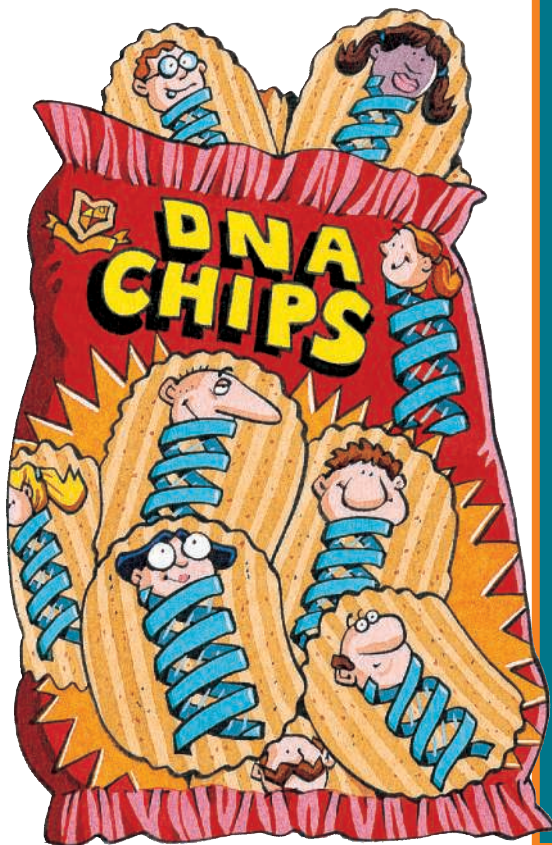
Who was Tutankhamen's dad? Genetic testing on the mummified body of Pharaoh Akhenaton could determine whether he was Tutankhamen's father. However, Akhenaton's mummy is missing, but his portraits show a swollen abdomen and thighs and a very distorted body, signs that he may have been affected by Frolich's disorder. If the gene responsible is found on one of Tutankhamen's chromosomes, the mystery of mummies and daddies may be solved!



'Froliching' chromosomes in ancient Egypt?

DNA chips

DNA chips help 'fish out' portions of your DNA so that parts of your genetic code may be identified. This new genetic technology involves the attachment of probes made of short fragments of selected genes to a wafer to make up a DNA chip. Simply squirting an individual's DNA over a particular type of DNA chip will result in a 'light up' response if it has one of those genes for which the DNA chip is 'searching'. This response occurs as the matching DNA locks on to the relevant probe in the chip, causing the positive response. For example, a chip that carries probes for cystic fibrosis will search for this particular gene while ignoring the 19 999 others. Are you aware that there are currently patents for devices that can detect a variety of genes — including those that affect behaviour and intelligence?



DNA chips can be considered as critical for making sense of the enormous amount of information that current research will supply us with about the human genome. Although the most obvious use is for diagnosis, there are important implications of the easy accessibility of genetic information using this technique. With increasing

knowledge about the human genome, probes for genes with known particular phenotypes (both favourable and unfavourable) may be incorporated into these DNA chips. Which gene probes should be used? Who should make this decision? Who 'owns' the resulting genetic information and decides what is done with it?

activities

REMEMBER

- 1 List three different techniques that can be used to find out about your genetic information.
- 2 State six reasons for using genetic tests.
- 3 State three genetic disorder screening tests performed on newborn babies.
- 4 What are the aims of the Human Genome Project?
- 5 What is a genome?
- 6 What do genome maps tell us?
- 7 Which has the largest genome, a nematode worm or a fruit fly?
- 8 What do you need to consider when involved in a bioethical discussion?
- 9 How are the following types of chromosome abnormalities different?
 - (a) Down syndrome
 - (b) Turner's syndrome
 - (c) Klinefelter's syndrome
- 10 Describe the difference between amniocentesis and chorionic villus sampling.
- 11 Describe the relationship between Down syndrome and the age of the mother.
- 12 What are DNA chips and what do they do?
- 13 What are some of the issues surrounding gene therapy?

THINK AND DISCUSS

- 14 Who do you think should know if you had a higher risk of dying from a genetic disorder in 15 years time?
- 15
 - (a) Read each question on the signposts in the illustration on page 221 and record your immediate response. Try to make up your mind! What kinds of implications (ethical, legal, social, etc.) are associated with each question?
 - (b) In a group of about four, discuss each question. In separate lists record the arguments involved in answering each question. Research additional information if necessary.
 - (c) Consider the arguments carefully, then each person in the group is to review their first set of responses.
 - (d) Did you change your mind about any of your responses? What factors influenced your decision? Did any member of the group change his or her response?
 - (e) If you were a different person involved in the debate, such as a parent or medical scientist, would you give different responses? Explain.

Science at work:

Domesticating biotechnology?

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

Tools of the trade

Genetic engineering is one type of biotechnology which involves working with DNA, the genetic

material located within cells. Genetic engineers use special tools to cut, join, copy and separate DNA.

Do-it-yourself creation kits

Imagine if everyone was able to access genetic engineering tools to create and develop new forms of life. Imagine being able to create new plants for your garden, and pets that you could only dream

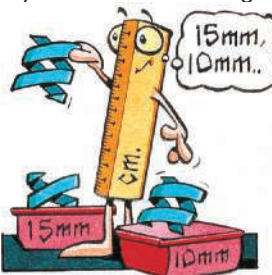
about. Will designing genomes become a personal thing — a type of art form or expression of creativity?

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

Restriction enzymes to cut DNA into fragments at precise locations



Electrophoresis to separate these fragments by their size and charge



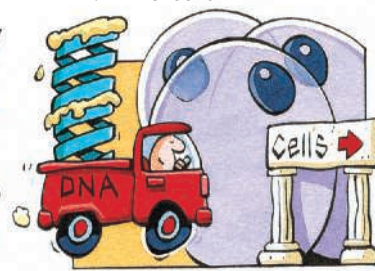
DNA probes to find particular DNA fragments



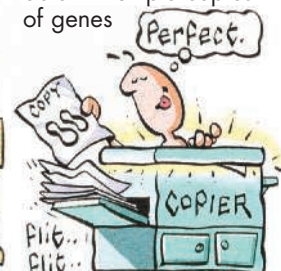
Ligase enzymes to join DNA fragments together



Vectors to transport DNA into cells



Gene cloning to obtain multiple copies of genes



Genetic engineering tools

Counting sheep

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

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

1859

Charles Darwin publishes his explanation of evolution by natural selection.

1866

Gregor Mendel's work on pea plants marks the birth of modern genetics.

1882

Chromosomes are discovered in animals.

1902

Chromosomes are found to carry hereditary information.

1905

Discovery of sex chromosomes

1926

Studies show that X-rays can induce mutations in genetic material.

1944

Geneticists prove that DNA is hereditary in bacteria.

1951

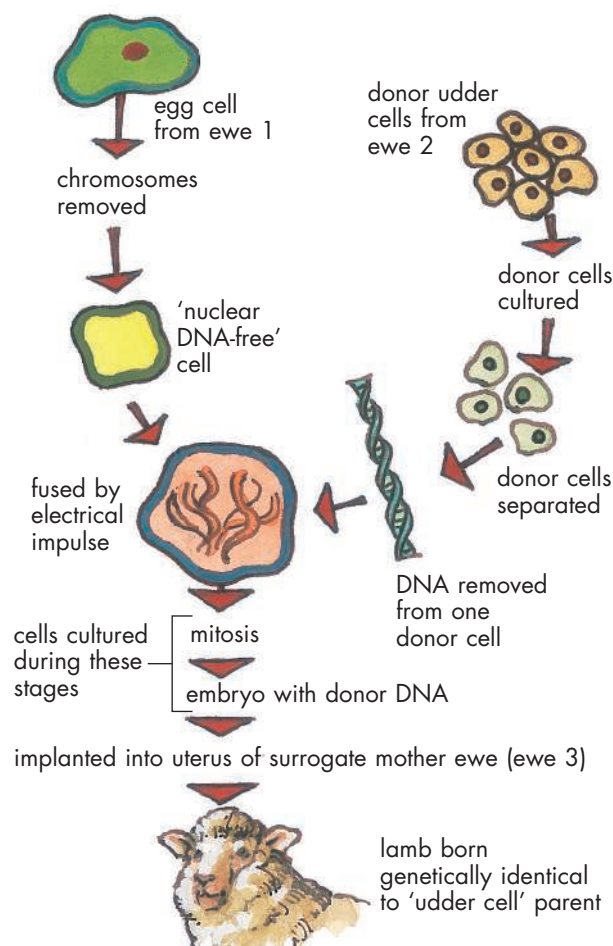
X-ray diffraction images of DNA are captured for the first time by Rosalind Franklin.

1953

James Watson and Francis Crick deduce the double-helix structure of DNA.

Dolly dared to be different! She has shown that it is possible to reprogram adult cells to develop differently. Follow her story in the diagram below.

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



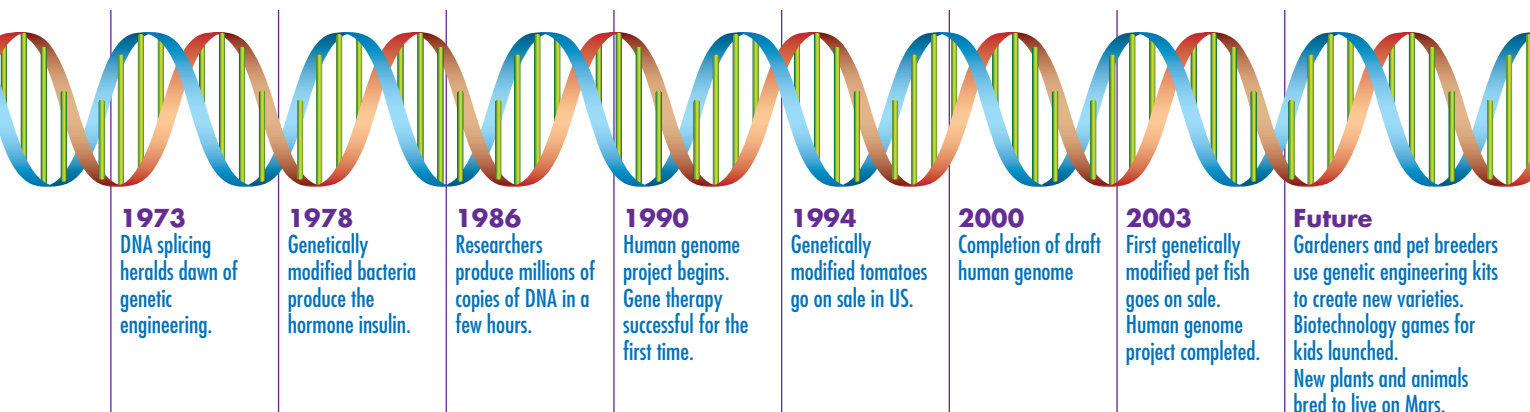
activities

REMEMBER

- 1 What are clones?
- 2 Why is Dolly famous?
- 3 Refer to the diagram showing Dolly's creation.
 - (a) What had to be done to the unfertilised egg taken from ewe number 1?
 - (b) How did the donor cell DNA (from ewe 2) get inside the 'empty cell' from ewe 1?
 - (c) What was ewe 3's role?
- 4 Summarise the tools of genetic engineering into a mind, concept or cluster map. Include diagrams or figures to help you remember what each of the tools does.

INVESTIGATE, THINK AND DISCUSS

- 5 Why is Dolly called a clone?
- 6 Which ewe was the original Dolly?
- 7 Does Dolly have any genetic material from a ram?
- 8 There is now no scientific reason why cloning of humans, even of dead ones, is not possible. Some people have suggested that cloning of humans should be permitted while some countries, such as the United States, have outlawed it.
 - (a) What reasons would people give for wanting to clone humans?
 - (b) Research media stories that raise issues related to Dolly and cloning, particularly of humans. Draw an issue map to identify some of the issues and implications. Keep a record of your references.
- 9 Since Dolly was cloned, a number of other animals have also been cloned. Find out more about the cloning of at least one other animal. Report on:
 - (a) the reasons for cloning that particular animal
 - (b) how the animal was cloned
 - (c) the advantages and disadvantages associated with the cloning of the animal.
- 10 Use the timeline to answer the following questions.
 - (a) In which year were sex chromosomes discovered?
 - (b) Find out when Dolly was born and add it to the biotechnology timeline.
 - (c) Find out two other biotechnological events to add to the timeline.
 - (d) Research one of the events on the timeline in more detail and share your findings with your class.
- 11 Design a biotechnology game of the future.
- 12 Find out the environmental conditions on Mars and think about what sort of life form could survive them. Then design a plant or animal that you think would have a very good chance of surviving on Mars.



Science at work: All mixed up

Human genes in bacteria?
Insect genes in plants?
Cotton plants producing granules of plastic for ultra-warm fibre?
This sounds bizarre but it is not in the realms of fantasy — it is happening now.

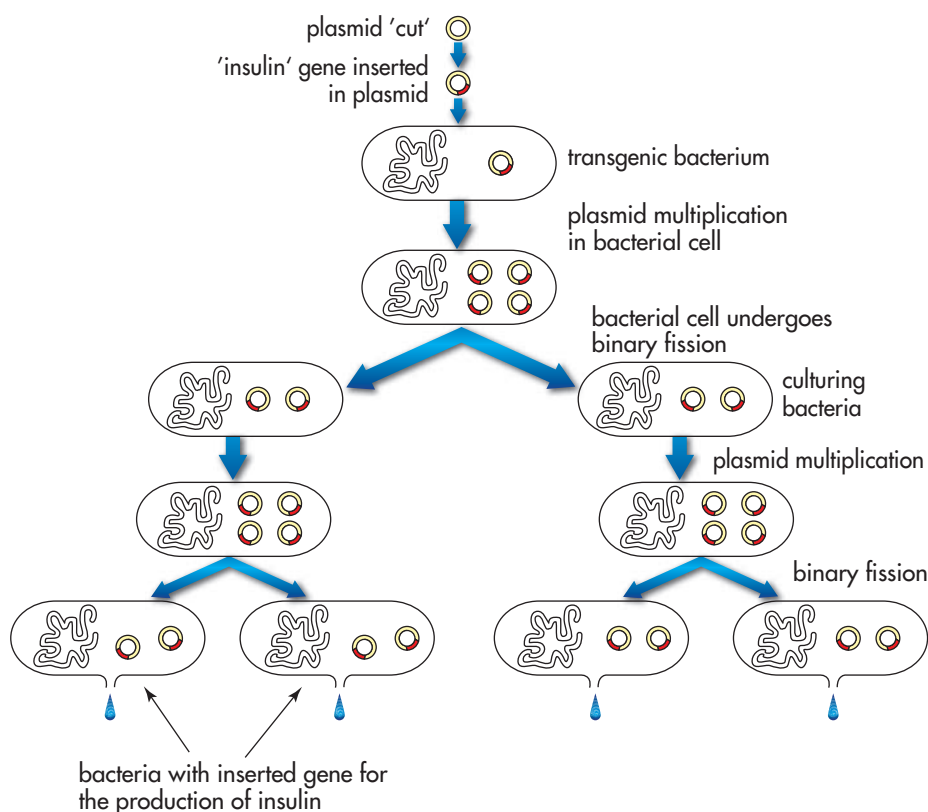
Transferring the code

DNA can be manipulated in different ways using **gene technology**. Sections of DNA can be cut using specific enzymes and the 'desirable' genes of a species inserted into the DNA of the same or another species. Plant genes can be inserted into animal cells and animal genes into plant cells. A plant, animal or other kind of organism with a foreign gene is known as **transgenic**. The foreign gene will behave in the same way as it would in its own species.

The addition of a gene from Arctic fish to tomato plants, for example, results in the tomatoes being frost resistant.

Busy bacteria and insulin

Before developments in gene technology, sufferers of diabetes had to use insulin extracted from the pancreas of pigs. Immune reactions were common. Now the human gene for the production of insulin is inserted into the **plasmids**, rings of DNA, of bacteria. The transgenic bacteria and their plasmids divide rapidly when cultured in large vats and produce large quantities of insulin.



Busy bacteria: production of human insulin

The plant invader

Agrobacterium tumefaciens is a soil bacterium. It is able to get inside and infect many plants such as vines and fruit trees. In doing so, it transfers a tiny piece of its DNA into the host cell. This programs the host cell to make chemical compounds for the sneaky bacterium to feed on. Genetic engineers saw the possibility of using this bacterium as a **vector** to carry the genes they wanted from one plant into another.

Other kinds of bacteria and viruses act as vectors and carry genetic information from one organism (or synthesised to be like that organism) to another organism. Vectors can be used to carry genes for producing protein in soybean and sunflower plants, producing enzymes to control chemical processes, and producing compounds that keep insects or pathogenic viruses at bay.



A gene technologist at work

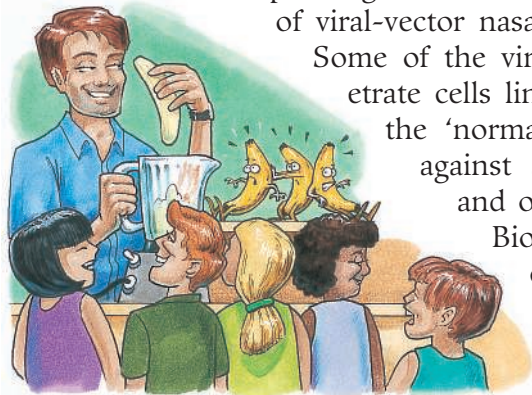
Take aim . . . fire!

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

Biotechnology involving gene technology is a rapidly expanding branch of science. Already there have been trials of viral-vector nasal sprays to help treat CF sufferers.

Some of the viruses carried by these vectors penetrate cells lining the respiratory tract and insert the 'normal' gene into those cells. Vaccines against a number of diseases in humans and other animals are being investigated.

Biotechnologists are trying to genetically engineer bananas to produce a vaccine against the hepatitis B virus. Will genetically engineered vaccines eventually treat diseases such as AIDS?



Bananas as a vaccine against hepatitis B?

Should or shouldn't we?

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

There are also legal problems. Should companies be allowed to patent particular genes so that they 'belong' to those companies? Who should decide?

activities

REMEMBER

- 1 What is gene technology?
- 2 Draw a simple flow diagram to summarise how bacteria are used to produce insulin.
- 3 Explain what transgenic organisms are and give an example.
- 4 What is a vector?
- 5 What methods are used to transfer 'foreign' genes into other organisms?

THINK AND DISCUSS

- 6 What are some of the advantages and disadvantages of gene technology? Use specific examples to support your views.
- 7 Would you eat genetically manipulated food? Why or why not?

INVESTIGATE

- 8 In a group, compile a folio of about 10 journal and other media articles relating to gene technology. Make sure you note the date and source of each one. Each person will choose an article to analyse. In your analysis include:
 - (a) the kind of gene technology being reported on
 - (b) a simple description or explanation of the particular example of technology
 - (c) any issues relating to the example.

- 9 Any gene technologist or researcher conducting experiments in an institute has to have the approval of the biosafety and bioethics committee. Imagine you and your research team are applying for permission to develop a particular transgenic organism for the company you are working for. Working in a group of about five, discuss the following questions.
 - (a) What will be the name of your company?
 - (b) What transgenic organism would you like to develop and why?
 - (c) Who would you expect to be represented on the committee evaluating your submission? Allocate roles within the group for the research applicant(s) and members of the committee.
 - (d) What guidelines would the committee have to draw up to ensure:
 - (i) safety to researchers and technicians in the lab?
 - (ii) safety to the environment?
 - (iii) ethical issues are addressed?
 - (e) What questions do you think each member of the committee should ask the applicant(s) before the committee makes the decision? Make sure the applicant(s) prepares appropriate answers to the questions.

Present your findings as a role-play or a debate. Ask your audience to decide whether the committee should or should not agree to the proposal.

Science at work:

Genetic fingerprints

Think that you can hide your identity? Maybe today, but definitely not tomorrow. DNA technology is rapidly providing techniques that will bring out into the open your deepest secret about yourself.

Has your DNA got the stutters?

Did you know that about 90 per cent of the DNA in your chromosomes has no known function? These non-functional parts vary in length and consist of about 20–40 bases in a sequence that is often repeated many times; for example: GATTACAGATTACAGATTACAGATTACA. These repetitive sequences are called **microsatellites**.

It is the variation in each individual's base sequence patterns in this non-functional DNA that forms the basis for **DNA fingerprinting**. This technique produces a kind of 'bar code' of the natural variations found in every person's DNA. It is this 'bar code' or DNA fingerprint that enables the identification of the individual to be made. It has been calculated that only one in a million persons are likely to have identical DNA fingerprints.

Accused in double murder case extradited after DNA match

FOR two days a smiling Tuan Dang Nguyen insisted it was a case of mistaken identity. Victorian homicide squad detectives had arrested him in Cabramatta over the machete murders of two women fruit pickers.

Sydney Morning Herald, 21 April 2006

FIGHTING AN HEREDITARY FOE

CANCER has been known as the silent killer, but advances in genetic testing are alerting women to family mutations and giving them a say in their future. Some people are wired for breast and ovarian cancer. Within their DNA lurks a gene flaw that was present at their conception in the egg or sperm.

Illawarra Mercury, 3 September 2005

Football clubs consider genetic tests for potential recruits

TWO of the AFL's most powerful clubs are considering DNA testing young recruits to test their likely height, strength, speed and stamina.

The Age, 21 June 2005

DNA tests bring sadness to fathers

BIOLOGY is everything — and nothing — to men who discover they are not the biological father to children they believed theirs.

Sydney Morning Herald, 20 May 2006

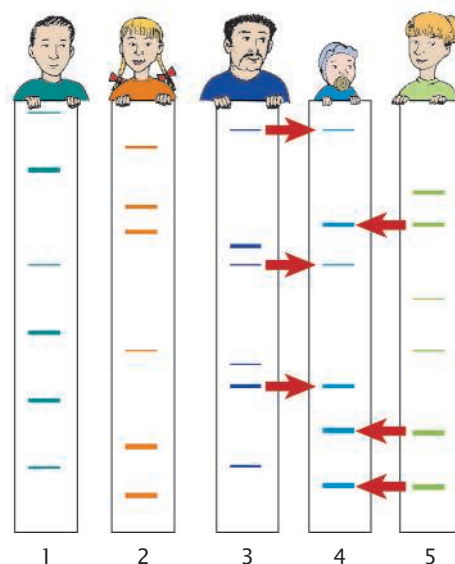
AFL TO BAN GENE TESTS ON RECRUITS

AFL clubs that conduct genetic tests on potential recruits will face heavy financial sanctions, and possibly loss of premiership points, after the league yesterday moved to outlaw the practice.

The Age, 23 June 2005

DNA fingerprinting

DNA fingerprinting involves analysing DNA fragments. After the extraction of these fragments from biological material, restriction enzymes are used to cut the DNA into specific fragment lengths. DNA patterns are then obtained using a process called electrophoresis.



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

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

GENETICS TELL STORY

THE uniqueness of Australian Aboriginal people and their long association with the continent has been revealed in a landmark genetic study.

Newcastle Herald, 15 April 2006

Collect the sample.



Extract the DNA from the sample.



Separate the DNA fragments on the basis of their length, using electrophoresis.



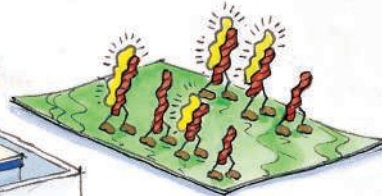
Cut the DNA into fragments using restriction enzymes, which cut at certain base sequences they recognise.



Split the DNA into single strands and transfer them onto a nylon sheet.



Immerse nylon sheet in bath with DNA radioactive probes. Probes bond to the core sequences of the sample DNA fragments.



Expose nylon sheet to X-ray film. The radioactive probes attached to DNA fragments show up as dark bands.



Making a DNA fingerprint using the tools shown in the cartoon on page 224.

activities

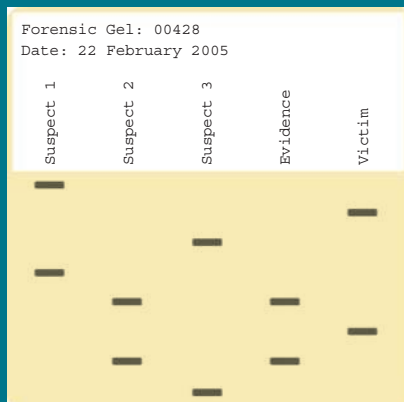
REMEMBER

- 1 About what percentage of the DNA in your chromosomes has no known function?
- 2 What are microsatellites?
- 3 Which part of the DNA forms the basis for DNA fingerprinting?
- 4 State some applications or uses of DNA fingerprinting.
- 5 Outline the steps involved in the technique of DNA fingerprinting.

THINK, EVALUATE AND DISCUSS

- 6 The diagram on the right shows the DNA fingerprint of the victim,

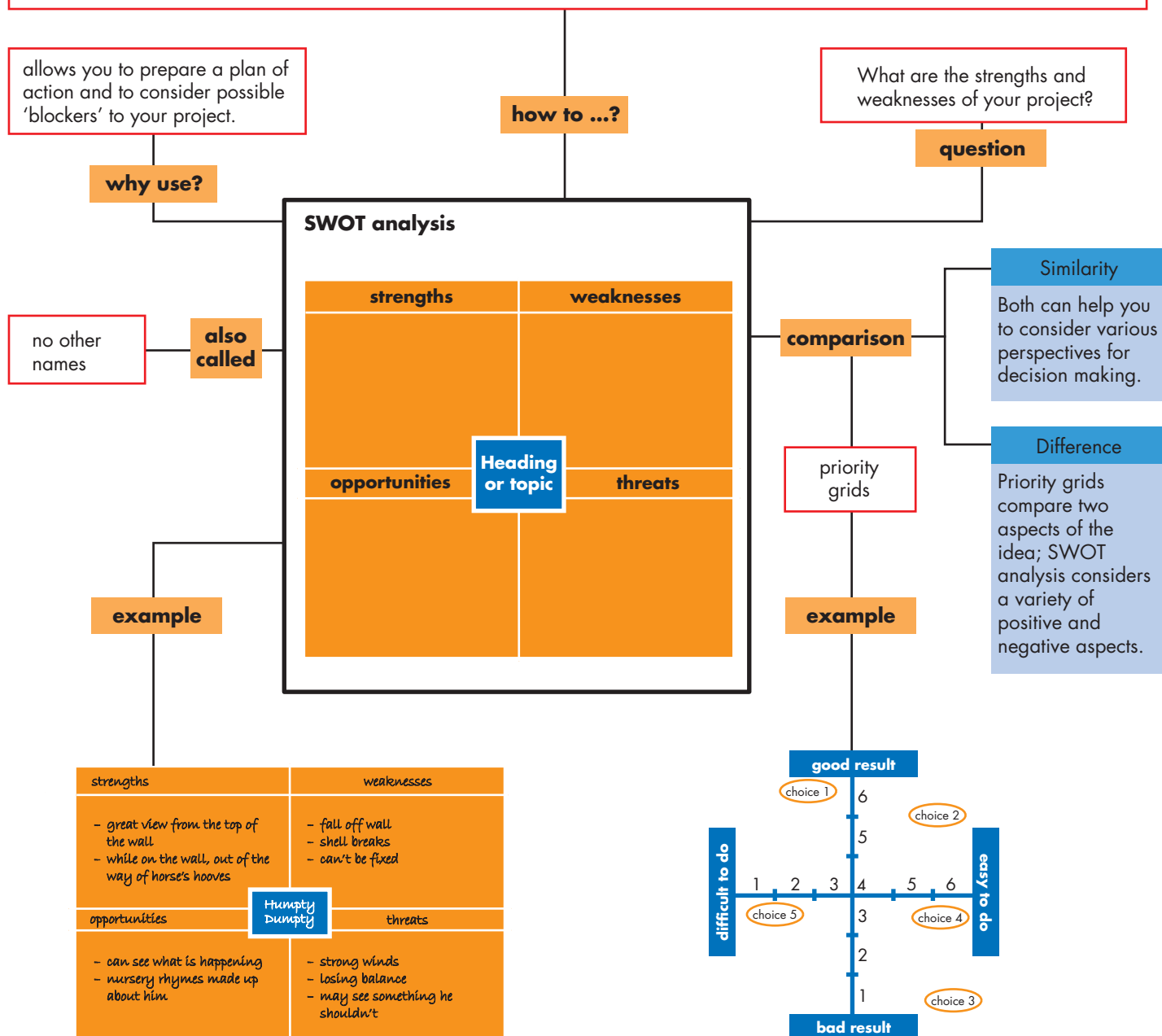
the DNA fingerprint from evidence taken from her body after an attack and the DNA fingerprints of three suspects.



- (a) Using the information in the DNA fingerprints, which of the three suspects is most likely guilty of the crime against the victim?
- (b) Give reasons for your response to part (a).
- (c) Suggest why a sample was taken from the victim as well as the foreign DNA sample collected from her body.
- (d) (i) State some other forensic diagnostic tools that exist to identify those guilty of crimes.
(ii) How do these compare with DNA fingerprinting?

Thinking tools: SWOT analyses and priority grids

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



Adapted with permission from Hawker Brownlow Education © 2005 modellearning.com *Thinking Skills and EyeQ: Visual Tools for Raising Intelligence* by Ian Harris, Oliver Caviglioli and Bill Tindall

THINK, DISCUSS AND CREATE

- 1 Read through the article *IVF test for gene killers* then answer the following questions.
 - (a) What does PGD stand for?
 - (b) What does the PGD test identify in embryos? Include four specific examples in your response.
 - (c) Outline the opportunity that this test offers families with histories of genetic disorders.
 - (d) Outline a negative aspect of the PGD test.
 - (e) At which stage is the embryo when a single cell is removed from it?
 - (f) Are you aware of any bias in the article? How many different perspectives were included? If you were to write the article, what other information or details might you include?
- 2 (a) In your team construct a SWOT analysis on **pre-implantation genetic diagnosis**.
 (b) Share your SWOT with another team and discuss any

similarities and differences between your SWOTs.

- (c) On the basis of your discussions, construct your own SWOT on the article.

- 3 In your team, brainstorm statements or choices related to genetic testing of embryos. Select five of these statements and position them on a priority grid with the following labels.

Horizontal

left-hand side — difficult decision
right-hand side — easy decision

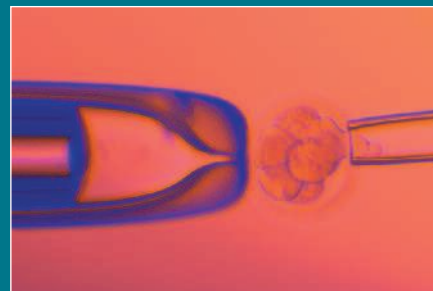
Vertical

top — good result
bottom — bad result

INVESTIGATE AND DISCUSS

- 4 Find out more about two of the genetic disorders listed in the article. Share and discuss your findings with your team or class members.
- 5 Genetic tests can be ordered on the Internet!

- (a) Find out examples of tests that are currently available.
- (b) Suggest some implications of being able to buy genetic tests in this way.
- (c) Is the unregulated sale of genetic tests potentially misleading or unethical? How can the accuracy and privacy of these tests be ensured?
- (d) Who owns the information of genetic tests that are ordered on the Internet? Comment on your findings.
- (e) Construct your own SWOT analysis of your findings, and then share it with others in your class.



A single cell being extracted from an eight-cell embryo.

IVF test for gene killers

by Robyn Riley

A TEST pioneered by Victorian scientists could reduce the incidence of 40 deadly genetic diseases, including breast cancer and cystic fibrosis.

Called pre-implantation genetic diagnosis (PGD), the controversial test identifies embryos carrying gene mutations for more than 30 inherited cancers as well as those destined to develop fatal lung and blood disorders, even deafness or blindness.

The team from Monash IVF said the diagnosis was accurate and could save parents a lot of heartache.

It offers hope to families with a history of genetic disorders, allowing them to have their children through IVF and screen out the killer diseases.

But the test has its critics. The Rev Norman Ford, director of the Caroline Chisholm Centre for Health Ethics, said the diagnosis led to the unethical destruction of embryos.

He said the life of every human embryo was morally inviolable, regardless of its genetic heritage.

'There are credible scientific and philosophical grounds for holding that early embryos are human individuals from conception and as such should be safeguarded as persons,' Mr Ford said.

Dr David Cram, director of Molecular Genetic Services at Monash IVF, said the diagnosis could be used to test for 'pretty much any genetic condition where the exact genetic cause was known'.

The test is carried out on an IVF embryo when it has developed eight cells.

A single cell is removed and its DNA is tested for gene mutations.

Dr Cram said despite the test being readily available, many Victorian couples at risk were still unaware it was available.

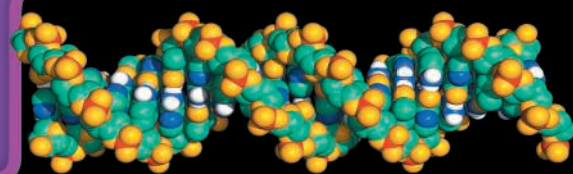
A free community PGD education program was held in Melbourne yesterday run by Monash IVF, Melbourne IVF and the Infertility Treatment Authority.

It's all in the genes

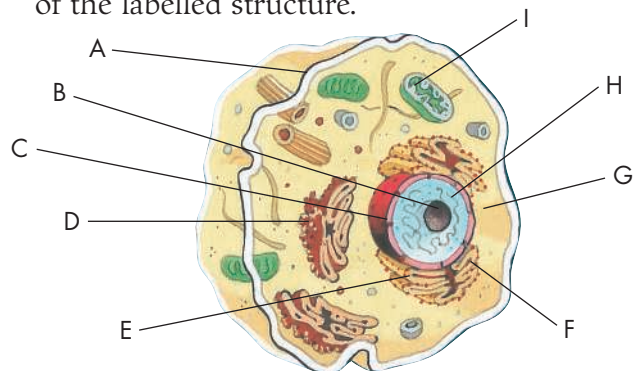
Among other conditions, the test can be used to identify:

- Chromosomal imbalances that cause infertility
- Chromosomal imbalances and translocations that cause recurrent miscarriage and disorders, including:
 - Alports Syndrome
 - Beckers Muscular Dystrophy
 - Duchennes Muscular Dystrophy
 - Haemophilia
 - Kenney Disease
 - X-linked Mental Retardation (non-syndromic)
 - Alagille Syndrome
 - Amyloidosis
 - Beta Thalassaemia
 - Cystic Fibrosis
 - Diastrophic Dysplasia
 - Huntington's Disease
 - Lesch Nyham Syndrome
 - Retinoblastoma
 - RhD Factor Incompatibility
 - Spinal Muscular Atrophy
 - Tuberous Sclerosis

Looking back



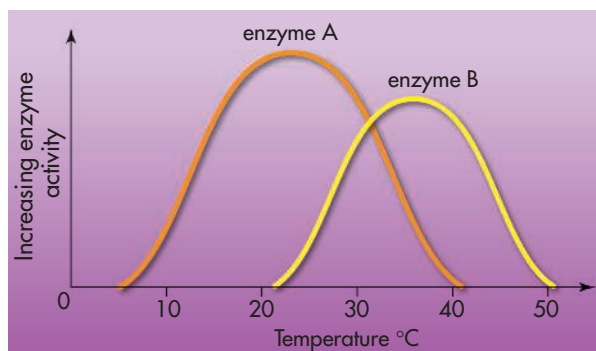
1. Match the letters on the diagram of the cell below with the corresponding names in the table. Write a brief description of the function of the labelled structure.



Structure of a cell

Letter	Name	Description/function
	endoplasmic reticulum	
	nucleus	
	mitochondrion	
	cytoplasm	
	ribosomes	
	nuclear membrane	
	Golgi body	
	cell membrane	
	nucleolus	

2. Graphs are useful for showing if there is a relationship or pattern between variables or factors. The following graph shows the relationship between the activity of two enzymes and temperature.



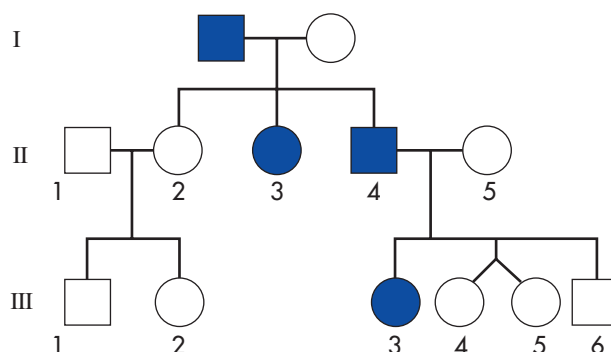
Graph showing the relationship between enzyme activity and temperature

- (a) Describe the trends shown by the graph.
 (b) Which enzyme, A or B, functions best at human body temperature (37°C)? Explain your answer.
 (c) What are enzymes composed of and what function do they have in cells?
 (d) Enzymes are made up of protein. Suggest the relationship between DNA and enzymes.
 (e) What effect may a mutation in the DNA of an organism have on the enzymes' production?
 (f) Suggest a possible relationship between gene therapy, mutations, enzymes and DNA.

3. The pedigree chart below shows the inheritance pattern of Huntington's disease. This disease is due to a dominant HD gene on chromosome 4.

Key

	normal male		affected female
	affected male		normal female
	identical twin	1, 2, 3, etc.	sequence of individuals



Pedigree chart showing the inheritance pattern of Huntington's disease

- (a) How many generations are shown?
 (b) How many females are in the pedigree?
 (c) How many males are in the second generation?
 (d) Identify three individuals who have Huntington's disease.
 (e) If 'H' represents the allele for Huntington's disease, state the genotypes of:

- (i) individuals 1 and 2 in the first generation
- (ii) individuals 2 and 4 in the second generation.

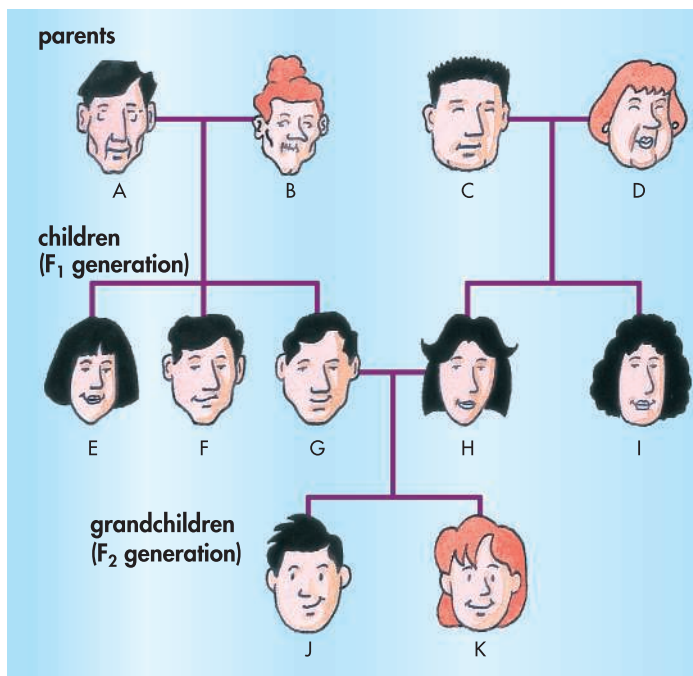
(f) How would the pattern in the pedigree be different for a recessively inherited trait?

4. (a) Using the letters 'B' or 'b' to represent the gene for coat colour, predict the results of the following crosses. Draw diagrams or Punnett squares to show your predictions.

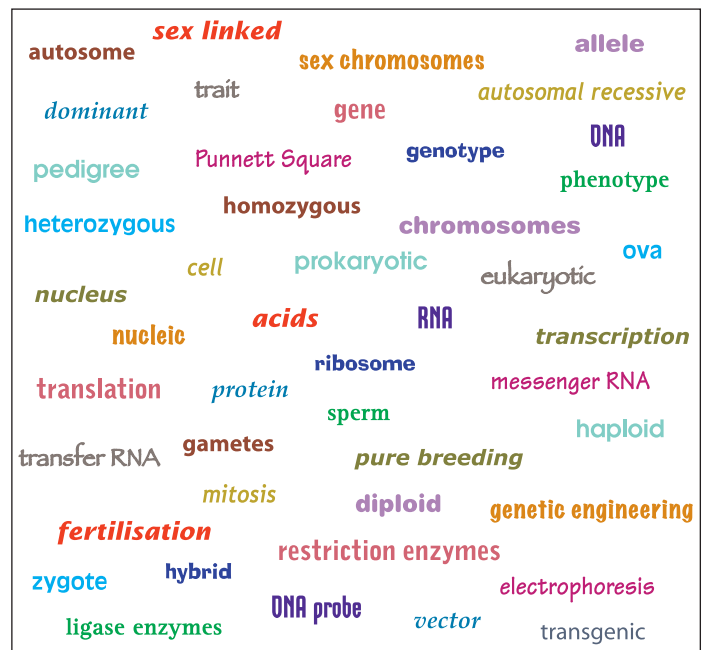
- (i) A pure breeding (homozygous) black mouse with a hybrid (heterozygous) black mouse.
- (ii) A pure breeding black mouse with a pure breeding white mouse.

(b) Is black dominant to white or white to black? Support your answer.

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



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



- 6. (a) Create a concept map that uses as many of the above terms as you can.
- (b) Select at least five terms and create a poem or song to help teach others how the terms are linked or connected. Share your creative piece with others in your class.
- 7. (a) For each of the following statements, rate your opinion on the scale shown below:

0	1	2	3
(strongly disagree)	(disagree)	(agree)	(strongly agree)

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

