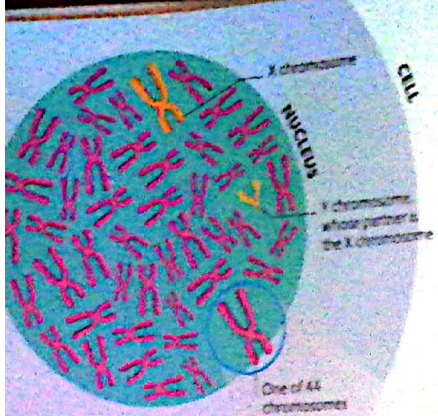
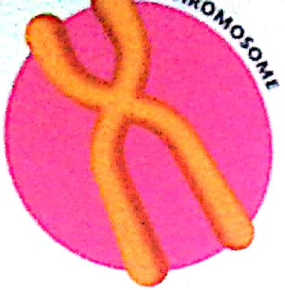
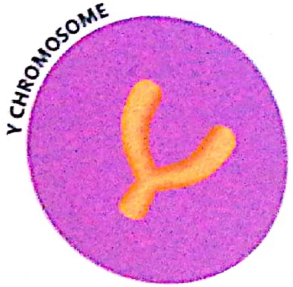
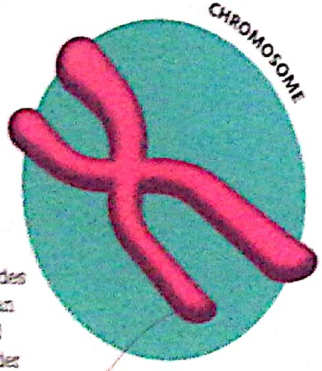


Control centre
DNA is stored in the nucleus of every cell (except for red blood cells, which lose their DNA as they mature). In each cell nucleus, there are 2 m (6 ft) of DNA tightly coiled into 23 pairs of chromosomes – making a total of 46. We inherit one chromosome of each pair from our father, the other from our mother.

Boy or girl?
While 22 of our chromosome pairs are duplicates – with only a slightly different version of each gene on each chromosome – the final 23rd pairing is different. In most people, it determines our sex. Females usually have two X chromosomes, while males have an X and a Y. Only a few of the X chromosome genes are repeated on the shorter Y chromosome, which mostly carries genes that produce masculine features.





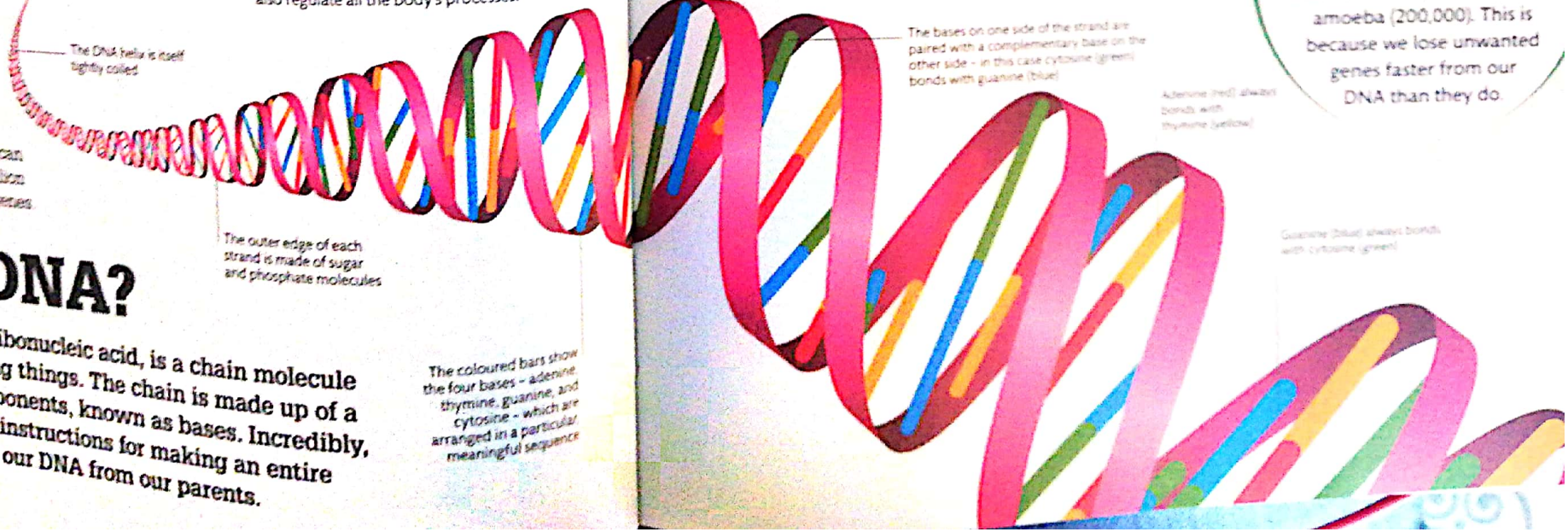


Human library
DNA is a long molecule that provides all the information necessary for an organism to develop, survive, and reproduce. It is like a twisted ladder with rungs made of a pair of chemical bases. These bases form long sequences called genes that are coded instructions for building proteins. When a cell needs to duplicate its DNA or make a new protein, the two halves of the ladder unzip so that a copy of the gene can be made. Humans have more than 3 billion bases in their DNA and nearly 20,000 genes.

Body builders
The genes that build our bodies may range from a few hundred bases to more than 2 million bases in length – longer than the small section shown here. Each gene produces a single protein. These proteins are the building blocks of the body, forming cells, tissues, and organs. They also regulate all the body's processes.

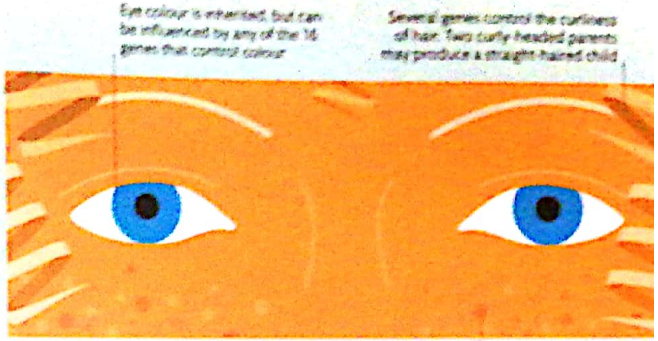
What is DNA?

DNA, also known as deoxyribonucleic acid, is a chain molecule that exists in nearly all living things. The chain is made up of a sequence of molecular components, known as bases. Incredibly, the sequence acts as coded instructions for making an entire living organism. We inherit our DNA from our parents.



Express yourself

The majority of genes are the same in everybody because they code for molecules that are essential for life. However, around 1 per cent have slight variations – known as alleles – that give us our unique physical characteristics. While many of these are harmless traits, such as hair or eye colour, they may also result in more problematic conditions, such as haemophilia or cystic fibrosis. Because alleles come in pairs, one may override the effect of the other so that the trait stays hidden.



Unpredictable outcomes
Many of our physical features are under the control of more than one gene. This may result in unexpected combinations.

Unravelling DNA

Chromosomes help package DNA to fit into the nucleus. The DNA is wrapped around spool-like proteins that run through the centre of each chromosome. The helix is made of two strands of sugar phosphate linked together by a pair of bases. The bases always form the same pairs, but the sequences of bases along the strand are specific to the proteins they will eventually produce.

DO HUMANS HAVE THE MOST GENES?

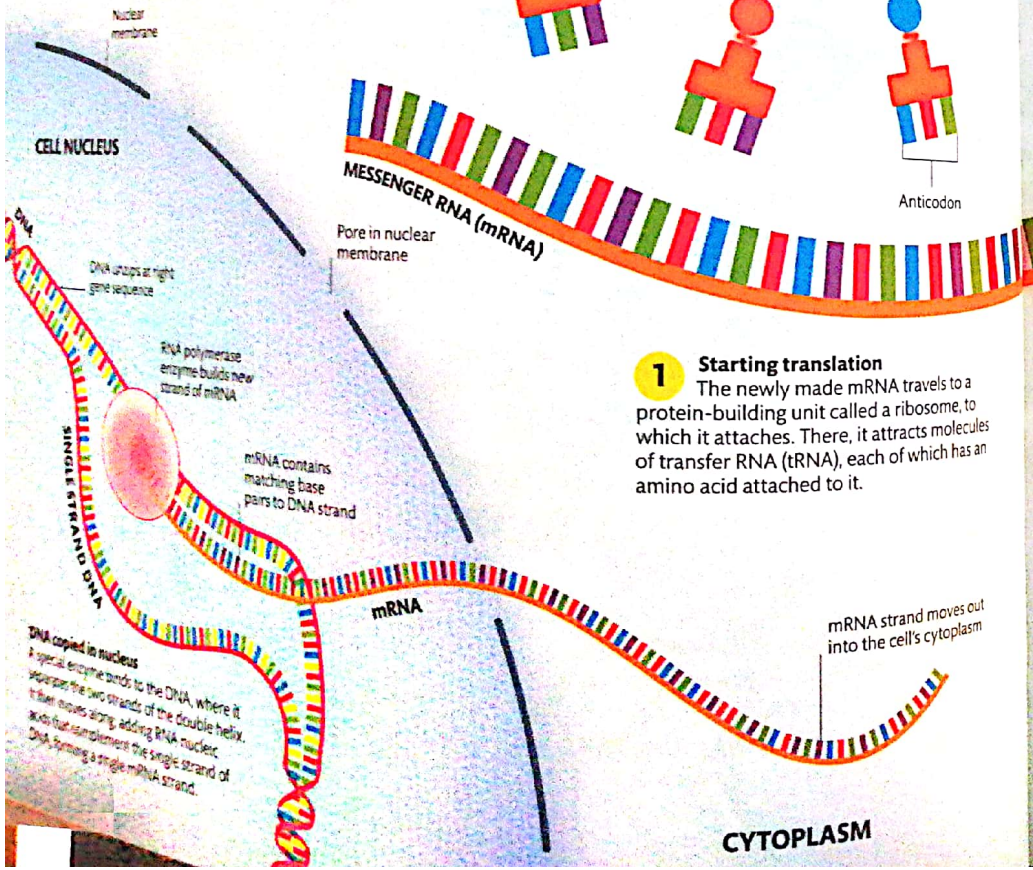
Humans have a relatively low number of genes. We have more than a chicken (16,000) but fewer than an onion (100,000) or an amoeba (200,000). This is because we lose unwanted genes faster from our DNA than they do.

How genes work

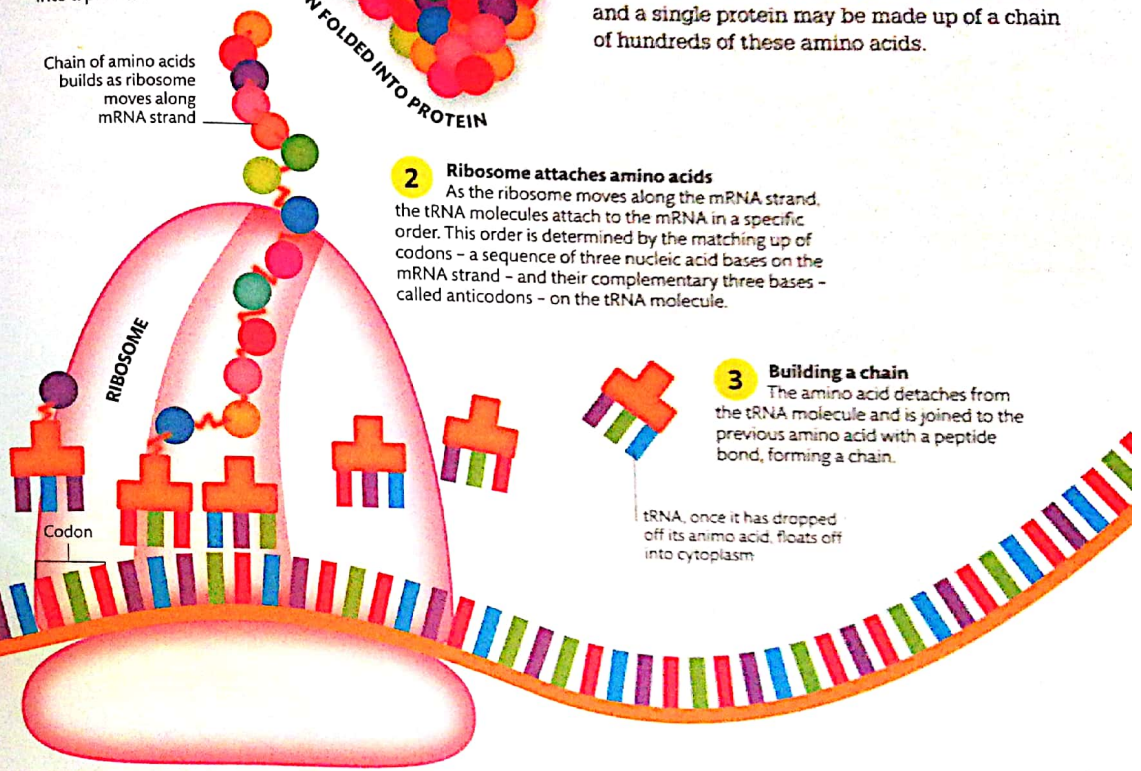
If our DNA is the body's recipe book, then a gene within that DNA is equivalent to a single recipe in the book; it is the instructions for building a single chemical or protein. It's estimated that humans have around 20,000 genes that code for different proteins.

Genetic blueprint

To translate a gene into a protein, the DNA is first copied (transcribed) in the nucleus of a cell by enzymes, forming a strand of messenger RNA (mRNA). The cell will only copy those genes that it needs, not the entire DNA sequence. The mRNA then travels outside the nucleus, into the cell's cytoplasm, where it can be translated into a chain of amino acids, which will build the protein.



1 Starting translation
The newly made mRNA travels to a protein-building unit called a ribosome, to which it attaches. There, it attracts molecules of transfer RNA (tRNA), each of which has an amino acid attached to it.



2 Ribosome attaches amino acids
As the ribosome moves along the mRNA strand, the tRNA molecules attach to the mRNA in a specific order. This order is determined by the matching up of codons – a sequence of three nucleic acid bases on the mRNA strand – and their complementary three bases – called anticodons – on the tRNA molecule.

3 Building a chain
The amino acid detaches from the tRNA molecule and is joined to the previous amino acid with a peptide bond, forming a chain.

4 Amino acids folded into proteins
When the ribosome reaches a stop codon at the end of the mRNA strand, the long chain of amino acids is complete. The order of the amino acids determines how the chain folds up into a protein.

Making proteins

Every three bases in the mRNA is known as a codon and each codon specifies a particular amino acid. There are 21 different amino acids and a single protein may be made up of a chain of hundreds of these amino acids.

LOST IN TRANSLATION

Gene mutations can cause changes in the amino acid sequence. A single mutation in the 402nd base of the gene that codes for the hair protein keratin causes the amino acid lysine to be put in place of glutamate. This changes the shape of the keratin, making the hair look beaded.

WHAT HAPPENS TO mRNA AFTER TRANSLATION?

A strand of mRNA may translated into a protein times before it eventu degrades within the c

How genes make different cells

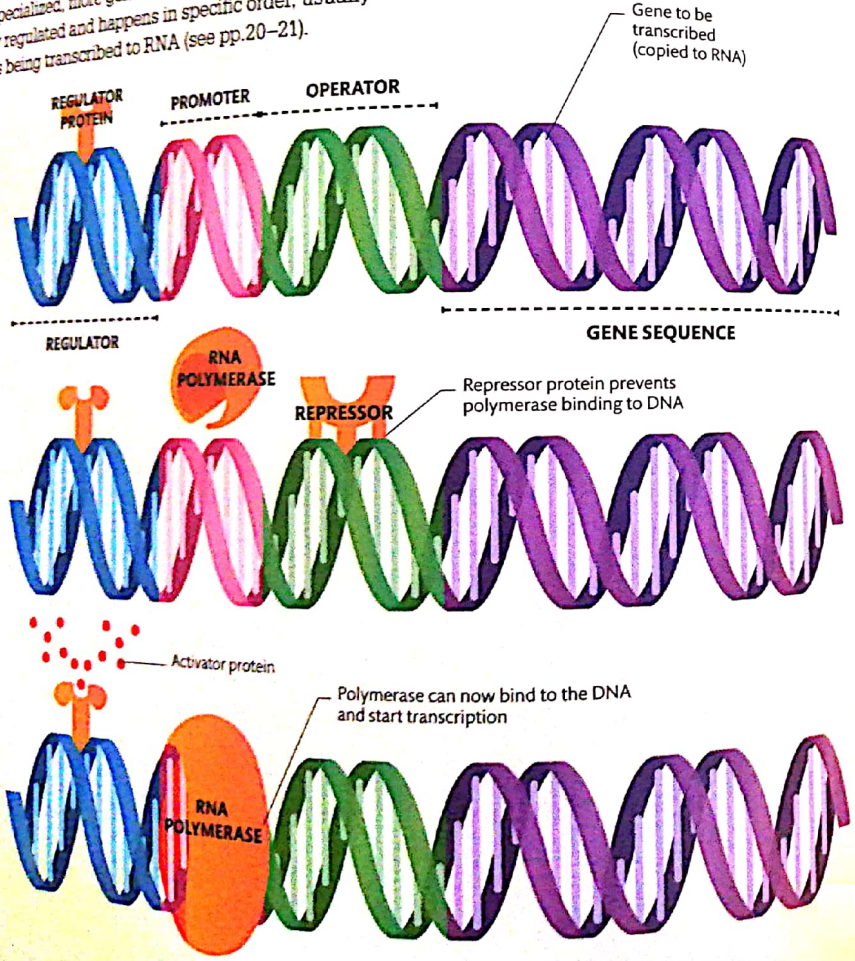
DNA contains all of the blueprints for life, but cells pick and choose only the plans (genes) they need. These genes are used by the cell to build the proteins and molecules that not only define what the cell looks like, but what it does within the body.

HOW DO CELLS KNOW WHAT TO DO?

The chemical environment around the cell, or signals from other cells, tell it that it is part of a particular tissue or organ, or in a certain stage of development.

Gene expression

Each cell uses, or "expresses", only a fraction of its genes. As it becomes more specialized, more genes are switched off. This process is highly regulated and happens in specific order, usually when the DNA is being transcribed to RNA (see pp.20–21).



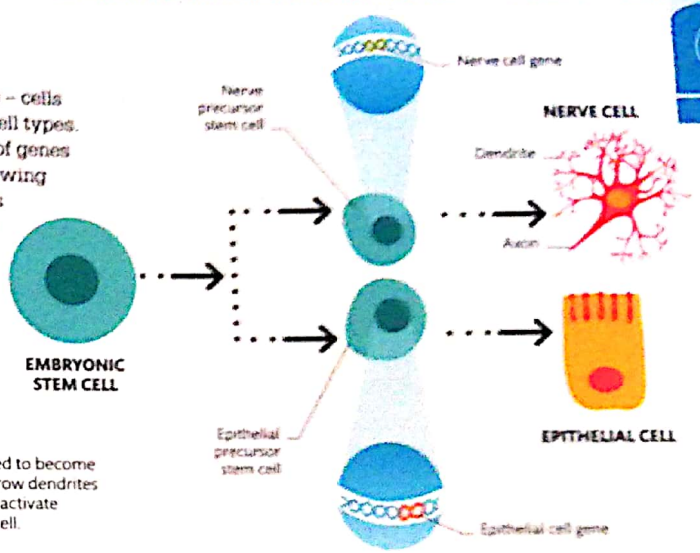
1 Regulation
Transcription of a required gene is controlled by a series of genes that sit in front of it. These include regulator, promoter, and operator genes. The gene won't be transcribed until conditions are right.

2 Repressor protein
If a repressor protein is blocking the gene, transcription can't take place. The gene can only be turned on when a change in the environment removes the repressor protein.

3 Activation
When an activator protein binds to the regulator protein and there are no repressor proteins blocking the gene, transcription can start.

On or off?

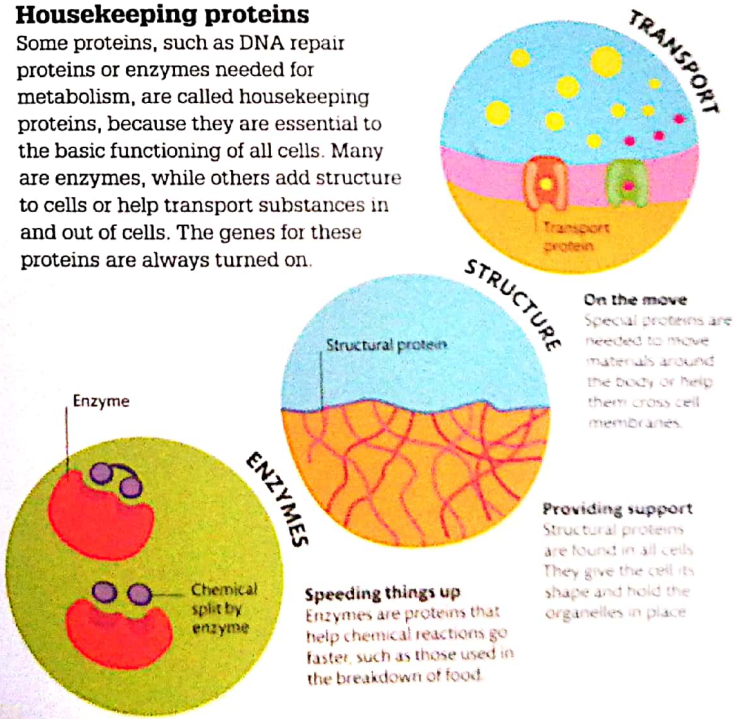
Embryonic cells start out as stem cells – cells with the ability to turn into different cell types. Stem cells initially have the same set of genes switched on and they simply keep growing and dividing to produce more cells. As an embryo develops, it needs its cells to specialize and organize into tissues and eventually organs. So when signalled, the cells start shutting off some genes and switching on others to turn into a specific type of cell.



Making a difference
As an embryo is developing, a stem cell destined to become a nerve cell will turn on the genes needed to grow dendrites and an axon, whereas another stem cell might activate different genes to become an epithelial (skin) cell.

Housekeeping proteins

Some proteins, such as DNA repair proteins or enzymes needed for metabolism, are called housekeeping proteins, because they are essential to the basic functioning of all cells. Many are enzymes, while others add structure to cells or help transport substances in and out of cells. The genes for these proteins are always turned on.



On the move
Special proteins are needed to move materials around the body or help them cross cell membranes.

Providing support
Structural proteins are found in all cells. They give the cell its shape and hold the organelles in place.

Speeding things up
Enzymes are proteins that help chemical reactions go faster, such as those used in the breakdown of food.

BOY OR GIRL?

At 6 weeks, an embryo has all the internal organs needed to be either male or female. If it is genetically a male embryo, a gene on the Y chromosome will turn on at this stage and produce the hormones that develop the male reproductive organs and cause the female organs to degenerate. The reason why men have seemingly pointless nipples is that these are also formed in the first 6 weeks, but their further development depends on whether they are in a male or female hormonal environment.

Environmental assault
Each of our cells is inundated daily by chemicals and energy that can cause damage to our DNA. Solar radiation (UV), environmental toxins, and even the chemicals produced through our own cellular processes can cause changes to our DNA that affect how it works, including how it can be copied or how it produces proteins. If this damage becomes a permanent change in the DNA, it is called a mutation.

20,000
THE NUMBER OF **DAMAGED BASES REMOVED** AND **REPLACED** IN EVERY CELL **EVERY DAY**

CAN THE DAMAGE ALWAYS BE REPAIRED?
Our ability to repair DNA diminishes as we get older. Damage starts to accumulate and this is thought to be one of the main reasons behind ageing.

UNDER ATTACK
This DNA strand is shown under many kinds of stress. However, some types of DNA damage can be used to advantage. Many chemotherapy drugs are designed to cause damage to the DNA in cancerous cells. Cisplatin, for example, forms crosslinks in the DNA, which triggers cell death. Unfortunately it also causes damage in normal healthy cells.

When DNA goes wrong

Every day, the DNA in cells is damaged – whether by natural processes or environmental factors. This damage can affect DNA copying or how specific genes function and if it can't be repaired, or is repaired incorrectly, it can lead to disease.

REPAIRING DNA

Cells have built-in safety systems that help to identify and repair damage to their DNA. These systems are constantly active and if they are unable to fix the damage quickly, they will stop the cell cycle temporarily so they can take some extra time to work on it. If it's not repairable, they will trigger the death of the cell by apoptosis (see p.15).

Gene therapy

When DNA damage causes a mutation, it can stop a gene from working properly and result in disease. While drugs might help treat the symptoms of the disease, they can't solve the underlying genetic problem. Gene therapy is an experimental method that's exploring ways to fix the defective gene.