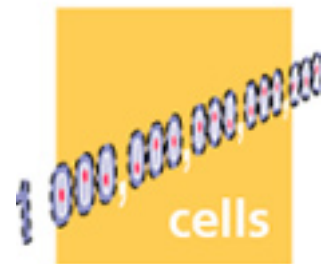


DNA, genes and genomes - General

Genomes

What is a genome?

Your body is made up of about one hundred, million, million cells (100,000,000,000,000). Each of these cells has a complete set of instructions about how to make your cells, their components and their components' components. This set of instructions is your genome.



Your genome is quite similar to everyone else's genome, which is why we all turn out to be human beings. But every other living thing also has a genome.



All living things have genomes. Beetles each have a beetle genome, which is a complete set of instructions for making a beetle. Cabbages have a cabbage genome and bacteria have a bacteria genome.



Extinct species, such as dinosaurs also had their own genomes.

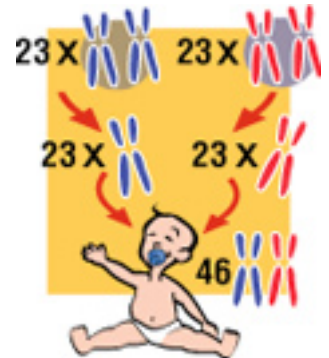


DNA, genes and genomes - General

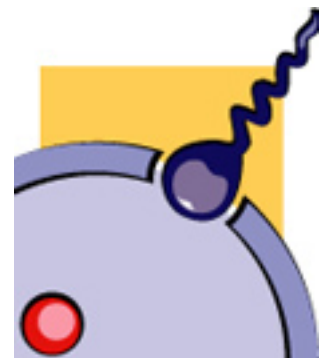
Genomes

Where do genomes come from?

We actually have two genomes each. We get one copy of our genome from each of our parents. A sperm cell (from the father) has only one copy of a genome and an egg cell (from the mother) also only has one copy. The genome in each sperm or egg cell is a mixture of the father's or mother's two genomes.



At the moment of fertilisation, the sperm cell and the egg cell join together to make a cell containing two genomes. The fertilized egg then has a complete set of instructions to make a new person.



DNA, genes and genomes - General

DNA

What are genomes made of?

Your genome is made of a chemical called DNA. The letters of DNA stand for **d**eoxyribo**n**ucleic **a**cid but that's not really important. What is important is the unique shape of DNA.



Why is DNA's shape important?

DNA is shaped like a twisted ladder. Imagine a ladder made of rubber. If you held the bottom of the ladder firmly and twisted the top, the shape you would create is the same shape that your DNA has. Researchers call this shape a 'double helix'.

The rungs on the DNA ladder are called base pairs: these do the important job of being the instructions. These base pairs can break, allowing the sides of the helix to unravel (roll your mouse pointer over the image). This special property is what allows DNA to copy itself, and to act as the instructions.

DNA, genes and genomes - General

Comparing genomes

How much DNA?

DNA is, of course, much smaller than a ladder. We measure DNA's dimensions in thousands of millionths of metres, known as 'nanometres'! To put these tiny measurements into perspective: in each of your cells, you have about 6,000,000,000 'rungs' of DNA. That means if the base pairs were as far apart as the rungs on a real ladder, then the DNA from just one cell would stretch half way to the moon!

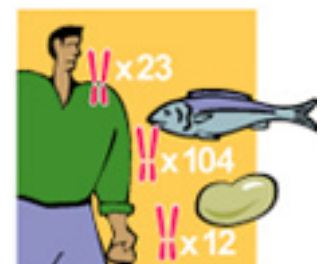


All living things have DNA, packaged into their cells. In tiny organisms like bacteria, the DNA is coiled into tiny circular packages. In plants and animals, DNA is tightly packaged into bundles, wrapped around a scaffold of protein. If we look at DNA under a microscope, we can sometimes see these bundles of protein and DNA, called chromosomes.



How many chromosomes?

The amount of DNA and the number of chromosomes in an organism's cells depends on the species it comes from. Humans have 46 chromosomes (23 pairs), but carp fish have 104 chromosomes (52 pairs) while broad beans have 12 chromosomes (6 pairs).



DNA, genes and genomes - General

DNA's code

What is DNA's alphabet?

We use codes everyday; alphabets are also codes. Let's take the word "koala". In English, the letters 'k', 'o', 'a', 'l' and 'a' in that particular order mean an animal that lives in Australia and eats eucalyptus leaves. If you didn't know any English, you wouldn't be able to guess what the word means from the letters that are in it. The letters 'k', 'o', 'a', and 'l' appear in lots of other words where they don't mean anything to do with koalas. Different languages use different alphabets to convey meaning.



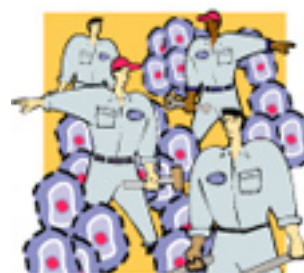
DNA's code is written in only four 'letters', called A, C, T and G. The meaning of this code lies in the sequence of the letters A, T, C and G in the same way that the meaning of a word lies in the sequence of alphabet letters. Your cells read the DNA sequence to make chemicals that your body needs to survive.



What does DNA code for?

A gene is a length of DNA that contains the instructions to make a chemical in your body. The DNA in a gene usually codes for a protein.

In our cells, proteins are the workforce; they get everything done. Proteins break down our food to release energy. Proteins organise the transport of useful chemicals between cells. Often, these useful chemicals are themselves proteins.



DNA, genes and genomes - General

DNA's code

What does DNA code for?

As well as doing things, proteins are the building blocks for most of your body. In the same way that a wall is made mostly of bricks, your body is made mostly of protein.

We talk about genes having different characteristics. For instance, if you hear about 'genes for eye colour', it means that these genes code for protein pigments in the iris of each of our eyes. Genes can come in different versions. Some people's versions code for proteins that make their eyes look blue while other people's versions make proteins that make their eyes look brown.



DNA, genes and genomes - General

Making proteins

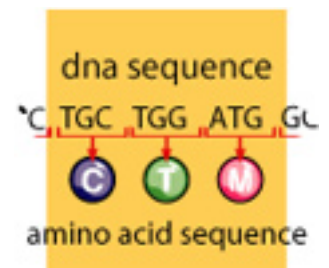
What are proteins made of?

The ingredients of a protein are amino acids. To build a protein we need to build a long chain of amino acids. There are 20 different types of amino acids, so there are lots of different protein chains we can build. Biologists give amino acids a code letter, as for DNA. This is much easier than writing out the whole name each time. For example, M is methionine, L is leucine, F is phenylalanine (because P is proline).

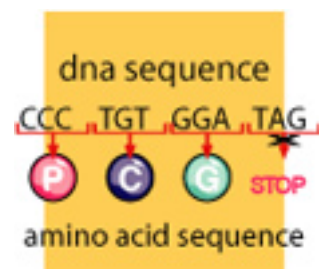


Three DNA letters, one amino acid?

The DNA code uses groups of three 'letters' to make meaning. This means that when the cell reads the instructions encoded in the DNA sequence to make a protein, it reads it three letters at a time. Most groups of three letters - known as triplets or codons - code for an amino acid.



Since there are four different DNA letters (A, G, C and T), there are $4 \times 4 \times 4 = 64$ different combinations that can be used. However, as there are only 20 different types of amino acid, some of these 64 codons code for the same amino acid. Some of the 64 codons don't code for any of the amino acids. Instead they provide the punctuation and grammar, like where the cell should start and stop reading the sequence.



DNA, genes and genomes - General

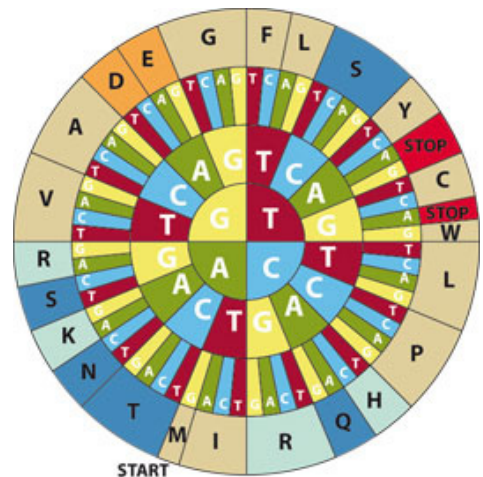
Making proteins

An Example:

An example of a DNA sequence might be

CCCTGTGGAGCCACACCCTAG

If you use our DNA decoder (codon wheel), you can decode this triplet by triplet (you can get a copy from our downloads section). Start from the inside of the wheel: find the first letter of your codon in the centre of the wheel and work outwards, through the second ring (with the next letter) and so on, to find the corresponding amino acid.



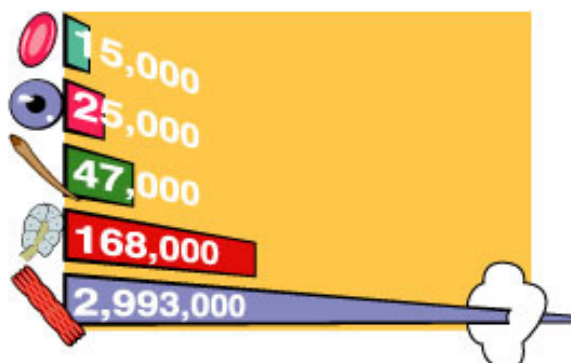
This would make the amino acid chain:

P - C - G - A - T - P

Proline-Cysteine-Glycine-Alanine-Threonine-Proline

We talk about genes having different characteristics. For instance, if you hear about 'genes for eye colour', it means that these genes code for protein pigments in the iris of each of our eyes. Genes can come in different versions. Some people's versions code for proteins that make their eyes look blue while other people's versions make proteins that make their eyes look brown.

Most proteins are actually much longer than this. Many of the proteins that make your body makes contain hundreds of amino acids. So if we know what the DNA sequence is, we can work out which amino acids the protein must contain and in what order.



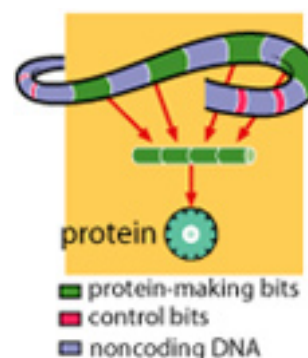
DNA, genes and genomes - General

Genes and 'junk'

What does the rest of the DNA do?

Most of the DNA sequence in our genome is not used to make protein; amazingly only 2% of it is! The rest of the DNA is made up of the same letters as coding DNA but it doesn't have the same meaning.

Quite simply, we don't know what some of our genome does. The bits of DNA we don't understand have often been called 'junk DNA'. However, the more we learn about what's in the 'junk', the more it seems better to call it 'noncoding DNA' instead.



Controls and repeats

Some noncoding sequences enable our cells to produce different amounts of proteins at different times. For example, control sequences contain instructions to tell the cell how to switch genes on and off. Other noncoding sequences are part of genes, but don't directly code for proteins. These are thought to help the cell to generate a number of different proteins from one gene.

More than half of the DNA in our genome is made up of repeated sequences. The result is as if a printer had made a mistake and scattered lots of copies of one page of a book throughout the story. Some of these repeated areas appear to stabilize the chromosomes; others may have a role in spacing out the coding sequences so that they can be activated independently.



We don't know all the answers yet, but stay tuned for the next update . . .

DNA, genes and genomes - General

Variation

Am I unique?

Yes. Although, at the level of our DNA, researchers currently think that any two human beings are more than 99% alike, there are differences between us. These differences can be small changes in a single DNA letter or duplicates and deletions of much larger chunks of DNA.



Can genomes change?

Genomes change - between generations or over a lifetime - these changes are called mutations. Mutations can be helpful, harmful or make no difference at all.

You can inherit mutations from your parents. Mutations are also happening in your cells all the time. Environmental factors like smoking and sunlight can increase the rate of DNA mutation in your cells.



Mutations can happen anywhere in the DNA - in the noncoding DNA as well as the coding DNA of our genes. Many mutations will do nothing at all because they occur outside the important coding and noncoding regions of our DNA.

At this stage, we know most about mutations that we can detect. These are the relatively few mutations that directly cause disease or an obvious change in the way our bodies work. In the future, researchers expect to learn more about mutations with small or indirect effects. For example, small changes that switch a gene on at the wrong time, or mean that not quite enough protein is produced, could contribute to complex diseases like diabetes or hypertension.

DNA, genes and genomes - General

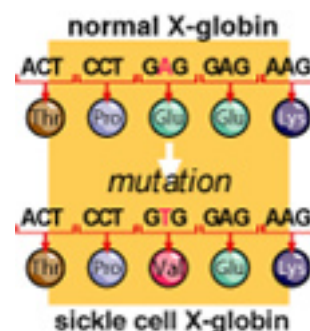
Variation

How can mutations cause disease?

Sometimes, one of the DNA letters is accidentally swapped for another letter. This mutation might have a very serious effect, or none at all.

If the letter T is swapped for an A in the codon GCT then the protein will still be the same, since both the old codon (GCT) and the new codon (GCA) code for the amino acid Alanine.

However, swapping an A for a T in a gene for haemoglobin causes a serious disease called sickle cell anaemia. Haemoglobin does the job of carrying oxygen around the body in the blood. In people with sickle cell anaemia, the sequence of amino acids in haemoglobin is different, and so it doesn't work as well.



There are other types of mutations as well. Sometimes, a bit of the DNA sequence is missed out by mistake, or a new bit added in. Sometimes, parts of the sequence are swapped over, even between different chromosomes.

Inheriting mutations

Each of our genes is a copy from either our mum or our dad. If there is a mutation in one of these genes, this can be passed on from parent to child along with the rest of the gene. This is why diseases can run in families.

Small inherited changes can make big differences in our bodies. For example, the most common mutation to cause cystic fibrosis - a disorder where a person's internal organs become clogged with thick mucus - is the loss of three letters in a gene called CFTR.