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FAQ: Genetics

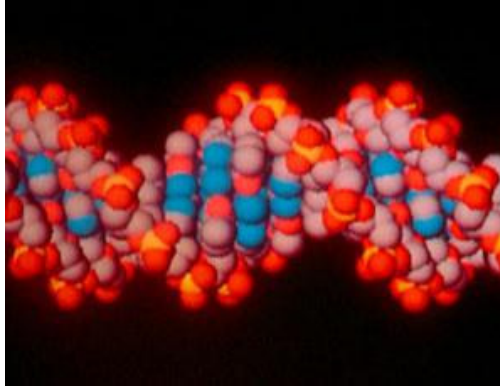
11:53 04 September 2006 by [Philip Cohen](#)

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1. What is DNA?

DNA is the storehouse of genetic information for every known organism, with the exception of a few viruses. It's a long, thin molecule - picture two strands that curve around each other, forming a double helix. Each strand spells out the genetic code as a chain of four chemical letters called bases: adenine (A), thymine (T), cytosine (C) and guanine (G).

Bases facing each other across two strands are always paired as follows: A with T and C with G. When cells duplicate their DNA, the two strands of the helix are "unzipped" and enzymes use them as a template to create a new version of the opposite strand.



Computer graphic of part of the DNA molecule, showing its twisted double helix structure (Image: Dr A Lesk, Laboratory of Molecular Biology / SPL)

2. What is a gene?

This is not a simple question.

For decades, biologists like the late Francis Crick - co-discoverer of DNA's structure - confidently proclaimed that genes were regions of DNA that served as blueprints for proteins through a simple process: DNA is copied into the related chemical RNA, which then is whisked away to the cell's protein manufacturing facility. Crick famously dubbed this definition of the gene the "central dogma of molecular biology".

A gene's protein sequence is spelled out as a series of three letter "words" - codons - composed of the four DNA bases. The codon "GGG" for instance, encodes the amino acid glycine.

Regions of DNA that do not produce proteins were therefore generally dismissed as functionless "junk DNA". But it turns out that Crick's dogma may have been a bit too, well, dogmatic.

Insights from the sequencing of the human genome have led some experts to argue that the purpose of many human genes is not to encode protein, but to spin out RNAs that serve many functions beyond that of middleman between DNA and protein. And the evolutionary conservation of some types of junk DNA suggests they serve important, if unknown, functions.

3. How do genes create organisms?

Genes are not always active. Sometimes they are busy churning out their encoded protein or RNA, sometimes they are shut off completely. And their activity can be tuned at different levels. During the development of a complex organism from a single cell, thousands of genes flash on and off in complicated patterns.

One of the most important jobs genes have is to create proteins called transcription factors, which coordinate the activities of other genes. As an eye or a finger is created, for example, transcription factors ensure that a characteristic series of genes get activated in surrounding tissue to build that structure.

Proteins and structures they create can also serve many other functions: generating energy,

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creating new molecules or serving directly as the brick and mortar of structures like muscle. Genes also shape organisms by driving the replication, movement, activity and death of cells.

4. What can go wrong with the process?

The genetic code is so precise that even a change in a single DNA base can have profound effects. The mutation which causes the disease sickle cell anaemia, for example, was tracked down to the substitution of a T for an A in the gene for the protein haemoglobin, which carries oxygen in red blood cells. As a result, a single protein building block called an amino acid is changed, resulting in a crippled protein.

Sometimes the problem is not the gene sequence, but the location or number of genes. Whole regions of chromosomes can be missing or duplicated, resulting in missing genes or inappropriate activity. Cancer cells, for instance, often have the wrong number of entire chromosomes.

5. How are genes inherited?

Our genes and the 23 pairs of chromosomes they reside on are inherited, with one of each pair coming from each parent. This means that sperm and eggs must contain half the number of chromosomes of any other cell in the body. Otherwise when sperm and egg fused to form an embryo it would contain twice the number of chromosomes needed.

Sperm and eggs receive their half-portion of genes through a chromosomal choreography called meiosis. First the 23 parental pairs of chromosomes match up at the centre of the sex cell. When the cell divides, each daughter receives only one half of each pair. Since this process is random, it generates a staggering 70 trillion possible combinations of chromosomes in the offspring.

In fact, the true degree of possible variation is higher because the maternal and paternal chromosomes exchange DNA when they pair, creating new gene combinations within the chromosomes. So rest assured of your genetic uniqueness - unless you are an identical twin.

6. What other factors control how our genes work?

It seems reasonable that if two genes with the same sequence are in the same cell, they should act the same way. But that is not always true. So-called epigenetic factors can alter how a gene works regardless of its DNA sequence.

One well studied example is parental imprinting. Certain genes are marked with chemical tags via a process called methylation while they are still in a sperm or egg, meaning that only the maternal or paternal copy is active in the offspring. As a result, certain traits are inherited exclusively from one side of the family.

There is also some evidence that environment can influence epigenetic factors. For example, Dutch women who were pregnant during the famines of the World War II gave birth to small babies. But, surprisingly, the next generation also spawned small babies even though they ate well, as if they "inherited" their mother's experience.

7. Do genes control everything about an organism, or is environment important?

The debate over the relative importance of nature and nurture would fill several encyclopedias, but modern genetics predicts that both should play a role. That should come as no surprise to anyone who views genes as a piece of cellular machinery. Dangerous chemicals, such as cigarette smoke, can jam that machinery or interfere in its workings.

Equally, a therapeutic environment can compensate for a faulty gene. For example, babies who are born with the disease phenylketonuria (PKU) lack an enzyme that metabolises the amino acid phenylalanine. It therefore builds up to toxic levels causing mental retardation. But babies are now screened for the defect at birth and those with two copies of the defective gene are given special diets low in phenylalanine. As a result, they develop normally.

8. How genetically similar are we to primates and other organisms?

Chimpanzee genes differ, on average, by roughly just 1% from human genes. Other apes' genes are 95% to 98% identical to ours, too. Rodent genes are 88% identical and chickens come in at 75% identical.

Once you leave the animal kingdom, wholesale comparisons between human genes and those of other species becomes trickier. About one-third of the genome of the fruit fly *Drosophila melanogaster*, for example, contains genes that are only shared by other arthropods, and one-quarter of human genes are shared only by vertebrates.

The function of some genes in flies, plants or worms appear close enough to their human counterparts that these animals can serve as models to study human biology and disease.

9. Most traits are controlled by a complex array of genes. But which human features depend entirely on single genes?

You already know some of your single gene traits like the back of your hand. Specific versions of different single genes cause: hair growth on the middle segments of the fingers, the top of the little finger to bend dramatically to the ring finger, and determine whether the left thumb crosses over the right - or vice versa - when fingers are interlocked.

The result of other single gene traits are as plain as the eyes, ears, and hair on your face. People with blue eyes, non-dangly ear lobes or a straight hairline have inherited specific gene varieties. The ability to roll your tongue into a tube and also to taste certain bitter chemicals is also conferred by certain types of single genes.

Defective versions of single genes can also cause disease such as cystic fibrosis, sickle cell anaemia and Huntington's disease.

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