

What is genomics?

Put simply, "genomics" is the study of all of the genes within an organism.

In contrast, genetics looks at specific genes.

Genomic researchers determine the specific sequence of DNA, which contains the complete set of genetic instructions for that organism e.g. genes to determine hair colour, height, skin colour etc. [1]

It's all in the DNA

Virtually every single cell in the body contains approximately **3 billion DNA base pairs** which make up the human genome. These base pairs act like letters of the alphabet to spell out the organism's genetic makeup.[1] In living organisms the genome is stored in the **nucleus** of each cell in thread-like structures called **chromosomes**. Human cells contain 46 single chromosomes (or 23 pairs). Each chromosome is made up of tightly coiled **deoxyribonucleic acid (DNA)** molecules (Figure 1).

The DNA bases are **adenine (A)**, **thymine (T)**, **guanine (G)** and **cytosine (C)**. Bases on opposite strands of the DNA pair specifically: an A always pairs with a T; a C always pairs with a G. The order of the As, Ts, Cs and Gs determines the meaning of the information encoded in that part of the DNA molecule just as the order of letters determines the meaning of a word.[3]

What are genes and proteins?

A **gene** refers to the unit of DNA that carries the instructions for making a specific **protein** or set of proteins. Located on 23 pairs of chromosomes packed into the nucleus of a human cell, genes direct the production of proteins with the assistance of enzymes and messenger molecules.[3]

Did you know?

The first draft of the human genome was published in Nature 20 years ago. Today using high-performance technology scientists can analyse enormous amounts of DNA much more rapidly than ever before.

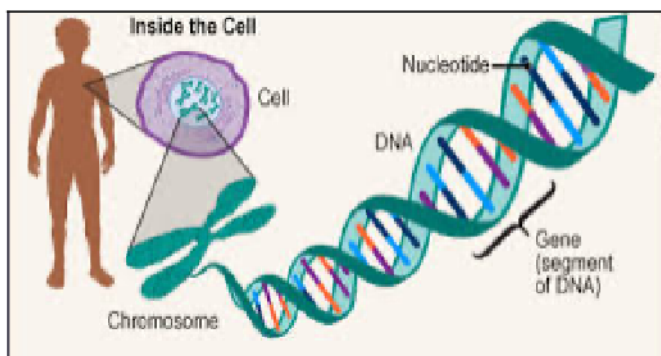


Figure 1. DNA found in the cell nucleus is arranged into chromosomes, composed of double stranded DNA

How do genes code for proteins?

Specifically, an enzyme copies the information in a gene's DNA into a molecule called **messenger ribonucleic acid (mRNA)**.^[3] The mRNA travels out of the nucleus and into the cell's cytoplasm, where the mRNA is read by a tiny molecular structure called a ribosome, and the information is used to link together small molecules called amino acids in the right order to form a specific protein (Figure 2).^[3]

Why are proteins important?

Organs and tissues of living things are made up of proteins. Proteins also control the chemical reactions and signals between the organism's cells. If a cell's DNA is mutated (i.e. changed or altered), an abnormal protein may be produced, which can disrupt the usual processes and lead to diseases such as cancer.^[3]

Modern technology advancing genomics

Modern technology advances such as CRISPR-Cas-based approaches, machine learning and big data analysis is making genomics easier and more economical.^[4]

Scientists predict that in the near future "genomics will be everywhere".^[4] Genomics has the potential to enable medical scientists to better predict, diagnose and treat diseases earlier and more precisely and personally than ever before.^[6]

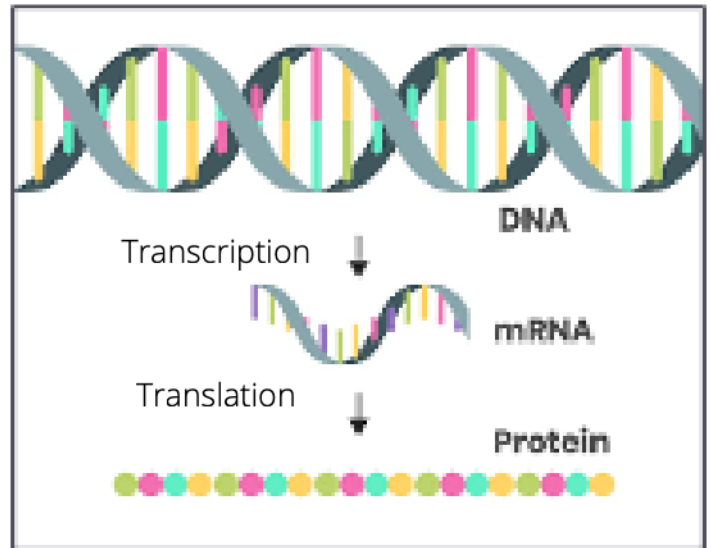


Figure 2. DNA, RNA and protein

Potential benefits of genomics:

- Rapid drug discovery
- Improved disease risk assessment
- Personalising drug therapy
- Individualised drug dosing

“ Understanding our own complete code and being able to manipulate it is the ultimate evolutionary milestone in 14 billion years of evolution” ^[4]

“ In the future we'll see every cancer patient sequenced, and we'll develop specific drugs to target their particular genetic alteration.