Australian Government
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The human genome

The human genome consists of the complete set of human genetic material that is contained in a human cell. In most human cells, the genetic material is made up of long DNA strands that are packaged into 23 pairs of chromosomes. In contrast, eggs and sperm have 23 unpaired chromosomes that parents pass on to their offspring. The offspring then inherit one copy of each chromosome from each parent and this means that siblings have, on average, about half of their DNA in common. More distant relatives have less DNA in common. For example, on average, first cousins have in common about one eighth (12.5%) of their genetic material.

Humans have two kinds of chromosomes: sex chromosomes (X and Y) and autosomes. Of the 23 paired human chromosomes, 22 are autosomes and one is the sex chromosome. The paired sex chromosome in females are two X chromosomes, whereas males have one X and one Y chromosome.

Each chromosome is an organised structure that contains DNA. DNA contains the instructions for building different parts of the cell and body. The instructions are in the form of a chemical code (the genetic code), made up of sequences of four building blocks known as nucleotide bases. The four bases are always paired in DNA molecules, with adenine (A) always paired with thymine (T) and guanine (G) always paired with cytosine (C) to make base pairs.

The human genome inherited from each parent is made up of over 3 billion DNA base pairs. Genes are formed from DNA base pairs that are arranged in sequences and instruct the cell to build the proteins that make up the human body. The genes also contain the coding regions of the human genome that are known collectively as the exome. There are approximately 20,000 genes in humans and these represent only 1–2% of the human genome.

What is the relationship between DNA mutations and genetic conditions?

A genetic disease or condition is caused by one or more genetic changes, which scientists refer to as mutations. A mutation is a permanent change in the DNA code. Inherited mutations are those that are passed on to

children from a parent and are called germ line mutations. Some of these mutations occur spontaneously, for unknown reasons.

Mutations also build up in a person's DNA over their lifetime (for example, DNA damage due to sun exposure). These mutations are not passed on to children and are called somatic mutations.

Changes in the DNA sequence do not always lead to health problems because some mutations occur in DNA that is not a part of a gene. Even so, studying these types of mutations can be useful in other types of genetic research.

Why is genetic testing a potentially powerful tool in medicine?

Mutations in inherited genes can result in genetic diseases or conditions that may cause problems at any stage of life, depending on the type of mutation. On the other hand, some genetic changes do not cause health problems for the individual but may cause health problems for their children.

Genetic testing examines the genes of an individual and looks for mutations. This information can be used to work out the future possibility of a disease or condition developing for that person or the future risk of a disease or condition in that person's children.

Many diseases are thought to be genetic in nature. However, genetic tests are usually only recommended when there is a history of a genetic health problem in an individual or the family.

Blood relatives share regions of their DNA with each other. If one member of a family is found to have a genetic mutation, other members of the family may have the same mutation. If the mutation causes health problems, then the health of other family members could be at risk. Doctors usually encourage people to share this type of information with their family so that other members can seek medical advice.