Genetic conditions account for many of the health and development conditions seen at birth, in childhood, adolescence and adulthood.

IN SUMMARY

- Genetic conditions can be seen from birth or develop during childhood, adolescence or adulthood
- There are a number of different causes of genetic conditions and they vary in the range of symptoms seen
- Genetic conditions can affect more than one person in a family and may be passed down the generations of a family.

A genetic condition can be linked to health, growth or developmental concerns that are present from birth, or they may not be noticed until childhood, adolescence or adulthood.

There are thousands of known health conditions caused by variation in one of the 20,000 or so gene pairs in the human cell, and hundreds of conditions due to a difference in the usual number or structure of the chromosomes.

There are a growing number of genetic conditions being identified that are due to an interaction between a genetic susceptibility (**predisposition**) and environmental factors or other influences on the expression of the genes (**epigenetics**). Many of these conditions are common health concerns.

A number of genetic conditions occur more frequently in some population groups and in people with a particular ancestry.

CHROMOSOMES, GENES AND DNA

Our bodies are made up of billions of cells. Each cell contains a complete copy of a person's genes.

Chromosomes can be thought of as being made up of strings of **genes** (DNA that codes for proteins) with non-coding DNA between them. The chromosomes, including the genes, are made up of a chemical substance called DNA (**D**eoxyribo**N**ucleic **A**cid) and are found in the nucleus of the cell.

Apart from the DNA found on chromosomes, another place in the cell where DNA is found is in very small compartments called **mitochondria** (the energy centres of the cell).

In each human cell, except the egg and sperm cells, there are usually 46 chromosomes. Chromosomes are found in pairs and each pair varies in size. Therefore there are 23 pairs of chromosomes, one of each pair being inherited from each parent.

There are over 20,000 gene pairs found in the DNA of each person. Each gene has its own specific location or address in the DNA, and the genes (coding DNA) plus the non-coding DNA make up that person's **genome**.

People generally have the same number and sequence of genes so that the same messages are sent to the body.

There can, however, be small variations between individuals in the information contained in specific DNA codes.

Members of the same family tend to be more similar in their genes than unrelated individuals.

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The genes in our DNA provide the instructions for proteins, which are the building blocks of the cells that make up our body. Although we all have variation in our genes, sometimes this can affect how our bodies grow and develop. Generally, DNA variations that have no impact on our health are called **benign variants** or **polymorphisms**. These variants tend to be more common in people. Less commonly, variations can change the gene so that it sends a different message. These changes may mean that the gene does not work properly or works in a different way that is harmful. A variation in a gene that causes a health or developmental condition is called a **pathogenic variant** or **mutation**.

Figure 5.1:
Diagram of a human cell showing nuclear DNA which is found on chromosomes in the nucleus of a cell and the mitochondrial DNA which is found in the energy centres of cells known as mitochondria.

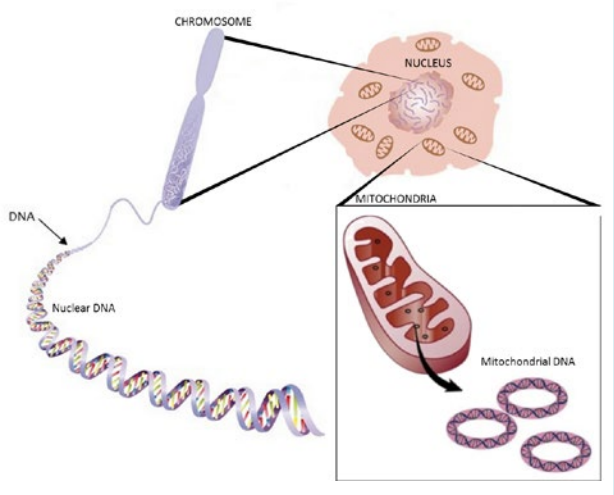

Figure adapted from the NHS National Genetics and Genomics Education Centre, https://www.genomicseducation.hee.nhs.uk/image-library

A DNA variation may cause a problem for one cell type but not another, since not all cells use all of the proteins that may be possible in the body.

When a DNA variation results in the protein not being made properly in cells that need that protein, this may result in a genetic condition.

WHAT ARE THE CAUSES OF GENETIC CONDITIONS?

Gene variants

There are two copies of each gene, one on each of the chromosome pairs located in the nucleus of the cell.

Genes are also located in the mitochondria.

A variant in one or both copies of one or more genes found in the nucleus or mitochondria of a cell can be inherited from a parent.

Variants can also happen either during the formation of the egg or sperm, or during or soon after conception.

The gene variant can be passed on to a child and may be the direct cause of a genetic condition.

Some examples of genetic conditions caused directly by a gene variant include cystic fibrosis, Huntington disease and haemophilia.

Chromosome changes

It is important that the chromosome balance in the cells of our body is right so that the right amount of DNA and genes is present.

Changes in the number or structure of chromosomes can be inherited from a parent who has the chromosome change in their cells.

Chromosome changes can also happen during the formation of the egg or sperm or during, or soon after conception.

Some examples of genetic conditions caused by a chromosome change include Down syndrome (where there is an extra copy of chromosome number 21) and Turner syndrome (where females have only one X chromosome instead of the typical pair).

Epigenetics

The term epigenetics comes from the words **epi-** meaning upon or over and **genetics** meaning our genes.

The way a cell reads the DNA message is controlled in a number of ways and one of these ways is by adding tags, like post-it notes to the DNA bases, or structures that DNA wraps around to change the instructions within a gene. Sometimes these tags give messages to switch on the gene and create the protein, while others stop the protein from being created and switch off the gene.

These tags are not permanent and can change quite a lot over time. There are a number of different types of tags or ways in which the DNA messages are controlled.

Epigenetic changes are the cause of some genetic conditions.

WHAT CAN BE DONE ABOUT GENETIC CONDITIONS?

Prevention and early detection

For some common conditions knowing about it early and finding ways to stop or slow down signs and symptoms can make a difference. This may be relevant to people with a family history of those conditions.

In some genetic conditions, early diagnosis, sometimes even before the symptoms appear, can lead to specific treatment. For example, all newborn babies in Australasia are screened for a condition called phenylketonuria (PKU) by a simple blood test. Diagnosis and treatment within the first month of life in a baby is important to prevent learning disability.

In some cases of breast cancer, bowel cancer, melanoma and prostate cancer, it may be possible to test for the genetic cause for cancer in a family. This can then be used to help find cancers early enough to treat, in people with a higher chance of developing cancer.

Genetic Counselling

Genetic counselling is available to families and people who have concerns about a condition in their family that may have a genetic basis.

A team of health professionals, such as clinical geneticists, other medical specialists and genetic counsellors, work together with other health care providers to give information and support so that families can better understand and adjust to the diagnosis of a genetic condition.

If appropriate, genetic testing can also be organised on the basis of informed consent.

Genetics services are available throughout Australasia and provide genetic counselling to help with informed decision making regarding genetic testing.

Support Groups

Support groups provide affected people and families with information and community resources about the condition in their family, as well as an understanding and empathic ear. There are many support groups and some of these are specific to a particular genetic condition.