

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

### In summary

- **Genetic conditions can be seen from birth or develop during childhood, adolescence and 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 through the generations and your family.**

Genetic conditions can cause many of the health, growth or developmental problems that are present from birth or they may not be noticed until childhood, adolescence or adulthood.

There are over 6,000 known conditions due to an inherited variation in just one of the 20,000 or so gene pairs in the human cell and hundreds of syndromes due to a difference in the usual number or structure of the chromosomes.

There is also 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 problems.

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

### DNA, GENES AND CHROMOSOMES IN THE BODY

Our bodies are made up of millions 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 46 chromosomes. Chromosomes are found in pairs and each pair varies in size. Thus there are 23 pairs of chromosomes, one of each pair being inherited from each parent.

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

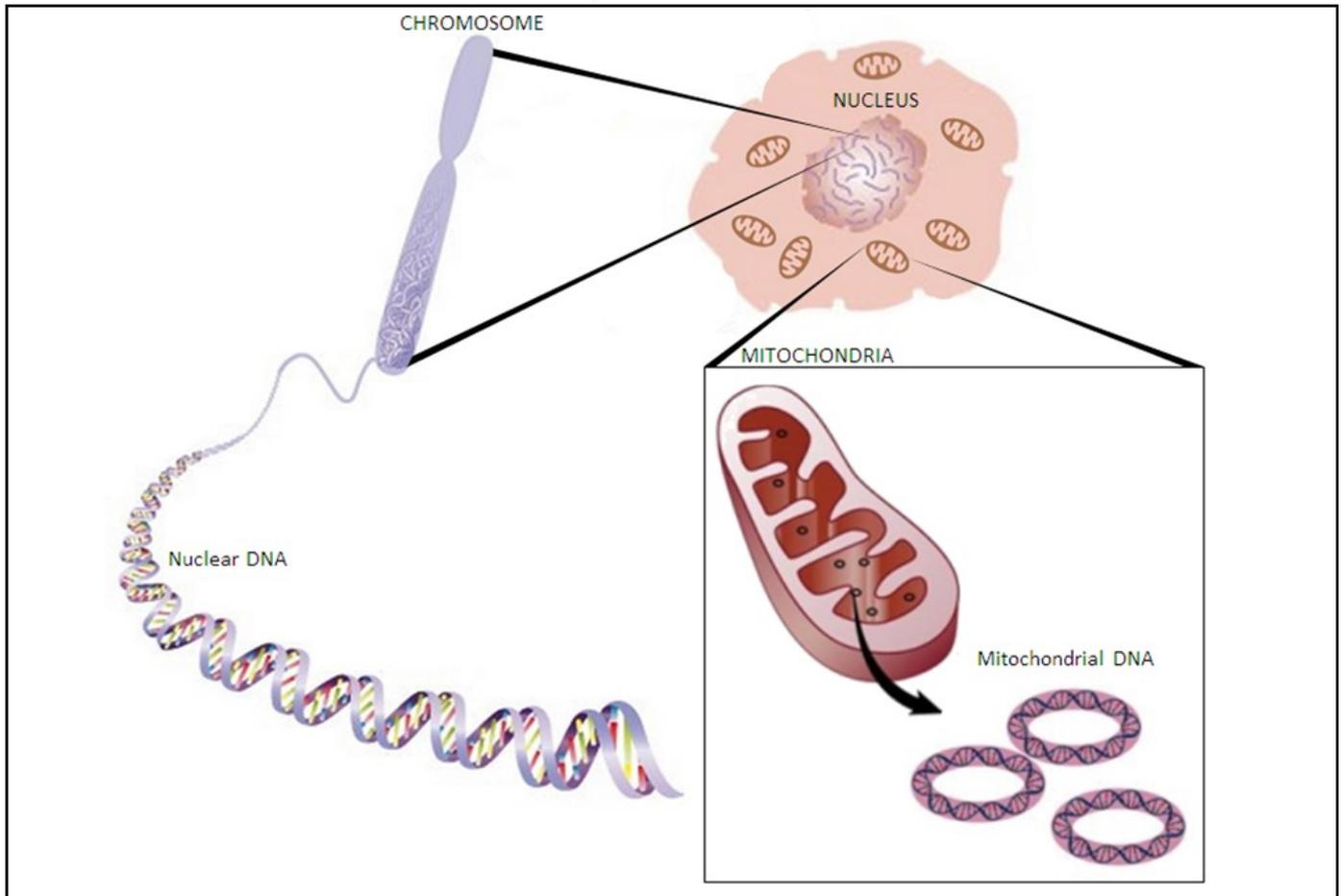
People generally have the same number and sequence of genes so that the same messages are sent to the body.

However, there can 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.

Most DNA variations do not have any impact on our health and are called **neutral variants** or **polymorphisms** (*poly* means many; *morphisms* means forms).

Other DNA variations may be associated with an increased risk of a health condition, for example diabetes or cancer.



**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

Some DNA changes can mean the instructions are incorrect so a faulty protein is made or the control switch is changed. A variation in a gene that creates a fault is called a **pathogenic variant** or **mutation**. These are quite rare.

A DNA mutation may cause a problem for one cell type but not another, since not all cells use all of the possible proteins.

When a DNA change causes a faulty protein in cells that need that protein, it usually results in a disease state or symptom and is often recognised as a genetic condition.

**WHAT ARE THE CAUSES OF GENETIC CONDITIONS?**

**Gene mutations**

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 mutation 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.

Mutations can also occur either during the formation of the egg or sperm or during or soon after conception.

The gene mutation 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 mutation include cystic fibrosis, Huntington disease and haemophilia.

**Chromosomal changes**

It is important that the chromosome balance in the cells of our body is correct in order for the correct amount of DNA and genes to be present.

Changes in the number or structure of chromosomes can be inherited from a parent who has the chromosomal change in their cells. Chromosomal changes can also occur 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 usual two copies).

### **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 post-it notes give messages to activate the gene and create the protein, while others stop the protein from being created.

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.

This is a very simple way of explaining the reason why some genes can be affected by something in addition to the DNA in a cell. Epigenetic factors are the cause of some genetic conditions.

### **WHAT CAN BE DONE ABOUT GENETIC CONDITIONS?**

#### **Prevention and early detection**

For some common conditions, prevention and early detection strategies are available. This is usually relevant for people with a family history of those conditions caused by the interaction of environmental factors with their inherited genetic information.

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 are crucial to avoid intellectual disability.

Also, for some cases of breast cancer, bowel cancer, melanoma and prostate cancer, it may be possible to determine the presence of the gene mutation in a family and adopt strategies to detect the cancer early enough to enable treatment to take place.

### **Genetic Counselling**

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

A team of health professionals which may include clinical geneticists, other medical specialists, genetic counsellors and social workers, work together to provide information and supportive counselling so that families may be better able to understand, and adjust to, the diagnosis of a genetic condition.

Genetic testing, if it is available and appropriate, can also be organised on the basis of informed consent. Genetics Services are available throughout Australasia and provide genetic counselling to assist in informed decision making regarding genetic testing.

### **Support Groups**

Support groups provide affected individuals and families with information about the condition and community resources, as well as an understanding and empathic ear. There are many support groups for specific conditions and also umbrella support groups for those rarer conditions which may not have a specific support group.