

DNA stands for deoxyribonucleic acid and forms the “instruction book” of our bodies. It is made up of very long chains of four basic building blocks called bases. The bases are named **A**denine (A), **G**uanine (G), **T**hymine (T) and **C**ytosine (C). Like a book, there can be changes in the “words”. Some changes are harmless but others can affect our health.

### In summary

- **DNA provides the instructions for the cells that make up our body**
- **Everyone’s DNA is somewhat different; variations in our DNA make us unique**
- **Some DNA variations are inherited from our parent/s, some appear from birth while others are acquired throughout life**
- **DNA variations that have no adverse effects on our cells and occur frequently in the population are called *polymorphisms***
- **DNA variations that do affect the function of the protein made from a gene and occur less often are called *mutations*.**

### DNA AND GENES

Our DNA is made up of sections that code for proteins, our genes, and sections that do not code for proteins. Proteins do the work in our cells and these are very important for normal cell function and our health. Our protein coding DNA, our genes, do not contain much variation. Sections of our DNA that do not code for protein contain much more variation. We do not understand all the functions of these non-protein coding sections of DNA, but we know sections can be important for controlling our genes.

People generally have the same number and sequence of protein coding 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.

An example of this is eye colour. Everyone has two genes that determine eye colour, but changes in these genes can make eye colour vary, this is why some people have blue eyes, some brown, some hazel. The same gene can send different messages because of variations in the messages.

Similarly, there may be small variations in our DNA that may affect how our bodies grow and develop.

Generally these variants do not have any impact on our health and are called **neutral variants** or **polymorphisms** (*poly* means many; *morphisms* means forms). Polymorphisms are quite common in the DNA code.

### DNA VARIATIONS

Variations in the DNA code can occur during our life for a variety of reasons including exposure to radiation, certain chemicals, by chance or other unknown factors. Ageing is one of the most common causes of genetic variation. Throughout our life, cells grow, divide and die. This means cells and their DNA are copied over and over again. As we age, our cells make mistakes which may not be corrected as efficiently as in younger people. DNA changes accumulate and can have an impact on our health.

Some DNA changes do not seem to make any difference to the way a cell functions. These types of changes are probably quite common.

Other DNA changes may be associated with an increased chance of developing a health condition, for example diabetes or cancer.

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 DNA site that creates a fault is called a mutation. These are quite rare.

A DNA mutation can 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.

### GENE MUTATIONS

Everyone is born with several gene mutations in their 20,000 or so total number of gene pairs. Depending on other factors, these mutations may or may not cause any health problems.

Most of the time, mutations cause no problem because genes come in pairs. This means that even when one gene copy has a mutation, the other gene copy is working and can still send the right message to the body. These are known as **recessive gene mutations**.

In some cases however, it is essential to have both gene copies working correctly as the amount of gene product may be critical. So in these cases, even though only one copy of the gene has a mutation, and the other is working, a health problem may still occur. These are known as **dominant gene mutations**.

Other mutations make a person susceptible to particular conditions but they will never develop the problem unless they are exposed to particular environmental triggers. These are known as **multifactorial or susceptibility gene mutations**.

In some cases, having a particular change in your DNA can be beneficial to a person. For example, in regards to the condition called thalassaemia, research has shown that people who have one copy of their thalassaemia gene working and the other with a mutation are less likely to develop malaria.

### INHERITED, SPORADIC AND ACQUIRED MUTATIONS

Mutations can occur in a number of ways in a person. They can be **inherited** from one or both parents, they can occur for the first time in a person for no apparent reason (**sporadic**) or they can come about in the body over time (**acquired**).

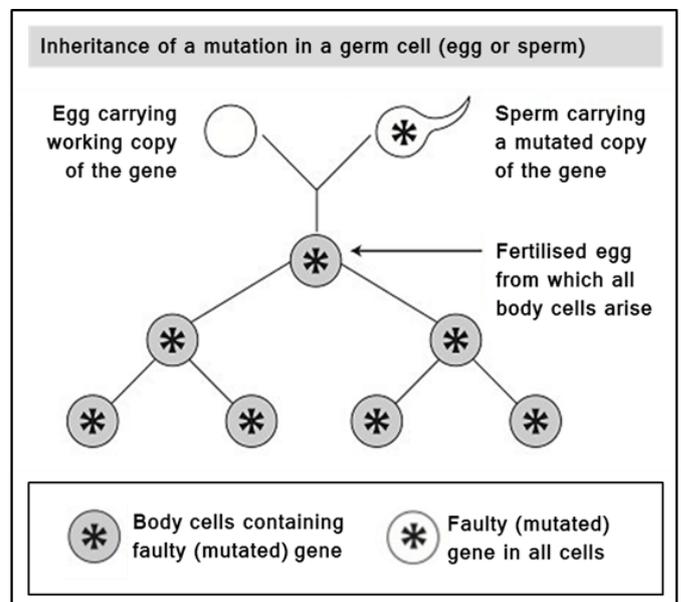
### Inherited

As mentioned above, everyone is born with several gene mutations in their 20,000 or so total number of gene pairs. These are usually passed down in the father's sperm or the mother's egg. Once that child has the mutation, they will have it for life and can also pass it on to their future children. This is an **inherited mutation** (see *Figure 2.1*).

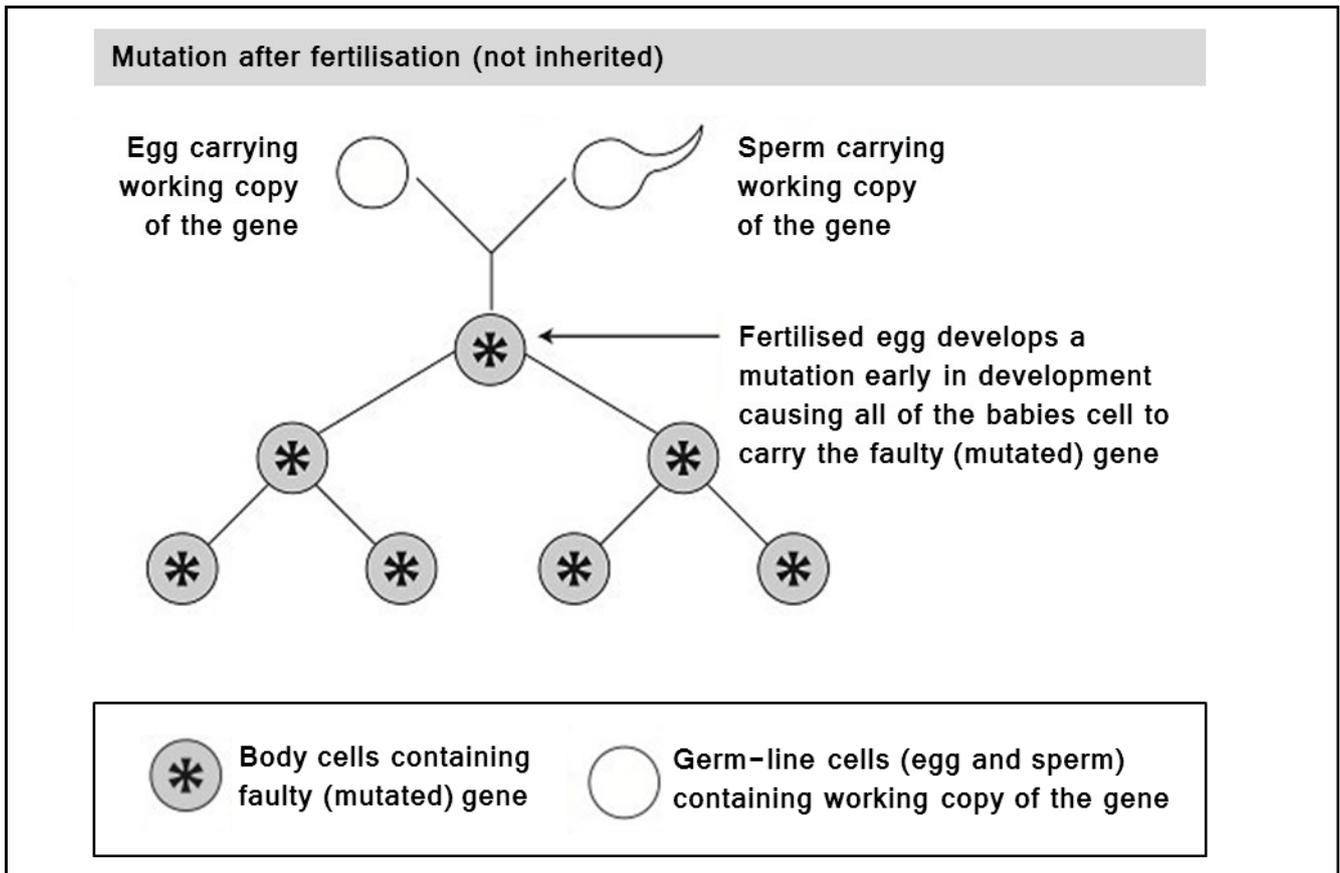
*Figure 2.1* shows a mutation being passed down through the sperm only. Inherited mutations can be passed down by only the father, only the mother or by both parents. The way in which the mutation/s will affect the child will depend on other factors as well as the mutation/s themselves.

Sperm and egg cells are called 'germ cells' and therefore mutations that are present in the genes of the egg or sperm are called germ cell or germline mutations.

If you have inherited a gene mutation from one or both of your parents, you will also possibly pass it on to your future children



**Figure 2.1:** Inheritance of a mutation from a germ cell (egg or sperm). The egg (from the mother) is carrying a working copy of the gene. The sperm (from the father) is carrying a faulty copy of the gene. Their offspring will have both a faulty copy and a working copy of the gene in all the cells of their body. This may also occur in the reverse situation.



**Figure 2.2:** Mutations can occur very early during fertilisation of the egg and sperm. The mutation is not inherited but all future cells of the baby have the mutation.

### Sporadic

When a health condition is caused by a gene mutation that appears for the first time in a family, it is said to be **sporadic**. Sporadic genetic conditions come about when a **spontaneous** mutation occurs at the time of fertilisation. Although it is not inherited from one of your parents, you can still pass it on to your future children. Your sperm or eggs will now contain this mutation. (see *Figure 2.2*).

### Acquired

DNA changes can come about due to things we experience during our life. For example, radiation produced by the sun, certain chemicals, or other environmental factors.

DNA changes may also occur as our cells are copied to enable us to grow or repair damaged cells throughout life as we age.

Mutations that occur in body cells (somatic mutations)

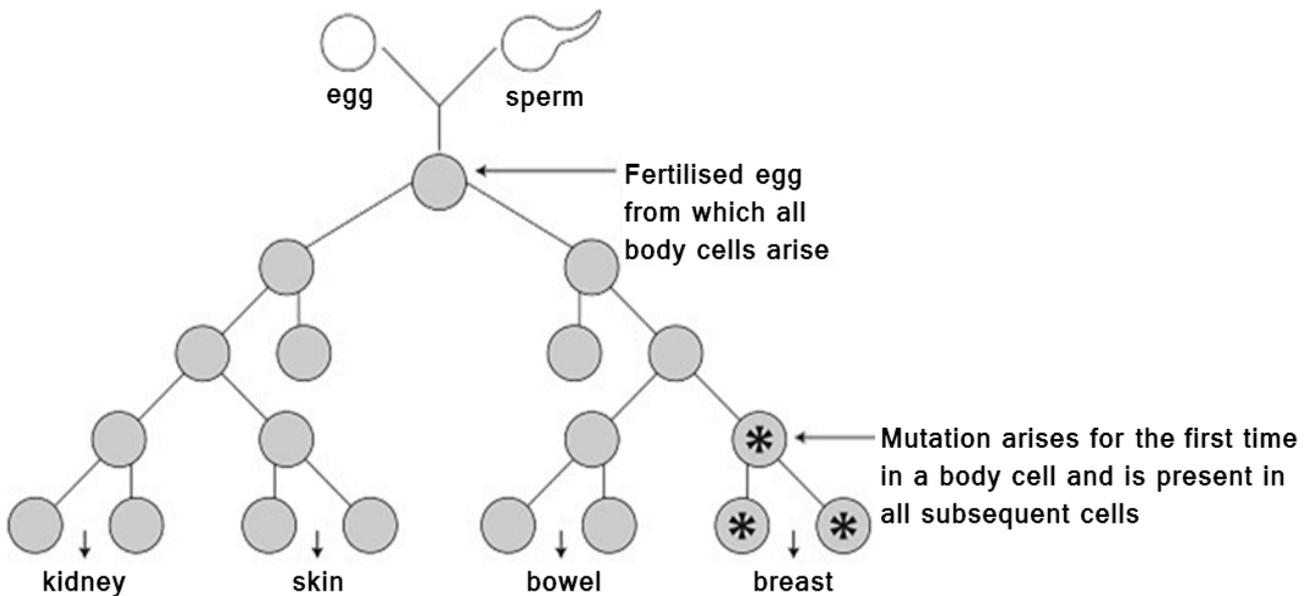


Figure 2.3: Mutations in somatic cells may only affect one specific area such as the breast tissue.

Although the body has an efficient system to repair these variations in the DNA as they occur, sometimes there is a breakdown in the cell's repair system.

If a variation to the DNA occurs and is not repaired, it will be copied into all the cells arising from that cell (see Figure 2.3).

As with other DNA variations, if the gene message becomes faulty, a mutation is said to have occurred.

Body cells (not the egg or sperm) are called 'somatic cells' and therefore a mutation that occurs in a somatic cell is called a **somatic mutation**.

In Figure 2.3, the somatic mutation has arisen in a gene in a cell of the breast tissue only and so cannot be passed on to a child.

A somatic mutation will only affect the part of the body in that particular individual. Somatic cells are therefore not passed down to children or inherited.