

This fact sheet describes mosaicism which refers to the situation where individuals have a mixture of cells in their bodies containing different genetic information.

In summary

- **Mosaicism means that a person has a mixture of cells with different genetic information**
- **Mosaicism can occur in the cells in one part of the body (such as in the egg or sperm) or in a generalised way throughout all of a person's cells.**

Our bodies are made up of millions of cells. Each cell contains a complete copy of our genetic information or DNA. Our DNA contains the instructions for growth and development and is packaged into chromosomes that contain all our genes.

A variation in a gene that creates a fault is called a **pathogenic variant** or **mutation**. Genes are sections of DNA that code for the proteins our body needs to function.

A mutation in a gene will affect the body differently depending on how much it changes the resulting protein, how critical that protein is to the body and how much of that protein is needed in the body.

A CLOSER LOOK AT MOSAICISM

Most individuals have the same amount of DNA with the same structure in all the cells in their body, whether they are blood cells, skin cells or muscle cells. Mosaicism refers to a situation where there is different DNA in different cells of the body.

Some people with a genetic condition have some cells in the body with the right DNA structure and other cells with a DNA change.

Just as mosaic tiles on a floor have a mixture of patterns, someone who is mosaic for a DNA change will have a mixture of cells in their body.

When there is mosaicism in the egg or sperm cells, it is referred to as **gonadal** or **germline mosaicism**.

WHAT DOES IT MEAN IF YOU HAVE MOSAICISM?

All of the genes are contained in every cell but only the genes that produced proteins necessary for the cell will be switched on. For a gene mutation to cause a problem, its protein must have an impact on the cells of the tissue or organ in which it is present.

A person may have a mutation detected in their blood cells but not have the condition associated with it.

For example, a gene mutation that would usually impact on brain development may be found in the blood of a normally functioning person. This means that the blood cells contain the gene mutation but the brain cells contain only the working copy of the gene.

This person has a mixture (mosaic) of cells in his/her body. Some contain the mutation (blood cells) and some have the working copy (brain cells).

In rare cases, mosaicism may explain the variability of symptoms in people with the same genetic condition.

It is, however, impossible to study the genes in every cell in the body, and so we cannot always be certain if someone is mosaic for a gene mutation or not.

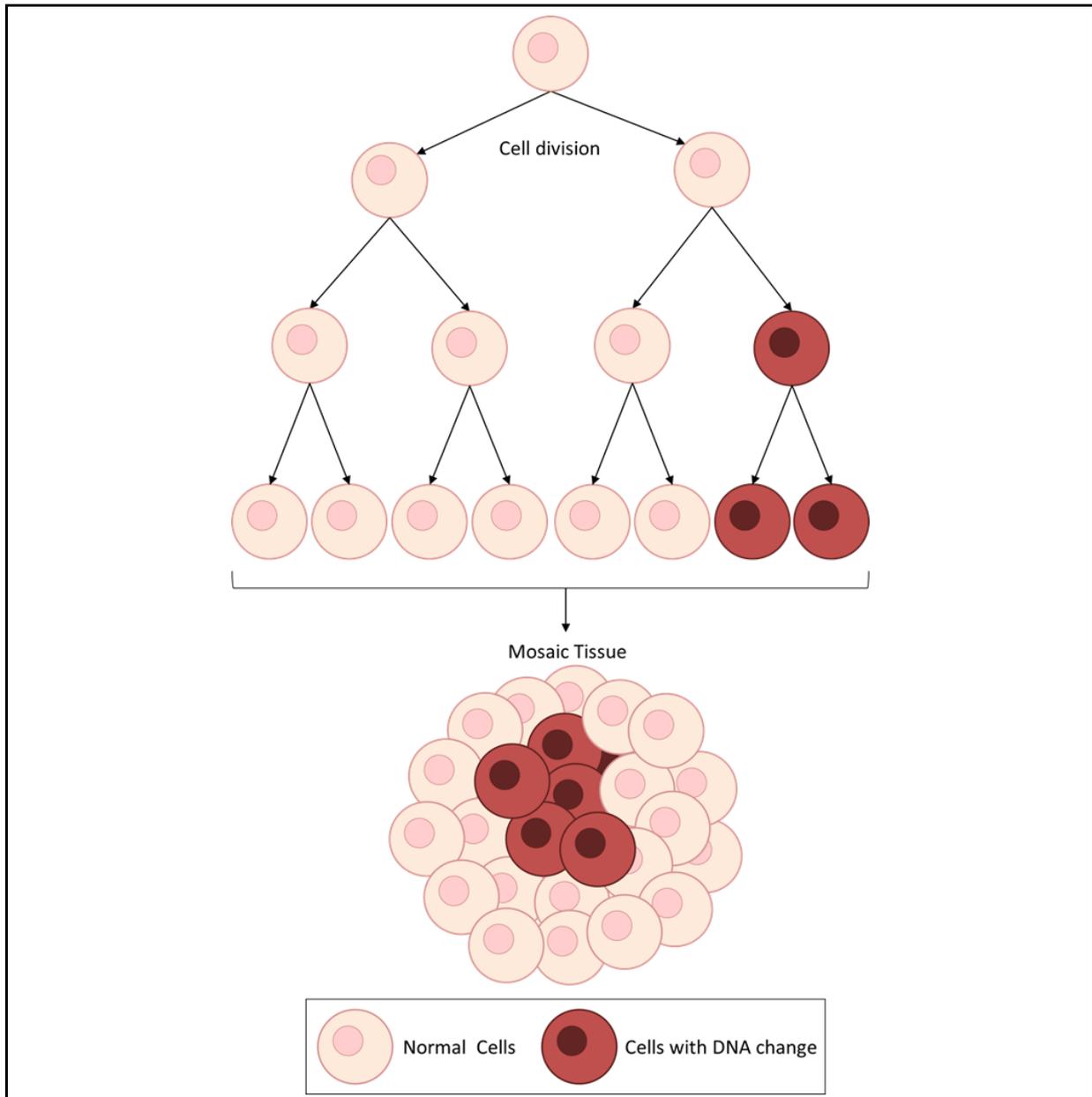


Figure 13.1: Mosaicism occurs when a cell acquires a DNA change or mutation during cell division. All cells that arise from the cell with the mutation will also carry the mutation, while other cells will contain the normal gene.

GERMLINE MOSAICISM (MOSAICISM IN SPERM CELLS AND EGG CELLS)

A DNA mutation may not be present in the body cells, but is in the germ cells (egg or sperm cells). An indication that this is possible is when a healthy couple have more than one child with a condition that is usually passed down by an affected parent (autosomal dominant inheritance).

When parents have one child with a condition that is caused by an autosomal dominant gene mutation, but neither parent has the gene mutation on a blood test, it is usually assumed

that the condition in the child occurred due to a new or spontaneous mutation in the egg or sperm, or shortly after conception.

If they have a second child with the same condition, the chance of the condition occurring again because of another spontaneous mutation in the same gene is highly unlikely.

The explanation may be that one of the parents is mosaic for the mutation in their germ cells that produce the egg or sperm, but have a working copy of the gene in their body cells.