## **Gonadal (germline) mosaicism**

This communication aid has been produced for clinicians to help support and guide conversations about gonadal (germline) mosaicism with their patients.

Our bodies are made of trillions of cells, each with a specific role. Most cells contain a nucleus that holds our genetic information – including our genes, which are the instructions for how our body works.

During the development of a baby in the womb, cells divide. Sometimes small errors can happen when copying the genetic information – like a spelling mistake. This is called a 'variant' or '**change**'. As a result, some cells will have different genetic instructions than others.

If a genetic change occurs in cells that create eggs or sperm (gametes), this will mean either some or all of that person's sperm or eggs will carry the gene change. This is known as germline mosaicism, or gonadal mosaicism.

As a result, the person's children could inherit the genetic change. The likelihood of this happening depends on how many gametes carry the genetic change. If the genetic change is inherited by a child, all the child's cells will have the change.

## **Key terms**

**Gametes:** Reproductive cells – sperm or eggs. A sperm cell fertilises an egg cell at conception of a pregnancy.

**Gonads:** Ovaries or testes – the organs that produce gametes.

**Gene change:** Changes in a gene or chromosome used to be referred to as 'mutations.' Now, they are more commonly called changes, alterations or variants.

## Want to learn more?

Scan to read or download a guide from Unique on mosaicism









## **Gonadal (germline) mosaicism**

This communication aid has been produced for clinicians to help support and guide conversations about gonadal (germline) mosaicism with their patients.







