

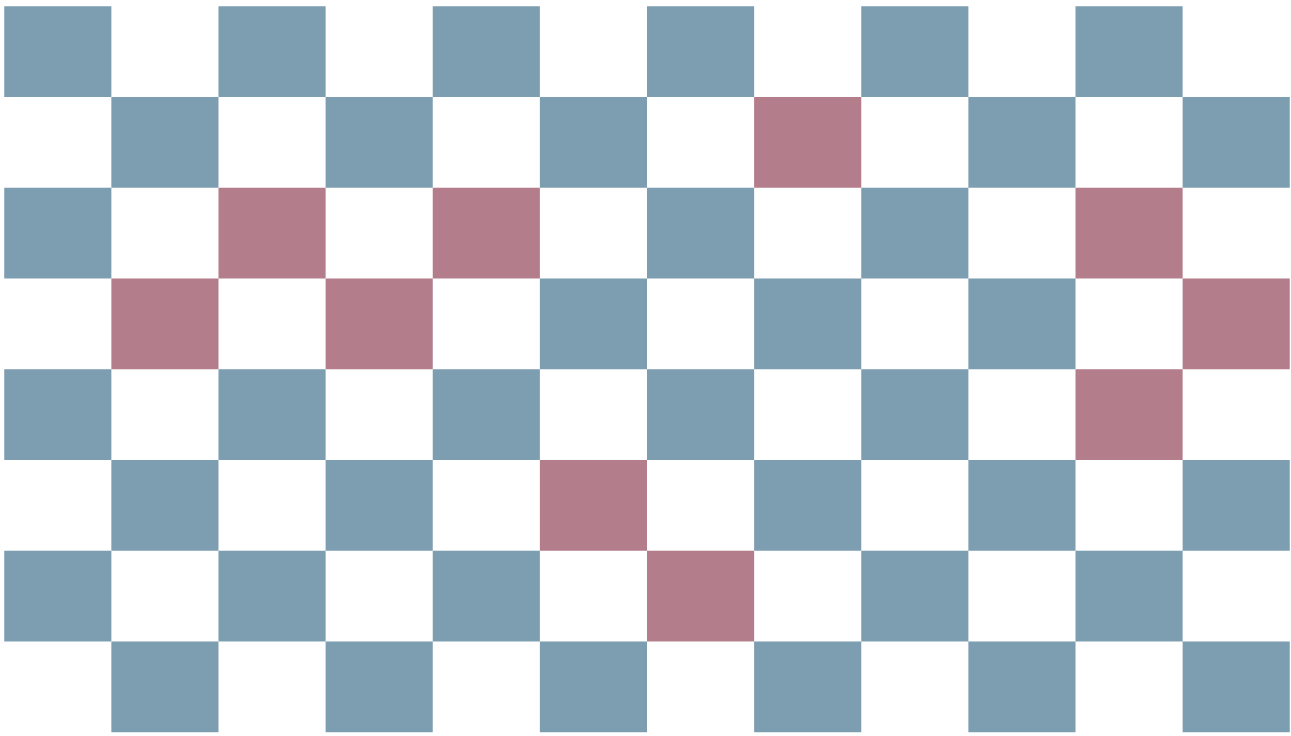


Mosaic Down Syndrome

Around 1–2% of people with Down syndrome will have mosaic Down syndrome. This means that in Australia there are approximately 120–240 people with mosaic Down syndrome.

What is mosaic Down syndrome?

Mosaicism occurs when a person has two or more genetically different sets of cells in their body. People with the more common form of Down syndrome have three copies of the additional twenty-first chromosome in each cell. However, people with mosaic Down syndrome have some cells with an additional twenty-first chromosome while other cells have two copies rather than three. Mosaicism occurs when the chromosomal variation occurs after fertilisation.



These different cell lines exist together like different coloured mosaic tiles.

How is mosaic Down syndrome diagnosed?

Usually mosaic Down syndrome is diagnosed through genetic blood tests. If non-invasive prenatal screening (NIPS) is undertaken a high chance result for a chromosomal difference may be returned. Further diagnostic testing would be required to confirm the diagnosis. In the case of a post-birth diagnosis, if Down syndrome is suspected a sample of the baby's blood can be taken for genetic testing. For other children a diagnosis may be made later in childhood when other health or developmental issues are being investigated.

What does it mean to live with mosaic Down syndrome?

People with mosaic Down syndrome are all different. Just like people with 46 or 47 chromosomes, those with a mixture of the two have a wide range of strengths, abilities and areas where they can benefit from support.

The physical features associated with Down syndrome may be less pronounced in some people with the mosaic form. Most children with mosaic Down syndrome experience delays in meeting developmental milestones. While some people with mosaicism experience less significant developmental and physical impacts than those with the more common form, this varies widely and is impacted by both the level of mosaicism and environmental factors. The majority of children with mosaic Down syndrome will benefit from Early Intervention supports such as speech therapy or occupational therapy and from the knowledge base in relation to the typical learning profile of people with Down syndrome.



Some people with mosaic Down syndrome might feel they have an invisible disability, or that it can be hard to fit in.

People with mosaic Down syndrome and their loved ones are welcomed as part of the Down syndrome community where we celebrate the differences that make us who we are. You're part of the family!

Where can I go for support?

People with mosaic Down syndrome and their families can access all supports offered by state and territory Down syndrome associations. Call **1300 881 935** to connect to your local association.

If you would like to make contact with other people with mosaic Down syndrome and their families, get in touch with the family support worker at your state or territory association.

Other resources:

Another option for connecting with others who have mosaic Down syndrome and their families is through online forums. The **Official Mosaic Down Syndrome Group** is a large closed Facebook support group run by the **International Mosaic Down Syndrome Association** with members from all over the world.

My Greta: finding a world of possibilities, is a personal reflection written by a mother following her daughter's mosaic Down syndrome diagnosis.

Disclaimer

The information in this resource is general in nature and does not constitute advice. Down Syndrome Australia will not be held responsible for any decisions made as a result of using this information. The contents of the resource do not constitute medical and legal advice and should not be relied on as such.



1300 881 935



18/71 Victoria Crescent, Abbotsford VIC 3067



info@downsyndrome.org.au



www.downsyndrome.org.au



Down Syndrome
Australia