Down syndrome

Summary

- Down syndrome is a common genetic condition caused by an extra chromosome 21.
- Some level of intellectual disability is the only feature common to all people who have Down syndrome.
- With the support and opportunities available to them today, people with Down syndrome are able to achieve and participate as valued members of the community.
- Support is available for people with Down syndrome and their families and for expectant parents who have received a diagnosis of Down syndrome for their unborn child.

What is Down syndrome?

Down syndrome (also known as trisomy 21) is a genetic condition in which the person has an extra copy of chromosome 21. This additional chromosome causes some level of intellectual disability and can cause a number of physical and developmental characteristics.

People growing up with Down syndrome have the ability to achieve and participate as valued members of their communities, with varying degrees of support. Local state or territory Down Syndrome Associations provide support for people with Down syndrome and their families, and for expectant parents who have received a diagnosis of Down syndrome for their child.

Down syndrome and the extra chromosome

Chromosomes are the blueprint for the body’s development. They are found in every cell in your body and determine your physical and mental characteristics. The usual number of chromosomes for human beings is 46 (arranged in 23 matched pairs). People with Down syndrome have an extra chromosome 21 in their genetic make-up.

Down syndrome is the most common chromosomal condition and occurs in all races and cultures at around the same rate. In Victoria, between 45 and 60 babies are born with Down syndrome each year.

Some level of intellectual disability is the only feature common to all people who have Down syndrome, although a range of other features are also associated with the condition. These include characteristic physical features and a number of health and developmental features.

Types of Down syndrome

Although we know how Down syndrome occurs, we do not yet know why. There are three forms of Down syndrome:

- trisomy 21 – where every cell in the body has an extra chromosome 21. Most people with Down syndrome (about 95 per cent) have trisomy 21. It is a condition caused at conception and is not hereditary
- mosaic Down syndrome – where there is an extra chromosome 21 in some (but not all) of the cells, while the rest of the cells have the standard genetic composition. Mosaic Down syndrome occurs in one to two per cent of people with Down syndrome. It can result in a milder level of intellectual disability and less obvious physical characteristics than the other forms of Down syndrome.
- translocation Down syndrome – where part of chromosome 21 is broken off and is then attached (translocated) onto another chromosome. This can occur before or at conception. This form of Down syndrome is uncommon, occurring in about three to four per cent of people with the condition. In about one third of these cases, the child has inherited the translocation from a parent. In such a case the family may be referred to a genetic counselling service.
Genetic tests can show what type of Down syndrome a baby has.

**Down syndrome – capabilities and potential**

Down syndrome affects a person’s development in certain ways, but it is not the most important influence on how that person develops and lives their life. People with Down syndrome demonstrate a wide range of capabilities. What happens after birth is far more important in shaping the outlook for a person with Down syndrome than the presence of the extra chromosome.

Children with Down syndrome attend childcare, kinder, primary and high school alongside other children of their age. Adults with Down syndrome attend post-school training, participate in the workforce and lead full lives as productive and valued members of the community. An increasing number of people with Down syndrome are achieving independent living, with some level of support, within the community.

People with Down syndrome often have their abilities and potential underestimated. A person with this condition will achieve in many areas when offered the opportunity and supported to do so.

**Physical characteristics of Down syndrome**

There are a number of physical characteristics associated with Down syndrome, although each person with Down syndrome may display only a few of these. The most common physical characteristics include:

- slight upward slant of the eyes – nearly all people with Down syndrome have a slight upward slant of the eyes. There can also be a small fold of skin on the inside of the eye (called an ‘epicanthic fold’) and small white patches on the edge of the iris of the eye (known as Brushfield spots)
- characteristic facial shape – the face of a person with Down syndrome is often rounded and tends to have a flat profile
- smaller stature – babies with Down syndrome are usually smaller and weigh less at birth than others. Children with Down syndrome tend to grow more slowly and are commonly smaller than other children their age. Adults with Down syndrome are commonly smaller than adults who do not have Down syndrome.

Despite some common physical characteristics, people with Down syndrome resemble other members of their family more than they resemble each other.

**Down syndrome and delayed development**

Babies with Down syndrome reach the same developmental milestones (such as smiling, sitting up, crawling, walking, talking and toileting) as all babies, but with some degree of delay. Speech and language development is often the area of greatest delay.

Everyone with Down syndrome will experience some delay in their development and some level of learning disability, but the extent and specific areas of delay vary from one person to another.

People with Down syndrome generally need more support than most other people in order to achieve their potential – some will need very little support, while others may require a high level of support. However, most people growing up with Down syndrome today will be able to achieve and participate as valued members of their communities.

**Diagnosis of Down syndrome**

Down syndrome is usually recognisable at birth and confirmed by a blood test. A doctor can usually tell if a baby has Down syndrome from their physical appearance.

Prenatal tests that can help to detect Down syndrome in a fetus include:

- **ultrasound scans**
- **maternal serum screening**
- non-invasive prenatal testing (NIPT)
- **amniocentesis**
- **chorionic villus sampling.**

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Read more information about [prenatal tests](#).

**Genetic counselling and support regarding Down syndrome**

Genetic counselling is available to parents in all states of Australia and may be helpful if screening suggests you may have a child with Down syndrome.

Genetic counsellors are health professionals qualified in both counselling and genetics. As well as providing emotional support, they can help you to understand Down syndrome and what causes it, and what a diagnosis means for your child's health and development, and for your family. Genetic counsellors are trained to provide information and support that is sensitive to your family circumstances, culture and beliefs.

Community support is also available for parents whose child has been diagnosed with Down syndrome at each state or territory's Down Syndrome Association. Visit [Down Syndrome Australia](#) for more information.

The [Genetic Support Network of Victoria (GSNV)](#), is connected with a wide range of support groups throughout Victoria and Australia and can connect you with other individuals and families affected by Down syndrome.

**Where to get help**

- Your [GP (doctor)](#)
- Your local community health centre
- [Maternal and child health nurse](#)
- [Down Syndrome Victoria](Tel. 1300 658 873)
- [Down Syndrome Australia](Tel. 1300 344 954)
- [Genetic Support Network Victoria](Tel. (03) 8341 6315)
- [Association for Children with a Disability](Tel. (03) 9880 7000 or 1800 654 013 for rural callers)
- [Better Start for Children with Disability](Tel. 1800 242 636)
- [Victorian Clinical Genetics Services (VCGS)](Tel. 1300 118 247)

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Down Syndrome Association of Victoria

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