

## Down syndrome explained

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 results in a number of physical and developmental characteristics and some level of intellectual disability.

These days, the majority of people growing up with Down syndrome have the ability to achieve and participate as valued members of their communities, with varying degrees of support. Support is available for individuals with Down syndrome and their families, and for expectant parents who have received a diagnosis of Down syndrome.

### Down syndrome and the extra chromosome

Chromosomes are the blueprint for the body's development. They are found in every cell in our body and determine our physical and mental characteristics. The usual number of chromosomes 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 chromosome disorder and occurs in all races and cultures at around the same rate. In Victoria, between 45 and 60 babies will be 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 indications.

### Forms 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** – every cell in the body has an extra chromosome 21. The majority of people (about 95 per cent) with Down syndrome have trisomy 21. It is an accident of birth and is not a hereditary condition.
- **Mosaic Down syndrome** – 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** – 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 also uncommon, occurring in about three to four per cent of cases. In about one third of these cases, one of the child's parents has the same translocation. For this reason, referral to a genetic counselling service is usually recommended.

Genetic tests can show what type of Down syndrome a baby has.

### Common physical characteristics

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:

- **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 (epicanthic fold) and small white patches on the edge of the iris of the eye (Brushfield spots).

- **Face** – this is often rounded and tends to have a flat profile.
- **Stature** – babies with Down syndrome are usually smaller and weigh less at birth than others. Children tend to grow more slowly and are commonly smaller than other children their age. Adults with Down syndrome are commonly smaller than in the general population.

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

## 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 individual 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.

## Capabilities and potential

Down syndrome affects, but does not determine, development and achievement in a person. People with Down syndrome demonstrate a wide range of capabilities. What happens after birth will be far more important in shaping the outlook for a person with Down syndrome than the occurrence of the extra chromosome at conception.

These days, children with Down syndrome attend childcare settings, pre-schools and primary and high schools 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 their communities. An increasing number 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 encouraged to do so.

## 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
- Amniocentesis
- Chorionic villi sampling.

## Genetic counselling and support are available

Genetic counselling is available to parents in all states of Australia and may be helpful if you are concerned that you may have a child with Down syndrome. Community support is also available for parents whose child has been diagnosed with Down syndrome.

## Where to get help

- Your doctor
- Your local community health centre
- Maternal and child health nurse
- Down Syndrome Victoria Tel. 1300 658 873 or email: [info@dsav.asn.au](mailto:info@dsav.asn.au)
- Genetic Support Network Victoria Tel. (03) 8341 6315

- Association for Children with a Disability

## Things to remember

- Down syndrome is a common genetic condition caused by an extra chromosome 21.
- People with Down syndrome are not different to other people except that they have an intellectual disability, which affects how they learn things.
- Children with Down syndrome have varying abilities to achieve and respond well to stimulation. With the support and opportunities available to them today, most people with Down syndrome are able to achieve and participate as valued members of their community.
- Support is available for people with Down syndrome and their families and for expectant parents who have received a diagnosis of Down syndrome.

**This page has been produced in consultation with, and approved by:**

Down Syndrome Association of Victoria

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