Down Syndrome
Australia

A guide for expectant parents

This document has been produced by
Down Syndrome Australia.

Down Syndrome Australia is the peak body for people with Down syndrome in Australia. Our purpose is to influence social and policy change, and provide a national profile and voice for people living with Down syndrome. We work collaboratively with the state and territory Down syndrome associations to achieve our mission. Our vision is an Australia where people living with Down syndrome are valued, reach their potential, and enjoy social and economic inclusion.

Acknowledgements

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The purpose of this resource is to provide balanced information about Down syndrome to expectant parents. This includes parents who have already had some prenatal testing which reveals their child has or may have Down syndrome, as well as those who want more information about Down syndrome for a range of reasons.

Depending on your circumstances some sections of this resource may not apply to you. You should feel free to read the parts of the document that are helpful and appropriate for you.

Down Syndrome Australia respects the right of families to make decisions appropriate to their own beliefs and circumstances.

Prenatal testing

This section of the resource provides information about prenatal screening and diagnosis. It covers the types of tests available and the different information these tests provide. It also looks at the next steps for parents who receive a diagnosis of Down syndrome and where to get useful support.

Permission to test

Prenatal testing is offered routinely to most women in Australia and can provide useful information about your pregnancy. This information may help families plan for their pregnancy or birth care. However, it is your choice whether or not to have prenatal testing.

It is also important to take time to ensure that you have understood all the correct information about the tests, their risks and limitations and the conditions that are being tested for. You should only have testing if you have given your permission.

Information about prenatal testing

Prenatal tests are divided into two types: screening tests and diagnostic tests. Screening tests are used to identify the likelihood of your baby having Down syndrome. Diagnostic tests provide a definitive diagnosis of Down syndrome based on genetic testing of cells from the baby.

Screening tests

There are a few different screening tests that are currently offered to pregnant women in Australia. They do not provide a definitive diagnosis.

The relevance (sensitivity and specificity) of any screening test is dependent upon a number of variables including the type of material that is being tested, i.e. biochemical analytes vs DNA, the particular test being used and other pregnancy related factors such as maternal age. It is important to ask about the specific performance of any screening test you are offered and to be aware that sometimes a test can suggest a 'high chance' result when the baby does not have Down syndrome; or a 'low chance' result when the baby does have Down syndrome.

Non-invasive prenatal testing (NIPT) is a blood test available from the 10th week of pregnancy and is available in the private health system in Australia. This test analyses both maternal and foetal cell-free DNA circulating in maternal blood to provide either a high probability or low probability of your baby having Down syndrome.

Combined first trimester screening is performed between 9 and 14 weeks gestation. This test combines a number of different measures; including measurement of chemicals in the blood, a scan to determine nuchal translucency, (which is a measure of the collection of fluid under the skin at the back of the baby's neck), and the mother's age to provide an estimate of the chance that a pregnancy is affected by a chromosomal condition, which includes Down syndrome, as well as others.

Second trimester screening is also available later in pregnancy through a blood test that can be done between 14–20 weeks. Most pregnant women are also routinely offered an ultrasound at 19–20 weeks of pregnancy, which can sometimes show other signs ofDown syndrome.

If the result of these screening tests shows there is an increased likelihood of your child having Down syndrome, you may be offered a diagnostic procedure such as chorionic villus sampling (CVS) or amniocentesis.

Diagnostic tests

Both CVS and amniocentesis are invasive procedures that involve obtaining a sample of the baby's genetic material for examination. They are the only way to get a definitive diagnosis of Down syndrome prenatally. These invasive tests increase the risk of miscarriage (above the usual background risk).

Regardless of the results of your screening tests, you are under no obligation to undertake further diagnostic testing. This choice is entirely up to you.

Your obstetrician, GP, midwife or a prenatal genetic counsellor can help you with information about all the different types of prenatal testing.

Making a decision

If diagnostic testing reveals Down syndrome, parents are usually offered a choice about continuing or terminating their pregnancy. It is important to take some time to carefully consider all the available information about Down syndrome and your options. Women report lower levels of regret either way when these decisions are made with access to high quality, up to date information about the condition, a lack of time pressure, and access to non-directive counselling and support by health care providers.

Genetic counsellors are available to discuss the results of a Down syndrome diagnosis with regards to medical, psychological and reproductive implications. They can also provide support for families while they consider the options available to them, whether that be continuing with the pregnancy, termination, adoption or permanent care placements.

Your local Down syndrome organisation can provide up to date information and support about Down syndrome. They have experience in providing balanced, non-directive information and supporting families in the different stages of the decision-making process. They can answer questions you may have and can also put you in touch with a family who has a child with Down syndrome. This can be a helpful opportunity to learn more about the day-to-day, lived experience of families and children who have Down syndrome and learn about people with Down syndrome living meaningful lives in their community at school, the workplace, sport or a range of different ways. You can contact your local Down syndrome organisation at **1300 881 935**.

There are also online support groups available for women where you can connect with other women with a prenatal diagnosis or high probability screening result and mothers of young children with Down syndrome. Contact your local **Down syndrome association** to find out about online support groups available.

Receiving a prenatal diagnosis can affect couples and family members in different ways at different times throughout the pregnancy. It is important to look after your own mental and physical wellbeing during these times. If you need emotional support, or you feel conflicted and are having trouble making a decision, you can reach out for support either from an understanding friend or family member, a support group or a professional such as a genetic counsellor, psychologist or a social worker.

What is Down syndrome?

Down syndrome is a genetic condition. It is not an illness or disease.

Our bodies are made up of millions of cells. In each cell, there are 46 chromosomes. Down syndrome is caused when there is an extra chromosome. People with Down syndrome have 47 chromosomes in their cells instead of 46.

Down syndrome occurs at the point of conception. It has nothing to do with anything the mother or father have done before or during pregnancy and it occurs across all ethnic and social groups and almost always occurs randomly. There is an increased chance of having a child with Down syndrome with increased maternal age, however having a child with Down syndrome occurs across all ages. The chances of parents in their early twenties having a child with Down syndrome is 1 in 2000, but this chance rises to 1 in 350 at 35 years of age.[1]

Most people with Down syndrome have happy and healthy lives. Research indicates that 99% of people with Down syndrome are happy with their lives.[2] People with Down syndrome go to school, get jobs, participate in sport and contribute to their communities in a range of different ways.

People with Down syndrome have:

- Some level of intellectual disability.
- Some delay in development which may include delays in speech development and motor skills.
- Some characteristic physical features, including a recognisable facial appearance and short stature.
- Some health concerns which may include:
  - vision and hearing problems
  - congenital heart condition

[1] www.ndss.org/about-down-syndrome/down-syndrome

[2] www.ncbi.nlm.nih.gov/pmc/articles/PMC3740159

- increased risk of leukaemia
- thyroid problems
- respiratory infections
- gastrointestinal issues.

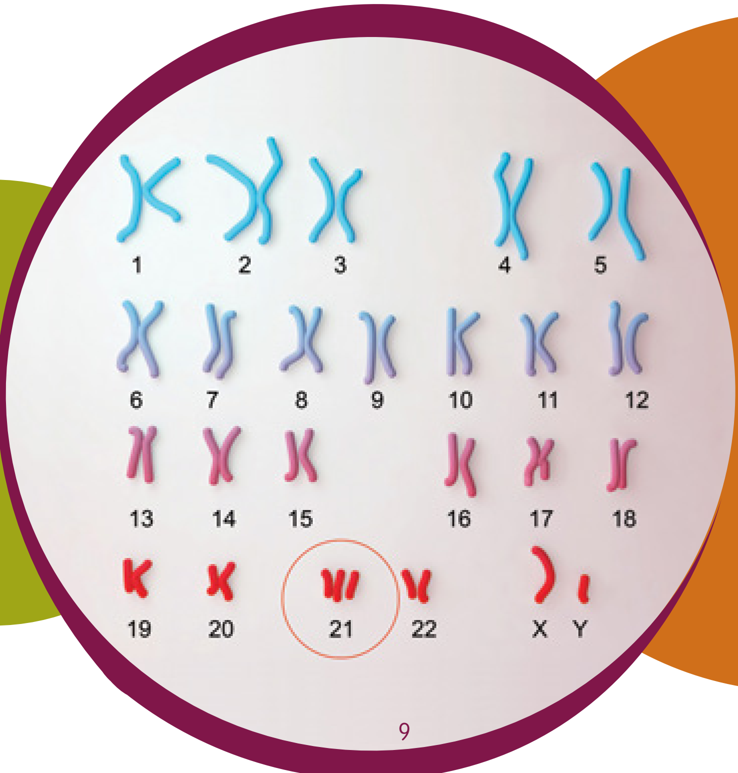
Not all people will experience these health concerns. Some people may have some and not others, others may not have any –and in many cases these health conditions are treatable and/or resolved with medical interventions and treatment.

Because no two people are alike, it is impossible to tell or predict how many features will present in each individual; this varies from one person to another.

There are three different types of Down syndrome – Trisomy 21, Translocation Down syndrome or Mosaic Down syndrome. This is identified from chromosome studies done at birth to confirm the diagnosis of Down syndrome:

Trisomy 21

Most children with Down syndrome (around 95%) have an extra whole chromosome 21 in every cell of their body. This is called trisomy 21.



Translocation Down syndrome

Translocation Down syndrome is caused when additional chromosome 21 material becomes attached to one of the other chromosomes, instead of being separate as in trisomy 21. You cannot tell whether a person has Translocation Down syndrome or trisomy except by looking at a chromosome test. Translocation usually happens by chance, but sometimes it can be inherited. Parents can have tests done to see if this is the case, as they would have a greater chance of having a baby with Down syndrome in subsequent pregnancies.

Mosaic Down syndrome

In about one percent of children with Down syndrome there is an extra whole chromosome 21 in only a percentage of their body cells – the rest of the cells don't have the extra chromosome. People with mosaic Down syndrome can have fewer characteristics expressed or can have the majority of features of Down syndrome, just as a person with all trisomy 21 cells could experience.

First feelings

All parents react differently to the news that their child has, or may have Down syndrome. Your reaction can depend on your cultural beliefs, background, and prior experience with children who have a disability.

There are some common feelings expressed by parents at the time of diagnosis or finding out the results of prenatal screening. You may find it helpful to know that others have felt the same. Some of these shared feelings are shock, a feeling of disbelief and sorrow, feelings of protectiveness, feelings of inadequacy and embarrassment, anger and guilt. Some parents don't have any of these feelings.

Some parents are very clear about the decision they want to make about their pregnancy, others feel more uncertain. Please know that however you are feeling at this moment in time is perfectly normal. It is normal to have worries, concerns and questions. It is equally normal to not have any of these things. Be patient with yourself and take one step at a time.

You can talk to your local **Down syndrome organisation** about your feelings and questions. The state associations have experience in providing balanced, non-directive information and supporting families in the different stages of the decision-making process. You can contact your local Down syndrome organisation at **1300 881 935**.

> **"I wish I'd known then that my baby would still be beautiful and would develop and do all the things typically developing babies do, albeit at a slower pace, and that he would teach me a lot of things"**
>
> **PARENT OF A CHILD WITH DOWN SYNDROME**

Talking to other parents

Many people find it helpful to speak to another parent of a child with Down syndrome, who can talk about their own experiences and feelings. While no one has exactly the same experience, there are others who share similar experiences. These parents may also be able to provide suggestions of local services or supports that have been useful for their family.

Keep in mind that their comments and views will be based on their own experiences and your experience may be different. You may, however, find that gathering knowledge and information enables you to more easily make decisions in relation to your child's health and wellbeing.

Your state or territory Down syndrome association can put you in contact with other parents who are happy to talk to you, so please get in touch when you feel ready.

> **"Other parents of children with Down syndrome helped us so much. There was this awful feeling that we were the only people in the world who had this happen to them."**
>
> **PARENT OF A CHILD WITH DOWN SYNDROME**

> **"Initially we thought there would be a million decisions to make right away, but we found that the decisions were spread over a long period of time, like with any other child. The decisions aren't knocking at your door every day."**
>
> **PARENT OF A CHILD WITH DOWN SYNDROME**

Dispelling myths

There are many common misconceptions about Down syndrome and what life will be like. It is important to understand that most people growing up with Down syndrome today will lead quite ordinary lives in the community. Some people with Down syndrome may not need much help to lead an ordinary life, while others may require more support. This section aims to address some of the common misconceptions.

Myth 1: People with Down syndrome have a short life expectancy

Over the last 30 years, the average life expectancy of a person with Down syndrome has increased from less than thirty years of age, to today when many people with Down syndrome live into their sixties. It is expected that the life expectancy of people with Down syndrome will continue to increase.

Myth 2: My baby won't be born healthy

There is an increased chance that some babies with Down syndrome will be born with health concerns. Your pregnancy will likely be more closely monitored and regular check-ups with your Paediatrician or GP once your baby is born are important. A number of health conditions occur more frequently in people with Down syndrome and your baby will be checked for some of these at birth, in particular heart and thyroid problems. This does not mean that your child will experience all, or even any, of these conditions. The main thing is to seek support from health professionals with a good understanding of Down syndrome, to ensure your child's health is managed as well as possible.

Myth 3: People with Down syndrome will be reliant on their parents for support for the rest of their lives

People with Down syndrome are able to live full lives as part of the community. With the right support they may live independently, be employed and participate in the community like any other person. With the introduction of the National Disability Insurance Scheme in Australia, there is support for people with Down syndrome to live an ordinary life and to be part of the community.

Myth 4: My child will be limited in what they can achieve

Your child with Down syndrome can and will learn. As with other children the range of ability in a child with Down syndrome is wide and their potential is an unknown factor. Children with Down syndrome may require assistance and support to achieve milestones and development may take longer. It is important to remember that people with Down syndrome continue to learn throughout their entire life. With support from professionals – such as a speech therapist or a physiotherapist – and with the appropriate supports, your child will be able to learn and achieve throughout their life.

Myth 5: Having a child with Down syndrome will negatively affect my family

The birth of a child with Down syndrome will bring about changes to the family, just like the arrival of any baby. When a new baby joins a family, parents should try to talk to each other right from the start. Share your feelings and support with each other and try to arrange things so that you spend time together. Many parents worry about how having a sibling with Down syndrome will affect their other children. Research shows a variety of positive outcomes for siblings of people with Down syndrome including more empathy and kindness towards their brother or sister, less conflict, and more warmth in their relationships.[3] Research has shown that a more mature level of empathy is developed and greater acceptance of diversity is expressed, not only towards their sibling but across the wider community.[4]

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[3] www.ncbi.nlm.nih.gov/pmc/articles/PMC3348944/

[4] www.ncbi.nlm.nih.gov/pubmed/17559293/

> “My daughter with Down syndrome is a very healthy little girl. If I focused on the list of all of the potential health problems I think I’d be a nervous wreck. When my other two children were born, I didn’t get a list of possible ailments that they may be afflicted with – at the same time I do think it’s important to be informed and we go to the doctor for regular check-ups.”
>
> **PARENT OF A CHILD WITH DOWN SYNDROME**

More information about talking to your other children about Down syndrome is available in Down Syndrome Australia’s New Parent Guide, which is available at: **www.downsyndrome.org.au/resources/life-stages/new-parent/**

The rewards and satisfaction of rearing a child with Down syndrome and the sense of positive achievement gained, are no less than with any other child – perhaps even greater.

> "No doubt about it, we've had our ups and downs since our son was born – but I really think that the fact he has Down syndrome has been fairly irrelevant. Having any child join the family is an adjustment."
>
> PARENT OF A CHILD WITH DOWN SYNDROME

> "One of my major concerns was about our other kids – I was worried they'd be teased, but it just hasn't happened so far and we take each day as it comes."
>
> PARENT OF A CHILD WITH DOWN SYNDROME

Informing family and friends

As well as the complex feelings you may have when you find out that your baby has Down syndrome, it is sometimes difficult to know how to share this news with family and friends. Some people may want to share this news during their pregnancy, others will wait until the child is born.

The following are some ideas and considerations to assist you when you are ready to start talking with family and friends about a Down syndrome diagnosis.

Tell relatives and friends about the diagnosis when you are ready

Give yourself permission to feel your emotions and allow yourself time to adjust. Friends and family may surprise you with their understanding, and their support can be invaluable.

Have information ready

Many people may not have met anyone with Down syndrome or they may have outdated information and ideas so providing them with up to date information about Down syndrome will help. Information and helpful resources are available at **www.downsyndrome.org.au**. Your friends and relatives are welcome to contact your state or territory Down syndrome office if they have any further questions or need additional information.

Remember... those around you are usually trying their best

Give grandparents, siblings, aunts, uncles and friends time to overcome their own initial reaction if they need it. They too are trying to adjust and will react in their own way. Some may say things that upset you, usually without realising. Try to remember that their initial response and attitude may not be the one they keep. Their feelings and reactions will most likely change as time goes on. Many parents report that they 'set the scene' for their friends and family by sharing their excitement about their baby and their high expectations for the new little one's future. The opportunity to spend time and bond with the baby may also help them see that babies with Down syndrome are more like other babies than different.

Supports available

The Government provides funding and support to assist people with disabilities.

National Disability Insurance Scheme (NDIS)

The National Disability Insurance Scheme plays an important role in support for families and children with Down syndrome. The goal of the NDIS is to support social and economic participation for people with a disability through their plans. For babies and children, it can provide funding to support a range of activities and services including physiotherapy, speech therapy and occupational therapy. Children with Down syndrome under the age of seven are automatically eligible for supports through NDIS if they also meet the residency requirements. Most older children and adults with Down syndrome will also have access to the NDIS. More information about the NDIS and accessing support is available in our Early Years booklet available at: **www.downsyndrome.org.au/resources/**

Carer Allowance

Parents or carers of a child with a disability may be eligible to receive a Carer Allowance. The forms are available through Centrelink or may be provided to you in the hospital. You can find out more by contacting Centrelink on **132 717** or visiting **www.humanservices.gov.au**

Life with Down syndrome

With the right supports people with Down syndrome live full, happy lives. Research has found that 99% of people with Down syndrome surveyed indicated that they were happy with their lives.

Things have changed dramatically over the last 30 years when it comes to health and public policy for people with Down syndrome. Life expectancy has increased dramatically, and people are being more involved in their communities in a range of different ways. There continues to be challenges around negative attitudes and low expectations, but many advocates are working to challenge these views. The implementation of the NDIS is creating new opportunities for people with a disability with a focus on helping people to live an ordinary life.

Many adults with Down syndrome are now living independently in the community, sometimes with support from friends, family or organisations. They are employed in valued roles in either supported or open employment, are enjoying friendships and relationships, and some are choosing to marry.

People with Down syndrome have the right to be included in the community and there is no doubt that the prospects for the future are bright.

For more information

Down Syndrome Australia has developed a range of information available through the website www.downsyndrome.org.au

You can find our prenatal resources on our website here:

www.downsyndrome.org.au/resources/life-stages/prenatal/

Download a **New Parent Information Pack**. These resources are designed specifically for parents and families with a new baby who has been diagnosed with Down syndrome.

Visit our website for a list of books to read about Down syndrome

www.downsyndrome.org.au/about-down-syndrome/faqs/booklist/

Useful resources from other organisations:

www.ndss.org/about-down-syndrome/down-syndrome

www.downsyndromepregnancy.org

www.downs-syndrome.org.uk/for-new-parents/being-pregnant

Associations

National: 1300 881 935

New South Wales

Down Syndrome NSW
T: 1300 881 935
E: admin@dsansw.org.au
W: www.downsyndrome.org.au/nsw

Victoria

Down Syndrome Victoria
T: (03) 9486 9600
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E: info@dsav.asn.au
W: www.downsyndrome.org.au/vic

Queensland

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T: (07) 3356 6655
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South Australia

Information Service South Australia (Down Syndrome Australia)
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Western Australia

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E: office@downsyndromewa.org.au
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Tasmania

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Northern Territory

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