Understanding Down Syndrome

A Guide from Down Syndrome NSW

DOWN SYNDROME
NEW SOUTH WALES

About Down syndrome

Down syndrome occurs at conception. People from all different backgrounds and ages have children with Down syndrome.

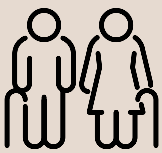
There is no national data collection on the number of people with Down syndrome in Australia.

Down syndrome is the most commonly occurring chromosomal condition.

The incidence of births of children with Down syndrome **increases** with the age of the mother. The chance of a woman conceiving a child with Down syndrome varies from **1 in 1400 for a woman 20 years of age to 1 in 30 at age 45 years.**

Younger women have babies more frequently, so the majority of babies born with Down syndrome are born to women **under 35 years of age**.

People with Down syndrome are living longer and healthier lives than they have in the past.



Life expectancy of people with Down syndrome has dramatically increased over the past 50 years, with the average life expectancy of a person with Down syndrome in Australia being **60 years of age.**

It is estimated there are

13,500

people with Down syndrome living in Australia.

Applying these numbers to the Australian population, every 10,000 people there are

5.2

people with Down syndrome.

This is similar to other countries like the UK.

Approximately

1 in every 1100

babies born in Australia will have Down syndrome.

Each year there are approximately

265

new babies who have Down Syndrome.

A short history of Down syndrome

Down syndrome is not a disease, it is a genetic condition. People with Down syndrome are not sufferers, they are people first with diverse talents, strengths, desires and contributions. Join us as we explore the history of Down syndrome, touching on a little biology along the way.

It is important to note that in the representation of the history of Down syndrome, many now outdated and inappropriate names have been associated with Down syndrome in the past. This information serves merely as a historical overview and in no way condones the use of this language or approach in the present day. We wish to reflect this history to show how far we have rightly come.

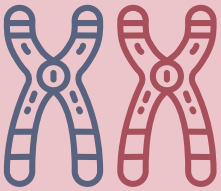
A Quick Lesson In Biology

An exciting discovery of the 20th century was DNA (deoxyribonucleic acid), the molecule that carries genetic information in our cells.

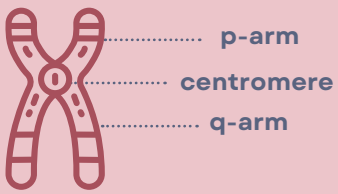
DNA is arranged in a double-helix (spiral) structure and contains chemical "bases" that form a code guiding cell activity and protein production. Most DNA is found in the cell nucleus, where it is organised into chromosomes.

Down syndrome occurs when there is extra genetic material on chromosome 21. Although the human genome contains over 30,000 genes, the affected region in Down syndrome involves only around 50–100 genes, with just a few currently understood. This additional genetic material influences how cells develop and specialise, contributing to the common characteristics seen in people with Down syndrome.

Down syndrome was first described in 1866 by Langdon Down, who used the term "mongoloid" based on outdated and incorrect assumptions.



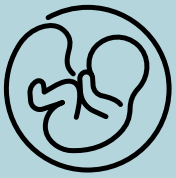
Humans typically have 23 pairs of chromosomes – 22 of which are identical pairs though varying in size to other pairs, while the 23rd pair of chromosomes determines the sex of an individual, often referred to as X and Y chromosome.



Each chromosome is made up of two strands of DNA – known as a chromatid – joined together by chiamata at a specific site known as the centromere.

Despite it's name indicating otherwise, this often is not in the centre, resulting in each chromatid having a long arm (referred to as q) and a short arm (referred to as p).

A short history of Down syndrome



Ideas about the causes of Down syndrome have changed over time. Until the late 1970s, it was wrongly thought that it was caused by "radical" foetal development or stress during pregnancy.

Earlier observations were made by Jean-Étienne Dominique Esquirol in 1838 and Édouard Séguin in 1844. However, the genetic cause was only discovered in 1959, when Jérôme Lejeune identified Down syndrome as being caused by an extra copy of chromosome 21 (trisomy 21).

Since then, research has shown that the features of Down syndrome are linked to a small section of chromosome 21.

The term "Down syndrome" was later introduced by geneticists and officially adopted by the World Health Organization in 1965.

While research continues, it is still not fully understood why the extra chromosome occurs or exactly how it affects development, though knowledge in this area continues to grow.

The identification of Down syndrome

In 1866, Langdon Down first described the characteristics of the most common cause of learning and intellectual disability. He set the condition apart from mental disability by coining the first term used to describe those with Down syndrome: "mongoloid". In Down's opinion, the children with the genetic condition shared similar characteristics to the people of the Mongolian race. The term "Down syndrome" was suggested later by a group of geneticists who wrote to the British medical journal, The Lancet, with four alternatives to "mongoloid." Down syndrome was later endorsed by the World Health Organization and officially accepted as standard terminology in 1965.

It remains unknown what may predispose cells to carry additional material during the splitting process, how this in turn affects development and function. There is, however, a current state of knowledge that provides some insights in what is an ever growing field of research. To learn more of this, see our fact sheet series.

In 1910, the expected age of survival for children with Down syndrome was nine years. After the discovery of antibiotics, the average age of survival had increased to roughly 19 or 20 years.

In 1980, the life expectancy was 25 years old. With recent advancements in clinical treatment such as heart surgery, about **80% of adults with Down syndrome are living to the age of 60** and at times are living beyond the age of 60.

Down syndrome represented in Renaissance art

Throughout history, some artworks have been interpreted as depicting people with Down syndrome.

Several Renaissance paintings, including works by Andrea Mantegna, feature children and figures with characteristics associated with Down syndrome. These artworks provide an interesting insight into how disability may have been viewed and represented in earlier societies, while also highlighting themes of humanity, inclusion, innocence, and dignity across history.

Andrea Mantegna's works (c.1455–1485), including Madonna and Child and Virgin and Child with Saints Jerome and Louis of Toulouse, have been suggested to show figures with Down syndrome characteristics.

In 1982, Dr Brian Stratford proposed that Mantegna may have used a child with Down syndrome as a model for the Christ child, based on facial features and hand shape.

A 16th-century Flemish painting, The Adoration of the Christ Child, has also been interpreted as possibly depicting individuals with Down syndrome.

Researchers Levitas and Reid suggest these depictions may reflect either early inclusion of people with disabilities or a lack of recognition of Down syndrome at the time.

Some interpretations propose that artists may have been highlighting qualities such as innocence, love, and gentleness in their subjects.

About Down Syndrome NSW

Once upon a time in NSW, Australia, there was a remarkable group of individuals who had children who were born with Down syndrome. Inspired by their children's journey and fueled by their desire to create a more inclusive society, the parents embarked on a mission to establish an organisation dedicated to supporting individuals with Down syndrome and their families. And so, the seeds of Down Syndrome NSW were sown.

As the word spread, more families joined these gatherings, forming a tight-knit community that offered support, understanding, and hope. The parent group worked tirelessly to run programs and collaborated with healthcare professionals, educators, and community leaders to raise awareness about Down syndrome and advocate for improved services and opportunities.

With the growing momentum, Down Syndrome NSW vision expanded beyond support groups and advocacy. They envisioned a dedicated organisation that could provide a wide range of services to individuals with Down syndrome, from newborn babies right through to support for the aging. With the help of passionate volunteers and the backing of the community, Down Syndrome NSW began to take shape.

Today, Down Syndrome NSW continues to evolve and adapt to the changing needs of the community. We have become a catalyst for change, breaking down barriers and championing the rights and abilities of individuals with Down syndrome. These parents and their story stand as a testament to the power of compassion, determination, and the enduring spirit of individuals, who make a profound impact on the world by turning their dreams into reality.

What we do

Advocacy and Research

Advocacy is at the heart of everything we do. Down Syndrome NSW works to identify and address the barriers and systems that impact people with Down syndrome and their families.

We advocate on key issues affecting our members across NSW through submissions, government relations, policy work and direct engagement with decision-makers. We do this both independently and in partnership with other disability advocacy organisations in NSW. We also work with Down syndrome organisations across Australia on national issues that affect people with Down syndrome and their families.

Down Syndrome NSW actively contributes to research that supports better outcomes for people with Down syndrome. We partner with universities and research organisations across Australia by providing feedback, supporting research applications, contributing member perspectives and participating in steering committees.

Information, Referral and Capacity Building

Down Syndrome NSW provides information, referral and capacity building support for people with Down syndrome, families, carers, health professionals, schools, educators and the broader community.

We offer a wide range of resources, including fact sheets, guides, books and information through our website **www.downsyndromensw.org.au**. People can also contact our Information and Support Line by phone or email for individual information and referral support.

We also deliver capacity building workshops and information sessions for people with Down syndrome, families, carers, professionals and mainstream services. These activities help build understanding, confidence and inclusion across the community.

Ageing Well

Down Syndrome NSW is committed to supporting adults with Down syndrome to live fulfilling, connected and healthy lives as they age.

People with Down syndrome may experience ageing differently from the general population. While improvements in healthcare have significantly increased life expectancy, adults with Down syndrome may also face specific health, social and support needs as they grow older.

Through our Ageing Well work, we provide information, resources, peer connection and practical support for adults with Down syndrome, their families and carers. This includes opportunities to connect, share experiences, build knowledge and plan for the future.

The Congratulations Initiative

The Congratulations Initiative supports families from prenatal diagnosis, postnatal diagnosis, birth and beyond. It provides families with up-to-date information, connection and support at an important time in their lives.

The initiative also works with medical professionals to support the delivery of a diagnosis in a respectful, informed and balanced way. Through our Medical Professionals Packs, we provide current information about Down syndrome, link families with Down Syndrome NSW as a key support partner, and promote the important message of saying “congratulations”, not “sorry”.

The Inclusive Education Initiative

The Inclusive Education Initiative supports the inclusion of students with Down syndrome by strengthening the capacity, knowledge and understanding of students, families and education staff, including teachers, principals and School Learning Support Officers.

The program works across mainstream and specialist school settings, building strong connections between students, families, schools, therapists and behaviour support specialists.

At the centre of this work is the student with Down syndrome, with support focused on their learning, inclusion, wellbeing and participation at school.

Up! Up! and Away

UP! UP! and Away supports children and young people of all ages, from toddlers through to teenagers.

Activities are specially curated for children and young people with Down syndrome and intellectual disability, with a focus on active engagement, social connection and participation.

The program offers enriching experiences through capacity-building workshops, activities and opportunities to learn new skills and develop confidence, wellbeing and social connection.

Employment Connections

Employment Connections supports adults with Down syndrome to explore work options, build confidence and find roles that match their skills, interests and personality. The project works with jobseekers, families and employers to create meaningful and sustainable employment pathways, while helping workplaces build their confidence, understanding and inclusive practice.

By partnering with employers, the project promotes workplaces where people with Down syndrome are valued for their strengths, given the right support to succeed, and able to contribute as part of a diverse team.

UP! Club

UP! Club provides social activities, camps and capacity building opportunities for adults with Down syndrome.

The program supports adults to learn, engage and connect with others. Activities focus on reducing isolation, building skills, supporting further education, promoting independence and assisting with future planning.

Our Stories

Lucy and Amelia's Story

For new parents Lucy and Roberto, the birth of their beautiful baby Amelia should have been a time of joy and celebration, but with negative messaging from medical professionals about a Down syndrome diagnosis, their pre-natal experience was tinted with fear and uncertainty.

Lucy recalled the moment they received the results from a Genetic Counsellor, who shared the news as if it was a devastating, terrible outcome. Instead of saying congratulations to the expectant parents, the medical staff said they were sorry – "I remember thinking my baby isn't dying, why is everyone so sorry?"

Instead of providing useful, accurate and up-to-date information, the Genetic Counsellor went on to describe what they should expect of a baby with Down syndrome, stating that the baby would have moderate intellectual disability and describing physical features including small ears and stature. It all sounded like a warning.

Until changing hospitals, their early experiences continued to be negative. For their 20-week scan, they saw an obstetrician who said she had noted some suspected abnormalities in the heart and due to a minor intestinal obstruction, the amniotic fluid was building up, due to this there was a very good chance their baby's heart could stop at any moment. Lucy recalls this conversation; "She then, in the most insensitive tone possible asked if we wanted to terminate. I was a mess; my partner was furious. He frustratingly asked if she had anything positive to say? She just stared at us, and we got up and walked out."

Lucy and Roberto spiraled into a very dark place after this. All the joy, excitement and celebration of becoming new parents had been taken from them.

A week later, now at a new hospital, the staff could see how distressed they were and quickly put their minds at ease that their baby's heart could not stop due to the buildup of amniotic fluid. Furthermore, they identified that she was perfectly safe, and her heart was functioning normally. There were no abnormalities with her heart, she had a minor intestinal obstruction which would require surgery after birth but everything else relating to the pregnancy was normal.

Knowing that her baby was safe, Lucy could now finally relax. But the fear she'd felt all week that she might at any point lose her baby had made that period of their lives one of the darkest and most challenging for their family. Not only did it impact their relationship, creating unnecessary added stress and worry, but it was also deeply traumatising for their parents and siblings.

Lucy and Roberto would like to see the medical professionals involved in their early care to be better equipped with up to date information to be able to offer them positive and constructive information so new parents could feel empowered and educated and enter this beautiful journey the right way so that we can give our children the best opportunities and support for the best life possible.

Connecting with Down Syndrome NSW was a turning point for their family. They were visited by staff with a shared lived experience, who were able to pass on positive and encouraging messages about their own children's achievements and challenges. With the right advice and information provided, Lucy and Roberto found comfort and support in the Down syndrome community.

Now, at 6-months old, Amelia is a thriving, happy, and beautiful baby girl. She loves playing on her play mat, she is active and strong. She can roll over onto her belly and looks so proud of herself when she does. She gives Lucy and Roberto endless smiles and sweet giggles and has enriched their lives beyond belief. Most importantly, they are so proud that she is their daughter.

"Knowing that they were just a phone call away was and is so helpful and we are very thankful for such a supportive and proactive organisation for our loved ones. We can't thank Down Syndrome NSW enough. We've already learned so much and met some amazing people who are part of this community. I really enjoy the online fortnightly New Parent sessions and got so much out of the sign language course we participated in during pregnancy. It is so wonderful that there are so many educational sessions made available to us and I really enjoy receiving Emily's organisation updates so we can plan ahead and feel so positive about Amelia's future. To know that DSNSW and so many inspiring people are advocating for our loved ones really means the world to us."

Our Stories

Melissa and Samuel's Story

Being a new parent comes with its common set of challenges for most families, but for parents whose children are diagnosed with Down syndrome, the start of their journey can feel particularly daunting. What should be a time of celebration and joy can sometimes be overshadowed by the cautionary messaging from healthcare professionals.

While there has been progress in recent years, there are still instances where healthcare professionals may struggle to deliver accurate and supportive information to parents upon the diagnosis of Down syndrome. In some cases, the news is delivered in a way that may emphasise the challenges rather than the potential for a fulfilling life. Unfortunately, the initial communication from healthcare providers can be framed in a negative light, using language that conveys sympathy or apologies instead of celebrating the birth of the child.

For Lee and Melissa, reflecting on the birth of their son Samuel, now 10 years old – a skilled ball player, bike rider and lover of music, they consider themselves lucky to have had an extremely supportive midwife who encouraged them about their baby's capabilities. In the early days, when Samuel was only a newborn, Lee and Melissa were visited by a team member from Down Syndrome NSW to offer support, guidance and celebration for the birth of their beautiful son with Down syndrome.

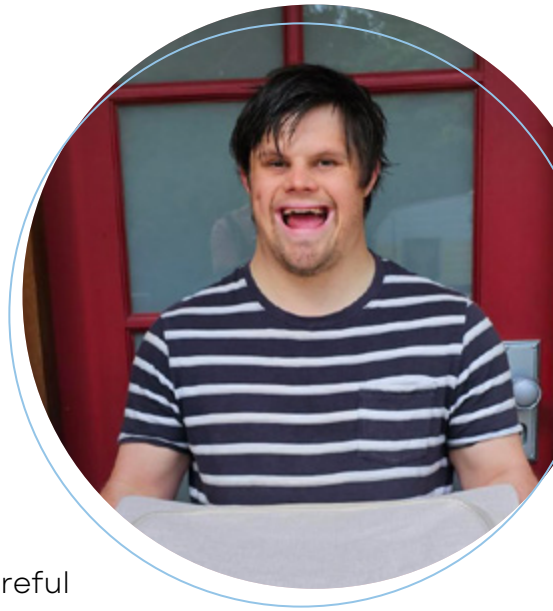
This first hospital meeting would start a much longer journey for Samuel's parents, who continued to contact the team at Down Syndrome NSW at all stages of Samuel's life. Throughout the years, they found a strong sense of community; connecting with other families, attending educational webinars and social events, and receiving guidance when new challenges came up.

Parents like Lee and Melissa know how important it is for parents of newborns with Down syndrome, to get access to support and connect with their community as soon as possible in those early years. Like many of the families Down Syndrome NSW work with every day, they experienced firsthand the benefit of medical professionals learning to say Congratulations, instead of sorry, and providing parents with accurate, up to date information to support their journey.

As Samuel grows up, with his parents beside him, Down Syndrome NSW will be with them along the way.

> "I know Down Syndrome NSW are advocating on our behalf in the wider community to improve the lives of all with Down syndrome."

Sam Stubbs' Story



Sam's journey towards moving out of home began in August 2022 at Down Syndrome NSW's empowering Housing Conference. Surrounded by support and guidance, Sam left with a newfound determination to venture out on his own, away from the easy comforts of home and the watchful eyes of his parents.

With unwavering encouragement from his family and the invaluable resources provided by Down Syndrome NSW, Sam embarked on a mission to find his own place. After careful planning and scouting, they discovered a perfect home just a stone's throw away, where Sam could spread his wings while still being within reach of his loved ones.

Following the Supportive Housemate model, two flatmates were found, and Sam transitioned seamlessly into his new abode at the age of 24, a milestone celebrated with pride by his family. The sense of achievement was palpable as Sam embraced this new chapter of his life with confidence and grace.

Six months later, with Sam settled comfortably into his routine, his parents embarked on a well-deserved holiday, reassured by the robust network of support they had established. Through meticulous planning and the assistance of dedicated caregivers, they could finally enjoy a vacation, knowing Sam had a great support network around him

Ongoing monthly house meetings have been a cornerstone of their arrangement, providing a platform for open communication and problem-solving. With Sam at the centre of discussions, along with his supportive flatmates, family, and caregivers, they tackled challenges head-on, ensuring a harmonious living environment for all.

Reflecting on their journey, Sam's family shared invaluable advice gleaned from their experience. They emphasised the importance of building a strong support team, reaching out for assistance, and fostering reciprocal relationships with his flatmates, neighbours and friends. Practical tips, such as organizing household services and maximising available funding, underscored their commitment to Sam's wellbeing.

Sam himself offered simple yet profound advice: "Just do it. You will love it!" His unwavering optimism and courage served as inspiration for others considering this important rite of passage.

Our Stories

Charlie's Story

Charlie was the first child with Down syndrome to attend his local school in a small regional town in NSW. It wasn't an easy start. Initially, his parents were encouraged to check out the 'special/ support unit' at the public school instead, where most of the children with Down syndrome had historically been directed. This is the message parents like Pearl and Adam so often receive from schools with only the best intentions: your child doesn't really belong here, try somewhere else.

But Charlie's parents were prepared for this response and were able to provide the school with evidence-based data to support their decision – and gratefully, the school accepted Charlie.

Over the years, Pearl and Adam worked collaboratively with the school, willing to try new approaches, equipping new teachers with supporting notes and ensuring the values of his school were embedded at home too.

Today, Charlie loves going to school and has maintained meaningful, long-term friendships along the way. His teachers and aids have been exceptional in the support they've provided, providing regular and open communication and offering multiple planning meetings at the start of each school term. Despite the school's early reluctance to take Charlie on, there's been a significant shift in mindset, and Charlie's community has come to recognize and celebrate the valuable role he plays in the community.

The message they receive from his school now? 'Thank you for gifting us with Charlie's presence.'

Without accurate research and information, many schools would not so warmly welcome children like Charlie, assuming that his needs couldn't be met in a conventional schooling environment. Here, at Down Syndrome NSW, we see our role as vital in supporting parents like Charlie's to have these difficult conversations with schools, change the narrative surrounding children with Down syndrome and stand behind their choices for inclusive education. For Pearl and Adam, inclusive education means no child gets left behind, and we're committed to making that vision a reality.

> "It is just wonderful to know that the team at Down Syndrome NSW are doing their utmost to support our kids in all aspects of life stages. Their commitment to research, innovation, collaboration and education/support for all involved is incredibly reassuring and inspiring."
>
> **– Pearl and Adam**

Teresa and Ben's Story

Ben's experience of Inclusive Education in high school has been a work in progress, with a combination of one-on-one classes in learning support and engaging in mainstream for music, drama, assembly, house and mentor groups and swimming.

Now in Year 9, Ben has chosen music and drama for his electives and loves these subjects. He is fortunate to train at the amazing pool the school has and use their gym with his physio. The teachers in learning support are very dedicated, caring and patient – ensuring Ben is best positioned for an enjoyable and meaningful school experience.

Although very social and active outside of school, forming meaningful school friendships can still have its challenges. However, Ben is highly involved in his wider community and has a range of interests that he enjoys. His school community has been caring and willing to look out for him, checking in on how he's going and appreciating the efforts he's making. Ben feels included in school sporting competitions in swimming and his teachers are happy to modify camps and excursions if needed so he doesn't miss out.

For Ben's parents, a core component of Inclusive Education has involved collaboration and openness with his school. Since Ben started High School, his school has been part of planning meetings with Ben's parents and therapists to help determine his goals each term, making adjustments to the curriculum and providing support where required. They welcome input, observations and visits from specialists, willing to adapt to suit Ben's needs.

For young people like Ben, Down Syndrome NSW is dedicated to providing ongoing support and resources to support families and schools, while ensuring the best outcomes for the student.

Our Inclusive Education Initiative is Primary and Secondary support program for mainstream and special schools aimed at building capacity of schoolteachers, principals, SLSOs as well as connecting key stakeholders of family, therapists and behavior support, with the student firmly at the centre.

"Down Syndrome NSW has been a great support and resource since Ben's birth from Welcome Packs, the parent line, information webinars, raising the profile and value of people with DS, and offering Ben some paid work to assist at an event now he's a teenager."

- Ben's mum

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