

down syndrome

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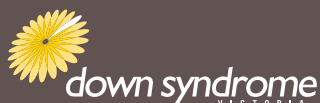
down syndrome
VICTORIA

Preface



Down Syndrome Victoria has a member library of publications on a range of topics related to Down syndrome and maintains an extensive website reference library at www.downsyndromevictoria.org.au

Please contact Down Syndrome Victoria for more detailed information on any of the topics covered in this brief introduction to Down syndrome. We will be pleased to offer you more information and answer any questions you may have.



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**Down syndrome
is a genetic
condition.**

**It is not an illness
or disease.**

1

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. The DNA in our chromosomes determines how we develop. Down syndrome is caused when there is an extra chromosome. People with Down syndrome have 47 chromosomes in their cells instead of 46. They have an extra chromosome 21, which is why Down syndrome is also sometimes known as trisomy 21.

The DNA in our chromosomes determines how we develop. You can read more detailed information about the genetics of Down syndrome on page 53.

Although we know how Down syndrome occurs, we do not yet know why it happens. Down syndrome occurs at conception, across all ethnic and social groups and to parents of all ages. It is nobody's fault. There is no cure and it does not go away.

Down syndrome is the most common chromosome disorder that we know of. One of every 700-900 babies born worldwide will have Down syndrome. This number has not changed a lot throughout the entire time that statistics have been collected. Down syndrome is not a new condition. People with Down syndrome have been recorded throughout history.

People with Down syndrome have:

- some characteristic physical features
- some health and development challenges
- some level of intellectual disability.

Because no two people are alike, each of these things will vary from one person to another.

A test for Down syndrome can be carried out before a baby is born. Down syndrome is usually recognised at birth and is confirmed by a blood test. It was named after Dr John Langdon Down who first described it.

You can find out more about the physical features of Down syndrome on page 56. Common health and medical matters are explained on page 58.





What does it mean to have Down syndrome?

Most of the young 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 a lot of support.

Having an intellectual disability

Down syndrome is the most common cause of intellectual disability that we know of. Everyone who has Down syndrome will have some level of intellectual disability. There will be some delay in development and some level of learning difficulty. Because everyone is unique, the level of delay will be different for each person.

When a baby is born, there is no way to tell what level of intellectual disability the child may have. Nor can we predict the way in which this may affect a person's life. However, we do know that having Down syndrome will not be the most important influence on how that person develops and lives their life. Instead, what happens after

Having Down syndrome will not be the most important influence on how that person develops and lives their life.

birth will be much more important and family, environmental, cultural and social factors will shape their life, just like everyone else.

For many people with Down syndrome, speaking clearly can be difficult. Although a lot of people with Down syndrome speak fluently and clearly, many will need speech and language therapy to achieve this. Very often, people with Down syndrome can understand a lot more than they can express with words. This often means that their abilities are underestimated, which can make them feel frustrated.

Some people with Down syndrome will find it very difficult to develop language skills and speak clearly. This may be made worse by hearing loss.

Living an ordinary life

People with Down syndrome are not fundamentally different from anyone else. They have the same needs and aspirations in life that we all do, including:

- a good place to live
- meaningful employment
- the opportunity to enjoy the company of friends and family
- intimacy
- having a role in our community.

However, achieving these goals is harder for people with Down syndrome than it is for everyone else. Many people with Down syndrome are likely to need some level of support to help them achieve the kind of life that most people take for granted.

In the past, many people with Down syndrome have not had the opportunity to develop to their full potential. Often, they have been separated from the rest of the community, living in segregated settings such as care institutions. Low expectations were placed on them and there were limited opportunities for learning and personal growth.

Today we recognise that growing up in families and communities, with the same rights and responsibilities as everyone else, is vital to the development of people with Down syndrome.

To be a part of a community you have to be in it. This means that people who have only experienced life in a segregated setting may find it difficult to be included in the general community. Life for people who grow up being included in families and communities will be very different than it has been for those who have always lived in care facilities.

Encouraging children with Down syndrome to go to a school with their peers from their community has many benefits. It opens the way for a smooth transition to adulthood and encourages meaningful inclusion in the community.

People with Down syndrome need opportunities to reach their full potential, like we all do. When given these opportunities, they become valued and productive members of their families and the community.

**To be a
part of a
community
you have to
be in it.**

Being an individual

One of the greatest challenges that people with Down syndrome face is the attitudes of other people who do not understand what it means to have Down syndrome. Despite much change, many people still don't see the individual person. Instead they just see 'Down syndrome' and expect everyone with Down syndrome to be more or less the same.

People with Down syndrome are very different from each other, just as we are all different. Every person with Down syndrome is unique, with their own talents, abilities, thoughts and interests. And, like everyone else, people with Down syndrome have strengths and weaknesses. While one person may read very well but find basic mathematics difficult, another might be a first-class cook and live independently in the community, but will have to work hard to speak clearly. Family passions, culture, interests and skills are also likely to be shared by people with Down syndrome, as they may be by other members of the family.

People with Down syndrome do not all look alike. In fact, people with Down syndrome look more like other people in their own family than they look like others with Down syndrome. Although there are some physical features associated with Down syndrome, there is large variation in how many of these features an individual may have. For some people, one feature may be very prominent while in another it may not exist at all. Importantly, the physical characteristics of Down syndrome that a person may have do not tell us anything about that person's intellectual ability.

Another common misconception is that all people with Down syndrome are happy and affectionate. People with Down syndrome experience all the same emotions as everyone else.

They get happy, sad, embarrassed, frustrated, thoughtful and fall in and out of love, just as we all do. They may, however, find it difficult to express their feelings in words. This can lead to frustration and the expression of feelings through behaviours.

You can find out more about the physical features associated with Down syndrome on page 56.

PERSONAL PERSPECTIVES

Paul Matley, 33



"I have just turned 33 and have been living in my flat in Northcote for five years. I'm an activity assistant in a nursing home in Coburg, which means I include the elderly residents in the activities. I do that part time. I do other jobs: I am the MC, master of ceremonies, at spring festivals, dance parties and other events in North Melbourne Town Hall. The last one I did was the '50s Swing Dance Party at the hall. I'm also an actor for the theatre companies called Rawcus and Weave Movement and a volunteer assistant and patron of e.motion21 dance group.

I act, dance, sing and carry on. I talk a lot. I socialise. I mingle.

In free time I catch up with family, friends and cousins. With family, we go out to Northcote Social Club for a drink and a meal. And I have had a girlfriend since 2008. Her friend introduced us.

Down syndrome is just another part of who I am. Everyone has a learning ability – it's just an ability that everyone has – including me. People like me have the power to do anything we want to do and to have a say and have a fair go. I'm just like everybody else. There are no good or bad things about Down syndrome, not at all. I just take it as it comes and go with the flow.

I would say to young people with Down syndrome that you need time to grow and just get out there. Seize the day, the moment! Do whatever you want to do. Don't let anyone tell you you're different ... you are who you are, exactly like everyone else. I would like to see people with Down syndrome keep up with their independence. But ask questions when you need help and help will come to you. And love will come to you too."



You can find out more about the health issues that people with Down syndrome may experience on page 58.

Some medical and health matters

As recently as the 1950s, life expectancy for people with Down syndrome was as low as 15 years of age. In recent times, progress in medical and social sciences has very much improved the health and the quality of life enjoyed by people with Down syndrome. In Australia today, most people with Down syndrome will enjoy a long and healthy life.

There are some common health issues and some more serious medical conditions that are more likely to occur in people with Down syndrome than in other people. People with Down syndrome often have lowered general immunity compared to the general population. This means that they may be more susceptible to infections and common illnesses, especially in early childhood. However, a diagnosis of Down syndrome does not mean that someone cannot have a healthy life. Some people with Down syndrome are very fit and healthy, while others experience a range of health issues.

Regular health checks may be required for specific issues. Living a healthy lifestyle is important, including keeping fit and getting regular exercise.

Research suggests that people with Down syndrome burn fewer calories in doing the same activities as the general population¹. This means that even with a healthy diet, there can be a tendency for both children and adults with Down syndrome to become overweight. An active lifestyle with plenty of physical activity helps to counterbalance this tendency and encourages general health and fitness.

A small percentage of people with Down syndrome will require a high level of living support. This may be because of complex health issues or a greater degree of intellectual disability.

¹ Medlen, J (1996) 'Looking at metabolism' *Disability Solutions* volume 1, issue 3



The most important influence on early development is daily interaction and activities within the family.



2

A BRIEF OVERVIEW OF LIFE STAGES

Early years

First and foremost, babies with Down syndrome are babies. They need the same things, especially love and stimulation, that all babies need. They grow and develop in much the same way as other babies. However, they usually reach milestones in development – such as sitting, crawling, walking and talking – at a slower rate.

It is a good idea to make sure that your baby has regular health and development checks. This will allow the professionals that you work with to identify and address any issues as soon as possible.

Although the most important influence on early development is daily interaction and activities within the family², babies with Down syndrome benefit also from structured learning opportunities. Families are encouraged to access the early learning and intervention services that are available from infancy. This will support

² Mahoney, G et al (2006) 'Responsive teaching: early intervention for children with Down syndrome and other disabilities' *Down Syndrome Research and Practice* 11(1) 18-28

the development of some of the most important early childhood skills. Early learning and intervention specialists work with families to encourage learning and development in the daily life of the child.

Areas of intervention include physiotherapy, occupational therapy (OT), speech and language therapy and early childhood intervention.

Physiotherapy assists with the development of:

- muscle tone
- motor control
- the balance our body needs for gross motor skills such as crawling and walking.

Occupational therapy assists with the development of:

- the way our hands work
- the way our eyes see and our brain perceives the world around us
- the way different parts of our bodies work together, such as hand-eye coordination
- the fine motor skills we need for moving our fingers and hands
- dressing and other self-care tasks.

Speech and language therapy assists with the development of:

- the muscles we use for speaking
- speech articulation skills
- language and communication skills
- feeding.

Early intervention teachers assist with the development of:

- the learning, social and play skills needed for early childhood activities.

Most children with Down syndrome will experience some delay in all areas of their development. However, the degree of delay will vary in each individual and will not be the same across all areas.

PERSONAL PERSPECTIVES

**Carolyn
McDiarmid –
mother of
Clem, 4**



“When I found out Clem had Down syndrome, 28 weeks into my pregnancy, I wish now I hadn’t been so sad and distressed. No one was able to tell us what life or he would be like. They seemed to be good at telling us all he wouldn’t be able to do or all the medical issues he’d have. They weren’t able to tell us all he’d be able to do, how much we’d love him, how he would just fit in with our family or the joy he’d bring to so many people.

When I think about the disadvantage I thought my other children would have because of Clem, I almost laugh. What greater gift can this family have? We learn about acceptance, diversity, ability, advocacy and many other things without even leaving the house.

Clem was born with a ‘pelvic’ kidney, meaning his right kidney is lower than normal and doesn’t drain properly. This has meant he’s had many urinary tract infections, the first being at three weeks old. Earlier on, it felt like I was taking him from one appointment to the next. But life has settled down a lot in the last year.

We feel we’ve been well supported with Clem. Both sides of the family accept and love Clem without question, as do our friends generally. Clem’s early-intervention program, with his physiotherapy, speech therapy, occupational therapy and education therapy for kinder, has also helped get him off to a great start. He’s so lucky to be born now because of the level of early intervention available to kids with disabilities.

My hopes and dreams for Clem are the same as they are for my other boys. I want them to grow into fine young men with integrity. I want them to be grounded in their faith. I want them to be happy in their life. I want them to have respect for themselves and for those around them.”

Speech and language development are commonly the areas of greatest delay. Some people have great difficulty speaking clearly and fluently. The difficulties that some people with Down syndrome can experience are related to a combination of physical factors, including muscular development and the activity of the brain that is required to produce clear speech.

Research shows that children with Down syndrome benefit a great deal from learning a system of keyword signs from very early in life. Key word signing helps to alleviate some of the frustration associated with taking longer to learn to speak (which in turn can lead to difficult behaviour). It also supports other areas of learning and helps to overcome many of the issues related to delayed language development. One widely used keyword vocabulary sign system is called Makaton.

Some children also benefit from augmentative systems, such as picture symbols, to enhance early communication. Many children with Down syndrome can learn to sight read simple words from an early age and this has been shown to assist the development of speech and language skills.

Both learning keyword signing and early reading can be pursued through early intervention services, or parents can access programs to teach their child at home. All children with Down syndrome will benefit from regular and ongoing speech therapy.

Because it takes longer for people with Down syndrome to learn things, skills that seem to happen effortlessly or automatically in other children may need to be carefully taught. New learning often requires more structure and greater repetition. New skills may need to be broken into smaller steps and repeated several times. A child with Down syndrome should be encouraged to learn all the things that other young children learn. And, as with all children, early learning forms the foundation for the skills and knowledge we need later in life.

Socially appropriate behaviour should be encouraged and expected right from the beginning. Children with Down syndrome benefit from boundary setting in the same way as other children. Parenting a child with Down syndrome will, in many ways, not be very different

- from parenting any other child. Good parenting practices
- apply to all children. And as with all children, consistency
- of approach is important.

- Most babies and young children can and do attend
- childcare centres, playgroups and pre-school settings
- alongside children of the same age. They will learn a
- great deal from joining in with other young children.



PERSONAL PERSPECTIVES

Shehani De Silva – mother of Akna, 9



“Akna is in grade 4. She has an integration aide working with her and takes part in almost all school activities. A big step for her last year was when she went on her first camp overnight with her school friends. She had a ball I am told and only asked for me once. This was a huge challenge for us as parents; it was the first time she was away from us and also, culturally, it was a challenge for us to send a child, especially a girl, on a night away. But this is in keeping with the goal we have for Akna, which is to get her to experience life to the fullest and to be able to go about it by herself one day.

Apart from the excursions, her other favourite time in school, I think, is when they learn the dance routine for the yearly concert.

She is sometimes a challenge for her school but they are happy for her to participate in any school activity and will give a listening ear anytime to any of our concerns. They have given her the opportunity to work with a speech therapist once a week. And an OT and a special education professional visit every term to support her teaching team.

A high point for me is when I see Akna going to school and getting the opportunity to experience everything like any other child and hoping she will be able to continue on this journey and enjoy it.”

School years

There is a very wide range of academic achievement in students with Down syndrome. With the right support, most children with Down syndrome learn to read and write and attain a variety of other academic skills.

A particular profile of learning strengths and weaknesses associated with Down syndrome has been developed³ and this will assist teachers in making appropriate modifications to their programs.

People with Down syndrome generally take longer to learn new things. New skills may need to be broken down into smaller steps than for other learners and more repetition may be needed to retain learned skills. Children with Down syndrome may require more structure in their activities so that they can work independently in class.

People with Down syndrome generally have some difficulties with short-term auditory memory. We use our short-term auditory memory to remember information we hear, for long enough to use that information straight away. Difficulties with short-term auditory memory affect learning spoken language, learning from listening and developing abilities in reasoning and problem-solving.

On the other hand, people with Down syndrome do not usually have difficulty with long-term memory and have a relatively strong visual memory. This means that students with Down syndrome do well with visual learning strategies and will benefit greatly from visual aids, cues and reinforcements.

Children with Down syndrome can attend the school of their parents' choice. In the past, many young people with Down syndrome have attended separate schools for students with intellectual disabilities. However, research shows that the majority of children with Down syndrome make the best progress when they are educated in

³ The learning profile developed by Prof Sue Buckley and colleagues at Down Syndrome Education International is described in Down Syndrome Victoria's (2009) *Learners with Down syndrome. A handbook for teaching professionals* pp10-11

mainstream schools alongside their peers. In a mainstream school there are particular benefits for spoken language and social behaviour.

A student with Down syndrome is more likely to experience success in a school where inclusion is embraced and supported as part of the school culture, and where the different learning needs of all the students are acknowledged and properly addressed.

Research shows that the whole school benefits from including students with disabilities as part of the school community. A range of students in every class will benefit from strategies developed to meet the learning needs of a student with Down syndrome. The best outcomes are achieved when appropriate support is provided to teachers to fully include the student in the class.

Children with Down syndrome should be provided with additional support to access the regular curriculum. The level of support and amount of program modification required will vary from one student to another. Children with Down syndrome can be included in all school activities and should have the same expectations placed on them for good behaviour and responsibility as other children do.

There are no behaviours specific to children with Down syndrome. However, sometimes the inability to express themselves with words can lead to frustration. Instead, children with Down syndrome will try to express themselves through behaviours – sometimes undesirable ones. It is often necessary to look beyond a behaviour and find the real message that the child is trying to express. This helps to understand and deal with the behaviour. It is often because of a lack of understanding about the underlying cause of a behaviour that people with Down syndrome are labelled as being stubborn.

The gap in skills and learning between children with Down syndrome and other children will grow with age. By secondary school the gap may be quite significant. People with Down syndrome do not plateau or stop learning new skills in their

PERSONAL PERSPECTIVES

Cameron McAlpine –
father of Julian, 10



“Julian has been in the Cub Scouts for almost a year and a half. We thought it would be good for encouraging more peer interaction and giving him a way to be more involved in our local community. Also to help him learn new skills and have a shared fun activity with his older brother.

The experience has been wonderful for Julian. He comes home full of information and smiles every Monday, and then eats a bowl of strawberries.

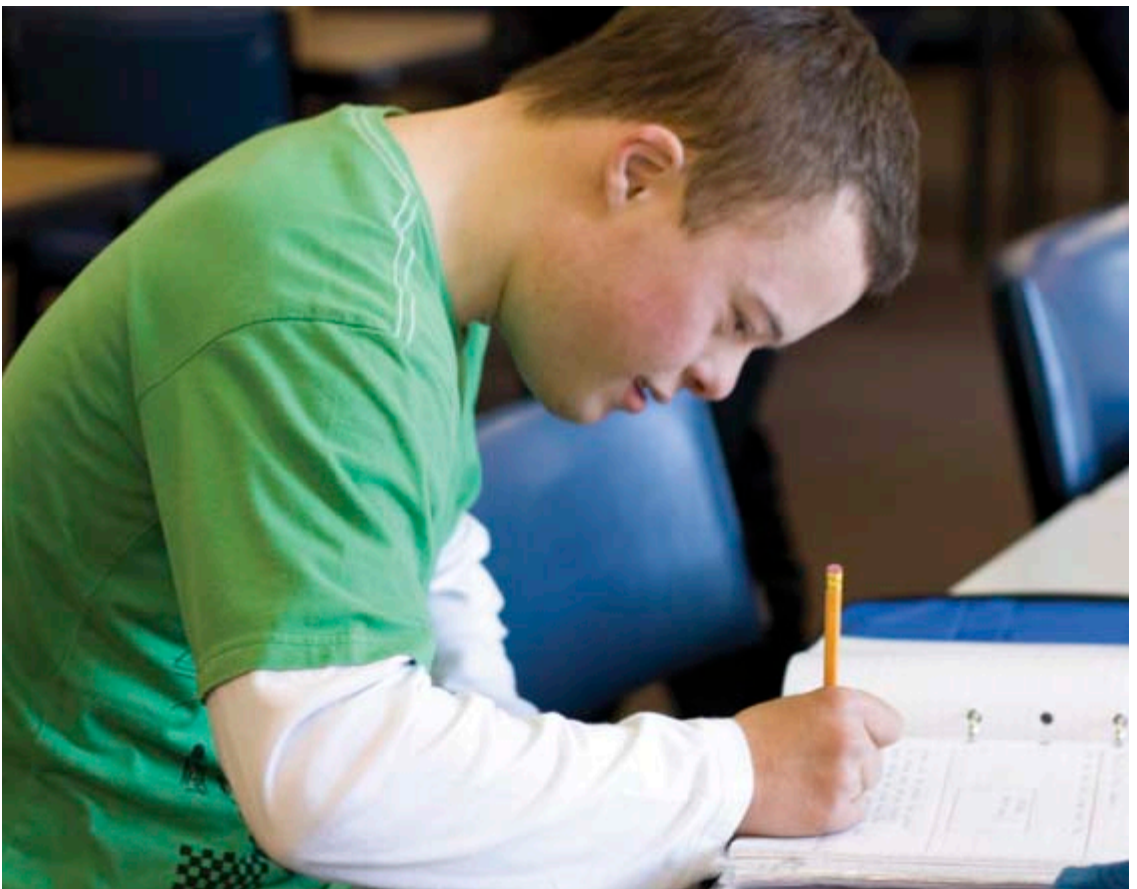
We’ve had a few difficulties, but we look for strategies to help work through them. For instance, it can be hard for Julian to keep concentrating for the entire time, and it has sometimes been hard for him to grasp the rules of the games that kick off each meeting. So we talk to the leaders regarding the program for the coming week and we prep Julian with a detailed rundown of the activities he’ll be doing.

Being a Cub Scout is great for Julian. He has become more independent, he has learnt the Cub promise, and it’s a great way of interacting with his peers and being part of his local community.”

teenage or adult years. They will continue to make steady progress and continue learning throughout life if given the opportunity to do so.

Many students with Down syndrome reach year 12 and go on to post-school training or tertiary education. Access to a range of work experience opportunities is very important in helping young people with Down syndrome to make informed choices about their life after school. Young people with Down syndrome face greater challenges in leaving school and making the transition to adult life than their peers, and more planning is likely to be needed than for other young people.

While everyone wants their child to experience success in school, it is also important to note that academic success is not the key to being able to lead an ordinary life. Many young people leave school with limited academic skills yet are well equipped to lead a happy, fulfilling and independent life as a productive member of the community.



Teenage years

Young people with Down syndrome reach puberty at about the same time as other young people. They experience the same physical changes and emotional turmoils of adolescence as do all young people.

It is important for teenagers with Down syndrome to be included in their family and the community in a way that is appropriate for their age. They can be expected to carry out a range of responsibilities, just like other teenagers. Adolescence is a time of moving towards increased independence and self-responsibility, and young people with Down syndrome need to have the opportunity to develop and practice the skills needed to make meaningful life choices.

Other important aspects of adolescence are social confidence and a positive sense of self. Socially appropriate clothes, hairstyles, interests and lifestyle become significant. Teenagers with Down syndrome, like all other teenagers, need a social life that involves time away from their family. They have an increased need for privacy and relationships with peers become more important. Social and leisure activities can have an important role in embedding young people within their peer group and the local community.

Lack of social maturity can make it hard for teenagers with Down syndrome to be socially included with teenagers of the same age

Socially appropriate clothes, hairstyles, interests and lifestyle become significant.



PERSONAL PERSPECTIVES

**Cailan Ford
Weinberg, 16**

“Being a teenager is awesome. I like to hang around with my friends and teenage boys – I have friends in school and friends from outside school. Before I didn’t like shopping at all but now I do. I like going out for dinner, going out to the movies, going swimming, going bowling, going to Luna Park. On weekends I work, I play Planet Earth, I have sleepovers, go on You Tube, hang out with friends, write songs, write, email my friends and snorkel at the beach.

At school I enjoy the choir and the school production. When I leave school I want to take care of animals or be a librarian. Perhaps I might want go to to College.”

who do not have a disability. It is therefore important to support young people with Down syndrome in developing personal care and safety skills, and age-appropriate social skills and behaviour.

Social confidence and acceptable behaviour are just as important as academic skills for a successful transition from adolescents to young adults. Developing a positive self image is particularly significant: during teenage years young people with Down syndrome start to become increasingly aware of the ways that they are different from their peers. They may need extra support to encourage and develop a positive identity and self-confidence.

Young people benefit from having friends who are the same age. It is important that they are given plenty of opportunities to socialise with other young people – including those who have a disability and those who do not. But it is not always easy to combine these friendship groups. Young people with Down syndrome may find they need to maintain two or more separate friendship groups.

The onset of puberty can bring about changes in health. As part of the transition to secondary school, a range of screening and health checks are advisable.

Health checks might include:

- thyroid hormone levels
- dental health
- diet and nutrition
- hearing
- vision.

A regular exercise and fitness regime is very important. For both health and social reasons, young people with Down syndrome should be encouraged to take part in a range of physical activities.

PERSONAL PERSPECTIVES

**Libby Mitchell –
mother of
Phoebe, 19**



“Phoebe went to mainstream primary school and is now in year 12 at a mainstream high school in Geelong.

She had a wonderful transition into high school. She was well supported, particularly by three girlfriends from primary school, and although they have made new friends and naturally moved on to a large degree, they have remained close and supportive.

Phoebe started Foundation VCAL (Victorian Certificate of Applied Learning) last year. Her subjects have included English, maths, personal development, recreation, information technology, visual communication and design, and cooking. Except for English and maths, Phoebe has attended her classes without an integration aide.

We’ve always been involved and part of all the decision-making at school. We have routinely had program or student support group meetings once a term and realistic goals have always been set. Phoebe has participated in a lot of extra-curricular activities so is well known and I believe is also well respected around the school.

Phoebe - “I love the teachers and integration aides. They’re fantastic, helpful and fun. I have lots of friends at high school and I can do lots of activities like running, swimming, cooking, drawing, maths and English. I feel really special at high school.”

At times over the past five and a half years there have been academic ‘moments’ when the school has questioned whether specialist schooling – where they manage the learning of ‘life skills’ in a more formal way – would be more appropriate. But we’ve always wanted Phoebe to remain in the mainstream system for a variety of reasons, not the least of which was that we felt she could handle it. We’ve always believed that she will have to mingle with a vast variety of people throughout life so she must practise and be equipped to handle that.

She has never ever said she has been ridiculed or unhappy at school and has yearned to get there every day with pride.

One of her friends from primary school who is her house captain sent me a text message after the school swimming sports and told me their house had won entirely due to Phoebe’s participation and awesome swimming. It wasn’t that she was fast by normal standards, it was that she had entered in as many events as possible and gained more points for her house. Her house participants certainly didn’t care that she had Down syndrome.”

Adult life

Increased life expectancy brings with it a host of new challenges, both to families and to health, education and community services. One of the greatest challenges faced by adults with Down syndrome today is being treated as adults with the same rights as everyone else.

In many countries there are now more adults with Down syndrome than children. Until quite recently, research had focussed more on supporting children with Down syndrome, but today with increasing studies relating to adulthood this imbalance is being addressed. With more information available, we have learned a lot about the ways we can support people with Down syndrome in their adult lives.

In the past, the majority of adults with Down syndrome have not had the opportunity to live in the community. Today, more and more are choosing to do this. The number of people with Down syndrome who live and play an active role in their community can be expected to rise dramatically over the next 20 years.

Achieving this goal can be complex for people who have an intellectual disability. Most people with Down syndrome will need some level of support to allow them to live and participate in the community. Some will continue to need a high level of support.

Adults with Down syndrome vary a great deal in their health, capacities and previous life experiences, and each of these factors can make a difference to the level of independence they can achieve in adult life.

Being included in a family and in the community throughout life is just as important for achieving independence as intellectual and academic skills. Growing up included within a family and the community will equip most young people with invaluable life skills and experience. These skills are likely to help establish a level of independence that will lead to a meaningful life in the community as an adult.

There are a range of services available to assist people with Down syndrome in their adult lives. Yet, while good services and funding

PERSONAL PERSPECTIVES

Anne Page –
mother of
Joanne, 33



Joanne – “Before I could not do what I wanted, I could not do the things I was good at, and I had little say. Now I have lots to say. I plan my life. And I feel great.”

Joanne has had to work harder than most to have her needs and rights met. More than 10 years ago, she had little choice over the work and lifestyle programs she took on through the Adult Training Support Services (ATSS) for people with disabilities. She felt unchallenged, undervalued and that her own interests were not being considered. But we set about changing that. We negotiated long and hard and won the right to change her funding to an individualised planning and funding package. This meant she was able to choose her own work and lifestyle programs, and the workers who would help her, to further her aim to be independent and lead a stimulating, full life.

Joanne’s week now consists of working in the community library, receiving lessons in art, computers, literacy and numeracy, cooking and photography as well as one day at an adult day training centre.

Winning an art prize at the Having a Say Conference this year and receiving \$800 for her efforts was a huge high. And, inspired by one of her planning days, she took her first overseas trip, visiting her sister in London and seeing the Mona Lisa in Paris.

As of this year individualised funding is open to all who attend ATSS’s, and

Joanne’s example of how she has been creative with hers has been used to encourage others.

Joanne is now also an advocate for self-determination. She has spoken in Sydney, Lakes Entrance, Benalla, Cobram and recently at the Down Under Institute disability conference in Lismore.”



are important – and desirable – achieving a fulfilling life does not depend on either of these.

In the past, adult life planning was mainly focussed on getting a job or daytime activity and securing accommodation. Today, life planning is more about helping a person with Down syndrome to take a full and valued role in society. Adult life planning is becoming more holistic, taking into account all aspects of the person's life, including further education, employment, interests, family life and residential options⁴.

Previously, a narrow range of options were available to people with Down syndrome. Adult life planning is today more individualised and is guided by personal preferences and choices. The aim is to support the person at the centre of the planning process to make responsible decisions and informed choices about their adult life.

Self advocacy is becoming increasingly important in the lives of adults with Down syndrome. A growing number of adults with Down syndrome are speaking up for themselves, and are increasingly represented on the governing bodies of the agencies and associations that serve and support them.

Employment

As more children and young people with Down syndrome are completing school and being given the opportunity to develop interests, skills and expertise previously denied to them, there is a steady increase in the number of people with Down syndrome entering further education and employment. Employment is not just about financial security but also about personal growth, being a contributing member of the community and living an ordinary life. Lack of meaningful employment has a significant impact on both physical and mental health.

In the past, employment for people with Down syndrome was largely restricted to adult training centres or supported employment in segregated group settings. Many people with Down syndrome, if they had access to work at all, had little choice about their workplace or type of work.

Today there is recognition that people with Down syndrome can fulfil valuable roles within a wide range of workplaces. This has led to a shift towards supporting school leavers with Down syndrome to transition into employment or further education based on individual interests and skills. While this is still far from being the norm, initiatives have been developed in many countries to support young people with Down syndrome to move into the workforce with appropriate, individualised supports.

People with Down syndrome still face several challenges in regard to entering the mainstream workforce. Work opportunities are often limited by a lack of flexibility in workplace culture and limited provision of support. And until recently there has been little emphasis on supporting people with Down syndrome to develop the skills they need to start work.

However, in the future, with increasing opportunity and appropriate support, there is no reason not to expect most young people with Down syndrome to transition successfully into the workforce with their peers when they complete their education.

PERSONAL PERSPECTIVES

Rennice Stanley
– mother of
Cameron, 29



“Cam loves being on the stage and has been involved with drama since he was at primary school in Warrnambool. When he was 20 he did a two-year TAFE course in Ignition Theatre Training with NMIT. Graduates of this course formed the Rollercoaster Theatre Ensemble and have performed street theatre, at corporate functions, festivals and La Mama Theatre. Currently the ensemble is working at St Martin’s Theatre and is developing work towards a performance for next year.

Cam has also been working part-time as a cleaner at Toll Holdings for two and a half years. Initially, he was employed there by a recruitment agency as a casual worker, but after two years was offered, and accepted, a position as a permanent part-time worker.

Once Cam started work the night shift manager worked alongside him to ensure he understood what was required. This happens with each new or different activity.

Cameron – “I am very happy with work. I enjoy having a meal with the boys (the fork lift drivers and bosses). I really like it when I drive the blue car (ride-on vacuum sweeper). I also love being on stage. I like my friends at Rollercoaster.”

He cleans the walkways and the many signs and helps others. If there is an unexpected need for another pair of hands, they will call on Cam and he responds well.

Cam has all the support he needs, his co-workers enjoy working with him and one of his managers has stated that he has been a positive influence in the workplace. Cam says he likes everything about work. He would go to work even if he were on death's door. He enjoys being with 'the boys' and he enjoys the football rivalry. Cam is a passionate Bulldogs supporter and is a cheer squad member. The only small downside to his job is missing the football when the Bulldogs play on a Friday night.

He went with a workmate to see the game in which their teams clashed. Before he left, the friend said that one of them would come home crying. Fortunately it wasn't the Bulldogs that lost.”

PERSONAL PERSPECTIVES

**Emily Rickards, 27,
and Michael Porter, 28**



“Michael and I have known each other about eight years. We live together. We are engaged and we’ll get married one day. I work as a waitress in a coffee shop once a week and Michael works at a bakery two days a week. We both go to the same training and support service the rest of the time. I love dancing and singing. I go to two different dance classes each week and I sing and play drums with The Bandits. Michael and I do most things together. We both love watching movies on TV and we’ve been to Bali twice. The first time we flew over by ourselves and we met Michael’s parents there. We really like it there.”

**Ellie Delafield,
Emily’s mother**

“While Emily and Michael have a strong support network of family and friends, they love their own home and the independent life they have. Their aim is to keep living independently.”



Personal relationships

Relationships and sexuality are as important for people with Down syndrome as they are for anyone else. The range of sexual needs will be just as varied among people with Down syndrome as in the general population. In the past, protective practices and restrictive interventions have limited the opportunity for people with Down syndrome to develop a positive sexual identity, enjoy sexual relations and develop long-term partnerships and marriages. The focus today is on appropriate education to support adults with Down syndrome as they enjoy safe and responsible sexual relations and make their own informed choices. Most people with Down syndrome who form long-term relationships choose a partner who also has a disability.

Just like other adults, people with Down syndrome need to make choices about contraception. To date, there have not been many cases recorded of parenthood in either men or women with Down syndrome and very little research has been conducted in this area. There is some evidence that both men and women with Down syndrome experience reduced fertility in comparison to the general population. However, restrictive intervention and a lack of social opportunities may have influenced past statistics in this area. Parents who have Down syndrome have given birth to children both with and without Down syndrome.

Accommodation

In the past, adults with Down syndrome frequently continued to live at home with their parents or in the house of another relative. Or, adults with Down syndrome moved from the family home into residential services offering various levels of care, where they may have had little or no independence or control over their lifestyle.

Many adults with Down syndrome neither need nor benefit from high levels of care, and they need the same access to appropriate and affordable housing arrangements as everyone else. Individual planning and self-determination are important in assessing adult housing options. More and more adults with Down syndrome today wish to live in the community. These people rightfully expect a choice of appropriate semi-supported or independent housing arrangements within their community. In the future, much work is needed and significantly more resources need to be channelled into this area.



Older years

A person with Down syndrome growing up in Australia today can expect to live to between 55-70 years of age⁵. Increased longevity in people with Down syndrome is offering up new challenges in the support we provide and the health challenges that need to be met.

In order to maintain a good quality of life in the older years, leading a full, active and healthy life is as important for people with Down syndrome as it is for everyone else. Research to date suggests a tendency to premature ageing, with associated physical and mental health issues, in people with Down syndrome. However, many of today's adults with Down syndrome have very different lifestyles to those of earlier generations. Improved quality of life, with more opportunities to be active and included in the community, may have a positive impact on ageing in people with Down syndrome in the future.

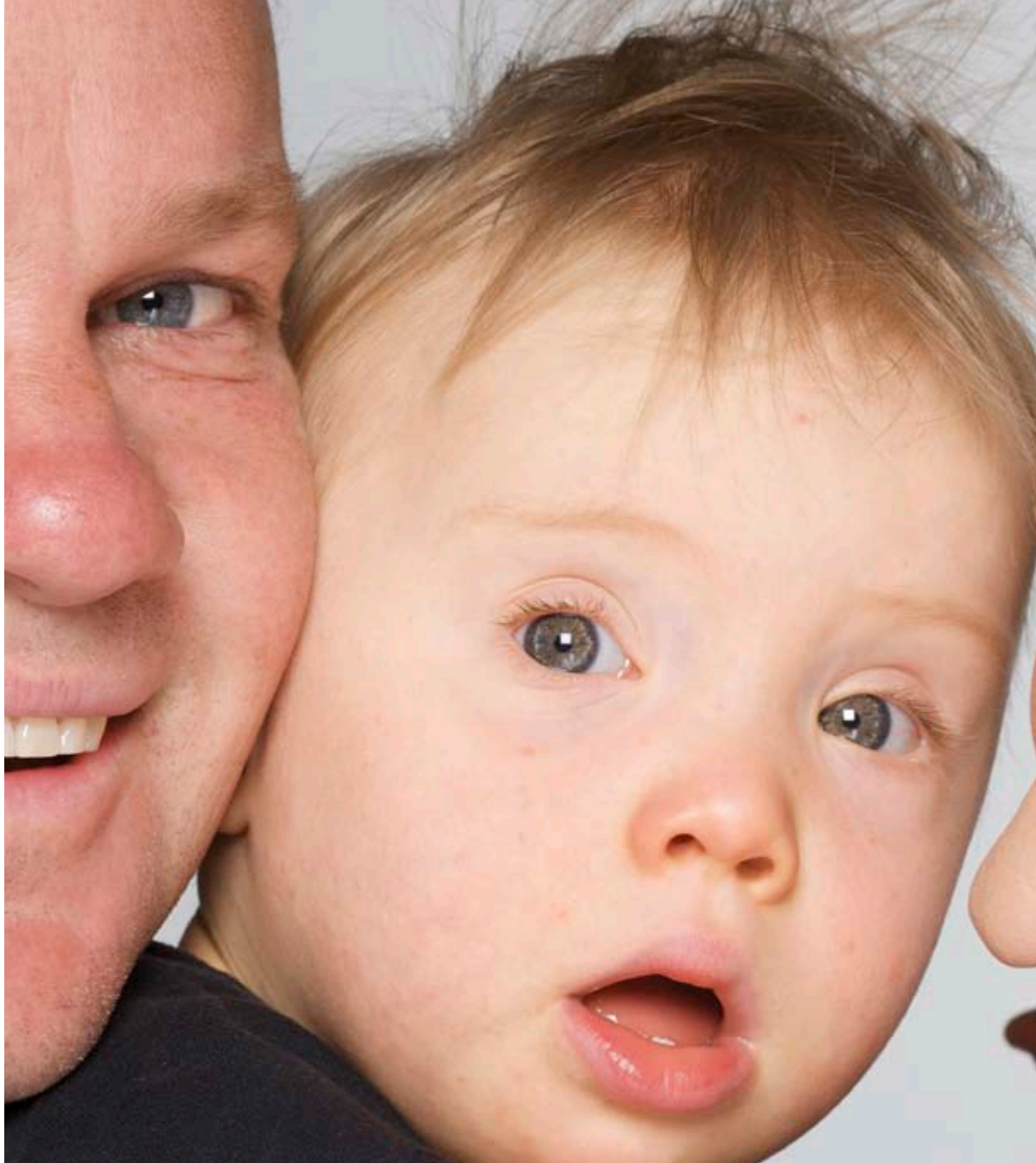
As they get older, adults with Down syndrome continue to require careful health monitoring. This relates both to general health concerns associated with ageing, and medical conditions that are more prevalent in people with Down syndrome. The importance of ongoing monitoring of mental health in adults has also been established. Both biological and environmental factors can place adults with Down syndrome at risk of emotional problems and other mental health issues⁶.

Scientists have established a complex connection between chromosome 21, of which people with Down syndrome have an extra copy, and Alzheimer's disease, which is the most common form of dementia⁷. This does not, however, mean that everyone with Down syndrome will develop the clinical symptoms of dementia. Research is continuing to determine the relationship between Down syndrome and Alzheimer's disease.

⁵ Brown, Roy *Life for adults with Down syndrome – an overview* p 12

⁶ McGuire, Dennis and Brian Chicoine *Mental wellness in adults with Down syndrome* p xi

⁷ You can find out more about this in *Down syndrome and Alzheimer's disease*, produced by Alzheimer's Australia, Down Syndrome Australia and the Centre for Developmental Disability Health Victoria (CDDVH)



Families have been the driving force behind many changes that have been made over the years.



3

FAMILY LIFE

Families are important

Family and home environment are an important part of the life of a person with Down syndrome.

For a person with Down syndrome, being included in all aspects of family life can pave the way to successful social inclusion within the community. Many of the practical and independent living skills that we carry with us into adult life come from our experience of family life.

Family members have an important role in encouraging a person with Down syndrome to show what they are capable of. If a family has high expectations of the person, better behaviour and higher levels of achievement can result. On the other hand, low expectations can limit the person's potential. Over recent decades, many more people with Down syndrome have had the opportunity to show what they can achieve and this has completely changed our understanding of what they are capable of.

Families are also crucial in providing leadership for the rest of the community. The attitude and expectations of the family are usually mirrored by the community at large. If families expect their child with Down syndrome to be treated with the same dignity and respect as everyone else, that is likely to be the outcome. If they create an impression of a person with various limitations who needs special treatment, this is most likely to be the way that person is perceived, and treated, by those around them.

Having realistic and high expectations of ongoing learning and independence help to ensure that these become the reality for the person with Down syndrome. Anyone who is constantly over-supported and helped in every aspect of their daily lives will become dependent on this kind of assistance. This is called 'learned helplessness' and has commonly been associated with Down syndrome.

Families can also have an important role as advocates for their family member with Down syndrome. By speaking up for a family member, families have been the driving force behind many of the progressive changes that have been made in the lives of people with Down syndrome over the years.



PERSONAL PERSPECTIVES

Antoinette Hammond – mother of Patrick, 8



“Patrick is a beautiful child; the innocence, honesty and earnestness with which he lives his life never fail to touch all who cross paths with him. He has an impish grin and a twinkle in his eye. He truly loves life, and participates fully in his own way in each experience that comes his way. He likes all the usual kid things; TV, sport, electronic games, animals, singing, dancing and jumping on the trampoline. He particularly loves ball sports, especially basketball and footy, which he has taken to with a fierce determination to succeed and better his abilities. He likes being in control and things going his way, loves playing up to any audience and is an absolute party animal. Patrick adores his three brothers, all of whom offer him something different. He loves being a part

of things, from playing together with his brothers, to walking off the field arm in arm with his older brother’s footy team, singing the team song he has learnt by heart.

The impact of having a child with Down syndrome in the family is enormous, yet negligible, since we know of no other life than that with Patrick. That’s just who we are as a family. I think life has been harder for all of us than it would have been if uncomplicated by disability – I feel guilty each time I have to ask Conor to help look after Patrick, so that I can manage all the boys in an unfamiliar environment. But I’m also so grateful for everything that Patrick has taught me about life, myself and other people. And I know that despite the difficulties, the other children benefit enormously from a life with Patrick too. The most significant gain is that they are relaxed in the face of disability.

Along the way we have celebrated all of his milestones heartily, not just because we have had to wait so much longer for them, but because he has had to work so hard to achieve them.”

Conor, 12, brother of Patrick

“Sometimes he’s annoying; sometimes he’s funny, and good to play with. He can make everyone laugh when they are sad. But he is a good brother. He makes all my friends laugh too.”

Families are resilient

While parents generally experience shock on learning that their baby has Down syndrome, the feelings immediately following diagnosis do not indicate the long term future for the family. The majority of families adjust relatively quickly to this new situation. Parents may experience some degree of 'appointment-stress' in the early days as a result of regular health and medical checks and early intervention appointments, but this period is usually short-lived.

Families generally find their feet and continue leading their lives, incorporating the additional needs of their child with Down syndrome into their family circumstances.

A number of studies of the experiences of families suggest that despite the very real demands of life with a child with a disability, families can and do make the adjustment, often with little disruption to family life. Families generally find their feet and continue leading their regular lives, incorporating the additional needs of their child with Down syndrome into their family circumstances.

Research shows a range of experiences that families raising a child with Down syndrome have. These are highlighted on page 45. Of course, every family is different. Some families will feel that their

lives have hardly been affected by the addition of a family member with Down syndrome, others may experience this as a life-altering event.

Almost all families report that there continue to be highs and lows, times of elation and times of despondency. Periods of transition, such as moving into school or between schools, the onset of puberty, leaving school and entering adult life tend to be where new peaks of stress and emotion occur in families. Support, either among individual families or through peer support groups, can contribute significantly to family wellbeing.

Many of the professional services available for people with Down syndrome recognise just how important families are. For this reason, many of the



Research with families of children with Down syndrome⁸

- Most families (70%) adapt and lead regular family lives.
- Most families report benefits of having a child with Down syndrome for the whole family.
- Marriage breakdown is no more frequent than for the rest of the population. It may even be less frequent.
- Brothers and sisters do not have more problems than in other families and are likely to be more caring.
- Resilient families tend to use practical coping strategies. They seek out information and services, and join parent support groups.
- Resilient families develop a supportive emotional climate and encourage open communication between family members.
- Vulnerable families can usually be identified in the first years of a child's life and will benefit from specific support from relatives, parent support groups and professional services.

services have a family-centred approach that allows families to take a lead role in directing the services that they need and use. This can help build resilience and develop independence. Sharing experiences with other families and taking part in peer support networks also help to build resilience.

Families are experts

Families quickly become knowledgeable about Down syndrome and what it means for their child and family. Maintaining a flow of clear and up-to-date information through the different life stages is important. Parents usually become proficient at researching and filtering information themselves or seeking help from support organisations and peer support networks. It is important to build open, honest and respectful communication between parents and professionals. The best support for the person with Down syndrome is achieved by combining the skills of both parents and professionals in partnership.

⁸ Buckley, Sue *Issues for families with children with Down syndrome*, p 8

PERSONAL PERSPECTIVES

Joel Deane –
father of
Sophie, 10



"We have three kids, Sophie, 9, Noah, 7, and Zoe, 3, and, yes, being their father is complicated. Child care, school, AusKick, gym classes, musical performances, late night hospital visits, juggling school holidays and work, birthdays, housework, homework, trying to remember everyone's names ... complicated. Add the fact that Sophie has Down syndrome and the complications go up a notch or two. Sophie, generally, is just like her siblings, with all the highs and lows, except the lows can sometimes seem to be a little lower. Those are the times I find toughest.

During those times, three things help. First, the knowledge that Sophie's low points are similar to those that Noah and Zoe experience, and that she can (and must) work her way out. Second, the understanding that Soph is the one who's been dealt the tough hand, not me, and that my job is to help her battle through and grow up to be an individual and (hopefully) independent woman. Third, the support of the other mums and dads of kids like Sophie because it's important to know you're not alone.

The impact of our interactions and friendships with other families has been overwhelmingly positive. We have a lot of fun, talk about a great many things besides Down syndrome and, when needed, call on each other for encouragement, information and support. The advantages are enormous, especially when it comes to sharing information about everything from the best doctors to the best schools to the best way to ensure the siblings of children with a disability don't miss out. Our relationships are priceless.

For me, the advantage of meeting other dads is that they are, in one very important respect, my peers. They know what it's like and I don't have to explain my daughter or myself, instead I can just be myself. In a complicated world, it's nice sometimes to have one less complication to worry about."

Siblings

Siblings are likely to have the longest relationship of anyone with their brother or sister with Down syndrome. They also frequently have a unique insight into this person.

Siblings may:

- have less difficulty than others in understanding their sibling's speech
- recognise an underlying cause of difficult behaviour
- see potential where others have failed to recognise it.

Many parents are initially anxious regarding the effect that having a brother or sister with Down syndrome may have on other children in their family. Sibling relationships can be a challenge in any family, and sibling relationships involving a sibling with Down syndrome will be as varied as those relationships where there is no disability involved. There is no single right way to guide these relationships although constructive family practice works as it does in other families.

Parents should be mindful that young people will tend to mirror the attitudes of the adults in the family. Other children outside the family will in turn take their lead from the attitude shown by the siblings to their brother or sister with Down syndrome. It generally helps to have open acknowledgement and discussion of Down syndrome and disability within the family. It is also important to discuss the ways in which we are all similar, and diverse, and each person's individual strengths and challenges.

It is also important to have the same fundamental expectations of behaviour, cooperation, family dynamics, responsibilities and rights for all family members.

- Siblings sometimes feel that the child with Down syndrome receives more time and attention from parents than other children in the family. It is important to acknowledge that this may appear to be the case, especially in view of additional appointments and therapy intervention in the early days.
- Siblings provide a range of benefits for a child with Down syndrome. They are a role model for language and behaviour, a childhood companion and, later, a social coach and mentor. Siblings in turn gain widened perspective from the experience of having a brother or sister with Down syndrome.
- Families need to guard against burdening a sibling with too many care responsibilities. Emphasising the need for mutual support in the family to meet everyone's needs and acknowledging and celebrating the differences in individual family members helps maintain perspective.
- All sibling relationships go through different stages as children grow. Many teenagers are likely to experience a phase in which their sibling causes them embarrassment. Parents need to be careful to handle this with sensitivity, knowing that it usually passes with time and developing maturity.
- A great deal of support material is today available for siblings of all ages. Sibling peer groups and web-based networks are also helpful.

PERSONAL PERSPECTIVES

Samar
Bakhsh, 16,
sister of
Abdullah, 20

"I'm grateful to have Abdullah as my brother; through him I've learnt the value of helping people, with and without special needs. We have a typical brother-sister relationship and it's always been like that. I don't think my relationship with Abdullah is different from my friends' relationships with their brothers; and my friends don't think so either."



**Down syndrome
is only a part of
who a person is – it
does not define that
person.**





4

CHANGING THINKING

Down syndrome is only a part of who a person is – it does not define that person. The most important thing we can do to support people with Down syndrome is to see the person first, and to treat Down syndrome as one aspect of that person. We need to think in terms of each person's qualities and abilities instead of placing our focus on what is different about them.

With the advances in health care, education and support services, and shifts in the way the general community views disability, the outlook for people with Down syndrome has altered significantly over the past 30 years or so. People with Down syndrome can today look forward to long and fulfilling lives, with increasing opportunities for self-determination and independence. Today, there is a growing focus on empowering and enabling people with Down syndrome to succeed in achieving their aspirations and dreams. We are still learning about just how much people with Down syndrome can achieve when they are afforded the same opportunities as everyone else.

The *United Nations Convention on the Rights of Persons with Disabilities*, which came into effect in May 2008, recognises disability as a social phenomenon, rather than an inherent attribute of the person. It also recognises that societies disable people. Unfortunately, for many people with Down syndrome, the biggest obstacle that they face growing up today is the society in which they live. People with Down syndrome are still routinely underestimated in their abilities and potential, and may be socially isolated because the community still has difficulty seeing past the disability to the person.

The biggest single factor that will impact on the lives of people with Down syndrome today and in the future is the degree of meaningful inclusion in the community. It has been established that inclusion in family and community life are among the most significant factors in determining the life path for a person with Down syndrome. This starts with inclusion in regular family life, continues through mainstream schooling opportunities and culminates in the person taking their place as a full and contributing member of their community.

All of us have a part to play in dismantling the social barriers that people with Down syndrome encounter, and in ensuring that Down syndrome adopts its rightful place as just another variation in a world of diversity.



5

MORE INFORMATION

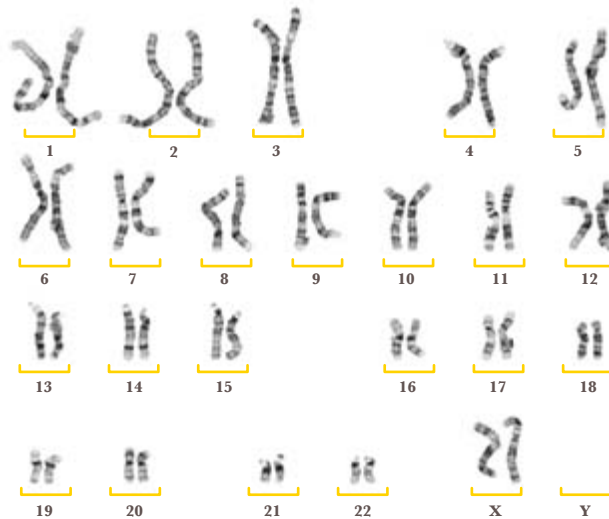
The genetics of Down syndrome

Down syndrome is a specific chromosomal disorder resulting from the presence of an extra chromosome.

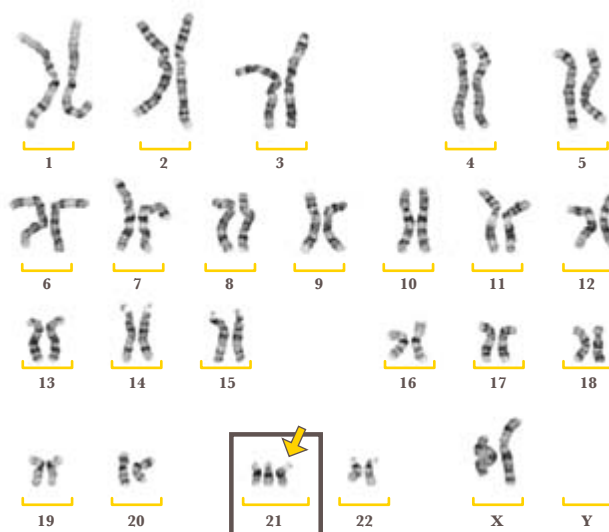
Chromosomes are structures that contain the genetic information people need to grow and develop. They are present in all the cells in our bodies. Each chromosome is made up of DNA, which contains encoded genetic instructions (genes) for the development of all the structures and functions in the body. Every chromosome contains thousands of genes. Our development is precisely controlled by our genes, so that if a person has either too much or too little chromosomal material this can have a significant effect on their development.

Each cell in the human body usually contains 46 chromosomes arranged in 23 pairs, which are labelled 1-23. For example, the 23rd pair are the so-called 'sex chromosomes' that determine whether a baby is a boy or a girl. In people with Down syndrome, an extra copy of chromosome 21 is present.

Normal karyotype (female)



Trisomy 21 karyotype (female)



Source: VCGS (Victorian Clinical Genetics Service) Pathology, Cytogenetics

There are three forms of Down syndrome.

1. **Trisomy 21:** Ninety-five percent of people with Down syndrome have trisomy 21. In this type of Down syndrome, every cell in the body has an extra chromosome 21.
2. **Mosaic Down syndrome:** One to two percent of people with Down syndrome have mosaic Down syndrome. In this type of Down syndrome, only some cells have the extra chromosome 21. The rest of the cells have the usual genetic composition. This sometimes leads to a milder level of intellectual disability and less obvious physical features of Down syndrome.
3. **Translocation Down syndrome:** Three to four percent of people with Down syndrome have translocation Down syndrome. In this type of Down syndrome, extra chromosome 21 material is attached – or translocated – to a different chromosome. This variation does not significantly change the effect of the Down syndrome. Translocation Down syndrome is sometimes hereditary.

The type of Down syndrome is identified by a blood test, usually taken shortly after birth.

Although the chance of having a baby with Down syndrome increases with maternal age, children with Down syndrome are born to mothers of all ages. Most babies with Down syndrome are born to mothers under 35 years of age, because this is the group to which the greatest number of babies are born overall.

Screening and diagnostic tests for Down syndrome are available before a baby is born.

The physical features of Down syndrome

There are as many as 120 features described in Down syndrome but no one person will have them all. Most people may have only six or seven of these features. People with Down syndrome resemble others in their family more than they resemble other people with Down syndrome. There is no known connection between the number of physical features associated with Down syndrome and the level of intellectual delay that the person experiences.

Body size

Babies with Down syndrome are usually smaller than average and therefore weigh less at birth. Children with Down syndrome grow slowly and are commonly smaller than their peers of the same age. Adult stature is also at the lower end of the general range. Body size is also influenced by the genetic characteristics of the family.

Muscle tone

Babies are frequently born with low muscle tone, which is called hypotonia. This means that the muscles are less firm. In some people, it is more noticeable than in others. Hypotonia improves with age and may disappear altogether as the child gets older. It is hardly ever noticeable beyond the teenage years. Muscle tone refers to the resistance capacity of muscles when relaxed. It is not related to muscle strength, which is normally similar to the general population.

Eyes

Nearly all people with Down syndrome have eyes that slant slightly upwards. There is often a small fold of skin that runs vertically between the inner corner of the eye and the bridge of the nose. This is called the epicanthic fold and it is often also seen in babies who do not have Down syndrome. The epicanthic fold becomes less prominent and may disappear as the child gets older. Eyes often have white or yellow speckles around the iris. These are called Brushfield spots. They do not affect vision and may also disappear in later life.



Face

The face is often rounded and tends to have a flat profile when viewed from the side.

Head and neck

The back of the head may be slightly flattened and the neck is usually short and broad. There may be excess skin over the back of the neck in newborn babies but this often disappears as the baby grows.

Mouth

People with Down syndrome generally have less space in the mouth and a larger tongue than the general population. This can lead to the tongue protruding from the mouth or resting on the lower lip. Simple techniques can be used from an early age to teach children to hold their tongue in their mouth.

Hands

Hands tend to be broad with short fingers. The little finger often has only one joint instead of two and may curve inwards. There may be only a single crease running across the palm.

Feet

Feet tend to be broad and short. There is commonly a gap between the first and second toe, known as the sandal gap.

Common health and medical matters

In the past, medical conditions associated with Down syndrome were responsible for significantly lowered life expectancy. Advances in surgical and medical intervention today mean that many people with Down syndrome will not have more ongoing health care requirements than everyone else. There are, however, a number of significant health and medical issues that are more prevalent in people with Down syndrome than in others.

Congenital heart defects

A large percentage of babies with Down syndrome are born with a congenital heart defect. This is a problem with the structure of the heart. The most common congenital heart defect for people with Down syndrome is an atrioventricular septal defect. How serious a heart defect may be usually depends on how much it affects the way blood flows around the body. In some cases, heart defects will cause no problems and eventually heal themselves. However, more commonly, heart surgery is required.

Babies are checked for heart problems at birth, and then examined again at six weeks of age.

Gastrointestinal issues

Some babies with Down syndrome are born with conditions which affect the stomach and gastro-intestinal system. These conditions include duodenal atresia, imperforate anus, Hirschsprung's disease and trachea-oesophageal fistula. Some of these conditions are evident at birth, and may require surgery straight away. Others are picked up by monitoring for ongoing unresolved feeding difficulties.

A relatively high proportion of babies with Down syndrome have reflux in the early days. Constipation is also common in people with Down syndrome. This can be made worse by low muscle tone in the stomach muscles.

Coeliac disease

People with Down syndrome are more likely than the general population to develop coeliac disease, which is an intolerance to

gluten. Routine blood tests are used to screen for this, and if coeliac disease is suspected, a biopsy from the small intestine is performed to confirm the diagnosis. Treatment is a diet free from gluten for life. Many people in the community have coeliac disease and a wide range of gluten-free products are today available in most Australian supermarkets.

Thyroid conditions

An over-active or under-active thyroid gland is caused by hormone imbalances or deficiencies. If left untreated, it can affect physical and mental wellbeing. The most common condition is an under-active thyroid, which is known as hypothyroidism. The symptoms of this condition include:

- lethargy
- lack of concentration
- weight gain
- dry coarse skin
- memory impairment
- intolerance of cold.

Newborn babies are tested for thyroid conditions. Ongoing screening should be done at least every two years throughout life as onset is gradual. Treatment is a thyroid supplement in tablet form.

Leukaemia

While leukaemia is more common in children with Down syndrome than in the general population, only around 1 in 100 children with Down syndrome will develop the disease. Onset is usually between the ages of one and four years. A particular form of transient leukaemia can occur in newborns with Down syndrome, in which the changes to blood and bone marrow associated with leukaemia appear, but then disappear again without treatment.

Upper respiratory tract infections

Children with Down syndrome tend to have relatively narrow ear and nasal passages. As a result, some children may be more prone to coughs, colds and ear infections than other children, especially

in early childhood and at times when there is an increase in mixing with other children, such as starting playgroup, pre-school or primary school. Older people are also more susceptible to chronic respiratory conditions.

Ears

Children with Down syndrome tend to have narrower Eustachian tubes than the general population. Eustachian tubes are the part of the ear that drain fluid from the middle ear.

Narrow Eustachian tubes can mean that the fluid cannot be drained easily. This can lead to blockages that cause ear infections and hearing loss. Blocked Eustachian tubes can also develop into a condition called 'glue ear'. Glue ear often clears up spontaneously as a child grows and the tubes enlarge naturally. However, the problem may be recurrent throughout childhood. If it becomes a persistent problem, it can be treated by surgery to insert tiny plastic tubes, called 'grommets,' into the ear that allow the fluid to drain.

For people with Down syndrome, hearing loss has been a significant health issue in the past. This was due partly to untreated ear issues in early life. It is less of an issue today with more consistent monitoring. Hearing should be monitored at least every two years throughout life. People with Down syndrome experience some degree of hearing loss with ageing, as do the general population.

Eyes

Visual defects are common but correctable and people with Down syndrome should have their vision checked regularly throughout life. Both long and short sightedness are more common than in the general population. Other conditions which occur more commonly include:

- squints
- nystagmus – involuntary movements of the eye that blurs vision
- cataracts – a clouding of the lens inside the eye
- keratoconus – vision becomes impaired because the cornea changes shape

A decline in sight with ageing occurs as in the general population.



Atlanto-axial instability

In a small minority of children, there is increased mobility of the atlanto-axial joint. This is the joint that connects the two neck bones directly under the skull (known as atlas and axis). This condition is known as atlanto-axial instability. In rare cases, atlanto-axial instability can lead to dislocation of the two bones which can cause compression of the spinal cord. This usually occurs gradually but can also occur suddenly.

Signs of spinal cord compression include:

- neck pain
- restricted neck movement
- unsteadiness in walking
- deterioration in bowel and bladder control.

Dental issues

The milk teeth of children with Down syndrome appear later than in other children, and they tend to keep them for longer, which results in increased wear and tear. Adult teeth may be irregularly spaced. Gum disease is more common in people with Down syndrome and regular dental check-ups are advisable. Ongoing and specific instruction may be necessary to maintain good oral hygiene.

Podiatric issues

Low muscle tone can lead to problems with the structure of the foot, poor gait and mobility issues. These can be corrected by the use of good supporting footwear and orthotics.

Skin and hair issues

The majority of people with Down syndrome have dry skin and hair, and routine monitoring for associated irritation, inflammation and infection is advisable. There is also increased likelihood of a number of common skin and hair disorders, such as atopic dermatitis, fungal and yeast infections, impetigo, eczema and alopecia.

Sleep apnoea

Sometimes, upper airway obstruction can lead to disrupted sleep patterns and resulting fatigue, stress and behaviour issues. Sleep apnoea can be identified by surveying sleep patterns. Treatment may involve the removal of tonsils and adenoids.

Alzheimer's disease

Alzheimer's disease is the most common form of dementia. It occurs more frequently and at a younger age in people with Down syndrome than in the general population, with the most common age for diagnosis being the mid 50s. It is characterised by a gradual change in ability to think, remember and perform daily living tasks.

General considerations:

Growth

People with Down syndrome grow more slowly than others. They are usually short in stature and may be prone to weight gain, partly as a result of metabolic differences. A physically active lifestyle is recommended for general health and to combat this tendency.

Lowered immunity

Many people with Down syndrome have lowered general immunity in comparison to the general population. This means that, especially in early and older years, extra care may be needed to ensure that common medical ailments are promptly attended to in order that they do not develop into more serious health issues.

Reduced expression of pain

People with Down syndrome do not always localise pain very well and may also not be able to express clearly the level of pain they may be experiencing. Care should be taken not to underestimate the discomfort a person may be in. The level of complaint may not adequately reflect the serious nature of a health or medical issue.



REFERENCES

Brown, Roy (2004) *Life for adults with Down syndrome – an overview* Down Syndrome Education International

Buckley, Sue (2000) *Living with Down syndrome* Down Syndrome Education International

Buckley, Sue (2002) *Issues for families with children with Down syndrome* Down Syndrome Education International

Buckley, Sue and Gillian Bird 'A comparison of mainstream and special school education for teenagers with Down syndrome: Implications for parents and teachers' *Down Syndrome Research and Practice* 9(3) 54-67
www.down-syndrome.org/reports/295/reports-295.pdf

Buckley, Sue and Ben Sacks (2001) *An overview of the development of infants with Down syndrome (0-5 years)* Down Syndrome Education International
www.down-syndrome.org/information/development/early/

Buckley, Sue and Ben Sacks (2001) *An overview of the development of children with Down syndrome (5-11 years)* Down Syndrome Education International
www.down-syndrome.org/information/development/childhood/

Buckley, Sue and Ben Sacks (2002) *An overview of the development of teenagers with Down syndrome (11-16 years)* Down Syndrome Education International

Capie, Angus, Anna Contardi and Diane Doehring (2006) *Transition to employment* Down Syndrome Education International

Chicoine, Brian and Dennis McGuire (2010) *The guide to good health for teens and adults with Down syndrome* Woodbine House

Cohen, William I, Lynn Nadel and Myra Madnick (eds) (2002) *Down syndrome. Visions for the 21st century* John Wiley

- Couwenhoven, Terri (2007) *Teaching children with Down syndrome about their bodies, boundaries and sexuality. A guide for parents and professionals* Woodbine House
- Down Syndrome Australia, Alzheimer's Australia and the Centre for Developmental Disability Health Victoria (2008) *Down syndrome and Alzheimer's disease*
- Down Syndrome Victoria (2009) *Learners with Down syndrome. A handbook for teaching professionals*
- Luecking, Richard (2010) 'The art of possibility: seamless transition from school to work and adult life' *Voice* vol 1 (3) 7-9
www.dsav.asn.au/resource/VoiceLueckingSept2010.pdf
- Mahoney, Gerald, et al (2006) 'Responsive teaching: early intervention for children with Down syndrome and other disabilities' *Down Syndrome Research and Practice* 11(1) 18-28
www.down-syndrome.org/perspectives/311/
- Medlen, Joan E (1996) 'Looking at metabolism' *Disability Solutions* 1/3 www.disabilitycompass.org/publications/back-issues-of-disability-solutions/volume-one/1-3.pdf/view
- McGuire, Dennis and Brian Chicoine (2006) *Mental wellness in adults with Down syndrome. A guide to emotional and behavioral strengths and challenges* Woodbine House
- Pueschel, Siegfried M (ed) (2006) *Adults with Down syndrome* Paul H Brookes Publishing Co
- Selikowitz, Mark (2008, 3rd edition) *Down syndrome: the facts* Oxford University Press
- Simons, Jo Ann (2010) *The Down syndrome transition handbook. Charting your child's course to adulthood* Woodbine House
- UN Convention on the Rights of Persons with Disabilities* (2008)
www.un.org/disabilities

About Down Syndrome Victoria



Down Syndrome Victoria is the state-wide peak membership organisation representing people with Down syndrome and their families. It is a not-for-profit organisation established in 1978 to provide support, encouragement, information and resources to people with Down syndrome, their families and the broader community.

We are a whole of life service offering:

- personal support and information to families
- advocacy, information, support, mentoring and training for adults with Down syndrome
- an education support service to assist students with Down syndrome and their teachers in mainstream schools
- peer support groups around the state
- an annual Family Fun Day and other events
- annual conference and education and information sessions
- a quarterly journal
- information and professional development for professionals and service providers.
- a library of resources relating to Down syndrome.

Down Syndrome Victoria is an active member of the Down Syndrome Australia network of state associations. Down Syndrome Victoria relies on public and private sector support to fulfil its mission of empowering people to achieve a lifetime of meaningful inclusion in the community.

Down Syndrome Associations around Australia

Down Syndrome Victoria

Phone 03 9486 9600 or 1300 658 873

www.downsyndromevictoria.org.au

Down Syndrome New South Wales

Phone 02 9841 4444

www.dsansw.org.au

Down Syndrome Association of Queensland Inc

Phone 07 3356 6655

www.dsaq.org.au

Down Syndrome Society of South Australia Inc

Phone 08 8369 1122

www.downssa.asn.au

Foundation 21 (South Australia)

Phone 08 8367 8022

www.f21.org.au

Down Syndrome Association of Western Australia Inc

Phone 08 9368 4002 or 1800 623 544

www.dsawa.asn.au

Down Syndrome Association of Northern Territory Inc

Phone 08 8985 6222

email dsant@octa4.net.au

Down Syndrome Association of ACT Inc

Phone 02 6290 0656

www.actdsa.asn.au

Down Syndrome Tasmania Inc

Phone 1300 592 050

www.downsyndrometasmania.org.au

