

Understanding Down Syndrome:  
Capturing family experiences through research

**Authors:** Helen Leonard, Jenny Bourke, Marie-Louise Collins, Sonya Girdler, Ami Bebbington, Seonaid Mulroy, Katherine Bathgate, Kitty Foley, Kate Povee, Kelly Thomas, Alinta Oates, Elizabeth Geelhoed, Aditya Deshpande, Olivia Knight, Divia Pillay, Carol Bower

## Acknowledgements

There are many contributors to the Down syndrome research project at the Telethon Institute for Child Health Research.

Every Family Counts: A huge thank you to the families who responded to the questionnaires. Each person's experience makes an important contribution to the knowledge base. Your contribution goes beyond families in WA: it will benefit future families, those yet to grow through the age of your child, as well as family and professional stakeholders nationally and internationally. It is vital that every family participates in order to produce valid and accurate representation in our community. These studies are part of a larger body of population-based research on intellectual disability using linked population data sources in WA.

We would like to acknowledge the wonderful support provided by Down Syndrome WA and the Disability Services Commission.

We would like to acknowledge the funders of the Transition Study which include:

ARACY, Australian Research Council, Disability Services Commission and Department of Education; as well as our collaborators Edge Employment Solutions and Down Syndrome WA.

The dedicated contribution from supervisors and research students from Edith Cowan University, Curtin University, and University of Western Australia which enabled much of the research to be undertaken.

We appreciate the generous input provided by members of the Consumer Reference Group.

## Contents

### Introduction

Down syndrome research in Western Australia – an outline .....	2
On the world stage.....	5

### Medical

Medical conditions and health service utilization of children with Down syndrome .....	
Health conditions in school-aged children with Down syndrome.....	
Direct health care costs of children and adolescents with Down syndrome .....	
Dental health in children with Down syndrome.....	
Puberty, menstruation and Down syndrome.....	

### Family

Physical and mental health in mothers of children with Down syndrome.....	
An investigation of the role of spirituality and organized religion in supporting parents of children with Down syndrome .....	
Leisure participation for school-aged children with Down syndrome.....	
The impact of having a sibling with Down syndrome.....	
Family functioning in families with a child with Down syndrome .....	

### Transition

Transition from school to post-school for young adults with Down syndrome.....	
--	--

## Down syndrome research in WA

So much has changed and continues to change in the lives of people with Down syndrome and their families. Identifying evolving issues is the work of researchers at the Telethon Institute for Child Health Research who, since 1997, have been investigating a broad range of topics relating to Down syndrome under the leadership of Dr Helen Leonard. The initial study in 1997 focussed on school-aged children and two hundred and twelve families (80% of those eligible) with a child with Down syndrome responded to a questionnaire asking about their child's medical conditions, illnesses and use of health services, schooling and functional ability as well as about their friends and family. This study led to important information being published internationally describing medical conditions, functional ability and survival of children with Down syndrome.

The 1997 study found that the most common conditions were eye (75%), ear (57%), cardiac (40%) and bowel (21%) conditions. Ear, nose, and throat specialists were the clinicians seen most often and the rate of tympanostomy tube (grommet) insertion was nearly 17 times that of the general childhood population. Whilst some support and supervision are required for complex self-care, communication and social skill tasks, severe functional limitations were found to be rare in school-aged children with Down syndrome. The likelihood of an infant with Down syndrome surviving the first year of life improved significantly over the study years from 89 to 94%. Whilst in the earlier years those born with a heart condition did less well than those without, this effect was not seen in the later years reflecting the benefits of early surgery.

A further study in 2004, titled Down Syndrome Needs, Opinions and Wishes (NOW) aimed to contact all families with a child aged up to 25 years with Down syndrome in WA. Three hundred and sixty three families (73% of those contacted) responded to this questionnaire, which again asked similar questions to the 1997 study but also included more structured questions on parental wellbeing, family functioning, child behaviour and costs associated with having a child with Down syndrome. This valuable body of information has already provided important feedback to families, clinicians and service providers in the form of a publication titled Down Syndrome Needs, Opinions, Wishes Report. A number of studies have also drawn on these data to investigate many aspects of life for a child with Down syndrome and their family. Findings from these studies are highlighted and presented in this booklet.



It has also become apparent that a time of particular concern for families with a young person with an intellectual disability is the period when their child leaves school and ventures into adult life. In an effort to explore this further, researchers at the Telethon Institute held a workshop in 2007 involving families, service providers and academics from across Australia to discuss the issues around this topic. As a result of this, funding was received by the Australian Research Council in 2009 to undertake a study focussing on Transition from School to Adulthood. The main purpose of this study was to describe what a "good outcome" is for the young person and the family and what factors may either help or interfere with this process. Consequently, at the end of 2009 families with a young person with Down syndrome aged 16 to 29 years were asked to participate in this study. Approximately 200 families have returned the questionnaire. The initial Transition Questionnaire again asked families about their child's medical conditions, illnesses and behaviour as well as family functioning, as these can all change over time and be important factors which may impact on the transition outcomes for the young person. Information on the experience of transitioning from school was also sought from the family. In 2011, this study will again contact families to seek follow-up information on their transition outcomes as well as to examine the important issues of obesity and nutrition.



The purpose of our research is to improve lives of people with Down syndrome and their families.  
The issues families confront need to be recognised and addressed by policy makers.  
Research translation is a tool for empowering those who may not have a popular voice.



## On the world stage

In parallel with our research in Western Australia the study team has worked with national and international researchers including Professor Dennis Hogan (Brown University, Rhode Island) and Michael Msall MD (University of Chicago).

In 2009 Dr Helen Leonard and her team were invited to submit a chapter to a publication on health issues in Down syndrome: *The International Review of Research in Mental Retardation: Health Issues in Down Syndrome*, edited by Dr Richard Charles Urbano from Vanderbilt University. The chapter, "Overview of Health Issues in School-aged children with Down Syndrome" was first authored by Kelly Thomas with contributions from Sonya Girdler, Jenny Bourke, Aditya Deshpande, Katherine Bathgate, Stephanie Fehr and Helen Leonard. Several of the authors were able to provide specific insights from their own areas of expertise such as nutrition and oral health. The Western Australian contribution is situated among articles from international authors focussing on health over the life span, particular diseases and body systems, and the needs of people with Down syndrome and their families.

Whilst acknowledging that published information is quite limited, the team researched the literature available and described the medical conditions most commonly experienced by school-aged children with Down syndrome. The clinical implications of these conditions were also covered and they are shown in Figure 1.

# The Researchers: Biographical statements

The researchers involved in these studies come from a diverse set of academic disciplines including Medicine, Dentistry, Public Health, Health Economics, Occupational Therapy, Psychology and Dietetics. Their interest in this field of study stems from many different reasons; at least two of our team have a personal connection and their stories follow.

## Katherine Bathgate

I am a PhD student in the research group at the Telethon Institute for Child Health Research (TICHR) focusing on nutrition and Down syndrome. My life's journey has brought me to this point where my research has both a personal and professional importance and I am delighted and honoured to be part of this team.

I am a dietician and lecturer; I have worked in hospitals, community health and private practice and helping people to embrace maximum health and well-being through choosing and eating nutritious foods is a passion of mine. Since 2000 I have been a lecturer at Curtin University, teaching undergraduate students studying nutrition and public health as well as furthering my own education.

Twelve years ago my first child Mark was born with Down syndrome, followed closely by his brother Cameron and more recently by his sister Imogen. Mark is a treasured and much loved member of his immediate and extended family and the way he manages his world around him is an inspiration. Mark teaches us the most important values in life, which are not the material items around us but the love and relationships we have with those close to us. He has also taught me to find joy in the present and in the smallest and most everyday occurrences that I might otherwise miss.

Soon after Mark was born I took both a personal and professional interest in nutrition and Down syndrome and sourced published research to educate myself about current knowledge. My first connection with the research team at TICHR was as a participant in the 2004 Down syndrome NOW study. Later I attended a family Down syndrome conference where Dr Helen Leonard spoke about the findings and I read the report that accompanied them. At the beginning of 2010 when I was presented with the opportunity to select an area of research for my PhD, I thought more about taking an active role in research in nutrition and Down syndrome and approached the TICHR research team.

Having been part of the research process both as a participant and now as a researcher I can truly see the value of our input into the research being conducted at TICHR, the ethical and respectful way that data is handled and the thorough investigation and dissemination of results. I hope that the findings of my research can be used to raise awareness of the importance of good nutrition and physical activity for people with Down syndrome and to provide the tools for nutritional assessment and support for families and young people.



## Kate Povee

For my Honours project I worked in collaboration with the Telethon Institute for Child Health Research (TICHR) to conduct research that explored the impact of having a child with Down syndrome on the family. Working on this project really made me reflect on my own experiences of having a sister with Down syndrome. For me, having a sister with Down syndrome has enriched my life immensely. Emma is my best friend. We have such a wonderful time cooking together or going out for coffee. She has such a great sense of humour and brings so much joy to everything she does.

Emma has made me see the beauty in life and appreciate the 'little things'. She has also taught me acceptance and to see the person, not the disability. I find the way Emma has overcome the barriers of having a disability truly inspirational. Emma has completed high school, studied at TAFE and works everyday at the local child care centre. I am so proud of my sister.

In my family, Emma has always been treated exactly the same as my brother and I. My parents have always provided Emma with the opportunities to grow to her fullest potential, just like my brother and I. Everyone is valued. I think my family are really close because of my sister. She is the glue.

As part of my PhD in Clinical Psychology at Curtin University I am hoping to conduct research with young people with intellectual disabilities that explores their experiences of stigma, the social roles they occupy and their self concept.

Ultimately, through my research and practice as a clinical psychologist, I hope to improve the quality of life of people with intellectual disabilities and their families.



# Medical

## Variation over time in medical conditions and health service utilization of children with Down syndrome\*

Children and young people with Down syndrome are at risk of a number of ongoing medical health issues, illnesses and infections. In recent times there have been many improvements in medicine that may have impacted on the management of these conditions. There have also been changes in community and health professionals' attitudes toward the treatment provided to people with Down syndrome.

This study sought to examine changes in the health of children with Down syndrome and how they have been using health care over time. Information from the 1997 and 2004 questionnaires was used in this study. Over this time period there was found to be a 70% reduction in currently reported heart problems. Routine screening for heart problems for newborn children and advances in surgical techniques have meant that cardiac disease is diagnosed and treated earlier. Thus more children with Down syndrome are surviving their first year and are likely to have improved health and a better quality of life. Regardless of whether the child was born with or without any cardiac condition, there was also a corresponding decrease in reported episodic illnesses with the greatest reductions being in tonsillitis and ear infections.

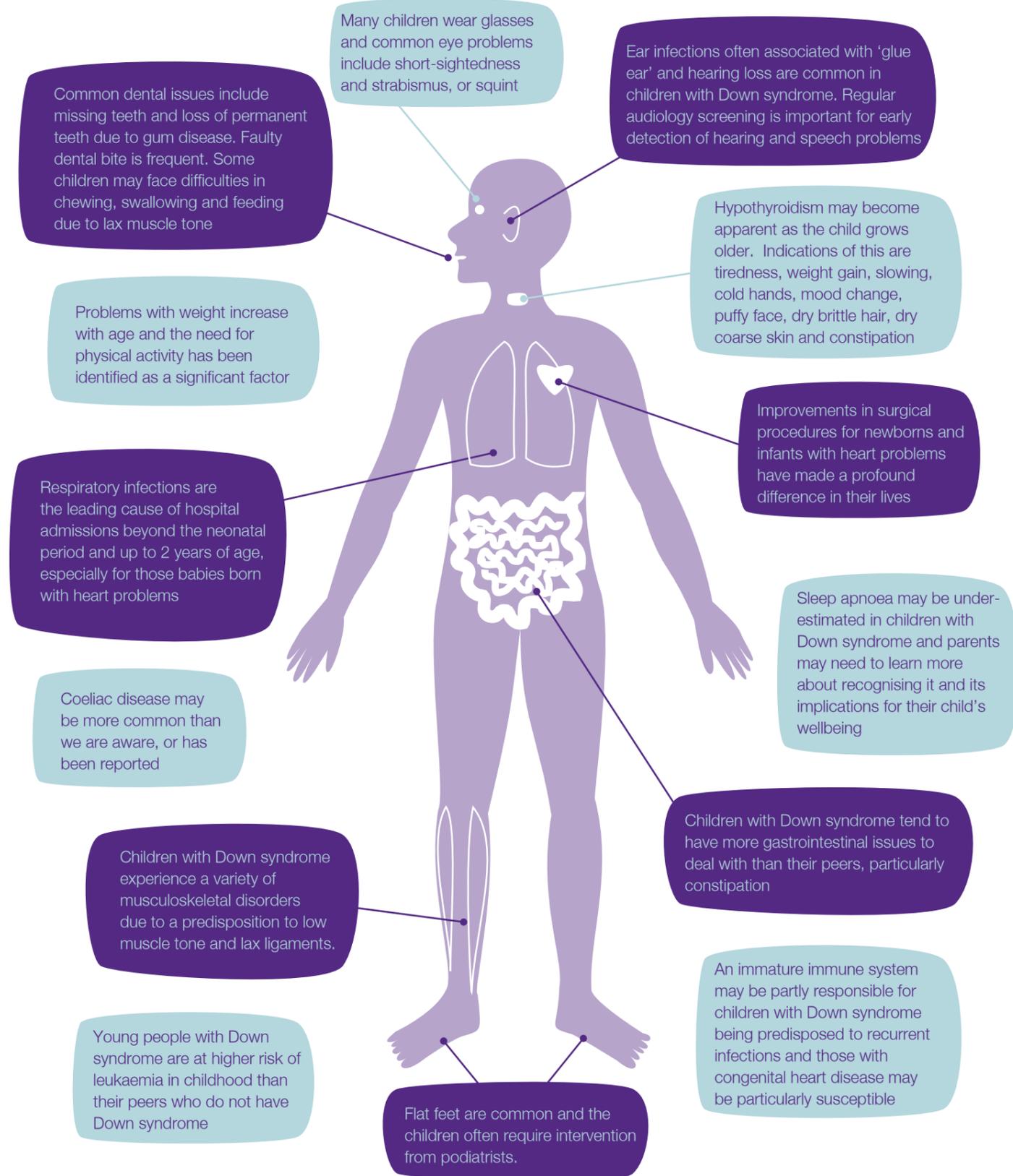
Subtle changes in the recognition and identification of some conditions were noted. Sleep apnoea seemed to be increasingly recognised over time with 20% of parents reporting this condition in 2004. Other conditions remained apparently unchanged over time. There seemed to be similar identification and treatment of thyroid problems with about 13% of parents reporting that their child had a condition in 2004 compared with 14% in 1997. Bowel problems continued to be of concern – particularly constipation. More than one third of the children were reported to have flat feet and half this number to wear orthotics.

In 2004 there was a reduction in the number of visits to GPs reported as well as in the use of specialist services. This could be partly due to the phasing out of some medical and other specialist disability medical services previously provided by Disability Services Commission. This is of concern as audiology and podiatry are important for the management of hearing and musculoskeletal concerns. These issues have a bearing on children's capacity for learning and participation in social relationships and sport.

So while survival is less of an issue for children with Down syndrome, identification and treatment of medical conditions now has more to do with enabling them to participate more in family and community life.

It is pleasing to report that the health status of children with Down syndrome appears to be improving over time in many areas. In 2004 compared with 1997 parents reported that their children had fewer illnesses, infections and hospital admissions and those born with congenital heart disease had fewer symptoms in later childhood. Medical conditions that these children experience and the supportive treatment they receive will impact on their quality of life and the choices that they can make as adults.

## Some facts about medical conditions which may be associated with Down syndrome\*



\* Thomas K, Bourke J, Girdler S, Bebbington A, Jacoby P, Leonard H. Variation Over Time in Medical Conditions and Health Service Utilization of Children with Down Syndrome. Journal of Pediatrics. 2011;158(2):194-200.e1.

\* Thomas K, Girdler S, Bourke J, Deshpande A, Bathgate K, Fehr S, et al. Overview of Health Issues in School-aged Children with Down Syndrome. In: Urbano RC, editor. International Review of Research in Mental Retardation: Academic Press; 2010. p. 67-106.

### Direct health care costs of children and adolescents with Down syndrome\*

The effects of Down syndrome involve a range of medical conditions which are either identified at birth or which develop through life. Consequently the use of medical services is not trivial and for children and adolescents has been shown to be greater for Down syndrome than for the equivalent general population in their early years.

In this study direct health care costs for families with a child with Down syndrome were estimated, based on information provided by parents in the 2004 Down syndrome NOW study. Costs were based on hospital admissions, medical and dental visits, therapy services, respite services, medications, complementary medicines and therapeutic devices used.

Whilst the total average annual health care cost was approximately \$4000, the median was around \$2000, indicating a wide variation amongst individuals, with almost half the total costs accounted for by about 10% of individuals. Average costs were shown to decrease with age, with most of the cost occurring in the first two years of life and rapidly decreasing after this. Whilst hospital costs are significant, much of the cost was represented by therapy in the younger age groups and respite in the older age groups.

An important finding from this study was that the demands on health services decline significantly with age to approach population levels during adolescence, indicating the general level of good health within this group.

Adolescents who live with Down syndrome generally enjoy good health. They place no more demands on health services than any other adolescent group in our society.

### Dental Health in children with Down syndrome\*

This research has focussed on the dental health of individuals with Down syndrome and the dental services used by them. The data used was obtained from families who had responded in the 2004 Down Syndrome NOW study. We found that only a quarter of children had visited a dentist in the year prior to the study and in particular very few in the 0-6 year age group. A slightly higher proportion of children had had extractions (16.25%) than fillings (11.25%). Additionally four parents were interviewed regarding their experiences in (1) maintaining dental health and (2) accessing dental services for their child with Down syndrome.



### 1. Maintenance of dental health:

#### a) At home

Although the four children in these families (aged 11-19 years) were independent in tooth brushing, there was some variation in the effectiveness:

*"He insists now because he is 17... cleans his own teeth, but they are not really clean .....there is still plaque there. ... I used to brush them myself once or twice a week just to make sure they are properly clean. He has an electric tooth brush... he does the best he can."*

*"He has always been interested in brushing his teeth, and that's one thing that he makes sure he does everyday. He just needs to be reminded to brush a little bit longer, which he is trying to do, but the dentist is quite happy with his oral health."*

#### b) Coping with dental procedures

Some parents expressed concerns regarding their child's ability to cope with complex dental procedures.

*"I have four children, my other three all had braces ...I just don't think he would cope with when they take the moulding, attach all the metal work. He just couldn't cope, he wouldn't be able to understand why and he wouldn't like it and he would probably just refuse to comply".*

*"...the visit before that he had a clean and scale under general anaesthetic. Every time he needs dental treatment it is a GA."*

### 2. Accessing dental services

Families also expressed some concerns about accessing suitable dental services particularly post school-age for their child.

*"We have had the school dentist up until recently. I have been very happy with the dentists and services we received, but I am not sure how it is going to be in future..."*

*"I guess the services that we receive for him are adequate but I am not in a position to comment if he was in the public system whether that would be adequate or not and that's one of the reasons why we have the private health insurance, because I know in other sort of speciality areas and other ancillaries there is a very long waiting list."*

We intend to use the findings for better understanding of the dental needs of this group of individuals and address them by suggesting more appropriate and accessible dental services.

\* Geelhoed E, Bebbington A, Bower C, Deshpande A, Leonard H. Direct health care costs of children and adolescents with Down syndrome (submitted for publication).

\*Deshpande A. (PhD Dental student) Factors associated with use of dental services by children with intellectual disability Perth: University of Western Australia; 2011.

## Puberty, Menstruation and Down syndrome\*

Puberty is intrinsic to the time of transition between childhood and adulthood in a young person's life. As it does for every family, for a family of a girl with Down syndrome this natural experience brings with it medical, educational and ethical concerns often relating to the management of menstruation. This study drew on 2004 data in the Down syndrome NOW study to investigate issues around puberty for girls.

Education and awareness-raising conversations are important for girls and young women with Down syndrome as they navigate their way through puberty. They can help manage any confusion and distress that may be accentuated by the combination of impaired cognitive ability and lack of information.

*"Our daughter needs a lot of direction and help in these areas; she has a lack of understanding connecting various issues."*

Menstrual management aims to maintain hygiene, privacy and dignity, minimize the physical and emotional distress of menstruation and maintain the young woman's general health and wellbeing. The three most usual approaches to managing the menstrual cycle are (a) self management, (b) medication and (c) surgery:

**(a) Self management:** Many factors influence menstrual management including the young woman's physical ability to change pads or tampons and her intellectual ability to understand when to change and implement necessary actions. Each person's experience is unique.

*"She needs supervision to handle the menstrual cycle. She seems to suffer from pre menstrual tension and cramps. She becomes more agitated and starts crying suddenly. Once her cycle starts she handles her period well providing she is reminded to use sanitary pads. Sometimes she will remove her pads after she has put one on. We sometimes have to remind her where to dispose of her pads."*

*"She does not handle having periods very well and her hygiene is affected when she has to deal with periods. She usually had periods every three weeks and would bleed for up to six days. It seemed very unfair...hence the need for the Mirena."*

**(b) Medication to suppress menstruation:** The most common form of medication is the continuous use of the oral contraceptive pill, however factors such as the individual's medical history and ongoing medical issues need to be taken into account before any medication is prescribed. It is an individual decision and choice which needs to be taken in conjunction with the young person's doctor.

*"We decided to put her on the pill as her periods were extremely heavy and she couldn't manage"*

*"My daughter suffered from terrible mood swings and Depo-Provera injections have calmed her down a lot. She quite often put herself in vulnerable situations."*

**(c) Surgery:** Whilst it is rare and consent issues may cause ethical dilemmas, sterilization in the form of tubal ligation or hysterectomy has occasionally been undertaken.

*"My daughter is on the pill. I would prefer that I could have her tubes tied. She is not sexually active but if that came and she became pregnant she would not have the intellectual capacity to cope with such an enormous thing and I feel it would affect her psychologically."*

*"Our daughter has had her tubes tied."*

The main study findings were:

- Girls with Down syndrome usually started having periods between 12 and 13 years
- The most common issue reported by parents was their daughter's lack of understanding of what was happening to their bodies at this time
- If there was a need to suppress menstruation the oral contraceptive pill was often used
- The oral contraceptive pill is also used as a means of managing pain and heavy bleeding.

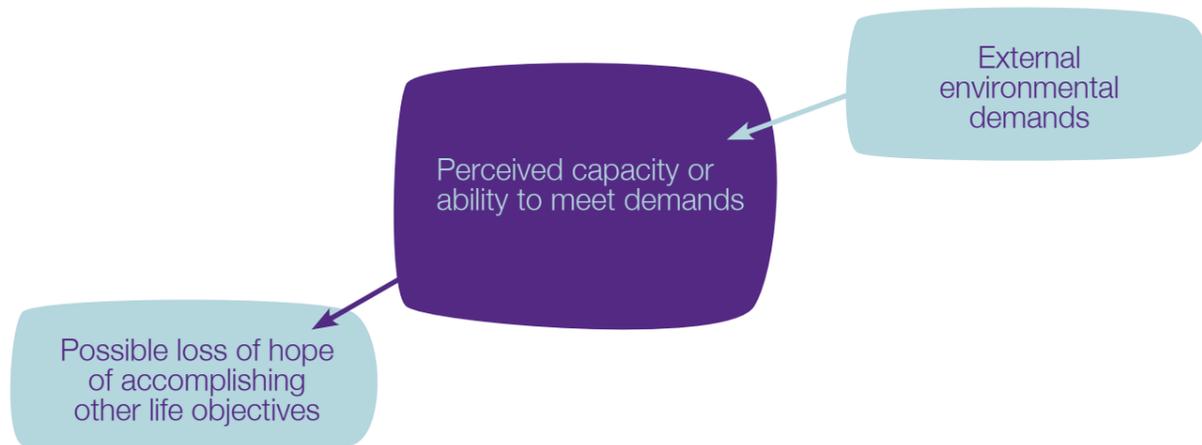
The ways in which we manage and educate, the decisions we make all contribute to the sense of wellbeing and acceptance.

\*Knight O. Pubertal Trajectory and the management of Menstruation in Females with Rett Syndrome and Down Syndrome. Honours Thesis: Edith Cowan University; 2010

# Family

## Physical and mental health in mothers of children with Down syndrome\*

This study explored the child factors which may impact on mothers' health as measured by the SF12, a widely-used set of questions which were included in the 2004 Down syndrome NOW questionnaire. The study looked at the role that their child's health and development has on mothers in the context of other factors such as mother's age, family income and number of siblings. For these mothers there is often a balancing act between "external environmental demands" and their own perceived internal ability to respond to these demands as well as the possibility that meeting these demands may interfere with satisfying or accomplishing other objectives in life.



In this study, the most important predictors of maternal health, particularly mental health, were found to be the amount of care required and the child's behavior. However mother's mental health was also shown to improve with mother's age. If a child is learning new skills, is independent in dressing and accessing public transport, and is participating in home and community life e.g. doing chores, shopping, using the telephone – then the mother tends to have better physical and mental health. Poorer mental and physical health was seen in mothers whose child had more behavioral problems. However mothers whose children had musculo-skeletal problems, multiple current health problems and four or more episodes of illness in the previous year also had poorer mental health. The compounding of issues increases the mother's experience of stress and can have an impact on her health.

Our study suggests that services aimed at enabling families to sort through parent-identified issues such as child behavior problems would be most beneficial. Programs for managing the health and well-being of children with complicated lives must be attentive and responsive to the challenges faced by the caregiver. Preventative strategies designed to help families both manage child behavior and maternal stress could have the potential to change caregiver health outcomes.

\* Bourke J, Ricciardo B, Bebbington A, Aiberti K, Jacoby P, Dyke P, et al. Physical and mental health in mothers of children with Down syndrome. *The Journal of Pediatrics*. 2008;153(3):320-6.

## An investigation of the role of spirituality and organized religion in supporting parents of children with Down syndrome\*

This research explored the role of religion and spirituality in the lives of mothers who have a child with Down syndrome. It involved a small number of face-to-face interviews as well as the use of data from the 2004 study.

Spirituality is a complex phenomenon and while there is overlap between organized religion and personal spirituality this study distinguished between the two: spirituality being understood as primarily an internal resource for coping and religious organizations as an external support from which people may draw strength to cope with stress and uncertainty.

Personal descriptions of spirituality were diverse, complex, intimate and self-reflective. The language used to describe spiritual experience was deeply relational and very often involved trying to express things that seemed contradictory. For example: grief and joy; highs and lows; the loss of a child hoped for and the treasuring of the one born; profound suffering, particularly when the child is sick, and deep love. Three broad themes can be identified in these interviews:

### a) The intensity of the experience begins at diagnosis and/or birth.

Each person interviewed described the birth-diagnosis period as most difficult. Each described their emotional state during diagnosis as feeling overwhelmed and traumatised. The mothers' words carry the raw elements of a spirituality marked by grief and pain: *"Just dreadful grief. Extreme distress ... it's a whole different pathway. So tremendous grief when she was first born for my normal baby that I didn't have."*

Again,

*"... Totally devastated and shocked, but it didn't change my love for her at all .... 'cause I already had that connection with her. I was scared too. Because, I didn't know what to expect and, I think I was devastated and scared."*

The experience of isolation may indicate a need for education of medical staff in their professional relationships at this time:

*"... he chose not to wait until I had my partner with me and told me by myself, so that's probably the most traumatic thing that's ever happened to me, to go through that by myself."*

*"My baby was in intensive care and nurses wouldn't talk to me but leave a pack about Down syndrome on my bed .....No one actually talked to me, but they would leave things for me to find, and I think I'd rather I'd had someone to talk to me."*

Watching their child suffer through surgery and life-threatening illness and being distanced from the child because of pain, equipment is described as "a nightmare".

*"...It was all just a rush of ambulances because he was going to die if he didn't have this surgery immediately. From then on, I didn't have a cuddle... and I didn't hold him for another two weeks."*

*"It was very traumatic. I spent a lot of time crying. I just couldn't handle seeing my child suffer."*

\* Pillay D. Spirituality and Organised religion in supporting parents of children with Down syndrome and intellectual disability. Honours Thesis: Edith Cowan; 2010.

**b) Spirituality is woven into the process of self-definition: Who am I? Who are my friends? Why has this happened?**

Mothers' incredible efforts to put their spirituality into words demonstrated how hard it can be to do this e.g. some describe their spirituality as a deep connection with or belief in a higher spiritual being or God which allows for expression of doubt: *"I have always had a belief in something higher than me. I question it constantly .... I quite haven't worked out what it is that I believe in, but I definitely do. I do think it makes me feel better that I have something to hold onto, I suppose."*

Others emphasize their personal beliefs or values which evolve with life's experiences: *"I think it is about your own internal belief system whether that be something tangible or not. I do not see myself subscribing to: "there's one higher power above all". I think there's something going on."*

Others again indicate their effort to integrate what they have received from formal religion with their own thoughts and experience: *"I suppose my personal beliefs are .... I think I have taken a lot from religion. I think it's the personal philosophy that you try to be nice, you try to be nice to people."*

Spirituality is part and parcel of the personal work of integrating the significant stressors of life in association with their child who has Down syndrome such as times when their child exhibits difficult behavior: *"We'd go somewhere and see families having a nice little picnic or doing something really just normal, that doesn't always work for us..... You'd tend to isolate yourself from everyone."*

Another stressor can be the pain which can be experienced in relation to family and friends: *"Oh Down syndrome, they're my favorite, they are all so happy".* Likewise, many experienced almost as a 'taboo' to speak of their child as a child with Down syndrome, as if 'there were this elephant in the room' and that they were being ungrateful for the child they had. The spiritual life involved learning to cope with loss and loneliness among family and friends and some respond by wanting to flee and protect their child from harsh judgments and misconceptions: *"Sometimes I wish I could take him and go and live on a desert island somewhere where there is no one else around to make him feel, and to make me feel, as if he was less than everybody else because he has a disability."*

Finances, work-life balance, schooling, fear of the unknown and uncertainty about the future are stressors that accompany the mother throughout this journey. This journey often involves an effort to cope with the profound sense of having been forgotten. *"My God, my God why have you forsaken me?"* and that is how I felt. *I felt like God had forsaken me.* Some worried: *"what did I do wrong?"* Or *"...what did we do to deserve this disabled child?"*

Through the difficult times of loneliness, anger, frustration mothers described developing a new identity and a strengthening of their capacity to parent and protect. *"Its more like a puzzle, or maybe a mosaic. It's never really finished. It just takes time and its bit-by-bit – the emotions. I guess its finding contentment in where she's at and also seeing the beauty in her."*

**c) Times when one can look back upon the experiences and reflect: being able to name the qualities which have hindered and/or enriched life.**

Upon reflection, mothers described the journey as entering a world previously unknown where friendships were made that may otherwise never have eventuated, where values were shaped differently, and wholesale changes in life plans and occupations were made. Having a child with Down syndrome spiritually challenged each mother who was interviewed. The challenges had the effect of reaffirming or weakening religious beliefs held prior to the diagnosis and birth of their child.

*"What tends to happen, I think for me, is when tragedy happens, that's when I do probably want to try and lean closer towards God or something that's out there, I think, almost as a comfort."*

Hope is important for gaining a sense of control, the capacity to adapt and a sense of direction. *"There's hope all the time, all the time when she does something new. I think you have to believe there is hope, otherwise you will go crazy."*

Prayer gave mothers the strength to deal with difficult situations and was used frequently when they felt isolated, anxious or depressed. Those who had turned away from their institutional religious beliefs still used prayer as a means of comfort during a crisis.

*"...I really was unnerved about having the baby .... In the end I remember ... saying (to myself) "Look, just pray about it. You're becoming so worked up about it..., through prayer you're given the strength to do it. I know I wouldn't have otherwise...."*

*"I said a prayer.....I think it's a reflex action...I didn't even believe in God; why am I praying to him but its ok. I just had that thought."*

As for friendship: *"My choice of friends depends on their relationship with my daughter. If they really don't get her, if they exclude her, then I don't have much time for them...."*

This study highlighted a very interesting divergence in outcomes: the responses provided in the 2004 questionnaire indicated that spirituality and organized religion had little effect in supporting the mental health of mothers of children with Down syndrome. However, the interview process yielded a quite different response whereby personal spirituality was described as a strong, dynamic source of support in coping with stressors and life's challenges.

Having a baby diagnosed as having Down syndrome may signify the beginning of a profound experience which will influence and be influenced by the spiritual dimension of life.

## Leisure Participation for school-aged children with Down syndrome\*

This study used data from the 2004 study to explore the participation of school-aged children with Down syndrome in leisure and experiences in play, friendships, sports and hobbies. Multiple factors can facilitate or impede a young person with Down syndrome's participation in these areas. For this reason, the World Health Organization's International Classification of Functioning, Disability and Health (ICF) framework was used to consider the numerous factors that can impact on children and young people at one point in time. The ICF takes into account the complexity of life and acknowledges that many things come into play which may affect a person's participation – in this case in friendships, sports and hobbies. The consideration of environmental factors, which included family characteristics such as income, availability of transport, parental health and family functioning, as well as the health of the child and their individual level of functioning, may all influence the level of participation by the young person.

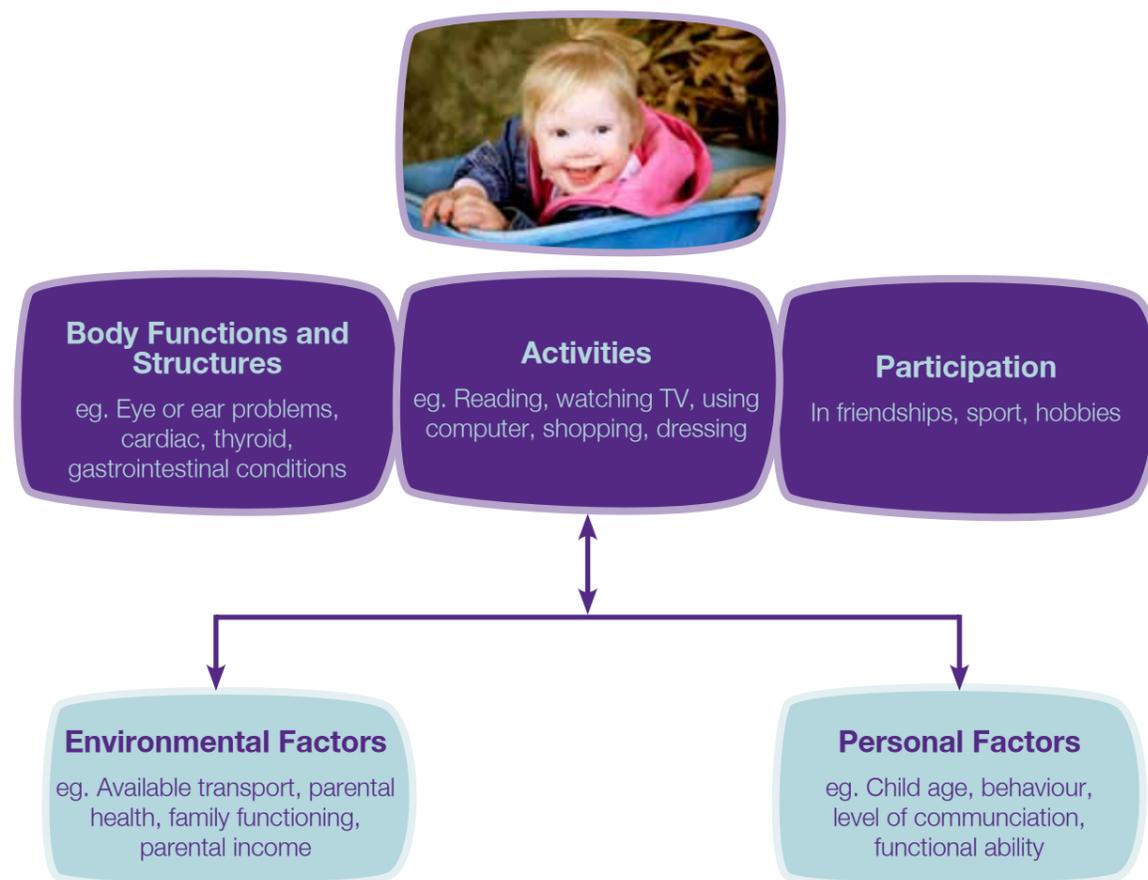


Figure 2: ICF Framework

\* Oates A, Bebbington A, Bourke J, Girdler S, Leonard H. Leisure participation for school-aged children with Down syndrome. Disability and Rehabilitation. 2011 in press.

Half of the parents reported that their child with Down syndrome had two or more friends, although one third reported that their child had no friends. Children who were more functionally independent at home, in school and in the community, were more likely to have friends. Children who experienced problems with social interactions, language and communication, or who had more behavioural and emotional difficulties, were also less likely to have friends. However, behavioural and emotional difficulties had more of a negative impact on the children's friendships than problems with social communication. Environmental factors which were associated with children having more friends were parents' better mental or physical health, more support from family or the community, and parents having more time for the family.

Overall, children in our study did not participate in many sports or hobbies. Hobbies in which children engaged included reading, using computers, drawing, games and playing musical instruments. Children whose parents had adequate access to public transport or to private transport were more likely to be participating in hobbies than those whose families had less access. Children with better functional independence and social communication skills also enjoyed more hobbies.



Overall the key messages identified in this study were:

- Although many children with Down syndrome have friends, they may not see their friends often.
- The health of the parent, the support available to the family and parent availability of time all impact on children's friendships.
- Children with Down syndrome were unlikely to participate in more than one sport or hobby and generally engaged in individual activities rather than in team sports. The most popular sports were swimming, bowling, soccer and basketball.
- Children with greater functional independence participated more in friendships, sports and hobbies. Grading and adaptation of activities to make it easier for children to participate could help to remove barriers and enable more involvement by children with lower skills.

### The impact of having a sibling with Down syndrome \*

Throughout much of the 20th century, families had been actively encouraged to place their child with a disability in residential care to minimize the impact on themselves and their other children. More recently, community attitudes have taken a more positive and inclusive position with most children remaining within the family. Much of the research in previous decades has focused upon parents (mainly mothers) of having a child with a disability. However, since the 1990s, awareness has been growing of the need to focus on the experience of siblings as well as parents. This study used data from the 1997 questionnaire to describe parents' perceptions of the advantages and disadvantages to their other children of having a sibling with Down syndrome.

The average age of children with Down syndrome in this study was 11.4 years. The number of siblings in each family ranged from one to nine. Nearly 80% of parents reported that there had been benefits to their other children from having a child with Down syndrome. However about three-quarters also reported that there had been disadvantages to their other children.

The descriptions given by parents of the positive and negative impact on siblings were considered in key themes. There was an overlap as some of these themes were identified as contributing to both benefit and disadvantage for brothers and sisters but they were separated into positive and negative impact for the sake of clarity (see table opposite).

Policies which have in the past favoured the "protection" of families from children with an intellectual disability like Down syndrome have been replaced by those which stress their inclusion in the family. This paper has sought to describe the experience of brothers and sisters of a child with Down syndrome through the eyes and concerns of the parents.



Parents perceived that brothers and sisters of children with Down syndrome experience both advantages and disadvantages that can often exist together. For example, they may develop a capacity for care and compassion at an early age but may also carry an extra burden of concern and possibly miss out on the "normal" reciprocal sibling relationship.

\* Mulroy S, Robertson L, Aiberti K, Leonard H, Bower C. The impact of having a sibling with an intellectual disability: parental perspectives in two disorders. *Journal of Intellectual Disability Research*. 2008;52(Pt 3):216-29.

Positive Impact:	Negative Impact:
<p>Tolerance and acceptance of difference: Parents frequently noted that their children were very much aware, tolerant and accepting of disability.</p>	<p>Time constraints: Some parents were concerned that they had had less time and energy for the brothers and sisters of their child with Down syndrome.</p>
<p>Capacity for care and compassion: The majority of parents observed that their other children were compassionate, caring and kind towards other people.</p>	<p>Routine/restriction: They also were concerned about the impact of extra 'rules' imposed on the other children such as keeping doors locked, small toys away, bedroom doors closed in order to deal with the behavioural challenges of the child with Down syndrome.</p>
<p>Mature personality: Parents noted that the brothers and sisters of the child with Down syndrome often presented as more mature than similar aged peers.</p>	<p>Parental emotion: The exposure of children to parental stress, anxiety and irritability arising from the pressures associated with caring for their child with a disability was also reported to be a disadvantage to siblings.</p>
<p>Patience: Brothers and sisters of a child with Down syndrome were reported to learn to exercise patience at a young age.</p>	<p>Financial: Many parents worried that costs as a result of the additional needs of their child with a disability meant their other children go without. This particularly related to not being able to afford holidays, some recreational and educational activities and material goods.</p>
<p>Insight into learning and teaching: Brothers and sisters of a child with Down syndrome were described as developing a role as teacher and they become expert at breaking down tasks into smaller units.</p>	<p>Burden/help: Some parents highlighted the burden on siblings in assisting with care.</p>
<p>Assistance/help/support: The ease with which siblings went to the aid of their parents in caring for the child was often noted.</p>	<p>They also expressed concerns about the long-term burden on them when they themselves were no longer alive.</p>
<p>Appreciation of self: one's own life, gifts and health: Parents described how the brothers and sisters of their child with Down syndrome tended to be strongly aware and appreciative of their own health and abilities. Many parents felt this sense of appreciation extended to valuing simple things they might otherwise have taken for granted. In relation to this heightened sense of appreciation, some parents acknowledged that their nondisabled children learned from an early age that life is sometimes very difficult. This was presented as having a positive impact on their child, in terms of being acutely aware of those less privileged.</p>	<p>Sibling relationship: Some grieved for their children at the loss of what they may have called "normal" brother-sister relationships particularly when it came to play and reciprocal emotionally intimate relationships.</p>
<p>"It has made them aware of people who may need a little extra help."</p>	<p>Peer acceptance: Brothers and sisters of children with Down syndrome often had to deal with misunderstandings about the syndrome and biases and insults towards themselves and their affected sibling. Parents may have felt powerless when they preferred not to have friends visit at home.</p>
<p>"They feel that they are better off than other friends who don't have a handicapped sibling because they've experienced more in life."</p>	<p>Dealing with behaviours: Behavioural issues were particularly identified by parents of children with Down syndrome as a problem for siblings.</p>
<p>"More emotionally mature because they have had to deal with a lot of issues early in their lives"</p>	<p>"Much time has been spent on therapies and working with my child at home. Sometimes this has meant less time for other children."</p>
<p>"They have to be more independent, an opportunity for greater self confidence."</p>	<p>"Some behaviours can be embarrassing to the adolescent siblings."</p>
	<p>"Unable to develop usual sibling relationship due to different abilities"</p>

### Family Functioning in Families with a Child with Down Syndrome \*

Early research on the impact of having a child with an intellectual disability on the family reflected a model whereby couples and the family as a whole were assumed to suffer greatly and experience inevitable negativity. Families that include a child with an intellectual disability face unique challenges however the wide variety of family responses to disability captures family strengths, adaptation and resilience. The increased emotional, physical and financial demands can have a profound impact on the functioning and wellbeing of the family. This study examined the impact of having a child with Down syndrome on the marital relationship and the wellbeing of the family unit.

This study also used data from the 2004 questionnaire particularly in relation to responses to the questions about the impact of having a child with Down syndrome on family activities. Overall, families in this study displayed healthy family functioning and a high level of marital satisfaction and adjustment comparable to families of children without disabilities in the general population. Most families felt that having a child with Down syndrome had no or minimal impact on the functioning of the family. The relationships within the family, daily family life and the involvement in family activities and family holidays were described as not being affected by the presence of a child with Down syndrome in the family:

*“No impact (on family life). He’s not hard to get along with. He walks and talks and can do anything we do. He’s just one of us”.*



Most families reported that the child with Down syndrome was included in all aspects of family life and was treated the same as the other children in the family:

*“She has always been treated exactly the same as our other children, she has always been included in anything that we do, so I believe no impact (on the family)”.*

This sense of normality was achieved by accepting disability and ‘getting on with it’. Family life was commonly adapted and modified to suit the unique interests and cognitive and physical capabilities of the child with Down syndrome. Other coping strategies identified in this study included relying on faith and having support from extended families and friends.

Some of the respondents in this study felt that the child with Down syndrome had a positive impact on the family. The child with Down syndrome was described as enriching family life by bringing family members closer together. Other respondents believed that having a child with Down syndrome taught them valuable life lessons and shaped their life philosophies; they were more patient, more accepting of difference and had a greater appreciation for life. As one parent explained:

*“He has taught us many important lessons about life and how no matter how tough we might think life is we should be thankful for what we have, as there is always someone who is worse off... people with disabilities are just the same as everyone else inside”.*

In contrast to the positive impacts and picture of normality commonly described, a smaller proportion of respondents in this study believed that family life revolved around the child with Down syndrome and felt disappointed that family life lacked freedom and spontaneity. Some were resentful that the needs of the child with Down syndrome were considered before the needs of the rest of the family. A commonly expressed concern was the disruption caused to the lives of the siblings. Some respondents felt that siblings received less attention, had more responsibility and a restricted social life:

*“I seem to have my middle son, all of 8 years, help a lot and I feel I load this responsibility too much on him. I forget he is only 8. But having an older brother with Downs can be pretty full on to deal with”.*

Other negative impacts on the family that were reported included an increased financial burden, worry for the future, marriage problems and a reduced social life. In this study child maladaptive and autism-spectrum behaviours were found to have a small effect on the functioning of families with a child with Down syndrome. A child wandering or running away was identified as a major issue during family activities and holidays. As one parent explained:

*“It is very hard to go places as he tends to run off and not listen. We are always worried he may get lost”.*

Other behavioural problems commonly identified by parents/carers included stubbornness, tantrums, aggressiveness and social inappropriateness:

*“... if there’s little kids around he’ll zone in on one then push, stalk, hug, strangle, whatever takes his fancy”.*

This study adds to our understanding of the impact of having a child with Down syndrome on the family unit and may have an important role in dispelling some of the myths that surround the functioning of families with a child with Down syndrome. The overriding impression of normality amongst the families in the current study strongly contradicts the historical views that having a child with an intellectual disability is a tragedy and inevitably detrimental to couples and families. It must however, be noted that child behaviour problems were associated with poorer family functioning in some families.

The overriding impression of normality amongst the families in the current study strongly contradicts historical views about the impact a child with an intellectual disability may have on family well-being.



\*Povee K. Family Functioning in Families with a Child with Down Syndrome: A Mixed Methods Approach. Honours Thesis: Curtin University; 2010

# Transition

## Transition from school to post-school for young adults with Down syndrome

Transition from school to post-school may be a time of upheaval, stress and important decisions. Currently services have been reported as disjointed and difficult to navigate for young adults with developmental disabilities and their families. This study aims to identify what is regarded as a “good outcome” for the young person with Down syndrome and their family, and what factors make a difference.

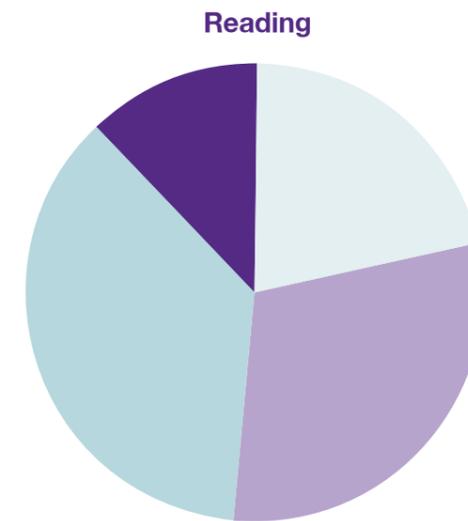
The International Classification of Functioning, Disability and Health (ICF as described earlier in Figure 2) provides an internationally recognised framework for this research. It takes into account the complexity of life and acknowledges that many things come into play which may affect a person’s ability to participate – in this case in post-school activities and/ or work. The ICF encompasses the health of the young person and any impairments; their level of functioning and the presence of ‘activity limitations’ which may impact on their ability to complete a task; and the description of their involvement in life situations described as ‘participation’. The ICF also takes into account personal factors such as gender, age and behaviour of the young person, as well as environmental factors such as the level of family resources, access to transport and parental health.

The questionnaire to families asked specific questions about this transition period, along with questions on the health and well-being of the young person with Down syndrome and their families. This information will be extensively analysed and explored to find those factors which positively and negatively influence transition outcomes. The first wave of questionnaires was sent out to 268 families in late 2009 and early 2010 and responses from 202 families (75.4%) have been returned. Forty-three percent of these young people were female (n=86) and 57% male (n=116). The age of the individuals varied from pre transition (16 to 17 years, n= 25), early transition (18 to 22 years, n=71) to late transition (23 to 31 years, n=106). The majority of families (75%) lived in the metropolitan area.

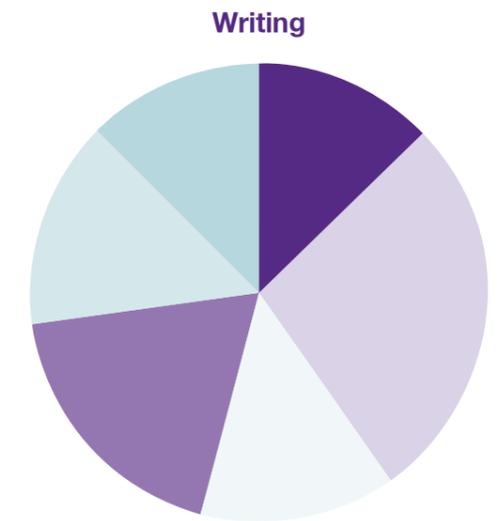
Most young people (91%) lived at home with their parents or guardian. Of those young adults living out of home almost all were aged over 23 years and lived either in group homes, a unit or house with other relatives or friends, in a unit or house living alone or in a hostel.

The variability between individuals in their ability to read, write, communicate and prepare meals can be seen through the following diagrams, which show the proportion of responses for each of these domains of functioning.

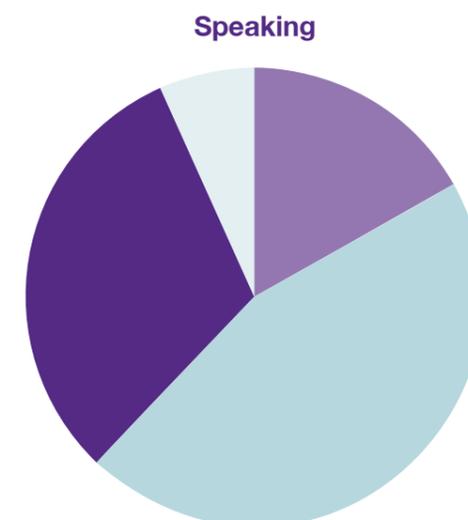
It is apparent that the post-school period is very dynamic and the activities undertaken, the options followed, the health, behaviour and perceived quality of life of both the young person and the family may change. In order to gain further insight into these changes that may be occurring in the lives of the young people the study will seek further follow-up information from the families in 2011. The study aims to identify both the barriers and facilitators for people with Down syndrome as they enter adulthood and navigate towards a fulfilled adult life.



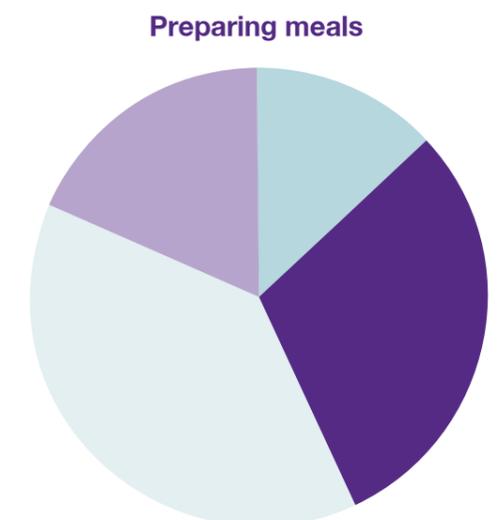
- Can read a series of instructions
- Can read signs
- Can recognise own written name
- Recognise 6 different labels on tins and boxes



- Has good writing skills
- Can write short notes
- Can write own name and address without help
- Writes full name without help
- Writes name and address from copy
- Unable to write



- Speaks well - intelligible to all
- Some difficulty in speaking - lack of clarity or fluency
- Only intelligible to those who know him/her well
- Does not use speech to communicate



- Can prepare a variety of meals without supervision
- Prepares simple hot food without supervision
- Prepares food which does not require cooking
- With supervision, can prepare simple foods
- Needs all food prepared for him/her



## Need for Research

A parent of a 40 year old son or daughter with Down syndrome who was born in the 1960s would find they have raised a child in an era before education was a right for children with Down syndrome, when diagnosis of Down syndrome was regarded as a heavy burden, and when the quality of care and the quality of life for those who lived in institutions was variable. The information from the studies presented here indicate that, while life for a child with Down syndrome and their families is very different today from the 1960s, there is still some way to go in relation to healthcare, education and complete community acceptance.

The evolving awareness of issues through research studies brings with it responsibilities which relate to:

- the need for parents of a child diagnosed with Down syndrome, whether prenatally or at birth, to receive accurate information about healthcare needs of children with Down syndrome.
- the importance of this information being delivered in a non-threatening, balanced, real and supportive manner.
- the reality that not every child with Down syndrome is sick, nor has multiple health problems.
- the importance of people with Down syndrome receiving the same healthcare as the rest of the population.
- anxiety about illness and noting that many symptoms experienced by children with Down syndrome are the same as any other child would experience.
- the importance of research delivering its findings accurately and not overwhelming and frightening families; rather that it be considerate of the real-life implications of a statistically significant finding.

On the research horizon we can identify the ongoing need for:

- up-to-date research on fertility, sexuality and relationships.
- understanding of implications for the management of the dual diagnosis of autism in children and adults with Down syndrome.
- “sophisticated” studies which allow us to tease out the effects of different family environments and individual characteristics so that we can understand and possibly predict individual risks and needs better.
- educating the medical profession in the healthcare needs of children and adults with Down syndrome and particularly to educate health professionals about the potential of people with Down Syndrome.

