“We all have a lot to learn.”

Prenatal screening for Down syndrome: A discussion paper

October 2021

Down Syndrome Australia

About Down Syndrome Australia

Down Syndrome Australia was established in 2011 as the peak body for people with Down syndrome in Australia. Our purpose is to influence social and policy change and provide a national profile and voice for people living with Down syndrome. Our vision is an Australia where people living with Down syndrome are valued, reach their potential, and enjoy social and economic inclusion.

The Down Syndrome Federation works together to provide support for people with Down syndrome and to make Australian society inclusive for people with Down syndrome. The Federation is made up of Down Syndrome Australia and the State and Territory Down syndrome organisations. We work in partnership to maximise the opportunities and support for people with Down syndrome and their families and support networks.

Down syndrome is a genetic condition in which the person has an extra copy of some or all of the 21st chromosome. This additional chromosome results in a number of physical and developmental characteristics and some level of intellectual disability. There are more than 15,000 Australians who have Down syndrome and approximately 1 in every 1,100 babies in Australia are born with Down syndrome.[1]

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[1] Down Syndrome Australia (2020). Down Syndrome Population Statistics. Retrieved from **https://www.downsyndrome.org.au/about-down-**

Background

Screening technologies, such as non-invasive prenatal screening, are becoming a routine part of early pregnancy care from ten weeks gestation. The ethical use of these technologies relies on prospective parents receiving accurate and balanced information pre and post screening, along with time to make informed decisions.

Around 400,000 women in Australia become pregnant each year. Families rely on accurate and balanced information from health care providers to make decisions about screening, including whether to continue a pregnancy based on their screening results. Most prospective parents undergo combined first trimester screening, however non-invasive prenatal screening (NIPT) is becoming more common (see Appendix 1 for an overview of prenatal screening and diagnostic test). An estimated 25–30 per cent of pregnant women in Australia choose NIPT,[2] which involves a blood test taken from a pregnant woman from 10 weeks gestation. The most common unexpected prenatal screening result is a high chance of Trisomy 21, also known as Down syndrome.

Down Syndrome Australia takes a pro-information stance in decision making,[3] whatever that choice may be, and demand all families are supported to make informed decisions. Medical practitioners must:

- deliver high chance or confirmed diagnosis results using neutral language (e.g., ‘chance’ rather than ‘risk’, factual delivery of a diagnosis without emotive terms such as ‘I’m sorry’ or ‘bad news’);
- provide up-to date, evidenced-based information on Down syndrome and other conditions being screened for, both before screening and after results are given;
- offer referrals to peer support services such as local Down syndrome associations;
- offer non-directive counselling and support services both before and after a decision is made on whether to continue the pregnancy;
- give respect for decisions made by families, and not revisit that decision once it has been communicated, while making it clear that they have a right to change their mind.

In Australia approximately 9 out of 10 pregnancies diagnosed with Down syndrome end in termination.

Down Syndrome Australia holds serious concerns about the lack of balanced, evidenced-based information given to prospective parents by health care providers about Down syndrome in the context of prenatal screening.

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[2] Medical Services Advisory Committee 2019. Public summary document: application no. 1492 – non-invasive prenatal testing (NIPT) for trisomies 21, 18 and 13. Public Summary Document., Australian Government, Canberra

[3] **Down Syndrome Australia (2019) Position Paper on Prenatal Screening.**

In July and August 2021, Down Syndrome Australia undertook a survey to understand contemporary prenatal screening experiences. The survey received 320 responses from parents who have a child under the age of 10 with Down syndrome. Disturbingly, the experiences shared reveal that it is still commonplace for prospective parents to be given misinformation about life with Down syndrome as they navigate prenatal screening decision making. **Nearly half of the parents surveyed felt pressure from health care providers to terminate their pregnancy.**

Information shared with families in the context of prenatal screening decision making rarely reflects the experiences of those living with Down syndrome and their families. Nearly half of parents who responded to the 2021 survey felt that the information presented to them by health care professionals did not give them an understanding of Down syndrome. 69 per cent did not think the information gave a clear understanding of the lived experience of people living with Down syndrome and their families. Very few families were given the opportunity to meet a person with Down syndrome.

Health professionals need to provide accurate, balanced information to prospective parents. They also need to allow sufficient time for decision making. Information delivered by health professionals must demonstrate both respect for diversity and for a family's right to informed choice, whatever that choice might be. Families must be given the opportunity to connect with local Down syndrome organisations which can provide information, support, and connection to other families.

Claire Mitchell, a self-advocate, member of the Down Syndrome Advisory Network and Director on the Down Syndrome Australia Board, says, "Don't screen for Down syndrome without telling families what it really means." When done right the information provided to parents during the prenatal screening process will support families to make an informed choice about their pregnancy and has the potential to contribute to a more inclusive society.

What do we know about experiences of prenatal screening in Australia?

Early pregnancy experiences are not often openly discussed. Decisions made after prenatal screening are highly personal, and prospective parents may fear that discussing their decisions publicly leaves them open to judgement. There is, however, growing public discussion and research focused on how prospective parents are experiencing prenatal screening and how their experiences could be improved.

A 2019 systemic review examined how prospective parents had experienced NIPT both in Australia and internationally.[4] The review focused on how informed choice was facilitated and described prospective parents identifying large gaps in clinicians' knowledge about the experience of raising a child with Down syndrome. While many women were satisfied with their understanding of NIPT, the review found 'several misconceptions still persisted, raising questions as to whether they were adequately informed about the test.'[5] The review concluded 'Widened availability to trustworthy information about NIPT as well as careful attention to the facilitation of counselling may help safeguard informed decision-making.'[6]

An Australian qualitative analysis focusing on non-invasive prenatal screening examined the perspectives of mothers of a child with Down syndrome.[7] Approximately half of the mothers in the study reported that they felt medical professionals expected that they would terminate following a positive screening test.[8] One of the mothers stated "I do know people who use NIPS (sic) just so they know and they can prepare. But I started to feel like, well, the best preparation is actually an inclusive society. Because then people aren't worried about it."[9]

Mothers in the study explained that the information about Down syndrome presented to them had a primary focus on the incidences of medical difficulties that a person with Down syndrome might experience. Participants felt that context and balance was needed.

One of the mothers said, "I think they hand out stats but they don't hand out any interpretations. They tell... parents 50 per cent chance of a heart defect, but they never tell them that a large number of those will just resolve themselves after birth."[10]

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[4] Cernat et al, 2019 Facilitating informed choice about non-invasive prenatal testing (NIPT): a systemic review and qualitative meta-synthesis of women's experiences. BMC Pregnancy and Childbirth, 19:27.

[5] Ibid.

[6] Ibid.

[7] Valentin et al, 2019. Mothers of a child with Down syndrome: A qualitative analysis of the perspectives on non-invasive prenatal testing. Midwifery, 76, 118-124.

[8] Ibid, p.120

[9] Ibid, p.122

[10] Ibid, p.121

Another mother, identified as Victoria, recalled her experience of receiving a diagnosis: "They took us into a little room. And you know that it must be bad, like it must be bad news because um, you have to go into the room and it's got nice furniture … you know so then you get to hear all the statistics about how bad your child's life will be … once they tell you thirty things, your feeling is, oh so I'll have all of these thirty things to deal with, and that's, crap."[11]

The mothers in the study spoke about the potential of NIPT to reinforce negative societal attitudes towards disability. All highlighted the importance of autonomy, appropriate support including balanced, evidenced-based information and 'time and space for individual reflection.'[12] The study concluded that health professionals hearing the stories of people with Down syndrome and their families may enhance their ability to provide informed, balanced information and support to prospective parents without an expectation of termination.[13]

In the recent ABC documentary, *The Upside* (2020), obstetrician Professor Steve Robson was interviewed by Julia Hales, an Australian actress with Down syndrome. Professor Robson said that a huge issue for doctors is the fear of missing a diagnosis of Down syndrome, and of being sued by parents. When Hales asked Professor Robson, "What do doctors know about life with Down syndrome?" he readily admitted: "A lot of doctors these days don't have a lot of experience [with Down syndrome] and in many ways they almost fall back on textbook descriptions."

A clinical environment in which doctors know little about life with Down syndrome – and fear litigation – is not conducive to enabling informed decision-making. Yet the stakes could not be any higher for prospective parents deciding whether to end or continue a wanted pregnancy.

The podcast *One Screened Every Minute*, presents a series of interviews with people across Australia who have received high chance screening results for a range of chromosomal differences, including Down syndrome. The interviews, which incorporate the experiences of prospective parents who have continued the pregnancy, as well as those who have chosen termination, reveal the immense emotional toil required to navigate decision making and, in many cases, the lack of timely and accurate information and support.

Within the Down syndrome community, prospective parents commonly report having received unbalanced information with a focus on the physical features of Down syndrome and the potential health complications. The 'extraordinary ordinariness' of life with Down syndrome is often left out.

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[11] Ibid, p.121

[12] Ibid. p.123

[13] Ibid. p.123

Down Syndrome Australia 2021 Survey

Down Syndrome Australia undertook a survey of 320 parents across Australia who have a child under the age of 10 with Down syndrome. The responses reflect the views of parents who chose to continue their pregnancy. Future work is needed to further understand the experiences and support needs of those who chose to terminate their pregnancy after a prenatal diagnosis of Down syndrome.

The responses reveal that it is commonplace for prospective parents to be given misinformation about life with Down syndrome and that nearly half felt pressure from health care providers to terminate their pregnancy.

Results showed that many families do not get access to balanced, evidenced-based information to make an informed decision as they make high stakes choices about the future of a wanted pregnancy. Many families described not receiving adequate support when they chose to continue their pregnancy after a high chance result or prenatal diagnosis.

The information provided by health professionals is often not consistent with the latest research on the experiences of people with Down syndrome and their families. People with Down syndrome report having a high quality of life, with one survey finding that 99 per cent of respondents indicated that they were very happy.[14] Similarly, a survey of parents and siblings of people with Down syndrome, indicate that most people have had a positive experience having a person with Down syndrome in their life. Less than 5 per cent indicated any regret about their decision to have their child.[15]

For many families, responding to the Down Syndrome Australia survey was their first opportunity to share their prenatal screening experiences in detail. Many noted they were motivated to share their story to ensure information and support is improved for families in the future.

Every state and territory were represented. Respondents lived in metro (59 per cent), regional (37 per cent) and remote (4 per cent) areas of Australia.

The majority of those surveyed had completed combined first trimester screening (82 per cent), with 42 per cent having completed non-invasive prenatal screening (some had undertaken both types of screening). Most respondents received the results of their screening test from their GP (52 per cent) or their obstetrician (40 per cent).

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[14] Skotko BG, Levine SP, Goldstein R. 2011. Self-perceptions from people with Down syndrome. Am J Med Genet Part A 155:2360–2369.

[15] Skotko BG, Levine SP, Goldstein R. 2011. Having a son or daughter with Down syndrome: Perspectives from mothers and fathers. Am J Med Genet Part A 155:2335–2347.

The most common information provided to them after receiving a high chance screening result was:

- Verbal information about termination (52 per cent)
- Verbal summary of Down syndrome (37 per cent)
- Flyer about Down syndrome (20 per cent)
- Links to other organisations (15 per cent)
- Information pack from a Down syndrome organisation (12 per cent).

A summary of the overall survey responses shows significant gaps in relation to the information and support that was provided to families during pregnancy, in particular:

- 49 per cent of families felt pressure from their healthcare provider to terminate their pregnancy
- 42 per cent of families said they received negative information about Down syndrome
- 42 per cent of families were told information about Down syndrome by health professionals that they know now to be untrue
- 45 per cent of families felt that they did not receive appropriate support during pregnancy
- 47 per cent of families felt that they did not get the information they needed to understand Down syndrome during their pregnancy
- 69 per cent of families felt that the information provided did not give them an understanding of the lived experiences of people with Down syndrome and their families.

Some participants reported having received good support and balanced information from health professionals. Approximately 36 per cent of respondents felt that they received appropriate support during their pregnancy in relation to the high chance result.

Survey respondents who indicated that they had received appropriate support were also more likely to:

- Receive more information including a Down syndrome information pack and links to websites and other organisations.
- Have an opportunity to meet a family with a child with Down syndrome or an adult with Down syndrome.
- Be connected to an online Down syndrome support organisation.

They were also less likely to:

- Report they received verbal information about options to terminate the pregnancy when told their high chance result
- Indicate that they felt pressure to terminate their pregnancy.

Information provided before screening – implications for informed choice

> *"We were just going along with what the doctor said."*
>
> Hayley, mother to 2-year-old Antonio

Informed choice about prenatal screening begins with prospective parents receiving pre-test counselling, which allows them to decide what, if any, prenatal screening options are right for them. Prospective parents reported that they had understood prenatal screening was a required part of pregnancy care. One mother said "I honestly thought you had to."

Many prospective parents approach screening with a focus on finding out the sex of the foetus, and are often not aware of the additional genetic information they may also learn.

The survey results describe insufficient information was provided to many prospective parents as they made decisions about screening. The survey data shows:

- 30 per cent were not provided any information about conditions being screened for
- 52 per cent were provided with information about the reliability of the tests
- 41 per cent were provided with information about the difference between screening and diagnostic tests
- 31 per cent were told that screening is a choice and not required as part of pregnancy care
- Many were given no information other than the costs involved
- Of those who chose not to undertake diagnostic testing after receiving a high chance screening result, 42 per cent felt pressure from medical professionals or others to do so.

Disability academic and activist Tom Shakespeare highlights the contexts within which prenatal screening decisions are made and the potential for free and fully informed choice to be compromised. He points to the following areas in which choice may be undermined:[16]

① **Information** – Where no information is provided about a condition being screened for such as Down syndrome, it can carry the implication that 'it is obvious that someone would want to avoid these conditions, the only question being whether the test is effective in providing the diagnosis.'[17] Shakespeare contends that it is only by providing full information about the lives of people who have a particular condition and their families that prospective parents can be enabled to make an informed decision about whether they wish to avoid this possibility for their own family.

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[16] Shakespeare,T., 2014. Disability Rights and Wrongs Revisited. New York: Routledge. p.131

[17] Ibid, p.132

② **Attitude and behaviour of medical professionals** – Choice is undermined if health professionals imply that women have a duty to have tests or terminations; if they are unsupportive to women who decline screening or if they are prejudiced about disability.

③ **Routinisation of screening services** – Sometimes referred to as ‘screening creep’, the autonomy of prospective parents is compromised when the focus of screening services is on a conveyor belt approach to the processing of women.

④ **Broader cultural context** – Prospective parents may be largely ignorant about disability. They may hold negative attitudes and fear that the life of their child will be marked by suffering. These fears may be reinforced by negative cultural stereotypes and messages about the benefits of genetic research and prenatal screening.

Responses to the Down Syndrome Australia survey raise concerns about each of the areas identified above as having potential to impact free and fully informed choice. One respondent, a paediatric doctor, discussed how the availability of pre-natal screening, which is often focused on the identification of Down syndrome, is likely to impact perceptions about the quality of life of those living with Trisomy 21. She said, “Because you can test for it, people must think it must be pretty bad.” As a paediatric doctor, she had experience working with children with life limiting diagnoses and also children with Down syndrome. Through her work with children with Down syndrome and their families, she said, “The children I saw had a good quality of life, they were happy. I’ve never met a family who had a child with Down syndrome who didn’t just love their kid.”

Unfortunately, recent research shows that people with intellectual disability still face stigma and discrimination in the community. A 2018 Victorian survey[18] reveals community attitudes towards people with disability. The following results provide an insight into the broader cultural context in which decisions about prenatal screening take place:

- 75 per cent of respondents indicated that they were likely or very likely to ‘pity’ a person with intellectual disability
- 56 per cent of respondents indicated that they were likely or very likely to avoid a person with an intellectual disability.
- One in five responded that children with disability should only be educated in special schools, and that employers should be allowed to refuse to hire people with disability.

One Down Syndrome Australia survey respondent remarked “Not many other parents have to be confronted with people who would rather terminate than have a child like yours.” Other parents reflected on the need to challenge incorrect information and assumptions made by health professionals during their pregnancy, and worried that others who were less informed would have made high stakes decisions based on misinformation.

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[18] Department of Health and Human Services, 2018. *Survey of Community Attitudes Towards People with Disability*, Victorian Government.

Pre-test counselling offers a unique opportunity for people to receive accurate information about Down syndrome and other chromosomal differences. Health professionals should be equipped to provide prospective parents with accurate information before and after screening tests, including the opportunity to connect with people with Down syndrome and their families.

The increased uptake of NIPT early in pregnancy means GPs are often responsible for providing information to prospective parents as they make decisions about screening. Often it is GPs providing the results of screening and providing initial information and counselling when results are unexpected. GPs may know little about the genetic conditions which are the focus of prenatal screening. The capacity of GPs to provide pre and post-test counselling needs to be enhanced by addressing the challenges of providing this service within the context of a busy general practice and by access to training, such as the University of Melbourne Genetics in Pregnancy online course.

An overview of information and support provided during pregnancy

Survey results showed:

49 per cent of families felt pressure from their healthcare provider to terminate their pregnancy

42 per cent of families said they received negative information about Down syndrome (43 per cent indicated they received neutral information and 15 per cent received positive information)

42 per cent of families were told information about Down syndrome by health professionals that they know now to be untrue

45 per cent of families felt that they did not receive appropriate support during pregnancy

47 per cent of families felt that they did not get the information they needed to understand Down syndrome during their pregnancy

69 per cent of families felt that the information provided did not give them an understanding of the lived experiences of people with Down syndrome and their families.

Lack of support

"I need to talk to someone; I don't know what to do.
What does this health condition mean?
What does Down syndrome mean?"

Louise, mother to 7-month-old Teddy

Many respondents described being told about their high chance result or diagnosis over the phone or while they were at work.

Surveyed families reported the most common information provided to them was verbal information about termination (52 per cent), with many reporting no information beyond details about termination was provided to them. Respondents shared the following experiences:

"I was told of the result over the phone while I was at work and then asked when I would like to book in for a termination because there was only a short window of opportunity."

"I was rung at work by the place I got the test done, told the OB had said was okay to tell me result by phone. Told the result. Vaguely remember asking questions it's all a blur of emotions. Don't remember much. Remember ringing OB office and first response was to arrange termination appointment."

"The voice mail said that there was a 1:2 chance our child had Down syndrome and to contact our doctor if we wanted further tests."

Only 37 per cent of respondents received a verbal summary of Down syndrome, and even less (20 per cent) were given a flyer about Down syndrome. Links to other organisations were given to 15 per cent of respondents and a Down Syndrome information pack was provided to only 12 per cent of the surveyed families.

Where families were given a verbal summary about Down syndrome, they often reported receiving only negative information about potential health complications or inaccurate information:

"We were told she would never walk, talk, she would be deaf, she wouldn't be able to breast feed, she would have numerous heart surgeries and be sick all the time. She would get cancer."

Respondents reported wanting information that could help them understand what a diagnosis of Down syndrome might mean for their child and for their family. Many families reported that they were required to seek out that information themselves.

Respondent Ellen recalls the night after receiving unexpected screening results as ‘the worst night of [her] life”, as she tried to find information online about the conditions the screening had indicated. It was in an online chat group that she learnt from other women that the results were not diagnostic.

Another respondent described there was a complete lack of support at the time the screening result was shared:

“They called me at work and let me know over the phone and then provided details to talk to a genetic counsellor. Which we couldn’t see for at least a week after hearing the news.”

Zoey’s experience

When Zoey received a call from her obstetrician with her amniocentesis results, she expected to be be asked to attend his office.

The call came when Zoey was at school teaching a room full of four-year-olds, when he stated over the phone “It’s not good news. Your baby has Down syndrome… and I’ve scheduled a termination for tomorrow.”

With twenty children watching on, Zoey had no chance to process what she had just been told. She made an appointment to see him in person later that day to talk further.

“We were in there for five minutes, it was awkward, we were in shock,” Zoey says. “He gave us an information sheet and from that point, it was neglect.”

Many parents described making a connection with their local Down syndrome association as a turning point, however, many had to seek this out themselves. As survey respondent Hayley noted, she would have benefited from “any information pack or connections to groups for support instead of being told they won’t have a quality life or walk or be able to do many other things which is completely untrue”.

Misinformation and negative attitudes about Down syndrome

The consultant's words were:

"I can fix his heart, but there is no way I can fix that he has Down syndrome."

Julia, mother to 3-year-old Grayson

Decisions about prenatal screening are made in the context of a society where stigma and bias towards people with intellectual disability exist. It is within these early pregnancy consultations that health care professionals can correct misconceptions about life with Down syndrome and provide evidenced-based, balanced, and up-to-date information.

The survey uncovered many examples of bias and stigma expressed by health professionals towards Down syndrome and disability and showed the negative impact this often had on how prospective parents experienced pregnancy and caring for their child post birth.

Some of the common themes that emerged from the survey include:

- Low expectations such as the child never being able to walk, talk, go to school, dying young, or not have a meaningful life
- A focus on potential medical complications, many of which can be treated and supported such as heart conditions
- Negative and inaccurate views about impact on relationships with partner or siblings
- Inaccurate stereotypes
- Inaccurate information about potential costs of therapies etc.

Respondents reflected on the misinformation and low expectations of Down syndrome from health care providers:

"It felt like the room went dark and it felt like they told us our baby died. That's how they acted as if they were delivering terrible news."

"No current information was provided about Down syndrome, and I was told that most children with Down syndrome end up living in an institution. While I wasn't specifically spoken to about termination, I was told a story about another patient who had declined the testing and ended up having a child with Down Syndrome – the insinuation was that if she had of known perhaps, she would have made a different decision. It wasn't at all helpful."

“The genetic counsellor called me and was very sad for me. I have been trying to track her down ever since to tell her that my son is the best thing that’s ever happened to me.”

“My doctor cried when she told me.”

“I felt like people were giving me sympathy. After waiting seven years for a positive result and now to be given sympathy instead of feeling nothing but extreme excitement was shattering.”

“We were given all the negatives about Down syndrome and that it would negatively impact our lives and I was given huge pressure to terminate. No support for my decision to continue.”

“Once we had the amnio though we were told outdated information that was scary and inferred we had a high chance of never being able to leave the house with our child and how it would possibly/likely negatively impact our older child and our parental relationship.”

“I had quite a bit of fear regarding having a child with Down syndrome. Would have been great to talk with someone about how wonderful life can be with a child with Down syndrome.”

“I was told my child was not really human, she would have a body like a human but nothing inside. I was told she would never be able to show she loved us. But we would love her like a family pet.”

One respondent recalled receiving a phone call from the ultrasound clinic to share the high chance screening result:

“There was dread in her voice… I could tell she was so uncomfortable and just wanted to get off the phone. In the end, she didn’t even tell me the gender, I had to ask her what the gender was. It was as if she thought I wouldn’t even care he was a boy because I must be so upset to have a baby with Down syndrome.”

Dismissing the lived experience of people with Down syndrome

> *"I thought surely there must be people with Down syndrome who have amazing fulfilled lives."*
>
> Julia, mother of 3-year-old Grayson

The lived experience of people with Down syndrome and their families must form part of the accurate and balanced information prospective parents deserve both pre-test and following an unexpected result.

Many respondents described messaging from health professionals focused on a burden of care narrative. Respondents shared that this was not the case in their lived experience and worried what this incorrect information would mean for other families receiving unexpected prenatal screening results.

42 per cent of survey respondents said they were told information about Down syndrome by health professionals that they now know to be untrue. Survey respondent Julia said, "Our life now is the complete opposite of the life they painted it to be." She was told by health professionals that she needed to consider the wellbeing of her six-year-old daughter. She says now her son and daughter are inseparable and have "a relationship like no other siblings I've known." Julia's daughter recently said, "Imagine how boring he would be if he didn't have Down syndrome."

Many survey respondents found health care providers showed a reluctance to offer information to prospective parents about how to connect to families who have a child with Down syndrome.

When Louise was trying to find information about life with Down syndrome, she was warned by one paediatrician to 'beware of true believers' who would just try to spread positive messages about Down syndrome. By chance, Louise heard the mother of a child with Down syndrome speaking on the radio and got in contact with her. This mother was able to provide her with information about how she could connect with other parents of children with Down syndrome. This connection to families with lived experience of Down syndrome was a significant support for Louise: "Finally I felt connected and grateful because I could get a realistic idea of where kids were at different stages."

An obstetrician and parent to a child with Down syndrome, said in the survey that she felt many other doctors might be concerned that by offering to connect parents with people with lived experience, it may be perceived as obstructing the path to termination. She said guidelines should be implemented to clarify expectations about presenting balanced information to prospective parents, as a means of protection for both the expectant parents and the doctors.

Another respondent was told by health professionals that people with Down syndrome seen in the media were the exception, and that most people with Down syndrome will not achieve what they have.

It's uncertain why some health professionals do not present information about people with Down syndrome having the ability to live full and happy lives, and that generally their families will continue to function well. While some people believe that positive assessments of family life involving a child with Down syndrome are inauthentic, current research shows that there is little reason to believe that bringing a child with Down syndrome into a family today will have an adverse effect on family life.[19]

Part of the challenge may be related to the limited training that health professionals receive about intellectual disability. Medical professionals generally have minimal training in intellectual disability health care, with only an average of 2.6 hours in medical degrees,[20] and there is variability in the amount of content included in nursing degrees. Curriculum development has been identified as a key short-term action in the National Roadmap on Intellectual Disability and Health.[21]

Community stigma and attitudes about intellectual disability also feed into the false narrative about people with Down syndrome not leading meaningful lives. Stigma and discrimination are social processes that reflect and reinforce power imbalances. According to Professor James Livingston, stigma researcher at Saint Mary's University in Canada:

> *'Stigma reveals more about society than it does about individuals who carry stigmatised characteristics or persons who harbour negative beliefs about others. It sheds light on who in society has access to the power and privilege necessary to define rules and apply sanctions for violating them – those who do become the beneficiaries of stigma; those who do not become its subjects.'*[22]

As highlighted above, the 2018 Victorian Survey of Community Attitudes towards People with Disability[23] revealed community attitudes that were particularly stigmatising of people with intellectual disability. There is clearly a need to challenge misconceptions and stereotypes about what it means to live with Down syndrome and to recognise that stigmatising attitudes, or fear of these attitudes, have the potential to impact the ability of prospective parents to make free and fully informed choices about the future of wanted pregnancies.

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[19] Kaposy, C., 2018. Choosing Down Syndrome: Ethics and New Prenatal Testing Technologies, MIT Press, Cambridge, Massachusetts.

[20] Trollor J, Ruffell B, Tracy J, Torr J,Durvasula S, Iacono T, Eagleson C and Lennox N (2016), Intellectual Disability Health Content within Medical Curriculum: an audit of what our future doctors are taught **bmcmededuc.biomedcentral.com/articles/10.1186/s12909-016-0625-1**

[21] **National Roadmap for Improving the Health of people with Intellectual Disability**

[22] Final report of the Royal Commission into Victoria's Mental Health System (2021) Volume 3, page 523

[23] Department of Health and Human Services, 2018. Survey of Community Attitudes Toward People with Disability, Victorian Government.

Pressure from health care providers to terminate the pregnancy

The GP said, *"Get it fixed and don't think about it."*

Ellen, mother of 10-year-old Owen

Nearly half (49 per cent) of families surveyed felt pressure from their healthcare provider to terminate their pregnancy. Many specific examples were provided by respondents where health care professionals gave them directional advice about termination following a high chance screening result or diagnosis. Often opinions about termination were given in the context of other inaccurate or unbalanced information about potential health complications or the potential impact on the family:

"The doctor said "just abort and try again. You are young. A disabled child will ruin your life."

"Every time we went for screening they always asked if we were terminating the pregnancy. They were more concerned about this and couldn't believe it wasn't an option for us."

"We were told having a child with Down syndrome would be a burden on the family, they would be teased, have behavioural issues and that it would not be fair on our daughter to have a sibling with Down syndrome."

"The doctor told us 'If you are asking my opinion, I would terminate.'"

Moya's experience

Like nearly half the survey respondents, Moya* felt pressure to terminate her pregnancy.

Moya had received a high chance of Down syndrome after non-invasive prenatal screening. Despite declining further invasive diagnostic testing, Moya was provided information about termination on the basis of her screening result, saying, "I got the impression that the doctor felt they needed to give me a way out."

The most unhelpful thing Moya found throughout her whole pregnancy was that even though she had clearly indicated she would not be terminating; she was asked to confirm that she was happy with that decision on every subsequent visit to her obstetrician and at ultrasounds.

"I lost count in the end, but the number of times I had to reiterate or confirm my decision was ridiculous.' She questioned this at the time, asking whether her decision could be noted in the file and that it not be revisited at each appointment but was told 'we just need to cover our bases and this is something we need to check."

"Having to so frequently confirm our decision not to terminate the pregnancy felt like our decision as a family was being questioned and invalidated each time, rather than respected."

*Name changed to protect privacy.

Many respondents said they wished their decision to continue their pregnancy had been respected by medical professionals. A survey respondent said, "I wanted the medical professionals to accept my decision not to terminate and be supportive, not judgemental."

Another respondent made it clear that her motivation for completing the survey was her desire for things to be different from her experience. She wrote: "I would like to be part of this to tell our story so that nobody else has to go through that pressure to abort."

Compromised clinical care

"My baby was having a tough time breathing when he was born. I got told to treat him like a vegetable. My heart sank."

Anonymous survey respondent

All prospective parents should be confident that the health needs of their child will be met and not overshadowed by a disability. This was unfortunately not the experience of some families, who felt the clinical care during their pregnancy was compromised following the screening results.

Survey respondent Zoey noticed that after her amniocentesis result confirmed Down syndrome, her obstetrician stopped taking measurements, only checking the baby's heart and "sending me on my way." When she was found to have dangerously low levels of amniotic fluid at her twenty-one-week scan, her obstetrician failed to follow up with her and only made an urgent referral to maternal foetal medicine after four weeks. Zoey chose to change obstetricians:

"It was like he was so cranky because we didn't do what he recommended. I'm so disappointed and sad that that was how my first pregnancy went. I needed a supportive doctor."

After changing obstetricians, Zoey had attended the public clinic on a couple of occasions for monitoring when she had noticed reduced foetal movement. On the third occasion the nurse in charge found the heartbeat and as Zoey was preparing to leave she said "Next time, perhaps we just listen for [the] baby's heart beat and say, "Yes, baby is with us" or "No, baby is not". Zoey was confused and asked for clarification, and the nurse went on to ask, "If there was something wrong with baby would you want sirens and helicopters to [the metropolitan maternity hospital]?"

Zoey became upset thinking, "here is somebody else who doesn't want me to have this baby."

The head nurse left the room, only to return a few minutes later, apologising. She explained to Zoey that there was some confusing or misleading information that had been included on her file suggesting Zoey did not want any lifesaving medical intervention for her baby. Zoey does not know how that information was included on her file, which potentially could have caused the death of her baby.

Another family described health professionals taking a negative view about outcomes:

"My baby was having a tough time breathing when he was born. I got told to treat him like a vegetable. My heart sank. I had no idea what I was getting into and being told that just made me focus on the negative. My strong baby is far from a vegetable, and I wish I could tell myself that everything would be fine."

Examples of good quality care

Some survey respondents described feeling well supported after receiving a high chance result for having a child with Down syndrome. While some families' experiences were mixed, pleasingly some described a wholly positive support experience. These examples provide an insight into the difference it can make when prospective parents receive the support they need to navigate prenatal screening decision making.

The key themes which emerged from families who felt well supported are consistent with the pathways of care for which Down Syndrome Australia has long been advocating. These include:

Informed consent prior to prenatal screening

"Our GP was fantastic and talked about why people screen and that it was important to make a decision about screening not just having a screen."

Families should be supported to make informed decisions about prenatal screening. This requires health professionals to spend time with families to discuss the screening process, provide information about the conditions being screened for, and to let families know what decisions they may have to make if they receive an unexpected diagnosis. Many respondents told us that prenatal screening was seen as routine and little information was given. There were examples, however, of this process working well.

Delivery of results using neutral language and factual information

"My obstetrician let me know my options in a neutral way."

Health professionals should provide families with a high chance result or diagnosis using neutral language such as 'chance' rather than 'risk'. Doctors need to be clear about the difference between screening and diagnostic tests and supported with education and training about providing appropriate, balanced information about the conditions which are the subject of screening.

Provision of up-to-date balanced information about Down syndrome and other conditions being screened for

"The information we were provided was just factual and neutral."

Health professionals should provide families with up to date, evidenced-based information about Down syndrome. Without balanced information, it is impossible for families to make an informed choice about the future of their pregnancy.

Referral to peer support services such as local Down syndrome organisations or online peer-support groups

“After having my daughter, she was in NICU and a social worker organised a visit from someone from our local [Down syndrome] organisation to come to the hospital for a visit. The person who came was so lovely, she gave me lots of support and I appreciated it because it was all positive and re-assuring. Engaging with someone who understands what you’re going through helps tremendously.”

Local Down syndrome organisations provide families with a child with Down syndrome with support, information and connections to other families including hospital visits, information packs and opportunities to meet with families. The Down syndrome organisations can also provide non-directive, supportive information for families who are considering their options during pregnancies. Families are not always connected with Down syndrome organisations and sometimes only get connected after searching for information or support on their own. When health professionals connect families to Down syndrome organisations, often families find the support and information provided to be invaluable.

Access to counselling and support services both before and after a decision is made as to whether to continue the pregnancy

“My GP was mostly concerned for me in the process of confirming the test results. Supporting me when I was shocked. She spoke kindly of my baby. Never suggested anything about my choices so never felt pressured. She sent me for NIPT test and also onto hospital for genetic counselling.”

Families should have access to appropriate support services to both assist in their personal decision-making and to provide support after a decision is made, regardless of that choice.

Respect for decision making

“My obstetrician knew I had declined prenatal screening and knew about my religious beliefs, she did mention that I had the option for termination once, but as I explained it was not an option, I would consider she did not mention it again and was quite respectful of my beliefs.”

When families have decided about the future of their pregnancy following unexpected screening results, this decision should be respected. They should have the opportunity to change their mind if they wish, but they should not be forced to revisit the decision at every appointment.

Existing policies and guidelines in Australia

Several national and local policies and guidelines are already in place in Australia aimed at ensuring prospective parents receive accurate information and are well supported as they navigate prenatal screening decision-making. They include:

Clinical Practice Guidelines: Pregnancy Care[24]

The Clinical Practice Guidelines (the Guidelines) were updated in 2020. The Guidelines were developed by the federal Department of Health 'to help ensure that women in Australia are provided with consistent, high quality, evidence-based maternity care.'

The Guidelines contain excellent information about how best to support women as they make decisions about prenatal screening including following unexpected prenatal screening results. Down Syndrome Australia provided significant input into the development of the section on prenatal screening as part of the consultation process in 2020.

The guidelines contain detailed information about pre-test counselling including:

'In discussing the tests so that women can give informed consent, it is important to talk in terms of 'probability' or 'chance' rather than 'risk' and to explain:

- *it is the woman's/couple's decision whether any testing takes place*
- *the chromosomal anomalies for which testing is available and the differences between these conditions*
- *the different pathways for testing (ie combined first trimester test alone, cfDNA testing as first-tier or second-tier test or in a contingent model; see Section 50.1) and the risks and benefits of each approach*
- *the testing pathway, the decisions that need to be made at each point and their consequences*
- *the need for accurate assessment of gestational age so that tests are conducted at the appropriate time*
- *that results of these tests alone indicate a probability of fetal chromosomal anomaly but do not give a definitive diagnosis of any anomalies*
- *the sensitivity, specificity and positive predictive value for the woman's age of the test and a full explanation of the reporting format of the test (eg high probability/low probability, 1 in 10, 1 in 300, 1 in 1,000)*
- *the options for women who receive a high-probability result, including information about chorionic villus sampling and amniocentesis*

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[24] Department of Health (2020) *Clinical Practice Guidelines: Pregnancy Care.* Canberra: Australian Government Department of Health.

- *a large nuchal translucency associated with normal chromosomes may indicate other anomalies which may be structural (eg diaphragmatic hernia, cardiac anomaly) or genetic (eg Smith-Lemli-Opitz syndrome, Noonan syndrome)*
- *factors that increase the probability of fetal chromosomal anomalies (advanced maternal age, family history of chromosomal anomalies)*
- *where and how tests can be accessed if the woman chooses to have them*
- *the availability of evaluated decision aids (eg the Ottawa Decision Framework) (Arimori 2006; Nagle et al 2006; 2008)*
- *the costs involved for the woman and the timeframe for receiving results.'*

'Women may choose not to have a test or may choose to proceed directly to a diagnostic procedure instead (e.g. due to a preference to receive definitive information and/or concerns about the sensitivity of available tests). The choice a woman and her partner make about testing should not influence the subsequent care she receives.'

The Guidelines contain similarly appropriate and useful information in relation to practitioners supporting pregnant women where a pre-natal diagnosis has been made including:

'It is very important that there is no negative commentary on the condition diagnosed and that women do not feel pressured into any course of action. Accurate information about the chromosomal condition should be given and women offered information about relevant support organisations.' (Organisations listed include Down Syndrome Australia)

'Counselling should be sensitive to the nature of decisions to be taken, should respect individual decisions and allow time to reach decisions.'

The Royal Australian and New Zealand College of Obstetricians and Gynaecologists statement *Prenatal screening and diagnostic testing for foetal chromosomal and genetic conditions*[25]

This statement is intended for use by health professionals providing antenatal care and notes *'All pregnant women should be advised of the availability of investigations for prenatal [screening] and diagnosis as early as possible in pregnancy to allow time to discuss the options available and facilitate an informed choice. An informed choice is 'based on relevant knowledge, consistent with the decision maker's values.'*[26]

The statement outlines the information that should be provided to prospective parents in relation to prenatal screening such as a description of the conditions that can and cannot be detected and a discussion of the difference between screening and

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[25] The Royal Australian and New Zealand College of Obstetricians and Gynaecologists, 2018. Prenatal screening and diagnostic testing for fetal chromosomal and genetic conditions.

[26] Ibid, p.10

diagnostic tests. While the statement refers to providing prospective parents details about 'support groups and sources of further information' it could be strengthened by more overt references to offering opportunities to connect prospective parents with people who have lived experience of conditions such as Down syndrome. The definition of Down syndrome included in the statement is a list of 'clinical manifestations' and conditions.

National Roadmap for Improving the Health of People with Intellectual Disability[27]

National Roadmap for Improving the Health of People with Intellectual Disability (the Roadmap) was launched in August 2021 and is a collaborative approach to addressing the needs of people with intellectual disability in Australia. The Roadmap forms part of the Primary Health Care 10 Year Plan and will support health and wellbeing outcomes under Australia's Disability Strategy (2021–2031).

Of particular relevance to prenatal screening is Key Action 3 of the Roadmap which notes: *Commonwealth Department of Health to work with PHNs, professional colleges and disability advocacy organisations to ensure balanced, evidence-based information about intellectual disability and related diagnosis (e.g Down syndrome) is available to parents during the pre- and post-natal period.*

National Women's Health Strategy 2020–2030[28]

This strategy highlights the need for women and girls to be 'informed and empowered to be part of the decision making process.' Priority area 1 – Maternal, sexual and reproductive health highlights the need to promote women's 'control over their own bodies, reproductive choices and lifestyle'. Detail within priority action point 3 'Ensure women planning pregnancy are aware of screening tests available to them prior to and during pregnancy' is particularly relevant to prenatal screening.

Medicare Rebates

Medicare rebates are available for up to three non-directive pregnancy support counselling sessions provided by eligible GPs, psychologists, accredited social workers and mental health nurses on referral from a GP, however they are not widely used.

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[27] Department of Health (2020) *National Roadmap for Improving the Health of People with Intellectual Disability*. Canberra: Australian Government Department of Health.

[28] Department of Health (2018) *National Women's Health Strategy 2020–2030*. Canberra: Australian Government Department of Health.

Implementation of Guidelines

The need to ensure not only the existence but the implementation of guidelines is recognised in the foreword of the Pregnancy Care Guidelines which notes *'The next challenge is to facilitate the uptake of the Guidelines and their incorporation into routine care so that the women of Australia receive the highest possible quality of maternity care.'* Unfortunately, this discussion paper provides many examples of current practice that demonstrates further, urgent work is needed to ensure women who are navigating prenatal screening decision making are receiving consistent, high quality, evidence-based care and support.

The UK *Tell it Right, Start it Right* program is an example of an effective approach which could be adapted within Australia to support the implementation of existing guidelines described above.

Tell it Right, Start it Right was developed by the Down Syndrome UK association in response to concerns that health professionals were not always confident and comfortable speaking to families about Down syndrome and disability and found delivering a high chance screen result or diagnosis challenging. The program offers training to clinicians to ensure they have accurate and balanced information about living with Down syndrome and to improve the way that news of a high chance screening result or diagnosis is shared.

The program has led to significant changes in how health professionals in the UK provide information and support to women around prenatal screening.[29] The qualitative data from the UK suggests that an educational program has the potential to have a significant impact both on health professionals as well as families.[30]

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[29] Bryant, L. D., Puri, S. C., Dix, L., & Ahmed, S. (2016). Tell it Right, Start it Right: An evaluation of training for health professionals about Down syndrome. *British Journal of Midwifery,* 24(2), 110-117.

[30] Bryant, L. D., Puri, S. C., Dix, L., & Ahmed, S. (2016). Tell it Right, Start it Right: An evaluation of training for health professionals about Down syndrome. *British Journal of Midwifery,* 24(2), 110-117.

Conclusion

Families have a right to accurate information about disability and Down syndrome before being asked to make decisions about the future of their pregnancies. The Down Syndrome Australia survey, together with other recent research and experiences, shows that there is an urgent need for change to ensure that prospective parents are enabled to make informed choices. Prospective parents should be able to rely on health professionals for accurate and balanced information and be given the time that they need to make informed decisions, both pre and post screening.

Guidelines alone are not enough to build the capacity of health professionals to keep pace with the wider adoption of rapidly evolving prenatal screening technology. A focus on the implementation of existing guidelines, including the recent recommendation made as part of the National Roadmap on Intellectual Disability and Health, will make a difference to how prospective parents experience prenatal screening together with improved access to education for health professionals.

The following seven recommendations for change outline a clear pathway to improve the prenatal screening and care experience for prospective parents who have received a high chance screening result or diagnosis of Down syndrome.

Recommendations for change

Acknowledge and Review

1. Disability Royal Commission to hold a hearing about prospective parents' experiences of prenatal screening including experiences of pressure to terminate their pregnancy and impacts on prenatal care.

Implementation of Guidelines

2. As part of the work on the Intellectual Disability and Health Roadmap, the federal Department of Health to work with Down Syndrome Australia to develop a national strategy focused on appropriate pre- and post- natal support for parents of children with Down syndrome including appropriate referral and connection to local Down syndrome organisations after a prenatal diagnosis or screening result.
3. State and territory departments of health to work with local Down syndrome organisations to support delivery of appropriate information, peer-connection, and support at the local level.

Improved Care Pathways

4. As part of the current Primary Care Enhancement Program within the National Roadmap on Intellectual Disability and Health, strengthen care pathways for prospective parents choosing to continue their pregnancy following unexpected prenatal screening results. This must include health professionals being linked to local Down syndrome organisations to access information, training, and support.
5. Inclusion of a Medicare Benefits Schedule item available to GPs and obstetricians for prenatal screening pre- and post-test counselling.

Improved training and awareness for Health Professionals

6. Training for health professionals on intellectual disability has been identified as a priority in the National Roadmap on Intellectual Disability and Health. The Department of Health must work in partnership with Down Syndrome Australia to ensure that the training developed and embedded in health professional curriculum includes comprehensive information in relation to prenatal screening decision-making and support.
7. Federal government to partner with the Down Syndrome Federation to create a targeted education and awareness campaign for health professionals which includes the lived experiences of people with Down syndrome and their families modelled on the UK *Tell it Right, Start it Right* UK programme.

Appendix 1

An overview of prenatal screening options in Australia

Prenatal testing is divided into two types: screening tests and diagnostic tests.

Screening tests

Screening tests are used to identify the likelihood of a baby having a chromosomal difference such as Down syndrome. They do not provide a definitive diagnosis. There are two types of screening tests: maternal blood screening and ultrasound examination of the baby.

Blood screening tests are used to identify the likelihood of a baby having Down syndrome and do not provide a definitive diagnosis. They are:

Combined first trimester screening

The first trimester combined screening test is available between 11 to 13 weeks of pregnancy. This is a two-part test which involves a blood test and a first trimester ultrasound.

By combining the age of the mother, the results of the ultrasound, together with the results of two proteins in the mother's blood, a calculation can be made of the chance of having a baby with Down syndrome. This test is 85 to 90 per cent accurate. If the resulting calculation indicates an increased chance of the baby having Down syndrome, prospective parents are offered a confirmatory diagnostic procedure such as chorionic villus sampling (CVS) or amniocentesis.

Second trimester screening

Second trimester screening is available from 14 to 22 weeks of pregnancy. It uses the measurement of several substances in the blood, gestation of the pregnancy, maternal age, and weight to provide an estimate of the chance of your baby having Down syndrome. If the resulting calculation indicates an increased chance of the baby having Down syndrome, prospective parents are offered a confirmatory diagnostic test (amniocentesis).

Non-invasive prenatal testing

Non-invasive prenatal testing involves a blood test taken from a pregnant woman, at least 10 weeks into the pregnancy. It looks at fragments of cells (DNA) found in placental blood. The woman's blood contains a mixture of her own cell fragments and those from the placenta. Therefore, NIPT screens for fragments of placental cells, NOT the baby's cells/DNA. Placental blood is not necessarily the same as the baby's blood which is why non-invasive prenatal screening – whilst considered up to 99 per cent accurate – is not a diagnostic test.

Ultrasound screening

Most pregnant women are also routinely offered an ultrasound examination at 19 to 20 weeks of pregnancy, which can sometimes show other subtle signs of Down syndrome. If this happens you may be offered an amniocentesis as a confirmatory diagnostic procedure.

Diagnostic tests

Chorionic villus sampling (CVS)

Usually conducted between 11 and 13 weeks gestation, CVS is an invasive procedure that collects a small sample of placental tissue. With ultrasound control, a fine needle is guided through the lower abdominal wall into the placenta and a biopsy of the placenta tissue (chorionic villi) is taken. While CVS is considered diagnostic, there is a small chance that the placenta and the foetus will have different chromosome counts – this is known as confined placental mosaicism.

Amniocentesis

Usually conducted from 15 to 18 weeks gestation, amniocentesis is an invasive procedure where a fine needle is passed through the maternal abdomen and uterine wall into the amniotic fluid around the foetus to obtain a sample of amniotic fluid.

These invasive tests are the only way to obtain a prenatal diagnosis. Both tests are associated with an increased risk of miscarriage (above the usual background risk).

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