

Position paper on prenatal screening and diagnosis

About Down Syndrome Australia

Down Syndrome Australia is the peak body and disability representative organisation for people with Down syndrome in Australia. Our purpose is to influence social and policy change, and provide a national profile and voice for people living with Down syndrome.

We work collaboratively with our member and partner organisations to achieve our mission. Our vision is an Australia where people living with Down syndrome are valued, reach their potential, and enjoy social and economic inclusion.

Summary

The purpose of this paper is to set out Down Syndrome Australia's position on prenatal screening and diagnosis for Down syndrome. This paper provides a basis for advocacy to government by identifying opportunities to improve the experiences of prospective parents navigating prenatal screening. Down Syndrome Australia stands ready to collaborate with government to effect much needed change.

Down Syndrome Australia understands that access to prenatal screening and diagnosis is important for many families. We respect the right of prospective parents to undertake prenatal screening, if they choose to do so, and to make decisions about diagnostic testing and whether to continue a pregnancy based on their own circumstances and beliefs.

Despite the wider use of prenatal genetic screening, research focusing on consumer experience consistently highlights deficits in the support and information available to prospective parents¹. These deficits compromise informed choice as consumers navigate high-stakes decision-making in time-pressured circumstances. All families have the right to accurate, up-to-date information and support.

Down Syndrome Australia calls on the Federal and state governments to urgently address the gaps in the provision of prenatal screening and diagnosis.

Prospective parents must be enabled to make informed decisions about prenatal screening and diagnosis at each key decision point.

¹ Lafarge C, Usher L, Mitchell K, Fox P. (2020) The role of rumination in adjusting to termination of pregnancy for fetal abnormality: Rumination as a predictor and mediator of posttraumatic growth. *Psychological Trauma* 2020;12(1):101-9; Down Syndrome Australia (2021) "We all have a lot to learn": Prenatal screening for Down (is there a word/s missing here after "Down")

Down Syndrome Australia calls for government to support:

1. A prenatal genetic screening support service pilot program

A pilot prenatal genetic screening support service would be embedded within an existing laboratory or healthcare setting to address the unique, and increasingly common, challenges faced by individuals navigating prenatal screening and diagnostic testing decision making. The outcomes of this pilot program should be regularly monitored and refinement of the approach to prenatal screening and diagnosis based on this evidence prior to a full roll out.

2. The establishment of a national coordination role

Down Syndrome Australia calls for funding to establish a national coordination role aimed at identifying and addressing barriers to the implementation of existing prenatal screening practice guidelines and key actions within the National Roadmap for Improving the Health of People with Intellectual Disability.

3. A national information and support project

Down Syndrome Australia calls for funding to deliver a national project to improve consumer-focused, accessible, consistent and coordinated information and support available to prospective parents navigating prenatal screening for Down syndrome.

4. A national public awareness campaign to tackle the stigma associated with conditions which are the subject of prenatal screening such as Down syndrome

Down Syndrome Australia recognises that the availability of screening and diagnostic tests for Down syndrome negatively impacts, or has the potential to negatively impact, people with Down syndrome and their families. Down Syndrome Australia asks government to recognise and address this impact by funding a public awareness campaign to tackle the stigma associated with Down syndrome and intellectual disability.

Background

Prenatal testing for Down syndrome has been available since the late 1960s when prospective parents over the age of 35 were offered amniocentesis to test for Down syndrome. Blood screening tests were developed in the 1980s after an association was discovered between changes in observed levels of biochemical markers and an increased chance of Down syndrome. In the early 2000s, ultrasound examinations were utilised to further enhance the reliability of the screening approach through the measurement of nuchal translucency thickness. These tests can identify prospective parents with an increased chance of having a child with Down syndrome.

In 2013 a new screening approach, non-invasive prenatal screening (most commonly referred to as NIPT) became available in Australia. NIPT screens for various chromosome conditions prenatally, including Down syndrome, through an examination of short fragments of DNA released into the maternal circulation from the placenta. The analysis of maternal blood can be performed from 10 weeks gestation. NIPT has higher performance (both sensitivity and specificity) than previous screening tests, but it does not provide a definitive result.

NIPT is often marketed as '99% accurate' and prospective parents may not understand the potential for false-positive screening results to occur. Following an increased chance screening result, the only way to determine a diagnosis of Down syndrome prenatally is via invasive diagnostic testing (amniocentesis or chorionic villus sampling).

In Australia, NIPT is currently not subsidised through the Medicare Benefits Schedule however is widely self-funded (current cost between \$400 and \$500). Uptake of NIPT is growing and differs across states and between socio-economic groups with >50% to 75% of prospective parents receiving private obstetric care utilising NIPT as a primary screen, compared with <25% of public patients².

As the use of NIPT expands, more prospective parents will receive results indicating their developing baby has an increased chance of Down syndrome or another chromosomal condition. Prospective parents have the right to a nationally consistent model of care that includes accurate, up-to-date information, free from bias and stigma, reflecting the lived experiences of conditions which are the subject of screening. Access to genetic counselling and attuned psychosocial support is vital as prospective parents navigate decisions about diagnostic testing and ultimately whether to continue or terminate a pregnancy.

Primary healthcare providers such as general practitioners are increasingly expected to share screening results with prospective parents and offer information and support as consumers seek to understand what the genetic information means for their pregnancy and to navigate diagnostic testing and decision-making about whether to continue or terminate the pregnancy.

Unexpected prenatal screening results can be distressing and traumatic for prospective parents³ and challenging for healthcare providers tasked with providing information and support.

It should be noted that access to balanced information was an issue of concern even before NIPT became available. However, NIPT means prospective parents are often making decisions around screening earlier in pregnancy. GPs are taking on the responsibility of providing information about screening and supporting prospective parents when increased chance results are returned. This evolving practice context means it has become even more critical to ensure that GPs can provide accurate information and access to relevant counselling and support.

² Gadsboll K, Petersen OB, Gatinois V, Strange H, Jacobsson B, Wapner R, et al. Current use of noninvasive prenatal testing in Europe, Australia and the USA: a graphical presentation. *Acta Obstetrica et Gynecologica Scandinavica* 2020;99(6):722-30.

³ Hodgson J, McClaren BJ. Parental experiences after prenatal diagnosis of fetal abnormality. *Seminars in Fetal Neonatal Medicine* 2018;23(2):150-4.

Down Syndrome Australia also recognises that prenatal screening and diagnosis has the potential to further entrench the bias and stigma experienced by people living with conditions which are the subject of screening such as Down syndrome. Careful attention to non-discriminatory framing and language must be given to avoid devaluing the unique contributions people living with Down syndrome make to their families and communities.

There are a number of Australian practice guidelines and position statements supporting evidence-based practice in relation to prenatal screening and diagnosis. Unfortunately, research continues to demonstrate significant barriers to implementation resulting in families not receiving the information and support they need. Negative community attitudes and outdated information about intellectual disability mean that many families and health professionals are unaware of the contemporary reality of raising a child with Down syndrome.

Discussion

This paper is organised around the three key decision-making points that demand tailored support and information for prospective parents:

1. When screening is offered
2. Following an increased chance screening result
3. Following confirmed diagnosis

At each of these points, prospective parents have the right to accurate, balanced and consistent information and psychosocial support.

The paper sets out gaps and opportunities for transformative change that would enable prospective parents to receive information and support which respects diversity as well as a woman or pregnant person's right to decide to continue or to terminate a pregnancy.

Key decision point 1: When screening is offered

The context, language used and the way prenatal screening is presented to prospective parents has the potential to enable or compromise informed decision-making. Health care professionals must attend to cultural, social and emotional considerations as well as the technical aspects and process of screening.⁴

Options available to prospective parents at this decision point are:

- Accept prenatal screening
- Decline prenatal screening

Research⁵ examining how prospective parents have experienced NIPT both in Australia and internationally, identified the need to safeguard informed decision-making by increasing the availability of trustworthy information about NIPT

⁴ McKinn S, Javid N, Newson AJ, Freeman L, Bonner C, Shand AW, et al. (2022) Clinician views and experiences of non-invasive prenatal genetic screening tests in Australia. *Australian and New Zealand Journal of Obstetrics and Gynaecology* 2022;62(6):830-7.

⁵ 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.

and enabling access to counselling. Large gaps in clinicians' knowledge about the experience of raising a child with Down syndrome was also identified.⁶

Choices about screening take place in the context of a society that displays bias and stigma against people with disability⁷. Narratives of people with disability 'suffering' or being a burden to families are common and at odds with the lived experience of most people with Down syndrome and their families.⁸

According to RANZCOG⁹, prenatal screening should 'be voluntary and only undertaken when the woman has been informed about the nature of the screening test, the possible results, and the options available to her.'

Responses to a 2021 survey conducted by Down Syndrome Australia of 320 parents of a child with Down syndrome highlighted significant gaps in the information provided to them as they made decisions about screening. Results included:

- 30% of respondents reported not being provided any information about the conditions which were the subject of screening
- Most respondents (59%) were not provided with information about the difference between screening and diagnostic tests

One respondent, a paediatric doctor, reflected on the meaning ascribed to life with Down syndrome more broadly due to it being the subject of prenatal screening saying 'Because you can test for it, people must think it is pretty bad.'

Pre-test counselling should offer an opportunity for all prospective parents to receive accurate, balanced information about Down syndrome and other chromosomal conditions including the opportunity to connect with Down syndrome organisations to understand perspectives of families or people with Down syndrome.

Opportunities to improve the experiences of prospective parents when screening is offered:

a) Enable access to evidence-based up-to-date information for consumers about prenatal screening and diagnostic testing for Down syndrome to allow prospective parents to decide which screening, if any, is right for them.

This should include updating, publicising and embedding existing resources such as the www.prenatalscreening.org.au website and the information and support offered by the Down syndrome federation across Australia.

b) Enhance the capacity of GPs and other health care professionals to provide pre-test counselling

⁶ Ibid.

⁷ Bollier, AM., Sutherland, G., Krnjacki, L., Kasidis, V., Katsikis, G., Ozge, J., & Kavanagh, AM. (2021). Attitudes Matter: Findings from a national survey of community attitudes toward people with disability in Australia. Centre of Research Excellence in Disability and Health, The University of Melbourne.

⁸ Skotko, B., Levine, S., & Goldstein, R. (2011). Self- perceptions from People with Down Syndrome. American Journal of Medical Genetics, Part A, 155: 2360-2369.

⁹ Royal Australian and New Zealand College of Obstetricians and Gynaecologists and the Human Genetics Society of Australasia. (2018). Statement on Prenatal screening and diagnostic testing for fetal chromosome and genetic conditions. <https://www.hgsa.org.au/documents/item/6110>.

GPs must be enabled to provide pre-test counselling in the context of a busy general practice. Access to high quality training, such as the University of Melbourne Genetics in Pregnancy online course, which attends not only to the reliability of the test, but also to the conditions which are the subject of screening, is vital.

c) Support increased understanding of and challenge bias and stigma about Down syndrome and intellectual disability amongst health care professionals

All health care professionals' training should include the perspectives of people with intellectual disability and an opportunity for health care professionals to reflect upon and challenge bias and stigma about disability. Down Syndrome Australia's Health Ambassadors are experienced in presenting to health care professionals and this program could be used as a model. Health care professionals need to understand that the language they use about screening and screened for conditions and the framing of screening options have the potential to compromise informed choice and reinforce bias and stigma against disability.

Key decision point 2: Following an increased chance result

When an increased chance prenatal screening result is received, prospective parents rely on timely and accurate information as they make decisions about diagnostic testing and the future of their pregnancy.

Options available to prospective parents at this decision point are:

- Diagnostic testing
- Await further clinical information
- No further testing

Too often, prospective parents receive misinformation and inadequate support following increased chance screening results with gaps reported in¹⁰:

- information about the implications of screening results such as the difference between a screening and a diagnostic test
- Access to genetic counselling and psychosocial support as consumers make meaning of new genetic information
- Up-to-date, balanced and accurate information about screened-for conditions including the opportunity to connect with people living with these conditions and their families

A lack of appropriate pre-test counselling, combined with the poor communication of increased chance NIPT results, have been acknowledged by clinicians as contributing to the potential psychosocial harm of prospective parents.¹¹

Increased chance prenatal screening results and diagnoses are often experienced by prospective parents as unexpected or shocking¹². Health care providers such as GPs, midwives and obstetricians are on the frontline,

10 Meredith, S., Brackett, S., Diaz, K. M., Freeman, K. G., Huggins, E., Khan, H., Leach, M. W., Levitz, M., Michie, M., Onufer, J., Skotko, B. G., Smith, L., Nicole White, A., Waller, T., & Ayers, K. (2023). Recommendations to improve the patient experience and avoid bias when prenatal screening/testing. *Disability and Health Journal*, 16(2), 101401–101401; Down Syndrome Australia (2021). "We all have a lot to learn" Prenatal screening for Down syndrome: a discussion paper. https://www.downsyndrome.org.au/wp-content/uploads/2021/10/DSA_Prenatal-Screening-Experiences.pdf; Valentin C, Smidt A, Barton R, Wilson NJ, How B. (2019). Mothers of a child with Down syndrome: a qualitative analysis of the perspectives on non-invasive prenatal testing. *Midwifery*, 76:118-24.

¹¹ McKinn S, Javid N, Newson AJ, Freeman L, Bonner C, Shand AW, et al. Clinician views and experiences of non-invasive prenatal genetic screening tests in Australia. *Australian and New Zealand Journal of Obstetrics and Gynaecology* 2022;62(6):830-7.

supporting expectant parents as they receive these results and navigate decision-making. Research shows that many health care providers do not feel confident discussing prenatal screening and feel they have inadequate access to resources and referral options to aid their conversations with patients¹³.

The 2021 Down Syndrome Australia survey found that the most common information (52 per cent) provided following an increased chance screening result was verbal information about termination.

“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”.

-Parent response to Down Syndrome Australia survey 2021

Only 37 per cent of respondents received a verbal summary about Down syndrome and only 20 per cent were given a flyer about Down syndrome. Where families were given a verbal summary, they often reported only receiving negative information about potential health complications or information that they now know to be untrue. The majority (69 per cent) of families feel that the information provided did not give them an understanding of the lived experiences of people with Down syndrome and their families.

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

-Parent response to Down Syndrome Australia survey 2021

Opportunities to improve the experiences of prospective parents following an increased chance prenatal screening result:

a) Enable best practice communication of increased chance prenatal screening results by GPs and other health care professionals

Together with access to training, GPs and health care providers would benefit from coaching at the time they are required to share increased chance screening results with prospective parents. Down Syndrome Australia supports the piloting of a coaching model intervention at the point an increased chance prenatal screening result is issued by the laboratory to the primary health care provider (most often a GP).

Understanding and communicating the difference between screening and diagnostic tests is vital as is the opportunity for prospective parents to connect with counsellors and people with lived experience.

b) Improve access to evidence-based, up-to-date and nationally consistent information for consumers and health care professionals about Down syndrome and prenatal screening which reflects lived experience perspectives

Down Syndrome Australia seeks funding to update and adapt an existing resource, the website prenatalscreening.org.au. The website was developed by Down Syndrome Queensland as part of a project funded by Queensland Health and informed by a multidisciplinary advisory group including clinical and lived experience expertise.

¹² Hodgson and McClaren, 2018

¹³ Cooke, E., Anderson, M., Langdon, G., Feltham, M., Staton, S., Coles, L., Thorpe, K. and Chawla, J. (2023). Clinicians' experiences of explaining prenatal screening and delivering genetic syndrome diagnoses. Brisbane, QLD, Australia: Down Syndrome Queensland.

As well as providing information to prospective parents, the website aims to improve the capacity of GPs and other health care professionals to provide accurate, up-to-date and balanced information about Down syndrome and to provide non-directional support to prospective parents as they navigate prenatal screening decision making. The site houses a comprehensive Practice resource endorsed by a wide range of organisations including RANZCOG and RACGP.

Down Syndrome Australia seeks to adapt the available information for a national audience and actively engage in the promotion of these resources.

c) Enable access to trained peer support and appropriate counselling services

Down Syndrome Australia receives no funding to support education and advocacy efforts relating to improving information about prenatal screening.

The capacity to establish a national information and support project would allow Down Syndrome Australia to develop a trained volunteer peer support network. Such a network would provide a valuable resource to prospective parents seeking to better understand the lived experience of people with Down syndrome and their families across a number of areas including the nature of Down syndrome, social, economic and familial impacts as well as specific areas including schooling and supports as an adult as examples.

In addition, a national information and support function would allow for the development and maintenance of a referral database of appropriately qualified counsellors where families are seeking additional psychosocial support.

Key decision point 3: Following confirmed diagnosis

Following confirmation of a prenatal Down syndrome diagnosis, options available to prospective parents are:

- Continue the pregnancy
- Continue the pregnancy with alternative care
- Terminate the pregnancy

The Down Syndrome Australia national survey (2021) found many examples of bias and stigma about Down syndrome and disability in the context of prenatal screening. Nearly half (49 per cent) of families reported that they felt pressure from their health care provider to terminate their pregnancy. There were many specific examples of directional advice given to respondents by health care providers following an increased chance screening result or diagnosis.

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

-Parent response to Down Syndrome Australia survey 2021

In Australia, research has found approximately 9 out of 10 pregnancies diagnosed with Down syndrome end in termination.¹⁴

¹⁴ Maxwell, S., Bower, C., & O'Leary, P. (2015). Impact of prenatal screening and diagnostic testing on trends in Down syndrome births and terminations in Western Australia 1980 to 2013: Impact of prenatal screening and diagnosis on Down syndrome, 1980-2013. *Prenatal Diagnosis*, 35(13), 1324-1330

A qualitative analysis focusing on non-invasive prenatal screening examined the perspectives of Australian mothers of a child with Down syndrome.¹⁵ Approximately half of the mothers in the study reported that they felt medical professionals expected they would terminate following a positive screening test.¹⁶

All the mothers highlighted the importance of autonomy, appropriate support including balanced, evidenced-based information and 'time and space for individual reflection.'¹⁷ 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.¹⁸

There is limited recognition of the mental health challenges faced by expectant parents who receive a suspected or confirmed diagnosis of a congenital difference such as Down syndrome. Where health care providers are not equipped to provide non-stigmatising care based on current evidence, there is an increased risk of trauma and distress. Down Syndrome Australia collaborates with and supports the work of Through the Unexpected, a registered charity which is currently developing the first national training programs.

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

Down Syndrome Australia successfully advocated for the following key action to be included in the 2021 National Roadmap for Improving the Health of People with Intellectual disability:

Commonwealth Department of Health to work with PHNs, professional colleges and disability advocacy organisations to ensure balanced, evidenced-based information about intellectual disability and related diagnoses (e.g. Down syndrome) is available to parents during the pre- and post-natal period. ^[17]

This action has yet to be implemented.

Opportunities to improve the experiences of prospective parents receiving a prenatal diagnosis for Down syndrome

In addition to Key decision point 2 opportunities:

a) **Enable access to up-to-date and balanced information about Down syndrome and non-direction support**

Training, information and guidance must be available to GPs and other health care professionals to ensure they are confident supporting prospective parents with accurate and balanced information following a prenatal diagnosis of Down syndrome. This includes awareness of additional patient resources to support pregnancy decision-making, referral options and how to connect to Down syndrome organisations to understand perspectives of families and people with Down syndrome.

Federal commitment and funding are required to implement the National Roadmap key actions and achieve tangible change in prenatal information provision to prospective parents.

¹⁵ 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

¹⁶ Ibid, p. 120

¹⁷ Ibid, p. 123

¹⁸ Ibid, p. 123

b) Improve the capacity of GPs and other health care professionals to understand and address the psychosocial impacts of prenatal screening and diagnosis

Training for GPs and other health providers must attend to the psychosocial impacts of receiving unexpected genetic information and decision making under uncertainty.

Down Syndrome Australia Position

It is not acceptable for prospective parents to be required to make high-stakes decisions about prenatal screening without accurate and balanced information and appropriate support.

Down Syndrome Australia calls on the Federal, State, and Territory governments to urgently address the gaps in the provision of prenatal screening and diagnosis. Down Syndrome Australia remains committed to engaging collaboratively with government and other stakeholders to ensure prospective parents are enabled to make informed decisions about prenatal screening and diagnosis at each key decision point.

In order to effect the urgent change required, Down Syndrome Australia calls for government to support:

1. A prenatal genetic screening support service pilot program

A pilot prenatal genetic screening support service would be embedded within an existing laboratory or healthcare setting to address the unique, and increasingly common, challenges faced by individuals navigating prenatal screening and diagnostic testing decision making. The outcomes of this pilot program should be regularly monitored and refinement of the approach to prenatal screening, diagnosis, and communication based on this evidence prior to a full roll out.

Such a service must:

- Evaluate the experiences (including psychosocial impacts) of prospective parents navigating prenatal screening and diagnosis and refine the approach based on this evidence.
- Consider the limited capacity of the genetic counselling workforce and adopt a multidisciplinary team approach to providing psychosocial support and information including, for example, counsellors, social workers and psychologists.
- Reflect the lived experience perspectives of people with conditions that are the subject of prenatal genetic screening including Down syndrome.

2. The establishment of a national coordination role

Down Syndrome Australia calls for funding to establish a national coordination role aimed at identifying and addressing barriers to the implementation of existing prenatal screening practice guidelines and key actions within the National Roadmap for Improving the Health of People with Intellectual Disability. This role would:

- Enhance the national availability and coordination of prenatal screening information, support and education as it relates to Down syndrome, supporting the National Roadmap key action relevant to prenatal information provision.
- Support national tertiary education curriculum development in intellectual disability health (as per the National Roadmap for Improving the Health of People with Intellectual Disability) to include learning outcomes and content related to pre-natal screening.

3. A national information and support project

Down Syndrome Australia calls for funding to deliver a national project to improve consumer-focused, accessible, consistent and coordinated information and support available to prospective parents navigating prenatal screening for Down syndrome. The project would include:

- Updating and adapting existing resources for a national audience (including the adaption of the website prenatalscreening.org.au developed by Down Syndrome Queensland and funded by Queensland Health).
- Enabling prospective parents the opportunity to access lived experience perspectives through trained peer supporters.

4. A national public awareness campaign to tackle the stigma associated with conditions which are the subject of prenatal screening such as Down syndrome

Down Syndrome Australia recognises that the availability of screening and diagnostic tests for Down syndrome negatively impacts, or has the potential to negatively impact, people with Down syndrome and their families. The availability of a screening test for a particular condition may further entrench existing stigma and bias against those living with the condition.

Down Syndrome Australia asks government to recognise and address this impact by funding a public awareness campaign to tackle the stigma associated with Down syndrome and intellectual disability. Such a campaign would highlight the negative impact of value-laden, deficit based language.

In particular a focus is needed on the language used to describe screening results (eg 'high risk' instead of 'increased chance'). In 2015 Down Syndrome Tasmania lodged a complaint with the Tasmanian Anti-Discrimination Commissioner regarding language used to describe prenatal screening, specifically the use of 'risk' to describe the probability of a Down syndrome diagnosis. The Anti-Discrimination Commissioner found this language to be a possible breach of the Anti-Discrimination Act 1998.

A national campaign would also contribute to changes in how health care and screening providers are presenting information in a way which respects diversity as well as a woman or pregnant person's right to decide to continue or to terminate a pregnancy following a Down syndrome diagnosis.