Position Paper on Prenatal Testing and Diagnosis

About Down Syndrome Australia
Down Syndrome Australia is 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. We work collaboratively with the state and territory Down syndrome associations 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 (DSA) position on prenatal testing (screening and diagnosis) for Down syndrome. This paper provides a basis for advocacy to government for systemic changes to the information and support provided during prenatal screening, and makes specific recommendations to improve access to balanced information.

Down Syndrome Australia understands that access to prenatal screening and diagnosis is important for many families. We respect the right of women to undertake prenatal testing, if they choose to do so, and to make decisions about whether or not to continue a pregnancy based on their own circumstances and beliefs.

Research suggests that, currently, many families may not be making fully informed choices about prenatal testing. We are concerned that some families may be making decisions that are based on negative community attitudes and inaccurate, outdated information about Down syndrome.

Down Syndrome Australia recommends that the government ensures that all women who are contemplating a test during pregnancy, have access to accurate and balanced information about Down syndrome including opportunities to connect with families. This should include improved training and education for doctors and midwives, improved access to post-screening counselling, and the development of a public awareness campaign to tackle negative community attitudes and stigma about Down syndrome and intellectual disability.

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1 Screening is not a diagnostic test and provides information on the likelihood of an outcome (e.g. chance of having a child with Down syndrome). Diagnosis of Down syndrome prenatally occurs through examination of foetal cells via amniocentesis or CVS.
Background

Prenatal testing for Down syndrome has been available since the late 1960s when women 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 are able to identify pregnant women with a higher chance of having a child with Down syndrome.

In 2013 a new screening approach became available in Australia which screens for various chromosomal conditions prenatally, including Down syndrome, through an examination of maternal blood known as a non-invasive prenatal screening (NIPS). This test can be performed as early as 10 weeks gestation and is more specific than previous screening tests, but it does not provide a definitive result. In Australia, women can access the test privately (cost is around $500). The government is currently considering applications for NIPS to be paid for publicly under certain circumstances. The outcome of this process will be decided over the next year or so.

The only way to determine a definitive diagnosis of Down syndrome is via amniocentesis or chorionic villus sampling (CVS) to obtain a sample of foetal cells for examination. These are invasive tests that also come with a potential risk of miscarriage (of around 1% above the usual background risk).

Prenatal testing can provide parents and health professionals with information in order to support the provision of the best care during pregnancy and immediately after the birth of the child. Some families access testing to obtain information to help them make a decision about whether to continue a pregnancy.

Down Syndrome Australia is concerned that women are not being given appropriate, balanced information both prior to NIPS and after receiving their results. Some families who receive a diagnosis of Down syndrome have told us that doctors questioned their choice to continue their pregnancy, or have told them that their child would have a lifetime of suffering.

It should be noted that access to balanced information was an issue of concern even before NIPS became available. However, NIPS means women now need to make decisions around screening earlier in pregnancy. This means GPs are taking on new responsibilities for providing information to women who need to make decisions about screening, and also providing feedback and support after the results are provided. As a result, it has become even more critical to ensure that GPs can provide accurate information and access to relevant counselling and support.

www.downsyndrome.org.au
Evidence

Informed Choice

Women have a right to make an informed decision as to whether they want to undertake any test during pregnancy. There are concerns that NIPS may be seen as a routine blood test that pregnant women are encouraged to have as part of 'typical' prenatal care. Women need to be able to give informed consent for all tests and understand that receiving a diagnosis of Down syndrome will mean they will be faced with further decisions about whether or not to continue their pregnancy. Many families report that they are not provided with sufficient information about the conditions that are being tested for. For example in a 2017 Down Syndrome Australia survey, nearly three out of four women reported not receiving any information about Down syndrome prior to screening.

Issues around the information provided and the process of informed consent have also been identified in the more traditional screening approaches. For example, an Australian study found that only 37% of women were fully informed regarding a prenatal blood serum screen and only 62% were aware that termination of pregnancy would be offered if Down syndrome were to be diagnosed. Similarly, a 2009 study found that approximately half of women surveyed, who underwent both ultrasound and biochemical screening, did not foresee that they might ultimately need to make the decision about whether or not to continue the pregnancy. Research suggests that the implementation of NIPS may further exacerbate the issue around access to informed consent as screening becomes more routine and available earlier in the pregnancy.

Information provided post-screening

Down Syndrome Australia is concerned about the information being provided to women after they receive a result which indicates a high chance of having a child with Down syndrome. A 2014 survey of women who had a child with Down syndrome found that nearly half (48%) believed that health professionals were giving biased or incorrect information about Down syndrome at the time of diagnosis. Approximately a quarter of women surveyed believed that this biased information was leading to more terminations of pregnancies of foetuses with Down syndrome than would occur if more balanced information was provided.

These findings are consistent with the results of a survey published by Down Syndrome Tasmania of 58 Australian women who have a child with Down syndrome and received a high chance result during

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pregnancy. Amongst these women, 62% indicated that negative language was used by health professionals when informing them of the high probability result. One woman described her experience: ‘They weren’t just negative, they told us our baby would most probably die in utero but if she survived she would be in hospital for most of her “short” life, never walk, talk or have any quality of life’.

Results from a 2017 Down Syndrome Australia survey, found that nearly half (47%) of the women who responded to the survey (who had a child born with Down syndrome in the last five years) felt that they were not given balanced information about Down syndrome by health professionals after receiving results. The parents responding to the survey also shared stories about the negativity of doctors and a focus on termination.

‘One medical professional listed every possible problem that could go wrong with our baby. Without including any of the lovely benefits of having a new baby with Down syndrome.’

-Parent response to DSA Survey 2017

“We were given the results then the first question was ‘Would you like to terminate?’ There was no information provided on what it would mean to raise a child with Down syndrome. No one offered to put us in touch with another family. There was a presumption we would terminate.

Doctors need more updated information and better skills to handle these matters’

-Parent response to DSA Survey 2017

Current Guidelines
There are a number of Australian health guidelines for prenatal care that clearly state the need for prescreening information and counselling, including up-to-date information about Down syndrome.¹⁰

For example the Royal Australian and New Zealand College of Obstetrics and Gynaecologists recommends that the ‘information provided should include:

- The difference between screening and diagnostic testing.
- The relative advantages and disadvantages of the available screening tests.
- Details of the nature, purpose, limitations and consequences of screening.
- That the decision whether to undertake screening or not is entirely that of the woman.
- Practical aspects of screening including the conditions that are being screened for, the type of tests, the timing of tests and the approximate costs involved.

• The possibility of diagnosing foetal genetic or structural conditions other than those for which the screening programs are designed.

• The nature of the results (often expressed as a numerical probability estimate) and the offer of a follow-up diagnostic test if an “increased probability” result is obtained.

• That continuing or not continuing the pregnancy are both options in the event that a foetal genetic or structural condition is diagnosed.

• An assurance that continuation of the pregnancy is a valid option should a foetal genetic or structural condition be diagnosed, and that couples will receive appropriate counselling and care in preparation for birth.11

Similarly, antenatal guidelines, currently under revision by the Department of Health, provide good guidance around the information that should be provided to parents about the screening tests, and also appropriate provision of results and access to support and counselling.

DSA Position
Down Syndrome Australia understands that access to prenatal screening and diagnosis is important for many families. We respect the right of women to have prenatal testing if they choose to do so and to make decisions about whether or not to continue a pregnancy based on their own circumstances and beliefs.

It is absolutely essential that any genetic screening program in Australia is done ethically and that families have access to balanced and timely information. The evidence suggests that despite the existence of guidelines, in practice many families are not getting the information and support they need to make informed decisions.

The World Health Organisation has published an updated synthesis of screening criteria for genetic screening12 as follows:

• The screening programme should respond to a recognised need.

• The objectives of screening should be defined at the outset.

• There should be a defined target population.

• There should be scientific evidence of screening programme effectiveness.

• The programme should integrate education, testing, clinical services and programme management.

• There should be quality assurance, with mechanisms to minimize potential risks of screening.


The programme should ensure informed choice, confidentiality and respect for autonomy.

The programme should promote equity and access to screening for the entire target population.

Programme evaluation should be planned from the outset.

The overall benefits of screening should outweigh the harm.

The current approach to prenatal testing in Australia does not meet the requirement to integrate education and clinical services into the screening program.

We are also concerned about a lack of sufficient safeguards to respect autonomy and facilitate informed decision-making. Negative community attitudes and outdated information about intellectual disability means that many families and health professionals are unaware of what raising a child with Down syndrome is actually like.

Without significant changes to the proposed approach to screening, there are serious concerns about the ethics of a government funded screening program.

Down Syndrome Australia recommends that the government take steps to ensure that women are able to make informed choices about prenatal tests. This includes access to balanced information, support and opportunities to connect with other families. The guidelines which have been developed are a start, but there needs to be a coordinated approach to implementation. This should include:

1. Support for a national training program and resources for relevant health professionals (including GP’s, midwives, Obstetricians etc.) to ensure that they are skilled in facilitating informed consent by provision of appropriate information and counselling before and after testing. This program will facilitate successful implementation of the revised antenatal guidelines.

2. Following a diagnosis of Down syndrome, all families should have access to counselling by appropriate health professionals (e.g. genetic counsellors). As part of this counselling, families should be offered support to connect to other families through Down syndrome organisations.

3. The government to work closely with representative organisations and people with Down syndrome to develop a public awareness campaign to tackle the stigma associated with intellectual disability and raise public awareness around Down syndrome and other chromosomal disorders.

4. Regular monitoring of the experience of women having prenatal testing and refinement of the approach to prenatal screening and diagnosis based on this evidence.