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**Submission to Bioethics and Disability
for the UN Human Rights Council**

September 2019



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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. 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.

Down syndrome is a genetic condition in which the person has an extra copy of some or all of chromosome 21. 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 http://www.downsyndrome.org.au/down_syndrome_population_statistics.html

Submission to Bioethics and Disability for the UN Human Rights Council

Down Syndrome Australia (DSA) welcomes the opportunity to provide a submission to **UN Special Rapporteur on the rights of persons with disabilities** regarding bioethics and disability. Our response focuses on issues relating to Down syndrome and prenatal screening. We are particularly concerned about the impact of prenatal screening and the lack of balanced and appropriate information provided to families within Australia.

Questionnaire on bioethics and disability

- I. **Please provide information on the legislative and policy framework in place in your country in relation to:**
 - a. **Prenatal diagnosis**
 - b. Disability-related abortion
 - c. Informed consent to medical treatment and scientific research
 - d. Protection of persons with disabilities undergoing research
 - e. Euthanasia and assisted suicide

RESPONSE

At present, Australia does not have any legislative frameworks in place regarding population genetic screening. Medical genetic tests are generally ordered by medical practitioners and, in some instances, may involve referral to a clinical geneticist as well as a genetic counsellor for pre-test and post-test counselling². Often genetic counselling services are only provided once a definitive diagnosis has been confirmed.

Current practice in Australia is to test for chromosomal conditions in the first trimester. An emerging practice is the use of non-invasive prenatal screening (NIPS). This test screens for various chromosomal conditions prenatally, including Down syndrome, through an examination of maternal blood. NIPS can be performed as early as 10 weeks gestation and is more specific than previous screening tests (has fewer false positives), but it does not provide a definitive result. In Australia, women can access the test privately (cost is around \$500).

² Australian Law Reform Commission, *Genes and Ingenuity: Gene patenting and human health* (ALRC Report 99)

The only definitive way to determine a 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).

There are several guidelines currently in place regarding prenatal care and the provision of information, referrals and counselling to patients undergoing prenatal testing. These include the *Clinical Practice Guidelines: Pregnancy Care* by the Department of Health³, the *Guidelines for Preventative Activities in General Practice* developed by the Royal Australian College of General Practitioners⁴, the *Prenatal Screening and Diagnostic Testing for Fetal Chromosomal and Genetic Conditions* produced jointly by the Royal Australian and New Zealand College of Obstetricians and Gynaecologists and the Human Genetics Society of Australasia⁵, and *Medical Genetic Testing: Information for health professionals* by the National Health and Medical Research Council⁶. These guidelines clearly outline the importance of the provision of comprehensive and unbiased information both pre- and post-screening so patients can make informed choices. These guidelines, however, are often not implemented in everyday practice.

It should be noted that access to balanced information on prenatal screening and associated conditions was an area of concern even before the availability of NIPS. However, NIPS means women often are asked to make decisions around screening and how to respond to unexpected results earlier in pregnancy. As a result, GPs are taking on new responsibilities for providing information to women who need to make decisions about screening, as well as providing feedback and support after the results are given. It is therefore critical to ensure that GPs provide accurate information and access to relevant counselling and support.

The Medical Services Advisory Committee (MSAC) is currently considering an application for NIPS to be paid for publicly under certain circumstances. They are considering a few different models of funding NIPS including as a primary screen or a contingent screen (available only for women at increased likelihood of having a chromosomal condition). MSAC will provide advice to the Minister of Health regarding whether NIPS should be publicly funded and under what circumstances. It is likely a decision on this matter will be made in 2020.

Down Syndrome Australia has been advocating that it is essential that any genetic screening program in Australia is implemented ethically and that families have access to balanced, unbiased, and timely information to assist them in making informed decisions. Currently this is clearly not the case. Some families who receive a diagnosis of Down syndrome have informed us that doctors questioned their choice to continue their pregnancy or have told them that their child would have a lifetime of suffering⁷. This raises serious ethical issues regarding autonomy and beneficence.

³ Department of Health, *Clinical Practice Guidelines Pregnancy Care*, 2019, Australian Government, Department of Health, Canberra

⁴ Royal Australian College of General Practitioners, *Guidelines for Preventive Activities in General Practice*, 2016, RACGP, Melbourne.

⁵ Royal Australian and New Zealand College of Obstetricians and Gynaecologists, 2015, RANZCOG, Australia

⁶ National Health and Medical Research Council, *Medical Genetic Testing: Information for health professionals* (2010), NHMRC, Canberra.

⁷ <https://www.mamamia.com.au/terminate-down-syndrome-pregnancy/>



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Down Syndrome Australia (DSA) believes there is a need for a funded approach to ensuring that prenatal guidelines are implemented appropriately, and families have access to suitable information and support both pre- and post-screening. International models may assist in the development of a prenatal screening program in Australia⁸. The UK Down Syndrome Society's *Tell it Right, Start it Right* Program has shown significant benefits in supporting families who receive an unexpected result regarding chromosomal conditions.⁹ This and similar international programs merit further consideration in the Australian context.

⁸ Royal Australasian College of Physicians, Newborn Screening in Australia: Position Statement, 2015, RACP, Sydney.

⁹ Bryant, LD, Puri C, Dix L, Ahmed, S Tell it Right, Start it Right: An evaluation of training for health professionals about Down syndrome

British Journal of Midwifery 2016 24:2, 110-117

Please provide any information and statistical data (including surveys, censuses, administrative data, literature, reports, and studies) in relation to:

- a. The availability, accessibility and use of prenatal diagnosis**
- b. The availability, accessibility and use of disability-related abortion**
- c. The practice of informed consent to medical treatment and scientific research
- d. The existence of measurements of quality of life which affect both clinical decision-making and health policy
- e. The practice of experimental, controversial and/or irreversible treatments
- f. The practice of euthanasia and assisted suicide on persons with disabilities

RESPONSE

The Department of Health (Australia) Prenatal-Guidelines recommends “In the first trimester, give all women/couples information about the purpose and implications of testing for probability of chromosomal anomalies to enable them to make informed choices” The majority of pregnant women undertake the combined first-trimester screening which includes screening for chromosomal conditions such as Down Syndrome. A smaller number opt to undertake the Non-Invasive Prenatal Screening (NIPS) which is available at 10 weeks of pregnancy.

There is considerable evidence, despite appropriate guidelines, that appropriate information about screening is often not provided to women and acceptance of screening is frequently presumed. Often families are not made aware of the decisions they will need to consider if they receive an unexpected result.

DSA has concerns that if publicly funded, NIPS could be treated as a routine blood test like one of the many that pregnant women are asked to have as part of ‘typical’ prenatal care¹⁰. If women are not given a clear choice about whether or not to undergo NIPS, they may not have an opportunity to consider whether they want the test results or the decisions they would have to make after receiving a high chance

¹⁰ Deans Z, Newson AJ. Should non-invasiveness change informed consent procedures for prenatal diagnosis? Health Care Anal. 2011;19(2):122–132.

result¹¹. Issues around the information provided and the process of informed consent have also been identified in the more traditional screening approaches¹².

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 diagnosed¹³. 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 be confronted with the need to make the decision about whether or not to continue the pregnancy¹⁴. 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 pregnancy¹⁵.

Data from Western Australia suggests that in Australia, most women for whom a prenatal diagnosis of fetal Down syndrome is made, choose to terminate the pregnancy (93%). The availability of prenatal tests for fetal aneuploidy over the last 30 years has significantly reduced the live-birth prevalence of Down syndrome in Western Australia, and these results are likely to be similar in other areas of Australia.¹⁶

There is significant concern that these termination rates are impacted by the lack of balanced information provided to families during prenatal screening. The lack of support from some medical professionals about continuing a pregnancy after a prenatal diagnosis also may influence parents' decisions. In 2017 Down Syndrome Australia conducted a survey and found that in our sample, more than a third of women who had a pregnancy with a prenatal diagnosis of Down syndrome felt pressure from medical professionals to terminate the pregnancy. For example, one respondent stated:

"We were very pressured to have a termination. They said our little boy was going to pass away due to his health issues, that he would end us going through so many surgeries and he would suffer, we were better off terminating before to save him the suffering. In the end we have a happy little boy because we refused to listen to this advice. I hate to think of all the parents who did listen to their doctors and missed the chance to meet their child"

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

¹¹ van Schendel RV, Kleinveld JH, Dondorp WJ, et al. Attitudes of pregnant women and male partners towards non-invasive prenatal testing and widening the scope of prenatal screening. *Eur J Hum Genet.* 2014 Mar 19;

¹² Constantine M, Allyse M, Rockwood T, Wall M, De Vries R. Imperfect informed consent for prenatal screening: Lessons from the quad screen. *Clinical Ethics.* 2014;9(1):17–27.

¹³ Rowe, H. J., Fisher, J. R. W. and Quinlivan, J. A. (2006), Are pregnant Australian women well informed about prenatal genetic screening? A systematic investigation using the Multidimensional Measure of Informed Choice. *Australian and New Zealand Journal of Obstetrics and Gynaecology*, 46: 433–439. doi:10.1111/j.1479-828X.2006.00630.x

¹⁴ Seror V, Ville Y. Prenatal screening for Down syndrome: women's involvement in decision-making and their attitudes to screening. *Prenat Diagn.* 2009;29(2):120-128.

¹⁵ *J. Clin. Med.* 2014, 3(2), 614-631; doi:10.3390/jcm3020614

¹⁶ 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. *Prenatal Diagnosis*, 35(13), 1324-1330. <https://doi.org/10.1002/pd.4698>

diagnosis. Approximately a quarter of women surveyed believed that this biased information was leading to more terminations of pregnancies of fetuses with Down syndrome¹⁷.

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 pregnancy. Amongst these women 62% indicated that negative language was used by health professionals when informing the women of the high probability result¹⁸.

Some anecdotal reports have indicated that women are being told that a child with Down syndrome can never go to a mainstream school and that parents will have to give up their careers to support the child. A post to the parenting blog Mamamia.com.au told of a woman who decided to terminate her pregnancy to avoid pain for her child¹⁹: “*We ended little Dot’s life because we didn’t want her to endure a lifetime of pain. We knew that she would need constant medical attention, and we felt we couldn’t stand by and watch her as she struggled physically and intellectually throughout her (possibly short) life, when we couldn’t do anything to help her.*” This description of life with Down syndrome is at odds with research findings and the experience of most families.

There are also concerns that in some cases families are seeking terminations without the appropriate follow up tests after a positive result on NIPS. The marketing of NIPS uses terminology such as “*unsurpassed accuracy*”²⁰ and “*Simple, fast, reliable*”²¹, and statements such as “non-invasive **prenatal** test offering **99%** accuracy”²² which can result in families having an view that NIPS provides a definitive result and that there is no need for additional testing..

While NIPS may have a high level of sensitivity and specificity a key issue to consider is the Positive Predictive Value (PPV) of the test. This refers to how likely it is that individuals who test positive are to be affected by the condition. PPV depends not only on test performance but also on the prevalence of the condition in the population studied. For example, if a women has a pre-test likelihood of Down syndrome of 1 in 2000, then the likelihood that a positive result from NIPS is a true positive is only around 55%. Information about PVV is often not shared with families, and inaccurate presumptions are made that a high chance result is likely to be a true result 99% of the time.

The UN Committee on Bioethics notes “... *there is the risk that pregnant women with a positive result don’t await the validation of the result through invasive diagnostics, but immediately choose to abort the embryo or foetus, without adequate counselling about the relevance of the detected abnormality*”²³.

¹⁷ Kellogg, G., Slattery, L., Hudgins, L. et al. J Genet Counsel (2014) 23: 805. doi:10.1007/s10897-014-9694-7

¹⁸ Kelly, R. (2016). Carrying a baby with Down syndrome: Women’s experiences of prenatal screening, diagnosis and pregnancy care. (published by Down Syndrome Tasmania).

¹⁹ <http://www.mamamia.com.au/terminate-down-syndrome-pregnancy/> (accessed 9/1/2017)

²⁰ https://www.sonicgenetics.com.au/nip/patients/harmony-prenatal-test/?gclid=EAIaIQobChMIxePX0I735AIV1RmPCh3IUwswEAAAYASAAEgIuIvD_BwE

²¹ <https://www.vcgs.org.au/tests/perceptnpt>

²² <https://www.ivf.com.au/fertility-treatment/genetic-testing-pgd/prenatal-testing>

²³ <http://unesdoc.unesco.org/images/0023/002332/233258E.pdf>

CONCLUDING REMARKS

Women in Australia have the right to make decisions about whether to continue their pregnancy. However, the way in which prenatal screening is being implemented in Australia means that women are not given the opportunity to make informed choices. Instead they are being provided with inaccurate information about Down syndrome and are not being given the opportunity to connect with other families or relevant organisations such as Down Syndrome Australia. In some cases, they are feeling pressured by medical professionals to terminate their pregnancy. The underlying social bias and discrimination of people with intellectual disability intersects with this lack of access to balanced information. Termination rates of pregnancies with Down syndrome in Australia are very high, and the number of births of people with Down syndrome has decreased over the last 30 years as a result. There is an urgent need to address the current information gap and to ensure that families are getting the full story about Down syndrome before deciding about whether or not to continue their pregnancy.