

Carrying a baby with Down syndrome: Women's experiences of prenatal screening, diagnosis and pregnancy care

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Key findings and recommendations

Over the last few years our capacity to test for genetic conditions such as Down syndrome before birth has exploded. We now live in a world where a simple blood test can be used to screen pregnancies for conditions such as Down syndrome, Trisomy 18 and even chromosome microdeletions. At the same time, there has never been a better time or place in history for a child with Down syndrome to be born than here and now in Australia. Medical advances, early intervention and the move away from segregated and institutionalised lives to full participation in the community mean that people with Down syndrome are living longer and more independent lives than ever. Overwhelmingly the lived experiences of people with Down syndrome and their families indicate positive outcomes for people born with the condition. For example, recent research shows:

- 99% of parents/guardians said they loved their child with Down syndrome; 79% said their outlook on life was more positive because of their child.
- 94% of siblings expressed feelings of pride in their sibling with Down syndrome; 88% said they felt they were better people because of their sibling.
- 99% of people with Down syndrome said they were happy with their lives; 97% liked who they are; 96% liked how they look and 86% indicated they could make friends easily.¹

Given these positive life experiences it is somewhat surprising the negative attitudes towards Down syndrome that many women are exposed to during pregnancy. This report summarises responses to a survey of 58 women who received their pregnancy care in Australia where their baby was either diagnosed or identified as highly likely to have Down syndrome during the pregnancy.

This survey shows significant failings in the way that prenatal screening programs are being run and the way results from screening and diagnostic tests are being delivered. While some women provide accounts of positive experiences where they felt their decisions were respected and that they were supported by their medical teams, all too often women provide accounts characterised by a lack of respect for their choices, provision of little to no information or support and out-dated, ableist and very negative perceptions of Down syndrome. From the survey it is clear:

- Many women do not realise that screening tests are voluntary or what the nature and purpose of these tests are. Very few women were provided with any relevant up to date information on the conditions, such as Down syndrome, being tested for before they undertook screening.
- The language used to discuss screening and diagnostic test results is often very negative, sending clear messages to women that having a baby with Down syndrome is a negative outcome.
- Many women are placed under immense pressure to undertake invasive diagnostic testing and to terminate their pregnancies. Women are often asked frequently to revisit decisions relating to testing and termination even after they've clearly indicated they're not interested.
- Women are being offered terminations on the basis of screening results only without any confirmed diagnosis of Down syndrome.
- Few women are provided with any up-to-date information to inform their decisions around invasive diagnostic testing and termination. Information given is often sparse, extremely negative and at odds with the lived experience of people with Down syndrome. Frequently

¹ <http://www.childrenshospital.org/news-and-events/2011/september-2011/parents-siblings-and-people-with-down-syndrome-report-positive-experiences>

the only information given is a very biased and outdated view of the limited value and potential of a life lived with Down syndrome.

- In some cases the negative attitudes of medical staff towards babies born with Down syndrome appear to have affected the quality of care provided to the extent that the baby and woman were put at risk.
- There appears to be discomfort amongst many medical professionals in providing results, with some doctors unsure how to deliver the news and to lead a balanced discussion of the options available.

In order to address these issues the following recommendations are made.

A Framework for Prenatal Clinical Care

Currently there is no standardized approach to the implementation of Australia's prenatal screening program. A Framework for Prenatal Clinical Care which provides for an informed consent process to be applied through the prenatal screening and testing process should be developed. This needs to involve:

- Provision of up-to-date resources to women before they undertake screening to inform their decision to screen. This should be supported by a Charter of Prenatal Care Rights (discussed below) which will make clear early on in the screening process women's rights to an informed choice and expectations they should see met through their prenatal care process.
- A 'sign to screen' approach so women explicitly sign to give informed consent to undertake screening, ensuring they have been provided with the appropriate resources and are aware of their rights.
- A clear pathway for practitioners to follow that guides them through the screening and testing process, indicating where and when women should be referred to for support to help inform and support their decisions.
- Clear standards around the use of neutral language in discussing screening and results as well as options around continuing or terminating their pregnancy. The option of adoption or long term foster placement should also be discussed with women.
- Guidelines on follow up testing and provision of support for women who decide to continue their pregnancies. This could include referring women to State based support organisations or counselling services as well as referral to paediatricians or other medical specialists who may be involved in the care of their child post birth.
- Guidelines for the provision of appropriate emotional support for women who choose to terminate their pregnancies as well as for those considering adoption or long term foster placement.
- Development of simple, easy to understand resources describing the conditions being tested for, life outcomes for people with the condition, links to good quality online resources with further information and contacts for relevant support organisations which are provided to women before screening as well as after a high probability result is found.

Adequate training

Training should be provided to all medical professionals involved in the care of pregnant women on the delivery of challenging results (such as a Down syndrome diagnosis) as opposed to delivering 'bad news'. This training should provide examples of the language that should be used, information about the conditions being tested for, appropriate resources and support and experience with children and adults with Down syndrome and their families. Ideally a training module should be developed that includes face to face contact with a parent of a child with Down syndrome or an

adult with Down syndrome who can talk with knowledge about the condition and its implications for the life of the person and family. Medical staff should be exposed to concepts of ableism and inclusion and encouraged to question their own beliefs about people with Down syndrome and other disabilities. Training should include a mix of parent/child talks or talks by adults with Down syndrome to medical staff and students and use of internet based resources, such as Karen Gaffney's TedX talk 'All lives matter' <https://www.youtube.com/watch?v=HwxjoBQdn0s>. Training resources should be developed in conjunction with the peak bodies representing relevant chromosomal and structural conditions being tested for (eg. Down Syndrome Australia, CleftPALS, HeartKids, Cystic Fibrosis Federation Australia). We suggest these could be modelled on resources already developed by the National Down Syndrome Congress² and a webinar already developed by Down Syndrome Australia for the Australian College of Midwives in 2015.

Standards and complaint processes

Women's experience of prenatal screening, diagnosis and care following identification of Down syndrome are variable, but results from this survey indicate there is a strong need for standards to be developed and adhered to around prenatal screening, diagnosis and pregnancy care. These standards need to cover aspects of informed consent, the use of unbiased language in delivery of results, quality information provision, emotional and social support for women whose babies are identified as likely to have Down syndrome or other chromosomal (or structural) conditions and demonstration of respect for women's decisions around screening, diagnostic testing and continuation of their pregnancy. A clear process for making complaints and having these addressed needs to be developed. Women need to be given information on their rights (discussed further below) and the complaints process.

Provision of resources and information to professional and patients

Medical professionals should be given easy access to a range of up to date resources (see the Suggested Resources at the end of this report for examples) on Down syndrome and the delivery of a pre or postnatal diagnosis. Women should be provided with access to these resources to inform their decisions to undertake screening, diagnostic tests and around the continuation of their pregnancy where Down syndrome or other conditions are diagnosed. Women should be presented with the option to continue their pregnancy and either keep and raise their baby, place it for adoption or long term foster care or terminate. Information about all of these options should be offered and women should be supported regardless of which of these options they choose.

Support

Women should be offered social and emotional support through their pregnancy regardless of whether they have a confirmed diagnosis (as opposed to a high likelihood of Down syndrome), whether they choose to continue their pregnancy or terminate. This support should include referral to professional counselling, referral to State based Support Associations and access to parents of children with Down syndrome or other identified conditions.

Respect for decisions

Women's decisions with respect to prenatal screening and continuation of their pregnancies should be respected. At each stage options should be provided in neutral language with the risks and uncertainties associated with tests clearly explained. It should never be assumed that the choice to screen implies the choice to undertake invasive diagnostic testing, nor should undertaking these tests be seen to imply a choice to terminate a pregnancy if results come back positive for Down

² <https://www.ndsccenter.org/physiciansguide/>

syndrome or other chromosomal conditions. Once women have declined invasive tests or termination these should not be offered or mentioned again unless and until the woman indicates she is reconsidering her previous decision and wishes to reopen the discussion. Even then, no presumption should be made about the choice she should or will make. Termination should never be presented as the logical or normal choice following a prenatal diagnosis. Women should be free to make their own decisions without bias from their medical team. All discussions should be balanced and not use negative language or perpetuate outdated perceptions of Down syndrome or other disabilities.

Informed choice – A Charter of Rights

All pregnant women should be provided with a statement of their rights early on in their pregnancy. These rights should include:

- You have the right to choose whether to undertake prenatal screening for chromosomal and structural conditions. You must be provided with access to up to date resources on the conditions being tested for and any risks associated with tests. You can expect your care giver to have a nondirective conversation about the negative and positive aspects of undertaking tests and your decisions in light of your own belief and value systems.
- You have the right to choose whether or not to undertake diagnostic testing. No assumptions should be made that choosing to screen implies you wish to undertake invasive testing.
- You have the right to make informed choices about continuation of your pregnancy. No assumptions should be made about what your choice would be based on your decision to undertake testing. You should not be placed under time pressure to make a decision and no arrangements for termination should be made unless you direct your caregiver to make them.
- Your decisions will be respected. In all cases once you've made a decision, that decision will be considered final until and unless you tell your healthcare provider you've changed your mind.
- Your screening and diagnostic test results should be relayed using neutral, unbiased language. You have the right to expect that the personal opinions of your care givers should not affect the way in which they deliver their results. You have the right to choose how results are delivered to you (eg. over the phone, in person) and who is present (eg. partner or friend).
- Before making decisions to undertake screening tests, diagnostic testing or around the continuation of your pregnancy, you should be:
 - Provided with up to date and accurate information on the tests being undertaken, what the results mean and the level of uncertainty (including the likelihood of false positive and false negative results), the conditions being tested for and likely implications for your pregnancy and the life of your child.
 - Offered a referral to both a genetic counsellor to discuss the implications of your results as well as a counselling service that can help you work through your feelings about your test results and help you make an informed choice about the next steps. Whether you take up this offer is entirely up to you. You can choose to take it up at any point in your pregnancy or after the birth of your child.
 - Offered contact information for relevant support groups. You may opt for your care giver to contact these groups on your behalf. You have the right to request to meet other parents and children with the condition before making any decisions.

- You have the right to high quality pregnancy care regardless of any decisions you make with regard to screening, diagnostic testing and continuation of your pregnancy.
- If these rights are not respected you can make a complaint to [insert appropriate complaint contact].

Introduction

This report summarises the experiences of 58 women who gave birth to babies with Down syndrome (DS) where DS was identified as either likely or as a confirmed diagnosis during their pregnancy. This survey aimed to understand and document the experiences of women with prenatal screening and diagnosis to underpin best practice guidelines being developed for pregnancies affected by Down syndrome and other chromosomal and genetic conditions. Women were recruited from Australian Facebook support groups for parents of children with Down syndrome. These groups are closed and members are vetted. Responses were sought from women where Down syndrome was identified during pregnancy, either as highly probable based on screening tests or diagnosed using invasive tests. The survey was conducted as an online survey using Survey Monkey. Survey questions were developed using the assistance of women who run online pregnancy support groups for women who have Down syndrome identified as highly probable or diagnosed during pregnancy. The survey questions are provided in Appendix 1.

Characteristics of the pregnancies covered by the survey are shown in Table 1. This shows that two thirds of survey responses are for pregnancies that resulted in births in the last 5 ½ years. Nearly half of the women had their pregnancy care in Victoria with a further 21% were cared for in NSW. At least one response was obtained from each State and Territory in Australia. The majority of pregnancies were managed in the public health system, with 3 women switching from public to private systems after their baby was diagnosed with Down syndrome. Half of the women received their pregnancy care from an obstetrician with 3 women shifting from other modes of care to obstetric once their babies were diagnosed.

TABLE 1. CHARACTERISTICS OF WOMEN AND THEIR PREGNANCIES IN THE SURVEY COHORT

Characteristic	Proportion of women in survey
Year baby was born	Before 2000 - 7% 2000 to 2005 - 7% 2006 to 2010 - 21% 2011 - 7% 2012 - 9% 2013 - 16% 2014 - 9% 2015 - 17% 2016 - 9%
State in which pregnancy care was given	Australian Capital Territory - 5% Northern Territory - 2% NSW - 21% Queensland - 16% South Australia - 2% Tasmania - 7% Victoria - 47% Western Australia - 2%
Health system in which pregnancy was managed	Public - 72% Private - 22% Public shift to private on diagnosis - 5%
Mode of pregnancy care	GP - 5% Obstetrician - 47% Shared care - 24% Team midwives - 14% Shifted from shared care/midwives to obstetricians post diagnosis - 5% Specialise clinic - 3% Other (unspecified) - 2%

Women undertook a range of screening tests with 10% of women opting out of screening (Table 2). Nearly 80% of women undertook routine first trimester screening, with a quarter of women having the morphology scan and 9% undertaking newer Non-invasive pregnancy screen (NIPS). Note that many women undertook more than one type of screening test.

TABLE 2. PROPORTION OF WOMEN WHO UNDERTOOK DIFFERENT MODES OF SCREENING

Screening tests undertaken	Proportion of women
I opted out of screening	10%
Morphology scan	24%
NIPS	9%
Routine first trimester screen (ultrasound and blood test)	78%

As shown in Table 3, nearly 50% of women undertook some form of diagnostic testing, with amniocentesis being the most common technique.

TABLE 3. PROPORTION OF WOMEN WHO UNDERTOOK DIFFERENT MODES OF DIAGNOSTIC TESTING

Diagnostic test	Proportion
Amnio	37%
CVS	9%
Unspecified	2%
None	53%

For those who had a confirmed diagnosis of Down syndrome, the timing of this diagnosis varied from before 12 weeks right up to term. Efforts have been made to retain the voices of the women who completed the survey through extensive use of quotes, in order for their experiences to be accurately recounted. Where a range of experiences were given, all detailed comments provided by women have been included. Summary data is also provided to give an estimate of the prevalence of different experiences. The survey and report focus on informed consent for testing and termination, experiences and attitudes around continuation of a pregnancy affected by Down syndrome and quality of care. Women were also asked for their opinions of what best practice care should look like.

The next section focuses on the concept of informed consent, both for undertaking screening and diagnostic testing as well as for decisions around termination and continuation of pregnancies.

Informed choice and support

There are three key points in time that women are asked to make decisions relating to chromosomal conditions:

- Before undertaking any screening tests – in deciding whether or not to have screening;
- Before undertaking diagnostic testing after receiving a high likelihood test result from screening – in deciding whether to have an invasive procedure to confirm a diagnosis;
- After a confirmed diagnosis – when asked to decide whether they intend to continue their pregnancy after a diagnosis or high probability result.

Clearly in order for women to make an informed choice at any of these points in time, women need to receive up to date information not only about the procedures and any risks involved but also about the conditions, such as Down syndrome, being tested for. Figure 1 shows the proportion of women who said they received up to date information about Down syndrome at each of these decision points in their pregnancy.

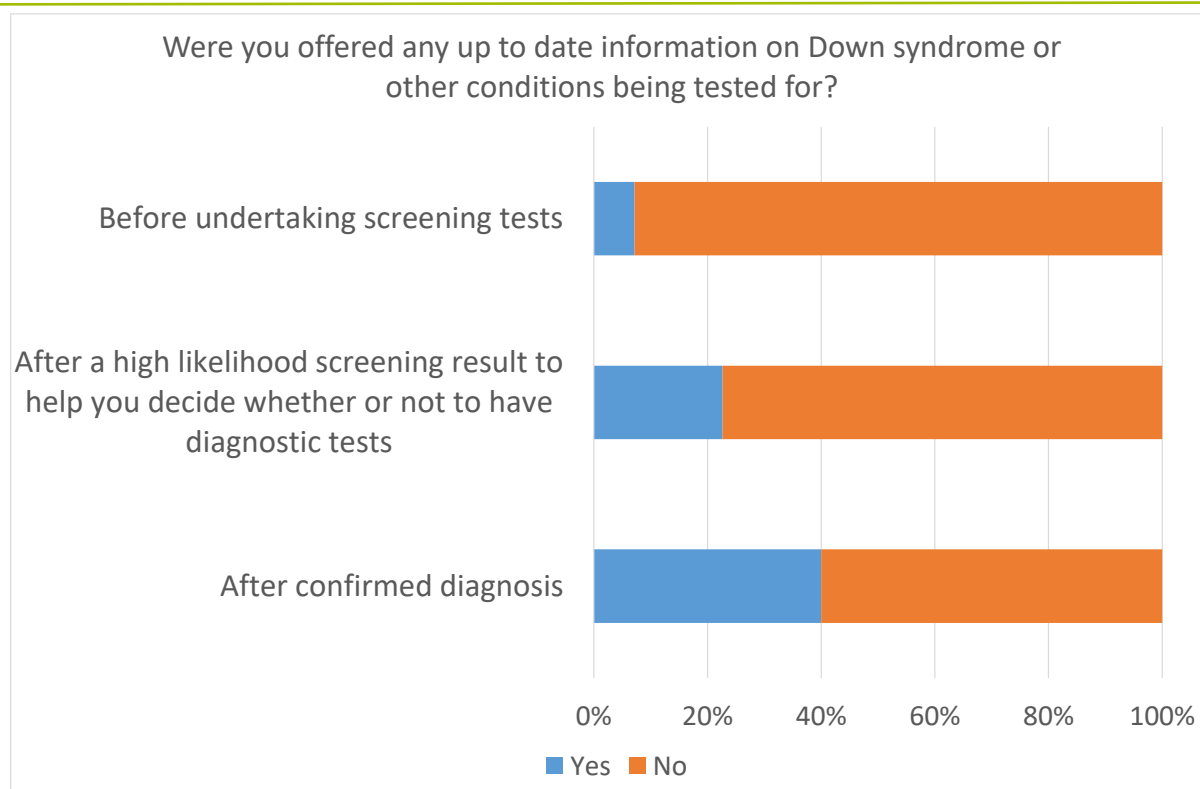


FIGURE 1. PROPORTION OF WOMEN PROVIDED WITH UP TO DATE INFORMATION ON DOWN SYNDROME (OF RELEVANT RESPONSES)

This figure shows that only 7% of women were offered any up-to-date information on Down syndrome before undertaking screening tests. Less than a quarter of women were provided with any information to inform their decision to undertake invasive diagnostic testing and only 40% of women were provided information after a confirmed diagnosis.

Women were also asked about whether they were offered any form of counselling and whether they were linked up with any parent support groups (see Figure 2).

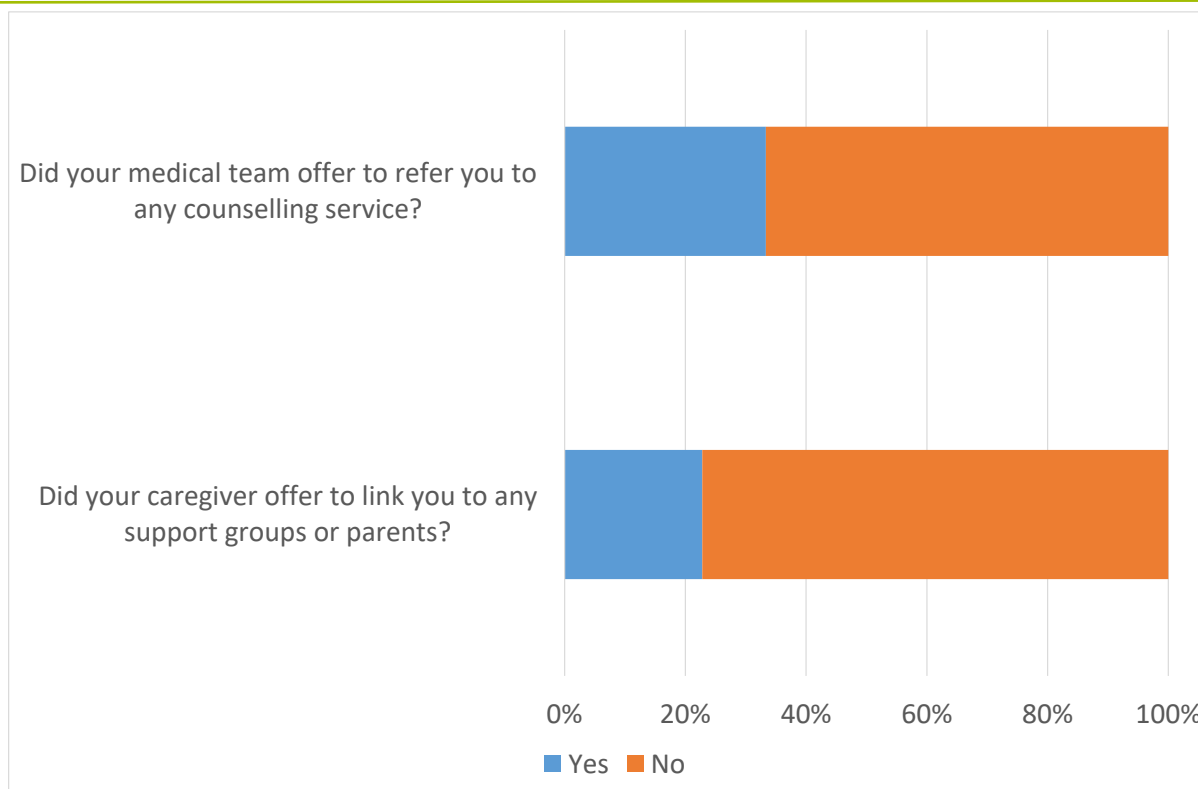


FIGURE 2. PROPORTION OF WOMEN LINKED WITH SUPPORT SERVICES AND GROUPS

This figure shows that 22% of women had their caregiver offer to link them up to support groups or other parents, with 33% offered a referral to a counselling service. Almost all counselling referrals were for genetic counsellors, as summed up by one woman *‘[It was] a referral to see a genetic counsellor who talked us through what Down Syndrome would mean for our son. She explained the financial assistance and support we could access to support us with our baby.’* – #8 Baby born 2015. Little support appears to be offered to help women deal with the emotional aspects of a diagnosis or screening results that indicate a high likelihood of Down syndrome. Another woman said she was told counselling would be offered *‘only after a termination (if I decided to go through with one) to assist me with grief of terminating a pregnancy.’* – #7 Baby born 2014. Another woman said her local hospital had *‘no idea where to start or who to contact at all.’* – #45 Baby born 2015.

Experiences of Medical Attitudes to Continuation of Pregnancies Affected by Down Syndrome

Figure 3 shows the proportion of women who were offered terminations at various stages of their pregnancy: after returning a screening result indicating a high probability of their child having Down syndrome but before undertaking any diagnostic testing to confirm a diagnosis; after a confirmed diagnosis of Down syndrome; and never. Note that in some cases women were offered terminations both before and after diagnostic testing. Interestingly over 40% of women were offered a termination on the basis of screening results only, without any confirmed diagnosis. 56% of women were offered a termination after receiving a confirmed diagnosis, while one third of women were never offered a termination. Results in the previous section (Figure 1) indicated that only 7% of women were offered up to date information before choosing to undertake screening tests, 23% to inform their decision to undertake invasive testing and 40% after a confirmed diagnosis. The results in Figure 3 shows that women are frequently being offered terminations based on screening test results alone, so for the majority of women being asked to decide whether or not they wish to

continue or terminate their pregnancies, they are making a decision without access to any up to date information on Down syndrome.

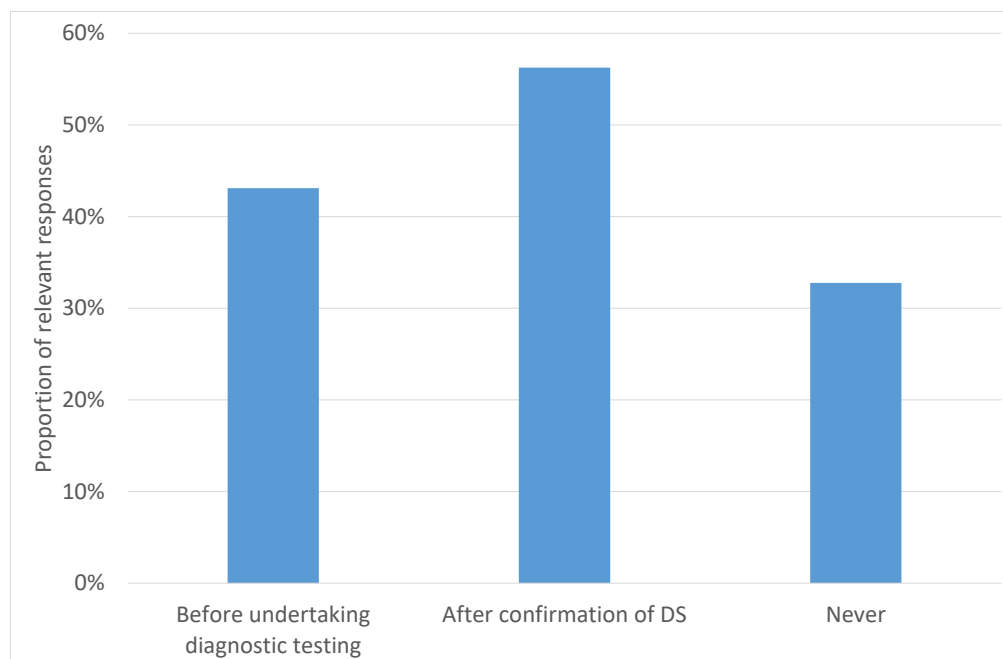


FIGURE 3. PROPORTION OF WOMEN (OF RELEVANT RESPONSES) WHO WERE OFFERED TERMINATION AT GIVEN STAGES OF THEIR PREGNANCY

Women were also asked whether they were given a timeframe within which they must make a decision around termination or continuation of their pregnancies (see Figure 4). Half of the women who were offered a termination were not given any time frame within which they must make a decision. One fifth of women were given a week or less to make a decision, with a further 13% given one to 2 weeks. Another 19% were given a timeframe of greater than 2 weeks.

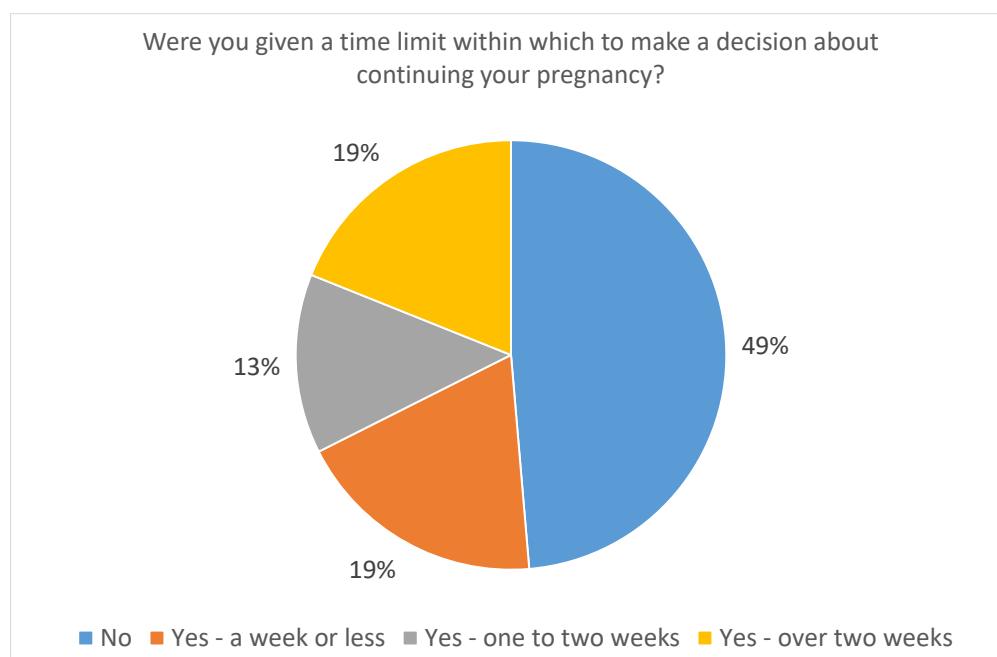


FIGURE 4. PROPORTION OF WOMEN GIVEN SPECIFIED TIME LIMITS IN WHICH TO DECIDE WHETHER OR NOT TO TERMINATE THEIR PREGNANCY

Women were asked about the frequency with which they were offered a termination, and whether they were offered again after first refusing the option. Only 35% had their initial decision accepted by their care givers, with 16% offered a second time after a first refusal. 27% of women say they were offered terminations several times after their initial refusal with a further 22% indicating they were offered terminations frequently through their pregnancies.

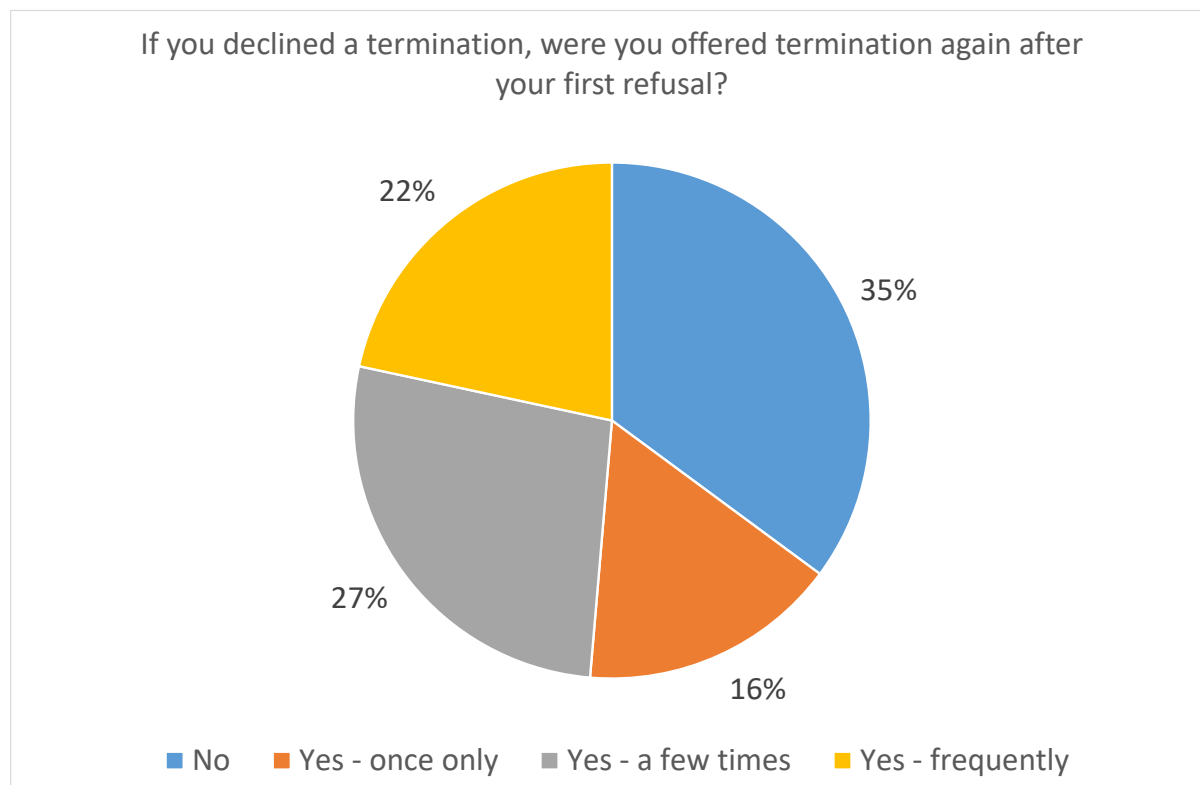


FIGURE 5. PROPORTION OF WOMEN ASKED ABOUT TERMINATION AT SPECIFIED FREQUENCY

The comments women made along with these responses make it clear that in some cases women are being placed under substantial pressure by medical caregivers to terminate their pregnancies. In some cases attitudes of medical staff are clearly coercive and designed to push women down the path of termination.

Several women made statements that indicate that medical caregivers indicated that termination was the most reasonable or commonly accepted outcome:

- *'One obstetrician asked why I was continuing with the pregnancy knowing the baby had Down syndrome.'* - #3 Baby born 2006.
- *'I guess I wasn't so much offered, as told it was a popular option.'* - #12 Baby born 2013
- *'We weren't offered a termination again. However various professionals asked us if we knew our baby would be born with DS.'* - #19 Baby born 2010
- *'Obstetrician said at first appointment "would have liked to have seen you sooner as we only have a small window of opportunity for a termination". I told him there would be no termination and I didn't want to risk the pregnancy with an amnio test.'* - #24 Baby born 1996
- *'Doctor only advised again on termination of pregnancy before 20 weeks, otherwise termination would be more complicated.'* - #47 Baby born 2016

Other women indicated that they endured attitudes and behaviour that constituted harassment, with women being made to constantly defend their decision to continue their pregnancies:

- *'I was harassed and treated negatively by medical staff when I refused to terminate.'* - #13 Baby born 2005
- *'[I was] offered termination every time [I] saw [the] obstetrician, also asked if I wanted everything done at time of birth or just let things take its course (baby born at 29 weeks), including the week before I had her.'* - #16 Baby born 2015
- *'It was decided I was at risk due to my age and should have a termination as I would not want a child like that at this stage of my life. I refused testing and would not listen to the doctor about termination.'* - #21 Baby born 1995
- *'I was told my child had 5% [chance] of living because she had Down syndrome. I was told to terminate or go home and wait for a miscarriage. One doctor asked upon my return if the baby was still alive. Very insensitive.'* - #22 Baby born 2015
- *'Shortly after my 28 week scan I learnt that my beautiful boy, that I waited 10 years for, happened to have Down syndrome. It was then that I was offered an "option B" - I could go home and have a still born! During the week before his birth at 29 weeks, I was visited by three more doctors (all on separate occasions and usually when I was alone) to discuss option B and that it was still available. They said they wanted to make sure I completely understood what my life would be like otherwise. By the third time, I was hysterically mad and demanded they tell me what they were hiding from me, as Down syndrome wasn't a concern of mine. I signed up to love my baby no matter what, so I was convinced they must have been hiding something else, something major. But they weren't. I guess they didn't think my baby was worthy of life given his diagnosis. To this day, I am heartbroken about that whole experience. Although the termination being offered wasn't a medical one, they seemed eager to push "the let nature take its course" option. I will never get over this. I look at the world differently and cannot believe when others believe their life is more important than others.'* - #30 Baby born 2013
- *'We were asked until quite late in the pregnancy (over 30 weeks) and this was with no confirmed Down syndrome diagnosis only a confirmed heart condition.'* - #40 Baby born 2013
- *'Yes!! Just about every time I had an appointment for a check up. I had to tell them I wasn't interested and didn't want to be asked anymore.'* - #41 Baby born 2008
- *'Our doctor was completely against us continuing with my pregnancy. She wanted us to terminate even though my son was born completely healthy with Down syndrome and no major health issues.'* - #45 Baby born 2015
- *'We were offered many times, as we had refused any invasive tests to confirm our diagnosis. We had already lost a child with trisomy 21 in a miscarriage, so with a chance of 1:2 we kind of acted like this child had it as well. The doctors were adamant we should know for sure but we refused, as we had already had two miscarriages, and had no live children.'* - #51 Baby born 2005

One woman outlined a more positive experience where her decisions around further testing were respected by her medical caregivers:

- *'My GP and OB were very supportive of my decision to not pursue further testing following a 'high risk' result on the 20wk combined ultrasound and blood test.'* - #52 Baby born 2014

Figure 6 shows the week in which women had a confirmed diagnosis of Down syndrome (vertical axis) versus the week they were offered pregnancy termination for the last time (horizontal axis).

Note that 36 responses were recorded for this question. This figure shows that while nearly one third of these women never received a confirmed diagnosis of Down syndrome, these women continued to be offered terminations right up to 36 weeks, with over 50% of them last being offered a termination later than 20 weeks. The bulk of confirmed diagnoses were made between 13 and 24 weeks, with terminations in this cohort being offered for the last time up to 32 weeks.

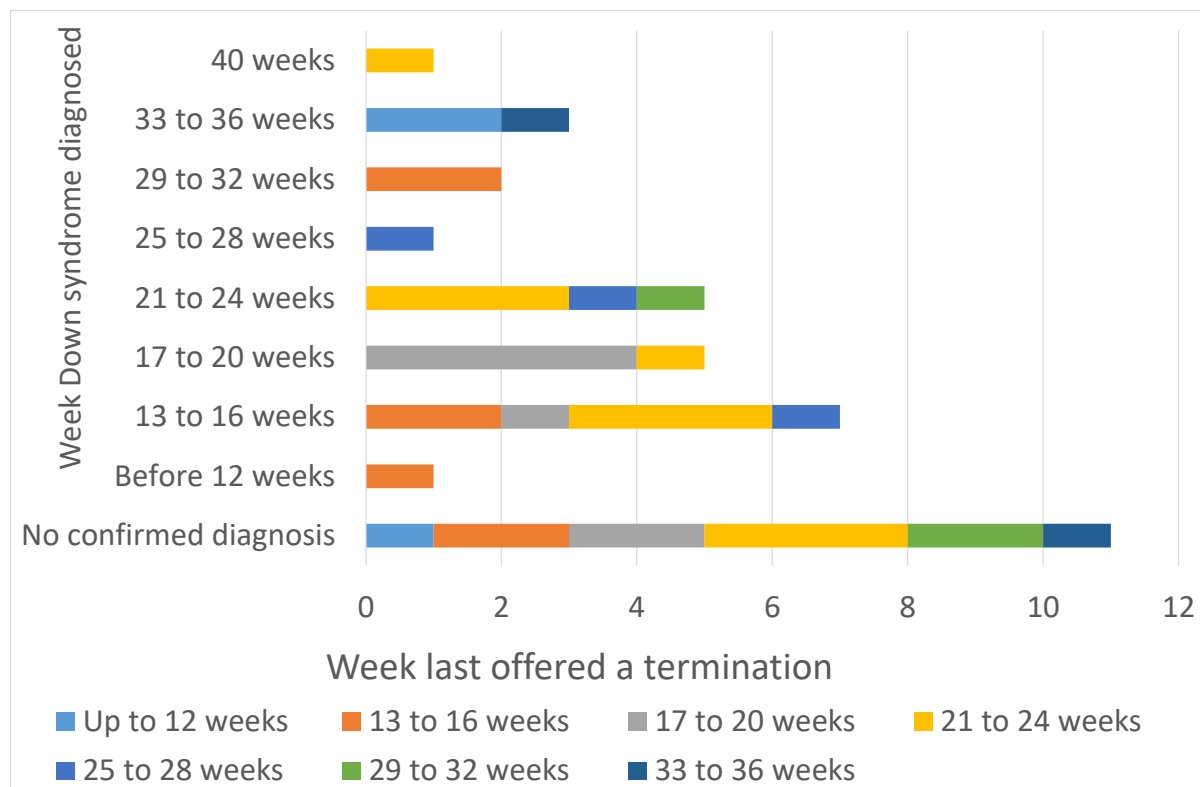


FIGURE 6. WEEK OF CONFIRMED DOWN SYNDROME DIAGNOSIS VERSUS WEEK IN WHICH WOMEN WERE LAST OFFERED A TERMINATION

As shown in Figure7, a small proportion of women (9%) were offered a further option of comfort care for their child rather than active termination of their pregnancy, where their baby would be kept 'comfortable' after birth but no active efforts would be made to keep them alive (eg. through denial of nutrition).

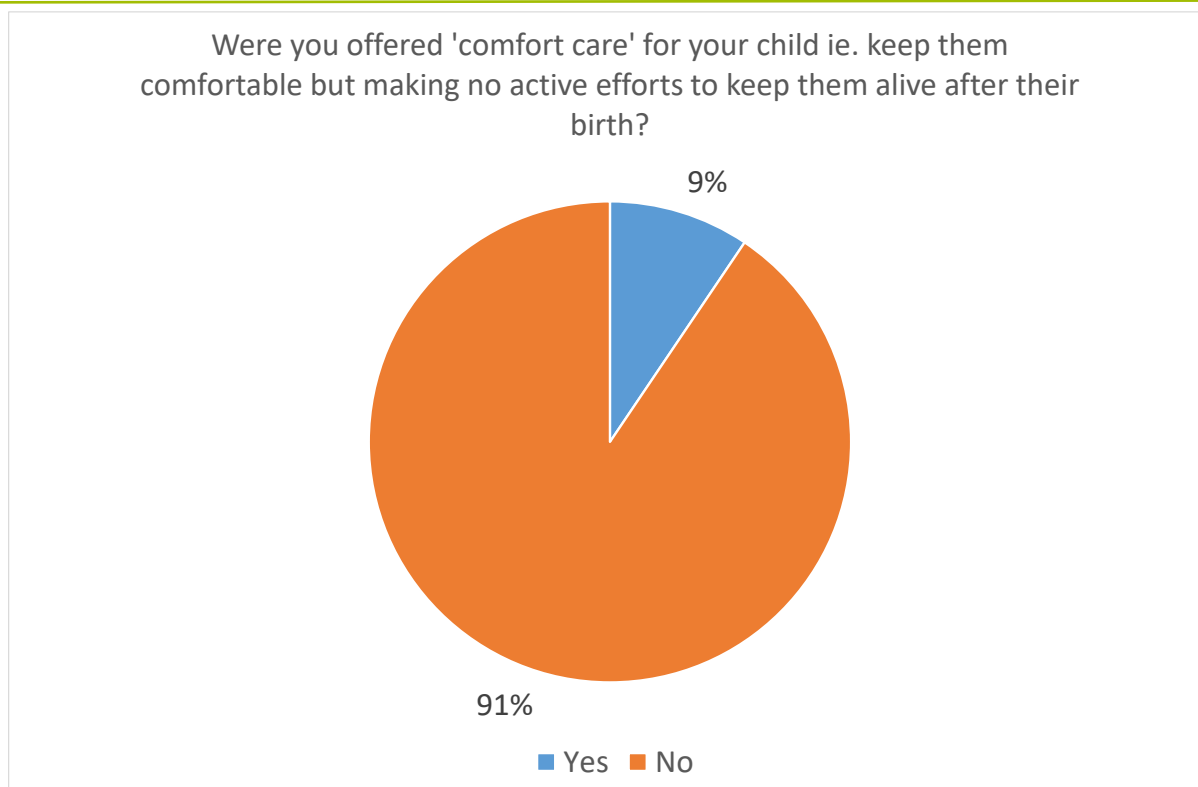


FIGURE 7. PROPORTION OF WOMEN OFFERED 'COMFORT CARE' (IE. KEEPING THE BABY COMFORTABLE BUT MAKING NO ACTIVE EFFORTS TO KEEP THEM ALIVE AFTER BIRTH)

These women expressed their fears for their babies and the pressure they felt to ensure appropriate care would be offered to their babies after birth, with one woman saying *'I made sure there would be paediatricians present at birth so she would be administered whatever help was needed. She required intubation at 3 and 20 minutes so lucky they were there.'* – #40 Baby born 2013. Another woman said *'I continuously told them as long as she has a strong heartbeat, termination was not an option.'* – #56 Baby born 2013. The view that this was a kind option to avoid their child suffering was also used to apply pressure, with one woman stating *'[It] wasn't worded as comfort care. I was asked if I wanted to watch my baby suffer or let things go naturally.'* – #16 Baby born 2015. One woman indicated that the option to withhold care after birth was offered even though she had stated clearly many times that termination wasn't an option they would consider *'E. was born at 32+6. Our previous fortnight appointment with an obstetrician (at 30/31 weeks) we were offered a termination despite stating that it was off the table, followed by information that it could be done up to term, and then were told that we could opt to withhold medical treatment instead. At this point we had a confirmed diagnosis of duodenal atresia so it was understood that this would kill her.'* – #54 Baby born 2015.

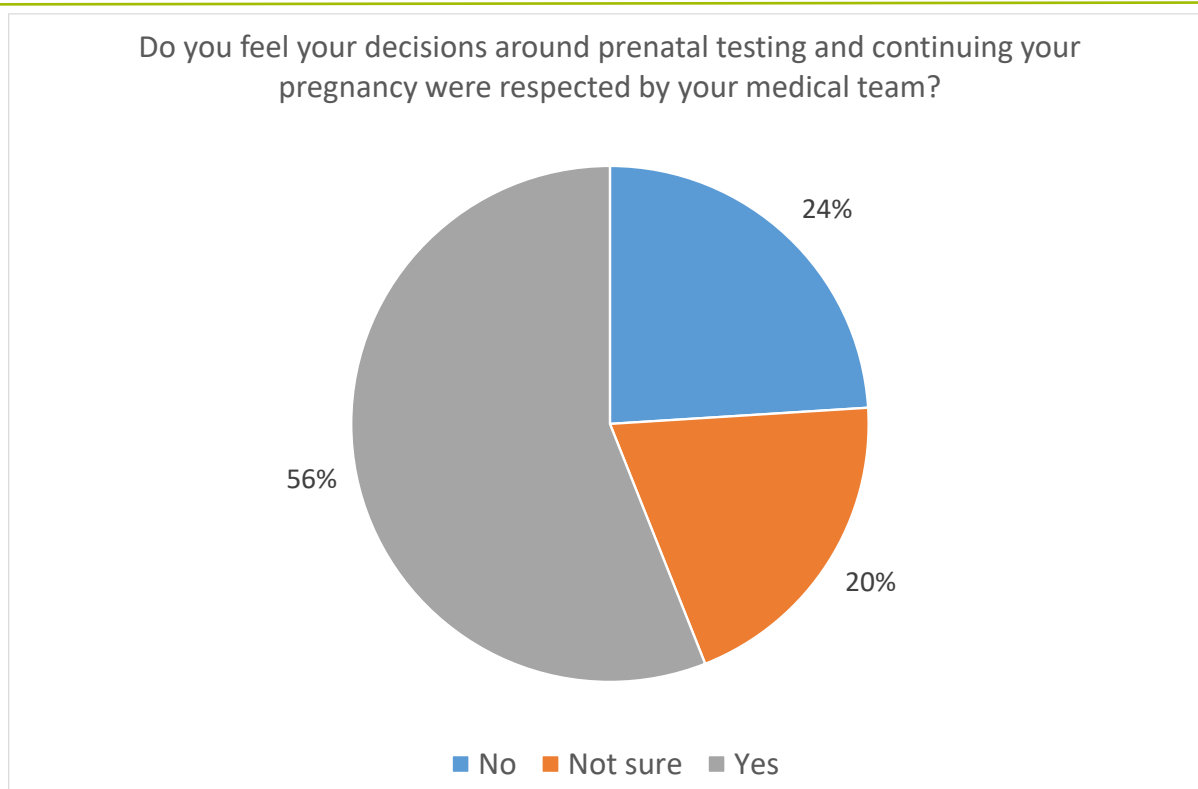


FIGURE 8. PROPORTION OF WOMEN WHO FELT THEIR DECISIONS AROUND PRENATAL TESTING AND CONTINUING THEIR PREGNANCY WERE RESPECTED BY THEIR MEDICAL TEAM

Women were asked whether they felt their decisions around prenatal testing and continuing their pregnancies were respected by their medical team (Figure 8). A quarter of women said they didn't feel their decisions were respected, with a further 20% unsure.

Where women felt their decisions weren't respected they recounted feeling harassed and coerced into undergoing diagnostic testing and/or termination. Practices included having their decision frequently questioned even after making it very clear that they did not want a termination, being told that diagnostic testing and/or termination is the normal pathway, having appointments made against their will and being told that they should abort to save their baby from a life of suffering.

- *'They tried to force me into having an amnio, even booking me in for one against my will and harassing me when I refused'* - #13 Baby born 2005
- *'I felt pressured to terminate that I was making the wrong choices'* - #16 Baby born 2015
- *'I was continually asked, even though I asked that my wishes be written clearly in my file.'* - #30 Baby born 2013
- *'Because he told me you don't want a child like that, he was very indifferent towards me from the point when we made the decision not to consider testing or termination.'* - #21 Baby born 1995.
- *'I chose not to have further testing and I was asked on many occasions why'* - #32 Baby born 2016
- *'My doctor at the hospital kept telling me to abort as it was not a good life for a baby with Down syndrome. I did ask for a new doctor.'* - #45 Baby born 2015
- *'Not really because they kept asking if we wanted to continue with this pregnancy.'* - #40 Baby born 2013

- *'One doctor was lovely, we saw her twice. The many other medical people doctors, midwives and nurses would ask me if we knew all our options and would push for amnio testing in a seemingly negative way. I was so sensitive to their negative attitudes. One said most people just get rid of them.'* - #46 Baby born 2013
- *'A few medical practitioners seemed surprised about our decision. There was often an uneasy atmosphere that we felt we needed to dissipate.'* - #19 Baby born 2010

One woman told of delivering her baby without a doctor present even though she had a high risk pregnancy, with consequences that could have been life threatening *'Because obviously they knew my daughter most likely had Down syndrome, they set a date to induce me for delivery because my blood pressure was high. On my delivery day there was no doctor around, only one mid wife delivered my baby, although I was high risk. My baby came out blue and not breathing. The mid wife pressed the emergency alarm. Around 5 to 10 doctors or nurses came to my wards and took my daughter to Intensive care.'* - #20 Baby born 2013

One woman who was unsure of whether her decision was respected or not said *'Medical professionals seem to be able to mask their feelings, not sure nor do I care what they thought about me keeping my baby and what they thought when some two years later we had our third child who also has Down Syndrome.'* - #2 Baby born 2012

Of the 56% of women that said their decisions were respected there were in some cases mixed experiences with various members of their team. In the words of one woman *'the midwives at home and the obstetrician who delivered her were amazing. All supportive. Once we ditched the negative obstetrician [our decisions were supported]'* - #3 Baby born 2006. Another woman said *'it was a mixed reaction. One doctor was good, the other one was a total jerk!'* - #9 Baby born 2009. Some women reported actively changing care givers after their screening or diagnostic tests, to a team who were more supportive of their choices

- *'The initial GP who delivered the screening results did not seem supportive. I then switched to an obstetrician who was supportive of our decision not to undertake further tests.'* - #10 Baby born 2015.
- *'Yes our decisions were respected by our GP, but the specialists, who we travelled to in our nearest city left a lot to be desired.'* - #51 Baby born 2005
- *'Genetic counsellor complete waste of time, obstetrician on the other hand very neutral, I would never have known her personal views. Very supportive and respectful for the remainder of the pregnancy. Although [I] was never given any info in regards to Down syndrome. But I was very proactive and sourced this through Down Syndrome Victoria'* - #43 Baby born 2015
- *'To be honest, I stopped caring what they thought and didn't really pay attention to their personal feelings. They were required by law to follow through on our wishes and that was all that mattered. The obstetrician we saw just a few days before E. came into the world was wonderful, and possibly instrumental in helping her cope with prematurity so well. He was proactive in caring for her and sent me for steroid injections to strengthen her lungs in case she came early, which she did! No one else seemed to care enough about there to do that sort of thing.'* - #54 Baby born 2015

One women recounted her experience as being largely positive with genetic counsellors and her obstetrician accepting her decision but indicated some discomfort with the ease with which termination was discussed - *'The genetic counsellors at the place that does the blood analysis notified me of my results fairly quickly after my blood test that morning. They told me my*

probabilities if doing further tests like CVS, amnio and Harmony but I declined. They said they would let my obstetrician know my decision but never pressured me into anything. A week later at my obstetrics appointment he only asked once more to confirm my decision. The only thing I would say is that they all talk about termination like it is an everyday natural occurrence. I guess they need to be unbiased as not every parent can cope with a child with special needs but the way they portray termination should be less nonchalant.' - #28 Baby born 2016

Another woman also indicated that she was left feeling termination was the 'normal' choice saying *'When we eventually made decision it was respected. However I felt they expected us to choose to terminate and no one really thinks anyone would choose not to terminate'* - #39 Baby born 2012. Another woman indicated she felt her decision was respected even though termination continued to be offered *'Although they supported my decision termination was still offered'* - #56 Baby born 2013

Other women had very positive experiences where their team embraced their decision to keep their baby.

- *'My team all knew that we would welcome a child with Down syndrome and respected that. We all worked towards a natural birth and were able to that in a low risk birthing suite. It never felt like a big deal to them.'* - #15 Baby born 2012.
- *'I was given all the options. Not once was I told to do this test or that test but I wanted to know for sure the diagnosis and they just supported me on how to go about that.'* - #7 Baby born 2014
- *'As soon as we told our obstetrician we were definitely keeping our baby he jumped straight into the excitement of the pregnancy. He was devastated when we lost our beautiful boy and told us that he had been really looking forward to supporting us through our journey.'* - #8 Baby born 2015
- *'Respected my choice not to have diagnostic testing not mentioned again and tried to reassure that all indicators (soft) were "normal".'* - #29 Baby born 2009
- *'We were generally well-supported & respected in our decision to continue the pregnancy & love our son'* - #33 Baby born 2011
- *'The obstetrician asked once if we were interested in doing an amnio, we said no and he never asked again. Now when I look back he knew but did not push as he realised that we were going to keep him.'* - #42 Baby born 2006
- *'Very much so.'* - #44 Baby born 2007
- *'Once they understood the pregnancy was wanted regardless, I was treated as per my previous pregnancy.'* - #49 Baby born 2012
- *'The first time I told my GP and obstetrician that I would be keeping the baby regardless of any further testing results and that I wasn't willing to risk my pregnancy for confirmation, they allowed me to continue with my pregnancy without any pressure.'* - #52 Baby born 2014
- *'My obstetrician said given my age the chances were unlikely so the pregnancy was just treated as if Down syndrome was never mentioned'* - #53 Baby born 2011

Quality of care

Women were asked to rate the delivery of their screening and diagnostic test results. As shown in Figure 9, 60% of women rated this as poor (27%) or very poor (33%). A further 24% rated it as OK with just 16% rating it as good or excellent.

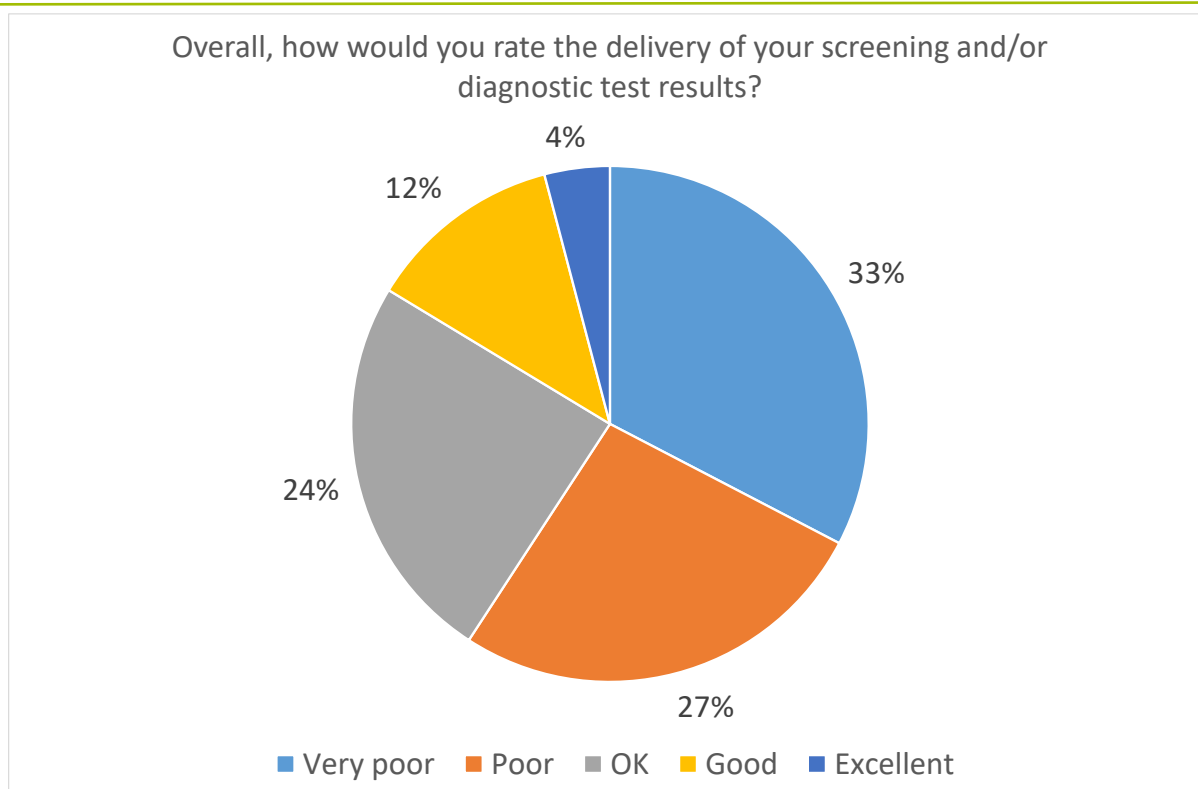


FIGURE 9. PERCEIVED QUALITY OF DELIVERY OF SCREENING AND DIAGNOSTIC TEST RESULTS

Common themes amongst those who rated the delivery of their test results as poor or very poor were the negativity with which the results were delivered and discussed, the presumption that the doctor made that either diagnostic testing or termination was the next logical step, the lack of up-to-date information on Down syndrome provided and the poor prospects attributed to their babies.

- *'I was in tears but shunted from person to person and they insisted I have the CVS then and there although I wanted to have my husband present and didn't really want the procedure. They insisted it was necessary and gave me horrific case scenarios to convince me. All while I was sobbing and my other children were waiting in another room.'* - #26 Baby born 2005
- *'I was made to feel like my screening test results (1/8) was terrible news and told that the next step was an amnio to confirm the diagnosis. It was assumed I would do this despite the small chance of miscarriage and regardless of having experienced two losses prior to this pregnancy. When I declined the doctor's body language and manner were not supportive of my decision. I never went back to that doctor.'* - #10 Baby born 2015
- *'My son wasn't diagnosed with Down syndrome until after he was delivered. Lucky for us we had a wonderful paediatric doctor who had a more positive approach to our son's future. The obstetrician was never supportive. Don't think he ever looked at our baby again after we left the delivery room. Certainly never asked how he was going or even looked at him when I went back for my 6 weekly check up.'* - #21 Baby born 1995
- *'Because it was initially based on the appearance of one finger joint and was presented to us as if our baby had something really terrible. After a scan of the baby's heart they were even more sure she had Down syndrome and more negative.'* - #13 Baby born 2005
- *'Termination was first mentioned, every bad thing about having a baby with special needs mentioned but then would add but oh they can have somewhat normal lives.'* - #16 Baby born 2015

- *'My obstetrician made himself scarce and didn't tell me a paediatrician was coming in to check her. After diagnosis he visited at 5am every morning and at one stage told me "there are good institutions for them these days".....I asked the nurses to tell him after that that I never wanted to see him again.'* - #34 Baby born 2007
- *'I was in NSW traveling and the nurse said: so are you going to terminate? You need to know what you want to do. She kept asking this question like she was talking about a sick pet.'* - #35 Baby born 2015
- *'The first words from the doctor who delivered [him] were 'I'm sorry'. Right from the moment he was born it felt like the doctors and most nurses/midwives were unsure of what to say.'* - #53 Baby born 2011
- *'We had some decent help (the genetic counsellor we saw after the duodenal atresia diagnosis at 23 weeks was positive and referred us to DSV), and some down right awful (pushing for amnio, repeated offers of termination etc).'* - #54 Baby born 2015
- *'No up to date information given, I was the one telling them about the local services and being told "I'm sorry" before delivering a confirmed diagnosis and other inappropriate views was not called for.'* - #56 Baby born 2013
- *'I was given the diagnosis over the phone. The tone of the doctor was negative and he seemed shocked that I was not upset. He did not refer us for genetic counselling or refer us to any other professional. I asked about making contact with a cardiologist.'* - #19 Baby born 2010
- *'They were very cold and said they were sorry. They painted Down syndrome in a negative way.'* - #30 Baby born 2013
- *'[They] made [it] all sound very hopeless.'* - #32 Baby born 2016
- *'[The doctor] who did 12 week scan told me this pregnancy should be terminated ASAP based on measurements and didn't want to give me recording of scan.'* - #32 Baby born 2012
- *'All we were told was our child would have lots of appointments and not be able to achieve much.'* - #40 Baby born 2013
- *'I refused an amniocentesis but was asked over and over to consider termination.'* - #41 Baby born 2008
- *'I was at work and the diagnostic ultrasound medical practice contacted me over the phone to inform my high risk (1:9) of having a baby with Down syndrome, also the lady said "I'M SORRY to inform you that you're a high risk".'* - #47 Baby born 2016
- *'My GP had no information in regards to Down syndrome. If it weren't for Facebook groups and Google I would have no idea what Down syndrome actually is.'* - #48 Baby born 2016
- *'Upon confirmed results the obstetrician immediately assumed termination.'* - #49 Baby born 2012
- *'The doctors must have had an idea, we were asked for amnio and we refused. My husband told them that he does not believe in termination even if the child had a disability. The ultrasound person at my 36 week scan said to me does your child have Down syndrome or a disability because it sure looks like it, and if not there is something wrong with it.'* - #42 Baby born 2006
- *'Results given over the phone, be told I'm sorry your baby has DS. Given no info about anything.'* - #43 Baby born 2015
- *'It was said that I would be in shock. I am going to give you the results. Are you sure you haven't got anyone here to support you when I tell you this news? Lots of talking.....when I finally convinced them to give me the facts.....she said my baby had 1:2 chance of Down Syndrome.....so what.....can you tell me if my child will have red hair? Nowell you can't*

tell me if my baby has Down syndrome.....is my baby alive ok thanks bye.' - #2 Baby born 2012

- *'Over the phone they kept saying "I'm so sorry" then in GC was positive about it.'* - #6 Baby born 2013
- *'After half an hour of delivering my baby I was so tired and exhausted they took me to ICU to my daughter and told me the news. They could not wait until I rest[ed] from my delivery.'* - #20 Baby born 2013

Several women recounted experiences that while not explicitly negative indicated a fairly passive approach to the delivery of their results.

- *'My GP told me my results were in my file, we could have a look and see what they were.'* - #3 Baby born 2006
- *'The person who told us refused to look us in the eye and kept asking questions about prenatal testing before actually saying that they suspected our child had Down syndrome.'* - #23 Baby born 2016
- *'They advised us of their suspicion, advised that they needed to do more tests and left us to deal with it. I rang my GP who came in later that day to try to help us.'* - #25 Baby born before 1990
- *'It was very off the cuff.'* - #5 Baby born 2013
- *'We were offered three choices in the order - Termination (the highlighted choice), Do nothing, Adoption.'* - #8 Baby born 2015
- *'A week after having amnio (when told it would be 2 days) I received a phone call to tell me it was Down syndrome.'* - #36 Baby born 2014
- *'I was told after my 20 week scan I needed to see a specialist as bub might have a heart defect. I get to Townsville only to be told that bub had no defect of his heart but he had Down syndrome. My local team never told me he had Down syndrome and they gave me my scan results over the phone about his heart.'* - #45 Baby born 2015

Where women rated the delivery of their results as excellent or good they indicate that their medical team were compassionate and understanding as well as positive. Women were offered choices and given information (although not always up-to-date) and then allowed time to make their own decisions.

- *'The sonographer was fantastic and looked at [angle] possible to see any other health conditions present eg. heart defects. Obstetrician was fairly nonplussed and said that most people go through with a termination but he was very supportive of our decision and said he would treat my pregnancy just like my other pregnancies, which he did.'* - #7 Baby born 2014
- *'I was informed about the test results in a matter of fact way with no judgement or presumption of the path I should take. I was given all options and left to make up my own mind. My decisions were respected. The delivery of the 20 week scan results which concluded my baby would almost certainly have DS were delivered over the phone while I was at home but I already had a good relationship with my midwife and was expecting the results. In hindsight I was very upset at home alone after the call but I don't think hearing it in person would have made a difference to me.'* - #15 Baby born 2012
- *'Found out after birth, had blood test done and results given following day - Hospital staff/doc very compassionate, gentle, understanding and positive.'* - #28 Baby born 2016
- *'I think my obstetrician was a little dubious with the results and therefore was not pushing for further testing. He was very positive overall. I have never asked whether it was because*

he did not believe the test results or that he did not see having a child with Down syndrome a big deal.' - #29 Baby born 2009

- *'The doctors involved were professional & fairly compassionate. They did not push us to have a termination. They offered fact-based (if somewhat outdated) info, but certainly not positive info offering hope for a good future.'* - #33 Baby born 2011
- *'We knew as soon as we saw him. It was a bit of a traumatic birth, he received resuscitation and taken straight to ICU. The next day the doctor asked us if we thought something was different about our baby, we were just worried about his health. Then we realised what she was asking us and we said "oh we know he has Down syndrome". She was trying to be gentle. The ICU nurses greeted us with congratulations and were very positive re: t21.'* - #46 Baby born 2013
- *'It's all a blur really but I can't remember anything but concern from our technician.'* - #51 Baby born 2005

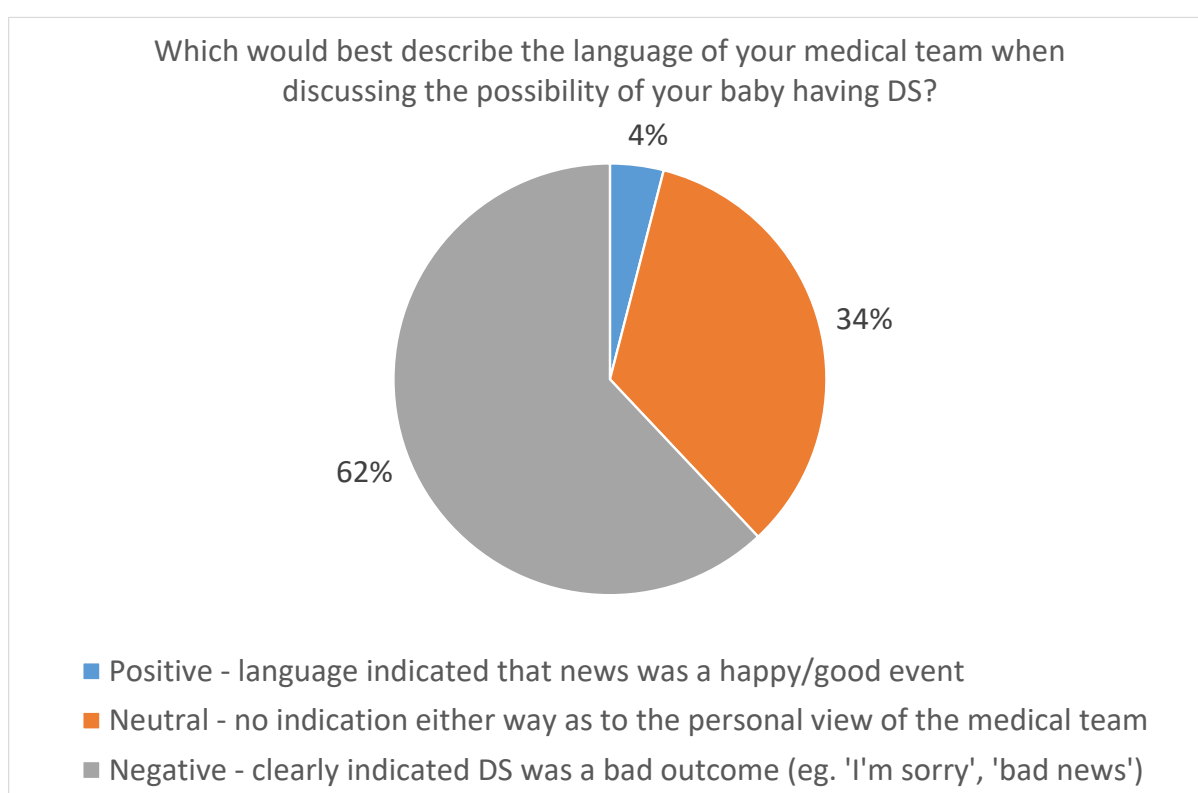


FIGURE 10. LANGUAGE USED TO DESCRIBE THE POSSIBILITY OF THEIR BABY HAVING DOWN SYNDROME

Women were asked about the language used to deliver their results (Figure 10). The majority of women (62%) indicated that negative language (eg. "I'm sorry, I have some bad news") was used to deliver their results, with 34% of women receiving their results in neutral language and 4% saying the language used was positive. Where negative language was used it conveyed that Down syndrome is a bad outcome, frequently with a presumption that diagnostic testing and termination would be the next steps. In these cases no up to date information on Down syndrome was provided and women were led to believe that people with Down syndrome tend to have poor quality lives.

- *'Lots and lots of negativity in the prenatal counselling, bad news, never.'* - #3 Baby born 2006
- *'Negative - clearly indicated Down syndrome was a bad outcome (eg. 'I'm sorry', 'bad news').'* - #4 Baby born 2010

- *'I suppose the future was not painted too rosy but then again they acted as neutral as possible given that they knew I was very knowledgeable of what to expect.'* - #7 Baby born 2014
- *'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.'* - #13 Baby born 2005
- *'Was given info over the phone and told I'm so sorry but it looks like your baby will have Down syndrome go have the NIP test tomorrow so that we can confirm so that way we can go look at termination after an amino in a few weeks.'* - #16 Baby born 2015
- *'The diagnosis was prefaced with 'I'm sorry' and no explanation was given or support services recommended. The doctor seemed to assume that we would not continue with the pregnancy or would be heartbroken. He did not seem to know how to meet my response. (This was a doctor from a private ultrasound practice that specialised in women and pregnancy. I was surprised at the lack of expertise in delivering a diagnosis).'* - #19 Baby born 2010
- *'Prenatal attitudes seemed very negative. Being told they make exceptions for these births to do them [terminate] further along.'* - #46 Baby born 2013
- *'The GP who called me in after our 12 weeks scan said "I'm sorry, but you've had an abnormal scan and your window for termination is rapidly closing" as I walked into the room. He was clearly panicked at the time.'* - #54 Baby born 2015
- *'... paediatrician was a little more negative but more ill-informed and uncomfortable, lost for words not necessarily overly negative.'* - #29 Baby born 2009
- *'My local medical team didn't want to treat me let alone want me to give birth to my son at the local hospital they wanted to hand me over to another hospital further away from my home town.'* - #45 Baby born 2015
- *'The ultrasound medical practice had a negative language, however my obstetrician was neutral.'* - #47 Baby born 2016

Women who received their results in a neutral or positive tone had a mix of experiences. Some indicated that while the news was not easy to hear, it was delivered in a matter-of-fact way but with compassion.

- *'The information was delivered very matter-of-factly with a lot of sympathy and time to ask questions etc. I don't remember feeling as though she thought it was bad news but the news itself was still difficult to hear.'* - #15 Baby born 2012
- *'The paediatrician who was there said to my husband that there were signs of our child having Down syndrome [and that we] must test, to be sure. But she was more concerned that he was born premature and had a heart condition. She did say many positives as he can do many things, that it just will take a little longer for them to learn.'* - #42 Baby born 2006
- *'Our GP who delivered our child, was excellent.'* - #51 Baby born 2005
- *'Spoke in matter of fact tone. Gentle but not negative, spoke of potential problems but also of improved life compared with decades ago.'* - #28 Baby born 2016
- *'Not all were negative. First obstetrician was neutral but told me she would terminate, but mentioned [she] know[s] people who have [a] child with Down syndrome and [are] happy.'* - #39 Baby born 2012
- *'[Positive] Despite being asked multiple times whether I was going to continue the pregnancy.'* - #48 Baby born 2016

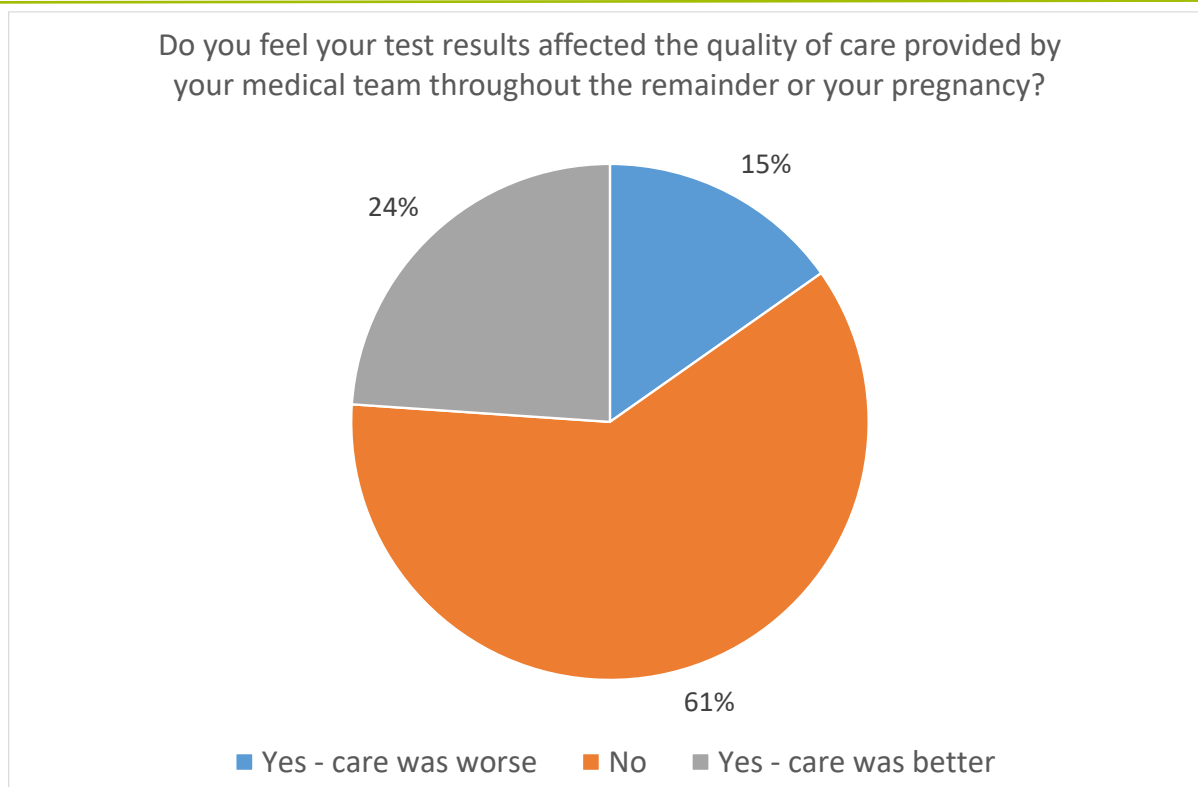


FIGURE 11. PERCEPTIONS OF THE EFFECTS OF PRENATAL SCREENING AND/OR DIAGNOSIS ON QUALITY OF PRENATAL CARE

Figure 11 shows how women perceived their prenatal screening and/or diagnostic test results affected the quality of care they received during their pregnancies. The majority of women (61%) felt their pregnancy care was unaffected by their results, while 24% felt it improved the quality of care they received. Only 15% felt they received poorer quality care as a result of Down syndrome being identified in their pregnancy.

For the majority of women who felt their care was not affected by Down syndrome being identified during their pregnancies, they indicated that their pregnancy was managed as normal, although in some cases additional care was taken with scans or around birth.

- *'My pregnancy was treated just like any other pregnancy and no special care was provided for. Although in saying this the obstetrician who delivered my baby was on call. In hindsight I think he rushed me through the end part of labour to get my baby out. Maybe that's the way he practices however I felt he just wanted to get my labour over and done with because of my baby's diagnosis. Happy to stand corrected on this.'* - #7 Baby born 2014
- *'I felt I would get good care regardless of the results.'* - #15 Baby born 2012
- *'No change, I did have paediatrician at my child with Down syndrome's birth but that was probably because he was born a month early - no apparent reason.'* - #29 Baby born 2009
- *'I was advised at 28 weeks and my boy was born at 29 weeks, so not enough time for me to notice this.'* - #30 Baby born 2013
- *'It feels like I received the same amount of care, just an extra scan here and there.'* - #48 Baby born 2016

Where women felt their care was better than it would otherwise be, they referred to extra scans to check on how their baby was as well as access to specialist support such as lactation consultants, tours of the NICU and other specialists (such as cardiologists).

- *'It was better in that we had more scans & testing. Some of the extra care I asked for (ie was not offered) eg appointment with a lactation consultant & a tour of NICU.'* - #33 Baby born 2011
- *'We both received a very high level of care, access to specialists and fantastic support.'* - #44 Baby born 2007
- *'My obstetrician did all tests and had all the care possible so my baby could be born healthy and well.'* - #47 Baby born 2016
- *'Our GP prepared us, and was very happy that our child was born healthy! The nurses in the hospital were very attentive.'* - #51 Baby born 2005
- *'The pregnancy was normal, so I was treated as such, however during labour I feel the team was particularly cautious, and took time to show me around special care nursery before etc.'* - #49 Baby born 2012
- *'I was able to go and have an Ultrasound whenever I wanted to check on how A. was doing.'* - #56 Baby born 2013

In some cases the better care resulted from women choosing to move their pregnancy care to a new medical team.

- *'Post screening results we found an obstetrician who specialises in high risk pregnancies and felt very supported by him during the pregnancy.'* - #10 Baby born 2015
- *'We had more regular monitoring due to our sons heart condition. Once we changed our prenatal care we felt more supported.'* - #19 Baby born 2010

Women who felt their results had led to worse care, recount experiences where they were treated with contempt for choosing to continue their pregnancy, the focus of their pregnancy care became Down syndrome and the standard basic monitoring of pregnancy health was not carried out, or they were put in situations in which their baby was placed at risk, due to either inaction or lack of appropriate medical staff being available at birth.

- *'They set a day to induce me and told me the doctor and midwife will be there for my delivery. On my delivery day only one mid wife was there although they knew I had high blood pressure and my baby had DS. When I delivered my healthy son in the same hospital there were two midwives and a doctor [who] delivered my son.'* - #20 Baby born 2013
- *'It was clear that the majority of my medical team felt strongly that we should have terminated. They voiced this on several occasions and were barely polite.'* - #13 Baby born 2005
- *'Yes [care was worse] my c section was consider an emergency at 8:30am they found issues at fetal monitoring blood flow going wrong way reduced movement to hardly anything but my emergency C section never happened until 5pm. While waiting for my C section a nurse couldn't find baby's heartbeat, got someone else they couldn't find it but instead of being concerned they said" it's ok she's off for [a] C section later" - yeah 4 hrs later.'* - #16 Baby born 2015
- *'The doctor who was treating me before I requested a new doctor, was always telling me to abort. She didn't care to treat me at all or talk about long term outcomes for bub.'* - #45 Baby born 2015
- *'We were bumped around to different hospitals. Care became so focused on Down syndrome and duodenal atresia that basic pregnancy things were left by the way side. No one was screening my urine, no gestational diabetes test was done, no testing of my iron levels etc. All the routine things I had with my first were totally ignored. I became the incubator of a*

child that they felt probably should have been terminated. This attitude continued into my postnatal care as well.' - #54 Baby born 2015

Women's views on best practice

Women were asked their views on what best practice should look like in terms of discussion of prenatal testing, delivery of test results and ongoing care for pregnancies with either a Down syndrome diagnosis or identification of a high chance of Down syndrome. In their eyes best practice should involve:

- Clear communication that undertaking screening and diagnostic testing is voluntary and that women are able to opt out if they wish. In these cases their choice should be respected.
- Women should be encouraged to make an informed choice around screening – to think about their own reasons for screening, the possible outcomes and what they will do with this information before undertaking screening tests.
- Use of neutral language and a factual approach to delivery of the diagnosis/screening results should be applied. Results should be delivered as 'news' and not tainted by judgement.
- Women being supported in their choice to continue their pregnancies.
- Accurate information on Down syndrome, both positive and negative aspects, including the positive effects a child with Down syndrome can have on families, modern expectations of what people with Down syndrome can achieve and what their lives can look like as well as health issues.
- Links to support organisations including State based Down syndrome Associations and an opportunity to meet with parents of children with Down syndrome.
- Uncertainties in screening and diagnostic test results should be adequately discussed and the risks to the pregnancy of invasive testing should be acknowledged.
- Termination should not be presented as the only, or the natural, option.
- Women should have support when being delivered results. Ideally results should be delivered in person or by someone who has a good relationship with the woman.
- Discussions around medical issues that may occur in a child with Down syndrome should be balanced with information about the possible outcomes and options available to manage these (eg. cardiac surgery) and their success.
- Social and emotional support should be offered to women regardless of whether they decided to undertake invasive tests or whether they choose to continue or terminate their pregnancy.
- Women should know that regardless of what decisions they make with regards to screening, diagnostic testing and continuing their pregnancy, they will be supported by their medical team.

In their own voices, the views of women about best practice are given below.

'Non-biased neutral advice and care which is in support of the continuation of the pregnancy.' - #5 Baby born 2013

'Say high chance instead [of risk]. Don't say I'm so sorry.' - #6 Baby born 2013

'You need to be given all the options as I think this is important. You also need to be aware of the health needs that can come with Down syndrome but just as importantly, the positives too ie. what they can achieve, what they bring to the family, support that is out there, it's not all doom and gloom. Gone are the days of institutionalised care and people with Down syndrome are active

members of the community with something to contribute. They also have a positive impact on siblings and at the end of the day are another sibling who happens to have an extra chromosome. Opportunity to talk to those who have recently had a child with Down syndrome would be good however, they need to be someone who will be honest and truthful about their journey. No good sugar coating things as it can be tricky in the beginning. Access to local State Down syndrome Association and linking the parent into them is important. I did this when pregnant and that really helped with giving me the most up to date information that was factual.' - #7 Baby born 2014

'More education from care givers about conditions that could affect my baby and what to look out for and ask for. Having said that, I don't know if I would have taken anything in as I was in denial and shock the whole time.' - #9 Baby born 2009

'Other mums are more articulate on this topic but it would be an improvement if neutral language was used to discuss results and information including contact details available for mums who choose to continue their pregnancy without confirming a diagnosis.' - #10 Baby born 2015

'Medical staff need up to date information, attitudes need to be changed. Delivery of diagnosis should be more factual and not negative. Information from people who have kept their babies with Down syndrome should be provided as well information on early intervention and positive outcomes. Termination (which is something I disagree with) should only be discussed in the light of up to date facts about Down syndrome rather than given as the primary option. The possibility of errors in the testing should also be mentioned, as well as the chances of the testing contributing to a loss of the baby (which is underplayed). Time should be given for parents to talk to each other, to parents who have children with Down syndrome if they want to, to paediatric specialists who can explain Down syndrome correctly rather than someone just saying the child will have no quality of life. The possibility of adoption also should be discussed if the parents don't feel they can raise a child with Down syndrome. Care provided should be at least as good as for a typical pregnancy.' - #13 Baby born 2005

'Judgement free. Delivery of facts with an explanation of what they mean and why they are given. Balanced information. Be guided by the parents. Refer to state based organisations. Be human.' - #15 Baby born 2012

'Understanding and caring giving informed up to date information about any diagnosis. Giving unbiased advice/information.' - #16 Baby born 2015

'Extra emotional support by qualified and experienced staff. Accurate and unbiased information presented.' - #18 Baby born 2015

'I think that everyone deserves the right to make an informed decision when it comes to the health of their children and themselves. Families should be presented with CURRENT evidence based information, professional and peer support, and a balanced delivery of the diagnosis.' - #19 Baby born 2010

'Our results (1 in 750 chance) were glossed over, we were offered further testing but as we said we wouldn't terminate it was never discussed again. I know that a lot of things can happen during pregnancy but I felt that maybe we should have been pushed to have more tests. I don't know if I would have rather known during pregnancy or not... but E. is my first child and I knew no different.' - #23 Baby born 2016

'Respect for family decision around knowing or not knowing, terminating or continuing. Subject of termination/ possibility of DS not raised again after first appointment.' - #24 Baby born 1996

'Usually best guided by medical staff, but needing to realise that they don't always get it right, when you think they should.' -#25 Baby born before 1990

'While health risks need to be discussed, a positive picture should be put forward for the life of a child with Down syndrome. It should be explained that they will still be a child and be loveable just the same.' - #26 Baby born 2005

'I really think information before testing like I received is so important. A lot of mums are just ordered screening and they haven't thought about why or what it means. In terms of delivery of results I think linking with State Down syndrome Association is important. Perhaps linking with a family who has a child with Down syndrome.' - #27 Baby born 2011

'I had no idea the first trimester combined screening was optional and this was my 4th pregnancy! I don't mind the testing or the fact the test exists as having information is useful. What I am unhappy about is the normalcy of terminating a child with Down syndrome or terminating based on a risk result when it could be a perfect child. I understand that people don't want a sick child to suffer or put stress on the family but what about the children with chromosomal abnormalities that don't have many issues and could potentially live out satisfactory lives? I would also like the error rate of the tests to be more openly stated as most people take the result and don't realise it is not only just a probability and that they need to account for an error rate as well. I would also like the doctors/genetic counsellors to deliver the option of termination of a pregnancy in a less blasé way, it is currently more of a scientific medical procedure but the way they portray it, you forget that it is a life they are taking.' - #28 Baby born 2016

'I think open discussion of what tests are for why you should or should have them. I think that the perception is that the testing is to weed out the "wrong ones" kind of like the bad nuts in Charlie and the chocolate factory, where the 'bad' nuts are trashed, needs to change. Language around Down syndrome needs to change too and so does society's expectations of what is achievable for people with Down syndrome. The available publications need to be up to date and informative, objective and not old and negative.' - #29 Baby born 2009

'Providing best care to each and every mother & baby. Provide support and positive information. RESPECT the mother's wishes! Understand that all life is worthy. Congratulate the parents on their beautiful baby. Don't ever talk to her when she is alone. Allow her to have support with her.' - #30 Baby born 2013

'Clear information, unbiased (i.e. not negative attitudes) contact with parents with Down syndrome kids. Not describing a Down syndrome pregnancy as a risk, better press for kids with Down syndrome. Not making it so easy to abort (and I'm not a pro-lifer). Legislation that sanctions the possibility of survival for babies with Down syndrome.' - #31 Baby born 1998

'It, of course, needs to be unbiased. The words 'chance' instead of 'risk' used. It needs to be about choice and control AFTER balanced information is provided. The NIPT should NOT be referred to as the 'Down syndrome' test but should be offered as a chromosomal disorders screen with balanced up to date information offered prior, in order for deliberation and reflection. The parents should be counselled as to the possibility of having to make a heart wrenching decision should the test come back positive or high chance. More supports for post diagnosis need to be readily available. It's not just about the information about Down syndrome, it's also very much about the human emotions, the complex and diverse reactions that people experience depending upon their backgrounds, experiences and their support networks. The diagnosis should be given as 'news' and not tainted with judgement. It should not be solely medicalised and all discussions about medical issues should be

balanced with discussions about how those medical issues can be treated or fixed (i.e. cardiac surgery).' - #34 Baby born 2007

'[I] think pregnant women should be given all the information, not just the bad part. Pro-choice should also include the choice NOT to terminate, not just a choice to terminate. Choosing to continue with my pregnancy was the hardest thing I ever did, but also the best thing!' - #39 Baby born 2012

'You should be given up to date info on Down syndrome. You shouldn't hear the words "sorry but" before you are told there's a "risk" your baby may have Down syndrome. Your wishes to not have further screening should be respected and not be asked every week "have you had your amnio yet?". You shouldn't be asked at every visit "do you wish to continue with this pregnancy ". Some positivity goes a long way. No scare mongering. Positive information.' - #40 Baby born 2013

'If there is a positive diagnosis, I think that doctors should give the positives as well as the negatives, and let the families to decide on their own. As I know it is an individual decision, it should be made in the family and not influenced by doctors, which seem to only give the medical but not what the child can do.' - #42 Baby born 2006

'My experience was very 'medical' and I think it would have been better if it included more social and emotional support.' - #44 Baby born 2007

'Doctors should be given more up to date information and a better education on Down Syndrome as well as list of people or support groups for expecting parents to make contact with. They also need to treat these mothers/ families the same as any other mother to make their experience more enjoyable.' - #45 Baby born 2015

'Non-judgemental, up to date terminologies, non-patronising, educational.' - #46 Baby born 2013

'The professional must try to give the news in a positive way. Who has to decide if they are sorry (with the news) not or whether this is a bad news, are not the doctors but the parents. We need more positive information in the doctors' room, hospitals etc showing kids with Down syndrome can have fulfilling and productive lives.' - #47 Baby born 2016

'I didn't even realise what the nuchal translucency screen was screening for. This information should be given right upfront. Needless to say the high risk came out of the blue. Upon giving results, there definitely needs to be more support, or at least a source of truth to be directed to. Termination should never be assumed!!! Ever!!! Frankly it should be up to the parent to request, not the doctor to offer. This devastates me, I wonder how many more parents make a fear based decision.' - #49 Baby born 2012

'Good pregnancy care is essential in not altering a person's decisions at all. All up to date facts should be given, so an informed decision can be made by the pregnant person/ couple. Supportive doctors and nurses should then respect the decisions that have been made, and they should read the file, instead of being insensitive, and keeping on asking questions, like if a termination is required.' - #51 Baby born 2005

'Eliminate the word 'risk' from any mention of tests or results. Risk is a negative word that is charged with connotations of a 'less than' child or a situation that will result in regret. Even if further invasive testing is declined, literature must be provided to expecting parents about Down syndrome. I ended up in denial that anything could happen to me which made the birth diagnosis incredibly difficult to handle.' - #52 Baby born 2014

'More neutral language needs to be used. Just because a doctor may see DS as a negative (which they shouldn't!!) doesn't mean the parents do too. Don't start by saying 'I'm sorry' as straight away it puts negative thoughts into a parents head! Give more information about Down syndrome and put people in contact with relevant groups/associations or other parents that have first-hand experience with Down syndrome.' - #53 Baby born 2011

'I think it all comes down to how the diagnosis is delivered and eliminating the "I'm sorry", being given your options rather than their opinions of "what is best" and being referred to Down syndrome groups along with up to date information that shows all of the positives rather than focusing on the negatives. One of the things I clearly remember being told is "whatever decision is made we will support you".' - #56 Baby born 2013

Recommendations

This survey shows significant failings in the way that prenatal screening programs are being run and the way results from screening and diagnostic tests are being delivered. While some women provide accounts of positive experiences where they felt their decisions were respected and that they were supported by their medical teams, all too often women provide accounts characterised by a lack of respect for their choices, provision of little to no information or support and out-dated, ableist and very negative perceptions of Down syndrome. From the survey it is clear:

- Many women do not realise that screening tests are voluntary or what the nature and purpose of these tests are. Very few women were provided with any relevant up to date information on the conditions, such as Down syndrome, being tested for before they undertook screening.
- The language used to discuss screening and diagnostic test results is often very negative, sending clear messages to women that having a baby with Down syndrome is a negative outcome.
- Many women are placed under immense pressure to undertake invasive diagnostic testing and to terminate their pregnancies. Women are often asked frequently to revisit decisions relating to testing and termination even after they've clearly indicated they're not interested.
- Women are being offered terminations on the basis of screening results only without any confirmed diagnosis of Down syndrome.
- Few women are provided with any up-to-date information to inform their decisions around invasive diagnostic testing and termination. Information given is often sparse, extremely negative and at odds with the lived experience of people with Down syndrome. Frequently the only information given is a very biased and outdated view of the limited value and potential of a life lived with Down syndrome.
- In some cases the negative attitudes of medical staff towards babies born with Down syndrome appear to have affected the quality of care provided to the extent that the baby and woman were put at risk.
- There appears to be discomfort amongst many medical professionals in providing results, with some doctors unsure how to deliver the news and to lead a balanced discussion of the options available.

In order to address these issues the following recommendations are made.

A Framework for Prenatal Clinical Care

Currently there is no standardized approach to the implementation of Australia's prenatal screening program. A Framework for Prenatal Clinical Care which provides for an informed consent process to be applied through the prenatal screening and testing process should be developed. This needs to involve:

- Provision of up-to-date resources to women before they undertake screening to inform their decision to screen. This should be supported by a Charter of Prenatal Care Rights (discussed below) which will make clear early on in the screening process women's rights to an informed choice and expectations they should see met through their prenatal care process.
- A 'sign to screen' approach so women explicitly sign to give informed consent to undertake screening, ensuring they have been provided with the appropriate resources and are aware of their rights.
- A clear pathway for practitioners to follow that guides them through the screening and testing process, indicating where and when women should be referred to for support to help inform and support their decisions.
- Clear standards around the use of neutral language in discussing screening and results as well as options around continuing or terminating their pregnancy. The option of adoption or long term foster placement should also be discussed with women.
- Guidelines on follow up testing and provision of support for women who decide to continue their pregnancies. This could include referring women to State based support organisations or counselling services as well as referral to paediatricians or other medical specialists who may be involved in the care of their child post birth.
- Guidelines for the provision of appropriate emotional support for women who choose to terminate their pregnancies as well as for those considering adoption or long term foster placement.
- Development of simple, easy to understand resources describing the conditions being tested for, life outcomes for people with the condition, links to good quality online resources with further information and contacts for relevant support organisations which are provided to women before screening as well as after a high probability result is found.

Adequate training

Training should be provided to all medical professionals involved in the care of pregnant women on the delivery of challenging results (such as a Down syndrome diagnosis) as opposed to delivering 'bad news'. This training should provide examples of the language that should be used, information about the conditions being tested for, appropriate resources and support and experience with children and adults with Down syndrome and their families. Ideally a training module should be developed that includes face to face contact with a parent of a child with Down syndrome or an adult with Down syndrome who can talk with knowledge about the condition and its implications for the life of the person and family. Medical staff should be exposed to concepts of ableism and inclusion and encouraged to question their own beliefs about people with Down syndrome and other disabilities. Training should include a mix of parent/child talks or talks by adults with Down syndrome to medical staff and students and use of internet based resources, such as Karen Gaffney's TedX talk 'All lives matter' <https://www.youtube.com/watch?v=HwxjoBQdn0s>. Training resources should be developed in conjunction with the peak bodies representing relevant chromosomal and structural conditions being tested for (eg. Down Syndrome Australia, CleftPALS, HeartKids, Cystic Fibrosis Federation Australia). We suggest these could be modelled on resources already developed

by the National Down Syndrome Congress³ and a webinar already developed by Down Syndrome Australia for the Australian College of Midwives in 2015.

Standards and complaint processes

Women's experience of prenatal screening, diagnosis and care following identification of Down syndrome are variable, but results from this survey indicate there is a strong need for standards to be developed and adhered to around prenatal screening, diagnosis and pregnancy care. These standards need to cover aspects of informed consent, the use of unbiased language in delivery of results, quality information provision, emotional and social support for women whose babies are identified as likely to have Down syndrome or other chromosomal (or structural) conditions and demonstration of respect for women's decisions around screening, diagnostic testing and continuation of their pregnancy. A clear process for making complaints and having these addressed needs to be developed. Women need to be given information on their rights (discussed further below) and the complaints process.

Provision of resources and information to professional and patients

Medical professionals should be given easy access to a range of up to date resources (see the Suggested Resources at the end of this report for examples) on Down syndrome and the delivery of a pre or postnatal diagnosis. Women should be provided with access to these resources to inform their decisions to undertake screening, diagnostic tests and around the continuation of their pregnancy where Down syndrome or other conditions are diagnosed. Women should be presented with the option to continue their pregnancy and either keep and raise their baby, place it for adoption or long term foster care or terminate. Information about all of these options should be offered and women should be supported regardless of which of these options they choose.

Support

Women should be offered social and emotional support through their pregnancy regardless of whether they have a confirmed diagnosis (as opposed to a high likelihood of Down syndrome), whether they choose to continue their pregnancy or terminate. This support should include referral to professional counselling, referral to State based Support Associations and access to parents of children with Down syndrome or other identified conditions.

Respect for decisions

Women's decisions with respect to prenatal screening and continuation of their pregnancies should be respected. At each stage options should be provided in neutral language with the risks and uncertainties associated with tests clearly explained. It should never be assumed that the choice to screen implies the choice to undertake invasive diagnostic testing, nor should undertaking these tests be seen to imply a choice to terminate a pregnancy if results come back positive for Down syndrome or other chromosomal conditions. Once women have declined invasive tests or termination these should not be offered or mentioned again unless and until the woman indicates she is reconsidering her previous decision and wishes to reopen the discussion. Even then, no presumption should be made about the choice she should or will make. Termination should never be presented as the logical or normal choice following a prenatal diagnosis. Women should be free to make their own decisions without bias from their medical team. All discussions should be balanced and not use negative language or perpetuate outdated perceptions of Down syndrome or other disabilities.

³ <https://www.ndscenter.org/physiciansguide/>

Informed choice – A Charter of Rights

All pregnant women should be provided with a statement of their rights early on in their pregnancy. These rights should include:

- You have the right to choose whether to undertake prenatal screening for chromosomal and structural conditions. You must be provided with access to up to date resources on the conditions being tested for and any risks associated with tests. You can expect your care giver to have a nondirective conversation about the negative and positive aspects of undertaking tests and your decisions in light of your own belief and value systems.
- You have the right to choose whether or not to undertake diagnostic testing. No assumptions should be made that choosing to screen implies you wish to undertake invasive testing.
- You have the right to make informed choices about continuation of your pregnancy. No assumptions should be made about what your choice would be based on your decision to undertake testing. You should not be placed under time pressure to make a decision and no arrangements for termination should be made unless you direct your caregiver to make them.
- Your decisions will be respected. In all cases once you've made a decision, that decision will be considered final until and unless you tell your healthcare provider you've changed your mind.
- Your screening and diagnostic test results should be relayed using neutral, unbiased language. You have the right to expect that the personal opinions of your care givers should not affect the way in which they deliver their results. You have the right to choose how results are delivered to you (eg. over the phone, in person) and who is present (eg. partner or friend).
- Before making decisions to undertake screening tests, diagnostic testing or around the continuation of your pregnancy, you should be:
 - Provided with up to date and accurate information on the tests being undertaken, what the results mean and the level of uncertainty (including the likelihood of false positive and false negative results), the conditions being tested for and likely implications for your pregnancy and the life of your child.
 - Offered a referral to both a genetic counsellor to discuss the implications of your results as well as a counselling service that can help you work through your feelings about your test results and help you make an informed choice about the next steps. Whether you take up this offer is entirely up to you. You can choose to take it up at any point in your pregnancy or after the birth of your child.
 - Offered contact information for relevant support groups. You may opt for your care giver to contact these groups on your behalf. You have the right to request to meet other parents and children with the condition before making any decisions.
- You have the right to high quality pregnancy care regardless of any decisions you make with regard to screening, diagnostic testing and continuation of your pregnancy.
- If these rights are not respected you can make a complaint to [insert appropriate complaint contact].

Suggested Resources for Practitioners and Patients

These resources are provided for use by both medical practitioners and women considering undertaking screening, diagnostic testing or making decisions around continuation of their pregnancy following a diagnosis of Down syndrome.

Information resources

Down syndrome Australia website - <http://www.downsyndrome.org.au/>

Down Syndrome Diagnosis Network - <http://www.dsdiagnosisnetwork.org/>

Raising Kids Network - <http://raisingchildren.net.au/>

Lettercase - <http://lettercase.org/>

Discussion Down Syndrome – A Physician’s Guide - <https://www.ndscenter.org/physiciansguide/>

Support services

State based Down syndrome Associations – links can be accessed off the Down Syndrome Australia website

Facebook – Down syndrome pregnancy Australia, pregnancy support group
<https://www.facebook.com/groups/536675403136412/>

Video interviews and talks of people with Down syndrome

Karen Gaffney – All lives matter - <https://www.youtube.com/watch?v=HwxjoBQdn0s>

Dear Future Mom - <https://www.youtube.com/watch?v=Ju-q4OnBtNU>

BBC Three – interviews – “Things people with Down’s syndrome are tired of hearing”
<https://www.facebook.com/groups/536675403136412/>

Appendix 1. Survey Questions

1. In what year was your child born?
 2. In what State was your child born?
 3. In what setting was your pregnancy care delivered?
 - a. Capital city
 - b. Regional city
 - c. Town
 - d. Rural or remote area
 4. Who managed your pregnancy care?
 - a. Obstetrician
 - b. Team midwives
 - c. GP
 - d. Shared care
 5. Was your pregnancy managed through the public or private health system? Public/Private
 6. Which prenatal screening tests did you have?
 - a. Routine first trimester screen (ultrasound and blood test)
 - b. NIPS (non invasive blood screen)
 - c. Morphology scan
 - d. I opted out of screening
 7. Did you have diagnostic testing in your pregnancy?
 - a. Amnio
 - b. CVS
 - c. I opted out of all screening and testing
 8. How far along in your pregnancy were you when it was first suggested your baby could have DS?
 9. How far along in your pregnancy were you when DS was confirmed? Enter 0 if it was never confirmed/you declined diagnostic testing
 10. Were you offered any up to date information on Down syndrome or other conditions being tested for:
 - a. Before undertaking screening tests?
 - b. After a high 'risk' screening result to help you decide whether or not to have invasive diagnostic tests (eg. Amnio or CVS)?
 - c. After confirmation of Down syndrome through a diagnostic test (eg. amnio, CVS)?
- If yes, what information were you provided with?
11. Did your care giver offer to link you to any support groups or parents? Yes/No If so, who?
 12. Were you offered a termination? (select all relevant)
 - a. Yes - before undertaking invasive diagnostic testing such as amnio or CVS to confirm the DS diagnosis

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- b. Yes - after confirmation of DS from an invasive test such as amnio or CVS
 - c. No
13. Were you given a time limit within which to make a decision about continuing your pregnancy?
- a. Yes – a week or less
 - b. Yes – one to two weeks
 - c. Yes – over two weeks
 - d. No
14. If you declined a termination, were you offered termination again after your first refusal?
- a. Yes – once only
 - b. Yes – a few times
 - c. Yes – frequently
 - d. No
15. How far along in your pregnancy were you when you were last offered a termination?
16. Were you offered the option of ‘comfort care’ for your child – ie. keeping them comfortable but making no active efforts to keep them alive after their birth?
17. Overall, how would you rate the delivery of your screening and/or diagnostic test results? (very poor, poor, OK, good, very good). Why?
18. Which would best describe the language of your medical team when discussing the possibility of your baby having DS? Negative – clearly indicated that DS was a bad outcome (eg. ‘I’m sorry’, ‘bad news’), Neutral – no indication either way as to the personal view of medical team as to whether the baby having DS was a good or bad thing, Positive – language indicated that news was a happy/good event. Comment.
19. Do you feel your decisions around prenatal testing and continuing your pregnancy were respected by your medical team? Yes/No/Not sure. Why?
20. Do you feel your test results affected the quality of care provided by your medical team through the remainder of your pregnancy? Yes – care was better, Yes – care was worse, No. Please explain how and why you think it was affected.
21. Please describe your experience in terms of screening, diagnostic testing, delivery of results and ongoing care.
22. How did this experience affect your feelings about your pregnancy and your baby?

23. What do you think good practice care looks like for discussion prenatal testing, delivering test results and ongoing care for pregnancies with either a DS diagnosis or identification of a high chance of DS?
24. Do you have anything else you would like to add?