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What is prenatal screening and what information can it provide families?

Prenatal screening describes the medical tests that are offered to all pregnant women/couples, and which usually take place towards the end of the first trimester of pregnancy.

The information that these tests provide includes the risk that the child to be born will have a chromosomal abnormality, such as Down syndrome (3 copies of chromosome 21), Edward’s syndrome (3 copies of chromosome 18) or Patau syndrome (3 copies of chromosome 13).

The results of the screening are usually reported as a probability, e.g. a 1 in 600 risk of one of these conditions. As the screening involves an ultrasound and maternal blood test that measures several biochemical markers, it is also possible that other problems with the viability of the pregnancy or health of the fetus may be detected, as may other risk factors for the pregnancy’s progression.

Based on the information received as a result of screening and other knowledge gained from reading information brochures or from pre-screen discussions, the pregnant woman/couple can then decide whether they would like to have further testing that can provide a more definitive result than a probability.

This is called prenatal diagnostic testing and currently takes place by way of chorionic villus sampling or amniocentesis, procedures which are described as ‘invasive’ as they require a needle to be inserted into the uterus to obtain a sample of fetal material.

Like screening, prenatal diagnostic testing cannot detect all conditions that a fetus may be developing.

What are the benefits for families of using prenatal screening?

The ‘benefit’ of prenatal screening is something that can be hard to grasp, and the answer to this question may depend on one’s personal or professional background. For families, the benefits of prenatal screening can be summarised as an opportunity to find out whether their fetus (some may prefer to call it their ‘baby’) is at greater risk of being born with one of the conditions described above.

This can then allow them to make decisions based on the information they have received. The actions resulting from this decision-making can be to do nothing, or to have further tests (with a new test now on the market – see below) to provide a more definitive result.

Based on further tests, couples may choose to use the information in their planning to have a child with a chromosomal trisomy, or to end the pregnancy.

For me, as an academic working on the ethical aspects of this technology, I see access to information as an important part of the experience of pregnancy. But just as the ability to access this information is important, so too is the option to decline the offer of this information. Some couples may have no intention to terminate a pregnancy on any grounds, or may have other reasons for not wanting prenatal screening.

While all families should be informed about it, the choice of whether or not to take up the offer of prenatal screening should be left to the woman or couple. Making these decisions is, however, not always easy and so the information given to women/couples needs to be balanced in terms of the test and the conditions being screened for.

Access to pre- and post-test counselling should therefore be an aim of all prenatal screening services.

Are there any potential harms associated with prenatal screening from a professional/ethical view point?

Prenatal screening is undoubtedly a sensitive topic. Two issues stand out for me in terms of potential harm. First, harm may occur if pregnant women/couples are not given the intellectual or temporal ‘space’ to make a well-considered decision. I talk about this further below.

Second, harm may occur if those living with the condition being tested for end up with less resources or recognition, such as meeting any specific education needs without families having to fund them from their own pocket. There is a long history of debate over the so-called ‘disability rights critique’ of prenatal screening and testing. Like ethical debates over the moral value of the fetus, this issue is somewhat intractable, as it depends on what value we ascribe to people living with different abilities and whether the very offer of prenatal screening is implying a devaluing of those who live with the condition being screened for.

It’s also interesting to note that for someone like me who works in bioethics, questions of harms like these are important – but they are also difficult because they rely on empirical evidence to inform the ethical aspects. So we also try to ask and answer questions, or work on issues, that don’t depend on these kinds of data.

For example, many in my field work on questions like these: “does prenatal testing inherently (as opposed to practically) devalue those living with the conditions being tested for?” and “if new methods of prenatal testing increase rates of termination, how should we respond?” – I don’t have the answers to those questions yet, but the work I’m doing is broadly informed by these kinds of overarching issues.
Some say that because prenatal screening is so routine and available, many people feel like they are expected to use it. Do families have a choice in undergoing screening?

Families do, and should continue to have a choice in whether they undergo prenatal screening. National standards specify that all women should be ‘offered’ screening, not that they should have it. Anecdotally, you do hear of some women saying that prenatal screening is something that was ‘recommended’ to them. It should definitely be ‘offered’, but should be a free and considered decision.

There is also a biotechnological critique of the ‘routineisation’ of prenatal screening, which looks at lots of different aspects of this process – from the offer of the test to what happens after the information is received. The ‘routine’ aspect of the test can be both beneficial and problematic. It’s beneficial because health providers have worked hard to ensure all women should have access to this testing. But it can also be problematic in that its routine nature can mean that its possible significance gets lost in the array of other decisions women make in pregnancy.

In some of my work, I have reflected on the importance of giving pregnant women/couples as much time as possible (within the necessary constraints of being pregnant) to reflect on the decision they are making. While we hope that most women are making a free and considered choice about prenatal screening, more theoretical and empirical research is definitely needed.

What do you think is the future of prenatal screening and testing in Australia?

The future of prenatal screening and diagnostic testing is currently at a turning point. A relatively new technology called non-invasive prenatal testing (NIPT) has now begun to be offered, mostly to pregnant women who are receiving their care in the private sector. This technology involves sampling fetal DNA fragments that can be found in the maternal bloodstream. Technical and other limitations to testing are still being ironed out, but initial data are suggesting that this technology will end up being an excellent screening tool for the presence of chromosome anomalies in a fetus. The test is low-risk in terms of harm to the fetus as it just involves a maternal blood test, like the existing screening tests. But, (NIPT) is much better at defining the risk of the fetus being affected by a chromosome abnormality than current screening tests are.

Indeed, it is almost as good as current invasive diagnostic tests, but it is not quite 100%. The question then becomes whether all women should be offered NIPT and when in pregnancy. Some of my recent academic papers have pondered these questions and I and my co-authors have concluded (so far!) that this technology should be offered to women, but that it should come as an ‘intermediate step’ between prenatal screening and invasive testing, to preserve women’s option to receive a probability-based result first and then give definitive information too early in pregnancy.

Initial estimates suggest that uptake of NIPT where it is offered is very high. NIPT is a transformative technology. As well as testing for trisomy conditions, it can also be used to predict gender, some genetic conditions caused by changes in single genes, and ultimately to obtain the sequence of a whole fetal genome (although this latter possibility is still technologically challenging and not available in a clinical context).

The ethical question is whether we should be using this technology for these kinds of applications, and if so, when and by whom.

For further information on screening and testing see: www.wcgs.org.au

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Down Syndrome Victoria

While the technology around screening for Down syndrome is moving at a phenomenal rate, the accuracy and quality of the information given to parents at the time of their diagnosis is not. It is very important that parents are made aware of the increased chance of certain medical and developmental conditions that can come with Down syndrome. The problem doesn’t come from giving this information, the problem is when this is the only information parents receive.

Unfortunately it is common for any positive or helpful information about Down syndrome to be withheld from the family until they have decided to proceed with the pregnancy. Health professionals see it as irresponsible to discuss the positives for parents in case it doesn’t happen for them. Down Syndrome Victoria believe that if it is prudent to inform parents of all the challenges they may face, whether it applies to them or not, it should be equally prudent to inform parents of the help and support that is available to them. They should go hand in hand, not handed out separately.

Parents need to know that health advancements, early intervention and inclusion in mainstream schools and work environments means that living with Down syndrome today is vastly different from the outdated stereotypes of the past. They need to know that many parents speak of the joy and happiness in their lives since welcoming a child with Down syndrome. This information is only useful to parents before they have made the decision to proceed, not after.

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