NIPT Fact Sheet

NIPT and prenatal screening: Understanding fact and fiction

NIPT stands for non-invasive prenatal testing. It involves a blood test taken from a pregnant woman, at least 10 weeks into the pregnancy. It looks at fragments of DNA found in maternal blood. A woman's blood contains a mixture of her own DNA and DNA released from the placenta as the baby grows and develops. Therefore, NIPT screens for fragments of DNA originating from the placenta. Sometimes this can differ from the DNA in the baby. NIPT does not look for all possible chromosome differences.

Down Syndrome Australia

Do I have to have NIPT or another screening test done?

No. When pregnant, you may be offered a screening test to see whether your baby shows signs of having a chromosomal/genetic difference (including Down syndrome). There are different types of screening tests such as a blood test, ultrasound or both. Some people are considered more likely to have a baby with Down syndrome (e.g. mothers over 35 years old or another test has already shown a higher chance) but it is important to know that babies with Down syndrome can be born to anyone.

It is your choice whether you wish to have these tests performed; some people like to be aware of any potential conditions their baby may have so they can explore all their options or feel prepared, but others do not. Some of these tests also involve costs (currently, NIPT does not have a Medicare rebate).

There is no right or wrong choice about prenatal screening tests and it is important that you do not feel pressured into having them if you do not wish to. You can learn more about screening tests on our Resource Hub or by watching this short video from the Genetic Support Foundation:

https://youtu.be/-vIJGFWJquk

Are screening tests accurate?

Screening tests are simply that – screening tools; which means they will estimate the chance (possibility) that the baby has a chromosome difference such as Down syndrome. It does not provide a definite diagnosis. NIPT is the most accurate screening test available for chromosome differences such as Down syndrome. All screening tests can give a high chance result when the baby does not have a chromosome difference. Therefore, it is suggested that a woman talk to her doctor, midwife or a genetic counsellor about the options available to confirm the results by diagnostic testing. For more detail about genetic counselling, see the section below 'What support is available?'.

Understanding the results of your screening test – Positive Predictive Value

If you do have a screening test during pregnancy, you are given the probability of whether your baby may have a condition such as Down syndrome. This may be explained to you as a high or low chance. Being told there is a high chance does not mean that the baby will have a chromosome difference; just as being told that there is a low chance does not mean the baby will not have a chromosome difference. The possibility of a 'false positive' or a 'false negative' exists.

In combination with your high or low chance screening result, you may be given a 'positive predictive value' – which 'represents the proportion of positive test results that are truly positive'. PPV varies for every woman, as it considers things such as your age, weight and how common Down syndrome is in the general population. For example, the PPV for younger women is generally lower based on some of the above factors, so if a younger woman is given a high chance screening result, there can be more incidences of this being a false positive. For example, if a woman's PPV is given to her as 60%, it means that in 40% of cases, the baby is found (either at birth; or with subsequent diagnostic testing) not to have a chromosome difference.

If you have already had NIPT testing done, you can work out your PPV at the following website:

https://www.perinatalquality.org/Vendors/NSGC/NIPT/

Does doing NIPT mean I don't need to have a chorionic villus sampling (CVS) or amniocentesis?

Diagnostic testing such as CVS or an amniocentesis is the only way to tell for certain whether your baby has a chromosome difference.

If you receive a high chance screening result you will be offered diagnostic testing to confirm the result. Your health care provider will review with you the options for diagnostic testing and the advantages and disadvantages of each test. It is your choice as to whether you would like to proceed with invasive diagnostic testing.

Does doing NIPT mean I don't need to have a 20-week morphology ultrasound scan?

Your GP or obstetrician is likely to encourage you to have a 20 week scan, as it is the best way to assess your baby's physical development (e.g. structural conditions such as heart conditions; bone growth issues; other concerns related to organs or structures of the body) and check the condition and position of the placenta. A blood test will not pick up any of these. However, it is your own choice as to whether you want to know such information. You should discuss the options and advantages and disadvantages of the morphology ultrasound scan with your GP or obstetrician.

What support is available?

You may be offered genetic counselling if scans/screening tests have shown signs that your unborn baby may have Down syndrome or another screened-for chromosome condition; or if a diagnostic test has confirmed this. It is a time when expectant parents are offered different choices (e.g. undergo further testing; continue the pregnancy and keep the baby; continue the pregnancy and consider alternative care; or consider termination of pregnancy). It can be an overwhelming and emotionally difficult time.

Genetic counselling aims to inform and support parents throughout the process of prenatal testing, through the provision of information and education about all testing options available and the risks involved in order to facilitate informed decision making. A genetic counsellor provides non-directive, non-judgmental and non-discriminatory counselling and their role is to support and respect the parents' personal values and decisions.

Most laboratories offering NIPT in Australia have genetic counselling services available for women to access. Talk to your doctor about this option should your result show a higher chance of a chromosome difference.

Prospective parents require up-to-date, evidence-based information about life with Down syndrome and other chromosome differences. All children including those with Down syndrome have strengths and weaknesses that cannot be predicted before birth. In general, people with Down syndrome have mild to moderate intellectual disabilities, low muscle tone and a higher chance of some health issues, particularly heart conditions. However, those living with Down syndrome and their families know life with an extra chromosome to be more than a list of symptoms. A genetic counsellor will be able to refer prospective parents to their state Down Syndrome Association. Speaking to people with Down syndrome and their families can help gain insight into everyday life with an extra twenty-first chromosome.

If you are seeking a genetic counsellor in your area, a list of possible options can be found here:

https://www.hgsa.org.au/asgc/find-a-genetic-counsellor

Your local Down syndrome association can offer non-directive information and support. Call or email to get in touch with one of the team. Your association can help with questions you may have, advise you on the options and services available to you, and assist you to navigate your next steps. We respect the right of all families to make decisions appropriate to you, your beliefs and circumstances.

Call **1300 881 935** or find your local State/Territory details here:

https://www.downsyndrome.org.au/about-us/contact-us/

Related resources

- https://ranzcog.edu.au/wp-content/uploads/2022/06/Prenatal-Screening-for-Chromosomal-and-Genetic-Conditions.pdf
- https://www.genetics.edu.au/publications-and-resources/booklets-and-pamphlets/prenatal-testing-special-tests-for-your-baby-during-pregnancy
- https://www.genetics.edu.au/publications-and-resources/booklets-and-pamphlets/safda-impact-options-afterwards
- https://www.genetics.edu.au/publications-and-resources/booklets-and-pamphlets/individuals-and-families-pregnancy-resources
- https://kidshealth.org/en/parents/genetic-counseling.html#:~:text=The%20counselor%20can%20help%20her,to%20screen%20for%20Down%20syndrome.
- https://www.pregnancybirthbaby.org.au/genetic-counselling
- https://www.dsrf-uk.org/nipt-facts/
- https://fetalmedicine.org/nuchal-translucency-scan?fbclid=IwAR3eRN1URefPQB0dZCCxIvIX_XDPa6lnrsSdnkT5vZJgZK0PVpMNtsMc-KM
- https://www.perinatalquality.org/Vendors/NSGC/NIPT/
- https://www.mcri.edu.au/images/documents/migrate/prenatal-screening-decision-aid.pdf

Pamphlet: Prenatal Testing and Information about Down Syndrome. 2017. Produced in conjunction by Global Down Syndrome Foundation and National Down Syndrome Congress, USA.

https://www.globaldownsyndrome.org/prenatal-testing-pamphlet/

Prenatal testing for Down syndrome – Fact Sheet.
Down Syndrome Association of Australia.

https://www.downsyndrome.org.au/blog/resource/prenatal-testing-for-down-syndrome-fact-sheet/

Genetic counselling and prenatal testing. (2013) Dr Jan Hodgson and Dr Melody Menezes. Voice magazine, April 2013 issue, Down Syndrome Victoria and Down Syndrome NSW Members' Journal.

Actor Julia Hales hosts *The Upside*, an ABC documentary available on iView. Julia, who has Down syndrome, speaks to a range of professionals and families about non-invasive prenatal screening.

https://iview.abc.net.au/show/upside

The podcast *One screened every minute* (**https://onescreenedeveryminute.com/**) includes stories from parents who have received NIPT screening results showing a high chance for Down syndrome and other chromosomal differences.

Disclaimer

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