Down's Syndrome Association
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Fact Checking

Non-Invasive Prenatal Testing (NIPT) for Down's syndrome



Publication date: 31 January 2020, Author: Colette Lloyd

This fact sheet clarifies facts about NIPT for Down's syndrome:

- **That NIPT uses placental DNA**
- **That NIPT is not diagnostic**
- **The importance of your "positive predictive value"**
- **What to do next**

Does NIPT test the baby's DNA?

No.

Many advertisements and even leaflets designed for use in studies around NIPT, claim that the DNA analysed during NIPT is from the baby. It is actually from the placenta. This is one of the reasons NIPT can be wrong, as sometimes, the DNA from the placenta, differs from that of the baby.[1]

Does NIPT mean that you can avoid an amniocentesis/Chorionic villus sampling (CVS)?

No, if you want to know for sure if your baby has Down's syndrome.

NIPT is a screening test, so similarly to the combined or quadruple test, it will only give a low or high chance of your baby having Down's syndrome. Amniocentesis or CVS are diagnostic tests, so they tell you if the baby does or does not have Down's syndrome. NIPT has false negatives and false positives. A recent study in the Netherlands,[2] identified 237 babies as having Down's syndrome. Of these, 227 were confirmed to actually have Down's syndrome, (so there were 10 false positives). 6 babies who turned out to have Down's syndrome had been incorrectly identified as not having Down's syndrome (false negatives).

Is NIPT a safer test?

No. If you compare NIPT with other screening tests, that give you a low/high chance, then it is simply "as safe as" other tests.

If you compare it with a diagnostic test, then yes, it is safer, but that would be like comparing apples with oranges. NIPT and diagnostics tests do not give you the same information, therefore are not comparable, and are unlikely ever to be so.[3]

Is NIPT always 99% accurate?

No.[4]

NIPT has a positive predictive value (PPV, the likelihood that a high chance result is correct) of about 46% if you are in your 20's. If you already have a 1:150 chance that the baby has Down's syndrome, then the PPV is 87%. If all these numbers confuse you, please just ask your health care provider for your "positive predictive value".

Should CVS be used to confirm a NIPT result?

CVS uses cells from the placenta, so the result from CVS can be wrong for one of the same reasons NIPT can be wrong, placental mosaicism.

So, ideally, you should not have CVS to confirm a NIPT result; you should instead wait for an amniocentesis. If you do choose CVS, you may still need an amniocentesis to be sure.[6] If you are a medical professional reading this, please make sure that a parent understands this. Sadly, not everyone does.[7]

Further questions?

If you have any further questions about screening, please don't hesitate to contact the Down's Syndrome Association on 0333 1212 300 or ask for a further discussion with your midwife.

If you would like a video to guide you through decisions around prenatal testing see the Genetic Support Foundation film *How to Decide About Prenatal Genetic Testing*: https://youtu.be/-vIJGFWJquk

If you would like further information about Down's syndrome please contact either the Down's Syndrome Association www.downs-syndrome.org.uk or Positive About Down Syndrome www.positiveaboutdownyndrome.co.uk

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References

1. Lestou, Kalousek. (1998). Confined placental mosaicism and intrauterine fetal growth. Arch Child Fetal Neonatal Ed 79:F223–6
2. Karow, (2018) ASHG: Dutch Nationwide NIPT Implementation Study Sees 42 Percent Uptake After First Year. Genome Web. Available at https://www.genomeweb.com/molecular-diagnostics/ashg-dutch-nationwide-nipt-implementation-study-sees-42-percent-uptake-after?fbclid=IwAR0mqcDAc3Oblcs-eetDv8X1Hle5BTbl09SSraxawPeo5kJG80k123dikpA#.W8-iFBbTWEc (accessed on 20th November 2019)
3. Stanislav Birko, Lemoine,Thu Nguyen, Ravitsky. (2018) Moving Towards Routine Non-Invasive Prenatal Testing (NIPT): Challenges Related to Women's Autonomy. OBM Genetics. Volume 2, issue 2 doi:10.21926/obm.genet.1802018 (4.2)
4. ITV (2019) Ads for three prenatal genetic test firms banned for using misleading statistics. Available at https://www.itv.com/news/2019-11-20/ads-for-three-prenatal-genetic-test-firms-banned-for-using-misleading-statistics/?fbclid=IwAR0t2mnXjnYxiGI91a7uxxvnG9MyEKWt5WYRlzhpQ2xp0RkrH3Hr-oBTVJA (accessed 21 November 2019)
5. Perinatal Quality.org (n.d.) NIPT/Cell Free DNA Screening Predictive Value Calculator. Available at https://www.perinatalquality.org/Vendors/NSGC/NIPT/ (accessed 20 Nov 2019)
6. Grati, F. R., Bajaj, K., Malvestiti, F., Agrati, C., Grimi, B., Malvestiti, B., Pompilii, E., Maggi, F., Gross, S., Simoni, G., and Ferreira, J. C. P. (2015) The type of feto - placental aneuploidy detected by cfDNA testing may influence the choice of confirmatory diagnostic procedure. Prenat Diagn, 35: 994– 998. doi: 10.1002/pd.4659. (accessed 4 January 2020)
7. McCrave C. (2019) The Journal.ie. Couple at centre of Holles Street abortion case 'utterly, utterly devastated. https://jrnl.ie/4641201 (accessed 21 May 2019)
