DOUGLASS HANLY MOIR PATHOLOGY

Information for Doctors

# Non-invasive prenatal test (NIPT)

Non-invasive prenatal testing (NIPT) screens for the presence of specific chromosome disorders in the developing fetus. The test analyses fragments of DNA in maternal plasma that have been released from both maternal and feto-placental cells.

By analysing the proportions of DNA fragments derived from different chromosomes or chromosome regions, NIPT can screen for the presence or absence of specific chromosome disorders.

NIPT is more accurate than first trimester maternal serum screening and ultrasound in identifying pregnancies with, or without, these disorders.

## Accuracy of NIPT

NIPT provides fewer false-positive and false-negative results than combined first trimester screening for trisomy 21, 18 and 13.

| Accuracy (T21, T18, T13) | Sensitivity* | False-positive rate# |
|---|---|---|
| Combined first trimester screening | 82% | 1 in 26 |
| NIPT | >99% | <1 in 1,000 |

*Proportion fetuses with trisomy correctly identified by the test as high probability of disorder
#Proportion of normal fetuses incorrectly identified by the test as high probability of disorder

It is important to note that NIPT is a screening test and does not provide a definitive genetic diagnosis, as NIPT cannot differentiate potential chromosome differences between the placenta and fetus. A definitive genetic diagnosis of the fetus requires cytogenetic analysis of either amniotic fluid or chorionic villus sampling (CVS).

## Targeted screening for specific common chromosome disorders

- **Trisomy 21 (Down syndrome)** is associated with moderate to severe intellectual disability, congenital heart defects and other malformations.
- **Trisomy 18 (Edwards syndrome)** and **trisomy 13 (Patau syndrome)** are associated with severe brain and cardiac malformations. There is a high risk of stillbirth or death during infancy.
- **Sex chromosome aneuploidy** (abnormalities in the number of X or Y chromosomes) can be associated with malformations and infertility e.g. Turner syndrome (45,X) and Klinefelter syndrome (47,XXY). This screen is optional (no additional cost).

In addition, NIPT can also assess fetal sex. This is optional (no additional cost).

NIPT does not screen for non-chromosome disorders, familial mutations, malformations, fetal growth or fetal viability.

## When to perform NIPT

NIPT should not be performed before a gestational age of 10 weeks. However, it is suitable at any time after that, preferably while there is sufficient time for further investigation or decision-making (should this be required). We recommend an ultrasound scan prior to NIPT to confirm dates and fetal viability, and to check for twins. Performing first trimester screening before NIPT may provide supplementary information regarding the status of the fetus.

Information for Doctors

Testing is performed in Australia at one of our NATA-accredited laboratories.

# Non-invasive prenatal test (NIPT)

## Reporting results

Sonic Genetics first checks that there is sufficient feto-placental DNA in the maternal sample and quality data to provide an accurate assessment. A re-collection may be recommended if the sample is not suitable or an assessment may not be feasible.

The report then summarises the screening assessment for each disorder specified by the requesting doctor (see example in table below).

| Chromosome | Result | Recommendation |
|---|---|---|
| **Trisomy 21** | **High probability** | **Genetic counselling and additional testing** |
| Trisomy 18 | Low probability | Review result with patient |
| Trisomy 13 | Low probability | Review result with patient |
| Sex chromosome aneuploidy | Not requested | — |
| Fetal sex | Male | Review result with patient |

A high probability NIPT result should always be confirmed by amniocentesis or CVS before making any decision regarding subsequent management of the pregnancy. **Please contact one of our genetic pathologists on 1800 010 447 should you wish to discuss your patient's result. We also provide free genetic counselling for patients with such results (terms and conditions apply).**

Sonic Genetics provides confirmatory cytogenetic testing (Rapid FISH and karyotyping) on amniocentesis and CVS samples with no out-of-pocket cost to eligible patients who have had NIPT with Sonic Genetics.

## If an assessment cannot be provided

On rare occasions, NIPT is unable to provide an assessment of the probability of specific chromosome disorders. This usually reflects the complex biology of genetics and pregnancy, and is not due to a failing in the laboratory.

If NIPT cannot provide a specific assessment, it is not worth repeating the NIPT (unless advised by the laboratory). A decision about other tests (maternal serum screening, detailed ultrasound, amniocentesis or CVS) should be based on the doctor's assessment of all risk factors identified, and may require specialist consultation.

The primary purpose of our NIPT is to screen for the common trisomies of chromosomes 21, 18 and 13; the sex chromosomes can be analysed at no additional cost. If NIPT cannot provide an assessment for these trisomies, the patient can apply for a full refund. If NIPT cannot provide an assessment for the sex chromosomes, a refund is not available.

## Arranging a test

1. Complete a Non-invasive prenatal test request form. This form is available electronically via a number of practice management systems or can be downloaded from the Sonic Genetics website. The test can be requested from 10 weeks' completed gestation.

2. Patients need to prepay and book a collection time online via sonicgenetics.com.au/nipt or by calling 1800 010 447.

3. Your patient can have a blood sample taken at select Douglass Hanly Moir Pathology collection centres.

4. Results are provided in 3–8 business days of the laboratory receiving the sample, and are available electronically via Sonic Dx or downloaded to your practice management system; fax and hard copy reports are also available.

## Cost

Medicare and health insurance do not cover the cost of NIPT.* Please refer to the Sonic Genetics website for current pricing.

**For further information regarding our NIPT service, please visit the Sonic Genetics website, sonicgenetics.com.au/dr/nipt**

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