Sonic Genetics

Service available through Sonic Healthcare practices:

Douglass Hanly Moir Pathology
Sullivan Nicolaides Pathology
Melbourne Pathology
Barratt & Smith Pathology
Capital Pathology
Clinipath Pathology
Bunbury Pathology
Clinpath Pathology
Hobart Pathology
Launceston Pathology
North West Pathology
Southern.IML Pathology

INSTRUCTIONS FOR THE PATIENT

To finalise the booking and payment for your NIPT, please visit **sonicgenetics.com.au/bookandpay**

All enquiries, please contact 1800 010 447 (Monday–Friday, 8 am–6 pm AEST).

Non-invasive prenatal test (NIPT) | Request form

FOR THE DOCTOR

This test should be requested by the doctor responsible for medical management of a patient's non-invasive prenatal testing.

Patient details

First name _____

Surname _____

Date of birth ____/____/____ Sex _____ **Female – Pregnant**

Address _____

_____

_____

Phone (mobile) _____

Test(s) requested

NIPT for: Trisomy 21, 18, 13 ☑ Yes

OPTIONS (no charge)

Fetal sex* ☐ Yes

Sex chromosome aneuploidy^ (singleton only) ☐ Yes

*Based on the presence or absence of the Y chromosome. For twin pregnancies this could indicate either two females (if absent) or at least one male.
^If sex chromosome aneuploidy is detected, the fetal sex will be revealed.

OPTIONAL SPECIALISED TESTING (additional charge)

Genome-wide NIPT# ☐ Yes

#The screening of autosomal aneuploidies, including gains and losses >7Mb. This option must be selected by the requesting doctor prior to sample collection. This option includes screening for sex chromosome aneuploidy in singleton pregnancies. See overleaf for information before ordering.

Is this a ☐ **RE-COLLECTION?** Previous Lab ID _____

Staff ID/Location	☐ 1 x NIPT tube	Date re-collected / /	Time re-collected :	Re-collect PAY CAT **SGUN**

Clinical information REQUIRED

This section must be completed for testing to proceed.

Please note: The requested clinical information is essential for test accuracy. If any of the clinical information you provide below needs updating, please notify the laboratory immediately.

NUMBER OF FETUSES

(assumed singleton, unless otherwise indicated)

☐ Twin pregnancy

GESTATIONAL INFORMATION

☐ LMP ____/____/____ (date) *or* ☐ EDC ____/____/____ (date)

The presence of any of the following invalidates the NIPT result; an alternative test should be considered.

- Taken at less than 10 weeks' gestation
- There are three or more fetuses
- There is known presence of a demised fetus
- There is known presence of maternal aneuploidy, maternal transplant or maternal malignancy

NIPT is not a test of fetal viability.

Requesting doctor

Name _____

Address _____

_____

_____

Phone _____ Provider No. _____

I confirm that this patient has been counselled about the purpose, scope and limitations of the test and has given consent.

DOCTOR SIGNATURE

X _____ Date

Copy reports to

Name _____

Address _____

_____

_____

FOR THE PATIENT – Patient and Financial Consent

I consent to the non-invasive prenatal test (NIPT) being performed and confirm that I have been advised about the purpose, scope and limitations of the test. I understand that I can request further information or genetic counselling before or after the test. I understand that NIPT is primarily a screen for an extra copy of chromosomes 21, 18 and 13, and can potentially examine other chromosomes as requested by my doctor on this form.

I understand that the result of this test should be interpreted by my doctor in conjunction with other clinical information and tests, and that it should not be the sole basis for making a decision about my pregnancy. I understand that a second blood collection may be required, that a small percentage of tests do not yield a result due to biological factors, and that I can seek a refund if there is no result for chromosomes 21, 18 and 13. A refund is not available if there is no result for sex chromosome abnormalities/fetal sex/other chromosomes.

☐ I do not agree to the laboratory contacting my treating doctors to obtain information and results regarding this pregnancy for quality assurance purposes.

PATIENT SIGNATURE

X _____ Date

Full payment is required prior to sample collection. Medicare benefits do not apply.
Following payment, you will receive an email and SMS confirmation of your booking.
Please make sure to bring this request form and booking confirmation with you on the day.
To locate a collection centre for your NIPT, please visit sonicgenetics.com.au/locations

FOR THE COLLECTOR

I certify that I established the identity of the patient named on this request, collected and immediately labelled the accompanying specimen(s) with the patient's name, DOB and date/time of collection.

Collector initials	☐ 1 x NIPT tube	Patient initials
Location code	Date collected / /	PAY CAT **SGU**
Collection type	Time collected :	

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Sonic Genetics

Non-invasive prenatal test (NIPT)

Information for patients

Purpose

The primary purpose of NIPT is to screen for common chromosome conditions that can affect the health of a baby, that is, Down syndrome (trisomy 21), Edwards syndrome (trisomy 18) and Patau syndrome (trisomy 13).

If requested by your doctor, NIPT can also screen for common conditions of the sex chromosomes, that is, Turner syndrome (45,X), Klinefelter syndrome (47,XXY), XXX and XYY in singleton pregnancies. NIPT is not primarily intended as a test of fetal sex.

NIPT can also screen for the gain or loss of other chromosomes or large chromosome regions, that is, genome-wide NIPT. The clinical implications and performance metrics for this option are different to those for the common autosomal trisomies and these should be considered and discussed before having this test. Please refer to sonicgenetics.com.au for further details.

Fetal fraction

The mother's bloodstream contains DNA fragments from both the mother and the developing fetus/placenta. The proportion of DNA fragments from the fetus/placenta is known as the fetal fraction. Numerous factors, including a gestation of less than 10 weeks and high maternal weight, can lower the fetal fraction. If the fetal fraction is too low the NIPT cannot provide a reliable result. Sonic Genetics will not provide a result unless the required quality thresholds are met. In some situations, we may offer to repeat the test (once, and for free) if there is a reasonable chance of providing a result for chromosomes 21, 18 and 13 on the analysis of a new blood sample.

Limitations

There are rare occasions when an NIPT result does not accurately reflect the chromosomal status of the fetus. All NIPTs rely on fragments of DNA found in the mother's bloodstream; these fragments come from the placenta. The placenta and the fetus develop from the same single cell (the fertilised egg) and usually have the same chromosomes. Sometimes, the DNA status of the placenta is not the same as that of the fetus and, as a result, the NIPT result may not reflect the DNA status in the fetus. For these reasons, among others, NIPT is considered a screening test. It is not designed to give a definitive result and all findings should be confirmed with the appropriate diagnostic test. An NIPT result should be carefully reviewed by your doctor, together with other information about your pregnancy, before basing any decision on that result. An ultrasound scan prior to the NIPT is recommended to confirm viability, number of fetuses and gestation. Please also note that NIPT is a test for the conditions noted above; it is not a test for every possible fetal condition or fetal viability. These considerations apply to any form of NIPT.

Fetal sex and sex chromosomes

For biological reasons, it is more difficult to count the numbers of sex chromosomes (X and Y) than other chromosomes. We do not routinely report fetal sex or analysis of sex chromosomes, but will do so on request. Please note that the request should be made by your doctor on the initial request form. Analysis for differences in the number of sex chromosomes is only available for singleton pregnancies.

In a small percentage of samples, a result can be provided regarding the probability of trisomy 21, 18 or 13 but not for fetal sex or sex chromosome conditions (or both). Various factors may make it difficult to provide accurate sex chromosome analysis, including poor quality of DNA in the sample, uncommon normal variations in the sex chromosomes, and a mixture of normal and abnormal cells in the placenta or mother. In these situations, we will report that a result is not possible for fetal sex or screening for a sex chromosome abnormality; alternatively, the accuracy of evaluation for sex chromosome conditions may be compromised. We do not recommend repeat testing, as the biological factors responsible for the lack of a result are unlikely to have changed. We do not offer a refund, as the primary purpose of NIPT (screening for trisomy 21, 18 and 13) has been achieved.

Genetic counselling

Various forms of prenatal screening tests have been available in Australia for more than 30 years. Your doctor will be able to provide you with information and advice regarding this test, and can help you decide if this is the right test for you. Your doctor may also recommend that you seek genetic counselling before or after the test; we can provide contact details for genetic counselling services nationally.

If you have a result indicating a high likelihood of a chromosome condition, a genetic counselling consultation by telephone is available through Sonic Genetics at no charge. This must be requested by your doctor, and is only available to patients who have paid Sonic Genetics directly for their test. Please refer to sonicgenetics.com.au/patient/nipt/gc for further details.

If you have any questions regarding your test result, we recommend that you discuss these with your doctor or genetic counsellor. Your doctor will know your situation and have the results of other tests and assessments. Your doctor can also contact our senior scientists and genetic pathologists to discuss technical aspects of your result.

Results

Your results will be delivered to your doctor typically within 3–8 business days of the laboratory receiving your sample.