



Sonic
Genetics

Melbourne Pathology

Non-invasive Prenatal Testing

Information for Patients



harmony®

PRENATAL TEST



Congratulations on your pregnancy

Pregnancy can raise a whole range of emotions, from delight and excitement, through to anxiety about the health of your developing baby.

During your pregnancy you will read and hear about a range of tests that can help you determine the health of your growing baby. These tests – known as First Trimester Screening (FTS) – include maternal blood tests and ultrasounds. The tests may reveal a higher chance of a disorder in your baby and, at this point, you may be offered an invasive procedure, such as a Chorionic Villus Sampling (CVS) or an amniocentesis.

These invasive procedures look directly at the DNA or chromosomes of your baby, but they do carry a risk of miscarriage.

A new type of screening test, the Non-invasive Prenatal Test (NIPT), has been developed. This is a blood screening test that has far greater accuracy than conventional screening methods. The non-invasive prenatal test provided by Sonic Genetics is called Harmony®.

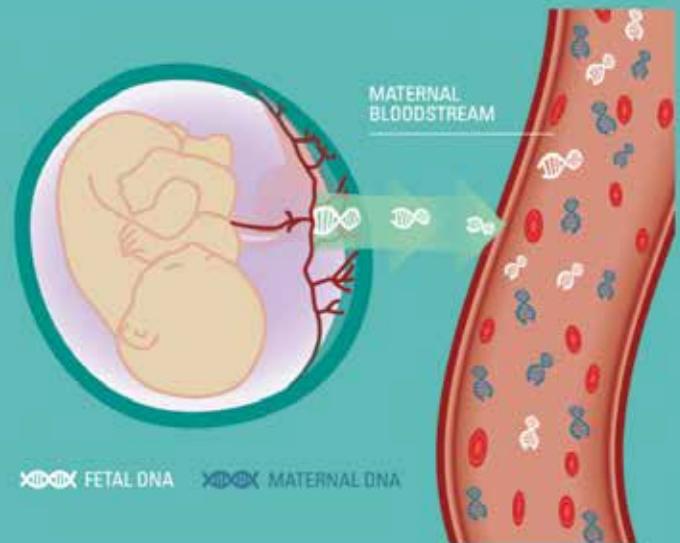
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PRENATAL TEST

What is a non-invasive prenatal test?

When you are pregnant, your blood contains fragments of your baby's DNA.

A non-invasive prenatal test analyses this DNA in a sample of your blood, to assess the chance of trisomy 21 (Down syndrome) and two other genetic conditions, trisomy 18 (Edwards syndrome) and trisomy 13 (Patau syndrome). While some genetic conditions run in families, these chromosomal disorders typically do not. They can happen in any pregnancy. Although the chance of such conditions increases with age, most babies with Down syndrome are born to women under 35 years of age.



Sonic Genetics provides the Harmony® Prenatal Test

➤ Performed in Australia

➤ Early answers

A non-invasive prenatal test is a single blood test that can be done as early as 10 weeks, or later in pregnancy.

➤ Greater confidence with a more accurate test

The Harmony® Prenatal Test is one of the few prenatal screening tests that includes a measurement of the amount of the baby's DNA present in the mother's blood. This is an essential measurement to ensure that there is sufficient DNA from the baby for the test result to be accurate.

➤ Clinically proven results

In April 2015, the New England Journal of Medicine (NEJM) published a study on the performance of the Harmony® Prenatal Test in a group of more than 15,000 pregnant women of various ages and risk, across many different countries. This is one of the largest NIPT studies ever published and showed that non-invasive prenatal tests significantly outperform the current screening standard for trisomy 21 (Down syndrome).

➤ Minimise the need and associated risks of invasive procedures

The accuracy of non-invasive prenatal testing means that far fewer women require an invasive test, such as CVS or amniocentesis. Nonetheless, confirmatory diagnostic testing, via either amniocentesis or CVS, is recommended following a high probability non-invasive prenatal test result.

Detection rate in the NEJM study

Pregnancies with trisomy 21 (Down syndrome) correctly identified:

Harmony® (38 of 38)	100%
First Trimester Screening (30 of 38)	79%

Harmony® correctly identified as high probability all pregnancies with trisomy 21 (Down syndrome). First Trimester Screening failed to identify as high probability eight of the 38 pregnancies with trisomy 21 (Down syndrome).

Pregnancies with trisomy 21 (Down syndrome) not identified:

Harmony®	0 of 15,794
First Trimester Screening	8 of 14,949

Of the 15,794 pregnancies identified as low probability by Harmony®, none had trisomy 21 (Down syndrome) i.e. none of 15,794 low probability results came from a pregnancy with trisomy 21. Of the 14,949 pregnancies identified as low probability by First Trimester Screening, eight had trisomy 21 (Down syndrome) i.e. one in 1,869 low probability results was from a pregnancy with trisomy 21.

Pregnancies without trisomy 21 (Down syndrome) correctly identified:

Harmony®	9 of 15,803
First Trimester Screening	854 of 15,803

In the 15,803 pregnancies without trisomy 21 (Down syndrome), Harmony® correctly identified 99.9% as being at low probability of trisomy 21. First Trimester Screening correctly identified only 95% as being low probability.

Common questions

What is a trisomy?

A trisomy is a chromosomal condition that occurs when there are three copies of a particular chromosome instead of the expected two.

What does Harmony® screen for?

Harmony® screens for the most common chromosomal conditions – trisomy 21 (Down syndrome), trisomy 18 (Edwards syndrome), trisomy 13 (Patau syndrome).

Does Harmony® test for fetal sex chromosomes?

Females have two X chromosomes and males have one X and one Y chromosome. The Harmony® Prenatal Test can screen for conditions caused by having an extra or missing copy of the X or Y chromosomes. The Harmony® Prenatal Test can also report the fetal sex chromosomes of your baby, if requested.

Can Harmony® be used for twins?

Yes, the Harmony® Prenatal Test can be used in twin pregnancies to test for trisomies 21, 18 and 13 and fetal sex chromosomes. The test can tell whether both twins are girls or at least one of the twins is a boy. It cannot tell if both twins are boys or if there is one boy and one girl. It is not possible to test for sex chromosome abnormalities in twin pregnancies. The Harmony® Prenatal Test is not available for triplet or higher order pregnancies.

Can Harmony® be performed on IVF pregnancies?

Yes, the Harmony® Prenatal Test can be used for both self-conceived and egg donor pregnancies.

How will I get my results?

Your results will be delivered to your doctor. The test result will give you a clear answer about the probability of your pregnancy having any of the genetic conditions screened for. If the test result shows that the probability of these disorders is low, it means that the conditions screened for were not detected. If the test result shows that there is a high probability of any of these conditions, we recommend further confirmatory diagnostic testing.

How much does Harmony® cost?

Current pricing is found on our website – www.sonicgenetics.com.au/nipt/pricing.

How long does it take?

Results are available 5–8 business days from your blood test.



Three steps to a Harmony® Prenatal Test result

1 At 10 weeks or later, get a blood test.

Visit your doctor, who will complete a Harmony® request form, then have your blood sample collected at one of our many Melbourne Pathology collection centres. Pre-payment is required, and current pricing and collection centre locations are found on the Sonic Genetics website.

2 Your blood sample is processed by the Sonic Healthcare genetics laboratory at Sullivan Nicolaides Pathology (SNP).

3 Your results are sent to your doctor in 5–8 business days from the day your blood sample was collected*.

*In rare cases (less than 3%), the laboratory is unable to obtain a result from the first sample. This can occur in samples where there is not enough of the baby's DNA. Further testing may be required (at no additional cost), and can therefore delay the result.

Why choose Sonic Genetics for your non-invasive prenatal test?

- Sonic Healthcare, our parent company, is Australia's largest provider of diagnostic services. We have extensive experience in the delivery of genetic testing across the country – with expertise in medically-led practices.
- We have Australia's largest network of collection centres – so you can choose the most convenient location to have your blood sample collected.
- The genetics laboratory at Sullivan Nicolaides Pathology was the third laboratory in the world to offer the Harmony® Prenatal Test. Until October 2015, Australian women had their samples sent to California. All testing by Sonic Genetics is performed in Australia and not sent overseas.
- We have developed systems to ensure that your blood sample is processed as quickly as possible, allowing your doctor to receive your result as soon as it is available.
- Sullivan Nicolaides Pathology is the only Australian provider that is NATA-accredited to perform the Harmony® non-invasive prenatal test. SNP provides Harmony® testing for Sonic Genetics and the Sonic Healthcare group. We test to Australian testing standards, so this means you can be assured of a high quality, accurate result.



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For further information, including scientific and peer-reviewed publications, please refer to our website www.sonicgenetics.com.au/nipt or contact us on: **T** 1800 010 447 | **E** info@sonicgenetics.com.au

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The published evidence that is the basis for this leaflet is detailed in other leaflets about Harmony® available on the Sonic Genetics website. Non-invasive prenatal testing (NIPT) services based on cell-free DNA analyses are not diagnostic; high probability results should be confirmed by a diagnostic test. The Harmony® Prenatal Test is developed by Ariosa Diagnostics. Testing is performed in Australia by Sonic Genetics in our NATA-accredited Sullivan Nicolaides Pathology laboratory. As with other laboratory-developed tests, this test is pending inclusion on the Australian Register of Therapeutic Goods (ARTG).

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