



Sonic
Genetics

Melbourne Pathology

Non-invasive prenatal testing

Information for patients

harmony[®]
PRENATAL TEST

Now including 22q11.2 deletion





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.

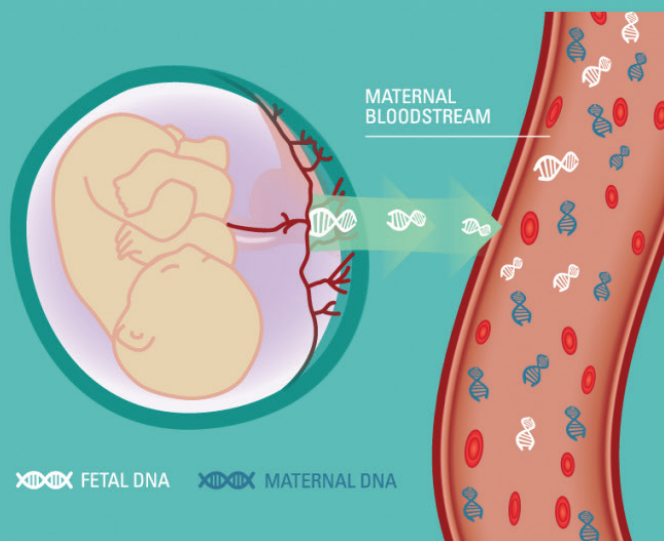
During your pregnancy you will read and hear about a range of tests that can help you determine the health of your growing baby. These tests – known as first trimester screening (FTS) – include maternal blood tests and ultrasounds. The tests may reveal a higher chance of a disorder in your baby and, 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 the Harmony® 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), trisomy 18 (Edwards syndrome) and trisomy 13 (Patau syndrome), plus 22q11.2 deletion (DiGeorge syndrome) if requested. 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)	>99%
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) incorrectly 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 or a deletion?

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

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) and 22q11.2 deletion (DiGeorge syndrome), if requested.

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 optionally 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. However, this is not the intended purpose of the test. Should the fetal sex not be determined due to biological limitations of the assay, repeating the test will not change that outcome and a refund is not appropriate.

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 or 22q11.2 deletions 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.

What if the result indicates a common chromosomal disorder?

Genetic counselling is important to help you understand the implications of the result from this test. If your Harmony result indicates a high probability of a chromosomal condition, including 22q11.2 deletion, Sonic Genetics provides access to an independent, qualified genetic counsellor, free-of-charge for patients who have prepaid us directly. Details on how to access the service will accompany the result sent to your doctor.

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.

Does it always work?

Occasionally, due to biological limitations, such as low fetal DNA in your blood stream, no result will be available and a repeat test will be warranted and offered free-of-charge. Approximately 1% of patients may not get a result and a full refund will be issued upon request if you have prepaid your local Sonic Genetics pathology provider. The intended purpose of the Harmony Prenatal Test is to screen for common chromosomal disorders. If such a result is provided then the test has achieved its aim. If, in such instances, there is no result for the optional fetal sex or sex chromosome abnormalities, no refund will be given.

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 Non-invasive Prenatal Test 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 could 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. 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 NATA-accredited to perform the Harmony Prenatal Test. SNP provides Harmony testing for Sonic Genetics and the Sonic Healthcare group. We work to Australian testing standards, so this means you can be assured of a high quality, accurate result.



MELBOURNE
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For further information, please refer to our website
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The Harmony non-invasive prenatal test is based on cell-free DNA analysis and is considered a prenatal screening test, not a diagnostic test. Harmony does not screen for potential chromosomal or genetic conditions other than those expressly identified in this document. All women should discuss their results with their healthcare provider who can recommend confirmatory, diagnostic testing where appropriate. The Harmony Prenatal Test is validated for use in women ≥ 18 years. The Harmony Prenatal Test was developed by Ariosa Diagnostics and is performed in Australia at our NATA-accredited Sullivan Nicolaides Pathology (SNP) laboratory. It is included on the Australian Register of Therapeutic Goods.

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