



Sonic
Genetics

Douglass Hanly Moir Pathology

Non-invasive Prenatal Testing

Information for Patients



harmony™

PRENATAL TEST

performed in Australia



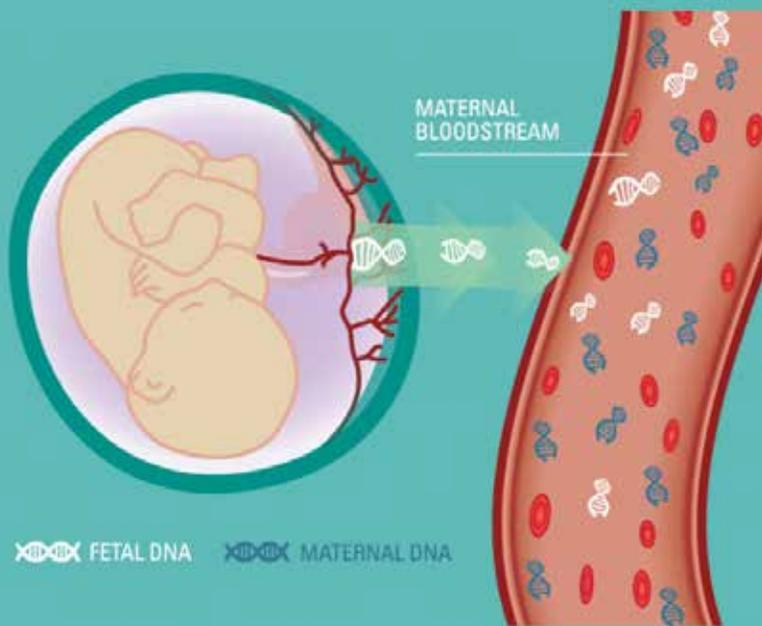
Congratulations on your pregnancy

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

harmony™

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What is the Harmony™ Prenatal Test?

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

The Harmony™ 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). Some genetic conditions run in families; others, (for example chromosomal conditions such as trisomy 21), 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.

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 include maternal blood tests and ultrasounds – known as First Trimester Screening (FTS). These 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 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 Harmony™ Prenatal Test, has been developed that is a non-invasive, simple blood screening test that has far greater accuracy than conventional screening methods.

Why choose Harmony™ Prenatal Test?

› Now performed in Australia

› Early answers

The Harmony™ Prenatal Test requires a single blood test that can be done as early as 10 weeks or later in pregnancy. Traditional First Trimester Screening tests are performed later in pregnancy – from 12–14 weeks.

› 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. Some prenatal screening tests do not include this measurement and potentially provide a result which reflects the mother's chromosomes rather than the baby's.

› 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 Harmony™ Prenatal Test significantly outperforms the current screening standard for trisomy 21 (Down syndrome).

The NEJM Study Results (Patients in study = 15,841)

Detection rate

Babies with trisomy 21 (Down syndrome) correctly identified.

T21: Harmony™ (38 of 38) 100%

T21: First Trimester Screening (30 of 38) 79%

Reduction in T21 false-positive rate by over 90-fold

9 of 15,803 False positives with Harmony™

854 of 15,803 False positives with FTS

A false positive is a test result which wrongly reports that a particular condition or attribute, in this case trisomy 21, is present. As shown above, in this study, 854 women were told they had a positive result following First Trimester Screening when they did not, compared to only 9 with Harmony™. False-positive results clearly cause significant and unnecessary stress and anxiety, and lead to unnecessary invasive procedures.

Minimise your risk of unnecessary invasive procedures

Confirmatory diagnostic testing, via either amniocentesis or CVS, is recommended following an abnormal FTS or Harmony™ result.

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 isn't possible to test for sex chromosome abnormalities in twin pregnancies. The Harmony™ Prenatal Test is not available for triplet or higher order multiple 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 chance in your pregnancy of having any of the genetic conditions screened for. If the test result shows that the chance 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 chance 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 simple steps to a Harmony™ Prenatal Test result

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

Visit your doctor, who will complete a special request form, then have your blood sample collected at one of hundreds of collection centres across Australia (visit: www.sonicgenetics.com.au/locations). Pre-payment is required, and current pricing is found at: www.sonicgenetics.com.au/nipt/pricing.

2 Your blood sample is sent to the Australian Sonic Genetics laboratory.

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 4%), 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, and can therefore delay the result.



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Why choose Sonic Genetics for your Harmony™ 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.
- Sonic Genetics was the third laboratory in the world to offer the Harmony™ Prenatal Test. Until October 2015, Australian women had to send their samples to California. All testing by Sonic Genetics is now 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 almost as soon as it is available.
- Sullivan Nicolaides Pathology (SNP) 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.

For further information, including scientific and peer-reviewed publications, please refer to our website, www.sonicgenetics.com.au/nipt or call us on 1800 010 447

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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 services (NIPT) based on cell-free DNA analyses are not diagnostic; positive 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 Sullivan Nicolaides Pathology Laboratory, which is NATA accredited. As with other laboratory-developed tests, this test is pending inclusion on the Australian Register of Therapeutic Goods.

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