harmony®

Clear ANSWERS to Questions that Matter

Clinical Labs is proud to be the exclusive Australian provider of Harmony NIPT, the most accurate non-invasive prenatal test, including 22q11.2 microdeletion.

AUSTRALIAN Clinicallabs

antenatal.clinicallabs.com.au/patient

During your pregnancy, your healthcare providers will offer you various screening tests to help assess your baby's health.

As part of your care, you will be given the option to screen for Down syndrome, a rare genetic condition that affects physical and mental development.

Experts support Down syndrome screening

Some genetic conditions run in families. Others, like Down syndrome, typically do not. They can happen in any pregnancy. Although the risk of Down syndrome increases with age, most babies with Down syndrome are born to women under 35 years of age.[3]

The Royal Australian and New Zealand College of Obstetricians and Gynaecologists (RANZCOG) recommends that all pregnant women be provided with information regarding screening for Down syndrome.[4]

What is the Harmony prenatal test?

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

Harmony NIPT is a screening test that analyses this DNA in a sample of your blood to assess the risk of Down syndrome (trisomy 21) and two other genetic conditions, Edwards syndrome (trisomy 18) and Patau syndrome (trisomy 13).

A more accurate test

Combined first trimester screening (cFTS) can detect 85-90% of pregnancies with trisomy 21 with a false positive rate of 3-5%[2].

Harmony has been shown in clinical testing to identify greater than 99% of Down syndrome cases and to have a false-positive rate of less than 0.1%.[1-2]

Suitable for women of any risk category*

The Harmony prenatal test was developed to be a more accurate prenatal Down syndrome screening test, is validated for use in women ≥ 18years and is suitable for women of any risk category*.

It is a DNA-based blood test that has been extensively tested in both the under 35 and over 35 age groups. Studies have included pregnant women ages 18-48 for trisomy 21.[1,2,5]

*Any risk refers to the average-risk population (age < 35) and high-risk population (age > 35). Pregnancies with more than two foetuses, a history of vanishing twin, maternal organ transplant, or maternal aneuploidy are not eligible for the Harmony test.

Harmony versus traditional Down syndrome tests[2]

HARMONY NIPT	**COMBINED** First Trimester Screening***
FALSE-POSITIVE RATE* Less than 0.1%	FALSE-POSITIVE RATE* 3-5%
DETECTION RATE** Greater than 99%	DETECTION RATE** 85-90%

* Reports a high risk for Down syndrome when it is NOT actually present
** Correctly indicates a high risk for Down syndrome when it IS present
*** Serum PAPP-A, total or free ß-hCG & Nuchal Translucency

Minimises need for invasive follow-up tests

The greater accuracy and low false-positive rate of Harmony compared to traditional tests may minimise the chance that further testing would be recommended due to a false-positive result.[6] Follow-up testing might include an invasive procedure, such as amniocentesis.

Clarity early

Harmony NIPT requires a single blood draw and is ideally performed between 10+ and 14 weeks' gestation. **Your referring doctor will receive your results 5-10 business days after the receipt of your sample at our lab.** Other commonly used screening tests for Down syndrome are performed later in pregnancy and may require multiple appointments.

Fetal sex chromosomes ♀♂

The Harmony prenatal test can also screen for conditions caused by having an extra or missing copy of the X or Y chromosomes, including Turner and Klinefelter syndromes. You also have the option to obtain information about the sex of your baby.[7,8]

22q11.2 microdeletion

Harmony NIPT can include an assessment for 22q11.2 microdeletion. 22q11.2 microdeletion is the most common microdeletion syndrome, occurring in as many as 1 in 1,000 pregnancies,[9] and is the second most common genetic cause of developmental delay and congenital heart disease after Down syndrome. Please discuss with your clinician if you wish to include the 22q11.2 microdeletion option with your Harmony prenatal test (an additional fee applies).

Who can have Harmony NIPT?

Pregnant women who are at least **10 weeks gestational age** with a singleton or twin pregnancy resulting from natural conception or IVF are eligible for this analysis. However, the additional fetal sex chromosome aneuploidy analysis and 22q11.2 microdeletion can only be performed in singleton pregnancies. The Harmony prenatal test is validated for use in women ≥ 18years.

What is genome-wide (GW) prenatal screening?

GW testing has the potential to identify rare autosomal trisomies and rare additional fetal segmental imbalances but with shallow analysis depth across all chromosomes.

Consequently, a GW approach is not recommended by clinical guidelines.

Is GW prenatal testing recommended?

The Human Genetics Society of Australasia (HGSA) and RANZCOG do not recommend GW prenatal screening. In their updated 2018 guidelines, they state that "*routine population-based screening for genome-wide chromosome abnormalities are not recommended due to the absence of well-performed clinical validation studies*".

The clinical utility of GW testing is unknown and unproven. The accuracy of the GW testing platform has not been established, but an increased rate of false positives has been demonstrated[10], which can lead to unnecessary invasive procedures and increased maternal anxiety.

GW testing also has limited ability to detect clinically relevant conditions such as 22q11.2 deletion syndrome.[11,12]

Harmony is the most proven NIPT[13]. It's test menu focuses on clinically relevant conditions and provides clear answers to the questions that matter most.[2]

Do I need to book for the test?

No, you do not need to make a booking for Harmony NIPT. However, for our collection centres in South Australia and the Northern Territory, we recommend calling ahead.

Where can I have my Harmony NIPT test?

Your doctor will provide you with a Harmony prenatal test request form. Payment is required before collection. All our collection centres across Victoria, New South Wales, Queensland, South Australia, Western Australia, the Northern Territory, and the Australian Capital Territory can collect a blood sample for Harmony NIPT.

For a current list of our collection centres, please visit clinicallabs.com.au/location.

How do I pay for my test?

Currently, neither Medicare nor private health insurers cover the cost of the test. For current pricing and to pay online, please visit pay.clinicallabs.com.au/harmony or scan the QR code.

Is genetic counselling available?

Genetic counselling will be offered to all women whose test result indicates a pregnancy with high probability of chromosomal abnormality within 48 business hours. The service may be provided, at your doctor's discretion, in one of the following ways:

- Your doctor may choose to provide the genetic counselling themselves;
- By the Australian Clinical Labs allied initial genetic counselling service (no additional fee applies);
- By referral from your doctor to a local genetic counselling service.

You are 10+ weeks pregnant and ready to have your HARMONY NIPT TEST

Simply follow these 4 easy steps:

1. Obtain a Harmony Request form from your doctor
2. Pay for your test online at pay.clinicallabs.com.au/harmony
3. Visit your nearest Clinical Labs collection centre (clinicallabs.com.au/location) with your request form and payment receipt
4. Your referring doctor will receive your results within 5-10 business days

Your results

The test result will give you a clear answer about the risk to your pregnancy of having any of the genetic conditions included in the test.

Once you have your Harmony test results, you can discuss your pregnancy care with your healthcare provider.

For frequently asked questions about Harmony NIPT testing, please visit our website antenatal.clinicallabs.com.au/patient/harmony/faq or scan the QR code.

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2. Norton et al. NEJM 2015; 372 (17): 1589-1597
3. The California Prenatal Screening Program March 2009. Provider Handbook 2009
4. RANZCOG Guideline (C-Obs 59) July 2018
5. Norton et al. Am J Obstet Gynecol. 2012 Aug;207(2):137.e1-8
6. Wax et al. J Clin Ultrasound. 2015 Jan;43(1):1-6
7. Nicolaides et al. Fetal Diagn Ther. 2014;35(1):1-6
8. Hooks et al. Prenat Diagn. 2014 May;34(5):496-9
9. Grati et al. Prenat Diagn. 2015 Aug; 35(8): 801-9
10. Van Opstal et al. Genet Med. 2018 Apr; 20(5): 480-485
11. Benn P and Grati F. Ultrasound Obstet Gynecol. 2018 Apr; 51(4): 429-433
12. Wapner et al. NEJM 2012 Dec 6; 367(23): 2175-84
13. Demonstrated by 72 peer-reviewed published studies using the Harmony prenatal test as of Jan 2021

Non-invasive prenatal testing (NIPT) based on cell-free DNA analysis is not diagnostic: results should be confirmed by diagnostic testing. Before making any treatment decisions, all women should discuss their results with their healthcare provider, who can recommend confirmatory, diagnostic testing where appropriate. The Harmony Prenatal Test is developed by Ariosa Diagnostics and it is performed in Australia.

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