Sonic Genetics
Quality is in our DNA

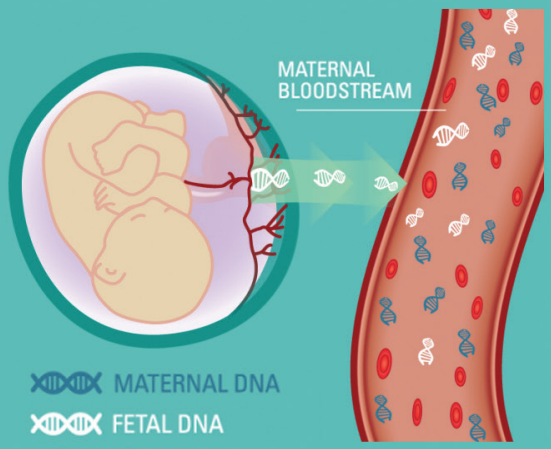
harmony®
PRENATAL TEST

Prenatal testing for your patients

Non-invasive prenatal testing (NIPT)

What is the intended purpose of non-invasive prenatal testing?

Non-invasive prenatal testing (NIPT) is a screen of a pregnant woman's blood to determine the chance that her developing baby has a specific chromosome abnormality. During pregnancy, DNA from the mother and fetus circulate in the mother's plasma. This 'cell-free DNA' (cfDNA) can be tested to estimate the risk of a fetus having a common chromosome abnormality. Sonic Genetics offers the Harmony® Prenatal Test.



What does Harmony screen for?

Trisomy 21 (Down syndrome) is associated with moderate to severe intellectual disability and may also cause congenital heart defects and other malformations.

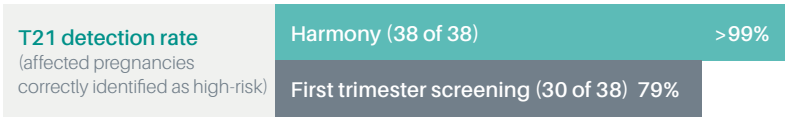
Trisomy 18 (Edwards syndrome) and Trisomy 13 (Patau syndrome) are associated with a high rate of miscarriage. These babies are born with severe brain abnormalities and often have congenital heart defects as well as other birth defects. Most affected babies die before or soon after birth, and very few survive beyond the first year of life.

22q11.2 deletion (DiGeorge syndrome) is the most common microdeletion syndrome. It is often associated with cleft palate and congenital heart disease.

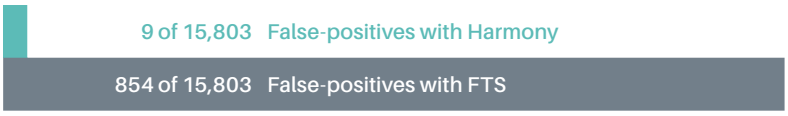
Sex chromosome abnormalities (SCA) (abnormalities in the number of X and Y sex chromosomes) can be associated with infertility (eg. Turner syndrome (45,X) and Klinefelter syndrome (47, XXY)), and may be associated with other malformations or developmental issues.

Why choose Harmony for your patient?

- **Performed as early as 10 weeks**
- **Supported by extensive clinical data:** In a 2015 study, published in the New England Journal of Medicine, that included more than 15,000 women, Harmony significantly outperformed the current screening for trisomy 21[1].
- **Exceptionally accurate results**
  - **Includes fetal fraction assessment:** Harmony will reveal if there is insufficient fetal DNA in the mother's blood to generate a result.
  - Higher detection rate[1]



  - Reduction in T21 false-positive rate[1] by over 90-fold minimises invasive procedures, such as CVS and amniocentesis

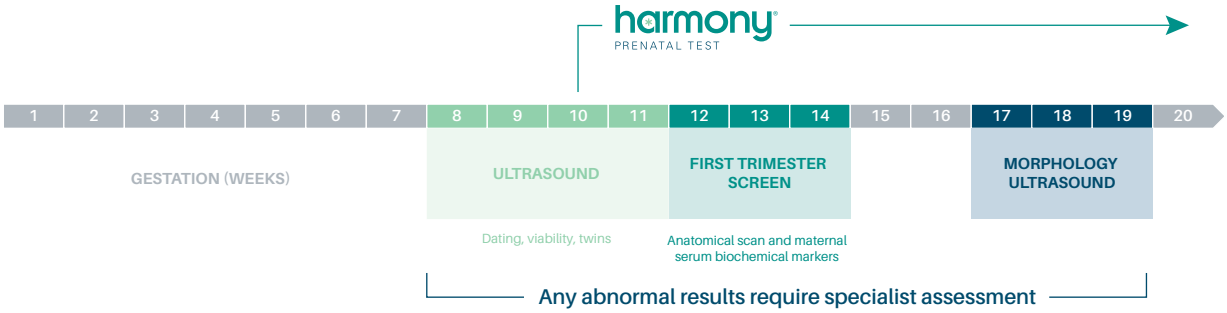


- **Free genetic counselling for eligible high-risk patients**

Ref: 1. Norton M et al., *N Engl J Med.* 2015; Published online https://doi.org/10.1056/NEJMoa1407349

Where does Harmony fit in prenatal care? Should a woman have conventional first trimester screening (FTS) and ultrasound as well as Harmony?

First trimester screening and screening by NIPT provide complementary information about the fetus. Harmony is an assessment of cell-free DNA from specific chromosomes, while first trimester screening assesses fetal anatomy and biochemical function (www.ranzcog.edu.au).



What does Harmony report?

- Fetal fraction in the maternal sample – it must be above 4% to generate an accurate result
- Probability for autosomal trisomies (T21, T18 and T13) and 22q11.2 deletion if requested
- Recommendation to clinician

In less than 3% of patients, Harmony may not give a result on their initial blood sample. The most common reason is biological – such as insufficient fetal DNA. Such samples can be recollected at no additional charge to the patient where indicated. This provides a result for 60-70% of recollected patients. If no result for chromosomal abnormalities is available even after a recollect, a full refund will be given to the patient upon request.

Harmony® Prenatal Test Report EXAMPLE

CHROMOSOME	RESULT	RECOMMENDATION
Trisomy 21 (T21)	High Risk	Genetic counselling and additional testing
Trisomy 18 (T18)	Low Risk	Review results with patient
Trisomy 13 (T13)	Low Risk	Review results with patient
Fetal Sex	Male	Review results with patient
Sex Chromosome Aneuploidy (SCA)	Low Risk	Review results with patient
22q11.2	No evidence of a deletion	Review results with patient

Why choose Harmony at Sonic Genetics?

- **Support for clinicians:** Clinicians can speak directly with one of our genetic pathologists or clinical geneticists to discuss their patient's results. We also have resources for doctors and patients at www.sonicgenetics.com.au/nipt.
- **Experience you can trust:** Our parent company, Sonic Healthcare, is Australia's largest provider of diagnostic services.
- **Quality assurance:** The Harmony Prenatal Test is included on the Australian Register of Therapeutic Goods (ARTG) and is performed in a laboratory accredited by local regulatory authorities (NATA).
- **Convenience for your patients:** We have the largest number of collection centres across Australia.
- **Results:** You can access and download your patient results via our online app: Sonic Dx – anywhere, anytime.

What if my patient's Harmony result is abnormal?

An abnormal Harmony result should always be confirmed by amniocentesis or CVS before making any major decision regarding further clinical management. You can call one of our genetic pathologists to discuss your patient's result at any time.

Additional diagnostic testing

In the event your patient's Harmony test returns a high-risk result, other services offered by Sonic laboratories include confirmatory cytogenetic testing (RAPID FISH and karyotyping). These services are offered at the Medicare rebate, resulting in no out-of-pocket costs to eligible patients who have had Harmony testing with Sonic Genetics. Patients who receive a 'high-risk' result for a fetal chromosomal abnormality will also be able to access telephone-based genetic counselling free-of-charge.

How do I arrange a Harmony test?

WHEN

Performed as early as 10 weeks' completed gestation.

HOW

1) Pretest counselling and patient consent.
2) Complete Harmony Test Request Form.
3) Patient payment and sample collection.

RESULTS

Provided in 5-8 business days from patient blood test. Available electronically via Sonic Dx or downloaded to Practice Management Software. Alternatively, fax or hard-copy reports may be requested.

COST

Medicare does not currently cover the cost of any form of NIPT. Current pricing for Harmony can be found on our website.

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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