Sonic Genetics
Douglass Hanly Moir Pathology

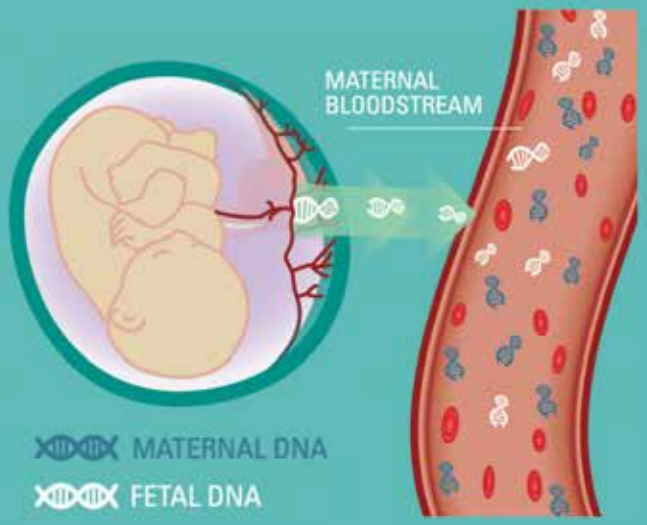
harmony™
PRENATAL TEST
performed in Australia

Prenatal testing for your patients:

Non-invasive prenatal testing (NIPT)

What is the basis of non-invasive prenatal testing?

Non-invasive prenatal testing (NIPT) is a test of a pregnant woman's blood to determine the chance that her developing baby has a serious 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™ NIPT.



What does Harmony™ screen for?

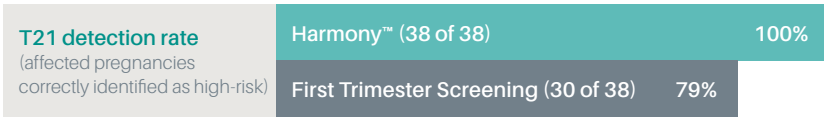
Trisomy 21 (Down syndrome) is associated with moderate to severe intellectual disability and may also cause congenital heart defects and other malformations.

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.

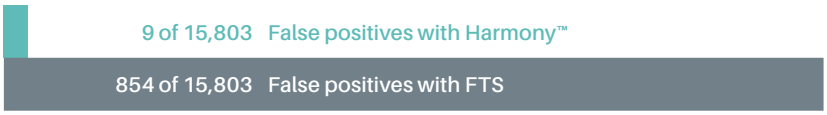
Sex chromosome abnormalities (SCA) (abnormalities in the number of X and Y sex chromosomes) can be associated with infertility (eg. Turner syndrome (45,X) and Klinefelter syndrome (47, XXY)), and may be associated with other malformations or developmental issues.

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 (N Engl J Med. 2015 Apr 23;372(17):1589-97).
- **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

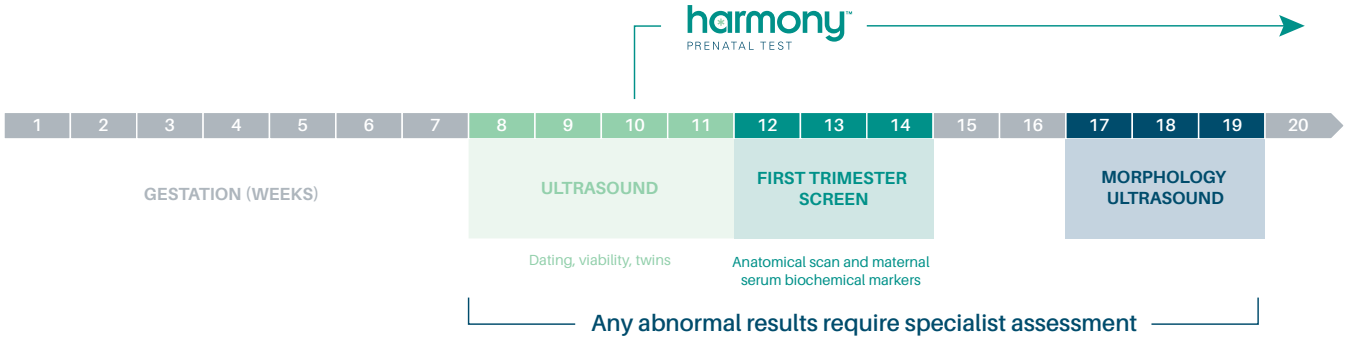


  - Reduction in T21 false-positive rate by over 90-fold minimises invasive procedures such as CVS and amniocentesis



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 the fetal genetic status, while First Trimester Screening assesses fetal anatomy and biochemical function (http://www.ranzcog.edu.au).



What does Harmony™ report?

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

In less than 4% of patients, Harmony™ will not give a result. The most common reason is biological – such as insufficient fetal DNA. Such samples can be recollected at no additional charge to the patient.

Harmony™ Prenatal Test Report EXAMPLE

CHROMOSOME	RESULT	PROBABILITY	RECOMMENDATION
Trisomy 21 (T21)	Low Risk	Less than 1/10,000 (0.01%)	Review results with patient
Trisomy 18 (T18)	Low Risk	Less than 1/10,000 (0.01%)	Review results with patient
Trisomy 13 (T13)	High Risk	Greater than 99/100 (99%)	Genetic counselling and additional testing
Fetal Sex	Female	Greater than 99/100 (99%)	Review results with patient
Sex Chromosome Aneuploidy (SCA)	Low Risk		Review results with patient
Fetal Fraction	12.0%	Fetal fraction refers to the percentage of fetal cell-free DNA in the mother's blood. The test is reported only if the fetal fraction is ≥ 4%.	

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:** We are the only Australian Harmony™ provider to be NATA accredited.
- **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. 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 patients who have had Harmony™ testing with Sonic Genetics.

How do I arrange a Harmony™ test?

WHEN

Gestational age of 10+ weeks

HOW

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

RESULTS

Direct to you (via Sonic Dx or fax) in 5-8 business days from patient blood test.

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

14 Giffnock Avenue, Macquarie Park, NSW 2113, Australia
T: 1800 010 447 | **E:** info@sonicgenetics.com.au
www.dhm.com.au

The published evidence that is the basis for this leaflet is detailed in other leaflets about Harmony™ available on the Sonic Genetics website.

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