SONIC GENETICS

Quality is in our DNA

Non-invasive Prenatal Testing

Information for patients

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Pregnancy is one of the most significant episodes in anyone's life – for you (the pregnant woman), your partner and your child.

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

Non-invasive prenatal testing (NIPT) is a major revolution in the type of prenatal screening that is now available to pregnant women, and the doctors caring for them. NIPT is a blood test that can tell you with remarkable accuracy whether your baby is at risk of developing the most common chromosomal abnormalities (Down syndrome, Edwards syndrome or Patau syndrome), as well as abnormalities with the X and Y sex chromosomes.

Sonic Genetics has reviewed the available NIPT tests and concluded that the Harmony™ Prenatal Test provides you with significant benefits over other NIPTs. These benefits have been substantiated in extensive international tests and reported in medical journals.

harmony™
PRENATAL TEST

What is non-invasive prenatal testing?

During pregnancy, a mother's blood contains fragments of her baby's DNA. Non-invasive prenatal testing, or NIPT, analyses the DNA in a sample of your blood to predict the risk of Down syndrome (trisomy 21), Edwards syndrome (trisomy 18) and Patau syndrome (trisomy 13). Collecting your blood sample for NIPT poses no threat to your baby.

Does it test for all genetic conditions?

No. NIPT specifically tests for trisomies 21, 18 and 13. These account for approximately 80% of chromosomal abnormalities. NIPT does not screen for all chromosomal abnormalities, nor does it screen for familial or inherited disorders (such as cystic fibrosis) or birth defects.

The Harmony™ Prenatal Test also offers optional testing of sex chromosomes.

When can NIPT be performed?

NIPT can be performed as early as 10 weeks or later in pregnancy.

How do First Trimester Screening and NIPT differ?

First Trimester Screening (FTS) and NIPT are very different types of tests, both in how they test and the accuracy of their results. However, together they offer you very important information about your developing baby. FTS combines the results of blood tests with the ultrasound findings to predict the risk that your baby has a **chromosomal** or **structural abnormality**.

The range of disorders that can be detected by FTS is broader than testing by NIPT, however, it is not as accurate as NIPT for detecting Down syndrome, Edwards syndrome and Patau syndrome. This is because NIPT is a genetic test that analyses your baby's DNA fragments from within your blood sample. Recent studies show NIPT to be more accurate in identifying certain chromosomal abnormalities, such as trisomy 21 (Down syndrome), with fewer false-positive results than FTS.[1,2]

Where does amniocentesis or Chorionic Villus Sampling (CVS) fit in?

If your FTS result is abnormal, NIPT is usually recommended as the next step. If the NIPT result is normal, this is reassuring and further analysis by an invasive technique would not be warranted.

However, if your NIPT result is abnormal, further analysis by an invasive technique, such as CVS or amniocentesis, would be recommended.

NIPT is much more accurate than FTS in determining the presence or absence of chromosomal abnormalities, but it is not perfect. Definitive diagnosis can only be achieved by examining cells from a baby, collected by invasive techniques such as CVS or amniocentesis. These procedures do involve risk of miscarriage.

What is the difference between chromosomal and familial genetic conditions?

Chromosomal abnormalities can happen in any pregnancy. They are caused by 'errors' in the way that chromosomes are copied and divide as new cells are formed, either at conception, or during the early stages of pregnancy. They are usually a random event.

Familial genetic conditions occur because the 'faulty' gene is inherited from one or more family members. These inherited genes are associated with conditions such as cystic fibrosis or Huntington's disease and require sophisticated tests for parents considering pregnancy.

Can NIPT test for sex chromosomes?

NIPT can also be used as early as 10 weeks to evaluate X and Y sex chromosomes.[3,4] This optional extra testing is for conditions caused by having an extra or missing copy of the X or Y chromosomes, including Turner (Monosomy X) and Klinefelter (XXY) syndromes. Abnormalities in the number of sex chromosomes are usually not as severe as abnormalities of chromosomes 21, 18 and 13. There is no additional cost for this optional testing, however, it can only be performed on single pregnancies.

Can NIPT be performed for twins?

Harmony™ Prenatal Test can be used in twin pregnancies to test for trisomies 21, 18 and 13. However, it isn't possible to test for sex chromosomes.

This test is not available for triplet or higher order multiple pregnancies.

Can NIPT be performed on IVF pregnancies?

Harmony™ Prenatal Test can be used for IVF pregnancies, including both self-conceived and egg donor pregnancies.

What are the benefits of the Harmony™ Prenatal Test?

There are many types of NIPT available and each one uses different methods to derive their results. These all have relative advantages and disadvantages. After thoroughly reviewing the available NIPT methods, Sonic Genetics has concluded that the Harmony™ Prenatal Test offers many significant benefits over other forms of NIPT.

Greater confidence

The Harmony™ Prenatal Test is one of the only NIPT to include the measurement of the amount of a baby's DNA circulating in the mother's blood. This is a critical measurement because if there isn't enough of the baby's DNA in the sample, the test will return results on the mother's chromosomes, rather than the baby's. This important measurement also allows the Harmony™ Prenatal Test to clearly distinguish normal from abnormal results, and improves the overall accuracy of the test.

Proven results

In April 2015, the New England Journal of Medicine 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 multiple international sites.[1] This is the largest NIPT study ever published, and illustrates the clear advantages of the Harmony™ Prenatal Test and its use for pregnant women of any age or risk category.

Quality assurance

The quality and stability of the Harmony™ Prenatal Test has been documented in more than 500,000 patient samples through the Ariosa Diagnostics laboratory in the USA.

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How do I organise a Harmony™ Prenatal Test?

The Harmony™ Prenatal Test must be ordered by your doctor using a special request form. Your doctor will explain the details surrounding NIPT to make sure that it is the right test for you.

Where can I have my blood sample collected?

You will need to book an appointment to have your blood sample collected. Visit our website www.sonicgeneticsnipt.com.au or call 07 3377 8354 or 1800 010 447 for more information.

How much does NIPT cost?

The price is available on our website www.sonicgeneticsnipt.com.au or call us on 1800 010 447 to obtain the price.

When will my results be available?

Your results will be sent to your doctor within about 5–8 working days from the collection of your blood sample.

In addition to the information contained in this brochure, we have a range of videos and detailed FAQs on our website www.sonicgeneticsnipt.com.au. These should help to inform you about all aspects of the tests and what you can expect.

1. Norton M, et al, NEJM DOI: 10.1056/NEJMoa1407349 (published online April 1, 2015)
2. Nicolaides et al. Am J Obstet Gynecol. 2012 Nov;207(5):374.e1-6.
3. Nicolaides et al. Fetal Diagn Ther. 2014;35(1):1-6.
4. Hooks et al. Prenat Diagn. 2014 May;34(5):496-9.

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