Non-Invasive Prenatal Testing (NIPT)

PATIENT INFORMATION

Non-invasive prenatal testing (NIPT analyses cell-free DNA circulating in the pregnant mother's blood. It is a new option in prenatal screening for Down syndrome (trisomy 21) and other common chromosomal conditions (trisomies 18 and 13), X and Y chromosome conditions. This test can be requested for any singleton pregnancy, including in vitro fertilization (IVF) pregnancies with egg donors. It can now also be requested for ALL twin pregnancies (without X or Y) conceived naturally or by IVF using the patient's own egg or a donor egg.

About the Ariosa Harmony™ Prenatal Test

DNA from the fetus circulates in the mother's blood. Cell-free DNA (cfDNA) results from the natural breakdown of fetal cells (presumed to be mostly placental) and clears from the maternal system within hours of giving birth.

During a pregnancy, cfDNA can be tested to give the most accurate screening approach in estimating the risk of a fetus having a common chromosome condition sometimes called a trisomy. This occurs when there are three copies of a particular chromosome instead of the expected two. The test looks to detect the following trisomies:

- Trisomy 21 is the most common trisomy at the time of birth. Also called Down syndrome, it is associated with moderate to severe intellectual disabilities and may also lead to digestive disease, 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 individuals die before or soon after birth, and very few survive beyond the first year of life.

- Sex chromosome conditions. The sex chromosomes (X and Y) determine whether we are male or female. X and Y chromosome conditions occur when there is a missing, extra, or incomplete copy of one of the sex chromosomes. The Harmony with X, Y test can assess risk for XXX, XYY, XXYY, XXY (Klinefelter syndrome), and a missing X chromosome in a girl (Turner syndrome). There is significant variability in the severity of these conditions, but most individuals have mild, if any, physical or behavioural features. If the mother is interested in having this optional testing, she should talk with her healthcare provider to determine if it is right for her. This option is not available for twin pregnancies.

Risk

The testing is non-invasive: it involves taking a blood sample from the mother. The pregnancy is not put at risk of miscarriage, or from other adverse outcomes that are associated with invasive testing procedures such as amniocentesis.

Accuracy

Clinical studies have shown that the Ariosa Harmony™ Prenatal Test has exceptional accuracy for assessing fetal trisomy risk.

A 'high risk' result is indicative of a high risk for a trisomy. The test identifies in singleton pregnancies more than %99 of fetuses with trisomy %98 ,21 of fetuses with trisomy 18, and %80 of fetuses with trisomy 13, and %96 of fetuses with Turner Syndrome. X and Y analysis provides >%99 accuracy for fetal sex. Accuracy for detecting other sex chromosome anomalies varies by condition.

After the test, the number of women required to have a CVS or an amniocentesis is less than ٪1.

It is important to note that if the test results show there is a high risk that the fetus has trisomy 21, 13 ,18 or sex chromosome conditions, it does not mean that the fetus definitely has one of these conditions, although it is highly likely. For this reason, in the event of a 'high risk' (or positive) result, follow-up testing by an invasive procedure is recommended.

In the same way if the test results show that there is a 'low risk' that the fetus has trisomy 13 ,18 ,21 or sex chromosome conditions, it is unlikely that the fetus has one of these conditions. However, there is a very small risk that not all trisomy fetuses will be detected.

All results should be interpreted by a clinician in the context of clinical and familial data. Patients should continue with their usual scan appointments following testing.

Who can have this test?

The Ariosa Harmony™ Prenatal Test can be ordered by healthcare professionals for women with pregnancies of at least ١٠ weeks' gestational age. The Harmony™ Prenatal Test can now be ordered for all IVF singleton pregnancies, including those with egg donors. Samples from pregnant women with twins naturally conceived, or those conceived using the patient's own egg or donor eggs, can also be accepted. This test does not assess risk for mosaicism, partial trisomies or translocations.

The results will be ready in approximately two weeks, at which time most women can have their ١٢week scan for a detailed examination of the fetal anatomy, including measurement of nuchal translucency, nasal bone and other important factors. In this visit, patients can discuss the DNA and ultrasound results with their obstetricians.

On the basis of the NIPT result and the ultrasound findings, a patient can decide whether or not she wants to have an invasive procedure (for example, CVS or amniocentesis).

Repeat samples

There needs to be enough fetal DNA in the maternal blood to be able to provide a result. If there is insufficient fetal DNA in the sample (occurring in ٪3 of cases), another blood sample from the mother may be required. This will be processed in the laboratory at no extra charge.

What is the process?

Once the mother has taken an independent personal decision that she wants to have the non-invasive prenatal test performed, she will be asked to sign a consent form and her blood sample can be taken from a vein in her arm.

Who carries out the analysis of the test?

Her sample and completed request form need to be sent to TDL Genetics, after which they will be referred to Ariosa Diagnostics Inc, USA. Ariosa performs the Harmony™ Prenatal Test on the DNA extracted from her blood sample.

Will the mother need to have any other tests?

The Ariosa Harmony ™ Prenatal Test does not provide information on other rare chromosomal abnormalities. If the ultrasound scan shows a high nuchal translucency or other major physical defects such as brain abnormalities, heart abnormalities, the risk for some rare chromosomal defects may be high. In such cases, the mother may choose to have a CVS or an amniocentesis.

The non-invasive prenatal test does not provide information on other physical defects such as spina bifida, or information on fetal growth. It is therefore advisable that the mother has all the usual ultrasound scans during her pregnancy.