



## 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 fetal chromosomal conditions (trisomies 18 and 13).**

#### About the 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.

#### 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 exceptional accuracy for detecting fetal trisomies.

A 'HIGH RISK' result is indicative of a high risk for a trisomy. The test identifies more than 99% of fetuses with trisomy 21, 98% of fetuses with trisomy 18, and 80% of fetuses with trisomy 13. 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, 18 or 13, 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.



(occurring in 3–5% 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 you have taken an independent personal decision that you want to have the NIPT prenatal test performed, you will be asked to sign a consent form and your blood sample can be taken from a vein in your arm.

In the same way if the test results show that there is a 'LOW RISK' that the fetus has trisomy 21, 18 or 13, 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 the test?

The test is suitable for any woman who has had an ultrasound scan to confirm that her pregnancy is singleton, that the fetus is alive, and that the fetal length is equivalent to a gestation of 10 weeks or more.

The test is not intended for use in cases of multiple pregnancies, IVF pregnancies using donor eggs.

The results will be ready in approximately two weeks, at which time most women can have their 12 week 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

### Who carries out the analysis of the test?

We will send your blood sample along with your personal information (including name, date of birth, gestational age) to The Doctors Laboratory based in London, UK, which has an arrangement with a company called Ariosa Diagnostics Inc, based in San Jose, USA. Ariosa will perform their NIPT test called Harmony™ Prenatal Test on the DNA extracted from your blood sample.

### Do I need to have any other tests?

The NIPT 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, you may choose to have a CVS or an amniocentesis.

The NIPT prenatal test does not provide information on other physical defects such as spina bifida, or information on fetal growth. It is therefore advisable that you have all the usual ultrasound scans during your pregnancy.

### Transfer of your information outside the European Union

For the purposes of carrying out the Harmony Prenatal Test, your personal information will be transferred outside of the European Union, to the USA. Please be aware that the laws applicable to your personal data in the USA are different from those operating in the UK, where The Doctors Laboratory is established.

### For further information, please contact:

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