Ultrasound Diagnostic Services
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Screening for chromosomal abnormalities
The SAFE, Panorama and Harmony Non-Invasive Prenatal Tests

The vast majority of babies are normal. However all women, whatever their age, have a small risk of delivering a baby with a physical and/or mental handicap. In some cases the handicap is due to chromosomal abnormality such as trisomy 21 (Down syndrome). Ultrasound Diagnostic Services has introduced a new approach to provide the most accurate way of estimating the risk of the fetus having trisomy 21 and other chromosomal abnormalities. This is based on the SAFE, Panorama and Harmony Prenatal Tests which are collectively referred to as non-invasive prenatal (NIPT) tests.

What are NIPT tests?
The NIPT tests analyze cell free DNA in maternal blood and gives a strong indication of whether the fetus is at high or low risk of having trisomy 21 (Down syndrome). It also gives an indication of whether the fetus is at risk of having trisomy 18 (Edwards syndrome) and trisomy 13 (Patau syndrome).

The only way to know for sure whether or not the fetus has a chromosomal abnormality is by having an invasive test such as chorionic villus sampling (at 11-15 weeks) or amniocentesis (at 16 weeks or later). However, these tests involve the introduction of a needle into the uterus to take a sample of the placenta (CVS) or amniotic fluid (amnio) and they carry a risk of miscarriage of about 1%.

What is trisomy 21, 18 or 13?
In humans, there are 23 types of chromosomes and most people have a pair of each one of these chromosomes (therefore a total of 46 chromosomes). In trisomy, there are three rather than two of a particular chromosome (total of 47 chromosomes). The most common trisomy is that of chromosome 21. Some other trisomies include those of chromosome 18 and chromosome 13.

- Trisomy 21 is found in about 1 in 700 births and the risk increases with maternal age. The condition is associated with intellectual disabilities and some physical defects, most commonly heart abnormalities. The life expectancy is about 60 years.

- Trisomies 18 and 13 are found in about 1 in 7,000 births and the risk increases with maternal age. The conditions are associated with severe mental handicap and several physical defects. Most affected individuals die before or soon after birth and they rarely survive beyond the first year of life.

Who can have NIPT tests?
The tests can be performed in all pregnancies of at least 10 weeks gestational age. However the test is not currently recommended for the routine detection of non-trisomic abnormalities as the accuracy of this testing has not been verified.

What are the necessary steps for the test?
If you wish to have the NIPT test for prediction of trisomies we will see you at Ultrasound Diagnostic Services for review of the results of any previous tests and in most cases carry out a detailed ultrasound scan to examine the fetal anatomy and growth. We will then take a blood sample from you for analysis.

Who carries out the analysis of your blood?
We will send your blood sample along with your personal information (including name, date of birth, gestational age) to St George's Hospital in London for the SAFE test, Natera who are based in the USA for Panorama or TDL Genetics based in the UK for the Harmony test.

When do I expect to get the results of the NIPT test?

Test Results for the SAFE and Harmony will generally be available in 5 days
Test Results for Panorama will generally be available in 10 days

We will notify you as soon as we receive them by phone or email / letter.

Please note that in up to 5% of cases the test does not give a result. We will repeat the test free of charge under these circumstances.

What would the results show?
If the NIPT test shows that there is a **high risk** that the fetus has trisomy 21 or 18 or 13 it does not mean that the fetus definitely has one of these defects. If you want to be certain if the fetus has one of these defects you should have CVS or amnio.

If the NIPT Test shows that there is a **low risk** that the fetus has trisomy 21 or 18 or 13 it is very unlikely that the fetus has one of these defects. The test identifies more than 99%, but not all, of the fetuses with trisomy 21, 98% of fetuses with trisomy 18 and 80% of fetuses with trisomy 13.

Do I need to have any other tests?
The NIPT test does not provide information on other rare chromosomal abnormalities. If the scan shows a high nuchal translucency (more than 3.5 mm) or major structural defects, such as heart abnormalities, the risk for rare chromosomal defects may be high. In such cases you may choose to have CVS or amnio.

The NIPT test does not provide information on physical defects, such as heart or brain abnormalities and spina bifida, or fetal growth. It is therefore advisable that you still have ultrasound scans at 11-13 weeks and at 20-22 weeks to examine the fetal anatomy and at 34-36 weeks to examine the fetal growth.

What is the cost of the NIPT test?

SAFE test	From 10 weeks (Blood Test only) £345			
SAFE test	Plus Early Viability Scan	£395	Plus Early Anomaly Scan	£495
Panorama	Plus Early Viabilty Scan	£450	Plus Early Anomaly Scan	£550
Harmony	Plus Early Viabilty Scan	£450	Plus Early Anomaly Scan	£550

What is the next step?

Consultants:
Prof Aris Papageorghiou, MD FRCOG
Prof Basky Thilaganathan, MD FRCOG
Mr Guy Thorpe-Beeston, MD FRCOG

You are invited to take the time to ask all the questions you might have to enable you to make an independent personal decision as to whether you wish to take the NIPT test. You are under no obligation to sign the consent form immediately; you can take this document away to read before deciding.

Once you decide you want to take the NIPT test, you will be asked to sign the informed consent form by one of our doctors or an appropriately trained member of our staff. We will then draw a small amount of blood from a vein in your arm. This may cause some discomfort but the sample is usually taken very quickly. Sometimes there can be some bruising after a blood sample is taken.

Who do I contact if I would like more information about the NIPT test?
If you have any questions about the NIPT test, please do not hesitate to contact a member of staff from Ultrasound Diagnostic Services.

Access to your information and correction
You have the right to request a copy of the information we hold about you. If you would like a copy of some or all of your personal information, please contact us at the address provided below. We want to make sure that your personal information is accurate and up to date. You may ask us to correct or remove information you think is inaccurate. You may contact the UK's data protection authority, the Information Commissioner's Office (www.ico.gov.uk), at any time if you feel we have not complied with your legal rights.

Transfer of your information outside the European Union
Your personal information may be stored and accessed by or on behalf of Ultrasound Diagnostic Services on computer systems located outside of the European Union, including the United States. Please be aware that these countries may not have laws that offer the same level of protection for your personal data as in the United Kingdom where Ultrasound Diagnostic Services is established.