What are the limitations of the test?

NIPT is the most accurate screening test currently available. However, the only way to know for sure whether the fetus has a chromosomal problem or not, is to have a diagnostic test such as CVS or an amniocentesis. These tests are invasive and carry a small risk of miscarriage (approximately 1.0% for CVS and 0.2% for amniocentesis).

If the NIPT test yields a low risk result, it is still highly recommended that an anatomy scan be performed at 20-22 weeks gestation to check for structural abnormalities. NIPT does not provide information on other rare chromosomal abnormalities or physical defects such as cardiac abnormalities or spina bifida.

There needs to be enough fetal DNA in the maternal blood sample to be able to provide a result. If there is insufficient fetal DNA in the sample, another maternal blood sample may be required. This happens in 3-4% of cases. There will be no extra charge for the second test; however, the result will be delayed.

The NIPT test currently being used in the Rotunda Hospital (Panorama® test) cannot be used to screen multiple pregnancies or those conceived with donor eggs. Information can be provided about other tests available.

If you would like more information on the conditions mentioned please visit:

www.rotunda.ie
www.panoramatest.com
What is the Non-Invasive Prenatal Test (NIPT)?

NIPT is a new screening test that helps us to identify if your baby is likely to have a chromosomal condition, for example, Down Syndrome (Trisomy 21), Edward Syndrome (Trisomy 18) or Patau Syndrome (Trisomy 13).

Chromosomes contain the genetic material or DNA and are usually arranged in pairs and number 46. The sex chromosomes (X and Y) determine whether you are male (46XY) or female (46XX). The syndromes mentioned above occur when a fetus (unborn baby) develops with three copies of a particular chromosome instead of the normal two. Depending on the syndrome, the fetus may develop with significant abnormalities or malformations. Some of the syndromes are fatal and the baby will die either before birth or shortly afterwards.

NIPT can also screen for some abnormalities linked to the sex chromosomes, for example, Turner Syndrome, which occurs when there is a missing X chromosome in a girl.

NIPT is non-invasive: it ONLY involves taking a blood sample from the mother.

What does the test involve?

A sample of the mother’s blood is taken from her arm. The blood sample is then sent to a special laboratory for testing. The results will be available 10 working days later. You will be contacted by one of the midwives or doctors from the Department of Fetal Medicine who will explain the test results to you.

If you decide to have the test done, you will be given an appointment to attend the Fetal Medicine Department. As this test is an opt-in service there is a fee involved.

What do the test results mean?

The test does not give a definitive result. Instead, the test will give a Low Risk or High Risk result for certain conditions. A High Risk result for a particular condition has 99% accuracy. In this situation, an invasive test such as chorionic villus sampling (CVS) or an amniocentesis will be advised to confirm the diagnosis.

If the test shows a Low Risk result, it is extremely unlikely that the fetus has one of the syndromes that have been tested.