Non-Invasive Prenatal Testing (NIPT) for Down’s syndrome is a new way of identifying pregnant women who are at higher risk of having a baby with Down’s syndrome (also known as Trisomy 21). The test detects DNA from a baby in a sample of blood taken from the mother.

Existing screening for Down's syndrome

All pregnant women in the UK are currently offered the “combined” test for Down’s syndrome as part of the NHS Fetal Anomaly Screening Programme (FASP). Pregnant women who are shown to be at a higher risk of having a baby with Down’s syndrome are then offered follow-up tests. These diagnostic tests can tell whether the baby will have Down’s syndrome but carry a small (1 in 100) risk of miscarriage.

Current availability of NIPT

NIPT is currently only available privately in the UK. Most studies conducted so far have been in high risk women and further work in larger groups of pregnant women is required to evaluate the accuracy of the new test, in particular the false positive rate (that is the number of women incorrectly identified as being at risk).

The UK NSC is therefore supporting a study to assess how the test performs in the NHS in England. Women in five hospitals will be offered Down's screening as normal, with those who have a medium-to-high risk of Down’s syndrome then being offered NIPT as a second stage. If that confirms the woman is at high risk then a diagnostic test would be offered. The introduction of NIPT should mean that many fewer women will need invasive diagnostic tests.

The study aims to discover whether the test can perform in large numbers as accurately as previous studies have indicated and to determine what information midwives and women need to allow them to offer and choose the correct course of action for themselves. It will report in 2015.
The UK National Screening Committee (UK NSC) will look at whether NIPT can be introduced as part of the NHS Fetal Anomaly Screening Programme once the results of the study are known.

**Additional considerations**

As NIPT is a blood test and women have many blood tests in pregnancy, one of the important aspects of the new pilot is looking at ways to ensure women understand the test and the implications of the results. Only then will they be able to make an informed decision about whether the test is right for them. Furthermore, it will be important to ensure that test results can be provided in a timely fashion without causing anxiety or distress.

The current research shows that a percentage of the tests do not produce results at all because there is not enough of the baby’s DNA present in the mother’s blood sample. This outcome is more common among larger women and can range from as few as 1% to as many as 12% of results. This is why NIPT is likely to work best as a second stage of the screening process, for women already found to be at higher risk.