Key messages and Q&A document for women

What are the key messages for women from this RCOG Scientific Impact Paper?

- Non-invasive Prenatal Testing (NIPT) uses an ordinary maternal blood sample and carries no risk to the mother or her baby.

- NIPT uses fetal DNA from the placenta and is much more accurate than previous screening tests. It does not have the risks of the current invasive tests, which carry a small but significant risk of miscarriage and will be needed much less often.

- It is already being used for some conditions, however, it is becoming a primary screening method for chromosomal abnormalities, such as Down syndrome, in pregnancy.

- The high degree of accuracy, however, means that pregnant women may be informed of findings of uncertain significance, such as placental mosaicism, when there is a discrepancy between the chromosomal makeup of the cells in the placenta and the cells in the baby, which can result in a healthy pregnancy, however, in some pregnancies it can lead to complications.

- Excellent communication between women and health professionals is therefore essential to understanding the implications of the test results and families need pre-test information and support for informed decision making.

Q&A

1. What is the difference between invasive and non-invasive prenatal testing?

Invasive prenatal testing such as amniocentesis involves needles being inserted into the uterus to detect chromosomal abnormalities. This method carries a small but significant risk of miscarriage.

Non-invasive prenatal testing is a new technique that uses DNA from the baby present in a pregnant woman’s blood from early pregnancy and is a much more accurate form of screening for a number of fetal disorders, such as Down syndrome. It is obtained via an ordinary blood test from the mother.

2. What is non-invasive prenatal testing currently used for?

Obstetricians currently use non-invasive prenatal testing to guide women whose baby is at risk of haemolytic disease of the fetus and newborn (HDFN), a condition where antibodies develop in a pregnant woman’s blood and subsequently destroy the blood cells of the baby she is carrying.

It is also used for fetal sex determination in pregnancies at high sex-linked genetic risk, such as Duchenne muscular dystrophy, an inherited condition which affects the muscles, causing muscle weakness.

In some parts of the world and in the UK in the private sector, NIPT is now available to detect Down syndrome and other chromosomal abnormalities.

3. What type of conditions will non-invasive prenatal testing screen for?
Non-invasive prenatal testing is able to detect the baby’s blood group, sex-linked diseases, and chromosomal abnormalities. There is the potential for other genetic conditions to be determined in this way.

4. **How accurate is non-invasive prenatal testing?**

Non-invasive prenatal testing is highly accurate. However, possible sources of error include the test being undertaken too early (before 10 weeks), or due to maternal obesity.

Furthermore, the test result does not predict the severity of the abnormality and further testing may be necessary.

5. **Are there any risks to the mother and baby if non-invasive prenatal testing is conducted?**

Non-invasive prenatal testing carries no risk to mother or baby. The sample required is an ordinary blood test taken from the mother.

6. **At what stage of the pregnancy would non-invasive prenatal testing be conducted?**

Non-invasive prenatal testing should be conducted after 10 weeks of pregnancy. If the test is carried out before 10 weeks there may be insufficient fetal DNA circulating in the mother’s blood to carry out the test.

7. **What support is available?**

Following the non-invasive test, if the result shows an abnormality, the woman would be offered further testing to re-confirm the diagnosis. A team of obstetricians and midwives would provide the necessary counselling and advice for families.

8. **Where can I find more information about non-invasive prenatal testing?**

The RAPID research programme has a guide to non-invasive prenatal testing: [http://www.rapid.nhs.uk/guides-to-nipd-nipt/introduction/](http://www.rapid.nhs.uk/guides-to-nipd-nipt/introduction/)

Public Health England has further information on non-invasive prenatal testing for Down Syndrome: [http://fetalanomaly.screening.nhs.uk/programmestatements](http://fetalanomaly.screening.nhs.uk/programmestatements)

9. **Where can I find out more about current screening?**

Currently all pregnant women in the UK are offered a test for fetal anomalies, including Down Syndrome, as part of The NHS Fetal Anomaly Screening Programme (NHS FASP).

More information on the current fetal anomaly screening programme can be found on the NHS FASP website: [http://fetalanomaly.screening.nhs.uk/testsabout](http://fetalanomaly.screening.nhs.uk/testsabout).