Fact sheet **27**

NON-INVASIVE PRENATAL TESTING (NIPT)

This fact sheet talks about a screening test available in pregnancy called non-invasive prenatal testing (NIPT). It can screen for whether the baby has certain chromosome conditions.

IN SUMMARY

- A screening test in early pregnancy to detect Down syndrome and other chromosome conditions
- The test is safe and does not pose any risk to mother or baby
- NIPT, like all tests in pregnancy, is optional
- The accuracy of NIPT tests is high although not 100%
- A definite diagnosis of a chromosome condition in the baby can only be made following a prenatal diagnosis test like CVS or amniocentesis.

WHAT IS NON-INVASIVE PRENATAL TESTING (NIPT)?

NIPT is a test that uses a sample of the mother's blood during pregnancy to determine if the developing baby has certain chromosome conditions that can affect health and development. During pregnancy, some of the DNA from the baby (fetal DNA) crosses into the mother's bloodstream. This DNA carries the baby's genetic information. It is this fetal DNA that is tested and analysed during NIPT to check for certain chromosome conditions.

WHAT DOES THE NIPT TEST FOR?

Chromosomes are the packages which contain the body's DNA. Problems with the way your bodies develop or work can happen when there is too much or too little chromosome material.

The most commonly known chromosome condition in newborn babies is Down syndrome (also known as Trisomy 21) which is caused by having an extra copy of chromosome number 21 in each cell of the body.

NIPT tests for conditions in the baby where an entire extra copy of a chromosome is present or missing. Most NIPT tests will test for conditions including Down syndrome (Trisomy 21), Edward syndrome (Trisomy 18), Patau syndrome (Trisomy 13) and certain sex chromosome variations.

Women who are considering NIPT should be aware that not all tests look for the same chromosome conditions and it is important to talk to your health care provider about the specific conditions the baby is being tested for.

WHO MIGHT WANT TO HAVE NIPT?

The test might be useful when:

- A screening test during the pregnancy has indicated that the baby is at increased chance of having a chromosome condition
- An ultrasound indicates a concern with the baby's growth and/or development
- A previous pregnancy has been affected by a chromosome condition
- Mothers aged 35 years or more at the date of delivery who have a higher chance of having a baby with a chromosome condition

HOW ACCURATE IS NIPT?

NIPT is highly accurate for the chromosome conditions that it tests for. The accuracy of the test, however, is not 100%.

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There are also differences in the accuracy, depending on the chromosome condition. For example, the result for Down syndrome may be more accurate than for the other chromosome conditions tested for.

There is also the possibility that following testing, no results are given. This may be because there is not enough fetal DNA in the mother's blood, or there is difficulty finding fetal DNA in the mother's blood.

DOES A 'NORMAL' (LOW RISK) NIPT MEAN THAT THE BABY IS HEALTHY?

No test can guarantee that a baby will be healthy at birth. NIPT looks for a number of chromosome conditions including Down syndrome. NIPT does not analyse all of the baby's chromosomes and DNA, therefore, will not rule out other genetic, chromosome or other health conditions. The health professional providing information about NIPT is the best person to ask about the limits of testing.

HOW EARLY IN PREGNANCY CAN NIPT BE USED?

The test can be used as early as 10 weeks of pregnancy though this may differ between companies offering the test.

WHAT IF THE RESULT OF THE NIPT SHOWS THAT THE BABY MIGHT HAVE A CONDITION?

If the NIPT result shows that the baby is at high risk of having a chromosome condition, information about the condition will be provided. It is likely that your health professional will discuss confirming any concerning test results using invasive diagnostic testing procedures such as chorionic villus sampling (CVS) or amniocentesis.

CAN NIPT HARM ME OR MY BABY?

The test uses a sample of the mother's blood. The mother may experience some discomfort when undergoing a blood test and there is no risk to the baby.

WHO CAN I SPEAK WITH TO ORDER THIS TEST?

This test is available through most general practitioners (family doctors).

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