

NONINVASIVE PRENATAL TESTING: INFORMATION FOR PRENATAL CONSUMERS

Illinois Department of Public Health

DEFINITION

Noninvasive prenatal testing (NIPT) is a blood test for pregnant women which look for chromosome abnormalities in the baby, such as Down syndrome. NIPT examines the baby's chromosomes, cells that normally are in the mother's blood, and is a screening method to help identify some birth defects and chromosome abnormalities.

HOW IS NIPT DONE?

A small amount of blood is taken from a mother who is at least 10 weeks pregnant. The blood is sent to a laboratory for testing. The laboratory looks for the baby's chromosomes that are in cells circulating within the mother's blood. If there is a higher than expected amount of the baby's chromosomes present in the mother's blood, this suggests that the baby may have a chromosome abnormality. The test results are usually available to the mother's doctor within two weeks.

WHO SHOULD CONSIDER NIPT?

Currently, this testing is routinely offered to women with certain characteristics such as:

- Advanced maternal age (women 35 years of age and older)
- A woman who has previously given birth to a baby with a chromosome abnormality
- Women who are carriers of an X-linked recessive disorder
- An abnormal first trimester screen
- A family history of chromosome abnormalities
- An abnormal ultrasound of the baby

WHAT ARE THE RISKS?

This screening test poses no risks to the baby. Risks of testing may include bruising or soreness at the site of the blood draw on the mother.

WHAT WILL THE TEST RESULTS TELL ME?

The test results will tell pregnant women if they are at a higher risk of having a baby with a chromosome abnormality such as Down syndrome, trisomy 18, or trisomy 13, but the results do not tell women if their baby actually has one of these conditions.

WHAT DOES AN ABNORMAL NIPT RESULT MEAN?

An abnormal test suggests a woman is at a higher risk for having a baby with a chromosome abnormality, but it does not mean there is a definite problem with the pregnancy. Further detailed testing such as chorionic villus sampling or amniocentesis is recommended to see if a chromosome abnormality is present.

HOW ACCURATE ARE THE RESULTS?

NIPT is 98-99% accurate for determining if a baby possibly has Down syndrome or trisomy 18. NIPT is 91% accurate for determining if a baby possibly has trisomy 13. There is a small risk that the results will indicate a woman is at high risk for having a child with a chromosome abnormality when her baby is actually fine. It is for this reason that further testing is recommended to confirm the results of NIPT.

IMPORTANT FACTS

- NIPT is a simple blood test on the mother's blood.
- This screening test poses no threat to the mother or her baby.
- Prenatal screening is optional. Questions regarding screening should be discussed with a physician or genetic counselor.
- A normal screening result can be reassuring but it does not guarantee a healthy baby. Not all birth defects or genetic conditions are identified through screening.
- An abnormal screening test does not always mean there is a problem with the baby. Further testing options can be discussed with a physician or genetic counselor.

ADDITIONAL INFORMATION

Illinois Department of Public Health
Genetics/Newborn Screening Program
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<http://www.dph.illinois.gov/topics-services/life-stages-populations/genomics/prenatal-diagnostic-tests>