NONINVASIVE PRENATAL TESTING
INFORMATION FOR PREGNANT PEOPLE

ILLINOIS DEPARTMENT OF PUBLIC HEALTH
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DEFINITION

Noninvasive prenatal testing (NIPT) is a blood screening test for pregnant people. It helps to find some chromosome abnormalities in the baby, such as Down syndrome. NIPT looks at the pregnant person's blood for DNA that has shed off the placenta. This test can also be called noninvasive prenatal screening (NIPS) or cell-free DNA screening (cfDNA).

HOW IS NIPT DONE?

A small amount of blood is taken from a person who is at least 10 weeks pregnant. The blood is sent to a laboratory for testing. The laboratory looks for DNA that has shed off the placenta. A higher-than-expected amount of DNA markers from any of the baby's chromosomes means that the baby may have a chromosome abnormality. The test results are usually available to the obstetric care provider within two weeks.

WHO SHOULD CONSIDER NIPT?

✓ Currently, NIPT is routinely offered to people at least 10 weeks pregnant with a single or twin pregnancy.

× NIPT is not recommended for people carrying three or more babies. NIPT is not recommended for twin pregnancies where either twin is suspected to have a problem.

WHAT ARE THE RISKS?

NIPT poses no risks to the baby. Risks for the pregnant person may include bruising or soreness at the site of the blood draw.

WHAT DOES AN ABNORMAL NIPT RESULT MEAN?

An abnormal result means a person is at higher risk for having a baby with a chromosome abnormality such as Down syndrome, trisomy 18, or trisomy 13. An abnormal result does not mean there is a definite problem with the pregnancy. Diagnostic testing, such as chorionic villus sampling (CVS) or amniocentesis, is recommended to confirm the abnormal NIPT result.

There is a small chance for a non-reportable result due to a low "fetal fraction."

- This means that there wasn't enough of the baby's DNA in the sample to run the test.
- In this case, you may be able to redo the test one or two weeks later.

HOW ACCURATE ARE THE RESULTS?

- NIPT has a 98-99% detection rate for determining if a baby possibly has Down syndrome, trisomy 13, or trisomy 18. This means that 98-99% of pregnancies that actually have those conditions will test positive on NIPT.
- There is a small risk that the results will indicate a pregnant person is at high risk for having a baby with a chromosome abnormality when their baby is actually fine; this is called a false positive.
- The positive predictive value (PPV) represents the proportion of positive test results that are truly positive. It answers the question: "If my test is positive, what is the chance my baby is affected?" This varies based on the condition detected, and age of the pregnant person.
- This is why more testing is recommended to confirm the results of NIPT.

IMPORTANT FACTS

- NIPT is a simple blood test on the pregnant person's blood.
- NIPT poses no threat to the pregnant person or their baby.
- This screening test is available to all people at least 10 weeks pregnant.
- A normal screening result can be reassuring, but it does not guarantee a healthy baby. Not all birth defects or genetic conditions are identified by NIPT.
- If NIPT detects a chromosome abnormality, then additional diagnostic testing – such as amniocentesis or chorionic villus sampling – should be discussed with an obstetric care provider or genetic counselor.
- There is a small chance for a non-reportable result.
- NIPT is optional. Ask your obstetric care provider or genetic counselor for more information about screening.

ADDITIONAL INFORMATION

Illinois Department of Public Health
Genetics/Newborn Screening Program
535 W. Jefferson St., Second Floor
Springfield, IL 62761
217-785-8101
http://www.dph.illinois.gov/topics-services/life-stages-populations/genomics/prenataldiagnostic-tests

REFERENCES

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