NONINVASIVE PRENATAL TESTING
INFORMATION FOR OBSTETRIC CARE PROVIDERS

ILLINOIS DEPARTMENT OF PUBLIC HEALTH
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DEFINITION

Noninvasive prenatal testing (NIPT) is a screening method for detecting chromosome abnormalities in a fetus. NIPT screens for Down syndrome (trisomy 21), as well as the less common chromosome abnormalities, Patau syndrome (trisomy 13) and Edwards syndrome (trisomy 18). This blood test can also screen for sex chromosome abnormalities and the fetus's Rh factor. NIPT examines fetal DNA within the pregnant patient's blood, which has shed off the placenta. This test is also referred to as noninvasive prenatal screening (NIPS) and cell-free DNA screening (cfDNA).

HOW IS NIPT PERFORMED?

At 10 weeks gestation or later, a blood sample is taken from the pregnant patient and sent to a genetic testing laboratory for analysis. The laboratory compares the pregnant patient's DNA with the fetus's DNA. A higher-than-normal percentage of certain fetal DNA markers may suggest a chromosome abnormality.

WHEN IS NIPT RECOMMENDED?

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

Testing is not recommended for pregnancies involving three or more fetuses. Testing is not recommended for twin pregnancies with fetal demise, vanishing twin, or an ultrasound abnormality in either twin.

HOW DOES NIPT COMPARE TO AMNIOCENTESIS AND CHORIONIC VILLUS SAMPLING?

NIPT is the only one of these three tests offered to pregnant people that poses no physical risks to the pregnant person or fetus.

Amniocentesis and chorionic villus sampling (CVS) are both invasive prenatal tests that carry a small additional risk for miscarriage. The risk for a miscarriage with an amniocentesis is 0.1%, whereas the risk for a miscarriage with CVS is 0.2%. Both diagnostic tests can detect chromosome abnormalities, as well as other genetic conditions that may not be detectable by NIPT.

Amniocentesis: An invasive prenatal test where a fine needle is inserted into the abdomen of the pregnant person and a small sample of amniotic fluid is removed from the amniotic sac surrounding the fetus.

Chorionic villus sampling: An invasive prenatal test that removes a sample of chorionic villi cells from the placenta at a place where it attaches to the uterine wall.

ADVANTAGES OF NIPT

- 98-99% of pregnancies that have a fetus with trisomy 21, trisomy 13, or trisomy 18 will test positive with NIPT.
- There is no risk of miscarriage with NIPT.
- There are no physical risks to the fetus or pregnant person.

LIMITATIONS OF NIPT

- The false-positive rate for detecting trisomy 21 and trisomy 18 is approximately 1 in 500 or 0.2%.
- The detection rate for sex chromosome abnormalities is 79-92%.
- The positive predictive value (PPV) refers to 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?" It varies with factors such as the age of the pregnant person, and the chromosome condition detected.
    - Find the NIPT PPV Calculator here.
- If NIPT detects a chromosome abnormality, additional invasive diagnostic testing, such as amniocentesis or chorionic villus sampling, is needed.
- There is the possibility of a "non-reportable" result, which happens in about 1 in 67 or 1.5% of samples. This means that there was not enough fetal DNA circulating in the pregnant person's blood to run the test.
    - This can happen due to a variety of factors, including a high body mass index (BMI), lower gestational age, maternal medication use (Lovenox in particular).
    - If a non-reportable result occurs, a re-draw can be done a week or two later.

IMPORTANT FACTS

- Obstetric care providers should discuss the option of prenatal screening with their eligible patients.
- Eligible patients should be given information about the advantages and limitations of screening.
- Not every blood draw site has the special tubes needed for NIPT. Coordinate with the blood draw site.
- 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.

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

Skotko, Allyse, M. A., Bajaj, K., Best, R. G., Klugman, S., Leach, M., Meredith, S., Michie, M., Stoll, K., & Gregg, A. R. (2019). Adherence of cell-free DNA noninvasive prenatal screens to ACMG recommendations. *Genetics in Medicine*, 21(10), 2285–2292. https://doi.org/10.1038/s41436-019-0485-2

Screening for Fetal Chromosomal Abnormalities. (2020). *American College of Obstetricians and Gynecologists Practice Bulletin*, *136*(4), 1–22.

To calculate PPV value: https://www.perinatalquality.org/vendors/nsgc/nipt/
Provided by the National Society of Genetic Counselors and Perinatal Quality Foundation