

NONINVASIVE PRENATAL TESTING: INFORMATION FOR PHYSICIANS

Illinois Department of Public Health

DEFINITION

Noninvasive prenatal testing (NIPT) examines fetal DNA within the mother's blood and is a screening method for detecting chromosome abnormalities in a developing fetus. NIPT screens for trisomy 21 (Down syndrome), as well as two other less common chromosome abnormalities, trisomy 13, and trisomy 18. This blood test can also screen for sex chromosome abnormalities and the fetus's Rh factor.

HOW IS NIPT PERFORMED?

At ten weeks gestation, a blood sample is taken from the mother and sent to a reproductive testing laboratory for analysis. The laboratory compares the mother's DNA with the fetus's DNA and examines the chromosomes present. A higher than normal percentage of chromosomes may suggest a chromosome abnormality.

WHEN IS NIPT RECOMMENDED?

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

- Advanced maternal age (35 years and older)
- A woman who has previously given birth to a baby with trisomy 21, trisomy 18, or trisomy 13
- A woman who is carrier of an X-linked recessive disorder
- An abnormal serum screen
- A family history of chromosome abnormalities
- An abnormal prenatal ultrasound

HOW DOES NIPT COMPARE TO AMNIOCENTESIS AND CHORIONIC VILLUS SAMPLING?

Currently, NIPT is the only one of these 3 tests offered to pregnant woman that poses no physical risks to the mother or fetus.

An amniocentesis and chorionic villus sampling (CVS) are both invasive prenatal tests that carry a 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 screenings can detect various birth defects, such as abnormal chromosome abnormalities, as well as other genetic diseases. These tests are only recommended for women with certain health characteristics as indicated above.

Amniocentesis: An invasive prenatal test where a fine needle is inserted into the abdomen of the mother 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 or trisomy 18 can be detected with NIPT
- There is no risk of miscarriage with NIPT
- There are no physical risks to the fetus or mother

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%
- If NIPT detects a chromosome abnormality, additional invasive testing, such as amniocentesis or chorionic villus sampling, is needed

IMPORTANT FACTS

- Prenatal screening is optional. Questions regarding the advantages and limitations of 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.

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/prenatal-diagnostic-tests>