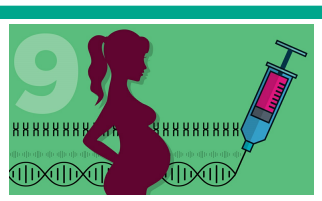

Non-invasive Prenatal Genetic Testing (NIPT)

THE AMERICAN SOCIETY OF HUMAN GENETICS

GENOMIC MEDICINE EDUCATION CONSORTIUM

National DNA Day APRIL 25 15th Anniversary

News you can use

What is it?

Non-invasive prenatal testing (aka: cell-free DNA screening) uses cell-free DNA floating in maternal blood to screen for fetal genetic conditions such as the common trisomies (i.e. Down syndrome) and deletion/duplication syndromes.

How does it work?

DNA fragments are isolated, sequenced, identified, and counted.

Each lab uses a slightly different approach and algorithm for analysis.

Labs may differ in the conditions they test for, turn-around time, and "no result" rates, but all have similar sensitivity and specificity.

Why it's important now

NIPT is more accurate, with fewer false positives for the most common trisomies, than other screening tests - leading to fewer invasive procedures.

Technology advances in NIPT can potentially lead to a screen for many different conditions.

Additional resources

ASHG's Prenatal Genetic Screening
www.pathlms.com/ashg/courses/4595

Genetic support foundation:
geneticssupportfoundation.org/videos

Coalition for access to prenatal screening
capsprenatal.com

In the clinic

NIPT is not diagnostic, but has higher sensitivity and specificity than traditional prenatal screens with high positive predictive values in high risk populations.

Women who test positive should be offered an invasive test (CVS or amnio) which have a small risk for miscarriage.

Women interested in more comprehensive analysis should consider diagnostic testing.

Who might be impacted?

Any pregnant person should be offered prenatal screening and/or testing, including NIPT, with genetic counseling to understand the risks, benefits and limitations of screening and diagnostic testing.

- ACOG/SMFM, Practice Bulletin, No. 162 and 163

What will it tell me?

Positive NIPT result indicates a high risk for a chromosome abnormality and should be followed up with a diagnostic test.

Negative NIPT result does not guarantee a normal pregnancy outcome.

A "no result" is possible and testing may have to be repeated.

NIPT screens for a limited number of conditions. More comprehensive testing is available through CVS/amnio and chromosome microarray analysis.

Did you know...

DNA sequencing now allows for testing for genetic variants in a fetus using DNA isolated from maternal blood?