NIPT now endorsed by ACOG/SMFM for all pregnant mothers regardless of age or risk[1]

- Also known as cell-free DNA (cfDNA) screening, noninvasive prenatal testing (NIPT) is a prenatal aneuploidy screening test
- Can noninvasively screen for the presence of fetal chromosomal aneuploidies as early as week 10

Noninvasive

NIPT is as safe and simple as a blood draw.

Accurate

Gain insights into genetic health risks with >99% accuracy for trisomies 21, 18, and 13.[2]

NIPT can also screen for[3]*:

- Common aneuploidies (trisomies 21, 18, and 13) and rare autosomal aneuploidies
- Partial duplications and deletions ≥7 Mb for all autosomes
- Copy number variants
- Sex chromosome aneuploidies

NIPT combined false positive rate for trisomies 21, 18, and 13[4]: **0.13%**

NIPT offers[1]:

- Highest reported detection rate for Down syndrome
- Low reported false positive rate for Down syndrome
- Ability to screen for additional chromosomal conditions

*NIPT offerings vary.

ACOG=American College of Obstetricians and Gynecologists; SMFM=Society for Maternal-Fetal Medicine.

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Early

NIPT can be performed as early as week 10 of gestation and until term, giving it the broadest screening window of any prenatal screening test.[1]

Supported by society guidelines[1,5]

> **“Screening ([...] cell-free DNA screening [NIPT]) and diagnostic testing [...] should be discussed and offered to all patients early in pregnancy regardless of maternal age or baseline risk.”**
>
> **“Cell-free DNA [NIPT] is the most sensitive and specific screening test for the common fetal aneuploidies (trisomies 21, 13, and 18) and can be performed at any time after 9-10 weeks of gestation.”**
>
> —ACOG/SMFM clinical management guidelines for obstetricians and gynecologists[1]

> **“There is now increasing evidence to show that the testing can also be applied to women with average risk… The following protocol options are currently considered appropriate: cfDNA screening as a primary test offered to all pregnant women.”**
>
> —International Society of Prenatal Diagnostics (ISPD)[5]

What are the limitations of NIPT?

NIPT based on cell-free DNA analysis from maternal blood is a screening test. False positive and false negative results do occur. Test results must not be used as the sole basis for diagnosis. Further confirmatory testing is necessary prior to making any irreversible pregnancy decision. A negative result does not eliminate the possibility that the pregnancy has a chromosomal or subchromosomal abnormality. This test does not screen for birth defects such as open neural tube defects, or other conditions, such as autism. Some NIPT tests do not screen for polyploidy (eg, triploidy) or single gene disorders. There is a small possibility that the test results might not reflect the chromosomal status of the fetus, but may instead reflect chromosomal changes in the placenta (ie, confined mosaicism [CPM]) or in the mother that may or may not have clinical significance.

References:

1. Rose NC, Kaimal AJ, Dugoff L, Norton ME; American College of Obstetricians and Gynecologists' Committee on Practice Bulletins—Obstetrics; Committee on Genetics; Society for Maternal-Fetal Medicine. Screening for fetal chromosomal abnormalities. *Obstet Gynecol.* 2020:136(4):e48-e69. **2.** Data on file, Illumina, 2019. **3.** Pertile MD, Halks-Miller M, Flowers N, et al. Rare autosomal trisomies, revealed by maternal plasma DNA sequencing, suggest increased risk of feto-placental disease. *Sci Transl Med.* 2017;9(405):eaan1240. doi: 10.1126/scitranslmed.ann1240. **4.** Gil MM, Accurti V, Santacruz B, Plana MN, Nicolaides KH. Analysis of cell-free DNA in maternal blood in screening for aneuploidies: updated meta-analysis. *Ultrasound Obstet Gynecol.* 2017:50:302-314. **5.** Benn P, Borrell A, Chiu RW, et al. Position statement from the Chromosome Abnormality Screening Committee on behalf of the Board of the International Society for Prenatal Diagnosis. *Prenat Diagn.* 2015;35(8):725-734.

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