

[Positive & Negative Predictive Value Calculator for Providers](#)

This [calculator](#) will allow you to estimate the Positive Predictive Value (PPV) and Negative Predictive Value (NPV) of cell-free DNA (cfDNA), also known as noninvasive prenatal testing (NIPT), based on estimates of population prevalence or by entering your own prevalence numbers. The results of this calculator only apply to patients who have a result from cfDNA.

Professional Guidelines and Policy Statements on the use of cfDNA

Noninvasive Prenatal Testing (NIPT) through analysis of cell free DNA (cfDNA) was first introduced in 2011 as a screening test for Down syndrome (trisomy 21). The list of conditions that cfDNA can screen for has grown to include trisomy 18, trisomy 13, sex chromosome conditions, and several microdeletion conditions. Several professional organizations have published practices guidelines and statements regarding cfDNA including:

- The American College of Obstetricians and Gynecologists (ACOG)
- The American College of Medical Genetics and Genomics (ACMG)
- The Society for Maternal-Fetal Medicine (SMFM)
- The International Society for Prenatal Diagnosis (ISPD)
- The National Society of Genetic Counselors (NSGC)
- The European Society of Human Genetics (ESHG)
- The American Society of Human Genetics (ASHG)

Group	First-tier test?	Sex chromosomes?	Microdeletions and microduplications?	Multiple gestations?
ACOG	No	If requested by patient	No	No
ACMG	Yes	Let patient know it’s available	Let patient know it’s available	Check with lab first
SMFM	No	n/a	n/a	n/a
ISPD	Yes?	Should be offered	Yes	Yes
NSGC	No	n/a	n/a	n/a
ASHG/ESHG	Yes	No	No	n/a

The key points from all recommendations are summarized below.

ALL guidelines advocate the following:

- Patients should have the opportunity to make an informed choice to decline or accept testing
- Pre and post-test counseling regarding the overall benefits, risks, and limitations is essential

Other important guideline information:

- In 2016, ACMG updated their cfDNA policy: ‘New evidence strongly suggests that NIPS can replace conventional screening for Patau, Edwards, and Down syndromes across the maternal age spectrum, for a continuum of gestational age beginning at 9-10 weeks, and for patients who are not significantly obese.’
- When comparing cfDNA to other screening methods, AGOG and SMFM state ‘no one screening test is superior to other screening tests in all test characteristics. Each test has relative advantages and disadvantages.’
- The ISPD discourages the use of maternal age as an indicator of prior risk and states that cfDNA is one possible primary screening approach for women, regardless of age or other risk factors.

Stay up-to-date with what the professional organizations are saying about cfDNA at the links below: