Noninvasive prenatal testing (NIPT) society statements table

Learn about recent society statements on NIPT (ie, cell-free DNA-based testing)

	ACOG/SMFM*	ISPD†	NSGC‡	ACMG§
Date of latest publication	October 2020[1]	August 2015[2], October 2020[3]	April 2021[4]	December 2022[5]
Gestational age	As early as 9-10 weeks–term	As early as 9–10 weeks	Not specified	10 weeks–term
Eligible patients for NIPT screening for common aneuploidies (trisomy 21, 18, 13)	All pregnant people	All pregnant people	All pregnant people	Preferred screening method for all pregnant people
Sex chromosome aneuploidy analysis	Not specified	Give option to separately accept or reject sex chromosome analysis	Consider test's positive predictive value	Should be offered to all patients with singleton gestations
Aneuploidies other than trisomy 21, 18 and 13, large copy number variation (CNV), and microdeletions	Not recommended	Testing should be limited to clinically significant disorders with a well-defined severe phenotype. Cumulative false positive rate (FPR) needs to be low. Individual positive predictive value (PPV) needs to be compatible with other disorders where prenatal screening is offered.	Consider test's positive predictive value	Suggests screening for 22q11.2 deletion syndrome be offered to all patients. No recommendation for routine screening for rare aneuploidies or other CNVs. CNV screening could be offered based on pregnancy or family history.
Multiples	Yes	Yes	Not specified	Preferred screening method for twin gestations, including vanishing twin

*American College of Obstetricians and Gynecologists/Society for Maternal-Fetal Medicine
†International Society for Prenatal Diagnosis
‡National Society of Genetic Counselors
§American College of Medical Genetics and Genomics

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Reporting fetal fraction	Preferred	No recommendation	No recommendation	No recommendation
Follow up after positive NIPT	Chorionic villus sampling (CVS) or amniocentesis	CVS or amniocentesis. CVS should consider potential for confined placental mosaicism (CPM). Amniocentesis is true indicator of fetal karyotype.	Confirmatory diagnostic testing	Prenatal or postnatal confirmation
Test failure	Recommend further genetic counseling, offering comprehensive ultrasound evaluation and diagnostic testing because of increased risk for aneuploidy	Added risk for trisomy 18, trisomy 13, monosomy X, and triploidy. Prior to redraw, consider cfDNA vs alternate testing.	Post-test genetic counseling; confirmatory diagnostic testing may be indicated.	Optimal management is unclear. Repeat testing may provide result.
Technology	Several molecular methods	Shotgun sequencing; targeted sequencing with counting; single nucleotide polymorphisms (SNPs)	Not specified	Not specified

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References

1. American College of Obstetricians and Gynecologists. Practice Bulletin No. 226: Screening for fetal chromosome abnormalities. *Obstet Gynecol.* 2020;136(4).
2. Benn P, Borrell A, Chiu R, 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.
3. Palomaki GE, Chiu RWK, Pertile MD et al. International Society for Prenatal Diagnosis (ISPD) Position Statement: Cell free (cf)DNA screening for Down syndrome in multiple pregnancies. *Prenat Diagn.* 2020 Oct 5. doi: 10.1002/pd.5832. Online ahead of print
4. Prenatal cell-free DNA screening. National Society of Genetic Counselors Web site. https://www.nsgc.org/Policy-Research-and-Publications/Position-Statements/Position-Statements/Post/prenatal-cell-free-dna-screening-1. Accessed March 28, 2023.
5. Dungan JS, Klugman S, Darilek S et al. Noninvasive prenatal screening (NIPS) for fetal chromosome abnormalities in a general-risk population: An evidence-based clinical guideline of the American College of Medical Genetics and Genomics (ACMG). Genet Med. 2023 Feb. 25(2):100336. doi: 10.1016/j.gim.2022.11.004. Epub 2022 Dec 16.

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