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Noninvasive prenatal testing (NIPT) society statements table

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

	ACOG/SMFM*	ISPD†	NSGC‡	ACMG§
Date of latest publication	May 2016[1]	April 2015[2]	October 2016[3]	July 2016[5]
Date of initial publication (if applicable)	NA	NA	January 2013[4]	April 2013[6]
Gestational age	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 women	High-, low-, or average-risk population	High-, low-, or average-risk population	High-, low-, or average-risk population
Sex chromosome aneuploidy analysis	Yes	Give option to separately accept or reject sex chromosome analysis	Yes to monosomy X with whole-genome sequencing (WGS). Others not addressed.	Yes
Microdeletions and expanded trisomies	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.	Not studied at time of publication. NIPT will likely expand to include additional chromosomal abnormalities and/or microarray analysis as future studies support clinical validity of such results.	Not recommended for expanded trisomies and genome-wide copy number variants (CNVs). Patients should be informed of availability of expanded panels.
Multiples	Not recommended	Twins: performance similar to singletons. When fetal fraction is measured for each fetus and lowest value is used to decide on interpretability, failure rate will be higher than in singletons.	Not specified	Contact specific laboratories for platforms and clinical validation studies

*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	No recommendation	No recommendation	No recommendation	Yes
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.	Not specified	CVS or amniocentesis
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.	No recommendation	Laboratories should specify a reason for a test failure. Women who receive a test failure due to low fetal fraction should be offered diagnostic testing. For obese patients, recommend alternative screening methods.
Technology	Three molecular methods	Shotgun sequencing; targeted sequencing	WGS; targeted sequencing	WGS; targeted sequencing

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References

1. American College of Obstetricians and Gynecologists. : Screening for fetal aneuploidy. *Obstet Gynecol.* 2016;127(5):e123-137.
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. Giammarco M. Prenatal cell-free DNA screening. National Society of Genetic Counselors Web site. http://www.nsgc.org/p/bl/et/blogaid=805#.WCTELTNRftw.linkedin. Published October 11, 2016. Accessed November 21, 2016.
4. Devers PL, Cronister A, Ormond KE, et al. Noninvasive prenatal testing/noninvasive prenatal diagnosis: the position of the National Society of Genetic Counselors. *J Genet Couns*. 2013;22:291-295.
5. Gregg AR, Skotko BG, Benkendorf JL, et al. Noninvasive prenatal screening for fetal aneuploidy, 2016 update: a position statement of the American College of Medical Genetics and Genomics. *Genet Med*. 2016;18(10):1056-1065.
6. Gregg AR, Gross SJ, Best RG, et al. ACMG statement on noninvasive prenatal screening for fetal aneuploidy. *Genet Med.* 2013;15(5):395-398.

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