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Rethinking the Burden of Traditional Informed Consent Prior to Prenatal Genetic Screening

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Appendix A.
Components of Informed Consent for Prenatal Genetic Screening, per Selected Professional Society Guidelines

	ISPD	ACOG	ACMG
		Pretest and posttest counseling is essential.	As with all prenatal screening and diagnostic testing, appropriate pre- and post-test counseling is an integral aspect of informed decision-making. Providers should emphasize the informed and shared decision-making that surrounds screening and/or testing in a supportive environment. Insufficient pre- and post-test counseling, rather than NIPS itself, represents potential and avoidable harms.
Exploration of personal values, followed by acceptance or rejection of prenatal screening		The ethical principles of autonomy, beneficence, and nonmaleficence require that the unique circumstances of each patient be considered before performing this type of testing. In some circumstances, this type of pretest counseling may require the assistance of an obstetrician–gynecologist or other health care provider with expertise in genetics.	We affirm the principles of counseling for NIPS that were highlighted in the 2016 position statement, including providing up-to-date, balanced, and accurate information and personalized, patient-centered counseling.
	Aim of prenatal screening: To provide pregnant individuals/expectant couples with more information about their unborn baby's health; to promote reproductive autonomy	The purpose of pretest counseling is to inform pregnant patients about chromosomal disorders, provide information regarding their specific risk of carrying a fetus with a chromosomal abnormality, review their relevant personal and family history.	
	Testing is optional: Assess the pregnant person's preferences and values, ascertain acceptance or rejection of screening; provide alternative screening methods if relevant.	After review and discussion, every patient has the right to pursue or decline prenatal genetic screening and diagnostic testing.	Pretest counseling should include a discussion of the optional and screening nature of NIPS.
Information provision, tailored to individual's information needs; followed by choice regarding participation in screening and range of conditions included (e.g., fetal sex)			
	Define screening: distinguish from diagnostic test in accuracy and range and pregnancy risks. Advise need for diagnostic confirmation if high chance result	Prenatal genetic screening (serum screening with or without nuchal translucency [NT] ultrasound or cell-free DNA screening) and diagnostic testing (CVS or amniocentesis) options should be discussed and offered to all pregnant patients regardless of age or risk for chromosomal abnormality.	Patients must receive thorough pretest counseling that details the benefits, limitations, and risks of NIPS, particularly highlighting the screening nature of the test, the possibility for false-positive results and the appropriate follow-up of results.
	Clinical features and variability of conditions: Consent for SCA and expanded NIPT should be discussed separately to consent for the common autosomal trisomies, due to differences in clinical implications and NIPT performance. Information should be tailored to information needs and the health literacy of each person. If MMS are being screened this should be discussed	Discuss the risks, limitations, and benefits of available testing options. Patients who prefer comprehensive prenatal detection of as many chromosomal aberrations as possible should be offered diagnostic testing and CMA.	Pretest counseling should include the types of conditions that can and cannot be screened for.
	Explain that both maternal and fetal DNA are analyzed, which can lead to maternal incidental findings such as chromosome conditions or neoplasia; tailored to individuals information needs		Because of the methodology of NIPS, there is the potential to incidentally identify certain maternal conditions. This includes the identification of maternal chromosome imbalances and unsuspected maternal malignancies.
	Sensitivity: Detection rate. False positive rate and confirmation of abnormal results: Possibility of a ‘false alarm’ exists for any screening test; hence need to have confirmatory diagnostic testing before management decisions	Has the potential for false-positive and false-negative results. Cell-free DNA testing is not equivalent to diagnostic testing.	Recommendation for confirmation of any abnormal results. Previously unknown parental chromosome abnormalities may be uncovered in mosaic or complete states, eg, 22q11.2DS
	Positive and negative predictive values: Chance of affected pregnancy after a low or high chance result	With reference to each patient’s specific a priori risk, the patient should be informed of the meaning and significance of positive, negative, or indeterminate test results, as well as results that are normal but may have variable phenotypes. This discussion of the positive predictive value and negative predictive value of the test result facilitates a discussion of the potential need for follow-up diagnostic testing.	Pretest counseling should include information about positive and negative predictive values.
	Limitations: Does not screen for every chromosomal or genetic condition. May result in a ‘no call’ result		Pretest counseling should include the types of test results (including no-call results and incidental findings) that can be received.
	Incidental findings: Maternal malignancy (1 in 10,000), maternal chromosome conditions, confined placental mosaicism	The potential for the discovery of variants of uncertain significance should be addressed with patients; such variants may prompt further testing or collecting of additional data from other family members or even necessitate long-term follow-up.	
Practical logistics			
	Timing, description of required blood draw	Although many genetic tests can be performed using a routine blood sample, such tests should not be adopted into routine practice without patient consent	
	Complementary role of ultrasound: Role of first trimester ultrasound to ensure accurate dates, detection of multiple pregnancy, live pregnancy prior to NIPT early detection of structural anomalies		
	Costs: Financial disclosure	Although cost should not be a driving factor in whether a medically indicated test is offered to a patient, many genetic tests are expensive and may not be covered by a patient’s insurance. Therefore, patients should be encouraged to discuss their eligibility for coverage with their insurance providers.	
	Reporting format: When and how their results will be provided	Patients also should be given a reasonable time frame within which they can expect to be informed about their test results, and they should be encouraged to call if they have not received their results at the end of that period.	