Genetic Counseling for Patients Considering Screening and Diagnosis for Chromosomal Abnormalities

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KEYWORDS

- Genetic counseling • Chromosome abnormalities • Prenatal care
- Noninvasive prenatal screening • Maternal serum multiple marker screening
- Chorionic villi sampling • Amniocentesis

KEY POINTS

- With the introduction of cell-free DNA (cfDNA) screening for fetal aneuploidy, as well as chromosomal microarray for prenatal diagnostic testing, options for pregnant women have become increasingly complex.
- Discussions regarding options for prenatal testing for aneuploidy should occur prior to any testing and need not be lengthy or complex but should include pertinent risks and benefits of each alternative test.
- It is also important that the family history be assessed so that a focus on aneuploidy screening does not occur at the expense of missing a serious condition in a family that warrants evaluation and formal genetic counseling.
- There is no single screening or diagnostic test option that is the right choice for all patients; patient decisions should be based on each individual woman's values and preferences after a discussion of all options.

INTRODUCTION

With the advent of cfDNA screening for fetal aneuploidy, and the transition of chromosomal microarray from the pediatric clinic to the prenatal setting, testing options for pregnant women have entered a new era, and the number of testing options is overwhelming. Although the continued expansion of available tests offers expectant parents more options than ever before, it becomes paramount to equip prenatal care providers with the resources and support they need to assure their patients are

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counseled adequately to make informed decisions about testing that best satisfies their needs. Ideally, as a result of genetic counseling, patients should not only be educated about the details of the testing protocols but also feel that their values and concerns have been heard and their provider understands and respects their decisions. To achieve this goal requires excellent pretest counseling that encompasses the components outlined in this article.

Professional societies, including the American College of Obstetricians and Gynecologists, agree that screening and invasive diagnostic testing for aneuploidy should be available to all women, regardless of maternal age.[1,2] The offer of prenatal testing requires a discussion of the risks and benefits of invasive testing compared with screening tests, including the potential for a false-positive result (the false-positive rate) as well as the possibility for a false-negative result and how many chromosomally abnormal fetuses will be detected (the detection rate). In addition, the detection rate for aneuploidies other than Down syndrome and the type and prognosis of the aneuploidies likely to be missed by different screening tests should be discussed. The practitioner providing this information should be familiar with these details and able to answer any questions that arise. The differences between screening and diagnostic testing should be discussed with all women; an individual's decision as to whether to have screening or diagnostic testing, or any testing at all, is based on many factors, including the risk that the fetus will have a chromosomal abnormality, the risk of pregnancy loss from an invasive procedure, and the consequences of having an affected child. Studies that have evaluated women's preferences have shown that women weigh these potential outcomes differently.[3] The decision to have invasive testing should take into account these preferences and the offer of an invasive test should not be based solely on age. It is generally agreed that maternal age of 35 years alone should no longer be used as a threshold to determine who is offered screening versus who is offered invasive testing.[1–4]

Since the introduction of amniocentesis, prenatal genetic testing guidelines have focused on identifying patients at increased risk of giving birth to an infant with Down syndrome or another chromosomal abnormality, for whom invasive diagnostic testing should be recommended. Although initially advanced maternal age was the only recognized risk factor, identification of serum and ultrasonographic markers that can better estimate the risk of an affected fetus has led to the incorporation of screening into routine prenatal care for women of all ages.[5–9] Introduction of cfDNA testing has intensified the complexity of prenatal testing decision making.

PRETEST COUNSELING

Discussions regarding options for prenatal testing for aneuploidy should occur prior to any screening or diagnostic testing and typically occur in the context of routine prenatal care. The conversation usually includes each woman's provider; this may be a midwife, nurse practitioner, family practitioner, generalist obstetrician, or perinatologist. A discussion of testing options should begin with the reminder that most babies are born healthy but that all fetuses have an estimated 3% to 4% chance of being born with a birth defect or intellectual disability.[10] Some women have risk factors, including maternal age, family history, underlying maternal medical conditions, and/or environmental exposures, that affect their a priori risks for specific conditions. Although there is no test that can identify all conditions, prenatal screening and diagnostic tests can shed light on some conditions. Some risk factors are not addressed by routine prenatal genetic testing, however, and patients with a family history of a genetic disorder should be referred for formal genetic counseling.