PATIENT HANDOUT

SHOULD I HAVE ULTRASOUND AND cfDNA TESTING?

Society for Maternal-Fetal Medicine

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The American College of Obstetricians and Gynecologists (ACOG) and the Society for Maternal-Fetal Medicine (SMFM) recommend that all women be offered the option of screening or diagnostic testing for fetal genetic conditions. Diagnostic tests are used to determine if a fetus *has* a specific genetic condition. Examples include chorionic villus sampling (CVS) and amniocentesis. Screening tests are used to evaluate whether there is a low risk or high risk that a fetus *may have* a genetic condition. These tests are not 100% accurate in determining whether a fetus actually has the disorder; more testing is needed to answer that question. Traditional screening with maternal blood testing and nuchal translucency (NT) measurement (a first-trimester ultrasound measurement of the area behind the fetal neck) is still the most appropriate option for low-risk patients.

For women at higher risk for common fetal aneuploidies (fetal chromosomal abnormalities such as trisomy 21 or Down syndrome), cell-free DNA (cfDNA) screening may be a more accurate screening option. cfDNA refers to small fragments of DNA found in the maternal blood during pregnancy. Most cfDNA fragments come from the placenta. These fragments can be tested for common chromosomal disorders in a developing fetus during pregnancy. Although cfDNA testing is most useful in women at higher risk, after appropriate genetic counseling regarding its benefits and limitations, cfDNA screening is an acceptable strategy for any woman, regardless of risk.

Q What is the role of nuchal translucency measurement if I am planning to have cfDNA screening or already had it done with a low-risk result?

CfDNA screening is very accurate in detecting trisomies 21, 18, and 13 and potentially some sex chromosome abnormalities. However, it does not provide information about additional chromosomal abnormalities that might be found with other methods of screening or diagnostic testing. An increased NT is associated with these additional chromosomal abnormalities, as well as with fetal structural problems and a variety of other genetic conditions, but its usefulness as a screening test for these conditions is limited. For this reason, if you are having cfDNA screening, NT measurement is not required to calculate the risk for a chromosomal abnormality.

First-trimester NT measurement can be useful if you are considering cfDNA screening. Doing a first-trimester NT measurement before you have the cfDNA screening, especially if you are at higher risk of

a fetal chromosomal abnormality, gives you the option of moving directly to diagnostic testing instead of cfDNA screening if an enlarged NT is found.

If you have already undergone cfDNA screening and received a low-risk test result, the benefit of also doing first-trimester NT measurement as a screening test for chromosomal abnormalities or structural problems is limited. Therefore, a first-trimester ultrasound solely for the purpose of NT measurement is not recommended. However, a first-trimester ultrasound examination may be performed for other purposes, such as confirming dating of your pregnancy.

Q Should I consider cfDNA screening if I am otherwise low-risk but "soft markers" are seen on an ultrasound exam?

Soft markers are ultrasound findings that are not structural abnormalities but might be clues to slightly increased risk for a fetal chromosomal abnormality. In general, isolated soft markers have limited usefulness for detecting a chromosomal abnormality in low-risk patients. In women who have already had traditional chromosome abnormality screening with low-risk results, the risk of trisomy 21 typically remains low even in the presence of an isolated soft marker, so more testing is usually not recommended. If you have already had a low-risk cfDNA screening test result and an isolated soft marker is seen on ultrasound, your risk remains low and many obstetric care providers will consider this ultrasound finding as a normal variant with no further testing needed.

Q If a fetal structural abnormality is detected by ultrasound, should I have cfDNA screening?

The presence of fetal structural abnormalities significantly increases the risk that a fetal chromosomal abnormality is present. Although cfDNA is an accurate screening test for common chromosomal abnormalities that may be present when a structural defect is detected (such as trisomy 21, 18, and 13), some fetuses with structural abnormalities may have chromosomal abnormalities that are not detectable by cfDNA screening. One study suggests that if a fetus has a structural defect and the cfDNA screening test result is low-risk, there is still a 1-in-15 risk that the fetus may have a genetic abnormality that was not detected by cfDNA. For this reason, all women in whom a structural abnormality is seen on an ultrasound exam are generally offered diagnostic testing. If you decline diagnostic testing, cfDNA screening can be offered as an alternative. You should be aware that your fetus may have a genetic abnormality that cfDNA screening cannot identify.

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