PATIENT HANDOUT

Prenatal Screening Using Cell-Free DNA

-What is Cell-Free DNA (cfDNA)?

CfDNA, also known as Cell-Free Fetal DNA or Non-Invasive Prenatal Testing (NIPT), refers to small fragments of DNA found in the mother's blood during pregnancy. Most of these DNA fragments come from the placenta and can be used to test the baby for common chromosomal disorders during pregnancy. When an extra chromosome is identified, the finding is known as a trisomy. CfDNA screens for trisomy 21, 18, or 13 and for missing /extra genetic material in the sex chromosomes X and Y. Sometimes other rare chromosome problems are also identified.

How accurate is cfDNA screening?

CfDNA screening is most accurate for diagnosing Trisomy 21 (Down Syndrome) and slightly less accurate at diagnosing other trisomies and sex chromosome disorders. There are several factors that influence the accuracy of cfDNA screening, including the women's baseline risk factors for having a baby with a trisomy. The chance of having a trisomy is most dependent on the woman's age at the time of screening. For example, a 40 year old woman has a 1% (1 in 100) chance of having a baby with Down Syndrome and a 30 year old woman has a 0.1% (1 in 1000) chance of having a baby with Down Syndrome. This means that the test is most accurate for women over the age of 35 who have a higher baseline risk of having a baby with a chromosomal abnormality. Women younger than 35 at delivery have a lower baseline risk and a high chance of a false-positive test result.

What does it mean to have a POSITIVE cfDNA screen?

CfDNA screening is not a diagnostic test. A positive result DOES NOT MEAN your baby definitely has a chromosome problem. A positive cfDNA result does mean that you have a higher chance of a chromosome problem. If you have a positive test result, you will be referred to a maternal-fetal medicine specialist, geneticist, and/or genetic counselor to review your results and offer additional testing if necessary. A diagnostic test may be offered such as Chorionic Villous Sampling (CVS) or Amniocentesis, to determine the actual fetal chromosomes.

What if my cfDNA screen doesn't give a test result?

Occasionally cfDNA screening will not be able to give a test result. This can happen when there is not enough DNA in the mother's blood or because there is a problem interpreting the result at the lab. The most common reasons for no test result are that the mom's blood was drawn before 10 weeks of pregnancy, the mom weighs over 250 pounds, or the baby does in fact have a chromosomal abnormality. A repeat test may be suggested by your doctor based on your age and risk factors. There is a chance that you might not get a result from the repeat test.

What are other limitations of cfDNA screening?

- At this time, cfDNA only evaluates for the three common trisomies and fetal sex chromosome abnormalities. It does not identify other birth defects.
- CfDNA is less accurate in twin pregnancies. No information is available on the test accuracy in women carrying 3 or more babies.
- Based on your risk factors, your doctor might also recommend MSAFP (Maternal Serum Alpha-Fetoprotein) screening for open spine defects (Spina Bifida).

More questions? Call us!
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