Prenatal Screening and Testing

In general, it is very likely that your baby will be born healthy. However, you should at least consider performing a test to try to identify if your baby carries certain problems in the chromosomes. These tests can be divided into two basic types: screening and diagnostic. Screening tests involve taking blood samples from you and a special ultrasound test. These are then used to estimate the risk of certain problems in the babies. These tests do not diagnose chromosomal problems. The results tell you if your baby is at an increased risk for certain chromosomal problems.

Diagnostic tests are used to diagnose certain chromosome or genetic problems. They involve taking a small tissue sample from the placenta or fluid from the amniotic sac. They are almost 100% accurate, but because they are invasive, there is a very small risk of possibly causing a miscarriage. The following are the options within each type of testing:

Screening:

The NIPS, non-invasive prenatal screen, takes fragments of DNA from your baby from your blood, and it identifies whether your baby is at an increased risk of a problem in certain chromosomes called “aneuploidies”. The most common of these are: Trisomy 21 “Down Syndrome”, Trisomy18 “Edward’s syndrome”, Trisomy 13 “Patau”, or abnormal number of the sex chromosomes. It can also reliably tell you the sex of your baby. This test detects certain aneuploidies with a high level of accuracy, but not as good as a diagnostic test. The benefit is that because it is blood work, there is no risk in doing this test. Also, in comparing the NIPS and the first trimester screen (below), the NIPS has a better detection rate and it is the recommended test by the American College of Obstetrician and Gynecology and The Society of Maternal Fetal Medicine.

The results of a NIPS can help you and your doctor decide the next steps, including whether to have a diagnostic test like chorionic villus sampling (CVS) or amniocentesis (“amnio”) described below or not. The NIPS test can be done in our office although we prefer that you do it at a maternal fetal medicine (MFM) office.

First Trimester Screen: The first part of this test involves doing a special ultrasound and collecting blood between 11 and 13 weeks. If this result comes back “screen positive” then a diagnostic test will be recommended. The First screen detects approximately 90% of babies with Down syndrome and 90% of trisomy 18. There is a 5% chance of a false “positive screen” result. This test is done only at a MFM office.

In addition, all patients have an MSAFP— a screening test done around the time of the anatomic ultrasound to check for open neural tube defects. A negative screen rules out approximately 80% of open neural tube defects.

Diagnostic:

Chorionic Villus Sampling (CVS): This test involves taking a small piece of placental tissue between 11 and 13 weeks. This tissue is sent for analysis of the chromosomes that make up the baby and it can also test for some specific medical conditions. It is close to 100% accurate for detection of these chromosomal abnormalities. The associated risk for miscarriage for the procedure is approximately 0.1-0.5 %.

Amniocentesis: This test involves taking a sample of the amniotic fluid between 16 and 20 weeks. The sample is sent for chromosome analysis. It is also close to 100% accurate for detection of chromosomal abnormalities. The associated risk for miscarriage for the procedure is approximately 0.1-0.5 %.

I understand the above testing options. My questions have been answered. I am aware that some of these tests may not be covered by my insurance.

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Patient Signature Date