Midwifery Care Associates, P.C.
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ANOMALY TESTING AND COUNSELING

Anomaly testing and counseling is offered to all clients. The midwife will order tests as clinically indicated or at the wishes of the client, after a discussion of the role of screening and diagnostic prenatal testing.

Ultrasound

An anomaly screening ultrasound is offered to all clients. This ultrasound performed at 20 weeks examines the fetus for any structural abnormalities or genetic markers. Ultrasound is also sometimes clinically indicated for gathering specific information such as estimated date of delivery, placental location, and size or position of the baby.

Women at or beyond age 40 at term and desiring homebirth are required to have an anomaly screening ultrasound to rule out any significant defects.

Alpha-Fetoprotein Triple Screen, Quad, or Tetra Screen

Genetic testing, of any sort, usually starts a large debate. These tests are no exception. Sometimes called the MSAFP or maternal serum AFP, this blood test is performed on blood drawn from the mother to check the levels of AFP. AFP is a protein secreted by the fetal liver and present in the mother's blood. It is generally used for screening for open neural tube defects. A low level of AFP is also associated with some cases of Down syndrome.

The Triple screen measures not only AFP, but HCG and estriol as well. This test is more sensitive than the AFP by itself, and screens for additional genetic problems. Generally speaking, any combination of the testing will identify 60% of babies with Down syndrome and 80-90% of babies with open neural tube defects.

The Maternal Serum Quad Screen is used to identify pregnancies that are at a higher risk to have Down syndrome, trisomy 18, or an open neural tube defect. The quad screen utilizes four biochemicals in the maternal serum for fetal assessment: alpha-fetoprotein (AFP), human chorionic gonadotropin (HCG), unconjugated Estriol (Ue3), and dimerlc inhibin-A.

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An “at risk” triple screen or quad screen may indicate that the risk of your baby being born with trisomy 21 (or Down syndrome) is as great or greater than that of a woman aged 35; it does not mean that your baby has Down syndrome. You may then be offered a very detailed ultrasound (Level II) or even an amniocentesis in an effort to determine your baby's exact chromosomal make-up and to rule out Down syndrome. NIPTS blood testing would also be an option. An elevated AFP indicates that further testing would be in order to rule out an open neural tube defect. This test would be a level II ultrasound to look for defects in the spinal cord.

First Trimester Screening: Ultra Screen

First trimester screening for Trisomy 21 (Down syndrome) and Trisomy 18 (a rare but severe chromosomal abnormality) assesses your risk of carrying a pregnancy with either of these abnormalities. It does not detect other birth defects. About 80% of the time a pregnancy affected with Trisomy 21 or 18 will result in a high-risk result. This test is a screening test only and a high-risk result does not mean your baby has Trisomy 21 or 18. It means that the risk is higher than average and further testing and genetic counseling should be offered. This test does not evaluate risk of neural tube defects and a blood test for AFP should be offered at the appropriate time.

First Trimester screening is a combination of ultrasound to measure nuchal fold thickness and a blood sample taken between 11 weeks 1 day and 13 weeks 6 days. The results take about 10 days to be reported if the blood test is done at the time of the ultrasound. You can choose to do the blood testing prior to the ultrasound (kits are available) and results will be immediate at the time of the ultrasound.

This test is available at Capital Health Maternal Fetal Medicine and Genetics at (609) 537-7253.

Genetic Counseling

Genetic counseling is offered to all clients over the age of 35 or those with risk factors. Genetic counselors at Capital Health will meet with the couple to evaluate the risk of genetic complications based on age and family/patient history. You will be counseled in what testing may be the best for your particular risk factors. A meeting with the counselors may be arranged by calling Capital Health Maternal Fetal Medicine and Genetics at (609) 537-7253.

NIPTS

NIPTS (Cell Free DNA) is a blood test that isolates and analyzes fetal DNA circulation in the mother's blood. This test is offered to women age 35 and over, or those with special risk factors for Trisomy 21 (Down syndrome). The test has sensitivity of 99.1% and a specificity of 99.9 %

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Amniocentesis/Chorionic villus Sampling

Amniocentesis is offered to all women over age 35, women who have had a baby with a chromosomal abnormality, women who are carriers of sex-linked diseases like hemophilia or muscular dystrophy, women with a strong family history of neural tube defect, women who have had an "at risk" screening test.

Amniocentesis involves withdrawing a small amount of amniotic fluid with a needle under ultrasound guidance. The fluid is sent to the lab for genetic analysis and AFP testing, and may take up to 1-3 weeks for results. The procedure is usually done between the 16th and 17th week of pregnancy. Amniocentesis, like all medical procedures, carries certain risks. Mild complications like cramping, bleeding or leaking fluid occur rarely in the mother and are usually not serious. However there is a small chance of miscarriage, infection or injury to the baby from the procedure. The risks of complication are about 1 in 200.

Some parents don't want to wait until 16 weeks for genetic testing. A Chorionic Villus Sample (CVS) can be performed earlier in pregnancy, usually around 11 weeks. CVS, like amniocentesis, is offered to women who will be 35 or older at the time of the birth, or with a family history of a child with a chromosomal abnormality. The cells collected in the CVS procedure are from the baby's placenta. A catheter is generally passed through the mother's cervix and some of the cells are withdrawn. Sometimes a needle is passed through the mother's abdomen and cells are withdrawn from the placenta. Like amniocentesis, CVS is done under ultrasound guidance. The risk of ruptured membranes, bleeding or infection is about 1 in 150. Results are generally ready in 3-4 days. CVS does not measure the amount of AFP in the amniotic fluid, so AFP testing is generally recommended later in pregnancy to evaluate risk for open neural tube defects.

Capital Health Maternal Fetal Medicine (609) 537-7253

I have read and discussed the above information with my midwife and I choose the following testing:

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To decline testing __

Signature_____ Date_____

Midwife_____ Date_____