*801 S. Milwaukee Avenue, Suite 100 Libertyville IL 60048 P: (847) 680-3400 F: (847) 680-348*6

*facts about…*

***Prenatal Screening and Diagnostic Testing Options***

During your pregnancy, you may be given the option to have any one of several prenatal tests. It is important to remember that prenatal testing is optional and you may elect to have no prenatal testing at all. Prenatal testing can be divided into two categories: screening tests and diagnostic tests.

***SCREENING TESTS***

Screening tests determine whether the fetus is at increased risk for certain conditions. It is important to remember that they cannot provide a definitive yes-or-no answer. Screening tests are considered noninvasive and pose no risk to the pregnancy. Screening results are typically reported as “high risk” or “low risk.” The available screening tests include:

**Non-invasive Prenatal Screening (NIPS):** NIPS is a maternal blood test that analyzes fetal DNA fragments to determine the risk for Down syndrome, Trisomy 18, Trisomy 13, Triploidy and Turner syndrome with a 92-99% detection rate. NIPS is not recommended for all women. This screen has not been validated in women carrying twins or higher order multiples, or in women whose pregnancies were conceived using donor eggs. NIPS can be done between 10-20 weeks gestation. \*

**First Trimester Screen (aka BUN screening/ Only performed at MFM):** The first trimester screen involves a maternal blood test and ultrasound to determine the risk for three chromosome abnormalities: Down syndrome, Trisomy 18, and Trisomy 13 (both more rare and more severe than Down syndrome). This screen is able to determine the risk for each of these conditions with a 90-95% detection rate. The blood test measures the amount of certain pregnancy-related chemicals, and the ultrasound measures the thickness of the back of the fetal neck (nuchal translucency) and looks for the presence or absence of a nasal bone. The ultrasound can sometimes provide risk information for additional abnormalities, such as heart defects. First trimester screening is done between 11-14 weeks gestation.\*

**Quad Screen:** The Quad screen is a maternal blood test that measures the amount of alpha-fetoprotein and other chemicals to predict the risk for Down syndrome, Trisomy 18, and open neural tube defects (e.g. spina bifida). This screen has a 60-75% detection rate for Down syndrome and Trisomy 18, and up to 90% for open spinal defects. The Quad screen is typically done between 15 to 19 weeks of pregnancy.

\**For patients who have the first trimester screen or NIPS, alpha-fetoprotein (AFP) alone should be measured in the second trimester to determine the risk for open neural tube defects.*

**Level II Ultrasound (Only performed at MFM):**  A comprehensive or level II ultrasound, usually performed between 18 and 22 weeks of pregnancy, is a thorough evaluation of the fetal anatomy targeted to identify anatomic or structural birth defects. Anatomic birth defects are present in 2-3 % of liveborn infants. Fetal growth, amniotic fluid volume, and placental anatomy are also evaluated during a comprehensive ultrasound. Chromosome abnormalities cannot be diagnosed or ruled out by ultrasound. However, fetal anatomic abnormalities are observed in 50-70% of fetuses with Down syndrome and over 90% of fetuses with Trisomy 18 or 13. Therefore, a normal comprehensive ultrasound may be associated with a reduced risk for chromosomal abnormalities. In contrast, identification of fetal anatomic abnormalities on ultrasound may be associated with a higher risk of chromosome disorders.

*If an increased risk of fetal abnormality is determined by any of the above screening tests, diagnostic testing such as amniocentesis or chorionic villus sampling may be offered to provide definitive diagnosis of fetal chromosome and/or genetic abnormalities.*

**DIAGNOSTIC TESTS**

Developed for pregnancies at “high risk” for a chromosome abnormality or other genetic disorder, diagnostic testing can provide a yes-or-no answer about whether the fetus is affected or not. Women who are considered to be at “high risk” to have a baby with a chromosome problem include women who will be 35 years old or older at the time of delivery, women who have had an abnormal screening result, and pregnancies in which abnormalities are identified on ultrasound. Diagnostic tests for chromosome abnormalities include:

**Chorionic Villus Sampling (CVS):** A technique that is offered for prenatal diagnosis of chromosome abnormalities (and other genetic syndromes if there is a family history or known risk). CVS is performed between 10-13 weeks of pregnancy. During the procedure, a small sample of the placenta is removed under ultrasound guidance. The cells from the placenta can be studied to analyze the fetal chromosomes. Results are available approximately 1-2 weeks after the procedure. There is a small increased risk of miscarriage associated with CVS, which is thought to be approximately 1 in 200 (or 0.5%) above the background risk of miscarriage in the first trimester. Abnormalities in chromosome number (like Down syndrome) or structure can be determined by CVS. Fetal gender can also be determined. Screening for open neural tube defects cannot be achieved through CVS. Women who opt for CVS should be offered maternal serum alpha-fetoprotein screening at approximately 16 weeks gestation to screen for open neural tube defects.

**Amniocentesis:** A technique that is offered for prenatal diagnosis of chromosome abnormalities (and other genetic syndromes if there is a family history or known risk). Amniocentesis is usually performed starting at 15 weeks of pregnancy and is available throughout the rest of the pregnancy. During the procedure, a small sample of amniotic fluid is removed with a needle under ultrasound guidance. The sample contains fetal cells floating in the amniotic fluid, which can be studied to analyze the fetal chromosomes. Results are available approximately 2-3 weeks after the procedure. There is a small increased risk of miscarriage associated with amniocentesis, which is thought to be 1 in 300 or less (<0.3%). Abnormalities in chromosome number (like Down syndrome or structure can be determined by amniocentesis. Fetal gender can also be determined. Additionally, the amniotic fluid can be tested to measure the amount of alpha-fetoprotein, which can detect the presence of open neural tube defects.

*Although diagnostic tests can provide a yes-or-no answer about the chromosome abnormalities, normal results do not guarantee a healthy baby.***For more information about prenatal screening and testing options, please feel free to consult your healthcare provider @(847) 680-3400.**