Prenatal Testing Options

There are several tests available to give you information about your pregnancy. All of these tests are optional since the decision to pursue prenatal testing can be very personal. The prenatal testing options can be broken down into two categories – screening and diagnostic tests.

Screening Tests – Screening tests do not diagnose chromosome conditions, but rather give a personalized chance that the pregnancy could have one of these conditions. Screening tests are non-invasive and do not have a risk for risk for miscarriage. However, there is a risk that these tests could falsely worry you (report that the risk for a condition is high when the pregnancy is not affected, which is also known as a false positive) or falsely reassure you (report that the risk for a condition is low when the pregnancy is affected, which is also known as a false negative). Some women use screening tests to help them make decisions regarding whether they would like to pursue diagnostic testing.

- *First Trimester Screen* – This screening test uses the information from a blood test (performed between 9 and 13 weeks of pregnancy) and an ultrasound (performed at 11 and 13 weeks of pregnancy) to screen for pregnancies affected with Down syndrome, trisomy 18 and trisomy 13. It is estimated that up to 91% of pregnancies affected with Down syndrome and up to 95% of pregnancies affected with trisomy 18 and 13 will be detected with first trimester screening.

- *Second Trimester Screening* – This screening test is a blood test performed between 15 and 21 weeks of pregnancy to screen for pregnancies affected with Down syndrome, trisomy 18 and open neural tube (spine) defects. It is estimated that 70-75% of pregnancies affected with Down syndrome, 60% of pregnancies affected with trisomy 18, and an 80-90% of pregnancies affected with neural tube defects would be detected with second trimester screening.

***Typically women choose to do either first trimester screening or second trimester screening and do **NOT** do both of these screening tests.

- *Enhanced Level I Ultrasound* – An ultrasound that can be conducted as early as 16 weeks. This ultrasound is primarily used to screen for neural tube (spine) defects; however, several other organ systems are also examined. An enhanced level I ultrasound can detect more than 90% of cases of neural tube defects when performed in a specialized center. Should a birth defect be suspected on an enhanced level I ultrasound, a level II ultrasound will be recommended.

- *Level II Ultrasound* – A detailed ultrasound that is typically conducted between 18-20 weeks of pregnancy. This ultrasound screens for birth defects and markers associated with Down syndrome and other chromosome conditions. Approximately 65-70% of cases of Down syndrome and 85-90% of cases of trisomy 18 and trisomy 13 can be detected by 20 weeks of pregnancy when performed by a doctor with specialized training.

Diagnostic Tests – Diagnostic tests are more than 99% accurate in diagnosing chromosome conditions. Testing for other specific genetic disorders known to be present in a family can often be performed. These procedures have a miscarriage risk of approximately 1 in 200.

- *CVS (chorionic villus sampling)* – A small sample of early placental tissue is obtained either cervically or abdominally, depending on the location of the placenta. This test can be performed between 10 and 12 weeks of pregnancy.

- *Amniocentesis* – A small sample of fluid is removed from the amniotic sac by inserting a very thin needle into a woman's abdomen. This test can be performed after 16 weeks of pregnancy. The amniocentesis can also detect more than 95% of all open neural tube defects.

***Women choose to do either CVS or amniocentesis and **NOT** both of these procedures.