Danielle S. Kiko, M.D., F.A.C.O.G.
Steven M. Willard, M.D., F.A.C.O.G.
Jessica Sovacool, M.D.
Heather A. Kreareas, MSN, WHNP-BC, IBCLC
Obstetrics and Gynecology
6555 Frank Ave. NW North Canton, Ohio 44720
PH: 330 956-5300 FAX 330 935-4603

Optional prenatal testing information:

Congratulations! You are entering a wonderful, exciting time of your life. There are numerous blood tests that are recommended at different times throughout your pregnancy. In addition to the recommended tests, there are some optional tests available. Below is a brief description of these tests. Please address any questions to your provider for further information.

Carrier Screening: Carrier screening is a blood test to see if you carry a gene or genes that carry certain genetic disorders. The four most common conditions screened for are cystic fibrosis, spinal muscular atrophy, fragile X syndrome and thalassemia. These tests may be recommended by advisory societies such as the American Congress of Obstetrics and Gynecology and the American College of Medical Genetics. Cystic fibrosis is found in 1 in 3500 births and is the most common fatal genetic disorder. The chance of being a carrier is approximately 1 in 30. Spinal muscular atrophy occurs in 1 in 6500-10,000 births and is the most common inherited cause of infant death. The chance of being a carrier is 1 in 50 to 1 in 90. Fragile X Syndrome occurs in approximately 1 in 3,600 boys and 1 in 6,000 girls and is the most common cause of inherited intellectual disability. Thalassemia is a term that refers to a group of genetic disorders characterized by insufficient production of hemoglobin. There are two proteins involved in the production of hemoglobin, alpha and beta. If there is a deficiency in either of these proteins, the red blood cells do not form properly and cannot carry adequate amounts of oxygen to all parts of the body. In pregnancies where both parents carry the gene, there is a 1 in 4 chance that the child will have the severe form of thalassemia, a 2 in 4 chance that the child will carry the gene for thalassemia and a 1 in 4 chance that the child will neither have the disease or be a carrier

Genetic Screening: There are optional tests for genetic screening including the Quad screen, Sequential/Integrated screening and Non-Invasive Prenatal Testing (NIPT). These tests are designed to look for specific genetic changes. They cannot detect all genetic changes that could cause health problems. A "normal" result does not guarantee a healthy pregnancy or baby. The sensitivity of a test is the likelihood of the test results being positive in someone who does have the disease (a true positive result). The specificity of a test is the likelihood of the test results being negative in someone who does not have the disease (a true negative result). The false positive rate is the rate of a test being positive when in fact the person does not have the disease. The false negative rate is the rate of a test being negative when in fact the person does not have the disease. The best tests have a high sensitivity and low false positive and negative rates.

Quad Screen: The Quad screen is a blood draw performed between 15-21 weeks of gestation to help determine the risk of genetic disorders such as trisomy 13, 18, and 21 (Down's Syndrome) and the structural disorder spina bifida. Some of the information can be used to indicate if the pregnancy is at a higher risk for problems with the growth of the baby. This is not a diagnostic test and only gives a risk of these conditions. It has the lowest sensitivity and specificity rates and highest false positive and negative rates. It gives the risk of these abnormalities in terms of numbers (1 in 10, 1 in 200, 1 in 10,000, etc

Sequential Screening/Integrated Screening: The sequential screen is a combination of ultrasounds and blood work that also identifies the risk of the above trisomies and spina bifida. Compared to the quad screen, it has a higher sensitivity and specificity, lower false positive and negative rates, but is still not a diagnostic test. It gives the risk of these abnormalities in terms of numbers (1 in 10, 1 in 200, 1 in 10,000, etc).

Non-Invasive Prenatal Testing (NIPT): NIPT is a blood test that can be performed after 10 weeks gestation. This test identifies fetal DNA that circulates in maternal blood. It has the highest detection rate for Trisomies 13, 18, and 21. To identify the risk of spina bifida, a second blood test can be obtained at ~15 weeks. NIPT has been validated in high risk populations, mothers age >/= 35, personal or family history of the above trisomies and certain other genetic disorders. In these populations, the sensitivity and specificity rates are 97-99% ad false positive and negative rates are <1%. It says with 99% certainty "yes" or "no".

Gender

At this time, there is not an available test to solely identify gender. **This is only available via the NIPT.**