CHROMOSOME TESTING

All pregnant couples are at risk of having a child with a chromosome problem or birth defect, but the chance increases as a woman gets older. Examples of chromosome conditions include Trisomy 21= Down Syndrome, Trisomy 13 and Trisomy 18. People with Down Syndrome can have developmental delays, heart problems, and learning problems. Trisomy 13 and 18 are conditions that cause severe complications and most babies do not survive the first year of life. Couples with an affected pregnancy typically have no family history of these conditions. If aware a pregnancy has a chromosome abnormality, we do follow the pregnancy more closely as there is an increased risk of miscarriage. Otherwise, pregnancy management is no different. We cannot change the genetic outcome. The American College of Obstetricians and Gynecologists (ACOG) recommends that all women be **offered** chromosome screening before 20 weeks of gestation. However, they do not state that this testing **should** be done as with other routine tests such as blood type, Hepatatis screening, etc.

We offer screening testing through our office. With all of these options, the answers are not definitive. There are false positive and false negative results. However, the tests allow couples who are at highest risk of having an affected child to be aware and pursue the definitive testing if they wish. In all circumstances, definitve testing would require either a chorionic villus sampling (CVS) or amniocentesis. Both of these tests involve putting a needle in the pregnancy to obtain genetic material and have some risk to the pregnancy. CVS and amniocentesis tests are performed by the maternal and fetal medicine (MFM) specialists. Screening options include:

1) 1st trimester nuchal screen- A blood sample is collected between 9-13wks, typically with a finger stick kit you are provided to bring home. You then arrange an ultrasound with the MFM office to measure the back of your baby's neck. This measurement is put in a calculation with the blood results to determine the risk you could have a child with a chromosome problem. The results return as a probability, such as 1:55, 1:420, 1:5,000. A risk >1:300 results in a positive test. Most positive results are false positives. A wide net is cast in order to identify pregnancies at high risk so further testing can be done.
2) 2nd trimester Tetra/Quad screen- A blood draw is collected between 15-21weeks looking at 4 markers and again calculating the risk. This test occurs later in pregnancy leaving less time for diagnostic options. It is also slightly less accurate than the 1st trimester screen. However, it is typically less expensive since there is no ultrasound and requires less time. Again, the result is a probability with most positive results being false positives.
3) Non-invasive Prenatal Testing (NIPT)- This is the newest test available. A maternal blood draw is collected any time after 10weeks. The test looks for extra copies of the following chromosomes: 13, 18, 21, and gender/ XY. This test is more accurate than the first two, specifically with less false positive results. However, it is screening ONLY the chromosomes mentioned and will not detect any other abnormality. Furthermore, because it is newer, insurance coverage varies widely.

We hope this information is helpful for you in navigating testing options available. Please be aware that information on the internet can be helpful as well but is not governed for accuracy. If you come across conflicting or confusing information, please bring it to our attention. Reliable sources for information are www.acog.org and www.uptodate.com for questions about testing and many of your pregnancy concerns.

CARRIER SCREENING

Carrier screening determines if you are at risk of having a child with a specific genetic disorder. A "carrier" is a person who shows no signs of a genetic disorder but could pass the genetic change on to her children. Carrier testing can be performed prior to conception or during pregnancy. Once pregnant, the genes cannot be changed. Unfortunately, genetic testing can be expensive and insurance coverage is inconsistent. Labwork that may be considered "covered" may be subject to high deductibles. Talk with your insurance company and know your benefits. Laboratory bills are not from our office, and we are not responsible for charges generated. We have listed the test codes for reference when talking with your insurance company. Be aware that a negative test DOES NOT ELIMINATE the possibility your child could have any of the disorders discussed here.

CYSTIC FIBROSIS (CF) - CF primarily affects lungs/breathing, digestion and fertility. The severity of the disease varies widely and is considered a chronic and progressive condition. European Caucasians have the highest risk of carrier status (1 in 24). African American, Hispanics, and Asian American are at lowest risk of being carriers (1:61, 1:58, and 1:94 respectively). The American College of ObGyns (ACOG) recommends that all couples be **offered** testing but does not have an opinion that couples **should** have it done. Procedure/ CPT code: 81220, Diagnosis/ Dx code: V77.6

SPINAL MUSCULAR ATROPHY (SMA) – SMA is a neuromuscular disorder with varying degrees of severity. People with SMA have progressive weakness and eventually are unable to breath on their own. It has four subtypes depending on age of onset and clinical course. The most severe type typically presents in the first months of life with infants dying before one year. Less severe types may not present until teens or 20s. There is no consensus among expert bodies regarding the usefulness of screening for SMA in those without a family history. For this reason, insurance companies may deny this testing when there is no family history. CPT Code: 81401, Dx code: V28.9

FRAGILE X SYNDROME (FXS) – FX is the most common inherited cause of intellectual disability. Affected individuals may have classic physical features (prominent forehead and chin), intellectual disability, and behavioral features (hyperactivity, anxiety, repetitive movements, etc.). FXS is an X-linked disorder with a complicated inheritance. Up to 3% of boys with special needs (such as autism and nonsyndromic intellectual disability) have been diagnosed with FXS. ACOG recommends screening for individuals with a family history of FX related disorders, unexplained mental retardation/ developmental delay, autism or premature ovarian failure. Again, without this history insurance coverage may be denied. CPT code: 81243, Dx Code: V28.9

HEMOGLOBINOPATHIES (Alpha & Beta Thalassemia, Sickle Cell) – These disorders affect the shape and function of blood cells. Affected people may have severe anemia, weakness, a large spleen, bone and heart problems and slower growth. These disorders are chronic and can be fatal. They are more common in individuals of African American, Mediterranean, Hispanic or Asian descent.

Jewish genetic disorders – This refers to a group of disorders known to occur with high incidence (20-25%) in the Ashkenazi (Eastern European) Jewish population. Anyone with one grandparent of Ashkenazi descent or any relative with one of the genetic conditions should consider testing. The four most severe disorders are Tay-sachs disease, Canavan Disease, CF, and familial dysautonomia but other disorders can be tested. If you have any question whether you are at risk for these disorders, please discuss with your provider.