Brookhaven OB-GYN Associates, P. A.

GENETIC SCREENING OPTIONS

What is Genetic screening in pregnancy? These screening tests evaluate for chromosomal abnormalities in the pregnancy. They do not give a definitive answer about whether or not a fetus has a genetic condition. The tests can only estimate whether the risk of having certain conditions is increased or decreased. These genetic screening tests do not cause harm to the fetus and are performed with a blood draw.

The two options offered at Brookhaven OB/GYN include:

1) Cell free DNA screen (NIPT - noninvasive prenatal testing)

a) Noninvasive prenatal testing (NIPT) is a method of determining the risk that the fetus will be born with certain genetic abnormalities. Obtained between 10 weeks and 36 weeks, it tests for small amounts of fetal DNA that are circulating in a pregnant woman's blood (cell free DNA). NIPT looks for chromosomal disorders (Down Syndrome or Trisomy 21, Trisomy 18 and Trisomy 13) and tests for extra or missing copies of the X or Y chromosomes (sex chromosome). The accuracy of the test varies by disorder.

b) If you choose this screening test, we will also recommend an additional separate screening test between 15 and 20 weeks for a one time blood draw of AFP. This will assess the fetal risk for neural tube defects and abdominal wall defects.

c) This test has varying coverage by insurance companies. Please call this number if you are interested to discuss the cost with a Harmony Care Specialist: 1-855-927-4672.

2) Quad screen (quadruple marker test)

a) The quad screen is a prenatal test that measures levels of four substances in a pregnant woman's blood: alpha-fetoprotein (AFP), human chorionic gonadotropin (HCG), estriol and Inhibin A. Obtained between 15 weeks and 20 weeks, these measurements can assess the risk that the fetus has certain conditions such as chromosomal abnormalities (Down Syndrome / Trisomy 21, Trisomy 18, Trisomy 13, neural tube defects and abdominal wall defects).

b) This test includes screening for neural tube defects and abdominal wall defects and does not need a second lab draw later in the pregnancy.

c) This test is covered by most major insurance companies.

Please note: options that are not offered at this clinic include nuchal translucency ultrasound screening and first trimester screening. Additionally, diagnostic testing is not performed here. This includes chorionic villus sampling (CVS) or amniocentesis. If any concern arises for the need for diagnostic testing, your doctor will discuss referral to a facility that can perform these.

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CARRIER SCREENING OPTIONS

What is carrier screening? Carrier screening can show if you or your partner carry a gene for a certain disorder. This screening can be done before or during pregnancy. Carrier screening often is recommended if you or your partner have a genetic disorder, have a child with a genetic disorder, have a family history of a genetic disorder, or belong to an ethnic group that has an increased risk of specific disorders.

The following three carrier screening tests are offered to *all* women of reproductive age because they are some of the most common inherited disorders:

1) **Hemoglobinopathy Screening: Hemoglobin electrophoresis**
    a) *Hemoglobin electrophoresis is used as a screening test to identify variant and abnormal hemoglobins, including Sickle cell disease or trait.*

2) **Cystic Fibrosis Carrier Screening**
    a) *Cystic fibrosis is a hereditary disease that affects the lungs and digestive system. The body produces thick and sticky mucus that can clog the lungs and contribute to persistent lung infections. This can limit the ability to breathe over time and can cause death.*

3) **Spinal Muscular Atrophy (SMA) Carrier Screening**
    a) *Spinal muscular atrophy is a genetic disorder that affects the nerves of the spine and is characterized by weakness and wasting in muscles used for movement. There are several different types of spinal muscular atrophy, which can vary in severity and onset of muscle weakness. SMA can cause severe disability and death. SMA does not affect mental ability.*

Please check with your insurance if you are interested in any of the above carrier screening tests. Once you know if the above three tests are covered by your insurance, please contact your doctor's office so these tests can be scheduled.

Please note: there is no risk of harm to the fetus with screening tests.