Chester County Ob/Gyn

Genetic Testing

During the course of your pregnancy, you may be offered **prenatal genetic testing**. In addition to the information you receive from our providers and nurses, this information is intended to serve as an extra resource. We are happy to answer any questions you may have about genetic testing. If we can be of any assistance, please do not hesitate to contact us.

What is Prenatal Genetic Testing?

Prenatal genetic testing refers to tests that are done during pregnancy to either screen or diagnose a genetic abnormality, birth defect or chromosomal abnormality. The goal of prenatal genetic testing is to provide expectant parents with information to make informed choices and decisions. Genetic testing also assists our providers in providing the best care and management of your pregnancy. The American College of Obstetricians and Gynecologists (ACOG) recommends that all pregnant women, regardless of their age, be offered screening for chromosomal abnormalities such as Down syndrome. It is important to remember that all genetic testing is optional. Many patients in our practice choose not to obtain genetic testing. It is your decision and we will always honor that choice.

There are two types of prenatal genetic testing that are routinely offered: **Aneuploidy Screening and Testing** (searching for abnormal chromosomes) and **Carrier Screening** (searching for abnormal genes that run in the family)

Aneuploidy Screening Tests
Aneuploidy Screening Tests do not diagnose a chromosomal abnormality. They only determine if a fetus is a high or low risk for the most common chromosomal abnormalities such as Down syndrome (Trisomy 21), Trisomy 18 and Trisomy 13. Examples of aneuploidy screening include:

- Sequential Screen (ultrasound + blood work)
- Quad Screen (blood work at 16 weeks)
- Cell Free DNA Testing (NIPT)

Aneuploidy Diagnostic Testing is definitive tests that reveal the actual chromosomal abnormality. Examples of diagnostic testing include:

- Amniocentesis
- Chorionic Villus Sampling (CVS)

Carrier Screening

All women who are pregnant or contemplating pregnancy should be offered screening for recessive genetic diseases based upon history. Cystic Fibrosis is a common recessive genetic disease that is recommended for screening in most patients. (click here to read more about Cystic Fibrosis Screening).
Other recessive genetic diseases can be screened for based upon history or ethnic background. Examples include Sickle Cell carrier testing which is offered to all African-American patients.

Chester County Ob/Gyn also offers optional universal recessive carrier screening for all patients. The optional test can detect over 90 genetic diseases that may run in your family. 30% of individuals will be a carrier for at least one recessive disease. Many of them are rare, some are severe and some are minor issues that can be corrected with treatment.

There may be a charge if you choose this optional test and your insurance company does not cover this test. Charges can be 100% covered or be as high as $275.00. We will be happy to discuss this with you in greater detail if you desire.

Neural Tube Defect Screening

Neural tube defects occur in only about 1 pregnancy out of 1,000. They are birth defects in which the brain or part of the spinal cord does not form normally. The risk does NOT increase with maternal age, but is more related to ethnic background and levels of folic acid in our diet. Screening is offered to all women at Chester County Ob/Gyn as a blood test at 16 weeks. It is included in women who are getting the sequential screening test or quad screening test. For those not interested in the blood test, your 20-week ultrasound that will be performed by Maternal Fetal Medicine is equally effective as a screening for Neural Tube Defects.

What are the individual Prenatal Genetic Tests that I may be offered?

First Trimester Sequential Screening

This screening test is offered to all women who are at low risk for chromosome abnormalities (fetal aneuploidy) such as women under the age of 35 who have no history of aneuploidy in their family. This is a combination of blood tests and ultrasound examination to help identify babies who may have Down syndrome, Trisomy 18 or Trisomy 13. These are the most common chromosomal abnormalities that babies can have. The test is performed at our Maternal Fetal Medicine office. At 12-14 weeks you will have an ultrasound examination and blood work. The ultrasound examination is to measure the skin fold around the baby's neck (Nuchal Translucency). If the results are normal, you will have additional blood work at 16 weeks. If all tests are normal, you are considered low risk for these chromosomal abnormalities.

Quad Screen Testing
A single blood test at 16-19 weeks, offered to low risk women who desire aneuploidy screening, but decided upon testing after the first trimester is over. The test gives an 80% detection rate for Down syndrome. The Quad Screen also provides risk assessment for Trisomy 13, Trisomy 18 and Neural Tube Defects.

Cell Free DNA Testing
This screening test is offered to all women who are at high risk for chromosome abnormalities (fetal aneuploidy) such as women 35 years of age or older at the time of the baby's due date, or have a family history of aneuploidy. Cell Free DNA testing is performed any time after 10 weeks gestation. Results return in less than 2 weeks. Fetal sex determination can be included. Detection rates in high-risk patients for Down syndrome, Trisomy 18 and Trisomy 13 are 99%, 90-97% and 80-99% respectively. The false positive rates are less than 0.1%. When combined with the Nuchal Translucency (Ultrasound of the baby's neck at 12-14 weeks), cell free DNA testing is a better screening test than the sequential screen. Although it is a very good screening test, it is not a diagnostic test and is not as definitive as the amniocentesis or CVS.

Many women at low risk prefer to have the cell free DNA testing performed. At Chester County Ob/Gyn we believe the testing to be superior to sequential screening and therefore offer it to all women. Many insurance companies are beginning to cover it 100%. Unfortunately, some are not. Some plans allow you to pay out of pocket for the test which can run anywhere between $150 and $1500. Some plans do not allow us to offer this out of network. Please speak to a provider directed if interested. Chester County Ob/Gyn works with many private labs that can offer the lowest out of pocket expense if you desire this test and it is not covered.

Chorionic Villus Sampling (CVS)
CVS involves obtaining a sample of the tissue surrounding the sac where the fetus develops. A sample of the cells from this tissue provides chromosomes and other material that may then be tested to diagnose chromosomal abnormalities, genetic birth defects and other conditions. There is a miscarriage rate of 0.5% or 1 pregnancy in every 200 that have the test. The test is usually performed between 11-13 weeks of pregnancy.

CVS is offered to all women 35 years of age or older at the time of their due date, as well as women with a family history of aneuploidy. Women with abnormal cell free DNA testing, or abnormal ultrasound testing may also be candidates for CVS.

Amniocentesis

Amniocentesis involves a thin needle place through the mother's abdomen into the baby's sac to remove a small amount of amniotic fluid. This fluid is tested for chromosomal abnormalities, genetic birth defects and other conditions. The miscarriage rate is quoted anywhere between 0.5% (1 in 200) and as low as 0.1% (1 in 1000). The test is typically performed after 15 weeks.

Amniocentesis is offered to all women 35 years of age or older at the time of their due date, as well as women with a family history of aneuploidy. Women with abnormal sequential screen testing, abnormal quad screen testing, abnormal AFP testing, abnormal cell free DNA testing, or abnormal ultrasound testing may also be candidates for amniocentesis.

AFP Screening

This blood test is used to screen for Neural Tube Defects such as spina bifida and anencephaly. It can also detect some abdominal wall defects. The test is included in the Sequential Screen and the Quad Screen. The incidence of this condition is NOT affected by maternal age and many experts believe a detailed ultrasound performed between 18-22 weeks of pregnancy is just as good as the AFP test for screening for neural tube defects.

What tests are right for me?

There are risks and benefits to all of these tests and it is an individual decision for each family whether to do any of these tests. You do not need to do any screening or diagnostic testing. The benefits of any prenatal testing include reassurance, or in the event of a problem, preparation, optimal medical management, or alternative options. The risks of getting testing also includes additional worrying if you have abnormal screening tests but don't do diagnostic testing and the miscarriage risks associated with diagnostic testing. The risk of not doing any testing is not knowing about a birth defect, or other problems, until after the baby arrives.

There is no correct answer regarding prenatal genetic screening, but we hope to assist you by providing the information needed for you and your family to choose the testing most suitable for you and your individual needs.

Whatever your plans, Chester County Ob/Gyn will respect your decision and provide you and your baby with the best obstetrical care around.