Tepeyac OB/GYN
Something More Than Medicine™

Prenatal Genetic Testing Options

Congratulations on your pregnancy! Our mission is to provide you with excellent medical care to maximize the health of you and your baby. In keeping with this mission, we have compiled the following information regarding your options for prenatal testing. There are many tests available to screen for genetic and neural tube disorders. Although they are available to all patients, they are completely optional. While a normal result is reassuring, abnormal results often lead to invasive procedures and may suppress of the joy of pregnancy. Many practices will use an abnormal result to justify terminating a pregnancy. Tepeyac Family Center is dedicated to the sanctity of all human life and does not recommend or perform abortions, even in the case of a child who may require special care. We are committed to walking with you through the remainder of your pregnancy in the context of respect for you and your child. Please feel free to ask us if you have any questions. For questions regarding cost and insurance coverage, please check with your individual plan.

Screening for Parental Carrier Status

Cystic Fibrosis

Cystic fibrosis is a genetic disorder that severely affects the respiratory and digestive systems. It is most common in the Caucasian, Jewish and Mediterranean populations. It is one of the most common inherited genetic disorders.

There is a blood test we can perform to see if the PARENTS carry the gene that causes Cystic Fibrosis. If the mom is negative, the likelihood that the baby will be affected is extremely low. If the mother does carry the gene, the father of the baby should be tested as well. If both parents carry the gene for CF, the baby has a 25% chance of having the disease itself. We will discuss this in greater detail if this is the case for you.

Other Inherited Disorders

There are many other inherited genetic disorders such as Fragile X, Tay-Sachs, and spinal muscular atrophy. If you are considered at risk for one of these genetic disorders, we may discuss testing to check your carrier status.

Non-Invasive Screening for Genetic and Neural Tube Disorders

Who should consider these tests?

ACOG recommends that all women be given the option for genetic screening. Screening tests do not provide a definitive diagnosis, but rather state if your baby is at *high risk* or *low risk* for one of these conditions. False positives (where test results are abnormal when the baby is perfectly healthy) and false negatives (where results are normal when the baby is actually affected by a disorder) are possible with all screening tests. If any of these come back abnormal, your options may include a diagnostic test with higher accuracy to confirm the result, referral to a specialist, or enrollment in our perinatal hospice program.

Cell-free Fetal DNA

What does it screen for? Down Syndrome, Trisomy 13, Trisomy 18, Monosomy X (aka. Turner Syndrome). It does not detect neural tube defects.

How is it done? This is a blood test that looks at the fragments of the baby's DNA that are in the mother's bloodstream. We can take the sample in our office any time after 9 weeks. It is currently recommended for high risk women with a singleton pregnancy.

How accurate is it? This test has the fewest false positives and false negatives. The false positive rate is 0.5%.

First Trimester Screen

What does it screen for? Down Syndrome, Trisomy 13, Trisomy 18, heart anomalies. It does not detect neural tube defects.
How is it done? This is performed at an outside facility and consists of a combination of a specialized ultrasound (nuchal translucency) and blood work. It must be done between 11weeks 3days – 13weeks 6days.
How accurate is it? This test is 95% accurate, with several false positives and false negatives

Alpha-Fetoprotein

What does it screen for? Open neural tube defects (where the brain and spinal cord do not fully close), gastrointestinal defects, Down Syndrome.
How is it done? This is a blood test done between 16-20 weeks gestation.
How accurate is it? Of women with an abnormal result, as few as 1 in 33 will have an affected baby. This means there are many false positive results.

Quad Screen

What does it screen for? Down Syndrome, Trisomy 13, Trisomy 18, & neural tube defects
How is it done? This is a blood test that is done in our office between 16-20 weeks. It includes AFP and other markers of the baby's health.
How accurate is it? This test is between 70-85% accurate, meaning it has the many false positives and false negatives.

Invasive Diagnostic Testing

A note about diagnostic tests

Diagnostic tests are the most accurate at detecting if the baby has a particular condition; however they are invasive and carry a small risk of losing the baby. ACOG recommends that all women over the age of 35, or women who have a family history of one of these disorders, be offered diagnostic testing. Due to the risks involved, few of our patients elect to pursue them. If you have an abnormal screening test, we may discuss if the knowledge gained from a diagnostic test is worth the risk involved. Depending on the nature of the diagnosis, we will discuss options including enrollment in our perinatal hospice program, specialist referral, or delivery at a tertiary care center.

Chorionic Villus Sampling (CVS)

CVS tests for genetic disorders, but not neural tube defects. This involves passing a needle through the abdomen or cervix to take a small sample of the developing placenta. It is done between 10-12 weeks gestation.

Amniocentesis

Amniocentesis detects genetic disorders and open neural tube defects. Using ultrasound guidance, a needle is passed through the abdomen to take a sample of the amniotic fluid which surrounds the baby. This can be done after 16 weeks gestation.

I have read the information regarding prenatal testing options, which was provided to me by the Tepeyac Family Center. I have had the opportunity to have my questions answered and my concerns have been addressed.

I am _____ weeks pregnant and would like the following **screening** tests:

- ○ No prenatal screening or diagnostic tests done during this pregnancy. If I have further questions or concerns, I will bring them up with my provider.
- ○ I am currently uncertain whether I will opt for prenatal screening during this pregnancy. I understand that many of these tests are time sensitive and recognize that by deferring my decision, I may miss the window to perform some of these tests. If I decide to pursue genetic testing, I will bring this up with my provider.
- ○ Cell Free Fetal DNA (genetic disorders only, for women at high risk)
- ○ First Trimester Screen (genetic & heart disorders, ultrasound & blood work)
- ○ Alpha-fetoprotein (neural tube & GI defects, Down Syndrome, many false +)
- ○ The Quad Screen (genetic & neural tube defects, low accuracy)
- ○ Cystic Fibrosis Carrier Screening (to see if mom carries the gene for CF)

I would like the following **diagnostic** tests. I have discussed these tests along with their use, risks, benefits, and alternatives with my provider:

- ○ No diagnostic testing during this pregnancy. If there is an abnormal screening test or ultrasound, we may revisit the topic of appropriate diagnostic testing.
- ○ Chorionic Villus Sampling
- ○ Amniocentesis

Patient Name: _____

Patient Signature: _____ Date: _____

Provider Name: _____

Provider Signature: _____ Date: _____