Gentle Hands OBGYN

F.A.Q. on Genetic Testing during your pregnancy

What does "genetic testing" mean?

Most women will have healthy pregnancies but 3-5% will have a baby with a congenital anomaly. **A congenital anomaly is a structural or functional abnormality present at birth.** Some of these are mild and corrected with relatively minor surgery- for example cleft palate, the most common anomaly. Some anomalies require major surgeries or long-term care and unfortunately, some are lethal. Many congenital anomalies occur because of an abnormality in the genes (chromosomes) of the baby.

There is no test that can identify all possible genetic abnormalities. Fortunately, we do have ways to test for some of the most common. There are two categories of testing we offer during pregnancy.

- Carrier screening: This can also be done before a pregnancy. This is a blood test to see if you are a "carrier" for a disease: you aren't sick from the disease but you have an abnormal copy of a gene you could pass to a child.

- Aneuploidy testing: This evaluates THIS pregnancy's risk for having an abnormality in the chromosomes. There are non-invasive **screening tests** which require a blood test and sometimes an ultrasound. The results will tell you the percentage risk THIS pregnancy has for an abnormality in the chromosomes. **Diagnostic tests** require sampling of tissue around the growing baby or the fluid around the baby. They are more invasive but give a more definitive answer of an abnormality.

Who should have testing?

The American College of Obstetrics and Gynecology recommends ALL pregnant patients be offered prenatal genetic testing. They recommend ALL pregnant patients or patients considering pregnancy be offered carrier screening.

If I am young and everyone in my family has had healthy babies, do I really need testing?

When a genetic disorder occurs in a pregnancy, most of the time, it is due to a random genetic accident. And it is true that these accidents are more likely as men and women age. But to give an example, 80% of children with Down Syndrome are born to women under 35 years of age. Young healthy people are still at risk for having babies with birth defects, even without a family history of problems.

Can I have an ultrasound instead?

Every patient has an ultrasound of the baby at 18-20 weeks gestation regardless of genetic testing. It is possible to have a baby with a normal ultrasound AND a genetic abnormality. Ultrasound alone is not sufficient testing.

Is this testing covered by insurance?

Frequently, yes. But "covered" doesn't necessarily mean zero cost to you. The cost is out of our control. WE WILL GIVE YOU A NUMBER TO CALL TO DISCUSS INSURANCE COVERAGE.

Why should I do testing if we can't do anything to change the diagnosis?

It is always your choice whether or not to do testing. And it's true that we can't change anything or treat the baby during the pregnancy. But we do increase monitoring during the pregnancy if we know there is an abnormality since those babies tend to have more heart problems and growth problems. We can also have a plan in place for after delivery, in case the baby would need to see a specialist. It also allows you and your family to be able to prepare mentally and emotionally.

What is tested for in carrier screening?

The following page lists the common diseases we screen for, their symptoms, and how common they are:

- Cystic fibrosis- a chronic lung disorder that can be very mild or very severe with a shorter life expectancy
- Fragile X syndrome- causes mental retardation, autism, and hyperactivity. Boys are usually more severely affected but it affects both sexes.
- Spinal Muscular Atrophy- destroys nerve cells that affect voluntary movement. The most common form causes death between 2 to 4 years of age
- Additional testing may be recommended based on family history or background (for example, Tay Sachs if history of Jewish ancestry)

What happens if I am a carrier?

In most cases, the next step is to test the father of the baby. If the father of the baby is a carrier, there is a 25% chance that this baby will have the disease.

What can you test for in aneuploidy testing?

This includes but is not limited to:

- **Trisomy 21 (also known as Down Syndrome)** is the most commonly occurring genetic condition and occurs in 1 in 700 births. It occurs in all people of all races and economic levels. People with Down syndrome have an increased risk for certain medical conditions such as congenital heart defects, respiratory and hearing problems, Alzheimer's disease, childhood leukemia and thyroid conditions. Many of these conditions are now treatable. All people with Down syndrome experience cognitive delays, but the effect is usually mild to moderate and is not indicative of the many strengths and talents that each individual possesses

- **Trisomy 18 (also known as Edwards Syndrome)** occurs in 1 in 5,000 births. Nearly all parts of the body are affected and there are severe intellectual disabilities. Many of those affected die before birth. Survival beyond the first year of life is 5-25%.

- **Trisomy 13 (also known as Patau Syndrome)** occurs in 1 in 16,000 births. Similar to Trisomy 18, nearly all parts the body are affected and there are severe intellectual disabilities. Only 5-10 % survive beyond the first year of life.

What are options for tests?

We use Cell free fetal DNA (also called non-invasive prenatal testing or NIPT)-This test is a blood test (from mom's blood) that can detect fetal blood cells. It is safe for mom and baby because it is just a blood test. It has a 99% detection rate for Down syndrome. It screens for Down syndrome, Trisomy 18, Trisomy 13, and abnormalities in the sex chromosomes. This test will also report fetal sex.

Can I just use the test to learn if I am having a boy or girl?

In our office, no. This is not the recommended use of the test. There are labs that offer "gender only" testing using a similar technology. If you want to know fetal sex as early as possible but do not want to do the screening test, we offer a "sneak peak" ultrasound around 15-16 weeks.

What happens if I have an abnormal screening test?

This means the pregnancy has a higher chance of having a genetic abnormality. We recommend an appointment with a Maternal Fetal Medicine specialist (high risk specialist) who will review the test and offer diagnostic testing.