# GUIDE TO OPTIONAL GENETIC TESTING

What should I consider when deciding whether to have optional prenatal genetic testing?

It is your choice whether to have prenatal testing. Your personal beliefs and values are important factors in the decision about prenatal testing.

It can be helpful to think about how you would use the results of prenatal screening tests in your pregnancy care. Remember that a positive screening test tells you only that you are at higher risk of having a baby with Down syndrome or another aneuploidy. A diagnostic test should be done if you want to know a more certain result. Some parents want to know beforehand that their baby will be born with a genetic disorder. This knowledge gives parents time to learn about the disorder and plan for the medical care that the child may need.

Other parents do not want to know this information before the child is born. In this case, you may decide not to have follow-up diagnostic testing if a screening test result is positive. Or you may decide not to have any testing at all. There is no right or wrong answer.

**Please review the information in this packet and decide what testing, if any, you would like to have performed.**

**ANEUPLOIDY TESTING**

**Trisomy 21 (Down Syndrome), Trisomy 18, and Open Neural Tube Defects (Spina Bifida). You may choose one of the following options:**

**NIPT** (Non-Invasive Prenatal Testing or Cell Free- Fetal DNA): This simple and accurate non-invasive prenatal screening blood test offers results for early risk assessment of Down syndrome (Trisomy 21), or Trisomy 18 and other aneuploidy conditions. The test also offers an optional analysis for fetal sex. The test analyzes the relative amount of 21, 18, 13; X and Y chromosome material in circulating cell-free DNA from a maternal blood sample. It can also tell you the sex of the baby. This test can be performed at any time after 10 weeks gestation. If you elect this test, you will also have an **AFP** (alpha-fetoprotein) blood test between 15-21 weeks to test for open neural tube defects, such as spinal bifida and anencephaly and abdominal wall defects (which would not be detected with the NIPT test). Recommended follow up to a positive result is genetic counseling and prenatal diagnosis. (CPT Code: 81420 for NIPT, 82105 for AFP)

**Quad Screen:** This test is also known as the quadruple marker test. It is a test that measures levels of four analytes in a pregnant woman's blood—Alpha-fetoprotein (AFP), a protein made by the developing baby; Human chorionic gonadotropin (hcg), a hormone made by the placenta; Estriol, a hormone made by the placenta and the baby's liver; and Inhibin A, another hormone made by the placenta. Typically, the quad screen is done between weeks 15 and 20 of pregnancy—the second trimester and the results indicate whether the baby is at higher risk for Down Syndrome (Trisomy 21), Trisomy 18, and open

neural tube defects, such as spinal bifida and anencephaly, and abdominal wall defects. This is a screening test. Recommended follow up to a positive result is additional testing that is more definitive, as well as referral for genetic counseling and prenatal diagnosis. (CPT Codes: 82105, 84702, 86336, 82677)

In addition, the analytes in the QUAD screen can help to evaluate placental dysfunction and can be used for determination of risk for preterm delivery, fetal growth restriction, preeclampsia, placental abruption, intrauterine fetal demise, and perinatal death. If abnormal, it would indicate a need for more intense monitoring of both mother and baby.

**Detection Rate for NIPT and Quad Screen**

| | **Trisomy 21** | **Trisomy 18** | **Trisomy 13** |
|---|---|---|---|
| **NIPT** (FPR 0.1%) | <99% | <98% | 80% |
| **Quad Screen** (FPR 5%) | 83% | 80% | N/A |

(FPR is False Positive Rate)

**CARRIER SCREENING OPTIONS**

**Carrier screening is done to determine if a patient carries a genetic mutation for a particular disease, that MIGHT be passed along to her baby. Because this testing is done on the mother, and not the baby, it would not need to be repeated in any subsequent pregnancy.**

**Cystic Fibrosis:** Cystic Fibrosis is the most common inherited disease of children and young adults. The carrier frequency is 1 in 24 to 1 in 97. Both parents need to be carriers for a child to be affected (25% chance). One in 3500, children born are affected. Cystic fibrosis is a disorder of mucus production and produces abnormally thick mucus leading to life threatening lung infections, digestion problems, poor growth, infertility, and more. Symptoms range from mild to severe, but individuals with severe disease may die in childhood. With treatments today, people with Cystic Fibrosis can live into their 30's. CF does not affect intelligence. Recommended follow up to a positive result is testing of the baby's father. CPT Code: 81220

**Spinal Muscular Atrophy (SMA):** SMA is the most common inherited cause of early childhood death. The carrier frequency is 1 in 47 to 1 in 72 in the US and both parents need to be carriers for a child to be affected (25% chance). 1 in 11,000 children are affected. SMA is a progressive degeneration of lower motor neurons. Muscle weakness is the most common type with respiratory failure by the age of 2 years old. Muscles responsible for crawling, walking, swallowing, and head and neck control are most severely affected. Recommended follow up to a positive result is testing of the baby's father. CPT Code: 81401

**Fragile X Syndrome** (the most common inherited cause of developmental delays): Fragile X syndrome is an "X-linked" genetic disease, which means it is only carried by the mom. Unfortunately, 1 in 250 females are carriers and a child has a 50% chance of being affected if this is the case. 1 in 4,000 boys is affected with Fragile X and 1 in 8,000 girls is affected. Approximately 1/3 of all children born with Fragile X also have autism and hyperactivity. Recommended follow up to a positive result is genetic counseling and prenatal diagnosis. CPT Code: 81243

**NIPT**– Noninvasive Prenatal Testing (Cell-free fetal DNA) is among the newest and most sophisticated of techniques used to screen for certain genetic disorders. This testing involves direct examination of the DNA molecule itself in the mother's blood. These have 98-99% accuracy in detecting Trisomy 18, 21, and other chromosomal aneuploidies. However, it is also among the most expensive, and is not usually covered by insurance unless the patient is over the age of 35 and/or has a family history of genetic defects or a previous history of a child with a genetic abnormality.

The Quad screen is covered by most insurance plans. Testing for Cystic Fibrosis, SMA, and Fragile X testing is a covered benefit in many circumstances.

**Coverage Information**

Due to the complexity of coverage guidelines, we are unable to quote benefits or guarantee insurance coverage for any of these tests. Insurance benefits are plan-specific and offer vastly different coverage based on your policy. This handout explains the benefits to each test and provides the billing (CPT) codes for each test.

Even if the testing is covered, it could be applied to any unmet deductibles, and copays may apply, resulting in a bill. You are encouraged to contact your insurance company to obtain your benefits based on your age and risk factors, so that there are no surprises. The laboratories will bill you separately for any testing.

The following are **ICD-10 DIAGNOSIS CODES** that we may use, depending on your history:

**ANEUPLOIDY TESTING**

- Encounter for Antenatal screen: Z36.9
- Encounter for other screening for genetic and chromosomal anomalies: Z13.79
- Advanced Maternal Age (over age 35 at the time of delivery), 1st pregnancy: O09.519
- Advanced Maternal Age (over age 35 at the time of delivery), 2nd or subsequent pregnancy: O09.529

CARRIER TESTING

- Screening test of female genetic disease carrier status (no risk factors): Z31.430
- Family History of Genetic Disease Carrier Z84.81

To obtain a cost estimate for your specific situation, please contact your insurance company regarding the test you desire. Please provide them with the appropriate CPT code, which is located at the end of each test description, and the appropriate ICD-10 diagnostic code from the list above that applies to you.