Prenatal Genetic Testing

Prenatal genetic testing gives parents-to-be information about whether their baby has certain genetic disorders. There are two types of tests: prenatal screening tests and prenatal diagnostic tests.

Prenatal **screening** tests can tell you if the baby is at increased risk for possibly having a genetic disorder.

Prenatal **diagnostic** tests can tell you whether your baby **actually has certain** disorders. These tests are done with a very thin needle to collect a small amount of fluid from around the baby or from the placenta; *amniocentesis (15-20 weeks)* or *chorionic villus sampling (10-13 weeks).*

SCREENING TESTS OFFERED BY OUR OFFICE

1.) **Carrier screening tests:** These prenatal tests can be done before or during pregnancy by a blood sample. These tests are used to find out whether a person carries a gene for certain inherited disorders, which could then be passed to your baby. They only have to be performed once in your lifetime, not with every pregnancy. These tests include

- **Cystic Fibrosis (CF)**
- **Spinal Muscular Atrophy (SMA)**
- **Fragile X syndrome**
- **Hemoglobinopathies: Beta-Thalassemia or Sickle Cell disease**

If you choose to have a full panel of screening (**Inheritest Carrier Screen Society Guided Panel**) can be selected which includes: CF, SMA, Hemoglobinopathies: Beta-Thalassemia and Sickle Cell disease, Fragile X syndrome, Bloom syndrome, Canavan disease, Familial dysautonomia, Fanconi anemia group C, Gaucher disease, Mucolipidosis type IV, Niemann-Pick disease type A and B, and Tay-Sachs disease.

2.) **Prenatal genetic screening:** These are tests that are done from a pregnant women's blood. These tests can screen the baby for abnormal chromosomes such as Down syndrome, defects of the brain and spine called Neural Tube Defects, and some defects of the abdomen, heart and face.

- **MaterniT21** with sex chromosomal analysis (cell-free DNA test)
  - Collection of blood anytime after 10 weeks
  - Cell-free DNA is the small amount of DNA that is released from the placenta into a pregnant woman's bloodstream. This is used to screen for Down syndrome (Trisomy 21), Edward's syndrome (Trisomy 13), Patau syndrome (Trisomy 18), and problems with the number of sex chromosomes. This test can be done between 9 weeks until the end of pregnancy. This testing is not recommended for a woman carrying more than one baby (twins or more).

- **Quad Marker**
  - Collection of blood between 15-22 weeks
  - Test that evaluates for substances in the pregnant woman's blood stream. This is used to screen for Down Syndrome (Trisomy 21) Edward's syndrome (Trisomy 13), Patau syndrome (Trisomy 18), and Neural Tube defects.

*** **Of the above tests, only ONE is to be selected.**

- If the MaterniT 21is selected, then alpha-fetoprotein (AFP) can be offered between 15-22 weeks to screen for Neural Tube or abdominal wall defects.

TEST RESULTS

A **negative** screening test result means that your fetus is at a lower risk of having the disorder compared with the general population. It does NOT completely rule out the possibility that your fetus has a disorder.

A **positive** screening test result means that your fetus is at higher risk of having a disorder compared with the general population. It **does NOT** mean that your fetus DEFINITELY has the disorder. After a positive screening result, we will refer you to the high-risk pregnancy specialist (Perinatologist/Maternal Fetal Medicine) who will perform a detailed ultrasound and offer you the option of diagnostic testing with *CVS or amniocentesis,* which are diagnostic tests that will tell if your baby has the actual disorder.

***These screening tests could result in a false-positive result (shows there is a problem when there isn't one) or a false-negative result (shows there is not a problem when there is one).

WHEN TO TEST

1). Who is considered a high-risk patient that may be more likely to consider doing a prenatal genetic screening test?

- Women with certain abnormalities found during ultrasounds of the baby.
- Women who have had a previous child with aneuploidy (abnormal number of chromosomes) such as Down syndrome.
- Women over the age of 35.

2.) How to decide whether to have a prenatal genetic test:

- 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.
- Some parents want to know before the child is born that their baby may 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. Some parents may decide to end the pregnancy in certain situations.
- Other parents do not want to know this information before the child is born, as they feel it wouldn't change any plans for them.
- There is no right or wrong answer. The choice to get prenatal genetic/diagnostic testing varies between parents.

If you desire prenatal genetic testing, please follow the instructions below BEFORE YOUR NEXT OB APPOINTMENT .

1.) Contact Integratedgenetics.com/transparency or 1-844-799-3243 to help clarify coverage.
   - Helpful questions and resources:
     - Will the test be covered?
     - Is the test subject to a deductible/copay?
     - Does this test require a pre-authorization? **--Integrated Genetics will commonly perform pre-authorization when you call but some insurance companies require a pre-authorization from our office which may take up to 2 weeks to perform prior to the date of desired testing.**

2.) The following is a list of insurance ICD10 and CPT codes that your insurance may ask about

- **ICD10 Codes**
  - High Risk Diagnosis Codes:
    - **O35.1xx0** Maternal care for suspected chromosomal abnormality in fetus
    - **O35.2xx0** Maternal care for suspected hereditary disease in fetus
    - **O09.519** Supervision of elderly primigravida, unspecified trimester
    - **O09.529** Supervision of elderly multigravida, unspecified trimester
    - **O28.3** Abnormal ultrasound findings on antenatal screening of mother
  - Low Risk Diagnosis Codes:
    - **Z34.90** Encounter for supervision of normal pregnancy, unspecified trimester
    - **Z31.430** Encounter for female for testing for genetic disease carrier status for procreative management.
- **CPT Codes:**
    - Inheritest society guided panel: 81412
    - Cystic Fibrosis (CF): 81220
    - Spinal Muscular Atrophy (SMA): 81329
    - Fragile X: 81243
    - Hemoglobin electrophoresis: 85660
    - Nuchal Translucency sono: 76813
    - Sequential 1: 81508 (Includes 84163, 84702)
    - Sequential 2: 81511 (82105,82677,84702,86336)
    - MaterniT21: 81420
    - Quad Marker: 81511 (Includes: 82105,82677,84702,86336)
    - Alpha-fetoprotein: 82105