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Healthcare For Women

OBSTETRICS and GYNECOLOGY

# Genetic Testing
# Obstetrics Booklet


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## GENETIC TESTING

PLEASE BE AWARE THAT THESE SCREENING TESTS ARE OPTIONAL. THEY ARE NOT NECESSARILY COVERED BY YOUR INSURANCE COMPANY. PLEASE BE SURE TO CHECK WITH YOUR INSURANCE PRIOR TO GETTING ANY OF THESE TESTS PERFORMED. At the end of this booklet, we are providing a list of the tests that include CPT codes and diagnosis codes. Refer to this handout when calling your insurance company to inquire about coverage.

## FETAL CHROMOSOME ABNORMALITY SCREENING

Fetal chromosomal abnormality screening is a personal decision since the usefulness of diagnosis depends on what one would choose to do with the result. The following tests can be performed in our office or with a Maternal Fetal Medicine (MFM) Specialist. **You may choose not to perform any tests** or have the option of the following:

1. **Non-Invasive Prenatal testing:** Cell Free DNA
2. **Invasive Tests:** Chorionic Villus Sampling (CVS), Amniocentesis

These tests are looking for chromosomal abnormalities of the baby. The risk of these abnormalities increase with maternal age. However, younger women give birth to the majority of these children because younger women have the majority of pregnancies.

First Trimester Risk of Down Syndrome or all chromosomal abnormalities

| AGE | Down Syndrome | ALL |
|---|---|---|
| 33 | 1/400 | 1/290 |
| 34 | 1/310 | 1/230 |
| 35 | 1/240 | 1/180 |
| 36 | 1/180 | 1/135 |
| 37 | 1/130 | 1/99 |
| 38 | 1/95 | 1/72 |
| 39 | 1/71 | 1/53 |
| 40 | 1/52 | 1/39 |
| 41 | 1/40 | 1/30 |
| 42 | 1/32 | 1/23 |
| 43 | 1/27 | 1/19 |

## NON-INVASIVE TESTING

Non-Invasive genetic testing is an elective test performed in the first trimester with the benefit of an early diagnosis and less possibility of complications in the event an abnormality is identified and intervention is requested. There are 3 parts to the test - maternal blood work at 10-12 weeks, an ultrasound of the baby at 12-14 weeks and maternal blood work again at 15-22 weeks. Coverage of each test can vary with insurance. Information for checking with your insurance about coverage is included at the end of the packet.

### Cell Free DNA

This screening test is based on the newest advances in non-invasive prenatal testing. This test is performed any time after 10 weeks and consists of a maternal blood draw. This test assesses the risk of Trisomy 21 (Down syndrome), and Trisomies 13 and 18 as well as the sex chromosome for abnormalities. It has a detection rate of up to **99%** and false positive rates as low as 0.1% for trisomy 21, 18 and 13.

### Nuchal Translucency Ultrasound

A nuchal translucency ultrasound is performed between 11 weeks 4 days and 13 weeks 6 days. This specifically looks at the neck thickness of the baby and very early limited anatomy. This test can identify other potential abnormalities that the cell free DNA blood work does not assess for.

### Maternal Serum AFP

This is a screening test performed between 15-22 weeks to screen for fetal spinal abnormalities and consists of a maternal blood draw.

## INVASIVE TESTING

Invasive testing is reserved for women who electively choose to do this testing, are over the age of 35, or those who have abnormal non-invasive testing or abnormal ultrasound findings. Invasive testing tests ALL possible chromosomes for abnormalities in comparison to the non-invasive testing which tests the 3 most common chromosomes (21, 18, 13). There are 2 different tests and which one is done depends on the trimester of the pregnancy. This testing is performed by a Maternal Fetal Medicine Specialist. Your Healthcare for Women OB/GYN will refer you to this specialist if you choose to pursue this test.

### First Trimester

#### Chorionic Villus Sampling (CVS)

Chorionic villus sampling generally is performed at 10-13 weeks of gestation. To perform this test, a very thin needle is used to withdraw a small amount of placental tissue. This is either done through the maternal abdomen or cervix. The primary advantage of CVS over amniocentesis is that results are available much earlier in pregnancy which allows for more time for advanced testing and to discuss options with specialists. CVS carries a diagnostic accuracy of greater than 99% with total pregnancy loss rates of 1/455.

### Second Trimester

#### Amniocentesis

Amniocentesis usually is offered between 16 and 20 weeks of gestation but can be performed up until delivery if indicated. To perform this test, a very thin needle is used to withdraw a small amount of amniotic fluid. This is done through the maternal abdomen. The cells floating in amniotic fluid are cultured to yield enough samples for chromosomal study. Amniocentesis also carries a diagnostic accuracy of greater than 99% with total pregnancy loss rate of 1/900. The advantage of amniocentesis over the CVS is the lower complication and fetal loss rate.

## CARRIER SCREENING TESTING

Carrier screening is a type of genetic test that can tell whether a mother or father carry a gene for a certain genetic disorder that can be passed on to their children. A carrier is a person who has only one gene for a disorder. Carriers usually do not have symptoms or have only mild symptoms. Because they often do not know that they carry a genetic disorder, they can pass the gene on to their children. The specific genetic disorders tested here require two genes for a person to have the disorder. Therefore, both the mother and the father would have to carry the same genetic abnormality, for the possibility that their child will be affected by that specific condition.

Carrier screening can be done at any time, pregnant or not pregnant. It is a blood test and it only needs to be done one time in an individual's life as the results will not change during a lifetime. Carrier screening is a voluntary decision. You can choose to have carrier screening or not choose to do so. Only one individual of a couple is tested and if a test result shows that the first partner is a carrier, the other partner is tested. The American College of Obstetrics and Gynecology recommends *offering* carrier screening for spinal muscular atrophy (SMA) and cystic fibrosis (CF) and Fragile X testing. Please see below for specific information regarding these conditions.

It is also possible to be tested for more than 100 different disorders. Please inform your OB if you are interested in this additional testing.

**Please be aware that these screening tests are optional and insurance may or may not cover them. You may call your insurance company and use the attached test codes to communicate what tests you are interested in obtaining to see what your coverage is. You may elect to bypass your insurance and pay for the testing out of pocket.**

**Spinal Muscular Atrophy**

A hereditary disease that progressively destroys lower motor neurons—nerve cells in the brain stem and spinal cord that control essential voluntary muscle activity such as speaking, walking, breathing, and swallowing. Over time, the ability to control voluntary movement can be lost. There is no cure for SMA. The prognosis varies depending on the type of SMA. Some forms of SMA are fatal.

SMA is caused by defects in the gene SMN1, which makes a protein that is important for the survival of motor neurons (SMN protein). SMA disorders in children are inherited in an ***autosomal recessive*** manner. Autosomal recessive means the child must inherit a copy of the defective gene from **both** parents. These parents are asymptomatic (without symptoms of the disease).

Kennedy's disease, an adult form of SMA, is **X-Linked inherited** which means the asymptomatic mother carries the defective gene on one of her X chromosomes and has a 50% chance of passing the disorder along to her sons. Daughters have a 50 percent chance of inheriting their mother's faulty X chromosome and will inherit a safe X chromosome from their father, which would make them asymptomatic carriers of the mutation. *Approximately 1 in 50 individuals is a carrier for SMA and one in every 10,000 births will be affected by SMA.*

**Cystic Fibrosis**

A hereditary disease that mainly affects the mucous of the lungs, pancreas, liver, intestines, sinuses, and sex organs. The mucus becomes thick and sticky to build up in these organs to block airways, increase risk for infections, as well as cause severe constipation, malnutrition, electrolyte imbalances, diabetes, and infertility. The symptoms and severity of CF can vary. There is no cure for CF. Respiratory failure is the most common cause of death. Improvements in screening and treatments mean people with cystic fibrosis may now live into their 60s.

CF is caused by a defect in the CFTR gene, which makes a protein that controls the salt and water in the body's cells. CF is inherited in an autosomal recessive manner. Autosomal recessive means the child must inherit a copy of the defective gene from both parents. These parents are asymptomatic (without symptoms of the disease). *Depending on heritage, approximately 1/25 to 1/200 individuals are a carrier for CF and one in 2,500 births will be affected by CF*.

**Fragile X**

A hereditary disease that causes a range of developmental problems including learning disabilities and cognitive impairment. Usually, males are more severely affected than females. They may also have attention deficit disorder (ADD) or disorders that affect communication and social interaction, such as autism or spectrum disorders. Most males and about half of females have characteristic physical features, including a long/narrow face, large ears and prominent jaw and forehead. There is no cure for Fragile X. Treatment involves training and education of the affected individuals and caregivers.

Fragile X is inherited in an X-linked dominant pattern. This means that the disorder is located on the X-chromosome and one copy of the gene is sufficient to cause the condition, therefore both females and males can be affected. Boys have only one X- chromosome, therefore a single fragile X is likely to affect them more severely. Parents do not have to have symptoms of Fragile X to be able to pass it on to their children. *Approximately 1/200 females is a carrier for Fragile X and 1/250-800 males*.

**Ashkenazi Jewish Heritage Screening**

This panel consists of the diseases frequent in the Ashkenazi Jewish population that have been recommended for population based carrier screening by the American college of Obstetricians and Gynecologists (ACOG). Most of these diseases are severe and can cause early death, but some can be treated to reduce symptoms and prolong life. Both parents would need to be carriers in order for their children to be affected by these diseases. This panel screens for Cystic Fibrosis, Tay-Sachs, Canavan Disease, Familial Dysautonomia, Gaucher Disease, Fanconi Anemia type C, Bloom Syndrome, Niemann-Pick Disease Type A, Mucolipidosis Type IV, Glycogen Storage Disease, and Maple Syrup Urine Disease.

## INSURANCE COVERAGE INFORMATION

**Since these screening tests are optional, insurance may or may not cover them. You may call your insurance company and use the attached test codes to communicate what tests you are interested in obtaining to see what your coverage is. You may elect to bypass your insurance and pay for any testing out of pocket.**

| Name of Test | Diagnosis Code* | CPT Code |
|---|---|---|
| Cell Free DNA (NIPT) | Z36.8 | 92777 |
| Nuchal Translucency Ultrasound | Z36.82 | 76813 |
| Maternal Serum AFP (msAFP) | Z36.1 | 0274-1 |
| Carrier Screen | Z13.71 | 94372 or 14230 |

***** If older than age 35 at time of estimated due date, please also provide diagnosis code O09.511 (Advanced Maternal Age)