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GENETIC TESTING BOOKLET

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GENETIC TESTING

PLEASE BE AWARE THAT THESE SCREENING TESTS ARE THE OPTIONS AVAILABLE TO YOU; BUT, 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 coverage.

FETAL CHROMOSOME ABNORMALITY SCREENING

Fetal chromosomal abnormality screening and genetic screening is a personal decision since the usefulness of diagnosis depends on what one would choose to do with the result. All of the following tests can be performed in our office or a Maternal Fetal Medicine Specialist. There are three options available to you:

1-**Non-Invasive testing:** Sequential Screens part 1 & 2, Maternal Serum Single and Quad Screen, cell-Free DNA

2-**Invasive testing:** Chorionic Villus Sampling (CVS), Amniocentesis

3-**You can choose to not have any tests performed at all**

**Note: Trisomy 21 (Down Syndrome), Trisomy 18, and Trisomy13 are chromosomal disorders that cause physical and mental retardation and birth defects. The risk of these abnormalities increases with maternal age. However, younger women give birth to the majority of these children because younger women have the majority of pregnancies. The *non-invasive screening* tests are used to identify those women who are not known to be at high risk but are nevertheless carrying a fetus with chromosomal abnormality. *The invasive screening* tests are usually offered to women who will be age 35 years and older at delivery; however, they may also choose to proceed with non-invasive screening tests while understanding the limitation of those tests.

Midtrimester Risk of Down Syndrome or all chromosomal abnormalities

AGE	DS	ALL
33	1/417	1/208
34	1/333	1/152
35	1/250	1/132
36	1/192	1/105
37	1/149	1/83
38	1/115	1/65
39	1/89	1/53
40	1/69	1/40
42	1/53	1/31
42	1/41	1/25
43	1/31	1/19

NON INVASIVE TESTING

First Trimester: the benefit of the first trimester screening is the early diagnosis and less complication with possible intervention in the first trimester.

1-Nuchal Translucency Screening Sequential Screen Part 1

This screening test consists of an ultrasound and a maternal blood test. The ultrasound is performed Between 11 weeks 4 days, and 13 weeks 6 days. The blood test is performed anytime from 10 weeks to 13 weeks 6 days. The ultrasound will measure the clear area (fluid accumulation) behind the neck of the fetus. The maternal blood is analyzed for free beta human chorionic gonadotropin and pregnancy associated plasma protein A. The results of the ultrasound will be combined with the results of the blood test to estimate a specific risk for Down syndrome and Trisomies 18 and 13. This test can identify up to 95% of Down syndrome pregnancies with a 5% false positive rate. There is a second blood draw dome between 15-22 weeks.

2-Cell-Free DNA

This test is based on the newest advances in non-invasive prenatal testing. This test is performed after 10 weeks. It is a simple maternal blood test that has been shown in clinical studies to detect fetal trisomies with high accuracy. This test assesses the risk of three fetal trisomies by measuring the relative amount of chromosomes in maternal blood. It has shown to have detection rate of up to 99% and false positive rates as low as 0.1% for trisomy 21, 18 and 13.

Second Trimester

1-Quad Screen: maternal blood test only

Maternal blood sampling can be performed between 15 and 20 weeks of gestation but is most accurate when performed between 16 and 18 weeks of gestation. Accurate pregnancy dating is essential. The maternal blood is analyzed for four different hormones:

- Maternal serum alpha fetoprotein (MS AFP)
- Human chorionic gonadotropin (hCG)
- Estriol
- Dimeric Inhibin A

This test will detect up to 85% of Down Syndrome pregnancies at 7% false positive rate.

2-Maternal Serum AFP

Performed between 15-22 weeks. This test screens for spinal abnormalities. This individual test is not necessary to perform if performing the sequential screen since this is included in the 2nd part of the sequential screen.

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 and 13).

First Trimester

Chorionic Villus Sampling

Chorionic villus sampling generally is performed at 10-12 weeks of gestation. Placental villi may be obtained through trans-cervical or trans-abdominal access to placenta. 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 diagnostic accuracy of greater than 99% with total pregnancy loss rates of 1/160,

Second Trimester

Amniocentesis

Amniocentesis usually is offered between 15 and 20 weeks of gestation. Amniotic fluid is obtained through trans-abdominal access under continuous ultrasound guidance. The cells floating in amniotic fluid are cultured to yield enough samples for chromosomal study. Amniocentesis also carries diagnostic accuracy of greater than 99% with total fetal loss rate of less than 1/400. 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 you whether you carry a gene for a certain genetic disorder that can be passed on to your children. For some genetic disorders, it takes two genes for a person to have the disorder. 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 have a gene for the disorder, they can pass the gene on to their children.

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 as the results will not change during your lifetime. Carrier screening is a voluntary decision. You can choose to have carrier screening or not choose to do so.

The American College of Obstetrics and Gynecology recommends *offering* carrier screening for spinal muscular atrophy (SMA) and cystic fibrosis (CF). We offer SMA and CF testing as well as 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. If you are interested in this, it can be done with a Maternal Fetal Medicine (MFM) specialist. Please inform your physician if you are interested in this additional testing.

Spinal Muscular Atrophy (SMA)

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 (SF)

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 now may live into their mid-to late 30s, on average, and some are living into their 40s and 50s.

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.

Ashkenazi Jewish Carrier

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.

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. Fragile X occurs in ~1 in 4,000 males and 1 in 8,000 females.

HOW TO DETERMINE IF YOUR INSURANCE WILL COVER PRENATAL DIAGNOSIS SCREENING TESTS

Provide your insurance company with all the Diagnosis Codes & CPT codes related to the prenatal screening test that you are interested in from the list below:

Fetal Chromosome Screening Tests: **ICD-10 (Diagnosis Code): z36**			
Test Name	**CPT Code**	**Test Compoments**	**Self-Pay Price**
Cell-free DNA	76813	Fetal NuchalTranslucency / Ultrasound	**$250.00**
	82105	AFP (maternal serum)	**$195/$150**

	81420 (add 81422w/ micros)	Fetal chromosomal aneuploidy (e.g., trisomy 21, monosomy X) genomic sequence analysis panel, circulating cell-free fetal DNA in maternal blood, can include	
Quad screen	86336	Inhibin A	**$102.00**
	82105	AFP (maternal serum)	
	84702	HCG	
	82677	Estriol	
Sequential Screen (P1&2)	76813	Fetal NuchalTranslucency / Ultrasound	**$250.00**
	84163	PAPP-A	**$181.50**
	84702	HCG	
	86336	Inhibin A	
	82105	AFP (maternal serum)	
	82677	Estriol	

Genetic Carrier Screening Tests:
ICD-10 (Diagnosis Code): z31.40, z31.5

Test Name	**CPT Code**	**Test Components**	**Self-Pay Price**
Spinal Muscular Atrophy	81400	SMA	**$50**
Cystic Fibrosis	81220	Cystic Fibrosis Carrier Testing	**$125**
Fragile X	81243	Fragile X	**$50**
Ashkenazi Jewish with CF	81243	Ashenazi Jewish carrier and CF	**$175**

How to Determine if Your Insurance Will Cover Common Prenatal Diagnosis Screening Tests

Provide your insurance company with all the Diagnosis Codes & CPT codes related to the prenatal screening test that you are interested in from the list below:

Fetal Chromosome Screening Tests: **ICD-10 (Diagnosis Code): z36**			
Test Name	**CPT Code**	**Test Compoments**	**Self-Pay Price**
Cell-free DNA	76813	Fetal nuchal translucency measurement	**$195/150**
	82105	AFP (maternal serum)	
	81420 (add 81422w/ micros)	Fetal chromosomal aneuploidy (e.g., trisomy 21, monosomy X) genomic sequence analysis panel, circulating cell-free fetal DNA in maternal blood, can include	
Quad screen	86336	Inhibin A	**$102**
	82105	AFP (maternal serum)	
	84702	HCG	
	82677	Estriol	
Sequential Screen (P1&2)	76813	Fetalnuchaltranslucency	**$181.50**
	84163	PAPP-A	
	84702	HCG	
	86336	Inhibin A	
	82105	AFP (maternal serum)	
	82677	Estriol	
Genetic Carrier Screening Tests: **ICD-10 (Diagnosis Code): z31.40, z31.5**			
Test Name	**CPT Code**	**Test Components**	**Self-Pay Price**
Spinal Muscular Atrophy	81400	SMA	**$35**
Cystic Fibrosis	81220	Cystic Fibrosis Carrier Testing	**$125**
Fragile X	81243	Fragile X	**$40**
Ashkenazi Jewish with CF	81243	Ashenazi Jewish carrier and CF	**$200**