Prenatal Genetic Screening

MaterniT 21 Non-Invasive Prenatal Testing – CPT: 81420

- Analyzes genetic information that enters the bloodstream from the placenta. It screens for certain chromosomal abnormalities that could affect your baby's health and development such as Trisomy 21 (Downs), 18, and 13. It can also detect the sex of the fetus. Most women will screen negative for chromosomal abnormalities and may not require further testing. *Recommended follow up to a positive result: genetic counseling and prenatal diagnosis*

First Trimester/Sequential Screen – CPT: 84163, 84702,86336, 84163, 84702

- First Trimester Screening offers early information about a baby's risk of Down Syndrome and Trisomy 18. Testing combines blood testing and an ultrasound to measure the baby's neck (nuchal translucency) between 11 and 14 weeks of pregnancy. This screen is performed at Maternal Fetal Medicine. The Sequential Screening is then performed between 15 and 18 weeks of pregnancy.

Maternal Marker Screen/AFP4/Quad AFP – CPT: 82105, 82677, 84702, 86336

- AFP is a screening test to calculate a baby's risk of Down Syndrome, Trisomy 18, and Open Neural Tube Defect. If the results are positive, you are considered at high risk for having a baby with Down Syndrome, Trisomy 18, or an open neural defect. If the results are negative, you are considered at low risk for having a baby with Down Syndrome, Trisomy 18, or an open neural defect.

INSURANCE COVERAGE: Coverage of these tests are subject to copays, coinsurance, and deductibles. Certain insurances may require pre-authorization and criteria to be met.

Visit www.integratedgenetics/transparency to determine your cost for MaterniT or call 844-799-3243.

I understand that:

- These tests do not detect all carriers of the diseases and cannot diagnose the condition definitively.
- The decision to have carrier testing is completely mine

I have read this form, or had the form read to me, and have been given the chance to ask questions regarding the above diseases and testing.

__ I accept MaternitT 21 Testing
__ I accept Frist Trimester/Sequential Screen Testing
__ I accept Maternal Marker Screen Testing

__ I decline MaternitT 21 Testing
__ I decline Frist Trimester/Sequential Screen Testing
__ I decline Maternal Marker Screen Testing
__ I decline all testing

_____
Patient's Name

_____ _____
Patient's Signature Date

_____ _____
Witness Signature Date

_____ _____
Provider's Signature Date

Screening in Pregnancy for Common Genetic Diseases

Cystic Fibrosis, Spinal Muscular Atrophy, Tay Sachs, and Fragile X are a few common serious disorders that can occur even without a family history. These tests are one time only simple blood tests that screen to determine if you are a carrier of the specific gene. A carrier is a person who has a gene that increases the risk to have children with a specific genetic disease. People do not know they are carriers until they have a blood test or an affected child. A negative test result significantly lowers but does not completely eliminate the risk of being a carrier. Carrier testing is not able to detect all the genetic abnormalities that cause a particular disease. These tests can be done either when you are planning a pregnancy or after you have become pregnant. The details of each genetic disease are listed below and are seen in all ethnicities and considered the most common.

Society Guided Screening Panel – CPT: 81200, 81209, 81220, 81242, 81251, 81255, 81260, 81290, 81330, 81361, 81401

- Includes 12 Genes
    - Beta Hemoglobinopathy
    - Bloom Syndrome
    - Canavan Disease
    - Familial Dysautonomia
    - Fanconi anemia group C
    - Gaucher Disease
    - Mucolipidosis
    - Niemann-Pick Disease (types A and B)
    - Cystic Fibrosis
    - Spinal Muscular Atrophy
    - Fragile X syndrome
    - Tay-Sachs Disease

Inheritest Core Panel – CPT: 81329, 81220, 511919

- Includes 3 Genes
    - **Cystic Fibrosis (the most common inherited disease of children and young adults):** CF is a disorder of mucus production and produces abnormally thick mucus leading to life threatening lung infections, digestion problems, poor growth and more. Symptoms range from mild to severe. The carrier frequency is 1 in 24 to 1 in 97 and both parents need to be carriers for a child to be affected (25% chance). One in 3500 children born are affected. *Recommended follow up to a positive result: test partner.*
    - **Spinal Muscular Atrophy (SMA) (the most common inherited cause of early childhood death)**: 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. The carrier frequency is 1 in 47 to 1 in 42 in the US and both parents need to be carriers for a child to be affected (25% chance). One in 11,000 children are affected. *Recommended follow up to a positive result: test partner.*
    - **Fragile X Syndrome (the most common inherited cause of developmental delays)**: Unlike CF and SMA, this is an x-linked genetic disease and only carried in 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 4000 boys are affected with Fragile X and 1 in 8000 girls are affected. Approximately 1/3 of all children born with Fragile X also have autism and hyperactivity. *Recommended follow up to a positive result: genetic counseling and prenatal diagnosis.*

Cystic Fibrosis 97 – CPT: 81220

- CF is a disorder of mucus production and produces abnormally thick mucus leading to life threatening lung infections, digestion problems, poor growth and more. Symptoms range from mild to severe. The carrier frequency is 1 in 24 to 1 in 97 and both parents need to be carriers for a child to be affected (25% chance). One in 3500 children born are affected. *Recommended follow up to a positive result: test partner.*

INSURANCE COVERAGE: Coverage of these tests are subject to copays, coinsurance, and deductibles. Certain insurances may require pre-authorization and criteria to be met.

Visit www.integratedgenetics/transparency to determine your cost for Society Guided Panel or call 844-799-3243

Carrier Screening in Pregnancy for Common Genetic Diseases

I understand that:

- These tests do not detect all carriers of the diseases
- The decision to have carrier testing is completely mine
- If I am a carrier for CF or SMA, testing of my partner will help determine the chance that my baby will be affected. If I am a carrier for CF, SMA, and/or Fragile X Syndrome, further prenatal testing is available to see if my fetus has inherited the abnormal gene.

I have read this form, or had the form read to me, and have been given the chance to ask questions regarding the above diseases and testing.

__ I accept Society Guided Carrier Testing
__ I accept Inheritest Core Panel
__ I accept Cystic Fibrosis Screen Testing

__ I decline Society Guided Carrier Testing
__ I decline Inheritest Core Panel
__ I decline Cystic Fibrosis Screen Testing
__ I decline all testing

_____
Patient's Name

_____ _____
Patient's Signature Date

_____ _____
Witness Signature Date

_____ _____
Provider's Signature Date

Definitions of Society Guided Screening Genes

Beta Hemoglobinopathy: Beta hemoglobinopathies are a group of inherited disorders of red blood cells characterized by mild to severe anemia. Individuals with beta hemoglobinopathies have defects in one of the beta-globin chains of hemoglobin, the oxygencarrying molecule in the blood. Symptoms of beta-hemoglobinopathies are due to structurally abnormal hemoglobins, or to reduced or absent production of hemoglobins.

Bloom Syndrome: Bloom syndrome is a rare genetic disorder characterized by short stature; increased skin sensitivity to ultraviolet rays from the sun (photosensitivity); multiple small dilated blood vessels (telangiectasia) over the nose and cheeks resembling a butterfly in shape; mild immune deficiency with increased susceptibility to infections; and most importantly, a markedly increased susceptibility to many types of cancer, especially leukemia, lymphoma and gastrointestinal tract tumors. Bloom syndrome is a prototype of a group of genetic conditions known as chromosome breakage syndromes. The genetic abnormality in Bloom syndrome causes problems with DNA repair, resulting in a high number of chromosome breaks and rearrangements.

Canavan Disease: Canavan disease is rare genetic neurological disorder characterized by the spongy degeneration of the white matter in the brain. Affected infants may appear normal at birth, but usually develop symptoms between 3-6 months of age. Symptoms may include an abnormally large head (macrocephaly), lack of head control, severely diminished muscle tone resulting in "floppiness," and delays in reaching developmental milestones such as independent sitting and walking. Most affected children develop life-threatening complications by 10 years of age.

Familial Dysautonomia: Familial dysautonomia is a genetic disorder that affects the development and survival of certain nerve cells. The disorder disturbs cells in the autonomic nervous system, which controls involuntary actions such as digestion, breathing, production of tears, and the regulation of blood pressure and body temperature. It also affects the sensory nervous system, which controls activities related to the senses, such as taste and the perception of pain, heat, and cold. Problems related to this disorder first appear during infancy. Early signs and symptoms include poor muscle tone (hypotonia), feeding difficulties, poor growth, lack of tears, frequent lung infections, and difficulty maintaining body temperature. Older infants and young children with familial dysautonomia may hold their breath for prolonged periods of time, which may cause a bluish appearance of the skin or lips (cyanosis) or fainting. This breath-holding behavior usually stops by age 6. Developmental milestones, such as walking and speech, are usually delayed, although some affected individuals show no signs of developmental delay.

Fanconi Anemia Group C: Fanconi anemia group C is an inherited disease characterized by physical abnormalities, bone marrow failure, and increased risk of cancer. Individuals with Fanconi anemia group C have defects in the FANCC protein, which is important in the process of DNA repair. Symptoms associated with Fanconi anemia group C are attributed to the inability to correct normally occurring DNA errors. This leads to abnormal cell death or uncontrolled cell growth, which may cause physical abnormalities and lead to bone marrow failure or cancers.

Gaucher Disease: Gaucher disease is an inherited disorder that affects many of the body's organs and tissues. The signs and symptoms of this condition vary widely among affected individuals. Researchers have described several types of Gaucher disease based on their characteristic features. Type 1 Gaucher disease is the most common form of this condition. Type 1 is also called non-neuronopathic Gaucher disease because the brain and spinal cord (the central nervous system) are usually not affected. The features of this condition range from mild to severe and may appear anytime from childhood to adulthood. Major signs and symptoms include enlargement of the liver and spleen (hepatosplenomegaly), a low number of red blood cells (anemia), easy bruising caused by a decrease in blood platelets (thrombocytopenia), lung disease, and bone abnormalities such as bone pain, fractures, and arthritis. Types 2 and 3 Gaucher disease are known as neuronopathic forms of the disorder because they are characterized by problems that affect the central nervous system. In addition to the signs and symptoms described above, these conditions can cause abnormal eye movements, seizures, and brain damage. Type 2 Gaucher disease usually causes life-threatening medical

problems beginning in infancy. Type 3 Gaucher disease also affects the nervous system, but it tends to worsen more slowly than type 2.

Mucolipidosis: The mucolipidoses (ML) are a group of inherited metabolic diseases that affect the body's ability to carry out the normal turnover of various materials within cells. In ML, abnormal amounts of carbohydrates and fatty materials (lipids) accumulate in cells. Because our cells are not able to handle such large amounts of these substances, damage to the cells occurs, causing symptoms that range from mild learning disabilities to severe intellectual impairment and skeletal deformities. Symptoms of ML can be congenital (present at birth) or begin in early childhood or adolescence. Early symptoms can include vision problems and developmental delays. Over time, many children with ML develop poor mental capacities, have difficulty reaching normal developmental milestones, and, in many cases, eventually die of the disease.

Niemann-Pick Disease (Types A & B): Niemann-Pick disease, type A or B, is characterized by enlarged liver and spleen, progressive lung disease, and failure to gain weight and grow as expected. Niemann-Pick type A also leads to progressive loss of intellectual and motor skills.1 The signs and symptoms of Niemann-Pick types A and B result from the inability of the body to properly break down a lipid known as sphingomyelin, which accumulates in various organs in the body.1 Niemann-Pick disease, type A or B, is also known as acid sphingomyelinase deficiency and belongs to a group of diseases called lysosomal storage disorders.2 This group includes Niemann-Pick disease type C, which is genetically and clinically distinct.

Tay-Sachs Disease: Tay-Sachs disease is a rare inherited disorder that progressively destroys nerve cells (neurons) in the brain and spinal cord. The most common form of Tay-Sachs disease becomes apparent in infancy. Infants with this disorder typically appear normal until the age of 3 to 6 months, when their development slows and muscles used for movement weaken. Affected infants lose motor skills such as turning over, sitting, and crawling. They also develop an exaggerated startle reaction to loud noises. As the disease progresses, children with Tay-Sachs disease experience seizures, vision and hearing loss, intellectual disability, and paralysis. An eye abnormality called a cherry-red spot, which can be identified with an eye examination, is characteristic of this disorder. Children with this severe infantile form of Tay-Sachs disease usually live only into early childhood.