

## Prenatal Genetic Screening

**Patient Name:** \_\_\_\_\_ **Date:** \_\_\_\_\_

Genetic Screening/ Teratology Counseling  
(Includes patient, baby's father or anyone in either family with: )

1. Patient's age greater than 35 years as of estimated date of delivery

Y                      N

If you answer Y (yes) to question #1, we recommend high risk counseling with a Perinatal Specialist to help you decide on further testing, such as first trimester screening, ultrasound or amniocentesis. First trimester screening includes free Beta, PAPP-A and nuchal translucency.

I **accept** high risk counseling. Please schedule an appointment for me. **Initials** \_\_\_\_\_

I **decline** high risk counseling. **Initials** \_\_\_\_\_

2. Thalassemia (Italian, Greek, Mediterranean or Asian background): MCV < 80

Y                      N

3. Neural Tube Defect (Meningomyelocele, Spina Bifida or Anencephaly)

Y                      N

4. Congenital Heart Disease

Y                      N

5. Down's Syndrome

Y                      N

6. Tay-sachs (Jewish, Cajun, French Canadian)

Y                      N

7. Sickle Cell Disease/ Trait

Y                      N

8. Hemophilia or other blood disorder

Y                      N

9. Muscular Dystrophy

Y                      N

10. Cystic Fibrosis

Y                      N

11. Huntington's Chorea

Y                      N

12. Mental Retardation/ Autism

Y                      N

13. Other inherited genetic or chromosomal disorder

Y                      N

14. Maternal Metabolic Disorder (Type I Diabetes, PKU)

Y                      N

15. Patient or baby's father had a child with birth defects not listed above

Y                      N

16. Recurrent pregnancy loss or greater than 2 stillbirths

Y                      N

My signature certifies that the above information is accurate and true to the best of my knowledge.

**Patient Signature** \_\_\_\_\_ **Date** \_\_\_\_\_

(Parent if minor)

## Sequential Screen: Early information and high detection rates for birth defects

### What is Sequential Screen?

Sequential Screen is a screening test which shows if you are at increased risk of having a baby with Down syndrome, trisomy 18, or an open neural tube defect. It requires a sample of your blood and an ultrasound measurement performed in the first trimester of pregnancy, and a second blood sample taken during the second trimester. It is the only screen that can provide you with two answers - an early, preliminary result in the first trimester, and a final result in the second trimester.

### What is Down syndrome?

Down syndrome is caused by the presence of an extra chromosome #21 and results in both mental and physical abnormalities. Approximately 1 in 800 babies is born with Down syndrome. The risk of having a child with Down syndrome gradually increases with the age of the mother, but can occur at any maternal age.

### What is trisomy 18?

Trisomy 18 is caused by the presence of an extra chromosome #18 and results in serious mental retardation and physical deformities, including major heart defects. Approximately 1 in 6500 babies is born with trisomy 18. Only 1 out of 10 babies affected with trisomy 18 lives past the first year of life. As with Down syndrome, the risk of having an affected child gradually increases with the age of the mother.

### What are open neural tube defects?

The neural tube, which forms very early in pregnancy, eventually develops into the baby's brain and spinal cord. If this tube does not close completely, an opening remains along part of the baby's spine or head. This can lead to paralysis and other physical and /or mental problems. Open neural tube defects occur in about 1 out of every 1,500 live births. The risk of having a child with an open neural tube defect does not increase with the age of the mother.

### How is Sequential Screen performed?

Part 1: Between your 10<sup>th</sup> and 14<sup>th</sup> week of pregnancy, a blood sample and an ultrasound measurement, called a nuchal translucency, will be taken. Sequential Screen will use these pieces of information to provide you with your risk of having a baby with Down syndrome or trisomy 18. If you are found to be at increased risk, your doctor will offer diagnostic testing to confirm the results of your screen.

Part 2: If you are not identified to be at increased risk in Part 1, you will come back between your 15<sup>th</sup> and 19<sup>th</sup> week of pregnancy for a second blood test. The second part of this screen will use the information collected during both Part 1 and Part 2 of the test to provide you with a final screening result. This final result will tell you what your risk is of having a baby with Down syndrome, trisomy 18, or an open neural tube defect.

**Do you wish to be scheduled for Part 1 of the Sequential Screen?**      Yes      No

**Patient Signature** \_\_\_\_\_ **Date** \_\_\_\_\_

## Carrier Screening in Pregnancy for Common Genetic Diseases

Everyone has a risk to have a baby with problems. There are a few common disorders that can occur even without a family history and can be tested for today. You can have one simple blood test before the baby is born to determine if you carry the gene (DNA change) that causes the disorders shown below.

### What is a carrier?

A carrier is a person who has a gene that increases the risk to have children with a genetic disease. People do not know if they are carriers until they have a blood test or an affected child. Some disorders occur only if both parents are carriers and other disorders occur only when the mother is a carrier.

### What is carrier screening?

Carrier screening involves a blood test from one or both parents to determine if they carry a specific gene that increases the risk that their baby is affected. If you turn out to be at risk, prenatal testing such as amniocentesis or chorionic villus sampling (CVS) is available to determine if your unborn baby is affected. All testing is optional and you can choose which disorder(s) to be tested.

Disease	Cystic Fibrosis (CF)	Fragile X Syndrome	Spinal Muscular Atrophy (SMA)
<b>Symptoms of Disease</b>	CF is the most common inherited disease in North America. A chronic disorder that primarily involves the respiratory, digestive, and reproductive systems. Symptoms include pneumonia, diarrhea, poor growth and infertility. Some people are only mildly affected, but individuals with severe disease may die in childhood. With treatments today, people with CF can live into their 20's and 30's.	Fragile X Syndrome is the most common inherited cause of mental retardation. It is a disorder that causes mental retardation, autism, and hyperactivity. It affects primarily boys. Women who are carriers are at risk to have a child with mental retardation.	SMA is the most common cause of inherited infant death. It destroys nerve cells that affect voluntary movement. Infants with SMA have problems breathing, swallowing, controlling their head or neck, and crawling or walking. The most common form of SMA affects infants in the first months of life and can cause death between 2-4 years of age. Less commonly the disease starts later and people can survive into adulthood. SMA does not affect intelligence. There is no cure or treatment.
<b>Inheritance</b>	If both parents are carriers, there is a 1 in 4 (25%) chance to have a child with cystic fibrosis	If a mother is a carrier, there is up to a 50% chance to have a child affected with fragile X syndrome	If both parents are carriers, there is a 1 in 4 (25%) chance to have a child with SMA
<b>General Population Carrier Frequency</b>	1 in 25 Caucasians 1 in 46 Hispanics 1 in 65 African Americans 1 in 90 Asian Americans	1 in 260 females.  Occurs in all ethnic backgrounds	1 in 41  Occurs in all ethnic backgrounds
<b>Do you wish to have carrier testing? (please circle one)</b>	Yes                      No	Yes                      No	Yes                      No

Patient Signature \_\_\_\_\_ Date \_\_\_\_\_