Sequential*Screen*SM

Early information and high detection rates for birth defects

Integrated GENETICS

LabCorp Specialty Testing Group

Sequential*Screen*

When a woman finds she is pregnant, she faces many choices. One important choice is whether to have a maternal serum screening test, such as Sequential*Screen*, to determine if she is at increased risk of having a baby with certain birth defects.

The good news is that most babies are born healthy and that Sequential*Screen* is a non-invasive test. This brochure provides some information to help you decide whether to have this test. If you have any additional questions, please speak with your doctor.

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 10th and 14th week of pregnancy, a blood sample and an ultrasound measurement, called 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 15th and 22nd 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.

What is nuchal translucency?

Nuchal translucency (NT) is a measurement of the fluid filled space at the back of the developing fetus' neck. Extra fluid in this space indicates that the fetus is at a higher risk for certain birth defects.

What do Sequential*Screen* results mean?

It is important to understand that a screening test does not provide a diagnosis; rather it predicts the likelihood of a defect occurring. Sequential*Screen* can only tell you if there is a greater chance of your baby having Down syndrome, trisomy 18, or an open neural tube defect. There are two types of screening test results in each part of the test:

Part 1

- **Final result pending second trimester sample:** This result means that your pregnancy is not in the highest risk group for Down syndrome or trisomy 18. You will be asked to complete the second part of this screen.
- **Screen Positive:** If the results show abnormal measurements, there is an increased chance of you having a baby with Down syndrome or trisomy 18. This is called a "screen positive" result. If your result is screen positive, your doctor will offer diagnostic testing to determine if your baby is affected with one of these birth defects.

Sequential*Screen* Part 1 leads to the detection of approximately 70% of Down syndrome cases and 80% of trisomy 18 cases.*

*According to several large, multi-center studies

Part 2

- **Screen Negative:** If the results show measurements within normal range, the chance of having a baby with Down syndrome, trisomy 18, or an open neural tube defect is low. This is called a "screen negative" result. In rare instances, screening will not detect these birth defects as it cannot detect all high-risk pregnancies.
- **Screen Positive:** If the results show abnormal measurements, there is an increased chance of you having a baby with Down syndrome, trisomy 18, or an open neural tube defect. This is called a "screen positive" result. If your result is screen positive, your doctor will offer diagnostic testing to determine if your baby is affected with one of these birth defects.

Sequential*Screen* Part 2 leads to detection of approximately 90% of Down syndrome cases, 90% of trisomy 18 cases and 80% of open neural tube defects.*

*According to several large, multi-center studies

If I am "screen positive" what additional testing is available?

If a screening test is abnormal, it does not necessarily mean that the baby has one of these birth defects. In fact, most women who have abnormal screening results will have normal, healthy babies. If you "screen positive", your doctor will offer you one of the following procedures:

- **Chorionic villi sampling (CVS)** is a procedure that takes a small amount of tissue from the developing placenta. The tissue is then sent to a laboratory for chromosome analysis. CVS is performed between 10 and 12 weeks of pregnancy. CVS is associated with a small risk of miscarriage.
- **Amniocentesis** is a procedure that withdraws a small amount of fluid that surrounds the fetus. The fluid is then sent to a laboratory to test for chromosome abnormalities and open neural tube defects. An amniocentesis is usually performed around the 16th week of pregnancy. Amniocentesis is also associated with a small risk of miscarriage; however, the risk is lower than that for CVS.

About Integrated Genetics

Integrated Genetics has been a leader in genetic testing and counseling services for over 25 years. This brochure is provided by Integrated Genetics as an educational service for physicians and their patients.

For more information on our genetic testing and counseling services, please visit our web sites:

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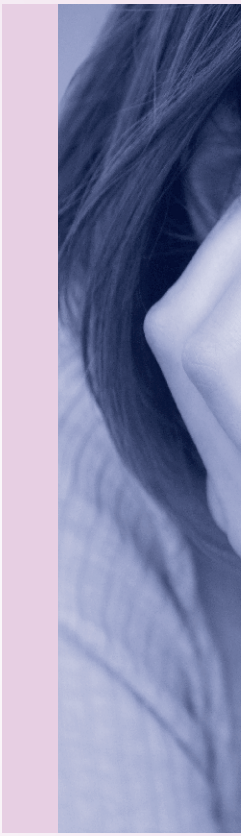

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Integrated Genetics Client Services
(800) 848-4436