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:

- **A more detailed ultrasound examination**, which can often identify an open neural tube defect.
- **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 associated with a small risk of miscarriage.

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:

www.mytestingoptions.com
www.integratedgenetics.com

Integrated GENETICS
LabCorp Specialty Testing Group

Integrated Genetics Client Services
(800) 848-4436

IntegratedScreen℠

Provides the highest detection rate for birth defects

Integrated GENETICS
LabCorp Specialty Testing Group

Integrated*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 Integrated*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 Integrated*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 Integrated*Screen*?

Integrated*Screen* is a blood 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.

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 Integrated*Screen* performed?

A small amount of blood is drawn from your arm and the levels of certain proteins are measured in a laboratory. These proteins are made by the developing baby and the placenta, and are found in every pregnant woman's blood. However, when a fetus is at risk for Down syndrome, trisomy 18, or an open neural tube defect, the amount of these circulating proteins may be abnormal. The results of an ultrasound measurement, called nuchal translucency, are combined with your blood test result to yield the final screening assessment.

Your specific test result is affected by:

- Exactly how far along you are in your pregnancy on the days that the blood samples are taken and the ultrasound is performed.
- Your weight, ethnic background, and age.
- Whether you are an insulin-dependent diabetic or take certain types of medications.
- Whether a close relative has Down syndrome or an open neural tube defect.

Since the first and second trimester results are combined, your risk assessment will not be available until your second trimester blood work is complete, between 15 and 22 weeks of your pregnancy.

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 Integrated*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. Integrated*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:

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 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.

Integrated*Screen* leads to the detection of approximately 92% of Down syndrome cases, 90% of trisomy 18 cases and 80% of open neural tube defects.*

*According to several large, multi-center studies