

If, however, these causes are not the explanation, your doctor may recommend the following:

- **A detailed ultrasound examination**, which can often identify an open neural tube defect.
- **Amniocentesis**, which is the withdrawal of a small amount of amniotic fluid from the uterus. The fluid can be tested for chromosome abnormalities, and also for open neural tube defects. Amniocentesis is associated with a small risk of miscarriage.

Am I required to have a maternal serum screening test?

Your doctor may recommend that you consider maternal serum screening, but you are not required to have it. The choice is yours.

About Genzyme Genetics

Genzyme Genetics has been a world leader in genetic testing and counseling services for more than 20 years.

This brochure is provided by Genzyme Genetics as an educational service for physicians and their patients.

For more information on our genetic testing and counseling services, please visit our web site at www.genzyme genetics.com.

Integrated Test Technology under license from Intema Ltd, UK

SerumIntegratedScreen™

- SerumIntegratedScreen provides information about the risks for having a baby with Down syndrome, trisomy 18 or an open neural tube defect.
- The test requires two blood samples from you, one taken in the first trimester and the other in the second trimester of your pregnancy.
- 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 14 and 22 weeks of your pregnancy.

The SerumIntegratedScreen leads to the detection of approximately:

- 87% of Down syndrome cases
- 90% of trisomy 18 cases
- 80% of open neural tube defects



The Facts.

When a woman discovers she is pregnant, she faces many choices. One important choice is whether to have a maternal serum screening test, such as *SerumIntegratedScreen*[™], to determine if she is at increased risk of having a child with certain birth defects.

The good news is that most babies are born healthy and that *SerumIntegratedScreen* is non-invasive. This brochure has some information to help you decide whether to have this test. If you have any questions about this test, please talk with your doctor.

What is *SerumIntegratedScreen*?

SerumIntegratedScreen 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 two samples of your blood, one taken in the first trimester and another in the second trimester.

What is Down syndrome?

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

What is trisomy 18?

Trisomy 18 is caused by an extra chromosome #18 and results in serious mental retardation and physical deformities, including major heart defects. 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. Open neural tube defects occur in 1 or 2 out of every 1,000 births. The risk of having an open neural tube defect does not increase with the age of the mother.

How is *SerumIntegratedScreen* performed?

A small amount of blood is drawn from your arm and certain proteins are measured. These proteins are made by your body, the fetus (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 proteins may be abnormal.

Each patient's specific result is affected by:

- Exactly how far along in your pregnancy you are on the days the blood samples are taken.
- 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.

What do *SerumIntegratedScreen* results mean?

It is important to note that a screening test **does not** provide a diagnosis; rather it predicts the likelihood of a problem to occur. *SerumIntegratedScreen* 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 results:

Screen Negative

If the results show normal measurements, the chance of your 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, these birth defects will not be detected through screening, 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.

What additional testing will be needed if the result is abnormal?

If a screening test is abnormal, it does not necessarily mean that your baby has one of these birth defects. In fact, most women who have abnormal screening results will have normal babies. An abnormal screening result is often caused by incorrect pregnancy dating or a previously unidentified multiple pregnancy (twins, triplets, etc.). An ultrasound can determine if one of these causes is present. If so, the screening result may be different.