



Prenatal screening *and you*



*Information about the
Maternal Serum Screen 4*



Quest
Diagnostics®

What is prenatal screening?

The health of her unborn child is often a pregnant woman's greatest concern. Fortunately, there is prenatal screening—simple blood tests that can help identify the small number of women whose babies are at greater risk for certain birth defects.

What do prenatal screening tests measure?

Maternal serum prenatal screening tests measure substances in the mother's blood such as alpha-fetoprotein (AFP), human chorionic gonadotropin (hCG), estriol, and inhibin A.

Maternal serum prenatal screening tests do not screen for all birth defects. The tests screen primarily for open neural tube defects, Down syndrome, and trisomy 18. Since the test results only indicate which babies *may* be at increased risk for these birth defects, additional testing is needed to know whether or not your baby really has a birth defect. On the other hand, a negative result does not guarantee the birth of a healthy baby.

This brochure will help you understand exactly what information can be obtained from the prenatal screening test and what the next steps might be if your results indicate that your baby is at increased risk.

What is a neural tube defect?

A neural tube defect is a birth defect that affects the spinal column or brain of the baby. The two main types of neural tube defects are anencephaly and spina bifida. In anencephaly, the brain and skull fail to develop normally. Babies with this birth defect usually die at birth or shortly afterward. Spina bifida results in a spinal column defect that may range from relatively mild to very severe. Mild defects may be corrected with surgery, resulting in minimal symptoms. More severe defects may cause difficulty in walking or an inability to walk, absence of bowel and bladder control, and frequent urinary tract infections. Unfortunately, test results may not predict the severity of the problem. Approximately 1 or 2 out of 1000 babies are born with neural tube defects.

What is Down syndrome?

Down syndrome (trisomy 21) is a condition in which the baby has extra genetic material (that is, one extra copy of chromosome number 21). Babies with Down syndrome have a distinctive physical appearance, mental retardation, poor muscle tone, and a higher frequency of heart defects. Approximately 1 out of 800 babies is born with Down syndrome. Although women who

are age 35 or older have a greater risk of having a baby with Down syndrome, most Down syndrome babies are born to women under the age of 35. This is because the majority of pregnant women are younger than age 35.

What is trisomy 18?

Trisomy 18 is another condition in which the baby has extra genetic material (one extra copy of chromosome number 18). Like Down syndrome, the risk is greater in women age 35 or older. The majority of pregnancies with an affected fetus end in miscarriage. Babies born with trisomy 18 have low-set ears, clenched fists, congenital heart disease, and severe mental retardation. Ninety percent of these infants die within the first year of life. Trisomy 18 occurs in only 1 out of 8000 births.

Who should have prenatal screening?

This is a decision that you and your doctor must make. Generally, all patients receiving prenatal care between 15 and 20 weeks of gestation are offered prenatal screening. If you would like to have more information about the health of your unborn baby,

you may wish to have prenatal screening performed. Women having an amniocentesis (see “What additional tests may be recommended?”) do not need prenatal screening tests. Women who have already had a chorionic villi sampling (CVS), however, should still consider prenatal screening since CVS will not detect neural tube defects.

If there is a history of neural tube defects, Down syndrome, or other abnormalities in your family or in the father’s family, prenatal screening may not be the best testing option. It is important to share your history with your physician or a genetic counselor and to discuss optimal testing options.

Remember that a negative screen does not guarantee a healthy baby and that a positive screen only indicates a need for additional testing to determine whether or not your baby has one of the conditions described above.

How is prenatal screening done?

The first step is to obtain information about your medical history and your pregnancy. This information is used to interpret the test results and includes any history of diabetes, your race, age, weight, and the number of weeks you have been pregnant

(gestational age). Gestational age is based on an ultrasound examination or on the first day (date) of your last menstrual period. If any of this information is incorrect, then the interpretation of your test results may be wrong.

The next step is for you to give a sample of your blood, which will be sent to the laboratory along with the medical information described above. The laboratory will test your blood for AFP, hCG, estriol, and inhibin A. These results will be combined with your age and the other information you provided to determine your baby's risk for an abnormality.

What does a negative screen mean?

A negative screen means that your baby probably does not have a neural tube defect, Down syndrome, or trisomy 18. Further testing is not required. A negative screen, however, does not guarantee that your baby will not have some form of birth defect.

What does a positive screen mean?

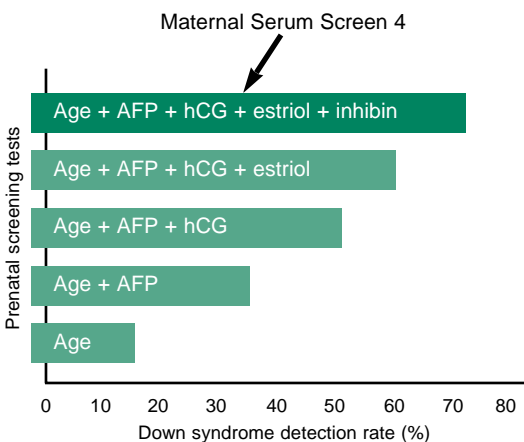
A positive screen means that your baby has an increased risk of having an open neural tube defect, Down syndrome, trisomy 18,

or some other problem. It does not mean that your baby definitely has a problem. In fact, the baby is fine in the majority of cases. For example, a positive screen is often simply due to an incorrect gestational age or twins. Your doctor or a genetic counselor can help you understand the risk and explain the additional tests that may be recommended.

How accurate is this screening test?

The Maternal Serum Screen 4 detects the same number of neural tube defects and trisomy 18 cases as other currently available maternal serum prenatal screens. Unlike the other blood screens, this screen includes a test for inhibin A, a substance produced by the ovaries and the placenta. When inhibin A is used with AFP, hCG, estriol, and the mother's age, approximately 10% to 15% more babies with Down syndrome can be detected before birth. Thus, the Maternal Serum Screen 4 is a more accurate screen. Remember that not even the Maternal Serum Screen 4 can detect all babies with Down syndrome before they are born.

The chart below shows that the Maternal Serum Screen 4 can detect about 75% of unborn babies with Down syndrome. The chart also shows that this detection rate is greater than that of other prenatal screens.



What additional tests may be recommended?

If your screen is positive, the first additional test that will probably be recommended is an ultrasound. Ultrasound utilizes sound waves to take an echo picture of your unborn baby. An ultrasound may help to more accurately determine the gestational age. It may also be able to visualize anencephaly (abnormal brain development), other types of birth defects, or the presence of more

than one baby. If the new information gained from the ultrasound leads to changing your positive screen to a negative screen, further testing will probably not be required.

If more testing is needed, AFP and chromosome analysis of amniotic fluid will probably be suggested. Amniotic fluid is the fluid that surrounds and protects your baby. A sample of amniotic fluid is collected during a procedure called amniocentesis. During amniocentesis, a needle is inserted into your anesthetized abdomen, and some fluid is withdrawn from the uterus. The level of AFP in the fluid will confirm or exclude the presence of an open neural tube defect. Chromosome analysis will determine the presence of additional genetic material, if any, as well as some other genetic abnormalities. Since there is a risk of miscarriage associated with amniocentesis, you should discuss both the risks and the benefits of the procedure with your doctor or genetic counselor.

What if the additional tests are negative?

If the additional tests are negative, your baby is likely to be healthy, but your doctor will follow your pregnancy very closely just to

be as safe as possible. Negative test results do not guarantee that your baby will not have some form of birth defect.

What if the additional tests are positive?

If an additional test is positive, genetic counseling will help you learn more about your baby's condition and will help you make decisions in the best interest of you, your family, and your baby.

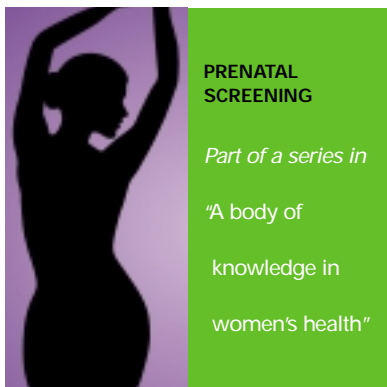
Summary

A negative prenatal screen means your baby probably does not have a neural tube defect, Down syndrome, or trisomy 18; however, it does not guarantee the health of your baby.

A positive prenatal screen means additional tests are needed to determine whether or not your baby has a birth defect.

If a birth defect is found, your doctor or a genetic counselor will help you make decisions in the best interest of you, your family, and your baby.

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