

What do the results mean?

Normal Blood test results do not guarantee a normal baby, nor do abnormal results definitely mean that your baby has Down syndrome, trisomy 18, or a neural tube defect.

Most women who have these screening tests will have normal results. Normal test results indicate that the risk of producing a child with open neural tube defects, Down syndrome, or trisomy 18 is minimal.

Abnormal results do not necessarily mean you will have an abnormal baby. It means your risk of having a baby with one of these birth defects is higher than average. Most women who are at increased risk deliver healthy babies. After receiving your results, you will be offered further information and testing. The information provided by screening tests is beneficial for some families; other families do not feel this information is helpful to have prior to delivery. This information will be useful as you and your family make decisions about the pregnancy and plan its management with your doctor.

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If you are a patient, please contact your obstetrician or call the **MUSC Prenatal Wellness Center** at

1-800-424-MUSC

or **792-1212.**

If you are a physician, please call through MEDULINE at

1-800-922-5250 or **792-2200.**

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**Prenatal
Wellness Center**

**Second Trimester
Maternal Screening**

A guide for expectant women

*Information provided by the MUSC Prenatal Wellness
Center and Laboratory Medicine*

The risk of producing a child with Down syndrome or an open neural tube defect concerns many parents. This risk can be assessed by screening the level of alpha-fetoprotein (AFP) in the mother's bloodstream.

AFP is a substance produced by the unborn baby. It circulates in the baby's bloodstream, and small amounts of it enter the amniotic fluid. Some AFP crosses the placenta and passes into the mother's bloodstream. The amount of AFP in the mother's blood can be measured by a blood sample taken between the 15th and 21st week of pregnancy.

Open neural tube defects

AFP screening can detect pregnancies with open neural tube defects because these defects allow greater than normal amounts of AFP to cross into the mother's bloodstream. There are two types of open neural tube defects – an absence of the skull, known as anencephaly, and an open spine, known as spina bifida. These defects generally occur without warning and without an affected family member.

Most women who have AFP testing will have normal results, meaning that their risk of having a baby with anencephaly or spina bifida is very low. The screening detects 80 percent of pregnancies with open neural tube defects.

Abnormal results

If the AFP level is higher than expected, the possibility of twins or incorrect dating (being further along in pregnancy than expected) must first be ruled out by an ultrasound. A repeat AFP blood test is usually performed if neither twins nor incorrect dating is found by the ultrasound.

If a woman has elevated AFP levels on repeat testing or the first AFP level is very high, she will be offered further information and testing to evaluate the baby for abnormalities. **IT'S IMPORTANT TO NOTE, MOST WOMEN WHO HAVE ELEVATED AFP LEVELS WILL HAVE HEALTHY BABIES.** Elevations usually can be explained by the differences in placentas and the amount of AFP that is allowed to cross into the mother's bloodstream.

Down syndrome

The AFP level and other blood levels also are used to detect pregnancies at increased risk for Down syndrome, a birth defect resulting in mental retardation. Down syndrome, like open neural tube defects, generally occurs without warning or without an affected family member. The risk of having a baby with Down syndrome increases as women age. While it is routine to offer prenatal testing, such as amniocentesis, to all women 35 years of age and older, Down syndrome pregnancies in women under 35 generally remain undetected until delivery unless a blood screening has been done.

To assess the risk for Down syndrome, AFP is measured from the same blood sample drawn between the 15th and 21st week of pregnancy. The levels of human chorionic gonadotropin (hCG) and estriol (uE3), which are placental hormones, and inhibin A, a placental protein, also are measured in this blood sample. Elevated levels of hCG and inhibin A and low AFP and estriol levels are associated with an increased risk of Down syndrome. The results of these tests are combined with the mother's age and reported as a risk figure for Down syndrome. If this risk figure is the same or greater than the risk for a 35 year old, the patient will be offered further information and more definitive testing. The results of this screening will detect 80-85 percent of pregnancies with Down syndrome.

Trisomy 18

Low AFP, hCG, and estriol levels are used to detect pregnancies at increased risk for trisomy 18, a birth defect that causes mental retardation, and in most cases, death. This screening will detect 60 percent of pregnancies with trisomy 18.

If you have questions about screening or diagnostic tests in pregnancy, please call the MUSC Prenatal Wellness Center at 792-1212.