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MATERNAL SERUM ALPHA FETOPROTEIN

A USEFUL SCREENING TEST FOR PRENATAL DETECTION OF BIRTH DEFECTS

Approximately 2 percent of babies will have a major birth defect. Some birth defects can be diagnosed in pregnancy. The maternal serum alpha fetoprotein (MS-AFP) test is a blood test which can give your doctor additional information about your pregnancy and can identify women who may be carrying a baby with certain types of birth defects.

WHAT IS AFP?

Alpha-fetoprotein (AFP) is a protein made by the baby as it grows in the uterus (womb). During pregnancy, AFP is present in the amniotic fluid that surrounds the baby. It is also found in small amounts in the mother's blood. In some women there may be more or less AFP than normally found. This DOES NOT always indicate a problem with the baby, but does warrant increased or decreased amounts of MS-AFP.

Some reasons for an increased amount of MS-AFP include a pregnancy further along than your dates suggests, twins, or the presence of a neural tube defect in the baby. Among the reasons for a decreased amount of MS-AFP included a pregnancy less advanced than expected or the presence of Down syndrome in the pregnancy. Other less common conditions may cause increased or decreased amounts of MS-AFP.

WHAT ARE NEURAL TUBE DEFECTS?

Neural Tube Defects (NTD) are birth defects in which the brain or part of the spinal cord does not form properly. The two major types of neural tube defects are anencephaly and spina bifida. Ancencephaly occurs when the brain and head do not develop normally. Babies with anencephaly are usually stillborn or die within a few days. Spina bifida (open spine) involves a defect in the closure of the spine. This defect may be quite severe and result in long term problems such as paralysis of the legs, lack of bowel and bladder control, hydrocephalus (water on the brain) and mental retardation. On the other hand, the defect can be mild and surgically correctable with little or no resulting handicaps. The incidence of open neural tube defects is approximately 1-2 per 1,000 births.

WHAT IS DOWN SYNDROME?

Down syndrome is a disorder of the chromosomes. Children with this condition are mentally retarded and have a variety of other birth defects that may include abnormalities of the heart and digestive tract.

WHO SHOULD HAVE THE MS-AFP TEST?

Many physicians believe this screening is appropriate for all pregnant women. The MS-AFP test is ideally performed between the sixteenth and eighteenth week after the first day of the last menstrual cycle. At this time, the test is most accurate. A small amount of blood is taken from the arm much like any other blood tests that you have had. The test causes no adverse health risks to you or the baby.

People with a positive family history of Down syndrome or neural tube defect should ask their doctor if the MS-AFP test is appropriate for them.

WHAT IF THE TEST RESULT IS NORMAL?

Normal results mean no further investigation is necessary. However, a normal result cannot guarantee a normal baby. Most birth defects are not detectable by this screening test. Estimates are that the MS-AFP test will detect approximately 85 percent of the neural tube defects, and up to 20 percent of Down syndrome for women under 35 years of age. For women over age 35, amniocentesis is advised because the risk of having baby with Down syndrome increases with advanced maternal age.

WHAT IF THE TEST IS ABNORMAL?

If the test shows either elevated or decreased amounts of AFP, it does not mean that the baby has a neural tube defect of Down syndrome. It only means that further tests are indicated. When the result of the test is too low, an ultrasound is recommended. Very often the ultrasound proves that the pregnancy is not far along as previously suspected. When the test result is too high, a repeat test may be requested. The test should be repeated within 7 to 10 days. If the second test also shows increased amounts of AFP, an ultrasound examination should be done. In many cases, the pregnancy is farther along than originally estimated or there is a twin pregnancy. If the ultrasound examination does not explain the high or low AFP level, you should discuss this with your doctor who may refer you for genetic counseling. Generally, amniocentesis will be offered at this time. Amniocentesis involves the withdrawal of a small sample of fluid that surrounds the baby. Elevated levels of AFP can be detected in the amniotic fluid and with other tests will accurately diagnose mos t cases of neural tube defects. A chromosome study is also preformed o the amniotic fluid cells which can accurately diagnose Down syndrome and other forms of chromosome abnormalities. Most women who undergo amniocentesis will receive normal results. If a birth defect is detected, genetic counseling is available to discuss the disorder and the various options available.

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Patient Initials Date