MATERNAL SERUM ALPHA FETOPROTEIN (AFP4) 

A USEFUL SCREENING TEST FOR PRENATAL DETECTION OF BIRTH DEFECTS

Most babies are healthy when they are born, however, 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-AFP4) 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. It is a screening test only. AFP4 results are not diagnostic.

WHAT IS AFP4?
Alpha-fetoprotein (AFP4) is a protein made by the baby as it grows in the uterus (womb). During pregnancy, AFP4 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 AFP4 than normally found. This DOES NOT always indicate a problem with the baby, but does warrant increased or decreased amounts of MS-AFP4. Some reasons for an increased amount of MS-AFP4 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-AFP4 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-AFP4.

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. Anencephaly 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 have varying degrees of mental retardation and have a variety of other birth defects that may include abnormalities of the heart and digestive tract.
WHO SHOULD HAVE THE MS-AFP4 TEST?
Many physicians believe this screening is appropriate for all pregnant women. The MS-AFP4 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-AFP4 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. Many birth defects are not detectable by this screening test. Estimates are that the MS-AFP4 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.