Amniotic Fluid Alpha-fetoprotein (AF-AFP) and Acetylcholinesterase (AChE)

Clinical Features:
In addition to fetal chromosome analysis and genetic testing, amniocentesis is also used to detect open neural tube defects including spina bifida and anencephaly in the fetus. Alpha-fetoprotein (AFP) is a protein produced by the fetal liver. The amount of AFP can be measured in amniotic fluid. If there is a higher level of AFP than expected in the amniotic fluid, an opening in the spinal column (neural tube defect or NTD) may be present. AF-AFP may also be elevated in fetuses with an omphalocele (ventral wall defect) or congenital Finnish nephrosis. Further testing may be indicated to determine the nature of the possible birth defect in the fetus.

Test Methods:
Fetal abnormalities suspected by ultrasound, such as spina bifida, ventral wall defects, anencephaly or abnormal maternal serum screening test result. AF-AFP is performed on all amniocentesis samples drawn between 15-24 weeks gestation. ACHE will automatically be done if the AF-AFP level is greater than or equal to 2.0 MoM. AF-AFP assays are performed using the Access AFP assay from Beckman Coulter. It is a quantitative test to determine the amount of AFP present in the submitted specimen. The assay is a two-site immunoenzymatic ("sandwich") assay. AFP is detected through a chemiluminescent reaction, and the amount of analyte in the sample is determined from a stored, multi-point calibration curve. Acetylcholinesterase is measured through electrophoresis.

Test Sensitivity:
Maternal red blood cell contamination of the amniotic fluid could lead to an incorrect result (false positive).

In cases with elevated AF-AFP, the risk for an open NTD or other fetal abnormality depends on the degree of elevation in the AF-AFP MoM (multiple of the median), the results of amniotic fluid acetylcholinesterase testing and other significant risk factors.