Screening Options for Genetic Tests

Option #1 Sequential Screening

Sequential Screen is a screening test which shows if you are at increased risk of having a baby with Down syndrome, Trisomy 18, or an open neural tube defect. It is a noninvasive test performed in two steps. This test is done at Akron Children's Hospital. You would receive an early preliminary result in the first trimester, and a final result in the second trimester. Sequential Screen leads to detection of approximately 90% of Down syndrome and Trisomy 18 cases and 80% of open neural tube defects. If the screening test is abnormal, it does not necessarily mean that the baby has one of these birth defects. If you have a "positive" screen you will be offered further testing. This typically includes a very targeted ultrasound and the possibility of Chorionic villi sampling (CVS)* or Amniocentesis*. A "negative" result does not guarantee the birth of a healthy baby.

How it is done:

Between 11-13 weeks of your pregnancy, a blood sample and ultrasound measurement, called nuchal translucency*, will be taken. If you are not identified to be at increased risk during the first part of the test, you will come back in at 16 weeks for a second blood test. You will receive preliminary results after the first part of the test and final results after part two.

Option#2 Quad Screen

The quad screen is an optional screening test offered around 16 weeks of pregnancy. The test screens primarily for open neural tube defects, Down syndrome, and Trisomy 18. The Quad screen offers about 75-80% detection rate. If the screening test is abnormal, it does not necessarily mean that the baby has one of these birth defects. If you have a "positive" screen you will be offered further testing. This typically includes a very targeted ultrasound and the possibility of Amniocentesis. A "negative" result does not guarantee the birth of a healthy baby.

How it is done:

A blood sample is obtained between the 16-20th week of pregnancy. A calculation is made based on certain medical information and this blood work.

Option #3 Alphafetoprotein (AFP) Blood Test

The AFP blood test screens for open neural tube defects only. The two main types of defects are Spina Bifida and anencephaly. Babies with anencephaly usually will die at birth or shortly afterward. Spina Bifida results in a spinal column defect and may range from mild to severe. Mild defects can be corrected with surgery.

How it is done:

A blood sample is obtained between the 16-20th week of pregnancy. A calculation is made based on certain medical information and this blood work.

Option #4 Decline all Optional screening tests.

Some families choose to decline all genetic screening. Choosing or declining screening is a very personal matter to which there is no right or wrong answer. Your decision may depend on religious beliefs, level of anxiety, or course of action that would be taken with abnormal screening results.

*Nuchal Translucency (NT) is a measurement of the fluid filled space at the back of the developing fetus' neck.

*Chorionic Villi Sampling (CVS) is a procedure that takes a small amount of tissue from the developing placenta. CVS is associated with a small risk of miscarriage.

*Amniocentesis is a procedure that withdraws a small amount of fluid that surround the fetus. This procedure is associated with a small risk of miscarriage; however, the risk is lower than that for CVS.