EMORY UNIVERSITY SCHOOL OF MEDICINE

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Abnormal First Trimester Screen Results

You have been informed that your first trimester screen indicates that your risk for having a baby with **Down syndrome (trisomy 21 – an extra copy of chromosome #21)** or **trisomy 18 or trisomy 13 (an extra copy of chromosome #18 or #13 respectively)** is increased.

This result does NOT mean that your fetus actually has one of these conditions. Screening tests do not diagnose a condition, but provide a risk estimate (probability) specific to the pregnancy.

What is the first trimester screen?
The first trimester screen is a test that combines a maternal blood sample with an ultrasound measurement at approximately 11-14 weeks gestation to identify pregnancies with an increased risk of having certain fetal chromosome abnormalities. The screen will occasionally also allow for the detection of other birth defects.

What does this screen measure?
The maternal blood sample measures two proteins that are made during pregnancy, **a form of hCG** and **PAPP-A**. The ultrasound measures the amount of fluid at the back of a baby's neck called the **nuchal translucency (NT)**. The overall risk is then calculated by a combination of the mother's age-related risk, the hormone levels in the maternal blood sample and the NT measurement.

What is the detection rate of the first trimester screen?
The detection rate is defined as the percentage of truly affected fetuses that are identified as "increased risk" by the test. The first trimester screen can detect over 90% of babies with Down syndrome or trisomy 18. The screen can also detect approximately 40% of babies with congenital heart defects, and many other anomalies, including additional chromosome abnormalities, other genetic conditions, or pregnancy complications.

A normal pregnancy is still the most likely outcome, even after an abnormal first trimester screening test.
It is important to remember that the first trimester blood and ultrasound test is a screening test, and that most of the time pregnancies with an abnormal first trimester screen will result in the delivery of a normal baby. In addition, first trimester screening will not detect all babies with these defects.

Should I have the first trimester screen repeated?
No, first trimester screening should NOT be repeated. It is also not recommended that you have second trimester maternal serum screening (the triple screen or quad screen) for Down syndrome or trisomy 18 as this testing will NOT improve the detection rate, and this additional test result can not be combined with the test result you have already received.

What are my options for follow-up testing?
There are two options for prenatal diagnostic testing depending on how far along in the pregnancy you are. **Chorionic villus sampling (CVS)** is available between 10 and 12 ½ weeks gestational age and **amniocentesis** is available after 15 weeks gestational age. These tests are able to identify a fetal chromosome abnormality with over 99% accuracy. You may also be offered a targeted fetal ultrasound and fetal echocardiogram at approximately 20 weeks gestation to screen for other birth defects. If you do not choose to proceed with diagnostic testing, or if you have a CVS procedure, it is still recommended that you have AFP-only maternal blood screening at approximately 15-20 weeks gestation to screen for open neural tube defects.