If your screening test indicates an increased chance, your doctor and/or genetic counselor will discuss diagnostic testing options with you. These options may include chorionic villus sampling (CVS), which is performed between 11 and 13 weeks and has an associated 1% chance for miscarriage, and amniocentesis, which is performed between 15 and 20 weeks and has an associated 0.5% or less chance for miscarriage. Both of these tests are able to diagnose 99.9% of all chromosome abnormalities, including Down syndrome, trisomy 13, and trisomy 18.

What if my test indicates an increased chance?

If your screening test indicates an increased chance, your doctor and/or genetic counselor will discuss diagnostic testing options with you. These options may include chorionic villus sampling (CVS), which is performed between 11 and 13 weeks and has an associated 1% chance for miscarriage, and amniocentesis, which is performed between 15 and 20 weeks and has an associated 0.5% or less chance for miscarriage. Both of these tests are able to diagnose 99.9% of all chromosome abnormalities, including Down syndrome, trisomy 13, and trisomy 18.

What if my test is reassuring?

This means that the chance for these chromosome abnormalities is low enough that diagnostic testing is not indicated. It is important to remember that screening tests cannot rule out the possibility that your baby has Down syndrome, trisomy 13, trisomy 18, other chromosome abnormalities, or open NTDs. Regardless of your results, or even if you decide not to have screening for chromosome abnormalities, you may choose to have diagnostic testing, by means of amniocentesis or CVS.

### Comparison of screening options:

<table>
<thead>
<tr>
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<th>First Trimester Screen</th>
<th>Quadruple Screen</th>
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<td>Trisomy 18: 65%</td>
<td>Trisomy 13: 90%</td>
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<tr>
<td></td>
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<td>Trisomy 18: 90%</td>
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<tr>
<td></td>
<td>NTDs: 85-90%*</td>
<td>NTDs: 85-90%</td>
<td>NTDs: 85-90%*</td>
</tr>
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</table>

Note: detection rates are slightly decreased in twin pregnancies.

* when AFP is measured separately between 15 and 20 weeks
What are open neural tube defects?
Screening for chromosome abnormalities often also includes screening for open neural tube defects (NTDs). The neural tube develops very early in pregnancy, forming the spinal cord and the brain. In some cases, this tube does not close properly, leaving an opening in the spine or head. Spina bifida, an opening in the spine, can cause features such as paralysis and problems of brain development. In anencephaly, where the brain and skull don’t develop, babies do not survive after birth.

Approximately 1 in 1,000 babies will have an open NTD, most of which aren’t related to chromosome abnormalities. The chance for open NTDs does not increase with maternal age. Screening for open NTDs generally includes second trimester ultrasound and measurement of a protein, called alpha-fetoprotein (AFP) from your blood. AFP measurement may also detect openings in a fetus’s abdominal wall.

What options are available to screen for chromosome abnormalities?

- **First trimester screen:** First trimester screening uses both blood work and ultrasound measurement of fetal nuchal translucency. Blood can be drawn between 9 weeks 0 days and 13 weeks 6 days gestation, and the ultrasound can be performed between 11 weeks 2 days and 13 weeks 6 days gestation. This test detects approximately 85% of fetuses with Down syndrome and approximately 90% of fetuses with trisomy 13 or trisomy 18. As this test does not screen for open NTDs, a second trimester blood test for AFP and/or ultrasound should be considered.

- **Quadruple screen:** Maternal serum quadruple screen, or second trimester screen, is a blood test that is performed between 15 and 20 weeks gestation. This test estimates the chance that a pregnancy will be affected with Down syndrome, trisomy 18, and open neural tube defects (NTDs). This test detects approximately 75-80% of fetuses with Down syndrome, 65% of fetuses with trisomy 18, and 85-90% of fetuses with open NTDs.

- **Sequential Screen:** The sequential screen incorporates both first and second trimester screens, allowing an increase in detection of Down syndrome and trisomy 18. This test begins with first trimester screen, as described above. Once you receive your results, you can decide whether or not you want to go on to have second trimester screening as well. If you do, you will receive a second set of results after your second blood draw. These final results will incorporate the information from both blood draws and the nuchal translucency measurement. This test detects greater than 92% of fetuses with Down syndrome, greater than 92% of fetuses with trisomy 18, 90% of fetuses with trisomy 13, and 85-90% of fetuses with open NTDs.

What are Down syndrome, trisomy 13, and trisomy 18?
These are the three most common numerical chromosome abnormalities. Although the chance for these conditions is increased for women age 35 and older, most children with these conditions are born to women under the age of 35. This is because more babies overall are born to younger women.

- **Down syndrome:** Down syndrome (also called trisomy 21) occurs when a fetus has an extra copy of chromosome #21. This causes mental retardation, changes in physical appearance, and, sometimes, birth defects.

- **Trisomy 13 and trisomy 18:** Trisomy 13 and 18 occur when a fetus has an extra copy of chromosome #13 or chromosome #18. These conditions are much more serious than Down syndrome and cause profound mental retardation and multiple birth defects. Babies with these conditions are often miscarried before the end of the pregnancy or survive for a very short period after birth.

What are chromosomes?
All of the cells in the body contain chromosomes. Typically, each person has a total of 46 chromosomes in each cell. The chromosomes come in pairs (23 pairs); one chromosome from each pair comes from the mother, and the other comes from the father. Every chromosome carries genes, the instructions for our growth and development. Problems arise when a fetus has extra or missing chromosomal material.

What is nuchal translucency?
The term nuchal translucency refers to an accumulation of fluid behind the neck of a fetus. While this fluid is always present in the first trimester, the accumulation tends to be larger in fetuses with chromosome abnormalities and certain other abnormalities, including heart defects. Nuchal translucency can be measured by ultrasound in the first trimester. Its size can be used to help determine the chance for chromosome abnormalities.

What is a screening test?
Screening tests estimate the chance for your pregnancy to have certain chromosome abnormalities, using both the age-related chance in the general population and factors that are specific to your pregnancy. Screening tests are used to identify pregnancies that have an increased chance to be affected with a chromosome abnormality but cannot definitively diagnose a chromosome abnormality in a fetus. Screening does not pose any risk to harm the pregnancy.

Screening
For more information or to schedule an appointment please call 617-667-7110.