



## California Prenatal Screening Program

The California Prenatal Screening Program is a set of screening tests offered to pregnant women to screen for certain genetic defects. A screening test is a method of determining who is at risk for a condition that may warrant further diagnostic testing. This screening test is a noninvasive test and carries no risk to you or the baby. A diagnostic test can tell if the fetus actually has a specific birth defect. The California Prenatal Screening Program tests for Down syndrome, Trisomy 18, anencephaly, open spina bifida, abdominal wall defects and Smith-Lemli-Opitz Syndrome (SLOS). Three different types of Prenatal Screening Tests are available and are detailed in the Prenatal Patient Booklet provided by your physician. A glossary of terms is located in the back of this book. This screening is optional and not all couples choose to have this screening.

### What are the three tests that are available?

- **Quad Marker Screening** - The Quad Marker Screening test is a screening test for pregnant women during the second trimester (between 15 and 20 weeks) of pregnancy who choose to do only a second trimester blood test. The detection rates for this test are 80 out of 100 Down syndrome and 67 out of 100 for Trisomy 18.
- **Serum Integrated Screening** – This is a combination of a first and a second trimester blood test that detect 85 out of 100 for Down syndrome and 79 out of 100 for Trisomy 18. This series of two blood tests does not include an ultrasound (NT).
- **Full Integrated Screening** - This includes the Serum Integrated Screening with nuchal translucency (NT) ultrasound and detects 90 out of 100 for Down syndrome and 81 of 100 for Trisomy 18.
- All three screening tests detect 97 out of 100 for anencephaly, 80 out of 100 for open spina bifida, 85 out of 100 for abdominal wall defects and 60 out of 100 for SLOS.

### What is the Nuchal Translucency (NT)?

An ultrasound is performed between 11 weeks 2 days and 14 weeks at a Prenatal Diagnosis Center to measure the clear (“translucent”) space in the tissue at the back of the developing baby’s neck. This measurement assesses the baby’s risk for Down syndrome (DS) and other chromosomal abnormalities. Babies with abnormalities tend to have more fluid accumulated at the back of their necks during the first trimester, causing this clear space to be larger. Based on statistical probability, the measurements are used along with the maternal age to calculate the baby’s chances of having a chromosomal abnormality. Along with the ultrasound, an accompanying blood test increases the accuracy of the risk assessment.

### What is a Preliminary Risk Assessment?

Results are delivered as a ratio to express your baby’s chances of having a chromosomal problem (based on your age, the baby’s age, and the nuchal fold measurement). For example, a patient who is 35 years of age at delivery has an average risk for a baby with

a chromosomal abnormality of 1 in 178. This risk gets higher as you get older. If your baby's nuchal fold measurement is found to be average for its gestation, the baby's risk stays the same: 1 in 178. A thicker than average NT increases the risk for an abnormality. If the nuchal fold is thinner than average, then the baby's risk of a chromosomal abnormality decreases.

This test does not directly test for chromosomal problems. It only gives a better indication of the baby's statistical risk of having a problem. A normal result (sometimes called "screen negative") is not a guarantee that your baby is normal, but it suggests that a chromosomal problem is unlikely. Nor does an abnormal result (sometimes called "screen positive") mean that the baby has a chromosomal problem—just that it has an increased risk of one. (Even so, most "screen positive" babies still end up being normal.)

Based on the screening risk, you will decide if you want to have diagnostic testing done. Individual parents-to-be have different feelings on what is an "acceptable" risk for them. The California State test considers a risk of 1 in 200 for Down syndrome as a "negative" test. Obstetrix Medical Group offers genetic counseling and the option of a diagnostic test if the risk is greater than 1 in 1000. It is your choice to have further testing or not and it is up to you to decide what your comfort level is for further testing. A detailed ultrasound can provide additional information, but definitive tests that can diagnose a chromosomal defect are chorionic villus sampling (CVS) or amniocentesis.

#### **What does it mean that the Full Integrated Screening test is "90% accurate?"**

You may have read that the results of this test are 90% accurate in detecting your risk of having a baby with Down syndrome. That means that if your baby has Down syndrome, there's a 90% chance that the test will pick that up and give a "screen positive" result that indicates further testing is recommended. It also means there is a 10% chance that the test will miss the Down syndrome and give a "screen negative" result and diagnostic testing will not be recommended. This does NOT mean that a "screen positive" baby has a 90% chance of having DS. It just means that 90% of babies who have DS will have screening results that are suspicious enough to recommend diagnostic testing. And 10% of babies who have DS will be shown to be at normal risk—that is, the results will be falsely reassuring. This screening test also has up to a 5% false positive rate. (A "false positive" result is when a test suggests there may be a problem when, in fact, there is no problem.) In this case, a 5% false positive rate means that 5% of all the babies with normal chromosomes who are tested will be "screen positive" meaning that the test will show them to be at an increased risk even though they are normal. Considering this "false positive" result, their mothers may opt for invasive diagnostic testing that they otherwise might not have done.

#### **What are the advantages the Full Integrated Screening?**

The advantage to these screening tests is that they can give you a better estimate of your baby's risk for chromosomal problems at an early date without subjecting you to the small risk of miscarriage from a more invasive diagnostic test like CVS. If the risk is low, you can find out as soon as possible and may be relieved. If the risk is high you can decide whether to have CVS (done between 10 and 12 weeks), or amniocentesis at about 16 weeks. These tests give a definitive answer while still early in the pregnancy. The NT is noninvasive and carries no more risk than an ordinary ultrasound. Even if you forgo

diagnostic testing (CVS or amniocentesis), you can get more information about your baby's health and development by following up with a routine second trimester ultrasound at 18 to 20 weeks that looks for "soft markers" of chromosome disorders, such as short limbs, a bright dot in the heart, bright intestines, cysts in a portion of the baby's brain, and certain problems in the kidneys.

**What's the downside of these screening tests?**

Like any screening test, they are not diagnostic—that is, they cannot tell you definitively if your baby has normal chromosomes. In some cases they will lead the patient towards additional intervention. In other cases the tests will be incorrectly reassuring. The NT does not detect neural tube defects, such as spina bifida and other anomalies that may be indicated by the Quad Marker Screening (done at 15 to 20 weeks) and the second trimester ultrasound.

**What is the cost of the various components of these tests, and what if my insurance does not pay?** Because this procedure is separate and additional from your global obstetric services, it may not be a covered benefit. Because the California Prenatal Screening Program offers these tests, they are usually covered by insurance – but not always, so it is important to check with your insurance company. If you screen positive, the initial fee covers additional diagnostic testing. The nuchal translucency ultrasound for the Full Integrated Screening is not included in the California Prenatal Screening Program fee.