



Prenatal Screening for Down Syndrome, Anencephaly, Spina Bifida and Trisomy 18

As technology has advanced, our ability to screen for certain birth defects for pregnant women has improved. A screening test uses certain well defined tests to adjust the risk that an individual woman may have a particular condition – such as a baby with Down Syndrome. A screening test does not give a final answer, it simply adjusts the statistical odds. If the screening test is above a chosen cut-off value it is considered a positive test, if it is below that cut-off, it is considered negative. If the screening test is positive, then a diagnostic test may be considered which does give “the answer”. Diagnostic tests can be more invasive – such as amniocentesis – and have some risk involved – even a small risk of pregnancy loss. In deciding whether or not to have a screening test, one must consider what would be done if, in the end, the baby were to be diagnosed with, for example, Down Syndrome. Would pregnancy termination be contemplated? Or, would this be more “to be prepared” for special challenges. If it would not change how the pregnancy would be managed, then is it worth the risk of possibly needing to have an invasive test? Confluence Health is interested in providing the most current testing available to our patients. The Integrated Prenatal screen is the most current and most sensitive prenatal screening available for Down Syndrome, Anencephaly, Spina Bifida and Trisomy 18.

INTEGRATED PRENATAL SCREENING

- **What Is The Integrated Prenatal Screening Test?**

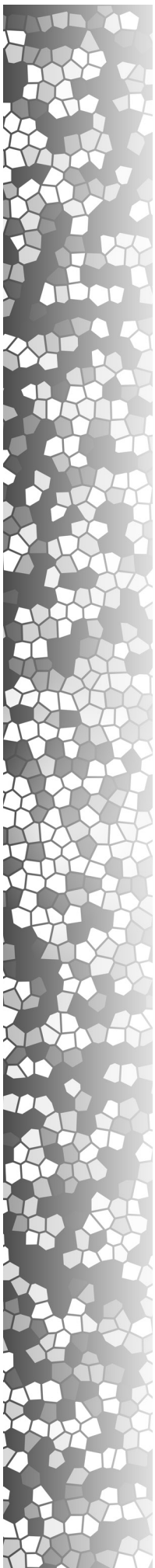
It combines the use of:

1. An ultrasound examination to measure the amount of fluid accumulation behind the baby’s neck (nuchal translucency).
2. A laboratory test performed on the mother’s blood at 10 weeks 3 days to 13 weeks 6 days weeks to measure the levels of a protein normally found in the blood of all pregnant women
 - PAPP-A (pregnancy associated plasma protein-A)
3. A laboratory test performed on the mother’s blood at 15 –18 weeks to measure the levels of 4 proteins normally found in the blood of all pregnant women
 - AFP (Alpha fetoprotein)
 - hCG (Human chorionic gonadotropin)
 - E3 (unconjugated estriol)
 - Inhibin A

The Integrated Screen will detect up to 92% of babies affected with Down Syndrome and up to 90% with Trisomy 18. It will also detect up to 80% of babies that have open neural tube defects such as Spina Bifida. Since the first and second trimester results are combined, your risk assessment will not be available until your second trimester blood work is completed.

- **What is Down Syndrome?**

In each of our cells, we have 46 chromosomes (23 from the mother and 23 from the father). They give instructions for our development. Down Syndrome results when there is an extra number 21 chromosome (three, instead of two). Any woman, at any age, can have a baby with Down Syndrome, although the risk increases with the age of the mother. People with Down Syndrome usually have mild to moderate mental retardation, as well as other medical problems.



- **What is Trisomy 18?**

Trisomy 18 is a severe chromosome abnormality that causes multiple birth defects and very severe mental retardation. Few babies with Trisomy 18 survive into childhood. Trisomy 18 results when the fetus has three, instead of the normal two, copies of chromosome 18. Like Down Syndrome, the chance to have a pregnancy affected with Trisomy 18 increases with increased maternal age.
- **What is an Open Neural Tube Defect?**

The neural tube, which forms very early in pregnancy, eventually develops into the baby's brain and the spinal cord. If the tube does not close completely, an opening remains along part of the baby's spine or head. If the opening is in the spine (Spina Bifida), the symptoms range from paralysis of the legs to lack of bowel and bladder control to hydrocephaly (water on the brain). If the opening is in the head (Anencephaly), brain development is incomplete. These babies are often stillborn or die shortly after birth. Open neural tube defects occur in 1 or 2 out of every 1000 births.
- **What does it mean if my screen is "Positive"?**

About 5% of women will have a "positive" screen. This increase in risk does not mean the baby has a chromosome abnormality. It simply indicates that further tests need to be considered. The options for further testing may include ultrasound examination and amniocentesis for fetal chromosome analysis. These additional tests will accurately diagnose Down Syndrome, Trisomy 18 and neural tube defects. Remember that the Integrated Screen is not a diagnostic test. It cannot tell you whether your unborn baby has or doesn't have a problem. It will simply tell you the odds (chance) that the baby has a problem. Keep in mind that the vast majority of women with "screen positive" results do not have babies with Down Syndrome, Trisomy 18 or a neural tube defect.
- **What does it mean if my screen is "Negative"?**

A negative result indicates that the risk that the fetus has one of these conditions is below the accepted cutoff value. However, a negative result does not completely eliminate the possibility that the fetus may have Down Syndrome, Trisomy 18, other chromosome abnormalities or a neural tube defect.
- **When should this testing be performed?**

Between 11 weeks 0 days and 13 weeks 6 days, 1st trimester blood and ultrasound nuchal (NT) measurement are performed.
Then: Between 15 weeks 0 days and 20 weeks 6 days 2nd trimester blood tests are done.
- **How can I get more information?**

Ask your physician. If you want the test, special arrangements for an ultrasound by a certified ultrasound technologist, along with appropriate blood testing, can be made.