First Trimester Screening

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The St. Cloud Perinatal Clinic offers First Trimester Screening to all pregnant women between 11 and 13 weeks 6 days gestation. This is a screen that uses blood and ultrasound markers to help us identify women who may be at an increased risk for having a child with Down syndrome, Trisomy 18 or Trisomy 13.

What conditions are screened for?

- Down syndrome is a medical problem that results from having too much genetic information. Individuals with Down syndrome have an extra chromosome 21. Children with Down syndrome have characteristic facial features, learning difficulties, intellectual disabilities, and an increased risk for birth defects.
- Trisomy 18 and Trisomy 13 also result from having too much genetic information, but these conditions are more severe than Down syndrome. Many babies or children with these medical problems die during the pregnancy or within the first year of life due to birth defects and respiratory troubles. Those that survive have serious learning difficulties. These children are often unable to walk or talk.

Risk associated with maternal age

Every woman has a chance of having a child with a chromosome issue like Down syndrome, Trisomy 18 and Trisomy 13. This risk increases with increasing maternal age. For example:

Maternal age at delivery	Chance for having a baby with an extra chromosome
20	**0.2%**
30	**0.3%**
35	**0.5%**
40	**1.5%**

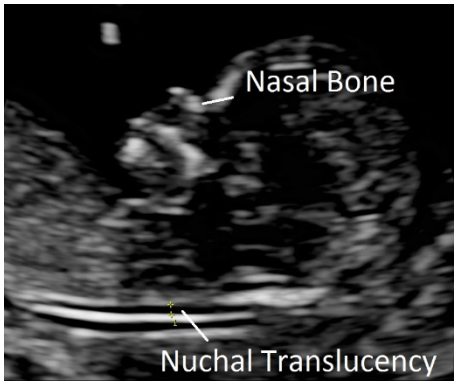
How does the First Trimester Screen work?

The First Trimester Screening gives a pregnancy an estimate of the risk for having a baby with one of the above conditions.

The First Trimester Screen measures chemicals that are produced by every pregnancy, free Beta hCG, PAPP-A (pregnancy-associated plasma protein-A), AFP (alpha fetoprotein), PIGF (placental growth factor) and dimeric inhibin A (DIA). These chemicals are measured from a **maternal blood draw** that is between 10 weeks 0 days and 13 weeks 6 days gestation.

Many babies with Down syndrome produce lower levels of PAPP-A, PIGF and AFP and higher levels of free Beta hCG and Inhibin. Babies with Trisomy 18 or 13 usually produce lower levels of hCG and PAPP-A.

The First Trimester Screen also includes an **ultrasound** measurement of a small amount of fluid that accumulates at the back of a developing baby's neck. This is called "nuchal translucency". Babies with a chromosome issue like Down syndrome, Trisomy 13, or Trisomy 18 often have more fluid than usual (an "increased nuchal translucency"). An increased nuchal translucency can also be seen in some babies with heart defects. Presence or absence of the nasal bone is also noted. Babies with Down syndrome may have an absent or short nasal bone.



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What will the results tell you?

The First Trimester Screen is considered "increased risk for Down syndrome" if the chance of carrying a baby with this condition is ≥ 1 in 300. A screen is considered "increased risk for Trisomy 18 or 13" if the chance of carrying a baby with this condition is ≥ 1 in 150.

In most cases, the test reveals that the baby is not at an increased risk for Down syndrome, Trisomy 13 or Trisomy 18. No further testing is offered to women with a negative screen. However, it is important to remember that a negative screen **DOES NOT** eliminate the chance of having a baby with the above conditions.

Some women will get an "increased risk" screen result. An "increased risk" screen **DOES NOT** mean the baby has the condition, but further investigation should occur. These women will be offered genetic counseling and further testing such as a detailed anatomic survey, non-invasive prenatal screening, chorionic villi sampling, or amniocentesis.

The First Trimester Screen detects:

- ~98% of babies with Down syndrome
- ~95% of babies with Trisomy 18 or 13

Limitations of testing

No test can guarantee a baby will not have any medical issues. First trimester screening only assesses the risk for Down syndrome, Trisomy 13, and Trisomy 18. This screen does not test for all genetic and non-genetic problems that may be present in a baby. If the test result is normal, it does not eliminate the risk for these conditions. False positives and false negatives can occur.

Additional Testing

Non-invasive prenatal testing (NIPT) is a test that measures the amount of genetic material in a pregnant woman's blood. This test can be done any time after 10 weeks gestation. NIPT detects:

- > 99% percent of babies with Down syndrome
- ~ 98% of Trisomy 18
- ~ 91% of Trisomy 13

Ultrasound is another form of screening for chromosome problems. Ultrasound detects:

- ~50% of babies with Down syndrome
- ~90% of babies with Trisomy 18 or 13.

First Trimester Screening does not assess the risk for having a child with spina bifida. Screening for this condition is offered in the second trimester by measuring the level of AFP ONLY between weeks 15- 22 gestation. Ultrasound may also be used.

Screenings like the First Trimester Screen, NIPT and ultrasound can tell you the chance of your pregnancy having a certain chromosome problem (for example, 1 in 50 or 1 in 5,000) but they do not provide a definitive answer. Invasive testing procedures (such as a CVS or amniocentesis) can provide a more definitive answer. With invasive testing there is a small risk of complications including miscarriage.

Why have First Trimester Screening?

Some parents do First Trimester Screening because they want more information about the risks for a chromosome problem specific to their current pregnancy. If additional testing confirms that their baby has a chromosome problem, they might find it helpful to use this information to plan and prepare for their child's condition. Others in this situation may choose not to continue a pregnancy.

There is no prenatal treatment or cure when we learn that a child has a chromosome problem during pregnancy, but we do follow these pregnancies more closely. This information may also impact plans for delivery. In some cases, early treatment can have great benefits for the child's long-term health.

Please feel free to ask your health care provider if you have further questions about this screening.

Insurance Information

You should check with your insurance company to see if they cover this screening. You will need to ask if they cover CPT codes: **36415** (venipuncture), 76801 and 76813 (ultrasound), and **84164, 84704, 83520, 86336** and **82105** (lab) using the diagnosis of **ICD-10 code Z36.**

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