What do the results of the FirstTrimesterScreen | Fβ mean?

Negative screen

A negative screen occurs in about 95% of all tests. **This means your baby is at low risk for Down syndrome, trisomy 18, or trisomy 13**. If you get a negative screen, your doctor may decide to stop screening or may continue with more tests in your second trimester for additional verification. A negative screen does not completely eliminate the possibility your baby may have Down syndrome, trisomy 18, or trisomy 13.

Positive screen

A positive screen occurs in about 5% of all tests. **This means your baby is at increased risk for Down syndrome, trisomy 18, or trisomy 13**. When you receive this result in your first trimester, you and your doctor may choose to consider additional testing options such as non-invasive prenatal testing (NIPT), or diagnostic testing options, like chorionic villus sampling (CVS) or amniocentesis.

About Eurofins NTD

For more than 30 years, Eurofins NTD has pioneered the research and development of prenatal screening protocols for open neural tube defects, Down syndrome, trisomy 13 and 18, and early onset preeclampsia screening Today, Eurofins NTD serves universities, medical centers, hospitals, laboratories, obstetricians and maternal fetal medicine specialists worldwide—providing risk assessment services that help healthcare professionals and patients make more informed medical decisions.

Learn more about Maternal Fetal Screen℠ | T1 by visiting **www.ntd-eurofins.com** or contacting us at **1-888-NTD-LABS** (683-5227).

NTD | Eurofins Clinical Diagnostics

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FirstTrimesterScreen | Fβ

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Why should I have prenatal testing?

According to the American College of Obstetricians and Gynecologists (ACOG), most children in the United States are born healthy. Only 2 or 3 out of 100 newborns have major birth defects. For the majority of babies the cause is unknown. **However, there are certain birth defects that can be tested for prenatally—before a baby is born**. These include Down syndrome, trisomy 18, trisomy 13, and open neural tube defects. The risk of having a baby with a chromosome abnormality, such as Down syndrome, increases with the mother's age. However, **ACOG recommends prenatal testing be offered to all pregnant women, regardless of age**.

The First Trimester Screen Fß is a test performed at NTD Labs to show if you are at increased risk of having a baby with Down syndrome, trisomy 18, or trisomy 13.

What is Down syndrome?

Down syndrome, also known as trisomy 21, is the most commonly occurring chromosome abnormality. It is caused by having an extra copy of chromosome 21—for a total of 3 copies instead of the normal 2. **Down syndrome often results in developmental problems and a higher risk of conditions including heart defects, mental retardation, breathing and hearing problems, and childhood leukemia**. The severity of these conditions varies greatly from individual to individual.

What is trisomy 18?

Trisomy 18, or Edwards syndrome, is the second most common trisomy—a condition in which someone has 3 copies of a certain chromosome. It is caused by having an extra copy of chromosome 18—for a total of 3 copies instead of the normal 2. **Trisomy 18 causes developmental problems associated with life-threatening complications in a baby's first months and years**. These may include deformities of the heart, intestines, esophagus, hands, and feet; kidney problems; delayed growth; and mental retardation.

What is trisomy 13?

Trisomy 13, or Patau syndrome, is another well-known chromosome abnormality. It is caused by having an extra copy of chromosome 13—for a total of 3 copies instead of the normal 2. **Trisomy 13 is associated with severe mental and physical problems that cause many infants to die during their first days or weeks of life**. Infants with trisomy 13 often have problems including abnormalities of the heart, brain, or spinal cord; small or poorly developed eyes; extra fingers and/or toes; deformed mouths; and weak muscle tone.

Why is prenatal testing important?

Being tested during pregnancy will allow you to be informed about the health of your baby, before your delivery. The information you receive will be extremely valuable in helping you plan for your baby's birth. You and your doctor will also be able to decide if you need more testing later in your pregnancy. If you learn you will have a baby with one of these birth defects, **you can proactively prepare your family for the future by making important medical and financial decisions about care**.

If I receive a positive screen, what additional tests may I take?

If you are at risk of having a baby with a birth defect your doctor may recommend additional testing.

Non-invasive prenatal testing, or **NIPT**, is a type of screening test that examines fetal DNA which is present in the mother's blood. It is performed after 10 weeks of pregnancy by a blood draw and detects the same type of chromosome abnormalities that are examined by the The First Trimester Screen Fß. It can also detect problems with the sex chromosomes (the X and Y chromosomes).

Chorionic villus sampling, or **CVS**, is performed at 10 to 12 weeks of pregnancy. The sample is sent to a lab and the cells are checked for chromosome abnormalities such as Down syndrome, trisomy 18, and trisomy 13. CVS is an invasive diagnostic test and is associated with a small risk of miscarriage.

Another test, known as **amniocentesis**, is performed between 15 and 20 weeks of pregnancy. The cells are checked for chromosome abnormalities such as Down syndrome, trisomy 18, and trisomy 13. Amniocentesis is a diagnostic test. Amniocentesis is an invasive diagnostic test and is associated with a small risk of miscarriage.

Be sure to discuss these additional tests with your doctor to determine which options may be right for you.

How is the FirstTrimesterScreen Fß performed?

The First Trimester Screen Fß is a simple blood test performed on you when a fetus is between 9 weeks and 13 weeks, 6 days of age. Your blood is analyzed for 3 markers normally found in all pregnant women.

The blood test is followed by an ultrasound examination, given when the fetus is between 11 weeks, 1 day and 13 weeks, 6 days of age. The ultrasound confirms your baby's age and measures the amount of fluid behind the baby's neck.

Results of the blood test and the ultrasound are combined to estimate the risk of Down syndrome, trisomy 18, and trisomy 13.

- *96% of Down syndrome cases are detected*
- *95% of trisomy 18 and trisomy 13 cases are detected*

If you are screened for chromosome abnormalities in your first trimester, **you should be sure to be screened for open neural tube defects (ONTDs) in your second trimester**. ONTDs, such as spina bifida, result from improper development of the brain and spinal cord, which may cause an opening to remain along the spine or head after the baby is born.

ONTDs can be detected through a maternal serum alpha-fetoprotein (MSAFP) screening. The maternal serum AFP test is noninvasive. Blood is drawn from the mother's arm and sent to the lab for analysis. High levels of AFP in the blood may indicate that the developing fetus has an ONTD.