

# Prenatal Screening Tests



**PERINATAL ASSOCIATES**  
OF NEW MEXICO, LTD

## **First and Second Trimester Screening Tests**

### **What is a screening test?**

A screening test is a test that may be able to find fetuses (unborn babies) that have an increased risk for certain birth defects or other conditions. Anyone can have a baby with birth defects. There is not one screening test that can find all birth defects or problems. Screening tests are designed to detect fetuses with specific conditions. The First Trimester Combined Screen (**FTCS**) and Integrated Screen (**IS**) are discussed in this brochure.





## **What is the First Trimester Combined Screen?**

First Trimester Combined Screening is a screening test done between 11 weeks and 13.6 weeks gestation (counting from the first day of the last menstrual period). It is a safe test which does not pose a risk for pregnancy complications. It involves an ultrasound and a sample of the maternal blood. A specific measurement of the fetus, called nuchal translucency, (NT), is taken. This is essentially the space between the back of the neck and the skin over it. This measurement is often thickened, or enlarged in fetuses that have chromosome abnormalities. The chromosome abnormalities screened for in this test include Down Syndrome, Trisomy 13 and Trisomy 18. These conditions are caused by an extra chromosome in every cell of the body, which causes mental retardation and birth defects. The vast majority of these three conditions are not inherited so there is usually no family history of these conditions. The chance of having a baby with one of these conditions increases with maternal age, but they can happen to anyone.

The nuchal translucency measurement is also often enlarged in babies that have heart defects, so if the NT is enlarged it may also mean the heart should be carefully examined with ultrasound later in the pregnancy when it is easier to see.

The blood sample is analyzed, measuring two substances found in the maternal blood (freeBeta HCG and PAPP-A). The level of these two substances is often different in healthy babies verses those that have Down syndrome, Trisomy 13 or Trisomy 18.

## Who is eligible for First Trimester Combined Screening?

Most women between 11-13.6 weeks pregnant from the first day of their last menstrual period and from the ages of teen to over 35 years old. This is an optional test but is not a substitute for a diagnostic test (yes or no answer test) when indicated.

## How accurate is First Trimester Combined Screening?

About 85% of fetuses who have Down syndrome, Trisomy 13 or Trisomy 18 will have an “increased risk” on FTCS. This means that the Detection Rate is 85%. About 15% of fetuses that are affected would be undetected. About 5% of unaffected (normal) fetuses will also show an “increased risk” on FTCS. This means that the False Positive Rate is 5%.

## When will I get my results for the First Trimester Combined Screening?

Results for the FTCS will be available in 3-5 business days and will be sent to your primary OB provider. Results are reported as “Increased Risk” (risk higher than cut off) or “Within Normal Range” (risk lower than cut off). The cut off is equal to the risk of chromosome problems for a woman who is 35 years old. You will receive a phone call either way.





## What do I need to do if my First Trimester Combined Screening results are negative?

A negative screen means that your risks of having a baby with Down syndrome, Trisomy 13 or Trisomy 18 are less than the screening cut off. **FTCS** can never eliminate the risk for Down Syndrome, Trisomy 13 and Trisomy 18.

It is recommended that all pregnancies be screened for open neural tube defects (such as spina bifida). Neural tube defects are birth defects that involve an opening on the spine. They can result in paralysis, hydrocephalus, and other complications. If you have **FTCS** you should also have a thorough ultrasound examination between 18 and 22 weeks gestation and a single marker AFP blood test. These two tests together will detect over 95% of all open neural tube defects.



## **What if my results from the First Trimester Combined Screening is positive?**

A positive FTCS result means that your risks of having a baby with Down Syndrome, Trisomy 13 and Trisomy 18 are greater than the screening cut off. *It does not mean a chromosome abnormality has been diagnosed.*

If you have a positive FTCS, Diagnostic testing is available by either chorionic villus sampling (CVS, between 10 and 13 weeks) or amniocentesis (after 15 weeks).

You will be offered genetic counseling to discuss the results of the screen and learn more about options for further testing in greater detail.





## **What is the Second Trimester (Integrated Screening)?**

Integrated Screening (IS) is a screening test that requires two visits to the doctor's office. One visit is in the first trimester (between 11 and 13.6 weeks gestation) and the second visit is during the second trimester (between 15 and 20 weeks gestation). The first visit, similar to the First Trimester Combined Screen, which involves an ultrasound (NT measurement) and bloodwork. The second visit involves a blood draw only. The blood draw is for an AFP4, or Quad Screen, which provides additional information about Down syndrome and Trisomy 13 and Trisomy 18 risks as well as open neural tube defects. The results from these two visits are integrated to provide you with a single result between 15 and 20 weeks gestation. As with FTCS this test does not pose risk for pregnancy complications. Integrated Screening may detect fetuses at increased risk for Down Syndrome, Trisomy 13, Trisomy 18 and open neural tube defects.

## **How accurate is Integrated Screening?**

The Detection Rate with **IS** is about 90% and the False Positive Rate is 2.5%.

One important fact about Integrated Screening is that the results are received **AFTER** the results return from the second appointment. No information is given after the first visit, with the rare exception of a severe abnormality on ultrasound. If there is a severe ultrasound abnormality, you would be told by a physician and testing options would be discussed, probably with a genetic counselor.

## **Can First Trimester Combined Screening or Integrated Screening tell me for sure if my baby will have one of these conditions?**

**NO.** No screening test is 100% accurate. The purpose of a screening test is to find out if your chance of having a baby with one of these conditions is higher than should be expected. If your unborn baby is determined to be at higher risk, additional testing would be offered to determine (with a very high degree of accuracy) if the baby actually has Down Syndrome, Trisomy 13, Trisomy 18, or an open neural tube defect. A test that has an extremely high degree of accuracy is called a diagnostic test. This can include CVS (chronic villus sampling) or amniocentesis. Both are a least 99% accurate.

Both are optional as well. The benefits and risks of these tests would most likely be discussed with a genetic counselor.

## **Can I do the First Trimester Combined screening in the first trimester and the AFP4 in the second trimester and get both results separately?**

This would be called sequential screening. While it can be done, our physicians prefer not to perform testing in this way at this time. The False Positive Rate could be higher (especially in women over 35 years of age), and if the results are conflicting, such as one result being normal and the other abnormal, it could cause additional confusion and anxiety.

## **Which test is right for me?**

First, it would be best to decide if this is the type of information you would like to have. If you would like to have additional information about your unborn baby's risks for Down Syndrome, Trisomy 13, Trisomy 18, or open neural tube defects, then you can choose First Trimester Combined Screening or Integrated Screening.

**FTCS** may be helpful to women who would like a screening test, who would like the information earlier in the pregnancy, and who would like immediate feedback regarding the ultrasound findings.

**IS** would be most helpful to women who want the highest detection rate and a lower false positive rate. You should understand that you will receive your results, and must be certain that you can keep **both** office visit appointments at the scheduled time.

## **What is a Genetic Counselor?**

A Board Certified Genetic Counselor provides information and support to women, couples and families. The genetic counselor will review your medical, family and pregnancy histories, and any risk factors present (such as screening test results). You can discuss genetic testing options, test results, and risks and benefits of many types of testing with a genetic counselor. A genetic counselor will not tell you which decisions to make. A genetic counselor encourages people to make decisions that reflect their own personal and cultural beliefs, values and goals. Genetic counselors support the decisions made by couples and individuals.



## **Perinatal Associates**

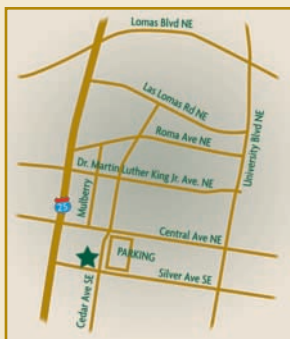
Perinatal Associates of New Mexico, Ltd., is a group of physicians specializing in high-risk pregnancies.

Our team of doctors, nurse practitioners, genetic counselors and other trained professionals are here to help you with your pregnancy. Working together, we want to assist you in overcoming problems during this potentially stressful time.

We welcome any questions you may have about your treatment and your pregnancy.



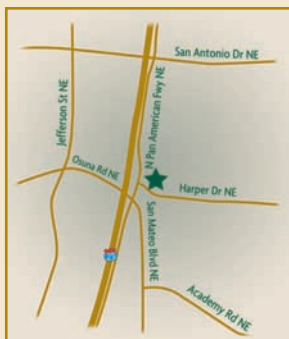
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