

First Trimester Screen

The First Trimester Screen is a new, optional non-invasive evaluation that combines a maternal blood screening test with an [ultrasound](#) evaluation of the fetus to identify risk for specific chromosomal abnormalities, including [Down Syndrome Trisomy-21](#) and Trisomy-18.

In addition to screening for these abnormalities, a portion of the test (known as the nuchal translucency) can assist in identifying other significant fetal abnormalities, such as cardiac disorders. The screening test does not detect neural tube defects.

The first trimester screen has been available in the U.S. for several years, but has only recently been determined an effective means of early chromosomal abnormality screening. A study published in the New England Journal of Medicine in November 2005 determined that first trimester screening was the most accurate non-invasive screening method available.

The combined accuracy rate for the screen to detect the chromosomal abnormalities mentioned above is approximately 85% with a false positive rate of 5%.

This means that:

- Approximately 85 out of every 100 babies affected by the abnormalities addressed by the screen will be identified.
- Approximately 5% of all normal pregnancies will receive a positive result or an abnormal level.
- A positive test means you have a 1/100 to 1/300 chance of experiencing one of the abnormalities.

It is important to realize a positive result does not equate to having an abnormality, but rather serves as a prompt to discuss further testing. The screen should not be confused with screens performed during the [second trimester](#) (often known as the [Quad Screen](#) or [Triple Screen](#)). These screening methods are less accurate and are performed between 15-20 weeks. The blood screen measures two pregnancy related hormones: [hCG](#) and PAPP-A. The ultrasound evaluation measures nuchal translucency (fluid beneath the skin behind baby's neck). This non-invasive procedure combines the results from the [blood tests](#) and the ultrasound, along with the mother's age, to determine risk factors.

What Is A Screening Test?

It is very important to remember what a screening test is before getting one performed. This will help alleviate some of the anxiety that can accompany test results. Screening tests do not look only at results from the [blood test](#). They compare a number of different factors (including age, ethnicity, results from blood tests, etc...) and then estimate what a person's chances are of having an abnormality.

These tests DO NOT diagnose a problem; they only signal further testing should be done.

How Is The First Trimester Screen Performed?

The blood screen involves drawing blood from the mother, which takes about 5 to 10 minutes. The blood sample is then sent to the laboratory for testing. The ultrasound is performed by an ultrasound specialist or perinatologist and takes between 20 and 40 minutes. The results are evaluated within a week of the testing.

What Are The Risks And Side Effects To The Mother Or Baby?

Except for the discomfort of drawing blood, there are no known risks or side effects associated with the First Trimester screen. There is a 5% false positive rate for the test. Parents should be aware of the possibility of receiving abnormal results and then finding, after further testing, the baby is normal.

When Is The First Trimester Screen Performed?

The First Trimester Screen is performed between the 11th and [13th week of pregnancy](#). Because the test is performed so early, it is often used to determine whether a mother should consider undergoing an early ([first trimester](#)) diagnostic test, such as chorionic villus sampling, or second trimester [amniocentesis](#).

What Does The First Trimester Screen Look For?

In babies who are at an increased risk for chromosomal abnormalities, increased fluid is often found in the nuchal translucency. Abnormally high or low hCG and PAPP-A levels are also often found. The first trimester screen combines the results from these three measurements (nuchal translucency, hCG, and PAPP-A) with maternal age risk factors and determines an overall risk factor for chromosomal abnormalities.

What Do The First Trimester Screen Results Mean?

It is important to remember the First Trimester Screen is a screening test and not a diagnostic test. This test only notes a mother is at risk of carrying a baby with a genetic disorder. Many women who experience an abnormal test discover later the test proved false. You will not be given specific quantitative values for the separate parts of the First Trimester screen. Instead, you will be told whether your results are “normal or abnormal”, and you will be given a risk level by your genetic counselor. The counselor will give you your risk factor for chromosomal abnormalities based on the test results (for example 1/250, 1/1300). Abnormal test results warrant additional testing for making a diagnosis. Your genetic counselor will discuss the results with you and assist you in deciding about diagnostic tests, such as CVS or [amniocentesis](#). These invasive procedures should be discussed thoroughly with your healthcare provider and between you and your partner. Additional counseling may prove helpful.

What Are The Reasons For Further Testing?

The First Trimester Screen is a routine screening that is not an invasive procedure and poses no known risks to the mother or baby. The First Trimester Screen results may warrant additional testing. The reasons to pursue further testing or not vary from person to person and couple to couple.

Performing further testing allows you to confirm a diagnosis and then provides you with certain opportunities:

- Pursue potential interventions that may exist (i.e. fetal surgery for [spina bifida](#))
- Begin planning for a child with special needs
- Start addressing anticipated lifestyle changes
- Identify support groups and resources
- Make a decision about carrying the child to term

Some individuals or couples may elect not to pursue testing or additional testing for various reasons:

- They are comfortable with the results no matter what the outcome
- Because of personal, moral, or religious reasons, making a decision about carrying the child to term is not an option
- Some parents choose not to allow any testing that poses any risk of harming the developing baby
- It is important to discuss the risks and benefits of testing thoroughly with your healthcare provider. Your healthcare provider will help you evaluate if the benefits from the results could outweigh any risks from the procedure.

<http://americanpregnancy.org/prenatal-testing/first-trimester-screen/>
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