

Patient Information: First Trimester Screening

What is first trimester screening?

The first trimester screen is an optional test that is available to all pregnant women. It is done between the 12th and 14th weeks of pregnancy. The first trimester screen is designed to estimate the chance of having a baby with Down syndrome (Trisomy 21) and the chance of having a baby with trisomy 13 or trisomy 18. This test cannot determine if the baby actually has one of these conditions; instead, it helps to find pregnancies that may be at a higher risk. If a higher risk is identified, further tests that can confirm or rule out these conditions are available.

What does the screening involve?

The test involves a combination of a special ultrasound examination and a blood test:

Ultrasound: The ultrasound examination measures a space found at the back of the baby's neck called the nuchal translucency (NT) as well as the presence or absence of the baby's nasal bone. A sonographer, certified by the Fetal Medicine Foundation, completes the ultrasound portion of the First Trimester Screen. A special doctor called a perinatologist reviews all ultrasounds and is available for any concerns.

Blood Test: For the blood test, a small amount of blood is drawn from a finger prick to measure levels of a hormone and a protein that are found in a woman's blood during pregnancy.

The information from the ultrasound and the blood test are combined together to estimate the chance that a baby has Down syndrome and the chance that a baby has trisomy 13 or trisomy 18.

When will I get results? How are they reported?

The final combined results are available about 7 days after the test is performed. Results are reported as an "increased risk" (risk higher than the cut-off) or "within the normal range" (risk lower than the cut-off). The cut-off for Down syndrome represents the risk for a woman who is 35 years old. If the results are in the normal range, you will receive a letter in the mail. If the results show an increased risk, a genetic counselor will contact you by phone to discuss the results of the screen and options for further testing. All results will be forwarded to your doctor or midwife.

What is Instant Risk Assessment?

An "Instant Risk Assessment" allows you to get your first trimester screen results more quickly and in person. To do this, you come to our clinic about one week prior to the ultrasound appointment for your blood test. You then return to the clinic for ultrasound. Once the ultrasound is completed, a genetic counselor enters your ultrasound results into the laboratory's secure website and obtains your combined test results. The genetic counselor then reviews the final results with you in person right after your ultrasound appointment. When you schedule your appointment you can ask for this option.

What do I need to do if my results show that the risk is in the normal range?

When the results are in the normal range it means that your risks for Down syndrome or trisomy 13 or trisomy 18 are less than the screening cut-off. First trimester screening can never eliminate the chance for Down syndrome, trisomy 13, trisomy 18 or other health problems. Many people feel reassured by a result that is in the normal range and they may decide to not have further testing such as a CVS or amniocentesis. Other people may wish to have a definitive diagnosis and so they may choose further testing.

What if my results show an increased risk?

A test result that shows an increased risk means that the chance for Down syndrome or trisomy 13 or trisomy 18 is higher than the screening cut-off. *An abnormal First Trimester Screen result does not mean that the baby has one of these conditions. In many cases, the baby does not have Down syndrome, trisomy 18 or trisomy 13.* If your first trimester screen results show an increased risk, diagnostic testing is available by either Chorionic Villus Sampling (CVS) until 14 weeks, or Amniocentesis after 16 weeks. Other screening options may also be considered such as Noninvasive Prenatal Testing or Targeted Ultrasound. The genetic counselor can discuss these options with you in greater detail.

What is the accuracy of the test?

First trimester screening can detect about 91-95% of pregnancies with Down syndrome and about 95% of babies with trisomy 13 and trisomy 18.

What if I have a family history of Down syndrome or other chromosome or genetic conditions?

Patients/couples with a family history of any genetic condition (including Down syndrome) should discuss testing options with their doctor and/or genetic counselor.

What else can first trimester screening detect?

If the nuchal translucency is larger than expected, there is a higher chance of other health problems such as a heart defect. If the nuchal translucency is larger than expected, you will meet with a genetic counselor to discuss your results.

Can all birth defects be detected by first trimester screening or any other screening?

No. No prenatal test can guarantee a child free of birth defects or learning problems.

Where is the First Trimester Screen performed?

This test is completed at the Center for Perinatal Care in Meriter Hospital. Valet parking or parking within Meriter's parking ramp are both available free of charge to our patients.

How long does the test take?

In general, we schedule 75 minutes for the total First Trimester Screen appointment, however this time can vary. You will first meet with a genetic counselor to review the test process and obtain a blood sample. Next, you will have your ultrasound. The blood sample and ultrasound measurement will be sent to the laboratory, and the combined results are available in about 7 days.

Questions?

Please feel free to contact us if you have any questions or concerns (608) 417-6667. Also, please visit our website (www.meriter.com) for more information about other services provided at Meriter's Center for Perinatal Care.

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