



If My First Trimester Screening is Normal, Can I Be Certain Everything's All Right?

First trimester screening, even with today's advanced techniques, cannot guarantee a healthy baby. First trimester screening will identify pregnancies in which the baby has Down syndrome about 85% of the time. This means that if first trimester screening was done on 100 women whose babies actually had Down syndrome, 85 of them would show an increased risk and follow-up testing would be offered. However, 15 of them would have a normal screening result and would not be offered any additional testing. First trimester screening identifies pregnancies in which the baby has Trisomy 18 in about two out of three Trisomy 18 pregnancies. First trimester screening cannot identify pregnancies in which a baby has other birth defects. But remember, most babies are born healthy.

If My Screening Is Normal, Do I Need Other Tests?

First trimester screening cannot detect all potential birth defects, so your doctor will offer you other routine tests later in your pregnancy. These will include a detailed ultrasound during the middle of your pregnancy, and a Maternal Serum AFP test (to check for neural tube defects like spina bifida) at 15 to 18 weeks.

For more information about First Trimester Screening, ask your doctor or call (585) 487-3480.

For detailed information, please visit our web page at: <https://www.urmc.rochester.edu/ob-gyn/genetics/services/tests-duringpregnancy.aspx>.

Prenatal Screening Program
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A close-up photograph of a woman with dark hair resting her chin on her hand, looking thoughtfully to the side. The image is set against a blue background with a yellow curved border at the top and bottom.

First Trimester Screening Is it right for you?

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First Trimester Screening

Would you want to know in your first trimester (2 1/2 to 3 months of pregnancy) if you had an increased risk of having a baby with certain birth defects? If you knew you had a higher chance, would you want additional testing to tell for sure?

If you think the answer to these questions is “yes,” first trimester screening might be right for you.

What is First Trimester Screening?

First trimester screening is done by combining information from a simple blood test and a specific ultrasound measurement of the fetus. Results can help you know if there is an increased chance for Down syndrome, or a different, less-common chromosome problem called Trisomy 18. If there is an increased risk you would have the option to choose additional testing such as a more-detailed ultrasound, amniocentesis or chorionic villus sampling, or non-invasive prenatal testing (by testing fetal cells in the mother’s blood).

The first trimester screening blood test measures two pregnancy proteins in a mother’s blood, hCG and PAPP-A. The ultrasound measures fluid in the skin at the back of the baby’s neck (Nuchal Translucency or “NT”). These measurements, in addition to the woman’s age, can be used to determine how high the chance is for Down syndrome or Trisomy 18. Most women will get a normal result and no further testing will be needed. However, about 3% of women will get a positive result, and these women will be offered additional testing.

Which Birth Defects May Be Identified by First Trimester Screening?

DOWN SYNDROME

Down syndrome is a condition in which the fetus has an extra 21st chromosome, meaning there are 47 chromosomes instead of the normal 46. Chromosomes contain the genes that determine our inherited characteristics. Down syndrome results in intellectual disability and sometimes physical or health problems such as heart defects.

Although the risk to have a child with Down syndrome is higher in older women, it also can occur in young women. While first trimester screening cannot tell for certain if a baby has Down syndrome, it can tell you if there is a higher chance for Down syndrome. If the risk is as high as a woman who is 35 or older, you may choose to have further testing.

TRISOMY 18

Trisomy 18 is caused by an extra 18th chromosome. It is much less common than Down syndrome, occurring in only about 1 in 5,000 births. It is a very serious birth defect that causes many health problems and severe intellectual disability. If first trimester screening shows an increased risk for Trisomy 18, you will be offered further more definitive testing.

HEART DEFECTS

Even if testing shows no evidence of Down syndrome or Trisomy 18, first trimester screening may reveal a higher chance for a baby to have a heart defect. This is the case in women whose Nuchal Translucency measurement is enlarged. In these cases the woman will be offered a fetal echocardiogram – a special ultrasound of the baby’s heart – during the middle part of the pregnancy (about 5 months).

How Do I Arrange to Have First Trimester Screening?

Your doctor may suggest first trimester screening at an early prenatal visit, or you can ask your doctor or health care provider about the screening. Here are some important details to remember:

- First trimester screening must take place between 11 to 14 weeks of pregnancy (3-4 months).
- Some doctors may perform the first trimester screening ultrasound right in their offices. Others may refer you to the UR Medicine Reproductive Genetics office (585-487-3480) to schedule an ultrasound.

Only ultrasound specialists with special certification may perform the NT measurement.



If you or your doctor need help arranging first trimester screening, please call (585) 487-3480 for assistance. Remember, this screening must take place before you are 14 weeks pregnant.

What Happens Next?

If your screening results show an increased risk, your doctor or genetic counselor will discuss your results and offer follow-up tests. If you’re still in the first trimester of pregnancy, chorionic villus sampling (CVS) may be an option. In this test, a small sample of tissue is taken from the placenta. These cells are genetically tested to show if the fetus has Down syndrome or Trisomy 18.

If you’re beyond your first trimester, amniocentesis is an option. In this procedure, a small amount of fluid is removed from the amniotic sac (bag of waters). Fetal cells in this fluid are examined to check for Down syndrome or Trisomy 18. Both CVS and amniocentesis are very accurate at identifying chromosome problems, but both tests have a very small chance of causing a miscarriage.

Another option for women in either the first or second trimester is an advanced blood test called “non-invasive prenatal testing.” This blood test is able to analyze fetal cells that are in the mother’s blood. It is not as accurate as CVS or amniocentesis. However, it will correctly determine if a baby has Down syndrome or Trisomy 18 about 99% of the time. Some women may choose to have only follow-up ultrasound or no additional testing at all.