If My 1st Trimester Screen is Normal, Can I Be Certain Everything is All Right?

The answer is "No." Although results from the 1st Trimester Screen are useful, they cannot guarantee a healthy baby. This is a screening test and it will detect pregnancies where the baby has Down syndrome or Trisomy 18 about 80% of the time. That means that for every 10 women who truly have babies with these chromosome problems, about 8 would have a positive 1st Trimester Screen, but 2 would have a normal screening result and would not be alerted to an increased risk. 1st Trimester Screening cannot identify pregnancies where the baby has other birth defects. But remember, most babies are born healthy and normal.

If My 1st Trimester Screen is Normal, Do I Need Any Other Tests?

Since there are birth defects that 1st Trimester Screening cannot detect, your doctor will want to schedule you for other routine tests later in your pregnancy. These will include a detailed ultrasound during the middle part of the pregnancy and a Maternal Serum AFP test (to check for neural tube defects like Spina Bifida) at 15 to 18 weeks of pregnancy.

If you would like more information about 1st Trimester Screening, ask your doctor or call (585) 487-3480.

Prenatal Screening Program
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URMC LABS

1st Trimester Screening

Is it right for you?

Prenatal Screening Program

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1st Trimester Screening

Would you want to take a test in the first trimester (2 1/2 to 3 months of pregnancy) to let you know if you have a higher chance to have a baby with certain birth defects?

If you learned that you had an increased risk, would you consider having other prenatal tests (CVS or amniocentesis) to tell for sure?

If you are over 35 and are already considering having prenatal testing because of your age, would you want a test that could help you know the chance that the baby has certain problems before you decide?

If you think the answer to these questions is "yes," and you are within the first 3 months of pregnancy, then 1st Trimester Screening might be right for you.

WHAT IS 1ST TRIMESTER SCREENING?

1st Trimester Screening is done by combining a blood test with a specific ultrasound measurement. With information from these tests, pregnant women can learn how high the chance is that the fetus has Down syndrome or a different chromosome problem called Trisomy 18. If a woman's chance to have a baby with one of these problems is high enough, she could choose to have a follow-up test to know for sure.

The blood test and ultrasound are safe for the mother and baby. The blood test measures two pregnancy proteins in the mother's blood, hCG and PAPP-A. The amount of these in the mother's blood can help know how high the chance is that the baby has Down syndrome or Trisomy 18. The ultrasound measures fluid in the skin on the baby's neck. This is called a "Nuchal Translucency" (NT) measurement. It is often enlarged in fetuses with Down syndrome or Trisomy 18.

By evaluating the hCG and PAPP-A in a pregnant woman's blood, and measuring the NT by a specially trained ultrasound specialist, many babies (about 8 out of 10) with Down syndrome or Trisomy 18, will be identified.

What Birth Defects Are Detected by 1st Trimester Screening?

DOWN SYNDROME

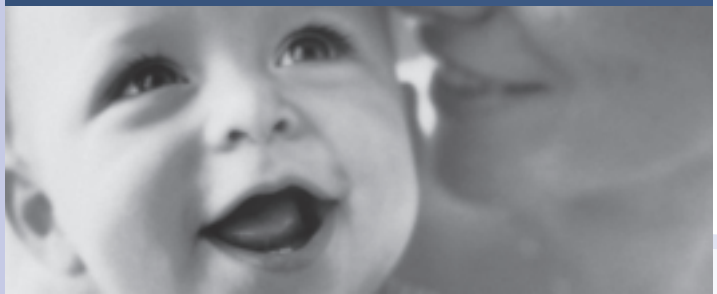
Down syndrome is a condition where the fetus has an extra 21st chromosome. Therefore, there are 47 chromosomes instead of the normal 46. The chromosomes contain the genetic material which determines our inherited characteristics. Down syndrome results in mental retardation and sometimes physical problems such as heart defects. Although the risk to have a child with Down syndrome is higher in older women, it also occurs in young women. 1st Trimester Screening cannot tell for sure if the baby has Down syndrome, but it can help women know whether they have a high or a low chance for Down syndrome. If a woman s chance for Down syndrome is as high or higher than a woman of 35, she may choose to have further definite testing.

TRISOMY 18

Trisomy 18 is caused by an extra 18th chromosome. It is much less common than Down syndrome. It is a very serious birth defect causing many health problems and severe mental retardation. If 1st Trimester Screening shows a high risk for Trisomy 18, the woman can choose to have further definite testing.

HEART DEFECTS

Even when further testing shows that the fetus does NOT have Down syndrome or Trisomy 18, there may be a higher chance for the baby to have a heart defect in women whose Nuchal Translucency measurement is enlarged. Therefore those women may wish to have a special ultrasound of the baby s heart (fetal echocardiogram) during the middle part of the pregnancy.



How Do I Arrange to Have 1st Trimester Screening?

Ist Trimester Screening must be done between about 11 to 14 weeks of pregnancy. Your doctor may discuss 1st Trimester Screening with you at your first prenatal visit. If not, and if you are interested, ask your doctor or health care provider about it. Some doctors' offices may be able to arrange to do the ultrasound right in their office. Others may refer you to the Strong Reproductive Genetics office to have it done. Only ultrasound specialists with special certification are able to do the NT measurement. If you or your doctor need help arranging this, call us at (585) 487-3480. We will arrange for you to meet with a genetic counselor to explain and discuss 1st Trimester Screening as well as other available tests. Remember, if you want this screening, it must be done before 14 weeks of pregnancy.

What Happens if My Result Shows an Increased Risk?

Your doctor or genetic counselor will discuss your results with you and discuss what follow-up tests are available. If you are still in the first trimester of the pregnancy, Chorionic Villus Sampling (CVS) may be an option. CVS is done by taking fetal cells from the placenta. Genetic testing can be done on these to show if the baby has Down syndrome or Trisomy 18.

If you are beyond the first trimester, amniocentesis would be an option. It is done by removing a small amount of fluid from the amniotic sac (the bag of waters). The fetal chromosomes can be examined in this fluid to check for Down syndrome or Trisomy 18. Both CVS and amniocentesis will show if the baby has a chromosome problem, but they both have a very small chance of causing a miscarriage. Some women may choose not to have either test.