What Are Down Syndrome and Trisomy 18?
Down syndrome and trisomy 18 are disorders caused by chromosome abnormalities. Chromosomes are packages of genetic material that exist in every cell in the body. They contain genes that act like sets of instructions that contribute to the way we look, how our bodies grow and develop, and our health.

Normally, the body’s cells contain 46 chromosomes arranged in 23 pairs. We inherit one copy of each chromosome from our mother and one from our father. Sometimes, however, a baby can be born with too many or too few chromosomes or with one or more chromosomes that are broken or rearranged. Errors in the number or structure of chromosomes cause a wide variety of birth defects ranging from mild to severe.

Down Syndrome
In Down syndrome, also called trisomy 21, the baby has an extra copy of chromosome number 21. All babies with Down syndrome have some degree of mental retardation and often have physical abnormalities, such as heart defects. About 1 in 800 babies is born with Down syndrome. First trimester screening detects 86% of Down syndrome pregnancies.

Trisomy 18
Trisomy 18 is also known as Edwards’ syndrome. Babies with this condition have an extra copy of chromosome number 18. Most babies with trisomy 18 die before their first birthday. Those who live longer are severely mentally impaired and have other serious health problems. Trisomy 18 is rare, occurring in 1 out of every 3,000 births. First trimester screening detects 75% of trisomy 18 pregnancies.

What Does It Mean If My First Trimester Serum Screening Test Is Negative?
A negative test result significantly reduces the likelihood that your baby has Down syndrome or trisomy 18; however, first trimester screening cannot completely rule out the possibility of these problems. In addition, first trimester screening cannot detect any other chromosome abnormalities or birth defects nor does it screen for open neural tube defects such as spina bifida. A different screening test called second trimester alpha-fetoprotein (AFP) screening can identify more than 80% of pregnancies with open neural tube defects.

Does a Positive First Trimester Serum Screening Test Mean My Baby Has Down Syndrome or Trisomy 18?
No. First trimester screening cannot diagnose problems with your baby or pregnancy. The test only tells you that you may be at an increased risk of having a baby with Down syndrome or trisomy 18. Typically, women who have a positive first trimester screening test result are offered additional tests that can provide a diagnosis.

If My Test Result Is Positive, What Happens Next?
Follow-up options are determined privately between you and your doctor. If your first trimester screening test is positive, your physician may recommend one or more of the following:

Genetic Counseling
Genetic counseling is designed to help you understand your test results and follow-up options and may include a discussion of your family and pregnancy history. Genetic counseling may be performed by a certified genetic counselor, a perinatologist (a specialist in high-risk pregnancies), or your own obstetrician.

Chorionic Villus Sampling (CVS)
CVS is a prenatal test procedure performed between 10 and 13 weeks of pregnancy by a trained specialist, such as a perinatologist. CVS removes a small piece of chorionic villus, which is the developing placenta. The placenta is special tissue that joins the baby to the mother and is sometimes called the afterbirth. The chorionic villus is made of the same genetic material as the developing baby. CVS can be performed in two ways: transcervically or transabdominally. During transcervical CVS, a thin tube is inserted into the vagina, threaded through the cervix (the bottom part of the uterus), and guided by ultrasound into the chorionic villus. Transabdominal CVS is performed by inserting a thin needle, guided by ultrasound, through the maternal abdomen into the chorionic villus. The chorionic villus cells are then analyzed to determine whether the baby has a chromosome abnormality such as Down syndrome or trisomy 18. There are some risks associated with the CVS procedure that you should discuss with your doctor. CVS identifies about 99% of chromosome abnormalities, but it cannot diagnose or identify all birth defects.

Ultrasound
Ultrasound is the use of high-frequency sound waves and a computer to create images of the developing baby. In the second trimester a detailed fetal anatomy ultrasound scan may be able to identify some birth defects, such as open neural tube defects (spina bifida). Babies with Down syndrome and trisomy 18 might have certain features that can be seen on ultrasound, but in general neither can be diagnosed from ultrasound alone.

Amniocentesis
Amniocentesis is a prenatal test procedure typically performed between 15 and 18 weeks of pregnancy by a perinatologist or obstetrician. During amniocentesis, a thin needle is inserted into the abdomen and uterus under ultrasound guidance, and a few teaspoons of amniotic fluid are removed. This fluid surrounds the baby and contains cells from the baby’s skin. The cells in the fluid are then examined in a laboratory to determine whether a chromosome abnormality like Down syndrome or trisomy 18 is present. There are some risks associated with this procedure that should be discussed with your physician. Amniocentesis helps to diagnose more than 99% of chromosome abnormalities, but, like CVS, cannot diagnose or identify all birth defects.
What Is Maternal Serum Screening?
Maternal serum screening can identify pregnant women who are at an increased risk for having a baby with certain birth defects. When a woman reaches a certain point in her pregnancy, a simple blood test can determine her risk of having a baby with an open neural tube defect, Down syndrome, or trisomy 18.

What Is First Trimester Serum Screening?
First trimester serum screening is a test that can determine whether your baby is at increased risk for Down syndrome and trisomy 18. The test is performed between 10 and 13 weeks of pregnancy and requires a blood test and an ultrasound exam. The blood test measures three proteins normally found in a pregnant woman's blood: (1) pregnancy-associated plasma protein-A (PAPP-A), (2) human chorionic gonadotropin (hCG), and (3) dimeric inhibin A (DIA). The ultrasound exam is performed by a credentialed specialist to measure the amount of fluid in the back of the baby's neck (nuchal translucency, or NT*). It is normal for all developing babies to have some fluid in the back of the neck, but larger amounts of fluid may be associated with an increased risk for Down syndrome and trisomy 18. The levels of PAPP-A, hCG, DIA, and the NT measurement are combined with information about you, such as your age and weight, to determine whether your baby is at risk of having Down syndrome or trisomy 18. First trimester screening provides an earlier screening option with improved detection of Down syndrome and trisomy 18 when compared to second trimester maternal serum screening tests.

* The NT should be performed by a health care professional credentialed by the Fetal Medicine Foundation or other equivalent entity.

Remember
First trimester serum screening is used to help your doctor identify pregnancies that may be at increased risk for Down syndrome or trisomy 18. It does not diagnose birth defects but helps identify those women who may benefit from additional testing.

References