

Maternal serum screening

What is maternal serum screening (MSS)?

MSS is a screening test that can tell you if there is an increased chance for Down syndrome, trisomy 18, or open spina bifida in the pregnancy. MSS is an optional test offered to all pregnant women between 15 and 23 weeks of pregnancy.

What is Down syndrome?

Down syndrome is the most common genetic condition seen in newborns. It is sometimes called trisomy 21. Babies with Down syndrome have intellectual disabilities and differences in their facial features. Babies with Down syndrome may also have problems with their heart and other organs. Down syndrome typically happens by chance and does not run in families.

What is Trisomy 18?

Trisomy 18 is another genetic condition sometimes called Edwards syndrome. Trisomy 18 is less common than Down syndrome. Babies with trisomy 18 usually have severe birth defects and often do not live very long after birth. Many pregnancies with trisomy 18 result in miscarriage or stillbirth. Like Down syndrome, trisomy 18 typically happens by chance and does not run in families.

What is open spina bifida?

Spina bifida is a condition caused by the incomplete closure of the spine and back. The opening in the back means that the nerves of the spinal cord are outside of the body. This can lead to nerve damage. Children with spina bifida typically have problems with walking. The opening in the back is closed with surgery either before or after the baby is born.

How is MSS done?

If you are interested in MSS, a sample of your blood will be taken between weeks 15 and 23 of pregnancy. The results of your blood test are combined with your age and other information about the pregnancy to determine the chance for Down syndrome, trisomy 18, or open spina bifida in the pregnancy. It takes about 5 days to get the results of the test. Your genetic counselor or primary healthcare provider will contact you when your results are available.

Why is a blood test done? The blood test measures proteins in the mother's blood which are made by the growing pregnancy. If levels of these proteins are different than the average pregnancy, it can mean there is an increased chance for Down syndrome, trisomy 18, or spina bifida.

How will the results of my screening come back?

The results will either be reported as screen positive or screen negative.

What if my test results are screen negative?

A screen negative result means that your risk for having a baby with Down syndrome, trisomy 18, or open spina bifida are below our cut-off. This does not completely rule-out the possibility of these conditions in the pregnancy. However, 80% of pregnancies with Down syndrome, 60% of pregnancies with trisomy 18, and 80% of pregnancies with open spina bifida will be detected by this screening test.

What if my test results are screen positive?

A screen positive result means that the chance for having a baby with Down syndrome, trisomy 18, or spina bifida is above our cut-off. It is important to remember that a screen positive result **does not** mean that your

pregnancy has any of these conditions. If your test results are screen positive, you will be scheduled for genetic counseling to review these results. You will also be offered further testing, such as amniocentesis, NIPT, or ultrasound.

What is an amniocentesis?

An amniocentesis is a test used to diagnose conditions such as Down syndrome, trisomy 18, and spina bifida. An amniocentesis is done by taking a small sample of amniotic fluid. Amniotic fluid is the liquid that surrounds the growing pregnancy. Amniocentesis has a risk for pregnancy complications or miscarriage. This risk is 1 in 300 to 1 in 500 (0.2 – 0.3%).

What is NIPT?

NIPT, or non-invasive prenatal testing, is a blood test that measures DNA that comes from the pregnancy and is found in the mother's blood. At this time, NIPT is only being offered to women who have an increased chance of having a pregnancy with Down syndrome, trisomy 18, or trisomy 13.