

Second trimester maternal serum screening

(Also called quad screen, triple screen, or AFP test)

What is maternal serum screening?

Maternal serum screening is an optional test to determine if a pregnancy has an increased chance for Down syndrome, trisomy 18, or open spina bifida. These conditions usually happen by chance, do not run in families, and do not happen because of anything either parent does or does not do.

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 difficulty learning and different facial features. Babies with Down syndrome may also have problems with their heart and other organs.

What is trisomy 18?

Trisomy 18 is less common than Down syndrome. Babies with trisomy 18 usually have multiple birth defects and often do not live very long after birth. Many pregnancies with trisomy 18 result in pregnancy loss.

What is open spina bifida?

Spina bifida occurs early in pregnancy when the spinal column and back do not close all of the way. Children with spina bifida typically have problems with walking. The opening in the back is closed with surgery either before or after baby is born.

How is maternal serum screening done?

Maternal serum screening is a blood test that is usually done between weeks 15 and 20 of pregnancy. The results of the blood test will tell you the chance that there is Down syndrome, trisomy 18, or open spina bifida in the pregnancy. Results take about 5 days to come back.

How will the results of my screening come back?

The results will either be reported as "screen negative" (low chance) or "screen positive" (high chance).

• A "screen negative" (low chance) result does not rule-out these conditions in the pregnancy. However, this test will pick up 60% of pregnancies with trisomy 18 and 80% of pregnancies with open spina bifida and Down syndrome.



This information remains the intellectual property of the University of North Carolina at Chapel Hill School of Medicine. This information cannot be reproduced in whole or in part without the expressed written permission of the school.

Updated by Reproductive Genetics 10/12/18 Approved by NC Women's Hospital Patient Education Steering Committee, April 16, 2019



• A "screen positive" (high chance) result *does not* mean your pregnancy has Down syndrome or another health problem. In this case, we would offer further testing that can get us closer to a yes or no answer.

Is maternal serum screening covered by insurance?

Maternal serum screening is covered by many insurance companies. The CPT codes (82105, 82677, 84702, and 86336) can be provided to your insurance company to check on coverage.

What other testing options are available?

- <u>First trimester screening</u>. A first trimester screen is a test that combines blood work and ultrasound and results are very similar to maternal serum screening. First trimester screening is done earlier in pregnancy. It is usually done between 11-13 weeks gestation.
- <u>CVS and amniocentesis</u> can test for Down syndrome and trisomy 18 with more accuracy. They are considered diagnostic. They can also test for more conditions. These tests involve taking a small sample from the placenta (CVS) or a small sample of the amniotic fluid (amniocentesis). Both CVS and amniocentesis have small risks for pregnancy complications or miscarriage.
- <u>Cell-free DNA screening</u> (cfDNA) or non-invasive prenatal testing (NIPT). This is a different blood test that screens for Down syndrome and trisomy 18. It has been shown to be more accurate at detecting Down syndrome than maternal serum screening (MSS). Insurance often does not cover cfDNA or NIPT in women unless they are over 35 years old or otherwise at high risk.
- <u>A targeted ultrasound</u> in the second trimester can also detect most pregnancies with open spina bifida.

How do I schedule maternal serum screening?

Tell your healthcare provider you are interested in MSS. It can be performed at your provider's office.

What if I have more questions?

If you have more questions or are not sure if you want screening, you can request an appointment to meet with a genetic counselor early in your pregnancy to review all of your screening and testing options.



This information remains the intellectual property of the University of North Carolina at Chapel Hill School of Medicine. This information cannot be reproduced in whole or in part without the expressed written permission of the school.

Updated by Reproductive Genetics 10/12/18 Approved by NC Women's Hospital Patient Education Steering Committee, April 16, 2019