

## New Options for Maternal Serum Screening for Birth Defects

Maternal serum screening is a way to determine if you are at a high or low risk for a baby with a specific birth defect such as Down syndrome, trisomy 18, or an open spine defect. It can also help determine if you are at risk for pregnancy complications from the way your placenta (afterbirth) functions. Today, there are several types of tests that can be performed at different stages of pregnancy to see if you are at increased risk for one of these disorders.

The test is performed by drawing one tube of your blood and performing an ultrasound of your pregnancy. It does not place your pregnancy at risk for miscarriage. Testing is voluntary and no special preparation is required.

This brochure is designed to give you information about the different tests that are available so that you can make the decision that is right for you and your family.

### 1. What types of testing are available to me?

There are three types of tests available:

1. First trimester screening
2. Second trimester (also called "quad") screening
3. Combined, Integrated screening performed in both the first and second trimester but gives a single result after both tests are completed.

### 2. How are these three tests different?

- a. **First trimester screening** is a blood test performed after an ultrasound in which your pregnancy is dated and the nuchal translucency (NT), or fluid measurement on the back of the fetus, is measured. This test is performed between 10 and 13 weeks of pregnancy.

The value of the markers found in your blood plus the ultrasound NT measurement, estimate the risk of carrying a pregnancy with Down syndrome. This early test can only test for your risk for Down syndrome.

- b. **Second trimester (quad) screening** is a test performed between 15-22 weeks gestation in which blood is obtained and a risk for Down syndrome, as well as for open fetal defects and trisomy 18 is calculated. Second trimester screening can also be a way to evaluate your pregnancy for placental abnormalities. This test is called "quad" screening, since four markers found in the mother's blood are evaluated. Some physicians will perform triple marker screening; quad screening is a little better at detecting Down syndrome. . Both test for the same problems in pregnancy including Down syndrome, placental abnormalities, trisomy 18, and open fetal defects. An

ultrasound does not have to be performed before this test is drawn, but accurate determination of the duration of your pregnancy by ultrasound makes the test more accurate.

- c. **Combined, integrated screening** is a combination of both first and second trimester screening. Blood is drawn between 10-13 weeks when a special ultrasound is performed , just like for first trimester screening. Then, blood is drawn again between 15-22 weeks. . As with second trimester screening, this combined test can give you information not only for Down syndrome but for trisomy 18, placental disorders, and open fetal defects. It is the best test available to test for Down syndrome in maternal blood.
  - i. Results are given to you when your second set of blood work is performed. If you choose the combined test but fail to have your second set of blood markers drawn, a first trimester screening result can be given to you based on your first blood test.

### 3. Why are so many tests available?

Patients should be given a choice, based on their personal desires and their gestational age, to choose the test that is right for them.

### 4. What kind of result will I be given?

The tests calculate a chance, or estimate of your risk of having an affected pregnancy. If your results place you at an increased risk for one of these disorders, this is also called a “screen positive” result.

- a. For **first trimester screening**, only a risk for Down syndrome is given. If you wish to be screened for open fetal defects, a second trimester ultrasound will be suggested.
- b. For **second trimester (quad) screening or combined, integrated screening**, risks for Down syndrome, trisomy 18 and open fetal defects will be given, as well as a risk for later complications caused by placental (afterbirth) complications.

### 5. What does a screen positive result for Down syndrome mean?

It means that you are in a high risk group to have a pregnancy with Down syndrome, and would be offered further testing. Most women with a screen positive test result do not have a pregnancy with Down syndrome.

- a. For **first trimester screening**, the chance that you will have a screen positive result is about 5 out of every 100 women.
- b. For **quad or second trimester screening** the chance of a screen positive result is also about 5 out of every 100 women.
- c. For **combined, integrated screening**, the chance of a screen positive result is 1 or 2 out of every 100 women.

**6. Do these tests detect all cases of Down syndrome?**

No, but they detect most of them

- a. For **first trimester screening**, they detect 85-90% of mothers carrying Down syndrome pregnancies.
- b. For **second trimester (quad) screening**, they detect about 75% of mothers carrying a Down syndrome pregnancy, 85% of open spine defects, and 60% of trisomy 18.
- c. For **combined, integrated screening**, they detect 90% of Down syndrome, 90% of open spine defects, and about 60% of trisomy 18.

**7. What can be offered if I have a screen positive test?**

You can be referred for genetic counseling where your risks will be addressed and information regarding testing your pregnancy can be given.

- a. **First trimester screening:** patients can have the option of an early test called chorionic villous sampling (CVS) in which a small piece of placental tissue can be removed and tested for Down syndrome and other common chromosomal disorders. It is usually performed between 10 and 12 weeks of pregnancy. A small (about 1%) risk of miscarriage is associated with CVS. The advantage is that fetal chromosome results would be available between 12 and 14 weeks of pregnancy.
- b. **Combined, integrated screening, and second trimester (quad) screening:** patients will have the option of genetic counseling, as well as a detailed ultrasound evaluation of their pregnancy and the option for amniocentesis to test the pregnancy. Amniocentesis is a test in which a small amount of fluid is removed from the sac surrounding the baby. It is usually performed between 15-20 weeks of pregnancy. The risk of miscarriage is very low at 0.5% (1/200 pregnancies or less).

**8. What is Down syndrome?**

Down syndrome is the most common form of mental retardation caused by extra chromosomal material in the developing baby. It is usually caused by an extra chromosome 21, and is therefore sometimes referred to as trisomy 21. Although there is a strong association between older age mothers and the chance of fetal Down syndrome, about 80% of Down syndrome pregnancies occur to women younger than age 35 years. Screening is designed to give mothers options to evaluate their pregnancies.

Down syndrome children also have a 30-40% risk of congenital heart disease, and also have a risk for intestinal problems, childhood leukemias, and Alzheimer's disease.

## **9. What are open fetal defects?**

The most common open defects detected by second trimester (quad or combined) screening are open spine defects and open abdominal wall defects. One of the proteins that is measured in second trimester screening with the quad and combined test, is maternal serum alpha fetoprotein (MSAFP). This protein is made almost exclusively by the fetus. If there is an open defect, more of this protein is found in the mother's blood. Screening can detect about 85% of open spine defects, and about 75% of open abdominal wall defects.

Open spine defects (spina bifida) means that the skin and bone covering the spinal cord has a part missing, leaving the nerves exposed. This occurs in about 1 out of every 1,000 pregnancies in the United States, and most occur in families without a prior family history. The severity of spina bifida is related to the size and location of the bony defect, but usually causes some lower limb paralysis and problems with bowel and bladder control.

Open abdominal wall defects can occur in different forms, but usually means that the fetal intestines are outside of the fetal body. This means that the baby will need surgery to fix the defect and can also have associated complications.

Sometimes, the MSAFP level will be elevated and the fetus will appear normally developed by ultrasound. Sometimes this demonstrates a risk for a placental problem that could cause the baby to be small or have other problems. These problems cannot be detected by the first trimester blood test.

## **10. What is trisomy 18?**

Trisomy 18 is another disorder that is caused by an extra chromosome, chromosome 18. This disorder is much more severe than Down syndrome. The affected children are much more severely disabled and the majority do not survive the first year of life. Second trimester (combined integrated or quad) screening can help detect this disorder, which may not have birth defects that can be identified with ultrasound. Trisomy 18 also cannot be detected by the first trimester test.

## **11. Do these tests make certain that my child does not have a birth defect?**

No test can guarantee a healthy outcome, but these tests can help place you into a low or high risk group for one of these disorders. If we find that your pregnancy does have a disorder, we will work with you and your family to find the right care and right pregnancy options for you.