

Increased Risk for Down Syndrome

What Now?

WHAT IS MATERNAL SERUM SCREENING?

Maternal serum screening is a blood test that measures the amounts of several different chemicals in a pregnant woman's blood. These chemicals are only present during pregnancy and are made by the fetus or placenta.

Why does the test show a high risk of Down syndrome?

By testing large groups of pregnant women, scientists have determined how much of each chemical is usually present during each week of pregnancy. The average amounts of the chemicals are different in pregnancies involving Down syndrome. In these pregnancies, some of the levels tend to be lower than average, while others are higher than average.

What is Down syndrome?

Down syndrome is a type of mental retardation caused by an extra chromosome within the fetus' cells. Normally we have 46 chromosomes within each of the cells of our bodies. Children with Down syndrome have 47. The extra chromosome causes birth defects and delayed mental and physical development.

People with Down syndrome vary in their abilities. Most learn to walk, talk and become toilet trained, although at a later-than-average age. Older children with Down syndrome may need special classes in school. Adults may need to work under special supervision or need assistance in everyday living.

How high is my risk?

The laboratory uses a statistical formula to compare each patient's results with those seen in affected and unaffected pregnancies then calculates the chance for Down syndrome. Your health care provider should have the laboratory report stating your specific risk. If the risk is higher than the risk at maternal age 35, the result is considered "increased risk." Further testing is then offered.

What further tests are offered?

If you have not already had an ultrasound or sonogram, this test should be done. Ultrasound uses sound waves to create a picture of the fetus. Sometimes the ultrasound will show that the fetus is much smaller than expected, indicating that the pregnancy is not as far along as originally thought. Since the amounts of chemicals in the mom's blood change during pregnancy, the blood test would need to be recalculated to account for the change in dating. The new result is often normal.

You may wish to have a genetic amniocentesis.

Amniocentesis is a test in which the physician withdraws a small amount of fluid from the womb. This fluid contains cells from the fetus. It can be sent to a laboratory where the cells can be studied for their chromosome content. In Down syndrome, there is an extra number 21 chromosome. Amniocentesis results take between ten and 14 days and are greater than 99 percent accurate.

Isn't amniocentesis risky?

There is a small risk involved in amniocentesis. Occasionally, the test may lead to a miscarriage or infection. The chance of serious complications from amniocentesis is approximately one in 500 or 0.2 percent.

Can Down syndrome be ruled out without amniocentesis?

Many fetuses with Down syndrome have physical features that can be seen with a comprehensive ultrasound. Features that are more common in fetuses with Down syndrome include a heart defect, shortened thighbones or an increased thickness of skin at the back of the neck, among others. Of all fetuses with Down syndrome, 50 to 80 percent will have one or more of these findings. However, some fetuses with Down syndrome appear normal on ultrasound. So, while high-resolution ultrasound can be reassuring, amniocentesis is still the most accurate way to detect Down syndrome.

WHAT IF I HAVE MORE QUESTIONS?

If you still have questions, please feel free to contact our genetic counselor at (515) 643-6888 or toll free at (877) 415-7447.



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