

Increased risk for Trisomy 18

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 Trisomy 18?

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 Trisomy 18. In these pregnancies, the amounts of all of the chemicals are lower, on average, than what is usually seen in the general population.

What is Trisomy 18?

Trisomy 18 is also known as Edwards syndrome. Most babies with this syndrome miscarry or live only a short while. In rare cases, a baby with Trisomy 18 may survive a few years. These children have severe developmental delay and intellectual disability. While they do continue to make developmental progress, most never learn to walk or talk.

Trisomy 18 is caused by an extra chromosome within the fetus' cells. normally we have 46 chromosomes in each of the cells of our bodies. Children with Trisomy 18 have 47. The extra chromosome causes abnormalities in fetal development, resulting in birth defects and delayed mental and physical development.

How high is my risk?

Most laboratories calculate a specific risk for Trisomy 18 for each patient. If your maternal serum screen was performed at one of these laboratories, your care provider should be able to tell you the exact risk. Some laboratories use only a "cutoff" to determine which patients are at increased risk for Trisomy 18. That is, if amounts of each of the three chemicals fall below a certain level, the patient is considered high risk. one study found that about seven percent of these patients had a baby with Trisomy 18. In Iowa, only around two percent of patients with an increased maternal serum screen risk for Trisomy 18 actually have a baby with the disorder.

In a more positive light, this means that 93 to 98 percent of women who screen positive for Trisomy 18 are not carrying an affected child. Therefore, further testing is always offered in this situation.

What further tests are offered?

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 Trisomy 18, there is an extra number 18 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 a serious complication from amniocentesis is approximately one in 500 or 0.2 percent.

TRISOMY 18

Can Trisomy 18 be ruled out without an amniocentesis?

Many fetuses with Trisomy 18 have physical features that can be seen with a comprehensive ultrasound. Features that are more common in fetuses with Trisomy 18 include a heart or kidney defect, a characteristic clenched fist, certain abnormalities within the brain and a characteristic appearance of the heel. Some fetuses with Trisomy 18 appear normal on ultrasound. While high-resolution ultrasound can be reassuring, amniocentesis is still the most accurate way to rule out the possibility of Trisomy 18.

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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