

# MSS – SCREEN POSITIVE FOR TRISOMY 18

NORTHWESTERN  
ONTARIO REGIONAL  
**GENETICS**  
PROGRAM



## WHAT IS MSS?

MSS is a blood test available to all pregnant women in Ontario. MSS measures four different hormones that have been produced by the baby's liver and the placenta. These hormones pass into the mother's blood stream in small amounts. MSS measures these amounts and compares them to the average for women in the same week of pregnancy. From this information, the chance of a baby having trisomy 18, Down syndrome, or open neural tube defect (spina bifida) is calculated.

## WHAT DOES SCREEN POSITIVE FOR TRISOMY 18 MEAN?

A screen positive result means the chance is higher than usual that your baby might have trisomy 18. It is important to remember that the MSS is a screening tool and not a diagnostic test. When the MSS is screen positive for trisomy 18, in most cases the baby is healthy and is not affected. Additional testing is needed to tell whether or not a baby actually has trisomy 18.

## WHAT IS TRISOMY 18?

Chromosomes are the packages of our genetic information. The genetic information is responsible for our body structure and function. Chromosomes are passed from parents to their children through the mother's egg and the father's sperm. The usual number of chromosomes is 46.

A baby with trisomy 18 has 47 chromosomes (an extra number 18 chromosome). The extra chromosome causes the variety of problems seen in a

baby with this genetic difference.

Babies with trisomy 18 will have a severe mental handicap. Often there are physical problems of the brain, heart, kidneys, and other organs. It is unusual for babies with trisomy 18 to survive beyond their first birthday.

## WHAT DOES YOUR RISK NUMBER MEAN?

You can ask your health care provider to give you your risk figure that is written down on the report. For example, the result might be 1:25 which means that your chance of delivering a baby with trisomy 18 is 1 in 25. Another way to look at this is that your chance of your baby **NOT** being affected with this chromosome change is 24 out of 25.

## YOUR RESULT IS SCREEN POSITIVE FOR TRISOMY 18; WHAT HAPPENS NEXT?

Your health care provider will verify information about the pregnancy and obtain any pieces of information that are missing.

*Further testing is available however it is your choice whether you wish to proceed.*

A detailed ultrasound will be offered to you to look closely at the baby's physical development. Amniocentesis, which is the diagnostic test that is used to confirm the presence of trisomy 18 in the baby, will be offered. Genetic counselling is available to you to discuss the implications of the screen result and to explain the testing options.

## WHAT IS AMNIOCENTESIS?

Amniocentesis is a test usually done to obtain a sample of amniotic

fluid surrounding the baby. The cells in the amniotic fluid will provide information about the chromosomes of the developing baby.

It involves the insertion of a needle through the woman's abdomen into the uterus and the removal of a small amount of fluid. Ultrasound is used to locate the position of the placenta and the baby, and to decide on the best "pocket" of the amniotic fluid to sample. There is a 1/200 or 0.5% chance of miscarriage associated with this procedure. The results are available in about two weeks.

## WHAT IS ULTRASOUND?

Sound waves are used to create a picture of the developing baby. This allows the specialists to look at the baby on a television monitor. Ultrasound is considered to be safe for the developing baby and mother.

It is important to be aware that every pregnant couple in the general population has a 2-3% risk of having a baby with a birth defect. While normal results of prenatal testing are reassuring, they do not guarantee the birth of a healthy baby.

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