What does a “screen positive” result tell me?

A “screen positive” means that there is a higher than usual chance that your baby might have Down syndrome, trisomy 18, SLOS, or an open neural tube or a ventral wall defect. However, most of the pregnant women who get a “screen positive” result go on to have a healthy baby.

A “screen positive” result may be due to confusion about how far along you are in your pregnancy. Once we have precise information about this, the result often becomes normal.

Only further tests can show if the baby really has a problem. Your doctor or midwife would tell you more about your test result and what it means. If you still have questions, you can get more information from the Manitoba MSS Clinical Coordinator at (204) 787-2097.

What further tests are available?

If the result is screen positive, a number of other tests are available to see if the baby does have a problem. You would be told which of these would be helpful. However, the decision to have any more tests carried out is up to you.

The first step is to check the baby’s age with ultrasound, if this has not already been done. An accurate estimate of how far along you are is essential to get an accurate MSS result.

If there is still an increased chance of your baby having Down syndrome, trisomy 18 or SLOS after the ultrasound, you will be offered an amniocentesis to test for these conditions.

If there is an increased chance that your baby has a neural tube or a ventral wall defect, you will be offered a very detailed ultrasound to look at the baby’s head, back and abdomen (tummy). Most often the additional tests will find that your baby does not have Down syndrome, trisomy 18, SLOS, a neural tube or ventral wall defect.

What if your baby really does have a problem?

You would be told what the problem is and what this would mean for the baby. Support would be available as you decide what to do next. Unfortunately, there is no cure for many of the problems that MSS identifies. Some babies with ventral wall defects are successfully treated with surgery, as are some with mild neural tube defects. There is no cure for Down syndrome, trisomy 18 or SLOS. Whether to continue or end the pregnancy is your choice. What you do is up to you.

These are difficult and personal decisions and there is no single answer that is right for every woman. Whatever you choose to do, the care you receive will not be affected in any way.

How can I get the MSS test?

MSS is available through your doctor or midwife. Make sure you talk about MSS with them early in your pregnancy.

When should I have the test?

When this test is done is very important. The best time for MSS is when you are 16 weeks pregnant, that is, 16 weeks after the first day of your last period. The complete test can not be done before 15 weeks or after 20 weeks and 6 days. The neural tube defect part of the test can be done up to 23 weeks and 6 days.

Who will get my results?

In about 1 week, your results will be sent to your doctor or midwife who will explain them to you.

Your results will also be kept in a database that permits us to keep track of the samples and helps us to evaluate the programme. We will record whether your pregnancy ended normally or if there were any problems with your or your baby’s health. This can tell us whether the screening programme is working properly.

The database may also be used in research projects studying fetal and maternal health problems. No information will be released that could identify you or your baby, and all such studies must be approved beforehand by a Research Ethics Board.

Maternal serum screening in the future

As other methods of maternal serum screening are developed, the Manitoba Maternal Serum Screening Programme will evaluate whether they should be added to our programme. Screening practices may therefore change in the future.

Definitions:

Amniocentesis: This is a test where a needle is inserted through the mother’s abdomen into the uterus (womb) and a sample of fluid from around the baby is taken. This test can check for chromosome problems such as Down syndrome or Trisomy 18 or other conditions such as SLOS.

Chromosomes: The carriers of genetic information. Chromosomes come in 23 pairs, one of each pair coming from the mother and one from the father. Down syndrome and trisomy 18 are due to extra copies of chromosomes.

Down syndrome: This is the most common chromosomal cause of mental disability. Children with Down syndrome have three copies of chromosome 21 instead of the normal two. They have a characteristic facial appearance and may have certain birth defects including heart problems. This condition occurs in about 1 in 700 babies and is more common in babies born to older mothers.

Neural tube defects: These are birth defects where the spinal cord or brain fails to form properly. They usually cause problems with bladder and bowel control or difficulties with walking due to abnormal spinal cord development (spina bifida) or a lethal brain malformation (anecephaly). About one in every 1200 babies is born with a neural tube defect.

Smith-Lemli-Opitz syndrome (SLOS): A genetic condition that causes birth defects and mental retardation.

Trisomy 18: Babies with Trisomy 18 have three copies of chromosome 18 instead of the normal two. About 1 in 4000 babies is born with Trisomy 18. It causes very severe birth defects and mental retardation.

Ventral Wall Defects: If the ventral or abdominal (tummy) wall is not properly formed, the intestines and other organs may protrude, most commonly through an enlarged umbilicus (belly button). Sometimes these problems can be corrected after birth with no long-term effects on the baby; in other cases, especially when there are other malformations, there may be serious complications or early death. About 1 in 1000 babies is born with a ventral wall defect.

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Ventral Wall Defects: If the ventral or abdominal (tummy) wall is not properly formed, the intestines and other organs may protrude, most commonly through an enlarged umbilicus–belly button (omphalocoele) or through a defect in the skin on either side of the umbilicus (gastroschisis). Sometimes these problems can be corrected after birth with no long-term effects on the baby; in other cases, especially when there are other malformations, there may be serious complications or early death. About 1 in 1000 babies is born with a ventral wall defect.
Remember, with the Maternal Serum Screen:

- Most tests come back normal.
- If your test comes back positive, it does not mean your baby has a problem. It only means that additional tests may be offered.

If you require further information, please feel free to call:

Coordinator
Manitoba Maternal Serum Screening Programme
Health Sciences Centre
Winnipeg, Manitoba R3A 1R9
Phone: (204) 787-2097 or call collect.

The Manitoba Maternal Serum Screen is a test that:

- will give you important information about your baby
- involves only a blood sample taken from your arm
- is best done around 16 weeks
- is voluntary

IT’S YOUR CHOICE

What is Maternal Serum Screening?

Maternal Serum Screening (MSS) is a blood test available to pregnant women in Manitoba as part of their prenatal care. This test can tell you about your chance of having a baby with a problem such as Down syndrome, trisomy 18 or Smith-Lemli-Opitz syndrome (SLOS). It can also detect openings in the baby’s spine or head (known as neural tube defects) or in the abdomen or tummy (called ventral wall defects). Most of these problems occur in families where there is no previous history of babies born with such problems. The MSS helps identify families who may be at increased risk for these defects, and offers them further testing.

MSS does not tell for certain if your baby is healthy, but it can tell you if there is a higher or lower chance of your baby having a problem. Some women find having MSS is reassuring. Others feel that they do not need, or want, this information. It is your choice whether to have MSS and it will only be done if you want it. Your choice about having this test will not affect the care you receive.

How is the test done?

A small amount of blood is taken from your arm. Maternal serum screening is best done around 16 weeks of pregnancy.

What can this test tell me?

The test looks at various natural substances in the mother’s blood that are made by the baby and placenta. By taking a sample of your blood, we can measure the level of each of these substances. This will tell us if your baby appears to be developing normally.

If the levels are what is expected, your test is called "screen negative", and the chance of your baby having Down syndrome, trisomy 18, SLOS, a neural tube or ventral wall defect is low. About 90 out of 100 pregnant women who have this test get normal results and no further tests are suggested. Although most women who get a normal result have a healthy baby, this is not a guarantee. A screening test cannot identify all mothers who will have a baby with a problem.

When a baby has Down syndrome, trisomy 18, SLOS, an open neural tube or ventral wall defect, the levels of these substances are usually different.