Ask for the Maternal Serum Screening Counsellor at one of the following genetic centre locations

Hamilton
Prenatal Diagnosis
Hamilton Health Sciences
McMaster Campus
1200 Main Street.
Hamilton, ON L8N 3Z5
(905) 521-2100 ext 12649

Kingston
Division of Genetics and Molecular Medicine
Kingston General Hospital
20 Burue Street.
Kingston K7L 3J6
(613) 533-6310

London
Medical Genetics Program of S.W. Ont.
London Health Sciences Centre
800 Commissioners Road East.
London N6C 2V5
(519) 685-8140

Mississauga
Genetics Clinic
Credit Valley Hospital
2200 Eglington Ave West.
Mississauga L5M 2N1
(905) 813-4104

North Bay
North Bay and District Health Unit
Genetics Program
61 Commercial Street
North Bay ON P1B 4E7
(705) 474-1400

North York
North York General Hospital
4001 Leslie St. 3rd floor, SE Wing
Toronto, ON M2K 1E1
(416) 756-0055

Orillia
Orillia Soldiers Memorial Hospital
170 Colborne Street West.
Orillia L3V 2Z3
(705) 327-9154

Oshawa
Genetic Services
Lakehead Health Corporation
1 Hospital Court
Oshawa, ON L1G 2B9
(905) 435-2733 Fax (905) 721-4757

Ottawa
Eastern Ontario Regional Genetics Program
401 Smyth Road.
Ottawa K1H 8L1
(613) 737-7600 ext 2138

Peterborough
Genetics Program
Peterborough County-City Health Unit
10 Hospital Drive
Peterborough, ON K9J 8M1
(705) 743-1000

Sault Ste. Marie
Algoma District Health Unit
Civic Centre, 99 Foerster Drive.
Sault Ste. Marie P6A 5K6
(705) 541-7143

Scarborough
Rouge Valley Health System-Centenary Site
Genetics Clinic
2867 Ellesmere Road.
Scarborough M1E 4B9
(416) 281-7425

Sudbury
Sudbury Regional Hospital
Genetics Counselling Services
4 Boland Ave.
Sudbury, ON P3E 1X7
(705) 675-4786

Thunder Bay
Thunder Bay District Health Unit
999 Balmeral Street.
Thunder Bay, ON P7B 6E7
(807) 625-5900 ext 5924

Timmins
Porcupine Health Unit
169 Pine Street South,
Timmins P4N 8B7
(705) 267-1181

Toronto
Prenatal Diagnosis Program
Mount Sinai Hospital
700 University Avenue - Hydro Building.
Toronto ON M5G 1Z5
(416) 586-4946

Windsor
Windser-Essex County Health Unit
Genetics Services
1005 Quelleme Avenue,
Windsor N9A 4J8
(519) 258-2146

York
Genetics Clinic
York Central Hospital
10 Trench Street,
Richmond Hill L4C 4Z3
(905) 883-2097

For additional information see http://www.hrh.on.ca genetics/MSS
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Every pregnant woman wants to have a healthy baby. Although most babies are healthy, it is normal to worry about your baby.

This pamphlet is about one way you can find out your chance of having a baby with any one of three specific problems. Maternal Serum Screening (MSS) can tell you the chance of your baby having Down syndrome, trisomy 18 or an open neural tube defect. Maternal Serum Screening is your choice. Only you can decide if you want it.

MSS is best around 16 weeks pregnant. Talk to your doctor or midwife about MSS before this time.

What is MSS?

MSS is a blood test available to pregnant women as part of their prenatal care. MSS can tell you about your chance of having a baby with Down syndrome, trisomy 18 or an open neural tube defect.

Some women find the results of MSS to be reassuring. Other women feel they do not need or want this information. It is your choice whether to have MSS. It will be done only if you want it. Your choice about having this test will not affect the care you receive.

How is MSS done?

If you decide to have MSS, a small amount of blood is taken from your arm. The amount of certain hormones will be measured in the blood. These hormones are made by the fetus (the developing baby) and the placenta during pregnancy and are found in every pregnant woman’s blood.

When a baby has Down syndrome, trisomy 18 or an open neural tube defect, the amount of these hormones might be different from usual. The result will tell you the chance of your baby having Down syndrome, trisomy 18 or an open neural tube defect.

It takes about one week to get the MSS result.

What can the MSS result tell me?

MSS can tell you the chance of your baby having one of these problems. If the results show that the levels of the hormones follow the usual pattern, the chance of your baby having Down syndrome, trisomy 18 or an open neural tube defect is low. This is called a screen negative result. If the level of these hormones does not follow the usual pattern, your baby has an increased chance of having Down syndrome, trisomy 18 or an open neural tube defect. This is called a screen positive result. Occasionally, an MSS result might lead to finding another type of problem for your baby. Your doctor or midwife would receive this result and discuss it with you.

What does a “screen negative” result mean?

A result is called screen negative when a pregnant woman’s blood has the usual amount of these hormones. This means the chance of her baby having Down syndrome, trisomy 18 or an open neural tube defect is low. Most times no further testing is needed.

About 90 out of 100 pregnant women have a screen negative result. Although most women with a screen negative result will have normal babies, MSS can miss a case of Down syndrome, Trisomy 18 or an open neural tube defect. As well, MSS does not find most other types of problems. So, a screen negative result does not guarantee a normal baby.

What does a “screen positive” result mean?

A screen positive result means the chance is higher than usual that your baby might have Down syndrome, trisomy 18 or an open neural tube defect. Most women with a screen positive result have healthy babies. Only further tests can show if the baby really has one of these problems. The chance of your baby having Down syndrome or an open neural tube defect is written on your result. (The chance for trisomy 18 is written on the report only if it is higher than the usual chance.) Talk to your doctor or midwife about your result.
What happens if the result is screen positive?

If your result is screen positive, only more testing can tell if your baby really has any one of these problems. Deciding what to do next can be difficult. Speak to your doctor or midwife about further testing. For additional information you can ask for a referral to your regional genetic counselling centre. Make sure all your questions are answered.

Do you want further tests?

When the result is screen positive, you can choose to have more testing to find out if the baby really has one of these problems. Each woman will be given choices about different tests, depending on her results.

If the result is screen positive, the first step is to check the age of the baby with an ultrasound, if this has not already been done. Knowing the age of the developing baby is important because the hormone levels change from week to week in pregnancy.

If the MSS result shows an increased chance of Down syndrome or trisomy 18, you will be offered amniocentesis. During amniocentesis, a small amount of fluid surrounding the baby is taken and tested for Down syndrome or trisomy 18. Amniocentesis can cause a miscarriage in about one out of 200 women.

If the MSS result shows an increased chance of an open neural tube defect, you will be offered an ultrasound. At the ultrasound, the head and back of the baby will be examined. However, an ultrasound cannot see all cases of open neural tube defects. If an open neural tube defect is not seen on the ultrasound, amniocentesis might be offered. During amniocentesis, a small amount of fluid surrounding the baby is taken. The fluid is tested for an open neural tube defect.

Most times the results of the ultrasound or amniocentesis will show that your baby does not have Down syndrome, trisomy 18 or an open neural tube defect.

What if the further tests show your baby really has Down syndrome, trisomy 18 or an open neural tube defect?

You will be given the information and support you need to make a decision about this pregnancy. Unfortunately, there is no cure for these problems. If a baby is found to have Down syndrome, trisomy 18 or an open neural tube defect, you can choose if you want to continue or to end the pregnancy. It is your choice.

How can I get MSS?

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

When do I get MSS?

Timing 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 menstrual period. It should not be done before 15 weeks and not after 20 weeks.

Analysis of pregnancy outcomes is part of the MSS quality assurance program. This information is confidential and is collected in such a way that neither a woman, nor her baby, can be identified. You will not be contacted.
Open Neural Tube Defects

Open neural tube defects happen when the brain or spine does not develop properly. There are two kinds of open neural tube defects - spina bifida and anencephaly.

Spina bifida is an opening in the bones around the spinal cord. Usually the opening is not covered by the skin. Spina bifida causes physical problems and sometimes mental disabilities. There is no way to predict how serious these physical problems will be or if the person will have mental disabilities. There is no cure for the problems, but often the problems of spina bifida can be helped. Many people with spina bifida lead productive lives.

When a baby has anencephaly, the brain and skull do not develop completely. A baby with anencephaly will die shortly after birth.

Between one and two babies out of 1,000 babies are born with a neural tube defect. The chance does not depend on the age of the mother. Talk with your doctor or midwife about your chance of having a baby with a neural tube defect.

Down syndrome

People with Down syndrome have physical and mental disabilities. They are more likely to have medical problems, such as heart problems. Each person with Down syndrome is different. Many have productive lives, but some have very limited physical and mental abilities. There is no way to predict how serious any of the disabilities will be. There is no cure for Down syndrome.

Any woman at any age can have a baby with Down syndrome. However, the chance increases with a woman’s age. For example, a woman who is 40 years old has a much higher chance of having a baby with Down syndrome than a woman who is 20. Talk with your doctor or midwife about your chance of having a baby with Down syndrome.

Trisomy 18

Each person with trisomy 18 has very serious physical and mental disabilities. Most infants with trisomy 18 die by one year of age. There is no cure for trisomy 18.

Trisomy 18 is not a common problem, but it can happen in any pregnancy. The chance of trisomy 18 increases with a woman’s age. For example, a woman who is 40 years old has a much higher chance of having a baby with trisomy 18 than a woman who is 20. Talk with your doctor or midwife about your chance of having a baby with trisomy 18.

MSS Flow Diagram

- discuss MSS with doctor or midwife before 16 weeks
  - blood taken for MSS best at 16 weeks
  - screen positive 
    - discuss further tests with doctor or midwife
      - ultrasound to check dates if not yet done
    - screen negative
      - no further testing (does not guarantee normal baby)

- Offer of amniocentesis
  - Down syndrome
    - 1/200 miscarriage chance
    - most don’t have Down syndrome
    - consider what to do if result is not normal
  - Trisomy 18
    - 1/200 miscarriage chance
    - most don’t have Trisomy 18
    - consider what to do if result is not normal
  - Open Neural Tube Defect
    - 1/200 miscarriage chance
    - most don’t have an open neural tube defect
    - consider what to do if result is not normal