

## PRENATAL SCREENING

Every pregnant woman hopes for a healthy baby. Although most babies are healthy, it's normal to wonder about your baby. This pamphlet describes a prenatal screening test called IPS (Integrated Prenatal Screening). A screening test does not tell for sure whether or not your baby is healthy. It can tell if you have a lower or higher chance than usual of having a baby with one of three problems: Down syndrome, trisomy 18 or an open neural tube defect. ***It's your choice whether you want to have IPS. Talk to your health care provider before deciding if IPS is right for you.***

## WHAT IS DOWN SYNDROME?

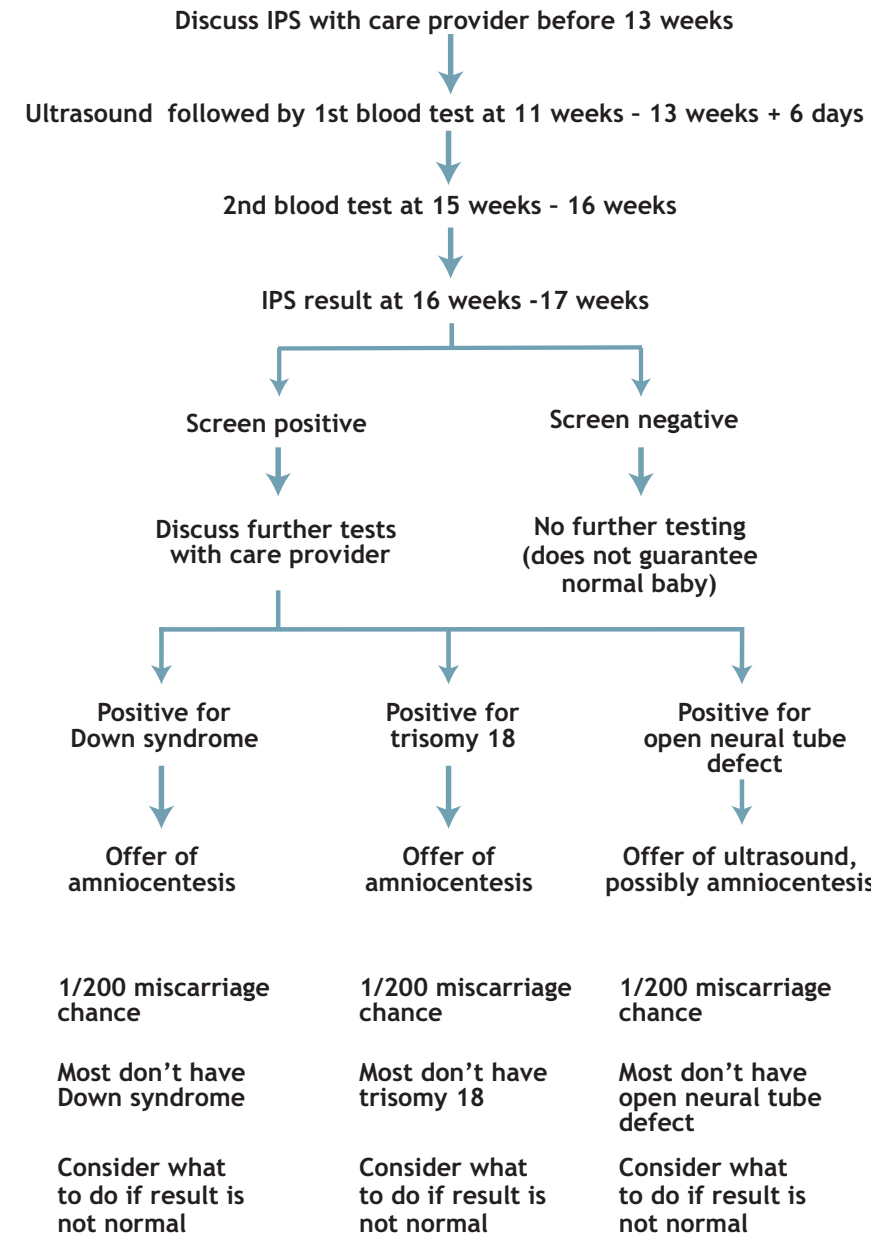
All people with Down syndrome have mental handicap and some have physical handicaps. They are also more likely to have medical problems. Each person with Down syndrome is different and there is no way to know how serious the mental handicap will be. There is no cure for Down syndrome but resources are available to help persons with Down syndrome and their families.

Any woman of 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 has a higher chance of having a baby with Down syndrome than a woman who is 20.

## WHAT IS TRISOMY 18?

Babies with trisomy 18 have serious physical and mental handicaps. Most do not survive beyond one year of age. The chance of having a baby with trisomy 18 also increases with a woman's age.

## IPS FLOW DIAGRAM



## ASK FOR THE SCREENING COUNSELLOR AT ONE OF THE FOLLOWING LOCATIONS

**Hamilton**  
Prenatal Diagnosis  
Hamilton Health Sciences  
1200 Main Street,  
Hamilton L8N 3Z5  
(905) 521-2100 ext 72649

**Kingston**  
Medical Genetics Unit  
Kingston General Hospital  
20 Barrie Street,  
Kingston K7L 3J6  
(613) 533-6310

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

**Mississauga**  
Genetics  
Credit Valley Hospital  
2200 Eglinton Ave West,  
Mississauga L5M 2N1  
(905) 813-4104

**North Bay**  
North Bay and District Health Unit  
681 Commercial Street,  
North Bay P1B 4E7  
(705) 474-1400

**North York**  
Department of Genetics  
4001 Leslie Street,  
Third floor, SE wing,  
Toronto M2K 1E1  
(416) 756-6345

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

**Oshawa**  
Genetic Services  
Lakeridge Health Corporation  
1 Hospital Court,  
Oshawa L1G 2B9  
(905) 433-2733

**Ottawa**  
Eastern Ontario Regional Genetics Program  
Children's Hospital of Eastern Ontario  
401 Smyth Road,  
Ottawa K1H 8L1  
(613) 737-7600 ext 3653

**Peterborough**  
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 Foster Drive,  
Sault Ste-Marie P6A 5X6  
(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  
Genetic Counselling Services  
4 Boland Avenue,  
Sudbury P3E 1X7  
(705) 675-4786

**Thunder Bay**  
Thunder Bay District Health Unit  
999 Balmoral Street,  
Thunder Bay 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  
Mt. Sinai Hospital  
700 University Avenue - Hydro Building,  
Toronto, ON M5G 1Z5  
(416) 586-4946

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

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

Are you pregnant?

Integrated Prenatal Screening (IPS)

it's your choice

## WHAT ARE OPEN NEURAL TUBE DEFECTS?

Open neural tube defects happen when the skull or spine does not develop properly early in pregnancy. 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. The opening can be anywhere along the spine and it's usually not covered by skin. Spina bifida causes physical handicap and sometimes also mental handicap. The amount of physical handicap depends on the size and location of the opening. It's not always possible to predict if there will be mental handicap. There is no cure for spina bifida, but often the problems caused by spina bifida can be helped by surgery and other treatments.

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

One to two babies out of every thousand born in Ontario have a neural tube defect. The chance of having a baby with a neural tube defect does not increase with a woman's age.

## CAN PRENATAL SCREENING FIND OTHER PROBLEMS?

Prenatal screening is not designed to find other problems. Occasionally the result of your prenatal screen can tell your health care provider about the possibility of other problems. If this happens, you will receive more information from your health care provider. In some situations, your health care provider will refer you for genetic counselling.

## WHAT IS INTEGRATED PRENATAL SCREENING (IPS)?

IPS is a prenatal screen that uses an early ultrasound and two blood tests to give your chance of having a baby with Down syndrome, trisomy 18 and an open neural tube defect in this pregnancy. IPS replaces MSS (Maternal Serum Screening) if you see your health care provider early in pregnancy.

## HOW IS IPS DONE?

You make an appointment with your health care provider as early as possible to discuss IPS.

If you decide to have IPS, your health care provider arranges an ultrasound between 11 weeks and 13 weeks + 6 days of pregnancy. The ultrasound is used to check the age of the baby and to measure the nuchal translucency (NT). The NT is the thickness of the skin at the back of the neck of the developing baby.

Next, you have a blood test to measure the level of a protein that's in the blood of every pregnant woman. This is also done between 11 weeks and 13 weeks + 6 days of pregnancy and usually after the ultrasound. It can even be done on the same day.

You have a second blood test at 15-20 weeks of pregnancy, ideally at 15-16 weeks for the earliest possible result.

***An IPS result is sent to your health care provider only after the ultrasound AND both blood tests are done.***

## WHAT DOES A 'SCREEN POSITIVE' RESULT MEAN?

About 4 in 100 women will have a 'screen positive' result. If your result is 'screen positive', the chance that your baby might have Down syndrome, trisomy 18 or an open neural tube defect is higher than usual. A 'screen positive' result can cause worry. ***However, most women with a screen positive result do not have a baby with Down syndrome, trisomy 18 or an open neural tube defect.***

## WHAT HAPPENS IF THE RESULT IS 'SCREEN POSITIVE'?

If your result is 'screen positive', deciding what to do next can be difficult. Your health care provider will discuss your result with you. You may also be referred to a genetic counsellor. Both can explain your result and tell you about your choices for more testing. Only more testing can tell you if your baby really has Down syndrome, trisomy 18 or an open neural tube defect.

## DO I WANT MORE TESTING?

If your result is 'screen positive', you can choose whether you want more testing to find out if the baby really has any one of these problems.

The tests offered depend upon your IPS result. If IPS shows an increased chance of Down syndrome or trisomy 18, you will be offered amniocentesis. During amniocentesis, a small amount of fluid is taken from the area around the baby and tested for Down syndrome and trisomy 18. Amniocentesis will cause a miscarriage in about one out of every 200 women who have it.

If IPS shows an increased chance of an open neural tube defect, you will be offered another ultrasound to look at the head and spine of the baby. There is no risk to you or your baby from ultrasound. Occasionally, amniocentesis is also offered to measure the level of a protein in the fluid around the baby. The results of these tests can tell you if the baby has an open neural tube defect.

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

## WHAT IF THE EXTRA TEST TELLS ME THAT THE BABY HAS DOWN SYNDROME, TRISOMY 18 OR A NEURAL TUBE DEFECT?

Your health care provider and/or genetic counsellor will discuss this with you in detail. Some people choose to continue their pregnancies, while others choose to end their pregnancies.

## WHAT DOES A 'SCREEN NEGATIVE' RESULT MEAN?

If the nuchal translucency measurement and the levels of the proteins in the blood are in the usual range, then the result is called 'screen negative'. Further testing is not offered.

About 96 in 100 women will have a screen negative result. Although most women with a screen negative result will have normal babies, ***IPS can miss a small number of babies with Down syndrome, trisomy 18 or an open neural tube defect.***

***If you have questions or are interested in having IPS, speak with your health care provider as early as possible in your pregnancy.***