



What is prenatal screening and testing?

There is a difference between screening and diagnostic testing.

Screening 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.

Diagnostic testing is done to find out if the baby really has any one of these problems.

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.



PRENATAL SCREENING AND TESTING OPTIONS



PRENATAL SCREENING AND TESTING

Every pregnant woman hopes for a healthy baby. Although most babies are healthy, it's normal to wonder about your baby. This pamphlet describes prenatal screening options as well as diagnostic testing options that may be available to you during your pregnancy. There is a difference between screening and diagnostic testing. Screening 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. Diagnostic testing is done to find out if the baby really has any one of these problems. It's your choice whether you want screening and/or testing during your pregnancy. Talk to your health care provider or a genetic nurse before deciding what is right for you.

WHAT IS DOWN SYNDROME?

Down syndrome is the most common chromosome change seen in newborn babies. Individuals with Down syndrome have some common physical characteristics, different degrees of mental handicap, and an increased risk for medical problems including congenital heart defects.

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.

WHAT ARE 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. It usually involves the skin and tissues around the spinal cord. This can result in a range of outcomes for the baby. Usually there is some weakness or paralysis of the legs. There may also be problems with bowel and bladder control as well as other medical problems such as hydrocephalus (an accumulation of fluid around the brain). In most cases

hydrocephalus can be treated, but the problem with the spinal cord cannot be fixed. There is no way to tell how serious the physical problems might be or whether or not the person will have mental disabilities.

With anencephaly the brain and back of the baby's skull are incompletely formed. This condition is not compatible with life and babies who have anencephaly are often stillborn or die shortly after birth.

WHAT OPTIONS ARE AVAILABLE TO ME? – SCREENING OPTIONS

If you are pregnant, you can choose to have prenatal screening to find out your chances of having a baby with the problems discussed above. In Ontario, pregnant women can have Maternal Serum Screening (MSS) or Integrated Prenatal Screening (IPS) which both look for the same problems but IPS is a better screen for Down syndrome.

1. Maternal Serum Screening – 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 level 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.

2. Integrated Prenatal Screening – Integrated Prenatal Screening (IPS) is another screening test that calculates the chance of a baby having Down syndrome, trisomy 18, and Open Neural Tube Defect. This test is done in two parts. Part one is done between 11 and 13 weeks plus 6 days of the pregnancy and involves a special ultrasound (called Nuchal Translucency) and a blood test. Part two of the testing is a blood test that is done between 15 and 18 weeks plus 6 days of the pregnancy. Results of the IPS are given after part two of the testing is completed. IPS cannot be done on a woman who is carrying twins, or if she has insulin dependent diabetes.

TESTING OPTIONS

1. 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. This first step in testing is offered after a "screen positive" is reported just to make sure

that the ultrasound dates agrees with your dates.

You may also be offered a detailed ultrasound which can look at the shape of the spinal bones and the skull of the baby. A detailed ultrasound can see some of the reasons why a screen result was positive. It cannot see all the problems a baby might have. If the ultrasound does not show the reason for the positive result, you may consider having amniocentesis as your next step.

2. Chorionic Villus Sampling: Chorionic villus sampling (CVS) is a test done to obtain a few cells from the developing placenta. This test is usually performed at 11-13 weeks of pregnancy. The cells from the placenta will provide information about the chromosomes of the developing baby.

There are two ways of doing CVS. One method inserts a needle through the mother's abdominal wall, and the other method puts a small tube into the vagina and into the uterus to sample the placenta. Ultrasound is used to locate the position of the developing placenta and the baby. There is about a 1% chance of miscarriage associated with this procedure. The results are ready in about 3-4 weeks. As CVS is not done in Northwestern Ontario, your health care provider would have to refer you to a larger centre.

3. Amniocentesis: Amniocentesis is a test done to obtain a sample of the amniotic fluid surrounding the baby. This test is usually done around the 16th week of pregnancy. The cells in the amniotic fluid will provide information about the chromosomes of the developing baby.

This test 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 about a 1/200 or 0.5% chance of miscarriage associated with this procedure. The results are available in about two to three weeks. Amniocentesis is done in Thunder Bay. Amniocentesis is offered to anyone over the age of 35 regardless of your choice around screening.

