The Complete Guide to Prenatal Testing

With the discovery of new markers and new technologies, the prenatal tests offered on the market have evolved considerably. It can be overwhelming when facing all these choices. This guide offers a description of the different prenatal tests offered in Canada. Its purpose is to help you understand these tests and help you make the best health care decision for you and your family.

Dynacare®

Prenatal Screening: An Informed Choice

Prenatal screening is used to assess whether your pregnancy is at higher or lower risk for certain chromosome differences, including Down syndrome. Currently, The Society of Obstetrics and Gynecology of Canada recommends that every woman should be informed of, and offered the option of prenatal screening. With that said, the decision to undergo a prenatal screening test is very personal. Some people may use prenatal screening tests for reassurance if the risk is low, or to make to make informed decisions regarding the management of their pregnancy in the event a chromosome difference or genetic condition is confirmed. Others may use this information to prepare for the birth of a child with special needs, and gather all the information and resources possible. Some couples also choose not to proceed with any screening tests.

Prenatal Screening Tests

Prenatal screening tests provide a risk assessment for certain conditions, such as trisomy 21 and trisomy 18. A screening test can tell if you are at higher or lower risk for a certain condition. It cannot tell for sure if the baby has the condition or not. Since these tests are done via ultrasound or blood draws, they represent no risk to the pregnancy. Some prenatal screening tests also provide a risk assessment for neural tube defects such as spina bifida. Results are classified as low risk (screen negative), or high risk (screen positive). There are 2 types of prenatal screening: conventional and non-invasive prenatal testing (NIPT).

Conventional Screening

Conventional screening tests are based on the combination of different factors: maternal age, blood markers and ultrasound markers.

Maternal Age

It is a well-known fact that the risk for a chromosome difference increases by a small amount each year as a woman ages. Maternal age is part of the risk calculation when doing prenatal screening tests.

Blood Markers

The amount of some proteins and hormones, which we call blood markers, can also help precise the risk assessment for chromosome differences. These blood markers are produced by the pregnancy and are found in small quantities in maternal blood. The level of these markers can be assessed in the 1st and/or 2nd trimester. Timing of the blood draw is very important.

Ultrasound Markers

Ultrasound markers are measurements and signs that can be assess during the 1st and 2nd trimester ultrasound. Markers are not malformations, but are indicators that can help estimate the risk for chromosome differences and certain genetic conditions. During the 1st trimester ultrasound, the measurement of the nuchal translucency (NT) and the examination of the nasal bone can be used in the risk calculation for chromosome differences. During the 2nd trimester ultrasound, the examination of other markers is done. If any are found, this information is used in connection with your prenatal screening test results to provide a personalized risk assessment.

There are many screening tests on the market. The markers included or the timing of the blood draw may vary depending on the test. The following is a short review of these different types of screening tests, as well as their benefits and limitations.

First Trimester Screening

As per the name of the test, the first trimester screening combines the blood and ultrasound markers of the 1st trimester with the maternal age. This test is usually performed between 11 weeks 1 day and 13 weeks 6 days. The results of this screening test are available in the first trimester and women with a higher risk can then be referred early in the pregnancy for NIPT or prenatal diagnosis.

Integrated Prenatal Screening (IPS)

Integrated Prenatal screening is the combination of maternal age with first trimester blood markers and the NT ultrasound done between 11 weeks 1 day and 13 weeks 6 days, as well as second trimester blood markers, done between 15 and 22 weeks gestation. It is very important to have both blood draws done in the correct time frame. Since this test includes all the possible markers for a conventional screening test, it has a high detection rate and the lowest false positive rate of all conventional screening tests. However, the results are available only in the second trimester, which can delay the referral for further testing.

Serum Integrated Prenatal Screening (SIPS)

In some Canadian centres, and in some remote or rural areas, the NT ultrasound is not routinely available. In this situation, Serum Integrated Prenatal Screening can still be performed using blood markers from the first and second trimester. The results are similar to those obtained by IPS, with a slight reduction in the detection rate and an increase in false positive rate.

Sequential Screening

In some Canadian centres, and in some remote or rural areas, the NT ultrasound is not routinely available. In this situation, Serum Integrated Prenatal Screening can still be performed using blood markers from the first and second trimester. The results are similar to those obtained by IPS, with a slight reduction in the detection rate and an increase in false positive rate.

Second Trimester Screening

Second trimester screening includes maternal age and blood markers of the 2nd trimester. Although the detection rate is lower and false positive rate higher than with the other screening tests, it may be used for pregnant women whose gestational age is too advanced for the first trimester screening tests.

Non-invasive Prenatal Screening

Non-Invasive Prenatal Testing or NIPT has been developed in the past decade and is now offered on the Canadian market. These tests are highly precise screening tests for the most common chromosome differences, including Down syndrome, Trisomy 18 and Trisomy 13. NIPT detects small pieces of chromosomes coming from the pregnancy (called cell-free DNA) that are found in a pregnant woman's blood, to determine if there is an extra chromosomes 21, 18 or 13. This type of test can be done earlier than conventional screening, starting at 10 weeks of pregnancy and requires a simple blood draw. These tests are also much more accurate than conventional screening. NIPT can reach a detection rate of >99% and a very low false positive rate for Down syndrome. This means that very few women will receive a positive result in an unaffected pregnancy, and very few pregnancies with Down syndrome will be missed. Although highly accurate, all high risk NIPT test results should be confirmed by diagnostic testing, such as an amniocentesis.

Prenatal Screening and Interpretation of the Results

These tests, either conventional screening or NIPT, are used to provide a personalized risk assessment for certain conditions. It is important to keep in mind that the results of a prenatal screening provides a probability and, while these tests represent no risk for the pregnancy, they cannot tell for sure if the pregnancy is affected with a chromosome difference or not.

Possible Results of Screening

A low-risk result means that the chance for your pregnancy to have a chromosome difference is reduced. However, some pregnancies which do have a chromosome difference will be missed by a screening test.

A high-risk result means that the chance for your pregnancy to have a chromosome difference is increased. Some pregnancies with a high-risk result will not be affected with a chromosome difference. As such, further testing is required to confirm the diagnosis.

Ultrasound Screening Tests

The use of ultrasound is also part of prenatal health care. In Canada, the second trimester ultrasound is the one recommended for every pregnant woman. However, other ultrasounds can also be offered depending on the provincial health care system, and the women's risk factors. Here is a review of the different ultrasounds and the information they can provide.

Dating Ultrasound

A dating ultrasound is usually done during the first trimester, between 7 to 9 weeks of pregnancy, and is primarily used to precisely date the pregnancy and to determine the number of fetuses. Depending on the gestational age, the heart beat can be heard during this ultrasound.

First Trimester Ultrasound

A first trimester ultrasound can done between 11 and 13 weeks and 6 days. During this ultrasound, they will assess the growth and the development of your fetus and also the gestational age. The measurement of the nuchal translucency (NT) is also possible at this stage of the pregnancy, which is used in the risk calculation for Integrated Prenatal Screening and First Trimester Screening tests. The nuchal translucency is the measurement of the fluid behind the neck of the fetus during the ultrasound. While it is normal to have some fluid, when the measurement is increased, this can indicate a higher risk for certain chromosomal differences or certain conditions. The presence or absence of the nasal bone is another ultrasound marker that can be included in the risk assessment for prenatal screening tests. The absence of the nasal bone at this stage of the pregnancy has been associated with a higher risk for Down syndrome. Sometimes the measurement cannot be taken because of the position of the fetus. As well, a first trimester ultrasound may also detect other rare malformations or conditions that may not be detected by conventional screening tests.

Second Trimester Ultrasound

The second trimester ultrasound is the most commonly offered ultrasound in Canada. At this stage of the pregnancy, the organs of the fetus are already well developed. The ultrasound technician will assess the different organs to make sure everything is developing properly. The technician will take measurement of the head, belly, arms and legs. They will also make sure that there is not too little or too much amniotic fluid surrounding the fetus. This is also another checkup for chromosome conditions, as other signs or markers can be seen at this stage. If any markers are seen, your doctor will review this information in connection with your prenatal screening results and see if any further testing is recommended. It is normally during this ultrasound that the gender can be revealed.

Third Trimester Ultrasound

The third trimester ultrasound may be offered for certain women. This ultrasound is use to determine the position of the baby and the placenta and also to measure the quantity of amniotic fluid. During this ultrasound, the estimation of the baby's weight is also possible.

Diagnostic Testing Options

Prenatal diagnostic tests can also be offered when the risk of having a genetic conditions are higher. Even though these tests can offer a yes or no answer for some genetic conditions, these tests use an invasive technique and present a risk of miscarriage. This is why these tests are usually offered to pregnant women with a higher risk of a genetic condition due to advanced maternal age, a previous pregnancy history, a high risk result after screening test or ultrasound findings suggestive of a chromosomal condition. A couple known to be carriers of a genetic condition could also be offered prenatal diagnosis.

Amniocentesis

Amniocentesis is a diagnostic test, meaning that it can tell with high accuracy whether or not the pregnancy is affected by a chromosome condition. Amniocentesis is the most commonly offered diagnostic test. It is offered after 15 weeks of pregnancy. Guided by ultrasound, a doctor inserts a needle through the mother's belly to remove a small amount of amniotic fluid. Cells from the fetus are found in the amniotic fluid, and these cells contain fetal chromosomes. Rapid results for chromosomes 21, 13, 18, X and Y are typically available within 2-3 days, with complete results available in 2 to 3 weeks. Amniocentesis has a greater than 99% accuracy and can detect differences in chromosome number, and other types of rare chromosome disorders. Amniocentesis is an invasive procedure, and it carries a risk of miscarriage of 1 in 200 (0.5%).

Chorionic Villus Sampling (CVS)

Chorionic villus sampling (CVS) is an alternate type of diagnostic test, offered between 10 and 13 weeks of pregnancy. It is performed by guiding a fine needle either through the mother's abdomen or a fine catheter transvaginally through the cervix. CVS takes a small piece of the developing placenta, rather than amniotic fluid, for chromosome testing. CVS is also very accurate, and rapid results for the most common chromosome differences are typically available within 2-3 days, while the complete results may take up to 3 weeks. However, there is a 2-3% chance that there will not be an informative result. This is due to chromosome differences which may be present in the placenta, but not in the developing fetus. As such, the accuracy of CVS is slightly less than amniocentesis (approximately 98%), and may require additional follow-up by amniocentesis. Additionally, although CVS provides an earlier result, the test has a higher risk of miscarriage, of 1 in 100 or 1%.

Carrier Tests for Specific Conditions

Carrier testing for specific genetic conditions may also be offered for some couples. When planning a family, some couples may be offered carrier testing based on their ethnicity or family history. The Society of Obstetricians and Gynecologist of Canada currently recommends this type of testing for all couples who are pregnant or planning a pregnancy. If both partners are found to be carriers of a specific genetic condition, prenatal diagnosis could be offered for the condition in the family.

Get the Information You Need Through the Dynacare Harmony Prenatal Test

When you are pregnant, your blood contains fragments of your fetus' DNA. Dynacare now offers the Harmony™ Prenatal Test. This is a new type of test that analyzes this DNA in a sample of your blood to determine the risk of trisomy 21 (Down syndrome), trisomy 18 (Edwards syndrome) and trisomy 13 (Patau syndrome).

The Harmony ™ Prenatal Test, a non-invasive prenatal test (NIPT), is a highly accurate prenatal test for all women regardless of age or risk, including women with twin pregnancies and pregnancies conceived by in vitro fertilization (IVF). It can be performed as early as 10 weeks in pregnancy and results are available within 10 business days.

Contact your doctor today to find out about Harmony Prenatal Testing for you.