Prenatal Screening and Ultrasounds

There are several prenatal screening options available to you during your pregnancy. A small genetic history is taken during your intake appointment with your midwives that may help guide your decisions with respect to prenatal screening. Most babies are born healthy. The intent of screening is to help identify major issues facing the development of the fetus. In Canada, about 4% of babies are born with a difference, a congenital anomaly, that may require medical or surgical intervention. The most common of these differences are heart defects, cleft lip and/or palate or a chromosomal anomaly such as Down Syndrome. Prenatal screening does not ensure a perfect outcome. Screening is not diagnostic. Screening is offered at a time (early enough) when parents can take action and make decisions about additional screening and/or diagnosis. It is important to consider all of the available options, the purpose of each screening tool, and the potential outcome of each test. Screening can result in considerable anxiety for parents that receive a 'positive' result, even if it is a false positive. Please read this document so that you can be informed of your decision.

11-14 week First Trimester Screen (FTS):
This test is offered to you as a screen for Down Syndrome (Trisomy 21), Trisomy 13, and Trisomy 18. It involves an ultrasound and a blood test. The Nuchal Translucency (NT) ultrasound measures the baby's nuchal fold (a small collection of fluid behind the baby's neck) and the accompanying blood work tests for 3 specific markers in your blood. The ultrasound and blood results are combined with your age to calculate your chance of having a child affected by Trisomy 13, 18, or Down Syndrome. This test is NOT diagnostic. A 'positive' result is one where your risk is greater than 1:150. This test is able to identify 85-90% of pregnancies with Down Syndrome. There are many false positives; approximately 4-5% of pregnant people taking the test will screen positive but the majority will NOT be carrying a baby with Down Syndrome. However, 10-15% of people taking the test will screen negative when actually the baby does have Down Syndrome, this is called a false negative. If you test positive, then further diagnostic testing will be discussed with you.

Non-Invasive Prenatal Testing (NIPT):
Offered through Panorama or Harmony. Tests for Trisomy 21, 18, 13, monosomy X (Turner Syndrome), sex chromosome aneuplodies, complete molar pregnancy, and finding out the fetal sex is optional. You can also choose to pay for additional screening for microdeletion syndromes. This test is not covered by Alberta Health Care . Private pay tests range from $495-$795. Tests are able to identify babies with Trisomy syndromes (13,18, and 21 aka Down Syndrome) 96-99% of the time. There is also much less of a chance of having a false positive compared to FTS, less than 0.1%. This testing is non-invasive because it involves taking a maternal blood sample and extracting fetal cells from the sample. This test is available any time after 10 weeks of pregnancy. For more information please visit this website: http://www.lifelabsgenetics.com/non-invasive-prenatal-testing/#whatis
https://www.dynacare.ca/what-s-next/prenatal/prenatal-testing.aspx

15-20 week Screening:
If you have already had the FTS or NIPT then this testing is not recommended for you. These tests rely on accurate dating of a pregnancy. There are 2 types of tests during this time period:

Maternal Serum Screening (MSS): A blood sample is taken at a lab and 3 specific maternal blood levels are tested to calculate your risk of having a pregnancy affected by Trisomy 18, Down Syndrome, and neural tube defects. This test correctly identifies Down Syndrome in 69 of 100 babies who have it.

Quad Screening: Same procedure as the MSS with the addition of one maternal blood test. This test correctly identifies Down Syndrome in 81 of 100 babies who have it.

Both tests correctly identify babies with neural tube defects (spina bifida/anencephaly) 80-90% of the time. However the primary screening tool for these neural tube defects is the 18-22 week Fetal Anatomy Scan.

15+ weeks Amniocentesis:
Amniocentesis is a diagnostic test for chromosomal abnormalities. Results are available 3-4 weeks after the test. It is not routinely offered to women. It is available for you if you:

- tested 'positive' for the FTS, MSS, or Quad Screen.
- have been deemed 'high risk' by NIPT
- have a history of a pregnancy or child affected by a chromosomal condition
- have a family history of an inherited disorder that can be detected by amniocentesis
- have a pregnancy where an abnormality has been detected by ultrasound or have multiple 'markers'

Amniocentesis is an invasive procedure whereby an ultrasound-guided needle is introduced into the mother's abdomen to withdraw a sample of amniotic fluid from around the baby. This technique is associated with a 1/200 risk of miscarriage.

18-22 week Fetal Anatomy Scan:
This ultrasound is recommended for all pregnant women. It is a screening tool to examine the baby's anatomy (heart, brain, limbs, organs) and measure the baby's growth. It also observes the location of the placenta and you may be able to find out the sex of the baby at this scan. Most birth defects can be identified with this ultrasound but not everything (3-4% are missed). This ultrasound is important for identifying an abnormally located placenta and any major birth defects where a prenatal diagnosis would be beneficial. This ultrasound is not considered a genetic screening tool but certain physical features may be seen in baby's with trisomy 13,18 and 21 that increase the suspicion of one of these trisomy's. This ultrasound can also sometimes detect soft markers. Soft markers are often of unknown significance and are part of a normally developing baby but they may be associated with chromosomal abnormalities, particularly if there are multiple soft markers. Often soft markers are incidental findings, especially if previous prenatal screening was negative.

Most babies are born healthy regardless of a woman's age, obstetric history or family history. The information from these types of tests can help you make decisions about whether to continue a pregnancy or not, or feel better prepared to parent a child with a disability. More often than not, these results cause stress and anxiety when a 'false positive' is reported. Genetic counselling is available if you have a pregnancy affected by a birth defect or chromosomal condition. Please give these choices careful consideration and do not hesitate to ask questions.

References

Genetics Education Canada. (2019). A guide to understanding prenatal tests, https://geneticseducation.ca/public-resources/prenatal-and-preconception-genetics/guide-to-understanding-prenatal-screening-tests/

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Society of Obstetricians and Gynaecologists of Canada. (2014). Prenatal screening, diagnosis, and pregnancy management of fetal neural tube defects. *Journal of Obstetrics and Gynecology Canada 2014; 36*(10): 927-939.

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