

MEDICAL GENETICS PROGRAM*of Southwestern Ontario*

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PRENATAL TESTING IN PREGNANCY

There are two prenatal genetic testing procedures available at LHSC:

1. Amniocentesis
2. Chorionic villi sampling (CVS)

Amniocentesis is performed after 15-16 weeks of pregnancy. An ultrasound machine is used to take a picture of the baby and the uterus. A thin needle is then inserted through mother's abdomen into the fluid around the baby. A small amount of amniotic fluid is removed (approximately 2-3 teaspoons). This fluid contains some cells from the baby. Genetic testing can be performed on these cells, called amniocytes. The risk for miscarriage associated with the amniocentesis is approximately 1/800 for a single baby, and approximately 1/400 for twins.

Chorionic Villi Sampling (CVS) is performed between 11+3 and 13+6 weeks of pregnancy. An ultrasound machine is used to take a picture of the baby and placenta. CVS can be done in two different ways. One way is to insert a catheter (a small tube) through the vagina into the uterus to obtain a small tissue sample from the chorionic villi, which is part of the placenta. The other way is to insert a thin needle through mother's abdomen into the uterus to obtain a small tissue sample from the chorionic villi. The doctor will decide which method is best based on the position of the placenta. Genetic testing can be performed on the chorionic villus cells. The risk for miscarriage associated with CVS is approximately 1/400. Other possible concerns with CVS will be discussed during your genetic counselling appointment.

What kind of testing can be done?

When testing is being performed because there is a risk of trisomy 21, 18 or 13 only, a technique called rapid aneuploidy detection (RAD) will be performed. This result will take up to 3 business days. When testing is being performed because of ultrasound abnormality or a family history of a chromosome rearrangement, a full chromosome study will be performed. This result will take approximately 2-3 weeks. Under special circumstances, other types of genetic testing may be performed. The accuracy of these tests and the time for results will be discussed with your genetic counsellor.

What is Rapid Aneuploidy Detection (RAD)?

RAD is designed to detect numerical abnormalities (including trisomies) of chromosomes 21, 18, 13, X, and Y. RAD cannot detect abnormalities of the other chromosomes, nor can it detect most structural chromosome abnormalities. A normal RAD result means that Trisomy 13, 18, or 21 was not detected in the sample tested, and if this was the reason for testing, no further analysis will be done. A normal RAD result does NOT mean there are no chromosome abnormalities.

How accurate are results?

The results of CVS and amniocentesis are very accurate for RAD and routine chromosome testing. For DNA testing the degree of accuracy depends on the specific test being ordered. Failure to get a result

after a successful procedure occurs about 0.1% of the time with amniocentesis, and about 1-2% of the time with CVS. This is because of non-informative molecular markers by RAD (QF-PCR) testing and difficulties with cell growth. There is a possibility with amniocentesis and CVS that the results are difficult to interpret. Occasionally we cannot be sure whether the chromosome difference we find will affect the development of the baby. For example, we may find mosaicism (a mixture of normal and abnormal cells). Further tests may provide us with a better understanding of how this could affect the baby's development, but there is a possibility of being left without a clear answer. There is also the possibility that we have grown only mother's cells (i.e., maternal contamination). If this is the case, we may have missed any chromosome abnormalities present in the baby's cells. These limitations are quite rare. Sometimes additional testing is required. It is also important to remember that there is no genetic test that can guarantee a healthy baby. In every pregnancy there is a 2-3% chance to have a baby with a birth defect or medical problem.

Who is eligible for prenatal testing?

Women are eligible for prenatal testing if they:

- Have a positive serum screen (MSS or eFTS)
- Have a fetal ultrasound abnormality
- Have another child with a known chromosome abnormality, metabolic condition or gene disorder
- Have a high risk to have a child with a known chromosome condition, metabolic condition, or gene disorder

Where is the testing done?

Both the amniocentesis and CVS procedures are done in Women's Ambulatory Care at London Health Sciences Centre. A map will be provided or visit our website at [http://www.lhsc.on.ca/Patients Families Visitors/Genetics/](http://www.lhsc.on.ca/Patients_Families_Visitors/Genetics/). RAD and chromosome analysis are performed at LHSC. More specialized testing may be performed at LHSC, or cells may need to be sent to other laboratories outside of London.

What happens at my appointment?

When you arrive you will be asked to show your Ontario health card to register. You will then be seated in the waiting room. The amount of time you have to wait depends on how busy the clinic is that day. Once you are called into the procedure room a nurse will review your information and ask you to sign a consent form for the procedure. You can bring one support person into the procedure room with you. There is no recovery time required after the testing. Most women can leave the clinic shortly after the procedure. Women are advised to rest for approximately 24-48 hours after the procedure.

What happens if the results indicate there is a problem?

If there is a problem identified on the amniocentesis or CVS you will be offered an additional genetic counselling appointment where you will meet with a genetic counsellor and/or a medical geneticist for further information.

If you have any questions or concerns, please do not hesitate to contact us at 519 685-8500, extension 52571 for further information. Additional information can be found at our website: [http://www.lhsc.on.ca/Patients Families Visitors/Genetics/](http://www.lhsc.on.ca/Patients_Families_Visitors/Genetics/)