NON-INVASIVE PRENATAL TESTING FOR CHROMOSOME ABNORMALITIES

You may have heard about the "new Down syndrome blood test". This is a non-invasive prenatal testing (NIPT) option available to women to better assess the risk that they are carrying a fetus with Down syndrome (trisomy 21). Currently, this testing is only supported by MOHLTC[+] funding in certain circumstances; however it is also an option available to those who are able to pay for the service themselves (minimum cost of ~$800).

WHAT IS IT?

NIPT is a way to *screen* for specific chromosome abnormalities (Down syndrome - an extra chromosome 21; an extra chromosome 13 or 18; and extra or missing sex chromosomes X & Y). NIPT quantifies fetal DNA that is circulating in maternal blood. Fetal DNA comprises approximately 10% of DNA in maternal blood and the amount increases with gestational age. It is a non-invasive test that poses no risk to pregnancy.

WHAT IS THE BENEFIT?

NIPT is <u>not</u> a replacement for diagnostic prenatal testing (via amniocentesis or chorionic villus sampling [CVS]). Currently, the SOGC[++] recommends NIPT as a second tier prenatal screen, demonstrating much higher detection rates and lower false positive rates than IPS and MSS[**]. The expected benefit of NIPT will be fewer women undergoing secondary invasive diagnostic tests associated with a risk of pregnancy loss.

HOW GOOD IS IT?

A series of NIPT validation studies have demonstrated high pick-up rates/sensitivity (which is company specific) for the detection of Down syndrome (sensitivity 99-100 %, with false positive rates [FPR] <1%), trisomy[*] 18 (sensitivity 97-100%), and trisomy[*] 13 (sensitivity 79-92%) and, with some companies, sex chromosome differences (sensitivity approximately 94-99%). A number of women have been required to have repeat blood draws due to initial test failure. Test failure may be as high as 6%.

WHO SHOULD CONSIDER HAVING THIS TEST?

Here in the Regional Genetics Program at CHEO we are discussing NIPT with all pregnant women who are eligible for invasive diagnostic testing. The indications include, but are not limited to: advanced maternal age; a positive prenatal screen result; a personal or family history of a common aneuploidy; soft ultrasound markers; pregnancies conceived via reproductive technologies. Eligibility is company-specific.

HOW IS THE TESTING DONE?

Women who are eligible for a genetic counselling appointment at CHEO may be candidates for MOHLTC funding of this test; their genetic counsellor will facilitate an application in these instances. Otherwise NIPT will not be organized through our program. Women who are interested are encouraged to discuss NIPT with their primary care providers. Currently, we are aware of the following ways to obtain testing in Ontario.

Company	Facilitating Service provider	Website	Contact information
Panorama™ by Natera	Life Labs	http://www.lifelabs.com/Lifelabs_ON/Patients/TestInfo/Special/Panorama.asp	1-877-849-3637
Verifi™ by Verinata Health	Medcan Clinic	http://www.medcan.com/services/genetics/non-invasive_prenatal_testing/	1-800-4MEDCAN
Harmony Prenatal Test™ by Ariosa Diagnostics	Gamma-Dynacare	http://www.ariosadx.com/	1-855-9-ARIOSA (855-927-4672)

As technologies and companies change rapidly in the field of genetics, it is important to know that companies offering NIPT may change.

We do not endorse any laboratory over another. You should do your own research about a specific company before using their services. Ordering providers should be prepared to provide complete pre-test and post-test counselling.

WHAT HAPPENS WHEN THE RESULTS COME BACK?

The companies report that their results take approximately 1-2weeks.

If the result is negative, this is reassuring. If eligibility for invasive diagnostic testing existed prior to NIPT, a woman would still be eligible to proceed.

If the result is positive, genetic counselling is available at the Genetics Clinic and providers can make a referral to us using the number below. The SOGC[++] recommends that no irrevocable obstetrical decisions should be made in pregnancies with abnormal NIPT results without confirmatory invasive testing.

If not already performed, maternal serum AFP measurement is suggested at 15-16 weeks since NIPT does not screen for open neural tube defects. A routine morphology ultrasound between 18-20 weeks is recommended in all pregnancies.

We are unable to provide telephone consultation to women in the community. NIPT will be discussed as an option during a genetic counselling appointment if an individual has already been referred to our clinic for an appropriate indication. As your local Genetics Centre, we are happy to be a resource for providers.

CHEO'S REGIONAL GENETICS PROGRAM CONTACT INFORMATION
TELEPHONE: 613-737-2275
FAX: 613-738-4822
WEBSITE: http://www.cheo.on.ca/en/genetics

MOHLTC – **M**inistry **o**f **H**ealth and **L**ong-**t**erm **C**are is the provincial healthcare funding plan for Ontario residents. For more information http://www.health.gov.on.ca/en/

[*]trisomy – refers to an extra copy of a chromosome

[++]SOGC – **S**ociety of **O**bstetricians and **G**ynecologists of **C**anada establishes produces national clinical guidelines for both public and medical education on important women's health issues. For more information http://sogc.org/

[**]IPS and MSS – **I**ntegrated **P**renatal **S**creening and **M**aternal **S**erum **S**creening are prenatal screening options that provide a woman with her own risk of having a child with: Down syndrome (also called Trisomy 21); Trisomy 18 and Open Spina Bifida (open neural tube defects). For more information http://www.prenatalscreeningontario.ca/

UPDATED FEBRUARY 2014