BC Prenatal Genetic Screening Program

Perinatal Services BC
An agency of the Provincial Health Services Authority

Non-Invasive Prenatal Testing (NIPT)
Comparison of NIPT and Amniocentesis

Revised February 23, 2016

NIPT is a blood test for the detection of Down syndrome (trisomy 21), trisomy 18, and trisomy 13 based on testing the cell-free fetal DNA circulating in maternal blood. **NIPT is now a funded benefit for women at high risk for trisomies based on one of the following:** a positive screen for Down syndrome or trisomy 18; a previous pregnancy affected with trisomy 13, 18 or 21; or specific ultrasound findings indicating an increased risk of Down syndrome or trisomy 18.

The BC Prenatal Genetic Screening Program (PGSP) has selected Dynacare, provider of the Harmony test, as the provincial vendor for funded NIPT for these women. For those eligible screen positive women who are interested in NIPT as a next step, health care providers can arrange funded NIPT by filling out the PGSP/Dynacare Funded NIPT Requisition (available on our website) with a patient-specific authorization code. Blood collection for pre-authorized funded NIPT is available through specific collection sites listed on our website.

Women with a previous trisomic 13, 18, or 21 pregnancy, or an ultrasound abnormality, who may be eligible for funded NIPT must be referred to Medical Genetics at either BC Women's Hospital (Tel: 604-875-2157 or fax referral to 604-875-3484) or Victoria General Hospital (Tel: 250-727-4461) for counselling and funded NIPT coordination.

All women at average risk, or those choosing NIPT as a first tier screen, continue to have the option of self-pay NIPT and can access it from a variety of locations listed on our website.

Comparison of NIPT and Amniocentesis	NIPT Blood Test	Amniocentesis
Detection Rate Down syndrome (Trisomy 21)	>99%	100%
Trisomy 18	~ 97%	100%
Trisomy 13	~ 93%	100%
Sex aneuploidy	96% for 45,X (Turner syndrome)*	100%
Other Chromosome Abnormalities	0	Depends on test done†
False Positive Rate (FPR)	<0.1% for T21, T18, T13** 1% for sex aneuploidy	0
Positive Predictive Value	>95% in high-risk pregnancies (40-80% in low-risk)	100%
Risk to Pregnancy	0	1 in 200 pregnancy loss
Failure Rate	~ 5% on first blood draw	< 1 in 1000
Result Turn-Around Time	Approx.10 business days	3-14 days depending on test done†

* Limited data available on detection rates for 47,XXY, 47,XXX, and 47,XYY.
** Given a small FPR, women with positive NIPT results are advised to have amniocentesis for diagnostic confirmation.
† When amniocentesis is done for a positive screen, only rapid aneuploidy testing is done. If procedure done for a fetal abnormality, a karyotype or microarray analysis will be done.

West Tower, 350-555 West 12th Avenue, Vancouver, BC V5Z 3X7
Phone: 604-877-2121 | Fax: 604-872-1987 | Email: psbc@phsa.ca | Web: bcprenatalscreening.ca