BC Prenatal Genetic Screening Program

Perinatal Services BC
Provincial Health Services Authority

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

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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 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) works with Dynacare Next, 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 the patient-specific authorization code provided on the screen positive report. Blood collection for preauthorized funded NIPT is available through specific collection sites listed on our website. Note – the screen positive patient-specific authorization code is for one-time pregnancy use only.

Women with a previous trisomic 13, 18, or 21 pregnancy may be eligible for funded NIPT. Medical Genetics at either BC Women's Hospital or Victoria General Hospital can be consulted (via telephone) to access an NIPT authorization code, or patient can be referred for counselling and funded NIPT coordination. A referral is required to either Medical Genetics sites for women with an ultrasound abnormality to be counselled on all options which may include funded NIPT. BC Women's Hospital (Tel: 604-875-2157 or fax referral to 604-875-3484); Victoria General Hospital (Tel: 250-727-4461).

Women at increased risk for trisomies based on a positive screen for Down syndrome or trisomy 18 with risk greater than 1 in 300; a previous pregnancy affected with trisomy 13, 18 or 21; or specific ultrasound findings indicating an increased risk of Down syndrome or trisomy 18 may be eligible for both funded NIPT or amniocentesis. This table compares both options and was created to facilitate counselling of these women.
Information about funded NIPT.

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	~ 2% on first blood draw	< 1 in 1000
Result Turn-Around Time	Approx.8 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 (BCCH, RCH). If procedure done for a fetal abnormality, a microarray analysis will be done.
# If amniocentesis in Victoria, women <20 wks GA and risk <5%, full karyotype done with results ~ 14 days. If GA >20 wks or risk >5%, rapid FISH done with results ~ 3 days.

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