NL Health Services

PROVIDER INFORMATION SHEET

Non-Invasive Prenatal Screening (NIPT)

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> "All pregnant women in Canada, regardless of age, should be offered, through an informed counselling process, the option of a prenatal screening test for the most common fetal aneuploidies (II-A)."
>
> No. 348-Joint SOGC-CCMG Guideline (2017)

Any person of any age may have a child born with a chromosome abnormality, but it is well known that the chances of this happening increase as a pregnant person gets older. The most common of these include Down syndrome (also known as Trisomy 21), trisomy 18 and trisomy 13.

Non-invasive prenatal screening (NIPT) on cell-free DNA is a screening test for Down syndrome, Trisomy 18 and Trisomy 13 that measures chromosome material reflecting the fetus present in a mother's blood during pregnancy. It can be performed at any point from 10 weeks gestation onward. In most cases, it is not currently covered by the public healthcare system and the different options cost between $349- $550 CAD. Although it is a much more accurate screening test for the three trisomies when compared to maternal serum screening (MSS), NIPT screen-positive results are not diagnostic and need to be confirmed by an invasive prenatal test (amniocentesis or chorionic villus sampling).

NIPT can include analysis of the fetal sex and/or sex chromosome aneuploidies for those who are interested. Many NIPT tests also include the option of adding on analysis of microdeletion syndromes. Testing for these microdeletions is *not* currently recommended by the Society of Obstetricians and Gynaecologists of Canada (SOGC) and the Canadian College of Medical Geneticists (CCMG) however this remains available from certain labs. Testing limitations should be discussed before proceeding with ordering.

Counselling Considerations

All pregnancies have a 3-5% risk of significant birth defects. **It is important to remember that no test can detect *every* type of physical or mental disability.** However, prenatal screening may help reassure

or help guide important decisions about the pregnancy. Screening options should be reviewed with all pregnant patients. It is helpful to ask the patient to think ahead about various outcome possibilities and the next steps they might take. This will help facilitate a decision about prenatal screening that is right for the patient.

Discuss what your patient's goals are for prenatal screening. Do they wish to include reporting of the fetal sex or sex chromosome aneuploidies? How would they feel about a screen-positive result?

Timing

NIPT can be done as early as 10 weeks gestation. Typically, turnaround times vary from 7-10 days. It is important to consider the implications of timing for follow-up in the event of a screen-positive result. Chorionic villus sampling (CVS) can typically be completed between 11-13 weeks gestation but can only be done outside of the province. The cost of the CVS procedure is covered under MCP. There is partial reimbursement available for certain travel costs under the provincial Medical Travel Assistance Program (https://www.gov.nl.ca/exec/las/medical-transportation-assistance-program-mtap/medical-transportation-assistance-program/). Amniocentesis is available in St. John's after 16 weeks gestation. Depending on the time results are returned, proceeding with an amniocentesis may involve a waiting period.

Cost

There are multiple companies who offer NIPT. Each test has its own cost, limitations, and benefits. The Provincial Medical Genetics Program does not recommend any test over another. We advise that physicians speak with their patients about the cost, limitations, turnaround times, and possible results for each available option to decide which is best for them.

Possibility of Abnormal Results

There is a possibility of abnormal results with any test. For NIPT this may include a failed test or high-risk/positive results. It is important to review that NIPT is a screening testing and is therefore not diagnostic. Most screen-positive results are associated with a positive predictive value (PPV, the chance the result is a true positive vs false positive) for each condition and these PPVs vary drastically between conditions and age of patients. Rare conditions, such as microdeletion syndromes, do not have a reported PPV.

For example: The PPV for a 30-year-old woman who screened positive for Down syndrome may be around 61%, whereas the PPV for a 42-year-old woman who screened positive for Down syndrome may be around 96%. You can estimate PPV values with the online prenatal calculator: https://www.perinatalquality.org/Vendors/NSGC/NIPT/

Possibility of a failed test

As many as 1-8% of patients will have a failed test. Approximately 40-50% of the time with failed tests, patients will get a result on repeat testing (typically offered at no additional cost.) However, it is important to note that there is an increased risk of aneuploidy with current estimates as high as 5%. Failed tests are typically because of low fetal fraction; this is often due to earlier gestational age than thought, or high BMI. Patients with a failed NIPT may have repeat testing ordered by their prenatal care provider. Patients wishing to consider invasive genetic testing can be referred to the Provincial Medical Genetics Program.

General Considerations when Ordering NIPT

- Ensure you have the correct kit available and have completed the requisition and payment form (if required) in full.
- Include your patient's EDD, height, and weight on the requisition. This helps ensure the most accurate test result.
- Blood samples for NIPT are time sensitive and must arrive at the testing facility in a certain number of days (usually 6 days or less). Therefore, blood can only be collected early in the week. Contact your local blood collection lab to confirm their policy for your area. In St. John's, blood can be collected Monday, Tuesday, Wednesday, and Thursday (before 11am on Thursday).

When Referring to Genetics:

1. **Always** include copy of positive/high-risk/abnormal/failed NIPT results and indicate patient's feelings toward:
    a. Pregnancy management
    b. Diagnostic procedures, such as amniocentesis
2. Do **not** refer solely for ordering NIPT
3. Continue to refer for typical considerations such as:
    a. Personal or family history of a known genetic condition
    b. Fetal anomalies or soft markers identified on prenatal ultrasound
    c. Screen-positive or inconclusive NIPT results
    d. Positive maternal serum screening result or history of aneuploidy in a previous pregnancy if the patient wishes to proceed with invasive testing, such as chorionic villus sampling or amniocentesis

Contact Information

Provincial Medical Genetics Program – T: 709-777-4363, F: 709-777-4190

Harmony (Dynacare) – T: 888-988-1888, F: 450-663-4428, E: dynacarenext@dynacare.ca

Panorama (LifeLabs) – T: 844-363-4357 ext 0, F: 647-943-2804, E: ask.genetics@lifelabs.com

Panorama (Natera) – T:844-778-4700

Current Options for Patient-Pay NIPT

	Harmony	Panorama	Panorama
Testing Lab	Dynacare	LifeLabs	Natera
Website	https://www.dynacare.ca/patients-and-individuals/health-solutions/prenatal-solutions/harmony-nipt.aspx	https://www.lifelabsgenetics.com/product/non-invasive-prenatal-testing/	https://www.natera.com/womens-health/panorama-nipt-prenatal-screening/
Lab Location	Ontario	Ontario	USA (Texas and California)
Turnaround time	10-14 calendar days	7-10 calendar days	10-14 calendar days
Reporting method	Fax	Fax	Fax
Suitable for twin pregnancies?	Yes – will not report zygosity	Yes - will report zygosity	Yes - will report zygosity
Use with vanishing twin?	No	Yes, with significant limitations	Yes, with significant limitations
Use for donor pregnancy?	Yes	Yes	Yes
Detects triploidy?	No	Yes	Yes
Price – Common Trisomies with or without fetal sex and/or sex chromosome aneuploidies	$349 CAD	$550 CAD	$350 USD (~$475 CAD)
Common trisomies + del22q11.2 only	$349 CAD	$650 CAD	$425 USD (~$575 CAD)
Common trisomies + mirodeletions	Not available	$795 CAD	$525 USD (~$710 CAD)

Information in this document may change without notice. Please contact Medical Genetics for updates (709) 777-4363.