

Physicians' Lab Update / Newsletter October 2013

LifeLabs Now Offers Panorama™ NIPT

What is a Non-Invasive Prenatal Test (NIPT)?

Non-invasive prenatal testing is a new, highly accurate screen for specific chromosome abnormalities like trisomy 21 (Down syndrome), trisomy 18 (Edwards syndrome), trisomy 13 (Patau syndrome), and monosomy X (Turner syndrome).

How does it work?

During pregnancy a small amount of cell-free fetal DNA (cffDNA) crosses into the mother's bloodstream. Panorama™ uses sophisticated DNA sequencing technology and bioinformatics to analyze the fetal DNA in the mother's blood and detect aneuploidies (abnormal chromosome numbers).

How early in pregnancy can Panorama™ be done?

The test can be done on a maternal blood sample as early as 9 weeks gestation.

Who should be tested?

The leading American and Canadian prenatal associations (ACOG¹, SOGC²) have recommended that NIPT be offered as a screening test to women at increased risk of fetal aneuploidy, prior to invasive diagnostic testing like amniocentesis, which carries a risk of miscarriage.

Indications for NIPT include:

- Advanced maternal age (≥ 35 years of age or ≥ 40 years of age, depending on the province)
- A positive prenatal serum screen
- Personal or family history of aneuploidy
- Abnormal ultrasound finding suggestive of an increased risk of aneuploidy.

The accuracy of NIPT has not been validated in low-risk women. Studies are ongoing.

Ordering Panorama™ is easy:

The physician downloads the Panorama™ requisition form and patient consent form from the LifeLabs website. The patient calls LifeLabs to arrange an appointment, a blood sample is drawn, and the result is sent to the ordering healthcare provider within 10 days.

What is the follow-up for a positive NIPT result?

Although it is highly accurate, it is recommended that positive NIPT results be confirmed by diagnostic testing (chorionic villus sampling or amniocentesis).

Is NIPT covered under provincial health plans?

Unlike amniocentesis, NIPT is not currently covered under any provincial health plan. The pregnant mother is responsible for the cost of the test. However, Panorama™ is one of the most affordable NIPTs in Canada.

How does Panorama™ compare to another NIPT and amniocentesis?

Features		Harmony test ^{3,4} (Ariosa)	Panorama™ ^{5,6} (Natera) offered by LifeLabs	Amniocentesis
Earliest Testing Possible		10 weeks	9 weeks	15 weeks
Down Syndrome (T21)	Sensitivity	>99%	>99%	100%
	False Positive Rate	0.1%	0%	0%
Edwards Syndrome (T18)	Sensitivity	98.1%	>99%	100%
	False Positive Rate	0.1%	0%	0%
Patau Syndrome (T13)	Sensitivity	80%	>99%	100%
	False Positive Rate	0.05%	0%	0%
Monosomy X (45,X)	Sensitivity	96.7%	91.7%	100%
	False Positive Rate	Unknown	0%	0%
Detection of Triploidy		No	Yes	Yes
Risk of Miscarriage		0	0	1 in 200
Results Turnaround Time		Up to 10 days	Up to 10 days	About 14 days

Points to Remember

The Panorama™ NIPT is:

- Simple and safe – performed on a maternal blood sample with no risk of miscarriage.
- Highly accurate for detection of Trisomies 21, 18 and 13 – significantly reduces number of unnecessary invasive procedures like amniocentesis after a positive serum screen.
- The only NIPT that can detect Triploidy.
- Performed as early as 9 weeks gestation.
- One of the most affordable NIPTs in Canada.

References:

1. American College of Obstetricians and Gynecologists. Committee Opinion No. 545: Noninvasive prenatal testing for fetal aneuploidy. *Obstet Gynecol* 2012;120(6):1532-4.
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4. Ashoor G, Syngelaki A, Wang E, Struble C, Oliphant A, Song K, et al. Trisomy 13 detection in the first trimester of pregnancy using a chromosome-selective cell-free DNA analysis method. *Ultrasound Obstet Gynecol* 2013;41(1):21-5. doi: 10.1002/uog.12299.
5. Nicolaides KH, Syngelaki A, Gil M, Atanasova V, Markova D. Validation of targeted sequencing of single-nucleotide polymorphisms for non-invasive prenatal detection of aneuploidy of chromosomes 13, 18, 21, X, and Y. *Prenat Diagn* 2013;33(6):575-9.
6. Samango-Sprouse C, Banjevic M, Ryan A, Sigurjonsson S, Zimmermann B, Hill M, et al. SNP-based non-invasive prenatal testing detects sex chromosome aneuploidies with high accuracy. *Prenat Diagn* 2013;33(7):643-9.

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