Non-Invasive Prenatal Testing (NIPT)
Frequently Asked Questions

WHAT IS NIPT?
NIPT (Non-Invasive Prenatal Testing) is a single blood test that uses cutting-edge technology to screen pregnant women for chromosome abnormalities as early as 10-weeks in pregnancy. NIPT analyzes fetal DNA in maternal blood to detect Trisomy 13, 18, 21 (Down Syndrome) and sex chromosome abnormalities (X and Y).

WHO CAN GET NIPT?
Women who are at least 10-weeks pregnant with a singleton or twin pregnancy can contact Medcan to arrange for an appointment. A physician referral is not required.

IS NIPT THE SAME FROM ANY PROVIDER?
No. Not all NIPT tests are equal. After an extensive review of the various NIPT options available, Medcan chose to offer the verifi® prenatal test by Illumina.

While the verifi® prenatal test is not covered by OHIP for any Ontario patient, Medcan offers this NIPT option exclusively because it is the most comprehensive and definitive test available, featuring:
- Fastest turn-around time (within approximately 5 business days)
- Lower sample failure rate
- Ability to test singleton or twin pregnancies
- Ability to test pregnancies using an egg donor
- Inclusion of sex chromosome abnormalities in analysis
- Option to test for microdeletion syndromes
- Use of the most advanced sequencing technology

NIPT with Medcan also includes a full session with a genetic counsellor, who will review the test in detail with you and answer all your questions based on their deep expertise in genetics and interpretation of genetic information.

HOW SOON WILL RESULTS COME IN FROM MEDCAN?
Results are provided within approximately 5 business days.
HOW ARE RESULTS PROVIDED?

Results regarding chromosome problems are provided as either: Detected or Not Detected. This is in stark contrast to the numerical ‘risk score’ (i.e., 1/150) commonly reported by other prenatal screening tests.

HOW ACCURATE IS NIPT?

Medcan-offered NIPT provides a high sensitivity rate (>99%) and low false-positive rate (<0.1%) making it highly accurate. It has the potential to reduce the number of invasive procedures, like CVS and amniocentesis, which carry a risk of miscarriage. Because NIPT technology directly analyzes fetal DNA circulating in the maternal blood, it has a higher detection rate and is more definitive than standard screening tests.

ACCURACY RATES

<table>
<thead>
<tr>
<th>Chromosome</th>
<th>NIPT detection rate</th>
</tr>
</thead>
<tbody>
<tr>
<td>21</td>
<td>99.49%*</td>
</tr>
<tr>
<td>18</td>
<td>97.23%*</td>
</tr>
<tr>
<td>13</td>
<td>97.98%*</td>
</tr>
<tr>
<td>Sex chromosomes</td>
<td>&gt;90%</td>
</tr>
</tbody>
</table>

*Based on Illumina’s outcome data for over 85,000 samples. Sex chromosome aneuploidies are rarer; large-scale data is not yet available. (Taneja PA, et al. Prenat Diagn. 2016; 36(3):237-243).

The Medcan-NIPT sample failure rate is <0.07%. Other NIPT failure rates are as high as 5-8%. Failed tests require a second blood draw and result in delayed report of results.

DOES THE TEST WORK IN TWIN PREGNANCIES?

Yes. Not all NIPT tests can be provided to women who are pregnant with twins – but Medcan-offered NIPT can. There are some differences in the information available for twin pregnancies. You will have the opportunity to discuss this with a Medcan genetic counsellor prior to proceeding with the test.

DOES THIS TEST REPLACE CVS OR AMNIOCENTESIS?

No. NIPT is a prenatal screening test, not a diagnostic test. Screening tests can help identify which women should be given the option for diagnostic testing in pregnancy. However, the major downside to current diagnostic tests is that they are invasive and carry a small risk of complications, including miscarriage. The aim of NIPT is to help reduce the number of women who proceed to CVS and amniocentesis.

DOES THIS TEST REPLACE THE NEED FOR AN ULTRASOUND?

No. Ultrasounds are performed for a number of different reasons, including dating the pregnancy, making sure the amount of fluid around the baby is sufficient and screening for physical abnormalities. The Nuchal Translucency (NT) scan done at 11 weeks may provide some information about the chance for a chromosome problem, but it can also be a red flag for other potential issues, such as congenital heart disease. Therefore, it is not recommended that you forgo ultrasounds because you choose to do NIPT.

IS THE TEST MORE ACCURATE IF DONE EARLIER IN A PREGNANCY?

After 10 weeks gestation, the accuracy of the testing is the same whether you are in your 10th or 23rd week of pregnancy.

CAN NIPT LOOK AT ANYTHING ELSE BEYOND COMMON CHROMOSOME ABNORMALITIES?

Yes, we are now able to screen for several microdeletion syndromes using NIPT. A microdeletion refers to a small chromosomal deletion spanning several genes. A few factors to consider:

- Microdeletions are not part of routine prenatal screening
- Microdeletion are common, affecting 1 in 1000 pregnancies
- Can affect anyone at any age, or ethnicity
- Can occur spontaneously without family history
- Microdeletion syndromes are often missed by routine ultrasound and by routine amniocentesis/CVS
Medcan clients undergoing NIPT now have the option to include screening for the following 6 microdeletion syndromes:

1. 22q deletion syndrome (DiGeorge)
2. Prader-Willi syndrome
3. Angelman syndrome
4. Cri-du-chat
5. 1p36 deletion syndrome
6. Wolf-Hirschhorn syndrome

Patients can discuss this Microdeletion Panel add-on in more detail during their genetic counseling appointment.

**WHY CHOOSE MEDCAN?**

With 30+ years experience in providing health services, Medcan was the first clinic in Canada to offer NIPT. After an extensive review of the various NIPT options available, Medcan chose to partner with Illumina to offer the verifi® prenatal test, the most comprehensive and definitive prenatal test available.

At our well-equipped and comfortable downtown Toronto location, you meet with a board-certified genetic counselor. A genetic counselor is a healthcare professional with deep expertise in genetics and interpreting genetic information within context of your personal and family history.

A Medcan NIPT appointment includes:

- **Full, comprehensive genetic counselling** – with up-to-date information in a non-rushed environment – allowing you to make informed decisions.
- A review of the test in detail, including potential risks, benefits and limitations, and an opportunity to have all questions answered. An appointment typically lasts **45 minutes**.
- Weekend appointments available.
- An on-site blood draw (one tube of blood).
- Review of results with a genetic counselor, which will be available within **about 5 business days**. Other tests can take 2 weeks to provide results.
- In the case of a positive result, a referral to a clinic providing prenatal diagnostic services (such as CVS and amniocentesis).

**IS A REFERRAL NECESSARY TO COME IN TO MEDCAN?**

No, a physician referral is not required. An appointment can be arranged within 24 hours of the request.

**WHAT IF I LIVE OUTSIDE OF TORONTO?**

Medcan offers the convenience of a remote NIPT appointment. Please call us to learn about this option.

**HOW MUCH DOES NIPT COST?**

An NIPT costs **$825**. This includes the cost of the test and the genetic counselling session and review of results. The cost of the Microdeletion add-on is **$100**.

To book an appointment, contact our Client Service Team at 416.350.3621 or Genetics@medcan.com