PRENATAL SCREENING
DÉPISTAGE PRÉNATAL
ONTARIO

BORN Ontario

Program Report

Prenatal Screening Ontario 2019 – 2020

Prenatal Screening Ontario
1-833-351-6490 | pso@bornontario.ca
prenatalscreeningontario.ca

Table of Contents

Figures and Tables

Figures

Tables

Definition of Terms

Term	Definition
Estimated due date (EDD)	The date that spontaneous onset of labour is expected to occur – 280 days after the first day of the last menstrual period.
False positive rate (FPR)	The proportion of pregnancies unaffected by the chromosome difference that received a “screen positive” or “high risk” result.
False negative rate (FNR)	The proportion of pregnancies affected by the chromosome difference that received a “screen negative” or “low risk” result.
Negative predictive value (NPV)	The probability that a screening result reported “screen negative” or “low risk” is a true negative.
Positive predictive value (PPV)	The probability that a screening result flagged as “screen positive” or “high risk” is a true positive.
Sensitivity	Also known as detection rate, this is the ability of a test to correctly identify those that have a condition. In the context of prenatal screening, sensitivity represents the percentage of pregnancies where the baby has a chromosome difference and the screening result is flagged as “screen positive” or “high risk”.
Specificity	The ability of a test to correctly identify those that do not have a condition. In the context of prenatal screening, specificity represents the percentage of pregnancies where the baby does not have a chromosome difference and the screening result is reported as “screen negative” or “low risk”.
Suppression	As per BORN’s privacy policy, cell sizes of five (5) or less are suppressed in aggregated data in order to ensure that the information does not identify an individual and that it is not reasonably foreseeable in the circumstances that the information could be utilized to identify an individual.

Key Messages

- In the 2019 – 2020 fiscal year, the overall uptake of Multiple Marker Screening (MMS) was 69.4%. As expected, uptake of MMS by those aged 40 and over decreased as expected because of the option of OHIP-funded Non-invasive Prenatal Testing (NIPT) for this group.
- OHIP-funded Non-invasive Prenatal Testing (NIPT) is available for pregnant individuals who meet eligibility criteria. Fifty-nine percent of NIPT performed in Ontario was OHIP-funded, with the remainder being self-paid.
- The performance of prenatal screening varies by modality and condition screened. For trisomy 21, enhanced First Trimester Screening (eFTS) has a sensitivity of 88.29% and NIPT has a sensitivity of 99.49%.
- Nuchal translucency (NT) measurements in Ontario demonstrate a tendency toward chronic under-measurement. PSO implemented a formal nuchal translucency quality assurance (NTQA) program for Ontario that is supporting sonographers in their NT practice, remediating poor NT measurement accuracy, and contributing to a high-quality eFTS program.
- Although the NT ultrasound can be performed as part of eFTS, it can be also be done on its own. For those declining eFTS or opting for NIPT, an NT ultrasound is still recommended.
- PSO engages stakeholders and provides resources to the prenatal community through multiple methods, including educational webinars, facilitating expert working groups and providing an information line via phone and email.

Prenatal Screening Ontario
1-833-351-6490 | pso@bornontario.ca
prenatalscreeningontario.ca

About Us

Prenatal Screening Ontario (PSO) was created and funded by the Ministry of Health in 2017 to coordinate prenatal screening services in Ontario. PSO is Ontario's resource for patients, families and providers looking for information and updates related to prenatal screening.

PSO is housed within the province's perinatal, newborn and child registry, the Better Outcomes Registry and Network (BORN) Ontario. BORN Ontario is a prescribed registry established in Ontario under the Personal Health Information Protection Act, 2004 (PHIPA). BORN collects, interprets, shares and rigorously protects high-quality data essential to making Ontario the safest place in the world to have a baby.

This Program Report aims to highlight the pillars of the PSO program and provide measurable performance indicators of prenatal screening in Ontario. The majority of data presented in this report is for records of pregnancies with estimated due dates from April 2019 to March 31, 2020; exceptions are noted for unique data holdings and where trends are illustrated. Please consider the timeline when comparing data from differing data holdings.

PSO Mandate

- Enhance access to high quality prenatal screening for all pregnant individuals in Ontario.
- Provide the education supports, information, and transparency needed for health care providers and pregnant individuals and their families to make informed decisions.
- Undertake ongoing quality assurance and system performance evaluation to support all components of the system in functioning effectively and meeting established standards.
- Facilitate the incorporation of evolving technologies or screening options, supporting evidence-based integration.
- Support the ongoing alignment of screening service provision.

PSO Team

The PSO team is a dedicated, experienced group of individuals committed to the support of high quality prenatal screening for families in Ontario. The PSO team consists of:

- Medical Directors (MFM/Geneticist)
- Program Manager
- Genetic Counsellors
- Clinical Content Specialists (Ultrasound and Congenital Anomalies)
- Epidemiologist
- Data Analyst
- Administrative Staff

Land Acknowledgement

BORN Ontario and the PSO Program recognize that its work, and the work of its community partners take place on traditional Indigenous territories across the province. We acknowledge that there are 46 treaties and other agreements that cover the territory now called Ontario. We are thankful to be able to work and live in these territories. We are thankful to the First Nations, Metis and Inuit people who have cared for these territories since time immemorial and who continue to contribute to the strength of Ontario and to all communities across the province. BORN is honoured to collaborate with Indigenous clients, stakeholders and communities throughout the various territories.

BORN would also like to acknowledge that it is a Provincial Program operating through the Children's Hospital of Eastern Ontario (CHEO), which is located in Ottawa. Ottawa is built on un-ceded Algonquin Anishinabe territory. The peoples of the Algonquin Anishinabe Nation have lived on this territory for millennia. Their culture and presence have nurtured and continue to nurture this land.

BORN honours the peoples and land of the Algonquin Anishinabe Nation.

Acknowledging territory is only the beginning of cultivating meaningful and reciprocal relationships with First Nations, Inuit and Métis peoples and communities. In 2021, all PSO staff will be supported with cultural safety training as a means to build a foundation of humility and safety within BORN/PSO.

With Indigenous partners, BORN is developing an Indigenous engagement strategy to better inform and support the submission, use, and governance of Indigenous people's data in a respectful, culturally safe, accessible way that ensures individual privacy and confidentiality and is responsive to the needs of individuals and communities. BORN recognizes that addressing inequity, improving outcomes and facilitating care for Indigenous people starts with data. Please visit our Indigenous Wellness section of our website to learn more.

Prenatal Screening in 2019-2020

Prenatal screening is an optional test that can determine the chance that a baby may or may not have trisomy 21 (Down syndrome) or trisomy 18 (Edwards syndrome). Anyone may have a pregnancy with trisomy 21 or trisomy 18, regardless of their family history.

Prenatal screening is available to all pregnant individuals in Ontario, and can be ordered through a health care provider. Currently, two categories of screens are available in Ontario:

Multiple Marker Screening

Enhanced First Trimester Screening (eFTS) involves both a nuchal translucency (NT) ultrasound and a blood test done between 11 weeks 2 days to 13 weeks 3 days gestation. The results combined with maternal age (or age of the egg donor) at delivery gives information about the chance to have a baby with:

- Trisomy 21 (Down syndrome)
- Trisomy 18 (Edwards syndrome)

Maternal Serum Quad Screening (MSS Quad), also known as second trimester quad screening (T2Quad), involves a blood test done between 14 weeks 0 days and 20 weeks 6 days gestation. MSS can be done if screening in the first trimester was missed or in geographic areas where NT ultrasound is not available. The blood test results combined with maternal age (or age of the egg donor) at delivery gives information about the chance to have a baby with:

- Trisomy 21 (Down syndrome)
- Trisomy 18 (Edwards syndrome)

Non-Invasive Prenatal Testing

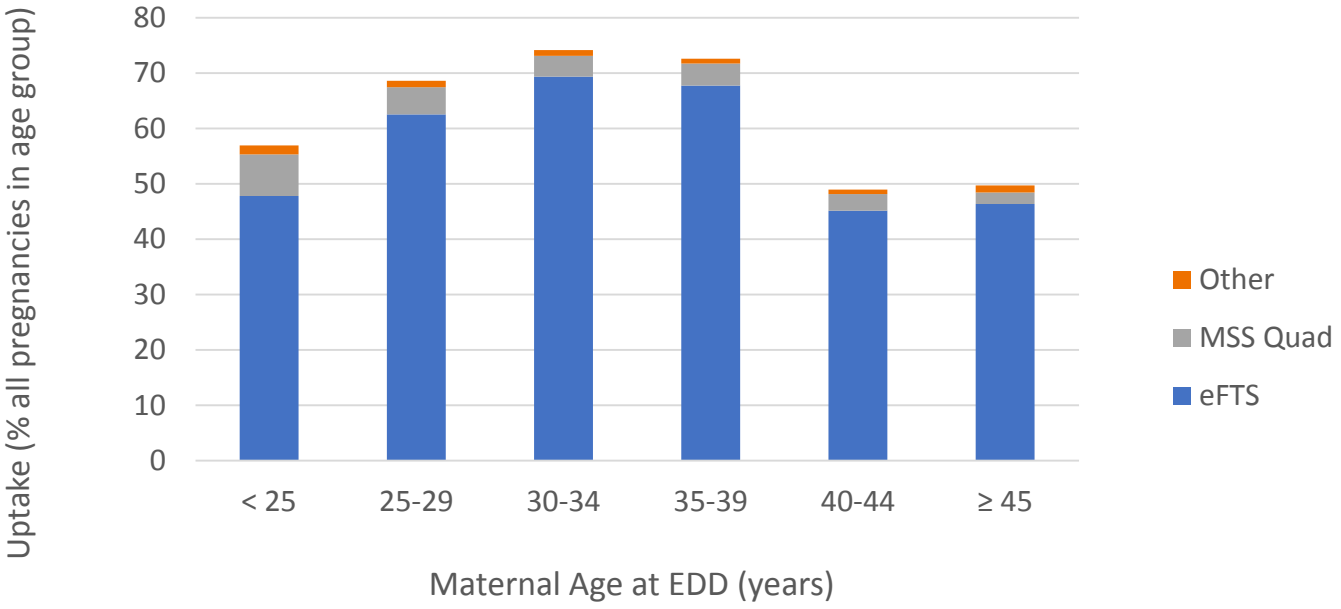
Fragments of genetic material (or DNA) from the placenta can be detected in a pregnant individual's blood stream by the end of the first trimester of pregnancy. **Non-invasive prenatal testing** (also referred to as **cell-free DNA screening**) is a blood test available from 9 or 10 weeks gestation (depending on the laboratory) until the end of the pregnancy, which analyses those DNA fragments to determine if there is a high or low chance to have a baby with:

- Trisomy 21 (Down syndrome)
- Trisomy 18 (Edwards syndrome)
- Trisomy 13 (Patau syndrome)
- +/- Sex chromosome differences

Multiple Marker Screening: Uptake by Maternal Age

In the April 2019 to March 2020 fiscal year, 146,947 pregnancies were recorded in the BORN Information System. Of those, 102,025 individuals (or 69.4%) had multiple marker screening.

Multiple marker screening is funded by the Ministry of Health for pregnant individuals of all ages. Uptake at age 40 and older decreased as expected because of the option of OHIP-funded Non-invasive Prenatal Testing (NIPT) for individuals aged 40 and older at the time of delivery.



Notes

1. The timeline for this figure was based on pregnancies with an EDD of 01-Apr-2019 to 31-Mar-2020

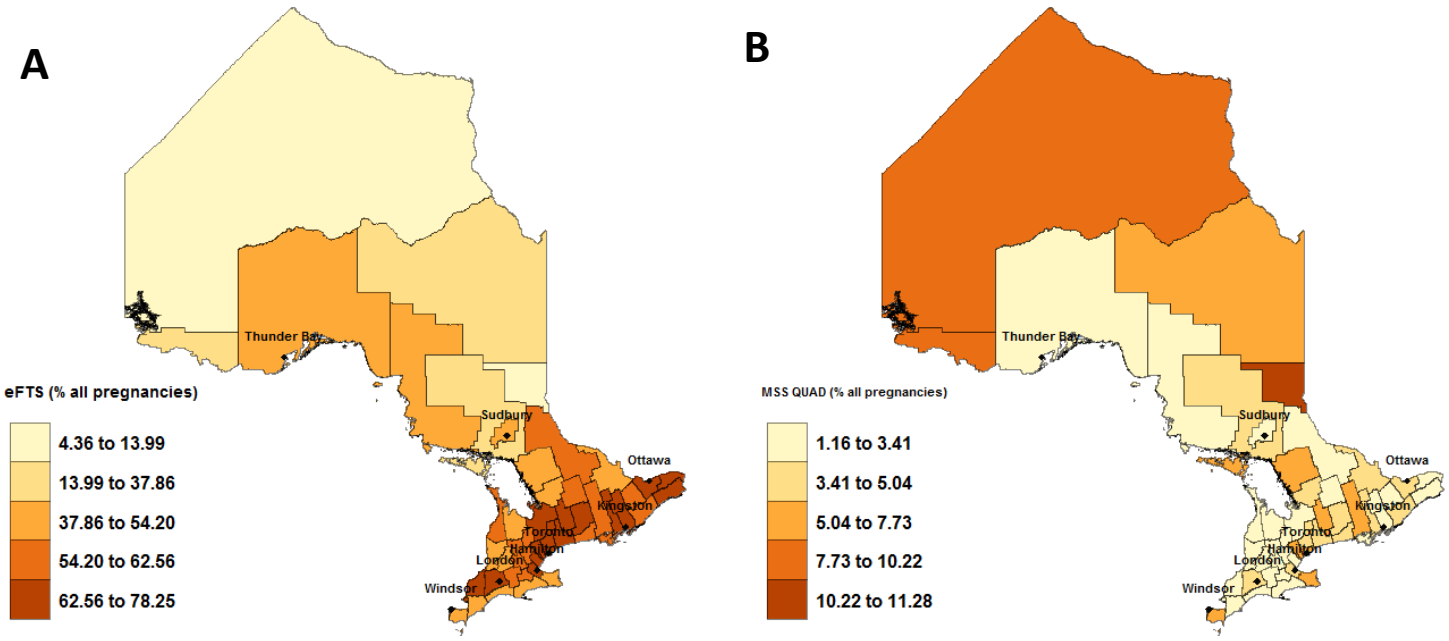
2. Only singleton pregnancies were included in this analysis

Figure 1. Multiple Marker Screening Uptake by Maternal Age.

Accessibility link: For the long description of Figure 1, see page 27

Although the NT ultrasound can be performed as part of eFTS, it can be also be done on its own. For those declining eFTS or opting for NIPT, an NT ultrasound is still recommended.

Multiple Marker Screening: Uptake by Geography



Notes

1. *These maps include all pregnancies who received A) eFTS or B) MSS Quad in Ontario, expressed as a percentage of all pregnancies in Ontario per census division*
2. *The timeline for this table was based on pregnancies with an EDD of 01-Apr-2019 to 31-Mar-2020*
3. *Only singleton pregnancies were included in this analysis*
4. *Census division could not be assigned for 12.9% of pregnancies*
5. *A total of 122,879 pregnancies were included in this analysis*
6. *A total of A) 79,292 pregnancies received eFTS in this analysis and B) 5517 pregnancies received MSS Quad in this analysis*

Figure 2. Multiple Marker Screening Uptake by Geography. Map A – eFTS; map B – MSS Quad.

Accessibility link: For the long description of Figure 2, see page 28

There were differences observed in the uptake of prenatal screening across Ontario. Overall, 69.4% of pregnancies utilized some form of multiple marker screening but the specific uptake of eFTS ranged from as low as 4.36% in the Northern Ontario census divisions of Kenora and Temiskaming to as high as 78.25% in the urban areas surrounding Toronto, Kingston, London and Ottawa.

MSS Quad is a less commonly utilized prenatal screening modality, with uptake never exceeding 11.28% of pregnancies in Ontario. However regional differences are apparent as well, with MSS Quad being utilized more commonly in Kenora and Temiskaming.

Future work of PSO includes examination of possible reasons for the geographic variance in uptake to ensure there is equitable access to high quality prenatal screening across the province.

Multiple Marker Screening: Performance

Multiple marker screening is not diagnostic; rather, it tells the pregnant individual about the likelihood of a baby having trisomy 21 or trisomy 18. For the time period of this Program Report, a calculated likelihood of 1 in 350 or greater for trisomy 21 and 1 in 200 or greater for trisomy 18 is reported as screen positive by eFTS and a likelihood of 1 in 200 or greater is considered screen positive by MSS Quad.

The performance of multiple marker screening is assessed through the comparison of screening results to diagnostic cytogenetic (chromosome testing) results and birth outcomes. eFTS has a higher sensitivity than MSS Quad overall, however, more precise data on MSS Quad is unavailable due to the lower number of MSS Quad screens performed in Ontario. Definitions of these performance metrics can be found on page 5.

Enhanced First Trimester Screening

Table 1. Enhanced First Trimester Screening Performance for Singleton Pregnancies.

Condition	Sensitivity (95% CI)	Specificity (95% CI)	PPV (95% CI)	NPV (95% CI)	FPR (95% CI)	FNR (95% CI)
Trisomy 21	88.29 (85.52,90.69)	93.66 (93.56,93.76)	3.65 (3.36,3.96)	99.97 (99.96,99.97)	6.34 (6.24,6.44)	11.71 (9.31,14.48)
Trisomy 18	83.96 (77.90,88.91)	99.76 (99.73,99.78)	21.63 (18.68,24.80)	99.99 (99.98,99.99)	0.24 (0.22,0.27)	16.04 (11.09,22.10)

Maternal Serum Screening - Quad

Table 2. Maternal Serum Screening Quad Performance for Singleton Pregnancies.

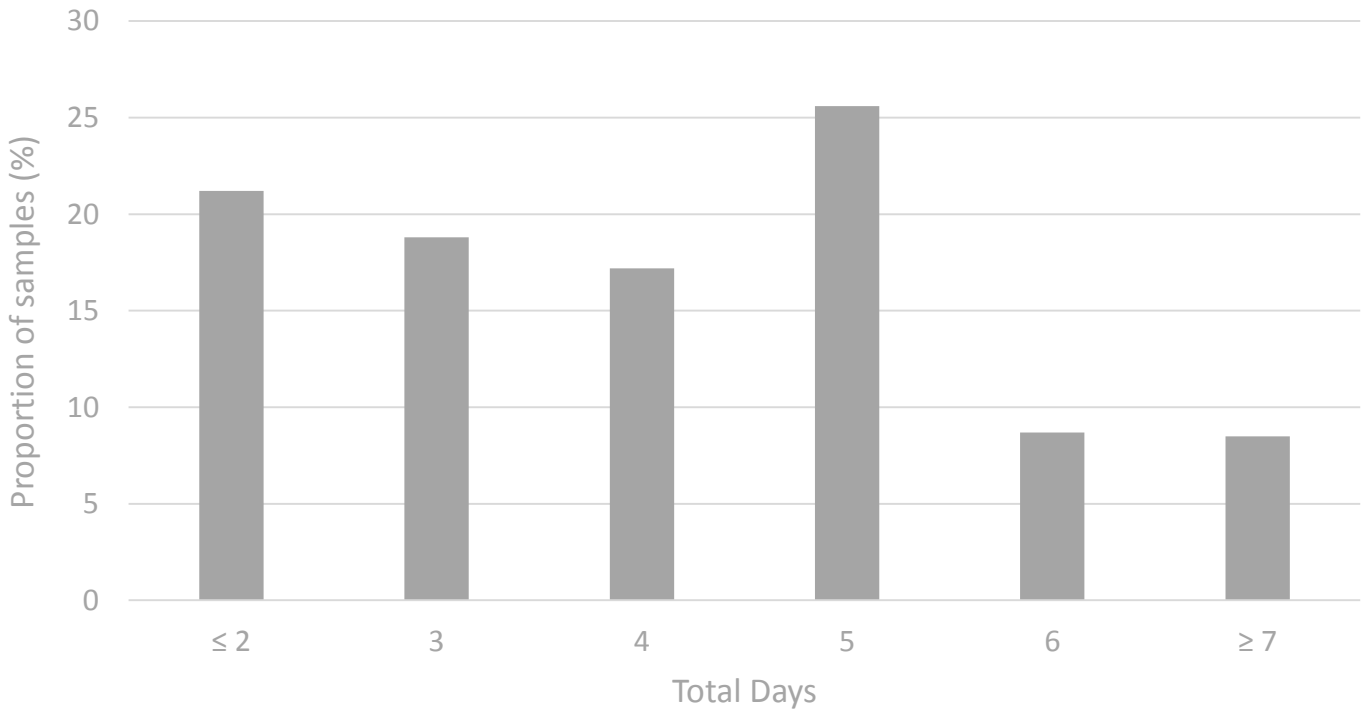
Condition	Sensitivity (95% CI)	Specificity (95% CI)	PPV (95% CI)	NPV (95% CI)	FPR (95% CI)	FNR (95% CI)
Trisomy 21	S (59.66, 86.81)*	94.66 (94.35,94.95)	2.74 (1.89, 3.83)	99.95 (99.91,99.97)	5.34 (5.05, 5.65)	S (13.19, 40.34)*
Trisomy 18	S (38.57, 90.91)*	99.44 (99.33,99.53)	6.82 (3.16,12.55)	99.98 (99.95,100.00)	0.56 (0.47, 0.67)	S (9.09, 61.43)*

Notes

1. *The timeline for these tables was based on singleton pregnancies with an EDD of 01-Sep-2016 to 31-June-2020*
2. *S = point estimate suppressed when confidence interval > 20%*
3. ** = performance data have a confidence interval greater than 20%. These performance metrics were calculated using small cell sizes from the available MMS and cytogenetic data in the BIS and are subject to change as more data are collected. Please interpret these data with caution.*
4. *PPV: positive predictive value; NPV: negative predictive value; FPR: false positive rate; FNR: false negative rate*
5. *"eFTS" includes both 4-marker and 5-marker eFTS*
6. *Uninterpretable, inconclusive, mosaic and partial cytogenetic results were excluded from this cohort*
7. *Outcome data for autosomes screened (chromosomes 21 and 18) were supplemented using data from the BORN information system (BIS) for negative results only, where the outcomes for pregnancies with no cytogenetic outcome were set to test-negative when their corresponding BIS record had no indication for the disorder during the perinatal period*

Multiple Marker Screening: Turnaround Times

Blood specimens for multiple marker screening are collected at hospital and community blood labs and the analysis is performed at one of three provincial hospital-based labs: Trillium Health Partners, Mount Sinai Hospital and North York General Hospital. Results are shared with provides within approximately 5 business days. Data collected by BORN show that the median turnaround time is 4 total days from the date of sample collection to the date the test report is issued.



Notes

1. *The timeline for this **figure** was based on pregnancies with an EDD of 01-Apr-2019 to 31-Mar-2020*
2. *Only singleton pregnancies were included in this analysis*

Figure 3. Multiple Marker Screening Turnaround Times.

Accessibility link: For the long description of Figure 3, see page 30

NIPT: Eligibility Criteria for OHIP-Funded NIPT in 2019-2020

NIPT is available to all pregnant individuals in Ontario, and is covered by OHIP if one of the following funding criteria* is met. For those meeting category II criteria, however, a consultation with a genetics or maternal fetal medicine specialist is required for a thorough discussion of the risk factors and appropriateness of NIPT. Individuals who do not meet any of the criteria can opt to pay for the testing themselves.

Category I criteria (can be ordered by any physician or nurse practitioner for singletons)

- A positive prenatal screening result from multiple marker screening for this pregnancy
- The maternal age will be 40 years or older at the expected date of delivery
    - in the context of in vitro fertilization, the maternal age is guided by the age at egg retrieval (whether own egg or donor egg)
- The nuchal translucency (NT) measurement is ≥3.5mm
- There is a personal history of a previous pregnancy or child with trisomy 21, 18 or 13

Category II criteria (must be ordered by a genetics or maternal-fetal medicine specialist for singleton and twin pregnancies)

- There are findings on ultrasound which are associated with an increased chance for trisomy 21, trisomy 18 or trisomy 13
- There is a chance for a sex-linked genetic condition
- The ultrasound shows findings suggestive of a sex chromosome difference
- The ultrasound shows findings suggestive of a disorder of sex determination

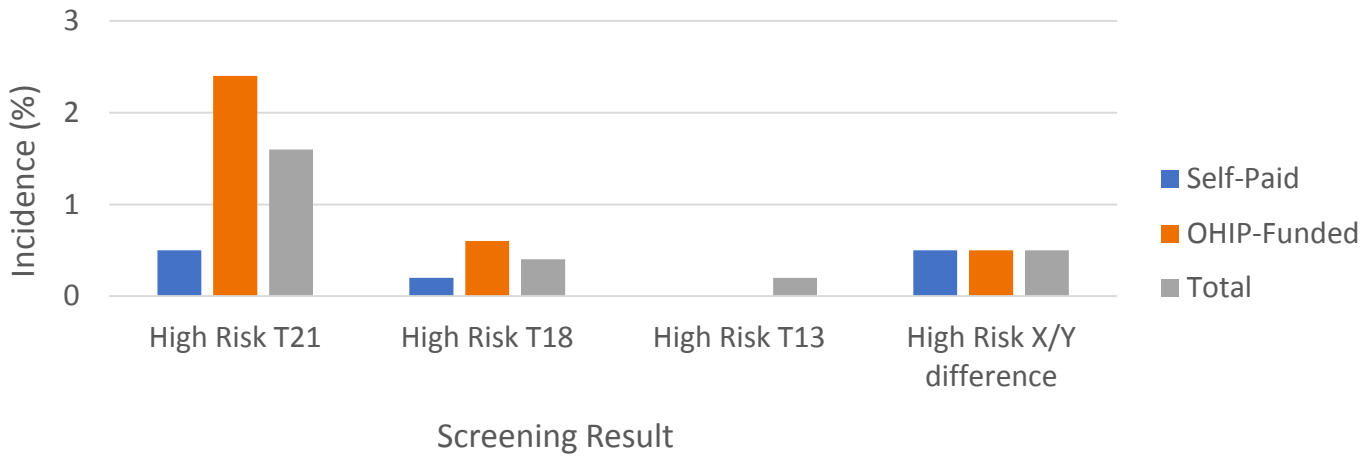
*This is the funding criteria that was in place pre-COVID19. Please visit the PSO website for current eligibility criteria.

NIPT: Uptake and Results by Funding Status

There were a total of 17,802 NIPT screens ordered in the 2019-2020 fiscal year, 59% of which were funded by OHIP while 41% were self-paid by the pregnant individual. The incidence of a high risk NIPT result was higher in the OHIP-funded group for trisomy 21, trisomy 18 and trisomy 13. The incidence of a high risk result for sex chromosome differences was 0.5% for both self-paid and OHIP-funded screens.

Table 3. NIPT Volumes and Results by Funding Status.

Funding status	Number of Screens	Number Reported as High Risk T21 (%)	Number Reported as High Risk T18 (%)	Number Reported as High Risk T13 (%)	Number Reported as High Risk X/Y difference (%)
Self-Paid	7,299	37 (0.5)	14 (0.2)	S	34 (0.5)
OHIP Funded	10,503	256 (2.4)	63 (0.6)	S	52 (0.5)
Total	17,802	293 (1.6)	77 (0.4)	35 (0.2)	86 (0.5)



Notes

1. *The timeline for this table was based on pregnancies with an EDD of 01-Apr-2019 to 31-Mar-2020*
2. *Only singleton pregnancies were included in this analysis*
3. *Clinical indicators for funding status are mutually exclusive*
4. *S = data suppressed due to small cell size, or suppressed to prevent back-calculation of another cell suppressed due to small cell size*

Figure 4. Incidence of High Risk NIPT Results by Funding Status.

Accessibility link: For the long description of Figure 4, see page 30

NIPT: Uptake by Maternal Age

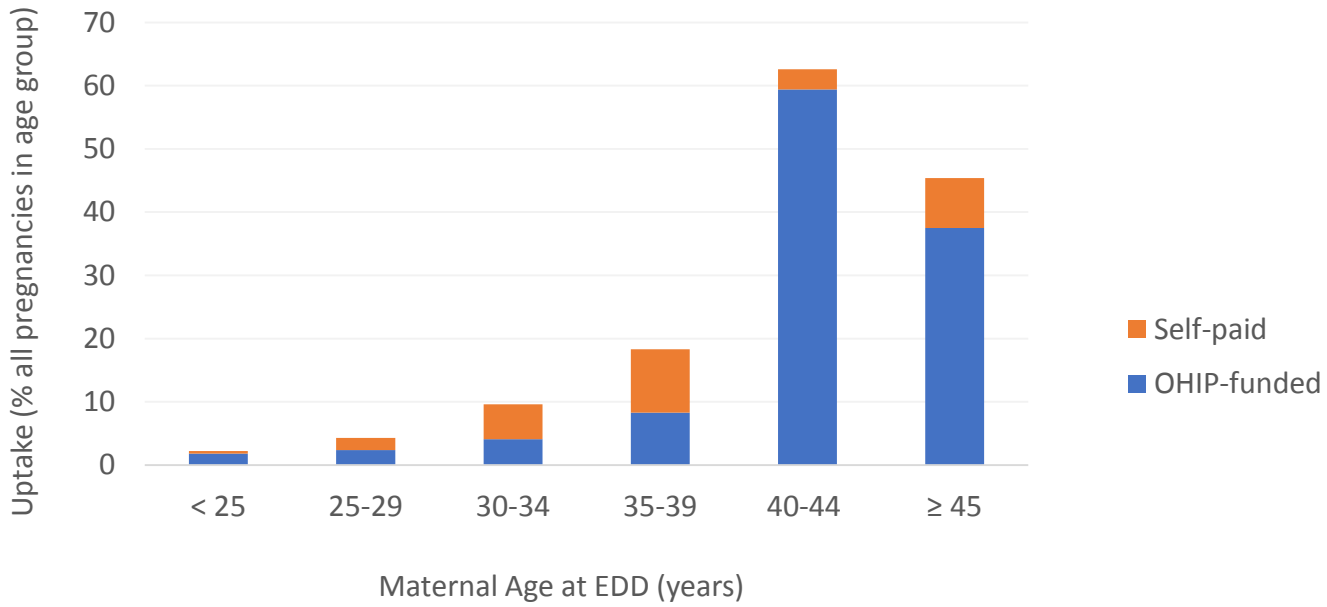
The proportion of pregnancies that are eligible for and elect to have NIPT generally increases with maternal age. This trend does not appear to persist at or above a maternal age of 45.

In the 30 to 34.9 year age group, 4.1% of pregnant individuals utilized OHIP-funded NIPT while an additional 5.5% opted for self-paid NIPT. In comparison, 10.0% of pregnant individuals ages 35 to 39.9 opted for self-paid NIPT.

In the same time period, there were 7,621 pregnancies reported in the 40 years and older age group and all of these pregnancies were eligible for OHIP-funded NIPT as per the criteria; 58% of these pregnancies had NIPT. An additional 3.2% of pregnant individuals in this age group self-paid for NIPT despite meeting OHIP eligibility criteria. While prenatal screening is optional, the number of self-paid tests in this age group highlights a need for continued health-care provider education and support to ensure individuals are receiving OHIP-funded services when eligible.

Table 4. Number of Pregnancies by Maternal Age.

Age Group	< 24.9	25 – 29.9	30 – 34.9	35 – 39.9	40 – 44.9	≥ 45
Number of pregnancies	14,415	37,341	55,813	31,757	7,138	483



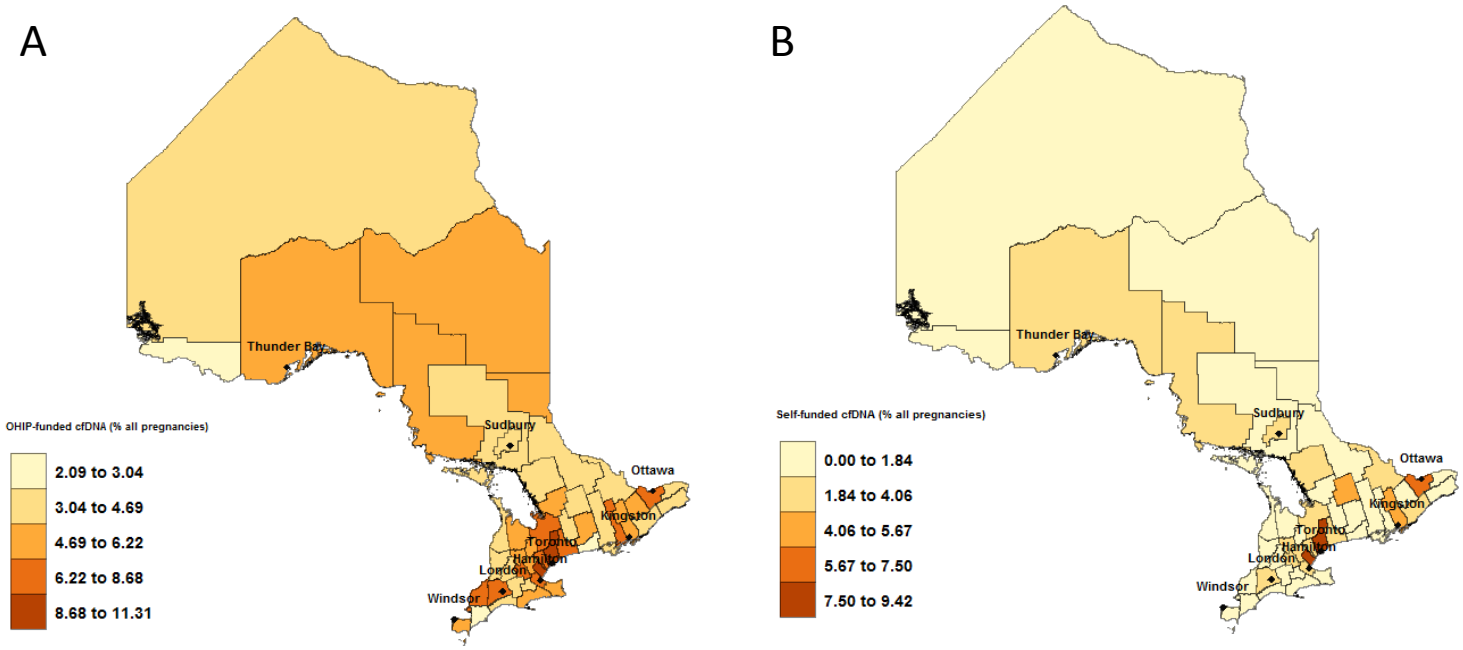
Notes

1. *The timeline for this table and figure was based on pregnancies with an EDD of 01-Apr-2019 to 31-Mar-2020*
2. *Only singleton pregnancies were included in this analysis*

Figure 5. NIPT Uptake by Maternal Age.

Accessibility link: For the long description of Figure 5, see page 31

NIPT: Uptake by Geography



Notes

1. *These maps include all pregnancies who received A) OHIP-funded NIPT in Ontario and B) Self-funded NIPT in Ontario, expressed as a percentage of all pregnancies in Ontario*
2. *The timeline for this table was based on pregnancies with an EDD of 01-Apr-2019 to 31-Mar-2020*
3. *Only singleton pregnancies were included in this analysis*
4. *Census division could not be assigned for 13.0% of pregnancies*
5. *A total of 122,879 pregnancies were included in this analysis*

Figure 6. NIPT Uptake by Geography. A – OHIP-funded NIPT; B – Self-paid NIPT.

Accessibility link: For the long description of Figure 6, see page 32

Geographic variance was also seen in the uptake of NIPT. Map A illustrates the proportion of pregnancies that were eligible for and subsequently elected to have OHIP-funded NIPT. This value ranged from 2.09% to 11.31%. A total of 9073 pregnancies received OHIP-funded NIPT in this analysis.

Map B illustrates the proportion of all pregnancies in a census division that elected for self-paid NIPT. Pregnant individuals in Toronto, York and Halton had the highest uptake, ranging from 7.5 to 9.42% of all pregnancies. In most other regions of Ontario a relatively low proportion of pregnant individuals elected for self-paid NIPT. A total of 5883 pregnancies received self-funded NIPT in this analysis.

NIPT: Uptake and Results by Indication for OHIP Funding

The table below summarizes the types of high risk results received from OHIP-funded NIPT. Only the funding categories with the highest volumes of screens are shown. PSO data show that the most common indication for OHIP-funded NIPT is a positive multiple marker screen, followed by maternal age greater than or equal to 40 years.

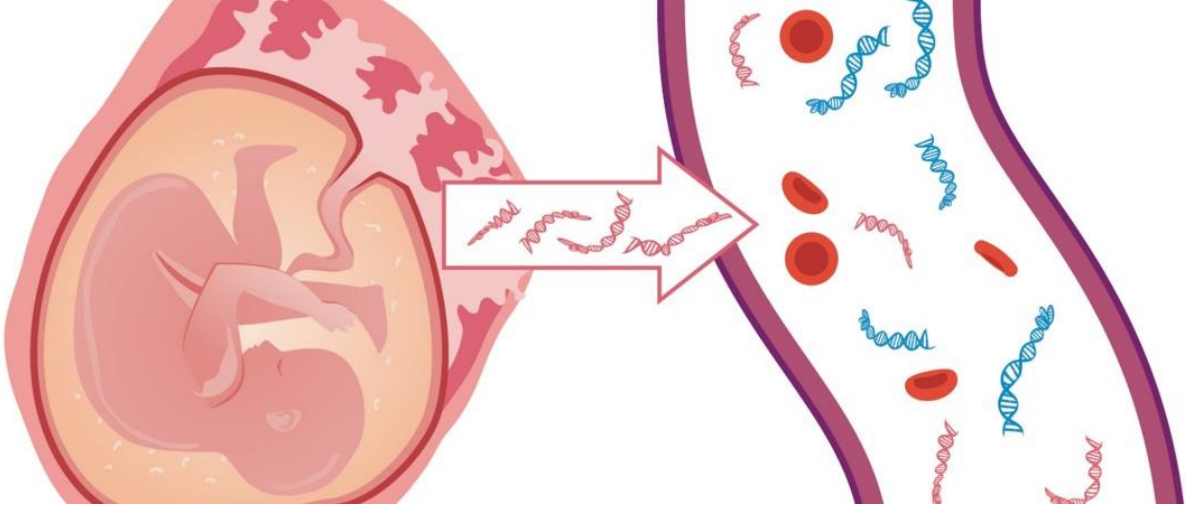
The likelihood of receiving a high risk NIPT result was lowest for pregnant individuals with a previous pregnancy with trisomy 21, 18 or 13. The overall positive rate was 1.3%, however, individual data for these trisomies is suppressed due to small cell size (numbers less than six). The likelihood of receiving a high risk NIPT result was highest for pregnant individuals with an increased fetal nuchal translucency measurement. The overall positive rate was 20.5%; for trisomy 21 specifically, the positive rate was 15.2%. The likelihood of receiving a high risk NIPT result was 3.7% for those of advanced maternal age and 2.8% for those with a positive multiple marker screen.

Table 5. NIPT Volumes and Results for Common OHIP-Funding Indications.

Indication for Funding	Number of screens	Number Reported as High Risk T21 (%)	Number Reported as High Risk T18 (%)	Number Reported as High Risk T13 (%)	Number Reported as High Risk X/Y difference (%)
Maternal Age ≥ 40	3,903	86 (2.2)	30 (0.8)	12 (0.3)	16 (0.4)
Positive multiple marker screen	4,221	85 (2.0)	14 (0.3)	6 (0.1)	16 (0.4)
Increased NT	132	20 (15.2)	S	S	S
Previous trisomy 21, 18 or 13	528	S	S	0 (0.0)	S
Fetal anomalies	198	6 (3.0)	S	S	S
Risk greater than a positive multiple marker screen result	684	14 (2.0)	S	S	S

Notes

1. *The timeline for this table was based on pregnancies with an EDD of 01-Apr-2019 to 31-Mar-2020*
2. *Only singleton pregnancies were included in this analysis*
3. *Clinical indicators for funding status are mutually exclusive. Screens ordered with multiple clinical indicators (e.g. positive multiple marker screen AND increased NT) are not included in the table*
4. *S = data suppressed due to small cell size, or suppressed to prevent back-calculation of another cell suppressed due to small cell size*



NIPT Performance

NIPT is a more accurate screening test than multiple marker screening but is still not a diagnostic test. The performance of NIPT in Ontario is assessed through the comparison of multiple marker screening results to diagnostic cytogenetic (chromosome testing) results and birth outcomes. The table below details the performance metrics for NIPT for singleton pregnancies in Ontario performed at Dynacare® and LifeLabs®.

Table 6. NIPT Performance for Singleton Pregnancies.

Result	Sensitivity (95% CI)	Specificity (95% CI)	PPV (95% CI)	NPV (95% CI)	FPR (95% CI)	FNR (95% CI)
Trisomy 21	99.49 (98.71,99.86)	99.93 (99.90,99.95)	95.50 (93.86,96.82)	99.99 (99.98,100.00)	0.07 (0.05,0.10)	0.51 (0.14,1.29)
Trisomy 18	96.26 (92.44,98.48)	99.97 (99.95,99.98)	92.31 (87.63,95.63)	99.99 (99.97,99.99)	0.03 (0.02,0.05)	3.74 (1.52,7.56)
Trisomy 13	90.91 (80.05,96.98)	99.96 (99.93,99.97)	S (56.56, 78.87)*	99.99 (99.98,100.00)	0.04 (0.03,0.07)	9.09 (3.02,19.95)

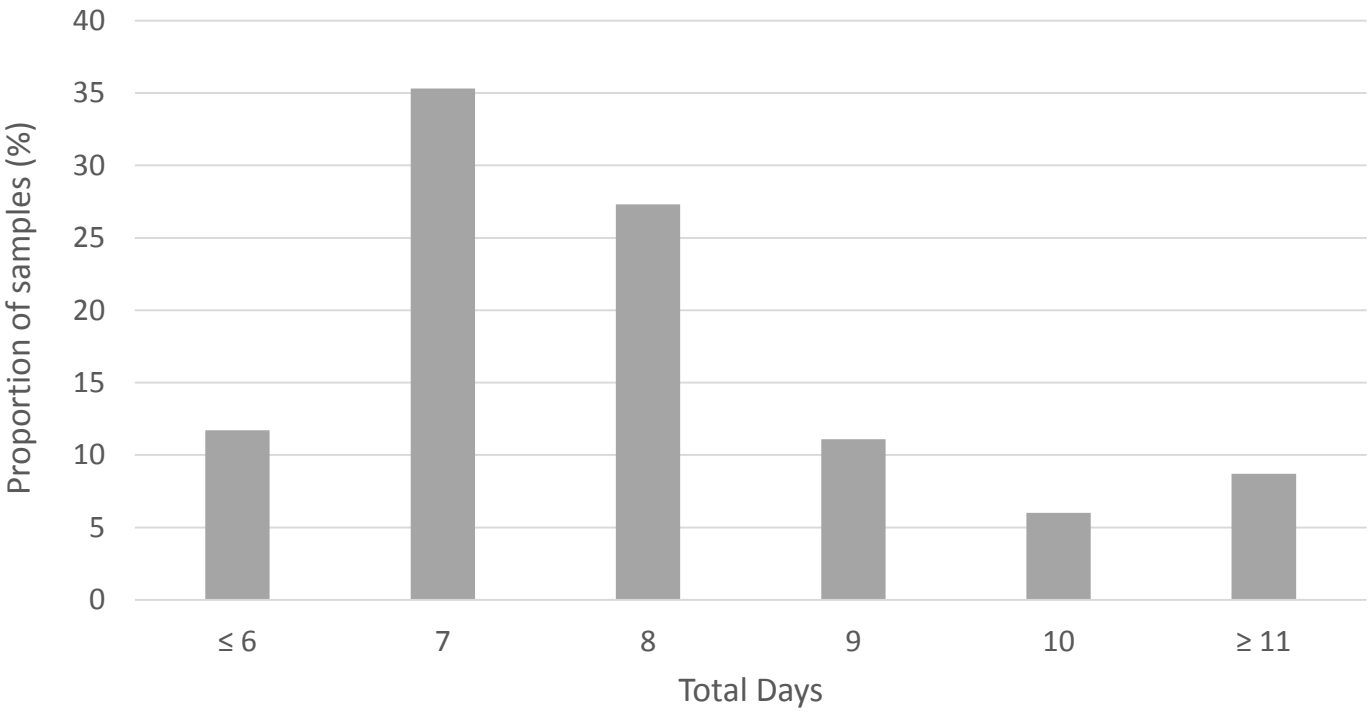
Notes

1. *The timeline for this table was based on singleton pregnancies with an EDD of 01-Sep-2016 to 30-Jun-2020*
2. *S = point estimate suppressed due to a confidence interval > 20%*
3. ** = performance data included in this release have a confidence interval greater than 20%. These performance metrics were calculated using small cell sizes from the available NIPT and cytogenetic data in the BIS and are subject to change as more data are collected. Please interpret these data with caution.*
4. *No-call and missing data screening results were excluded from this analysis*
5. *Uninterpretable, inconclusive, mosaic and partial cytogenetic results were excluded from this cohort*
6. *Outcome data for autosomes screened (chromosomes 21, 18, and 13) were supplemented using data from the BORN information system (BIS) for negative results only, where the outcomes for pregnancies with no cytogenetic outcome were set to test-negative when their corresponding BIS record had no indication for the disorder during the perinatal period*

NIPT Turnaround Times

Blood specimens are collected at Dynacare® and LifeLabs® locations across Ontario and performed at licensed, accredited and regulated facilities in Ontario. Dynacare® states that results are available within 10 business days from the time of sample collection. LifeLabs® states that that results are available within seven to 10 business days from the time that the sample is received by the laboratory.

BORN data show that the median turnaround time is eight days from the date of sample collection to the date the test report is issued (Figure 7) and six days from the date of sample receipt by the laboratory to report.



Notes

1. *The timeline for this figure was based on pregnancies with an EDD of 01-Apr-2019 to 31-Mar-2020*
2. *Only singleton pregnancies were included in this analysis*
3. *This table excludes all NIPT records with a data quality issue reported*

Figure 7. NIPT Turnaround Times.

Accessibility link: For the long description of Figure 7, see page 34

Where To Have An NT Ultrasound

Nuchal translucency (NT) measurements have been used since the 1990's to assist with the identification of pregnancies with Down syndrome. Nuchal translucency ultrasounds are performed by physicians and sonographers who have obtained NT certification from the Fetal Medicine Foundation (FMF UK). As of 2020, PSO recorded 313 facilities in Ontario that were able to offer this specialized ultrasound to pregnant individuals.

An interactive map is available on the PSO website to aid providers and patients in finding their nearest ultrasound facility that offers NT ultrasound. Please contact us if you are a facility offering NT ultrasound and would like to be included in, or removed from, the map on our website.

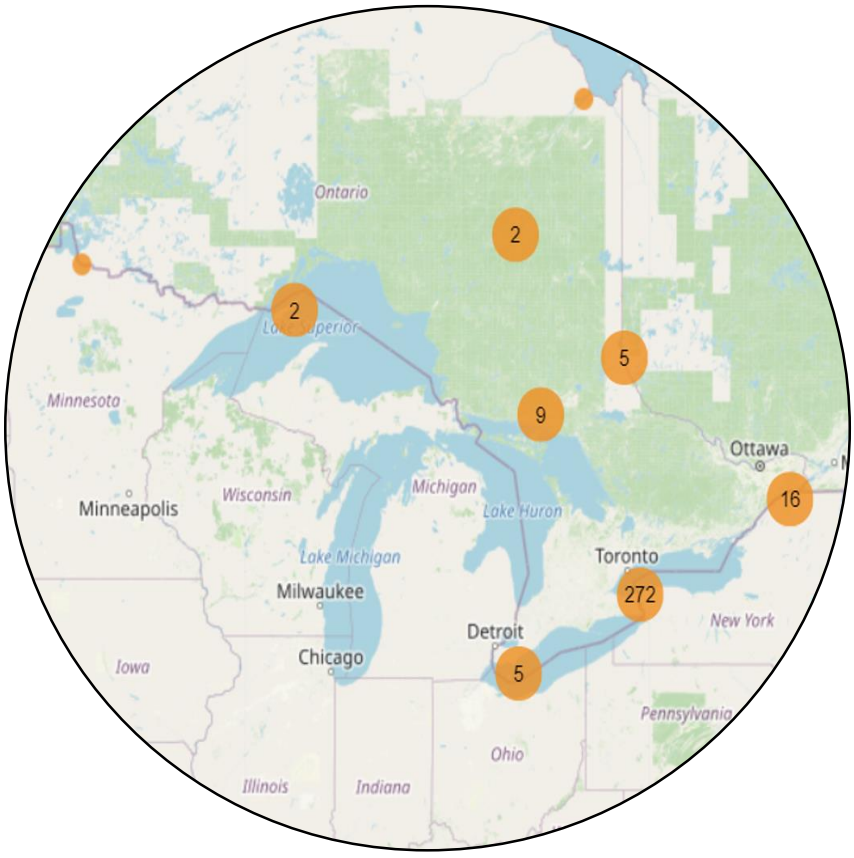
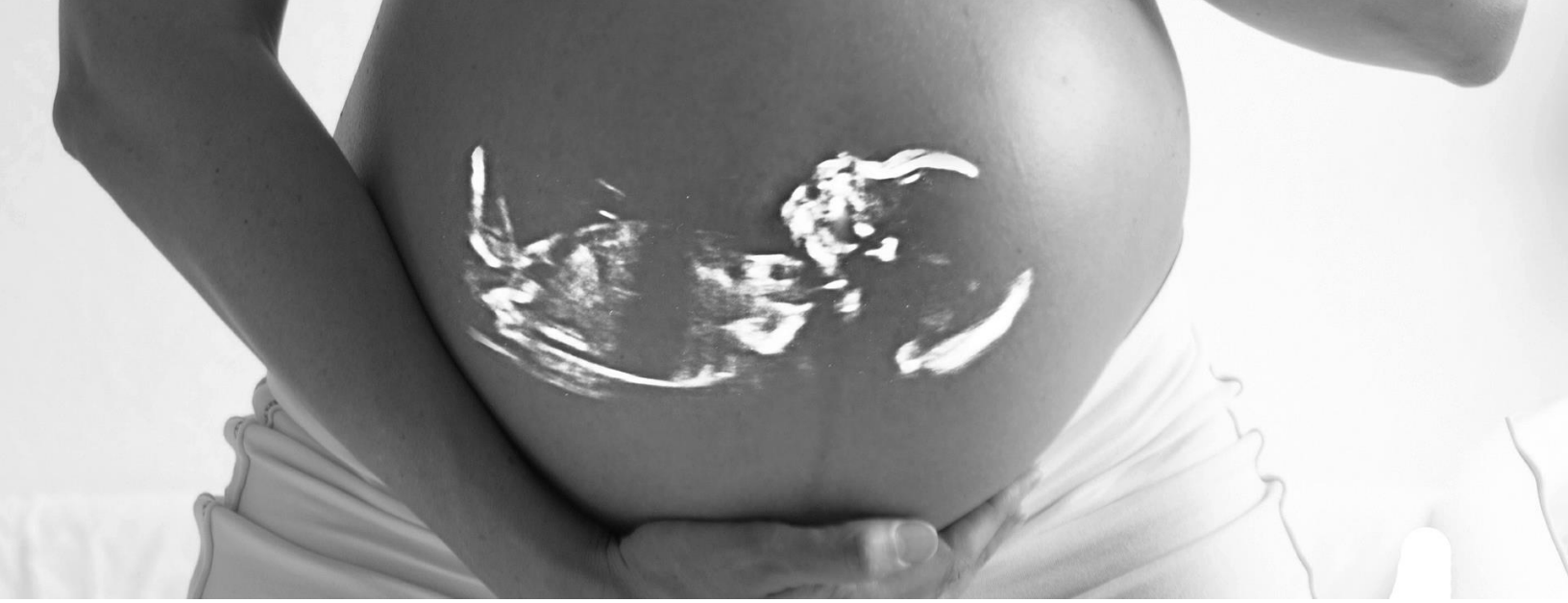


Figure 8. Map of ultrasound facilities in Ontario offering nuchal translucency ultrasound.

Accessibility link: For the long description of Figure 8, see page 34.



Nuchal Translucency Quality Assurance

Since 2012, BORN Ontario has been collecting NT ultrasound measurement data as recorded on pregnant individuals' eFTS requisitions. BORN used this data to map individualized distributions of NT measurements over the eFTS gestational age window for each sonographer. These NT performance distributions plot sonographers' cumulative NT data against the Fetal Medicine Foundation (FMF UK) standardized population curve, illustrating their NT measurement accuracy and helping to inform their practice.

Provincial audits have demonstrated a need for NT quality assurance (NTQA). There is a consistent discrepancy between sonographers' NT performance distributions and the FMF UK standardized curve with a tendency toward chronic under-measurement. Chronic under-measurement could reduce the detection rate of first trimester screening.

The PSO NTQA Program

Having identified the need for NT quality assurance, PSO implemented a formal program for Ontario that is supporting sonographers in their NT practice, remediating poor NT measurement accuracy, and contributing to a high quality eFTS program.

For more detailed information on NT certification and quality assurance in the province, please visit the "For Sonographers" section of the new PSO website.

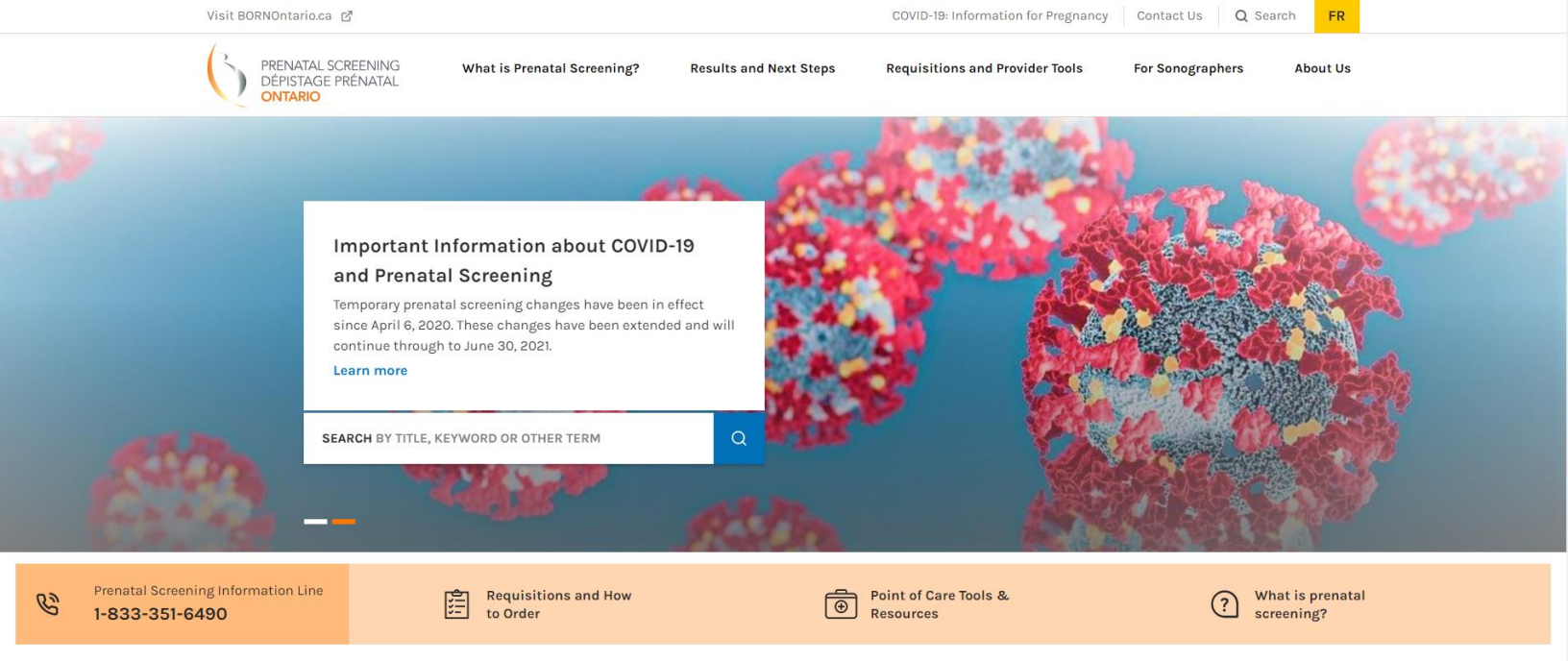
1522

Sonographers registered in the NTQA program

PSO Committees and Working Groups

PSO depends on input from experts and advisors from relevant fields to contribute to an ongoing optimal operational program. PSO has four groups engaged to facilitate stakeholder input and advice on important operational areas. Our working groups consist of a variety of dedicated individuals from varied specialties within the prenatal screening community to best enable the work to be done in a positive and effective way. Some of our completed projects this year include:

Advisory Committee	• Purpose: Advise PSO and the MOH on new conditions amenable to systematic population screening • Example: Submitted a request to Health Quality Ontario for a Health Technology Assessment for first trimester preeclampsia screening
Genetics Working Group	• Purpose: Advise on clinical issues related to prenatal screening for genetic conditions • Example: Introduced standardized multiple marker screening requisition with CRL cut-off in line with international guideline • Example: Improved language on NIPT requisition
Nuchal Translucency Quality Assurance Working Group	• Purpose: Ongoing strategy surrounding implementation and impact of provincial NTQA program on clinical practice for sonographers • Example: Advised on point-of-care tools and education tools to support sonographers in meeting NTQA requirements
Education Working Group	• Purpose: Advise in the development of educational strategies • Example: Advised on the content of the new PSO website • Example: Created three point-of-care tools to support conversations between primary care providers and patients



Communications

New Website

Creation of PSO's new website took place over the summer of 2019, with the launch of the website on August 30, 2019. Since the launch of the new site, PSO has averaged approximately 2000+ page views per week from users worldwide. From 2019 – 2020, the website was used by 52,548 individuals.

PSO Client Support

PSO has an email inbox and toll-free phone line (1-833-351-6490) that provides pregnant individuals, their families, health care providers and sonographers with direct access to:

- Genetic counsellors for questions related to prenatal screening results and navigating the prenatal screening system
- Ultrasound clinical content specialists for questions concerning NT ultrasound and NT quality assurance

Education and Outreach

Provider Tools

Printable point-of-care tools are available on the PSO website and serve as useful guides for patients and providers. Additionally, a presentation slide deck was made available in 2020 containing in-depth information about prenatal screening in Ontario for those who wish to learn on their own.

Private In-services

Our genetic counsellors and NT clinical content specialists are available for educational sessions and can tailor the content and platform to the specific needs of various health care provider groups. **Please contact pso@bornontario.ca to set up an educational session for your workplace!**

The list below highlights education sessions provided by genetic counsellors in fiscal year 2019-2020:

- August 16, 2019: Toronto Birth Centre
- September 18, 2019: Kingston, Frontenac, Lennox & Addington Public Health Unit
- October 3, 2019: City of Lakes Family Health Team
- November 14, 2019: Community Health Centre in Temiskaming
- November 21, 2019: GTA LHIN meeting for nurse practitioners
- December 5, 2019: Alongside Midwifery Unit at the Markham Stouffville Hospital
- January 16, 2020: Generations Midwifery Care in Ottawa
- January 20, 2020: Niagara North Family Health Team

Conferences and Webinars

Our genetic counsellors and NT clinical content specialists presented at numerous educational meetings, conferences and forums across Canada to increase awareness about prenatal screening best practices.

- April 8–10, 2019: Booth at the 68th Annual Refresher Course for Family Physicians
- April 26–27, 2019: NTQA presentation at the inaugural Wavelengths symposium hosted by The Michener Institute
- May 6–8, 2019: Booth at the Association of Ontario Midwives Annual General Meeting
- May 8, 2019: NTQA webinar hosted by the Ontario Association of Radiology Managers
- June 7, 2019: Presentation at the University of Toronto/Mount Sinai Hospital's course called Pregnancy and Birth: A Refresher for Primary Care Providers
- June 26, 2019: NTQA webinar hosted by Sonography Canada
- September 19–20, 2019: Booth at the Association of Family Health Teams of Ontario annual conference
- October 30–November 2, 2019: Joint presentation (with GEC-KO and the British Columbia Prenatal Screening Program) at the College of Family Physicians of Canada Family Medicine Forum
- November 28–29, 2019: Joint presentation (with GEC-KO) at the Ontario College of Family Physicians annual scientific meeting
- May 13, 2020: Presentation at the 28th Annual Symposium –New Developments in Prenatal Diagnosis and Medical Genetics

Accessibility Links: Long Descriptions

Figure 1 is a stacked bar graph with the y-axis representing the uptake of multiple marker screening, expressed as a percent of all pregnancies in the specific age group. Six bars on the x-axis present the maternal age groups at the estimated date of delivery. The six age groups are: less than 25, 25 to 29, 30 to 34, 35 to 39, 40 to 44, and 45 and greater.

Each bar is composed of three distinctly coloured, stacked sections. The first section, beginning at the x-axis illustrates the uptake of eFTS. Stacked on top is a section illustrating the uptake of MSS Quad and above that is a section illustrating the uptake of other modalities of prenatal screening. The height of the completed stack represents the uptake of all multiple marker screening for that maternal age group. Data are summarized in Table 1.

Table 1. Multiple marker screening uptake by maternal age, PSO 2019-2020

Age (Years)	Uptake of eFTS (%)	Uptake of MSS Quad (%)	Uptake of other multiple marker screens (%)	Total uptake (%)
< 25	47.8	7.5	1.6	56.9
25 to 29	62.5	4.9	1.2	68.6
30 to 34	69.4	3.8	1.0	74.2
35 to 39	67.8	4.0	0.9	72.6
40 to 44	45.1	3.1	0.8	49.0
≥ 45	46.4	2.1	1.2	49.7

Return to Figure 1 on page 10.

Figure 2 is a set of two maps of Ontario representing the uptake of multiple marker screening, expressed as a percent of all pregnancies in a specific geographical region. Both Map A and Map B are divided into Statistics Canada's 49 census divisions. There are uptake values are grouped into five ranges and coloured on a gradient with the lightest yellow representing the lowest uptake range and the darkest orange representing the highest uptake range.

Map A illustrates the uptake of eFTS. Data are summarized in Table 2A. Map B illustrates the uptake of MSS Quad. The uptake was in the lowest ranges, below 5.04% for the majority of the census divisions in Ontario. The highest uptake range was 10.22 to 11.28% and this was observed in Timiskaming. Data are summarized in Table 2B.

Table 2A. eFTS uptake by census division, PSO 2019-2020

Uptake Range (%)	Census Division
4.36 to 13.99	54 - Temiskaming 60 – Kenora
13.99 to 37.86	51 - Manitoulin 52 - Sudbury 56 - Cochrane 59 – Rainy River
37.89 to 54.20	26 - Niagara 28 – Haldimand-Norfolk 31 - Perth 34 - Elgin 36 – Chatham-Kent 37 - Essex 40 - Huron 42 - Grey 44 – Muskoka 47 - Renfrew 49 – Parry Sound 53 – Greater Sudbury 57 – Algoma 58 – Thunder Bay
54.20 to 62.56	7 – Leeds and Grenville 9 – Lanark 12 - Hastings 13 – Prince Edward 14 - Northumberland 22 – Dufferin 23 – Wellington 29 – Brant 30 – Waterloo 32 – Oxford 41 – Bruce 46 – Haliburton 48 – Nipissing
62.56 to 78.25	1 – Stormont, Dundas and Glengarry 2 – Prescott and Russell 6 – Ottawa 10 – Frontenac 11 – Lennox and Addington 15 – Peterborough 16 – Kawartha Lakes 18 – Durham 19 – York 20 – Toronto 21 – Peel 24 – Halton 25 - Hamilton 38 – Lambton 39 – Middlesex 43 – Simcoe

Table 2B. MSS Quad uptake by census division, PSO 2019-2020

Uptake Range (%)	Census Division
1.16 to 3.41	1 – Stormont, Dundas and Glengarry 2 – Prescott and Russell 9 – Lanark 10 – Frontenac 13 – Prince Edward 22 – Dufferin 23 – Wellington 24 – Halton 28 – Haldimand-Norfolk 29 – Brant 30 – Waterloo 31 – Perth 32 – Oxford 34 – Elgin 36 – Chatham-Kent 38 – Lambton 40 – Huron 41 – Bruce 42 – Grey 43 – Simcoe 46 – Haliburton 48 – Nipissing 53 – Greater Sudbury 57 – Algoma 58 – Thunder Bay
3.41 to 5.04	6 – Ottawa 7 – Leeds and Grenville 11 – Lennox and Addington 14 – Northumberland 15 – Peterborough 18 – Durham 19 – York 25 – Hamilton 39 – Middlesex 44 – Muskoka 47 – Renfrew 52 –Sudbury
5.04 to 7.73	12 – Hastings 16 – Kawartha Lakes 20 – Toronto 21 – Peel 26 – Niagara 37 – Essex 49 – Parry Sound 51 – Manitoulin 56 – Cochrane
7.73 to 10.22	59 – Rainy River 60 – Kenora
10.22 to 11.28	54 – Timiskaming

Return to Figure 2 on page 11.

Figure 3 is a bar graph with the y-axis representing the proportion of samples, expressed as a percent of all samples processed by the laboratory. Six bars on the x-axis represent the time between the date of sample collection and the date the test report was issued, also termed turnaround time, in days.

The six turnaround time groups are: less than or equal to two days, three days, four days, five days, six days and equal to or greater than seven days. The modal value (most commonly occurring) is five days and the median value is 4 days. Data are summarized in Table 3.

Table 3. Multiple marker screening turnaround times, PSO 2019-2020

Turnaround Time (days)	Proportion of samples (%)
≤ 2 days	21.2
3 days	18.8
4 days	17.2
5 days	25.6
6 days	8.7
≥ 7 days	8.5

Return to Figure 3 on page 13.

Figure 4 is a clustered bar graph with the y-axis representing the incidence of high risk NIPT results, expressed as a percentage of all NIPT results in the specific funding category. There are four clusters of bars, each cluster containing three bars. In each cluster, the first bar represents self-paid NIPT, the second bar represents OHIP-funded NIPT and the third bar represents all NIPT (both self-paid and OHIP-funded). The first cluster illustrates the incidence of results that were high risk for trisomy 21, the second cluster illustrates the incidence of results that were high risk for trisomy 18, the third cluster illustrates the incidence of results that were high risk for trisomy 13 and the fourth cluster illustrates the incidence of results that were high risk for sex chromosome (X or Y chromosome) differences. The first two bars are omitted from the high risk for trisomy 13 (3rd) cluster due to requirements for data suppression (see page 2 for details). Data are summarized in Table 4.

Table 4. Incidence of high risk NIPT results by funding status, PSO 2019-2020

Funding Status	Incidence of high risk result for T21 (%)	Incidence of high risk result for T18 (%)	Incidence of high risk result for T13 (%)	Incidence of high risk results for X/Y chromosome difference (%)
Self-Paid	0.5	0.2	Data suppressed	0.5
OHIP-Funded	2.4	0.6	Data suppressed	0.5
Total	1.6	0.4	0.2	0.5

Return to Figure 4 on page 15.

Figure 5 is a stacked bar graph with the y-axis representing the uptake of NIPT, expressed as a percent of all pregnancies in the specific age group. Six bars on the x-axis present the maternal age groups at the estimated date of delivery. The six age groups are: less than 25, 25 to 39, 30 to 34, 35 to 39, 40 to 44, and 45 and greater.

Each bar is composed of two distinctly coloured, stacked sections. The first section, beginning at the x-axis illustrates the uptake of self-paid NIPT. Stacked on top is a section illustrating the uptake of OHIP-funded NIPT. The height of the completed stack represents the uptake of all NIPT for that maternal age group. Data are summarized in Table 5.

Table 5. Uptake of NIPT by maternal age, PSO 2019-2020

Age (Years)	Uptake of OHIP-funded NIPT (%)	Uptake of self-paid NIPT (%)	Total uptake of NIPT (%)
> 25	1.8	0.4	2.2
25 to 29	2.4	1.9	4.3
30 to 34	4.1	5.5	9.6
35 to 39	8.3	10.0	18.3
40 to 44	59.4	3.2	62.6
≥ 45	37.5	7.9	45.4

Return to Figure 5 on page 16.

Figure 6 is a set of two maps of Ontario representing the uptake of NIPT, expressed as a percent of all pregnancies in a specific geographical region. Both Map A and Map B are divided into Statistics Canada's 49 census divisions. There are uptake values are grouped into five ranges and coloured on a gradient with the lightest yellow representing the lowest uptake range and the darkest orange representing the highest uptake range.

Map A illustrates the uptake of OHIP-funded NIPT. Data are summarized in Table 6A. Map B illustrates the uptake of self-paid NIPT. Data are summarized in Table 6B.

Table 6A. OHIP-funded NIPT uptake by census division, PSO 2019-2020

Uptake Range (%)	Census Division
2.09 to 3.04	14 – Northumberland 36 – Chatham-Kent 59 – Rainy River
3.04 to 4.69	1 – Stormont, Dundas and Glengarry 2 – Prescott and Russell 7 – Leeds and Grenville 12 – Hastings 13 – Prince Edward 16 – Kawartha 29 – Brant 31 – Perth 32 – Oxford 34 – Elgin 40 – Huron 41 – Bruce 46 – Haliburton 47 – Renfrew 48 – Nipissing 49 – Parry Sound 51 – Manitoulin 52 – Sudbury 53 – Greater Sudbury 60 – Kenora
4.69 to 6.22	9 – Lanark 10 – Frontenac 15 – Peterborough 22 – Dufferin 23 – Wellington 26 – Niagara 28 – Haldimand-Norfolk 37 – Essex 42 – Grey 44 – Muskoka 54 – Timiskaming 56 – Cochrane 57 – Algoma 58 – Thunder Bay
6.22 to 8.68	6 – Ottawa 11 – Lennox and Addington 18 – Durham 21 – Peel 25 – Hamilton 30 – Waterloo 38 – Lambton 39 – Middlesex 43 – Simcoe
8.68 to 11.31	19 – York 20 – Toronto 24 - Halton

Table 6B. Self-paid NIPT uptake by census division, PSO 2019-2020

Uptake (%)	Census Division
0.00 to 1.84	1 – Stormont, Dundas and Glengarry 2 – Prescott and Russell 9 – Lanark 11 – Lennox and Addington 12 – Hastings 13 – Prince Edward 14 – Northumberland 15 – Peterborough 16 – Kawartha 22 – Dufferin 26 – Niagara 28 – Haldimand-Norfolk 29 – Brant 31 – Perth 32 – Oxford 34 – Elgin 36 – Chatham-Kent 37 – Essex 38 – Lambton 40 – Huron 41 – Bruce 42 – Grey 44 – Muskoka 48 – Nipissing 51 – Manitoulin 52 – Sudbury 54 – Timiskaming 56 – Cochrane 59 – Rainy River 60 – Kenora
1.84 to 4.06	7 – Leeds and Grenville 18 – Durham 21 – Peel 23 – Wellington 25 – Hamilton 30 – Waterloo 39 – Middlesex 43 – Simcoe 47 – Renfrew 49 – Parry Sound 53 – Greater Sudbury 57 – Algoma 58 – Thunder Bay
4.06 to 5.67	10 – Frontenac 46 – Haliburton
5.67 to 7.50	6 – Ottawa
7.50 to 9.42	19 – York 20 – Toronto 24 - Halton

Return to Figure 6 on page 17.

Figure 7 is a bar graph with the y-axis representing the proportion of samples, expressed as a percent of all samples processed by the laboratory. Six bars on the x-axis represent the time between the date of sample collection and the date the test report was issued, also termed turnaround time, in days.

The six turnaround time groups are: less than or equal to six days, seven days, eight days, nine days, 10 days, and equal to or greater than 11 days. The modal value (most commonly occurring) is seven days and the median value is eight days. Data are summarized in Table 7.

Table 7. NIPT turnaround times, PSO 2019-2020

Turnaround Time (days)	Proportion of samples (%)
≤ 6 days	11.7
7 days	35.3
8 days	27.3
9 days	11.1
10 days	6.0
≥ 11 days	8.7

Return to Figure 7 on page 20.

Figure 8 is a map of Ontario with nine small circles placed throughout. Each small circle contains the number of nuchal translucency clinics in the region. There are 272 clinics in the Greater Toronto-Hamilton Area, five clinics in southwestern Ontario (around London), 16 clinics in areas surrounding Ottawa, nine clinics in the areas surround Sudbury, five clinics in the areas surrounding Timmins, two clinics around Kapuskasing, two clinics around Thunder Bay, one clinic in Moose Factory and one clinic in Rainy River.

Return to Figure 8 on page 21.

We'd love your feedback. Complete this survey and let us know what you liked about this report and how we can make it better.

PRENATAL SCREENING
DÉPISTAGE PRÉNATAL
ONTARIO