PRENATAL SCREENING
DÉPISTAGE PRÉNATAL
ONTARIO

BORN Ontario
Better Outcomes Registry & Network
Registre et Réseau des Bons Résultats dès la naissance

Prenatal Screening in Ontario

August 2019

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Genetic Counsellor, Prenatal Screening Ontario

The Road to PRENATAL SCREENING ONTARIO

1993
- Launch of Ontario Maternal Serum Screening Program

2003
- Shift to province-wide initiative with the introduction of nuchal translucency screening

2016
- Prenatal Screening Strategy Task Force struck by Provincial Council for Maternal and Child Health

2017
- Prenatal Screening Ontario launched as the formal provincial program

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Overview/Objectives

1. What is the current screening menu?
2. How has screening changed?
3. Benefits/limitations of different options
4. Practicalities of ordering
5. Provider tools and resources
6. Take home messages/key points

What you offer and what patients choose - is impacted by:

PERSONAL PREFERENCES & VALUES:

- What is the impact on pregnancy management decisions?

LOCATION/WHERE THEY LIVE:

- Is NT available &/or accessible in this region?
- Where are the closest referral centres?

GESTATION:

- Is NT still available vs. serum-only options
- What are the invasive diagnostic options?

NT= nuchal translucency

Current prenatal screening options in Ontario

1. Traditional multiple marker screening (MMS):
- Enhanced first trimester screen (eFTS)
- Maternal Serum Quadruple Screen (MSS)

2. Additional options:
- Non-invasive prenatal test (NIPT)

TRADITIONAL MULTIPLE MARKER SCREENING IN ONTARIO

Enhanced First Trimester Screening (eFTS)

- Single step, 1st trimester screen
  - performed between 11-13w6d gestation (CRL 45-84mm)
- Combines maternal/egg donor age with:
  1. nuchal translucency ultrasound measurement
  2. bloodwork
  - to measure maternal biochemical markers
    - Free beta human chorionic gonadotropin (hCG)
    - Placenta-associated plasma protein A (Papp-A)
    - Alpha-fetoprotein (AFP)
    - Placental Growth Factor (PlGF)* - (lab dependent)

* Combination of serum markers varies between labs

enhanced First Trimester Screen

Timing	11-14 weeks (CRL 45-84mm)
Screens for	trisomy 21 and trisomy 18
Detection rate	85-90%
False positive rate	3-6%
Results available	7 business days
Markers	• Maternal age • Nuchal translucency (NT) • Free beta human chorionic gonadotropin (hCG) • Placenta-associated plasma protein A (Papp-A) • Alpha-fetoprotein (AFP) • Placental Growth Factor (PlGF)

When is eFTS not possible?

- if gestational age ≥14 weeks
- if no access to NT or NT declined
- vanishing twin (see slide 25)

What are the options when eFTS is not possible?

Serum-only screening	Non-invasive prenatal testing (NIPT)
• If <14 weeks' gestation • If ≥15 weeks' gestation	• Ministry of Health funded • Patient self-pay

NT = nuchal translucency ultrasound
NIPT = non-invasive prenatal testing

Serum-only screening (no ultrasound)

Is lab & gestation-dependent

1. if patient is <14 weeks

sending to NYGH/CVH lab: first trimester serum Quad screen[1]
sending to MSH lab: second trimester MSS Quad screen[2]

2. if patient is ≥15 weeks

second trimester MSS Quad screen will be done (regardless of lab)

**Note – there is no multiple marker screening available for patients between 14w0d and 14w6d*

Maternal Serum Quadruple Screen

Timing	15 weeks 0 days – 20 weeks 6 days
Screens for	trisomy 21 and trisomy 18
Detection rate	80%
False positive rate	5%
Results available	7 business days
Markers	• Maternal age • Maternal serum alpha-fetoprotein (MS-AFP) • Unconjugated estriol (uE3) • Inhibin-A (DIA) • Human chorionic gonadotropin (hCG)

What about oNTD screening?

Current SOGC guidelines state:

"the primary screening test for the detection of fetal structural abnormalities including neural tube defects is a second trimester anatomical ultrasound with detailed fetal cranial and spinal imaging and assessment"[1]

1.Joint SOGC-CCMG Clinical Practice Guideline. J Obstet Gynaecol Can 2017

oNTD screening

MS-AFP (Maternal Serum Alpha Fetoprotein) screening for oNTD (open neural tube defects)

1. no longer routinely offered
2. available only when no access to a good quality ultrasound examination

Joint SOGC/CCMG updated guidelines Sept 2017:

"Second trimester serum alpha fetoprotein screening to rule out open neural tube defects is no longer necessary unless there is a barrier to good quality ultrasound examination (II-2A)."

Summary - what has changed?

1. Integrated Prenatal Screening (IPS)
   - no longer available (discontinued in Nov 2017)
   - largely replaced by enhanced First Trimester Screening (eFTS) given its earlier timing & comparable detection rate
2. Serum Integrated Prenatal Screening (SIPS)
   – no longer available
   – serum only options are still available (see slide 10 for details) for those with no access to NT ultrasound
   – lab & region dependent
3. Maternal serum alpha-fetoprotein (MS-AFP)
   – no longer used to screen for open neural tube defects (oNTD) (see slides 12 & 13 for details)

NON-INVASIVE PRENATAL TESTING (NIPT) IN ONTARIO

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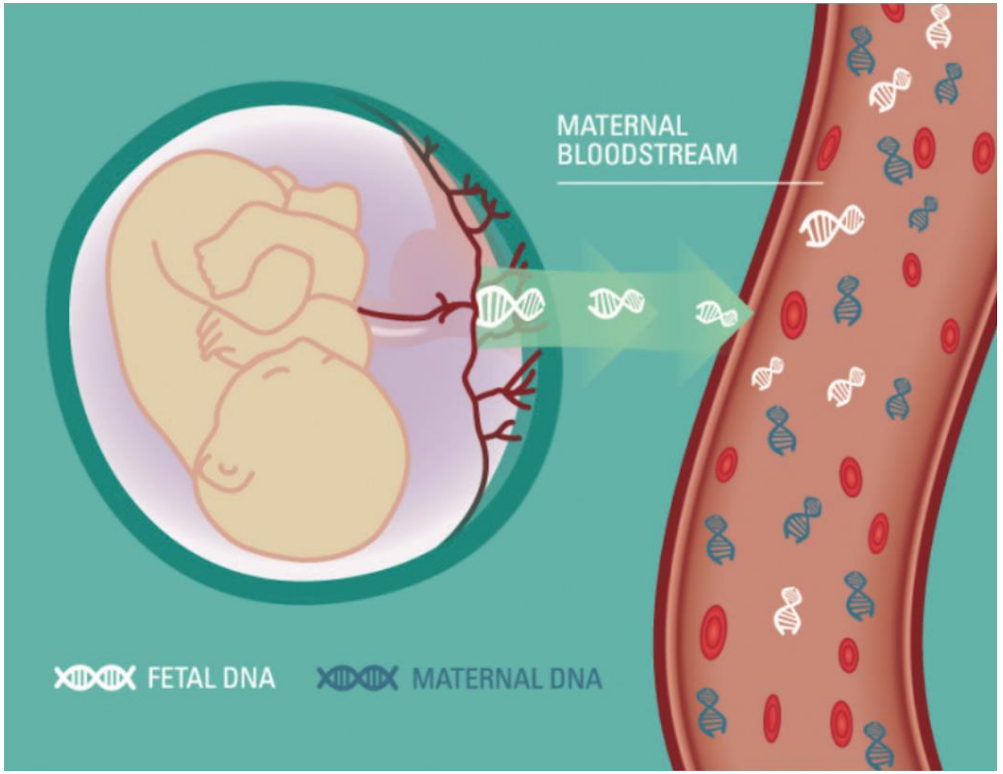
Non-invasive Prenatal Testing (NIPT)

What is NIPT & how are we using it in Ontario?

- NIPT analyzes circulating cell-free fetal DNA in maternal blood (cffDNA)
- Currently available to all pregnant persons in Ontario
  - MOH funding based on eligibility criteria (see next slide)
- 2 provincial labs performing MOH funded analysis (additional options for self-pay patients)
- SOGC guidelines suggest blood draw >10weeks (possible >9weeks)*



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Current NIPT MOH-funding eligibility criteria:

Category I criteria: (NIPT can be ordered by any physician)

- a positive multiple marker screening (eFTS/MSS) – in current pregnancy
- maternal (or egg donor) age is ≥40 years at the expected date of delivery
- nuchal translucency (NT) measurement is ≥3.5mm
- a personal history of a previous pregnancy or child with a specific chromosome condition

*Patient must only meet ONE of the above criteria

Current NIPT MOH-funding eligibility criteria:

Category II criteria

(to be ordered by a genetics or maternal-fetal medicine specialist):

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

*Patient must only meet ONE of the above criteria

Non-invasive Prenatal Testing (NIPT)

How does NIPT compare to traditional multiple marker screening?

Screening test	Detection rate for trisomy 21	False positive rate for trisomy 21
eFTS	88%	3%
MSS Quad	76%	3%
NIPT	more than 99%	less than 0.1%

eFTS = enhanced first trimester screen
MSS = maternal serum screen

NIPT Labs

How do they compare?

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	Harmony™ by Ariosa	Panorama™ by Natera
Where is blood drawn for this test?	DynaCare Next	LifeLabs Genetics
How early can blood be drawn?	10 weeks' gestation	9 weeks' gestation
When are results available?	10 business days	7-10 calendar days
Twins	Yes	Yes
In vitro fertilization (IVF)	Yes	Yes
IVF with a donor egg (not self)	Yes	Yes
Using surrogate	Yes	Yes
Triploidy	No	Yes
Higher order multiples	No	No
Vanishing twin	No	No
Sex chromosome reporting**	Opt-in for: Fetal sex, monosomy X, sex chromosome aneuploidy	Standard Fetal sex: optional
Other options (PRIVATE PAY)**	+22q11.2	+22q11.2 +microdeletion panel (x5)

Benefits of NIPT

1. Offered earlier than traditional multiple marker screening (MMS) (9/10 weeks)

   Leading to:

   - Earlier invasive diagnostic test options (CVS vs amniocentesis)
   - Earlier reassurance (when low risk result)
   - Earlier management options

2. Decreased invasive diagnostic procedures
   - Decreased procedure-related losses
3. Focused on most common aneuploidies
4. Much better detection rate/false positive rate than traditional MMS

Limitations of NIPT

1. Only 13, 18, 21, X & Y
   - Not able to give information about single gene disorders/aneuploidy of other chromosomes
2. Not diagnostic
   - NIPT is a screening test
   - Does not offer “yes” or “no” answers
3. “No call” results possible
   - most commonly due to high BMI, early gestation, poor placental function
     - Current literature reports 1-8% redraw rate

Limitations of NIPT

4. Not possible in all pregnancies
   - higher order multiples (>2), vanishing twins[1]
5. NIPT accuracy varies depending on the condition and the population screened
   - Positive predictive value – PPV[2]
   - a high risk NIPT result for trisomy 21 is more likely to be a true result than a high risk result for trisomy 13
   - a high risk NIPT result for trisomy 21 in a 40 year old is more likely to be a true result than a high risk result for trisomy 21 in a 27 year old

Prenatal Screening in: Multiple pregnancies & vanishing twins

	NT ultrasound only	NT + eFTS	MSS	NIPT
Twins		✓		✓
≥2 babies	✓			
Vanishing twin	✓		✓ *8 weeks post-demise	

* In cases of vanishing twin, NT + maternal serum screen (MSS) is available. It is recommended that the bloodwork be drawn at least 8 weeks post-demise.
* In pregnancies carrying more than 2 babies (higher order multiples), screening for aneuploidy is limited to nuchal translucency ultrasound measurement in combination with maternal (or donor egg) age.

PRACTICALITIES OF ORDERING

Provider Tools and Resources

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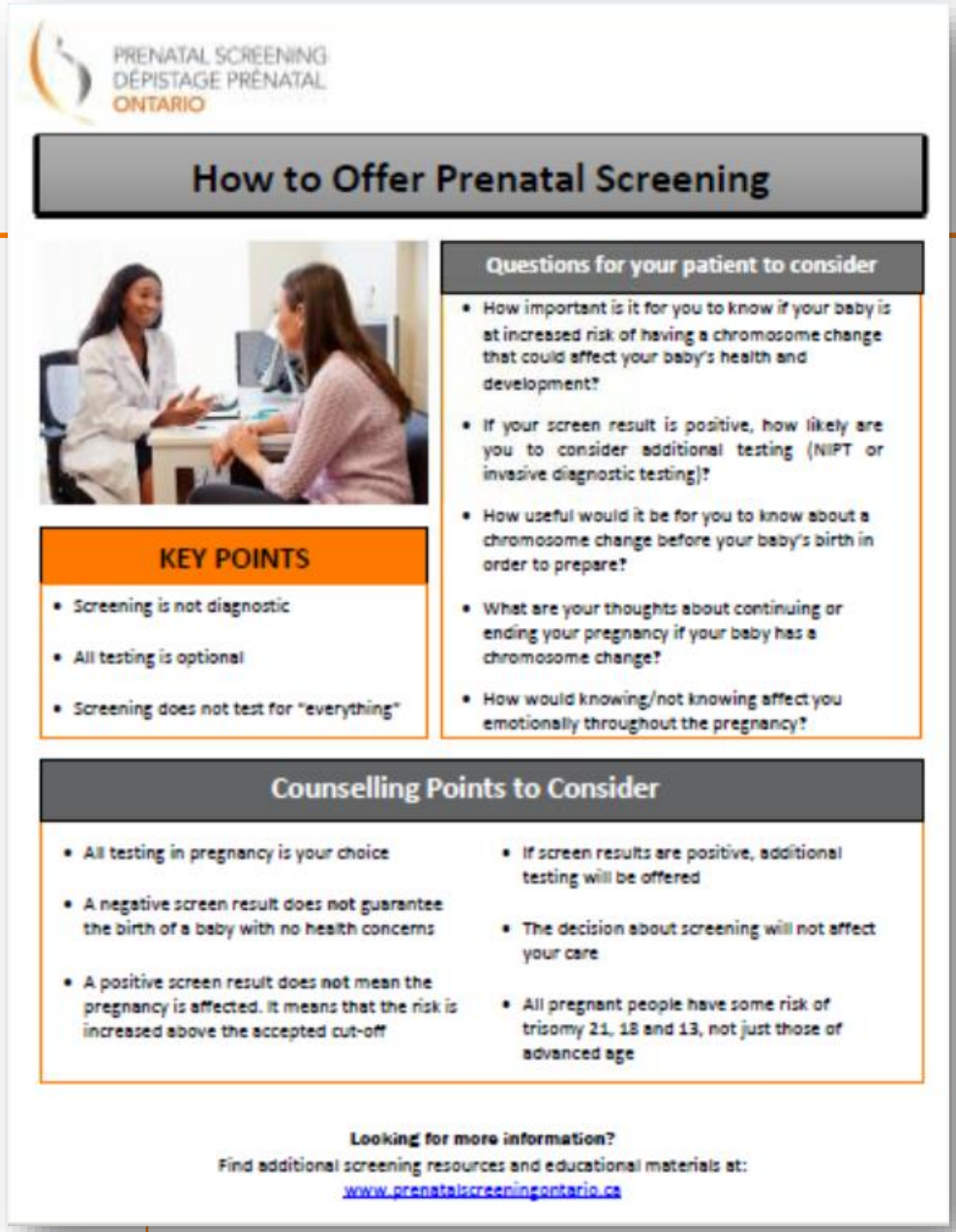
- How to order
- When to order
- Who qualifies for testing?
- What tests are being offered where?
- What requisitions to use
- Where can I refer?

How to Offer



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How to Offer Prenatal Screening

Questions for your patient to consider

- How important is it for you to know if your baby is at increased risk of having a chromosome change that could affect your baby's health and development?
- If your screen result is positive, how likely are you to consider additional testing (NIPT or invasive diagnostic testing)?
- How useful would it be for you to know about a chromosome change before your baby's birth in order to prepare?
- What are your thoughts about continuing or ending your pregnancy if your baby has a chromosome change?
- How would knowing/not knowing affect you emotionally throughout the pregnancy?

KEY POINTS

- Screening is not diagnostic
- All testing is optional
- Screening does not test for "everything"

Counselling Points to Consider

- All testing in pregnancy is your choice
- A negative screen result does not guarantee the birth of a baby with no health concerns
- A positive screen result does not mean the pregnancy is affected. It means that the risk is increased above the accepted cut-off
- If screen results are positive, additional testing will be offered
- The decision about screening will not affect your care
- All pregnant people have some risk of trisomy 21, 18 and 13, not just those of advanced age

Looking for more information?
Find additional screening resources and educational materials at:
www.prenatalscreeningontario.ca

-key points
-questions for patients
-counselling points

[Download this provider tool]

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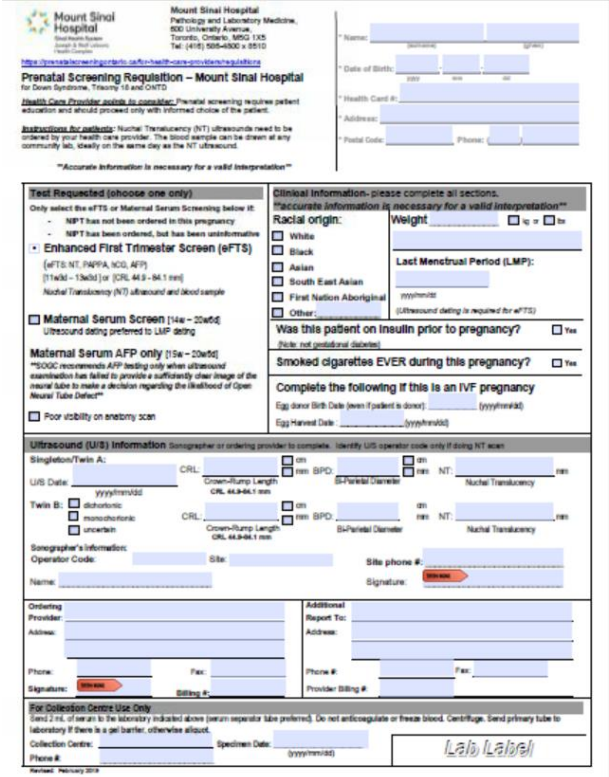
How to Order - Multiple marker screening (eFTS/MSS)

Mount Sinai Hospital


Mount Sinai Hospital
Pathology and Laboratory Medicine,
600 University Avenue,
Toronto, Ontario, M5G 1X5
Tel: (416) 586-4800 x 8510

Prenatal Screening Requisition – Mount Sinal Hospital
for Down Syndrome, Trisomy 18 and ONTD

Health Care Provider points to consider: Prenatal screening requires patient education and should proceed only with informed choice of the patient.

Instructions for patients: Nuchal Translucency (NT) ultrasounds need to be ordered by your health care provider. The blood sample can be drawn at any community lab, ideally on the same day as the NT ultrasound.

"Accurate information is necessary for a valid interpretation"

Test Requested (choose one only)
Only select the eFTS or Maternal Serum Screening below if:
- NIPT has not been ordered in this pregnancy
- NIPT has been ordered, but has been uninformative

Enhanced First Trimester Screen (eFTS)
(eFTS: NT, PAPPA, hCG, AFP)
[11w0d – 13w6d] or [CRL 44.9 – 84.1 mm]
Nuchal Translucency (NT) ultrasound and blood sample

Maternal Serum Screen [14w – 20w6d]
Ultrasound dating preferred to LMP dating

Maternal Serum AFP only [15w – 20w6d]
"SOGC recommends AFP testing only when ultrasound examination has failed to provide a sufficiently clear image of the neural tube to make a decision regarding the likelihood of Open Neural Tube Defect"

Poor visibility on anatomy scan

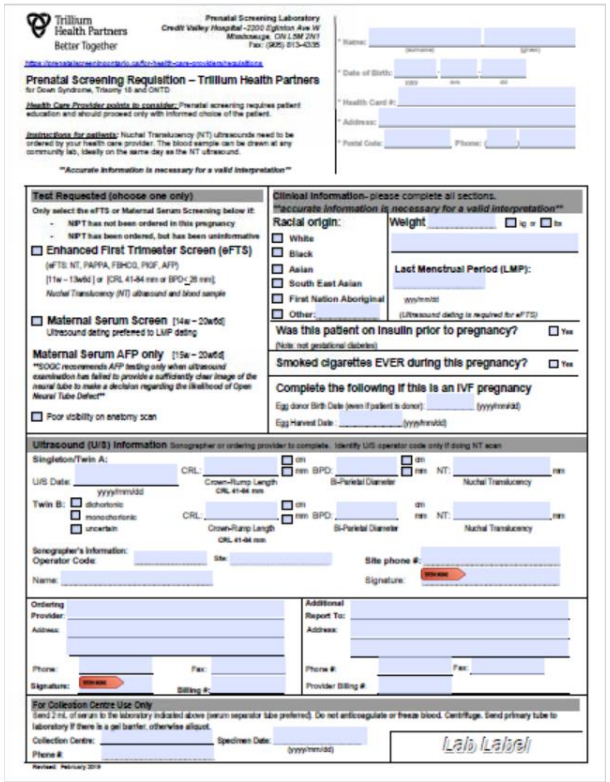
Clinical Information- please complete all sections.
"Accurate information is necessary for a valid interpretation"
Racial origin: White, Black, Asian, South East Asian, First Nation Aboriginal, Other
Weight
Last Menstrual Period (LMP):
Was this patient on insulin prior to pregnancy?
Smoked cigarettes EVER during this pregnancy?
Complete the following if this is an IVF pregnancy
Egg donor Birth Date (even if patient is donor):
Egg Harvest Date:

Ultrasound (U/S) Information
Singleton/Twin A:
U/S Date:
Twin B: dichorionic, monochorionic, uncertain
Sonographer's Information:
Operator Code:
Site:
Site phone #:
Name:
Signature:

Ordering Provider:
Address:
Phone:
Fax:
Signature:
Billing #:
Additional Report To:
Address:
Phone #:
Fax:
Provider Billing #:

For Collection Centre Use Only
Send 2 mL of serum to the laboratory indicated above (serum separator tube preferred). Do not anticoagulate or freeze blood. Centrifuge. Send primary tube to laboratory if there is a gel barrier, otherwise aliquot.
Collection Centre:
Specimen Date:
Phone #:
Lab Label

Trillium Health Partners


Trillium Health Partners
Better Together
Prenatal Screening Laboratory
Credit Valley Hospital - 2200 Eglinton Ave W
Mississauga, ON L5M 2N1
Fax: (905) 813-4335

Prenatal Screening Requisition – Trillium Health Partners
for Down Syndrome, Trisomy 18 and ONTD

Health Care Provider points to consider: Prenatal screening requires patient education and should proceed only with informed choice of the patient.

Instructions for patients: Nuchal Translucency (NT) ultrasounds need to be ordered by your health care provider. The blood sample can be drawn at any community lab, ideally on the same day as the NT ultrasound.

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Enhanced First Trimester Screen (eFTS)
(eFTS: NT, PAPPA, FBHCG, PIGF, AFP)
[11w – 13w6d] or [CRL 41-84 mm or BPD<26 mm]
Nuchal Translucency (NT) ultrasound and blood sample

Maternal Serum Screen [14w – 20w6d]
Ultrasound dating preferred to LMP dating

Maternal Serum AFP only [15w – 20w6d]
"SOGC recommends AFP testing only when ultrasound examination has failed to provide a sufficiently clear image of the neural tube to make a decision regarding the likelihood of Open Neural Tube Defect"

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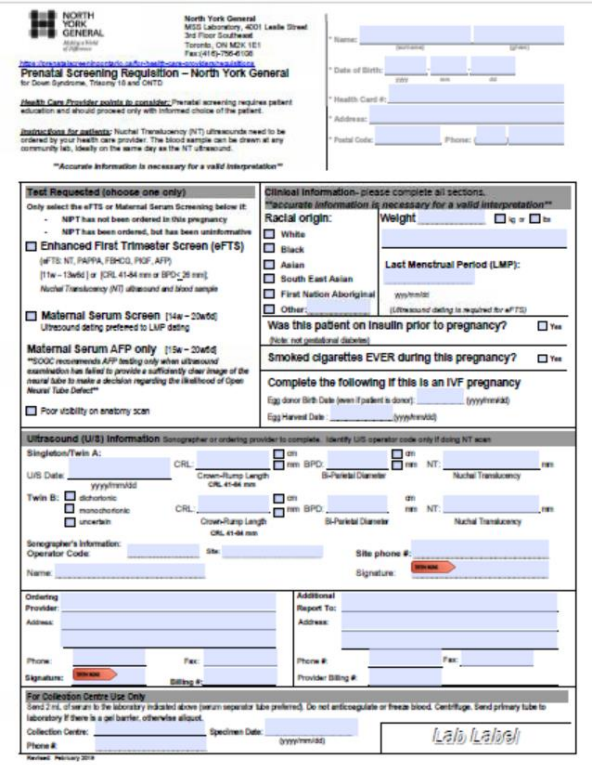
Was this patient on insulin prior to pregnancy?
Smoked cigarettes EVER during this pregnancy?
Complete the following if this is an IVF pregnancy
Egg donor Birth Date (even if patient is donor):
Egg Harvest Date:

Ultrasound (U/S) Information
Singleton/Twin A:
U/S Date:
Twin B: dichorionic, monochorionic, uncertain
Sonographer's Information:
Operator Code:
Site:
Site phone #:
Name:
Signature:

Ordering Provider:
Address:
Phone:
Fax:
Signature:
Billing #:
Additional Report To:
Address:
Phone #:
Fax:
Provider Billing #:

For Collection Centre Use Only
Send 2 mL of serum to the laboratory indicated above (serum separator tube preferred). Do not anticoagulate or freeze blood. Centrifuge. Send primary tube to laboratory if there is a gel barrier, otherwise aliquot.
Collection Centre:
Specimen Date:
Phone #:
Lab Label

North York General Hospital


NORTH YORK GENERAL
North York General
MSS Laboratory, 4001 Leslie Street
3rd Floor Southeast
Toronto, ON M2K 1E1
Fax:(416)-756-6106

Prenatal Screening Requisition – North York General
for Down Syndrome, Trisomy 18 and ONTD

Health Care Provider points to consider: Prenatal screening requires patient education and should proceed only with informed choice of the patient.

Instructions for patients: Nuchal Translucency (NT) ultrasounds need to be ordered by your health care provider. The blood sample can be drawn at any community lab, ideally on the same day as the NT ultrasound.

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Ultrasound dating preferred to LMP dating

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"SOGC recommends AFP testing only when ultrasound examination has failed to provide a sufficiently clear image of the neural tube to make a decision regarding the likelihood of Open Neural Tube Defect"

Poor visibility on anatomy scan

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"Accurate information is necessary for a valid interpretation"
Racial origin: White, Black, Asian, South East Asian, First Nation Aboriginal, Other
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Address:
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Collection Centre:
Specimen Date:
Phone #:
Lab Label

All requisitions and helpful hints for ordering can be found at:
www.prenatalscreeningontario.ca

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How to Discuss Prenatal Screening Results
Traditional Screening (eFTS / MSS)

Negative Screen Results

- A negative screen result is reassuring
- A negative screen result does **not** guarantee the birth of a baby with no health concerns
- A negative screen result would typically not prompt the offer of invasive diagnostic testing, in the absence of additional risk factors

Positive Screen Results

- A positive screen result does **not** mean the pregnancy is affected with a trisomy. It means that the risk is increased above the accepted cut-off
- If screen results are positive, both NIPT and invasive testing should be offered for the pregnant person to choose
- A referral for genetic counselling can be offered where available
- Only invasive diagnostic testing (chorionic villus sampling or amniocentesis) can provide a diagnosis

eFTS – enhanced first trimester screening
MSS – maternal serum screening
NIPT – non-invasive prenatal testing

KEY POINTS

- Screening is **not** diagnostic
- eFTS / MSS only screen for Down Syndrome and Trisomy 18

Looking for more information?
Find additional screening resources and educational materials at:
www.prenatalscreeningontario.ca

How to Discuss screening results

Traditional screening results (eFTS/MSS)

-key points
-counselling points

[Download this provider tool]

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How to Discuss NIPT results

-key points
-counselling points

[Download this provider tool]

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How to Discuss Prenatal Screening Results

Non-Invasive Prenatal Testing (NIPT)
for trisomy 21, 18, 13 ± sex chromosome differences

Low Risk Screening Results

- This typically means the risk for trisomy 21, 18, 13 is <1:10,000
- A low risk screen result is reassuring, but this depends on the indication for testing
- A low risk screen result does not guarantee the birth of a baby without any health concerns or other genetic conditions
- A low risk screen result would not prompt the offer of invasive diagnostic testing

Looking for more information?
Find additional screening resources and educational materials at:
www.prenatalscreeningontario.ca

High Risk Screening Results

- This typically means the risk for trisomy 21, 18, 13 is significantly increased
- The chance that a high risk screen result is a true abnormal varies by chromosome and the pregnant person's age
- A referral for genetic counselling should be offered
- NIPT is a screening test - only invasive diagnostic testing (chorionic villus sampling or amniocentesis) can provide a diagnosis

"No Call" or Failed Results

- There are different reasons why NIPT fails (e.g. not enough fetal/placental DNA [due to high maternal weight or blood drawn too early], chromosome difference in mother or baby)
- Repeating the bloodwork (i.e. redraw) will yield a result in most cases

KEY POINTS

- NIPT is a screening test, it is not diagnostic
- NIPT screens for trisomy 21, 18, 13 (± sex chromosome differences)
- Invasive diagnostic testing should be considered in the context of a high risk NIPT

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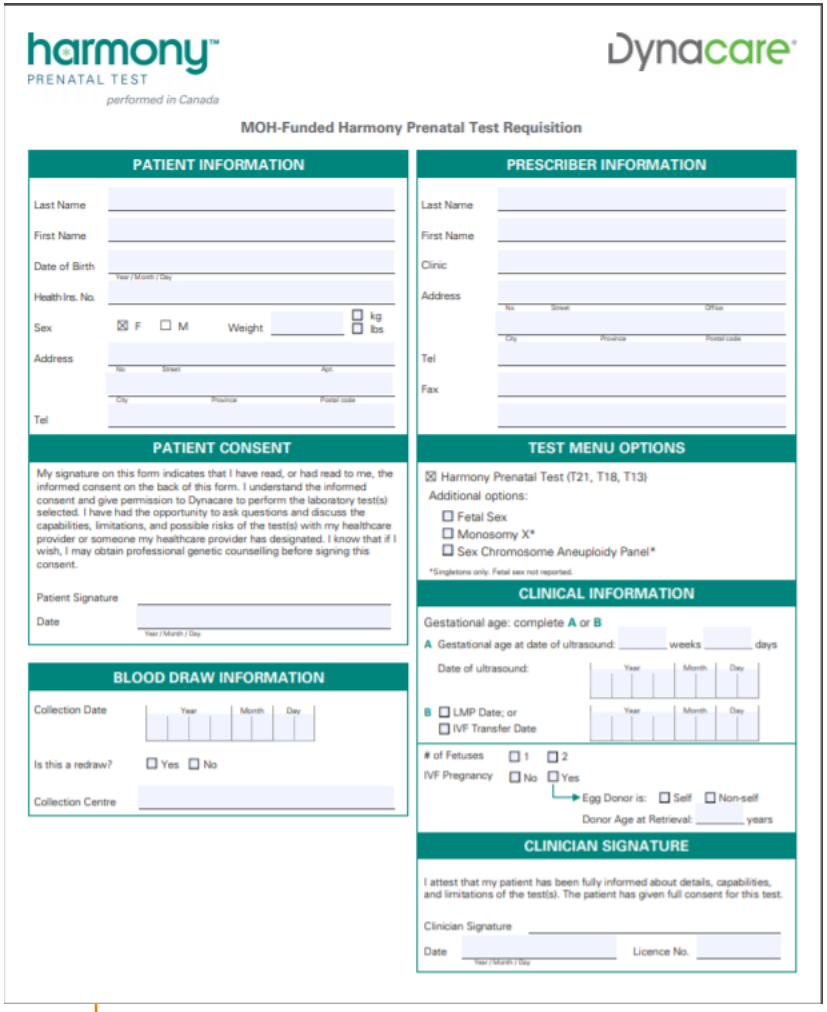
NIPT Requisitions

Harmony NIPT - Dynacare



harmony™ PRENATAL TEST performed in Canada

Dynacare

MOH-Funded Harmony Prenatal Test Requisition

PATIENT INFORMATION

Last Name
First Name
Date of Birth (Year / Month / Day)
Health Ins. No.
Sex ☒ F ☐ M Weight ☐ kg ☐ lbs
Address (No. Street Apt. / City Province Postal code)
Tel

PRESCRIBER INFORMATION

Last Name
First Name
Clinic
Address (No. Street Office / City Province Postal code)
Tel
Fax

PATIENT CONSENT

My signature on this form indicates that I have read, or had read to me, the informed consent on the back of this form. I understand the informed consent and give permission to Dynacare to perform the laboratory test(s) selected. I have had the opportunity to ask questions and discuss the capabilities, limitations, and possible risks of the test(s) with my healthcare provider or someone my healthcare provider has designated. I know that if I wish, I may obtain professional genetic counselling before signing this consent.

Patient Signature
Date (Year / Month / Day)

TEST MENU OPTIONS

☒ Harmony Prenatal Test (T21, T18, T13)
Additional options:
☐ Fetal Sex
☐ Monosomy X*
☐ Sex Chromosome Aneuploidy Panel*
*Singletons only. Fetal sex not reported.

BLOOD DRAW INFORMATION

Collection Date (Year / Month / Day)
Is this a redraw? ☐ Yes ☐ No
Collection Centre

CLINICAL INFORMATION

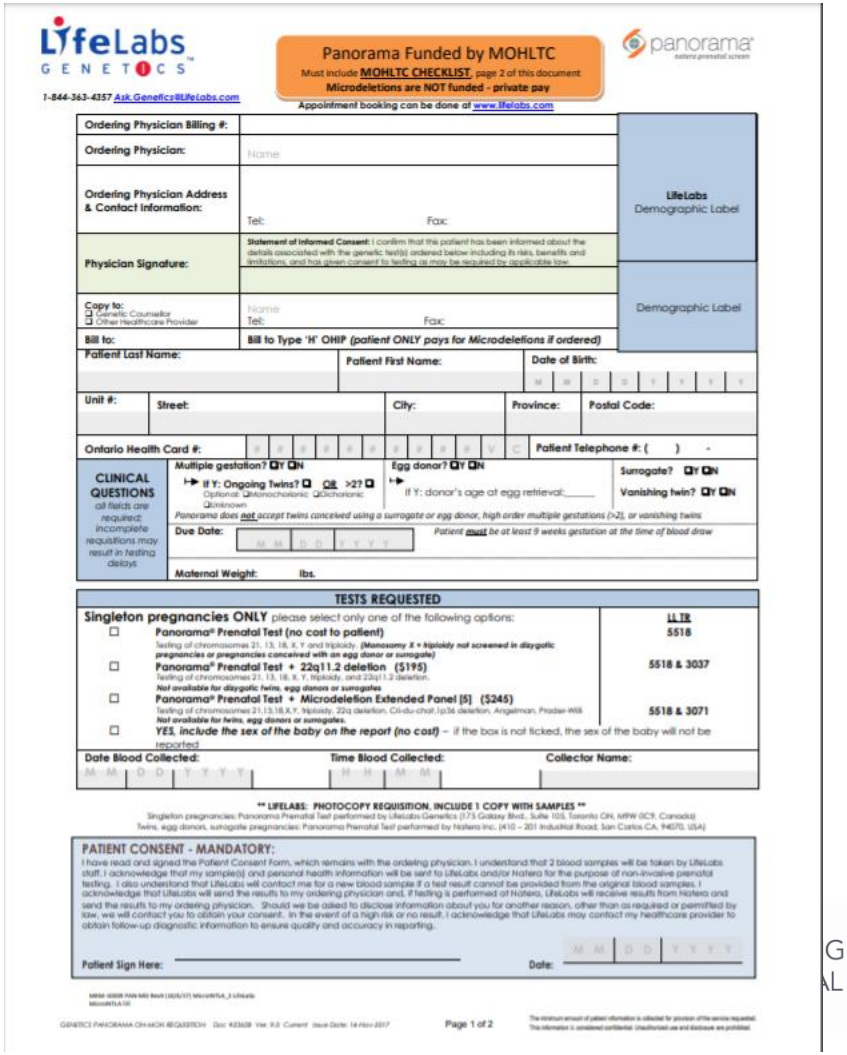
Gestational age: complete A or B
A Gestational age at date of ultrasound: _____ weeks _____ days
Date of ultrasound: (Year / Month / Day)
B ☐ LMP Date; or ☐ IVF Transfer Date (Year / Month / Day)
# of Fetuses ☐ 1 ☐ 2
IVF Pregnancy ☐ No ☐ Yes → Egg Donor is: ☐ Self ☐ Non-self
Donor Age at Retrieval: _____ years

CLINICIAN SIGNATURE

I attest that my patient has been fully informed about details, capabilities, and limitations of the test(s). The patient has given full consent for this test.

Clinician Signature
Date (Year / Month / Day) Licence No.

Panorama NIPT - LifeLabs



LifeLabs GENETICS

1-844-363-4357 Ask.Genetics@LifeLabs.com

Panorama Funded by MOHLTC
Must include MOHLTC CHECKLIST, page 2 of this document
Microdeletions are NOT funded - private pay

Appointment booking can be done at www.lifelabs.com

panorama™ noninvasive prenatal screen

Ordering Physician Billing #:	LifeLabs Demographic Label
Ordering Physician: Name	
Ordering Physician Address & Contact Information: Tel: Fax:	
Physician Signature: Statement of Informed Consent: I confirm that this patient has been informed about the details associated with the genetic test(s) ordered below including its risks, benefits and limitations, and has given consent to testing as may be required by applicable law.	
Copy to: ☐ Genetic Counsellor ☐ Other Healthcare Provider Name Tel: Fax:	Demographic Label
Bill to: Bill to Type 'H' OHIP *(patient ONLY pays for Microdeletions if ordered)*	

Patient Last Name: | Patient First Name: | Date of Birth: (M M D D Y Y Y Y)

Unit #: | Street: | City: | Province: | Postal Code:

Ontario Health Card #: | Patient Telephone #: ( ) -

CLINICAL QUESTIONS *all fields are required; incomplete requisitions may result in testing delays*

Multiple gestation? ☐Y ☐N
↳ If Y: Ongoing Twins? ☐ OR >2? ☐ Optional: ☐Monochorionic ☐Dichorionic ☐Unknown
Egg donor? ☐Y ☐N
↳ If Y: donor's age at egg retrieval:_____
Surrogate? ☐Y ☐N
Vanishing twin? ☐Y ☐N
Panorama does not accept twins conceived using a surrogate or egg donor, high order multiple gestations (>2), or vanishing twins
Due Date: (M M D D Y Y Y Y) *Patient must be at least 9 weeks gestation at the time of blood draw*
Maternal Weight: lbs.

TESTS REQUESTED

Singleton pregnancies ONLY please select only one of the following options:

		LL TR
☐	**Panorama® Prenatal Test (no cost to patient)** Testing of chromosomes 21, 13, 18, X, Y and triploidy. *(Monosomy X + triploidy not screened in dizygotic pregnancies or pregnancies conceived with an egg donor or surrogate)*	5518
☐	**Panorama® Prenatal Test + 22q11.2 deletion ($195)** Testing of chromosomes 21, 13, 18, X, Y, triploidy, and 22q11.2 deletion. *Not available for dizygotic twins, egg donors or surrogates*	5518 & 3037
☐	**Panorama® Prenatal Test + Microdeletion Extended Panel [5] ($245)** Testing of chromosomes 21,13,18,X,Y, triploidy, 22q deletion, Cri-du-chat,1p36 deletion, Angelman, Prader-Willi *Not available for twins, egg donors or surrogates*	5518 & 3071
☐	***YES, include the sex of the baby on the report (no cost)*** – if the box is not ticked, the sex of the baby will not be reported	

Date Blood Collected: (M M D D Y Y Y Y) | Time Blood Collected: (H H M M) | Collector Name:

** LIFELABS: PHOTOCOPY REQUISITION, INCLUDE 1 COPY WITH SAMPLES **
Singleton pregnancies: Panorama Prenatal Test performed by LifeLabs Genetics (171 Galaxy Blvd., Suite 103, Toronto ON, M9W 0C9, Canada)
Twins, egg donors, surrogate pregnancies: Panorama Prenatal Test performed by Natera Inc. (410 – 201 Industrial Road, San Carlos CA, 94070, USA)

PATIENT CONSENT - MANDATORY:

I have read and signed the Patient Consent Form, which remains with the ordering physician. I understand that 2 blood samples will be taken by LifeLabs staff. I acknowledge that my sample(s) and personal health information will be sent to LifeLabs and/or Natera for the purpose of non-invasive prenatal testing. I also understand that LifeLabs will contact me for a new blood sample if a test result cannot be provided from the original blood samples. I acknowledge that LifeLabs will send the results to my ordering physician and, if testing is performed at Natera, LifeLabs will receive results from Natera and send the results to my ordering physician. Should we be asked to disclose information about you for another reason, other than as required or permitted by law, we will contact you to obtain your consent. In the event of a high risk or no result, I acknowledge that LifeLabs may contact my healthcare provider to obtain follow-up diagnostic information to ensure quality and accuracy in reporting.

Patient Sign Here: Date: (M M D D Y Y Y Y)

GENETICS PANORAMA ON-MOH REQUISITION Doc: 423608 Ver: 9.0 Current Issue Date: 14-Nov-2017 Page 1 of 2

When to refer to Genetics

1.When nuchal translucency (NT) measurement is increased (≥3.5mm) [1]

2. If non-invasive prenatal testing (NIPT) result "high risk"[2]

3. If there are ultrasound anomalies suggestive of genetic condition other than trisomy 21, trisomy 18 or trisomy 13[3]

*You may also consider contacting your local Genetics centre with questions related to any of these indications.

NOTES:

1. Increased NT is known to be associated with an increased risk for aneuploidy, microarray abnormalities, single gene disorders and cardiac abnormalities
2. NIPT is a screening test & is not diagnostic – patients should be offered invasive diagnostic testing for confirmation
3. Ultrasound anomalies suggestive of a single gene disorder or microarray abnormality

Why refer to Genetics?

Geneticists & genetic counsellors will:

- Review and explain the screening result, tailored to the patient's specific clinical scenario
- Educate the patient regarding possible diagnoses, risks/benefits of further testing, pregnancy management options
- Answer questions and provide decisional support
- Review recurrence risks
  - "What are the chances that this will happen to me again?"
  - "This has never happened in my family before. Why has it happened now?"

Regional Genetics Centres

There are currently 15 regional Genetics centres offering prenatal care in Ontario.

Hamilton	Kingston
London	Mississauga
North York	Orillia
Oshawa	Ottawa
Peterborough	Richmond Hill
Scarborough	Sudbury
Thunder Bay	Timmins
Toronto	

Find your local Genetics clinic:
Canadian Association of Genetic Counsellors

Take home messages

1. Prenatal screening is useful for ALL, not just those of advanced maternal age

   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). [1]

2. Screening is not diagnostic
   - Positive MMS/NIPT needs to be confirmed with diagnostic testing

Joint SOGC-CCMG Clinical Practice Guideline. J Obstet Gynaecol Can 2017

Take home messages

3. NT is useful beyond screening for aneuploidy

   Where available with documented expertise, the first trimester ultrasound (11 to 14 weeks' gestation) offers many advantages including accurate dating, determination of twin chorionicity, early detection of major structural abnormalities, and aneuploidy screening (II-2A).[1]

   ***NT should still be offered even if low risk NIPT <12wks**

Joint SOGC-CCMG Clinical Practice Guideline. J Obstet Gynaecol Can 2017

Take home messages

4. Screening can still be useful if a patient would not consider interrupting a pregnancy
   - Information can be useful for preparation and management
5. 2 screens are not better than 1
   - Redundant/repeat testing not advised (or useful)
     - A positive MMS AFTER a low risk/negative NIPT can be harmful – NT alone should be offered

Take home messages

6. For further information or counselling regarding prenatal screening
   - consider a referral to a local Genetics centre
   - contact Prenatal Screening Ontario

For questions or to arrange a lunch and learn in your centre, please contact Andrea Staines at (437)688-7529 or Astaines@bornontario.ca

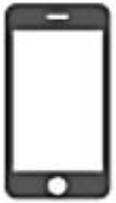
How to contact us:

www.prenatalscreeningontario.ca

PSO@BORNontario.ca

fb.me/PrenatalScreeningOntario

@OntarioPSO



Toll-free: 1.833.351.6490

613.737.2281

Did you know? PSO has on-call genetic counsellors to answer your questions about prenatal screening Mon-Fri 8-4pm

PRENATAL SCREENING
DÉPISTAGE PRÉNATAL
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