

Newborn Screening Bulletin 2023-9

December 14, 2023

Newborn screening over the holidays and upcoming changes

With the holidays approaching, we would like to inform you of the courier schedule and provide you with some tips to ensure timely newborn screening. Please forward this bulletin to any members of your team who will be involved in newborn screening collection or shipping over the holidays. We have also included upcoming changes regarding screen positive reports, and news about our website and resources.

Courier downtimes over the holiday period:

- No ad-hoc pick up requests on Saturday, December 23rd or Saturday, December 30th — regular Saturday deliveries will occur (shipments picked up on Friday will be delivered to NSO on Saturday).
- Purolator will NOT be operational during the following time periods:
 - Sunday, December 24th — Tuesday, December 26th inclusive
 - Sunday, December 31st — Monday, January 1st inclusive

****Special Notice to Midwifery Practice Groups****

If requesting a pick up on Friday, December 22nd or Friday, December 29th, please do so as early as possible and provide a wide window of time for the pick up.

If midwifery practice groups are not able to request a pickup on Friday, December 22nd or Friday, December 29th, samples can still be shipped to NSO by dropping the shipment off at an authorized Purolator location.

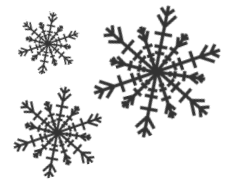
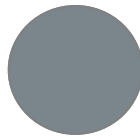
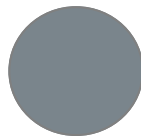
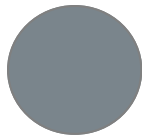
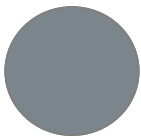
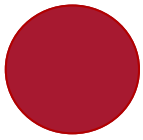
Please click [here](#) to find the most convenient drop-off location for you.

Tips to ensure timely newborn screening:

NSO will be working throughout the holiday period to ensure testing is done quickly despite any courier delays. To minimize potential delays caused by holiday downtimes, please do the following:

- Collect newborn screens between 24-48 hours of life. If an infant is discharged at <24 hours of age, please collect a newborn screen before discharge and ask the family to return in the next 24 hours for a repeat screen.
- Deliver samples to your shipping location (e.g. lab, unit clerk, practice office etc.) as soon as possible.
- Ship samples every business day, as soon as they are dry.
- If you are requesting a new pick up over the holiday period, a reminder to do so early in the day and to provide a wide window of time for Purolator to pick up your samples.
- Please closely review requisitions for completeness to reduce the likelihood a sample will be deemed unsatisfactory as a result of missing information. This is especially important for midwifery practice groups with offices that will be closed over the holiday period.





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Upcoming in 2024: Updated website and educational Submitter Hub

- Our website will have a new look in early 2024! In January, we will be launching our updated website and Submitter Hub, an updated online submitter manual and educational hub for health care providers involved in newborn screening. Stay tuned for more information early in the new year!

Change in practice: Faxing screen positive reports

NSO sends screen positive reports to hospitals and midwifery practices once NSO has referred the baby to a Regional Treatment Centre and the family is aware of the newborn screening result. Previously, these reports had been sent in an individual fax job with a cover letter.

Beginning December 18, NSO will fax screen positive reports along with other newborn screening reports that do not require follow-up. There will continue to be a note at the bottom of every screen positive report (see next page for example), indicating that a referral has been made and to which Regional Treatment Centre. The upcoming change will streamline the reporting workflow and allow these results to go out in a timely manner, with fewer manual steps.

We wish you Happy Holidays and all the best for the New Year!





Health Card Number/Type: Not Applicable Sex: M Birth: 2023-05-02 12:00 Collection: 2023-05-03 13:00 Receipt: 2023-05-05

Guardian's Name: Other Names:
Submitting Location: Location Fax: Submitter #:
Ordering HCP: Ordering HCP Fax:
Infant's HCP: Infant's HCP Fax:

Amino Acidemias:

Phenylketonuria and Variants / Bioppterin Defects Negative
Maple Syrup Urine Disease Negative
Homocystinuria (Hypermethioninemia) Negative
CitruUlinemias / Argininosuccinic Aciduria Negative
Tyrosinemias Negative
Amino Acidopathies, other Negative

Organic Acidemias:

Propionic / Methylmalonic Acidemias Negative
Isovaleric Acidemia / 2 Methylbutyric Acidemia Negative
Glutaric Acidemia Type 1 Negative
Guanidinoacetate Methyltransferase Deficiency Negative
Organic Acidemias, other Negative

Fatty Acid Oxidation Defects:

Medium Chain Acyl Dehydrogenase Deficiency Negative
Very Long Chain Acyl Dehydrogenase Deficiency Negative
Long Chain Hydroxy/ Acyl Dehydrogenase /Trifunctional Protein Deficiencies Negative
Carnitine Uptake Defect Negative
Fatty Acid Oxidation Disorders, other Negative

Galactosemia

Negative

Biotinidase Deficiency

Negative

Endocrine Disorders:

Congenital Hypothyroidism Negative
Congenital Adrenal Hyperplasia **POSITIVE**

Comments:

Screen positive for Congenital Adrenal Hyperplasia.

Sickle Cell and other Hemoglobinopathies Negative

Cystic Fibrosis Negative

Severe Combined Immune Deficiency Negative

Spinal Muscular Atrophy Negative

Mucopolysaccharidosis Type 1H (MPS1H or Hurler Disease) Negative

*Treatment Centre: Referred to HSC NBS Centre 416-813-7410 May 8, 2023

Comments:

Treatment Centre indicated above has informed family of results and is following up. No action on your part is required.

- o NSO's complete screening panel is available at www.newbornscreening.on.ca. Results for diseases not reflected above are pending.
- o Screen negative means that this infant is at decreased risk for the disease(s). Screen positive means that this infant is at increased risk for the disease(s). It does not mean that a disease is present, but further testing is indicated. If a test is positive and you and / or your patient have not already been contacted, please call NSO at (613) 738-3222.
- o This report does NOT contain information about this infant's carrier status for hemoglobinopathies. Families wishing to learn their child's carrier result should contact NSO or visit www.newbornscreening.on.ca
- o Infants with meconium ileus are at risk of having Cystic Fibrosis (CF) but will often have a screen negative result for CF; it is therefore recommended that they be clinically evaluated for CF regardless of the newborn screening results.

