

Newborn Screening Bulletin 2020-6

July 9, 2020

Screening for Spinal Muscular Atrophy and Mucopolysaccharidosis Type 1H (MPS1H or Hurler Disease)

We are pleased to announce Spinal Muscular Atrophy (SMA) and Mucopolysaccharidosis Type 1H (MPS1H or Hurler Disease) will soon be added to Ontario’s newborn screening panel. With a **launch date of July 27, 2020**, NSO will be the first newborn screening program in Canada to screen for these diseases.

Specimen collection

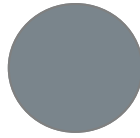
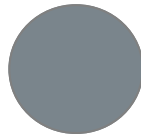
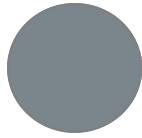
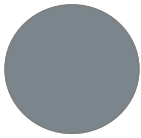
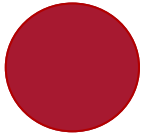
- Screening for SMA and MPS1H will be performed using the same samples currently being collected for newborn screening and no additional blood is required.
- In order to minimize the number of unsatisfactory samples, please do your best to completely fill as many of the 5 circles as possible and ensure the blood saturates to the back of the card. Please review the [Newborn Screening Manual](#) for a refresher of best practices in newborn screening specimen collection.

Reporting changes

- Once screening begins on July 27, 2020, SMA and MPS1H will be added to the newborn screening report; please see the mock report below.
- We recognize that changes may needed to be made to your internal information system to accommodate reporting of these results, and suggest that the LIS Codes “NBS-SMA” and “NBS-MPS1H” be used.
- Please note that the NSO screening pilot for SMA began on January 13, 2020 and will continue until the disease is formally included on the NSO panel on July 27, 2020. SMA pilot stickers will no longer need to be affixed to the Parent Information Sheet. A reminder that SMA screening results from the pilot phase are only available upon request.

Amino Acidemias:	
Phenylketonuria and Variants / Bioplerin Defects	Negative
Maple Syrup Urine Disease	Negative
Homocystinuria (Hypermethioninemias)	Negative
Citrullinemia / Argininosuccinic Aciduria	Negative
Tyrosinemia	Negative
Amino Acidopathies, other	Negative
Organic Acidemias:	
Propionic / Methylmalonic Acidemias	Negative
Isovaleric Acidemia / 2 Methylbutyric Acidemia	Negative
Glutaric Acidemia Type 1	Negative
Organic Acidemias, other	Negative
Fatty Acid Oxidation Defects:	
Medium Chain Acyl Dehydrogenase Deficiency	Negative
Very Long Chain Acyl Dehydrogenase Deficiency	Negative
Long Chain Hydroxyl Acyl Dehydrogenase /Trifunctional Protein Deficiencies	Negative
Camitine Uptake Defect	Negative
Fatty Acid Oxidation Disorders, other	Negative
Galactosemia	Negative
Biotinidase Deficiency	Negative
Endocrine Disorders:	
Congenital Hypothyroidism	Negative
Congenital Adrenal Hyperplasia	Negative
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





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Information about SMA and MPS1H

The information below about SMA and MPS1H should equip you to answer any questions you receive from parents about these diseases. We encourage you to review the additional information that will be available soon on the NSO website, and to contact us directly if you have any remaining questions.

About Spinal Muscular Atrophy (SMA)

SMA is a rare inherited (genetic) disease that results in muscle weakness and atrophy. Babies identified at a young age through screening can be treated as soon as clinically indicated. This screening uses a combination of MassArray and MLPA technologies, and will only detect cases of SMA caused by homozygous deletions of exon 7 in the SMN1 gene (~97% of cases of SMA). Only babies with homozygous deletions of SMN1, and a SMN2 copy number of 4 or less will be reported as screen positive. NSO will refer babies with positive SMA newborn screening results to specialists at one of the Newborn Screening Regional Treatment Centres in the province for diagnostic testing and treatment, if indicated. NSO anticipates that approximately 10-15 babies with SMA will be identified through newborn screening in Ontario each year.

About Mucopolysaccharidosis Type 1H (MPS1H or Hurler Disease)

Mucopolysaccharidosis Type 1H (MPS1H), often called Hurler Disease, is part of a group of rare genetic diseases called “lysosomal storage diseases”. MPS1H occurs when an enzyme called alpha-L-iduronidase (IDUA) is deficient. Early identification of MPS1H through newborn screening enables early treatment with bone marrow transplant or enzyme replacement therapy, which can prevent developmental disabilities, breathing and hearing problems. NSO will screen for MPS1H using a combination of biochemical and molecular technologies. NSO will refer babies with positive MPS1H newborn screening results to specialists at one of the Newborn Screening Regional Treatment Centres in the province for diagnostic testing and treatment, if indicated. NSO anticipates that between 4-8 babies with MPS1H will be identified through newborn screening in Ontario each year.

Please do not hesitate to contact us if you have any questions about the information included in this bulletin. We thank you for your continued dedication to newborn screening and hope you have a great summer.

