Newborn Screening Bulletin 2025-6

August 26, 2025

Update: Screening for X-linked Adrenoleukodystrophy (XALD)

In February, we announced that X-linked adrenoleukodystrophy (XALD) would soon be added to Ontario's newborn screening panel. For more information on XALD, you can refer to the initial submitter bulletin.

We launched the XALD screening pilot in March, with the original goal of formally adding XALD to the panel in August. However, due to delays in the legislative process required to support this addition, the official launch has been postponed. The pilot has been extended, and we anticipate implementation later this year. We will provide an update via a Newborn Screening Bulletin when an official launch date has been set.

Reporting changes

- Beginning September 29th, our newborn screening report will be updated to reorganize existing disorders. Once XALD screening is fully implemented, the report will also reflect the addition of XALD. Please see the mock report below.
- Please note that XALD will not appear on reports during the pilot phase.
- We understand that these changes may require updates to your internal information systems. To support this transition, we recommend using the LIS Code "NBS-XALD."

Existing:

Amino Acidemias:	
Phenylketonuria and Variants/Biopterin Defects	Negative
Maple Syrup Urine Disease	Negative
Homocystinuria (Hypermethioninemias)	Negative
Citrullinemias/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 Hydroxyl 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	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

New:

Disorders of Intermediary Metabolism:	
Phenylketonuria and Variants/Biopterin Defects	Negative
Maple Syrup Urine Disease	Negative
Homocystinuria (Hypermethioninemias)	Negative
Citrullinemias/Argininosuccinic Aciduria	Negative
Tyrosinemias	Negative
Propionic/Methylmalonic Acidemias	Negative
Isovaleric Acidemia/2 Methylbutyric Acidemia	Negative
Glutaric Acidemia Type 1	Negative
Guanidinoacetate Methyltransferase Deficiency	Negative
Medium Chain Acyl Dehydrogenase Deficiency	Negative
Very Long Chain Acyl Dehydrogenase Deficiency	Negative
Long Chain Hydroxyl Acyl Dehydrogenase /Trifunctional Protein Deficiencies	Negative
Carnitine Uptake Defect	Negative
Fatty Acid Oxidation Disorders, other	Negative
Additional Metabolic Disorders:	
Galactosemia	Negative
Biotinidase Deficiency	Negative
Mucopolysaccharidosis Type 1H (MPS1H or Hurler Disease)	Negative
X-linked Adrenoleukodystrophy	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

Thank you,
Newborn Screening Ontario

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