Newborn Screening Bulletin 2025-2

February 26, 2025

Screening for X-linked Adrenoleukodystrophy (XALD)

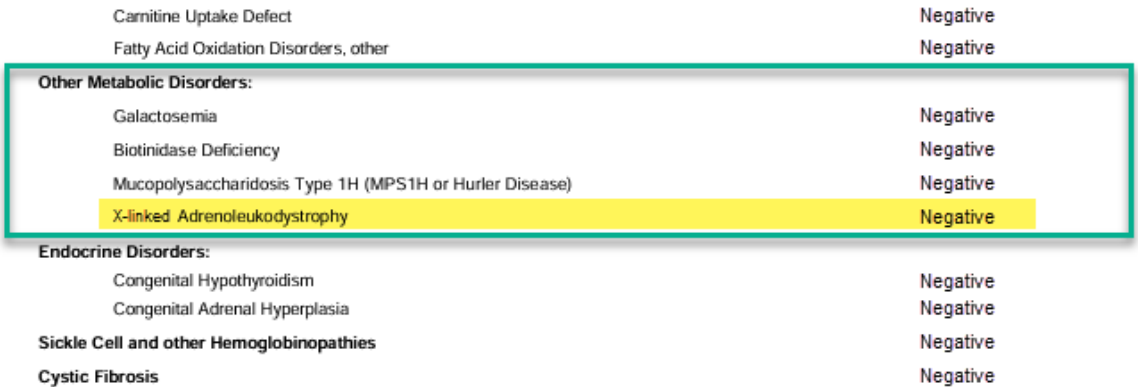
We are pleased to announce X-linked adrenoleukodystrophy (XALD) will soon be added to Ontario's newborn screening panel. The projected launch for our pilot implementation is **March 6, 2025.** During the pilot a random subset of the population will be screened for XALD (~270 samples/day). Any infant who has a screen positive result during the pilot will be referred to one of the retrieval centres at a tertiary care centre. The full implementation of XALD screening is projected to start in August 2025. NSO will be the first newborn screening program in Canada to screen for this disease.

Specimen collection

- No additional blood spots are required to be collected for XALD screening, either during the pilot or once it is fully implemented.
- To minimize the number of unsatisfactory samples, it remains important to 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 our Submitter Hub for a refresher of best practices in newborn screening specimen collection.

Reporting changes

- Once XALD screening is fully implemented (projected August 2025), this disorder will be added to the newborn screening report; please see the mock report below. During the pilot, it will not appear on reports. During the pilot, you will not know if any particular patient was screened for XALD and we will be unable to provide negative reports or results.
- 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 Code "NBS-XALD" be used.



Carnitine Uptake Defect	Negative
Fatty Acid Oxidation Disorders, other	Negative
Other 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

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Information about XALD

The information below about XALD should equip you to answer any questions you receive from parents about this disease. 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.

XALD is a rare inherited (genetic) disease that can cause problems with the function of the adrenal glands (adrenal insufficiency) and changes in the white matter of the central nervous system (leukodystrophy). Because it is an X-linked disorder, generally only boys are likely to develop the severe form of XALD (sometimes known as childhood cerebral adrenoleukodystrophy) that is the target of this screen. Babies who have XALD look normal when they are born. Without treatment, children with XALD will begin to have behavioural changes and loss of skills. These symptoms worsen over time, eventually leading to childhood death. Treatment is available. Children with XALD can benefit from being identified early, monitored for signs/symptoms and getting treatment sooner when needed.

XALD affects about 1 in every 15,000 male births in Ontario.

XALD happens when there is a genetic change in the *ABCD1* gene which causes the gene to not work properly. The job of the *ABCD1* gene is to help the body break down very long-chain fatty acids (VLCFAs). The genetic change causes the *ABCD1* gene to not work properly, resulting in the build-up of VLCFAs in the brain, nervous system and adrenal glands. The *ABCD1* gene is located on the X-chromosome. Individuals with one X chromosome (typically males) will be at risk of developing XALD. Individuals with two X chromosomes (typically females) are usually not at risk of developing XALD as they have two copies of the *ABCD1* gene.

Screening for XALD in Ontario will measure VLCFAs. If these are increased, counting of the *ABCD1* locus will be performed. Individuals with elevations in VLCFAs, who do not have two copies of the *ABCD1* locus (typically males), will be referred as screen positive.

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.

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