



Newborn Screening Bulletin 2019-2

June 12, 2019

NEWBORN SCREENING ONTARIO IMPLEMENTING A NEW LABORATORY INFORMATION SYSTEM

NSO is pleased to announce that we will be switching to a new laboratory information system (LIS) in July. While we anticipate a smooth transition, there may be delays in mailing and faxing reports as we adjust to the changes. We will ensure that these changes do not impact testing or referrals for infants who screen positive and will keep you updated about any changes that affect you.

PRIORITY SCREENING PANEL

NSO screens infants for rare, treatable diseases, some of which can present within the first few days of life. In our current system, we are unable to do any testing on samples with an insufficient quantity of blood to do the entire panel. The move to our new LIS in July will allow us to test these samples for the subset of diseases in our screening panel that can present with acute illness early on.

Samples that cannot be tested for the entire screening panel will be tested for the Amino Acidemias, Organic Acidemias, Fatty Acid Oxidation Defects, Galactosemia, and Congenital Adrenal Hyperplasia.

Repeat samples will be required for these infants to complete newborn screening testing. We anticipate this change will aid in the early identification of infants with critical illnesses. The newborn screening report will indicate “Negative/POSITIVE” for the diseases tested and “UNSATISFACTORY” for the diseases where testing could not be performed. It will also provide a reason for why the sample was unsatisfactory.

CHANGES TO THE NEWBORN SCREENING REPORT

The newborn screening report will have some minor changes in July. The Organic Acidemias section currently includes the C5OH related diseases, which were removed from the screening panel in 2017.

The C5OH related diseases will be removed and replaced with “Organic Acidemias, Other”.

Organic Acidemias:

Propionic / Methylmalonic Acidemias	Negative
Isovaleric Acidemia / 2 Methylbutyric Acidemia	Negative
Glutaric Acidemia Type I	Negative
3 Methylcrotonic / Hydroxymethylglutaric / Methylglutaconic / 2-Methyl, 3-Hydroxybutyric Acidemias, or β Ketothiolase Deficiency	No Longer Tested

Thank you in advance for your patience as we transition to our new system. Please do not hesitate to contact us if you have any questions about the information contained in this bulletin.

