



Newborn Screening Bulletin 2017-13

November 15, 2017

Removal of C5OH-Related Screening Targets from the NSO Panel

Please see the attached memo announcing the removal of C5OH-related targets from the NSO panel.

Please update your Laboratory Information Systems to reflect the following change in the Screening Report

- C5OH-related disorders are listed as the last line under **Organic Acidemias** and currently will have either a Negative or a Positive Screening Result

Organic Acidemias:	Negative
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	Negative

- After December 4th, the line will remain in the same place on the report, but the screening result will be noted as **"No Longer Tested"**

Organic Acidemias:	Negative
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

Please contact us at newbornscreening@cheo.on.ca if you have any questions about this change.

Thank you for your continued involvement in ensuring every newborn in Ontario receives newborn screening.

November 15, 2017

Newborn Screening Ontario Regional Treatment Centres and Submitters

RE: Removal of C5OH-Related Screening Targets from the NSO Panel

The Newborn Screening Ontario Advisory Council (NSO-AC) uses a formal review process to evaluate existing targets of the NSO panel to determine whether changes are needed to the current screening process, including potential removal of a disease from the panel. Based on the high number of false positives, the low positive predictive value, and the number of asymptomatic individuals that were being identified through screening, C5OH-related targets were nominated for review in 2016. C5OH-related targets include four primary screening target disorders: 3-Methylcrotonyl-CoA Carboxylase (3-MCC) Deficiency, Beta-Ketothialase Deficiency (BKT), Hydroxymethylglutaric (HMG) CoA-Lyase Deficiency, and Multiple Carboxylase Deficiency.

A task force of the Advisory Council undertook a thorough review of data collected between 2006-2014, as well as an extensive literature review. Upon completion of the review and following consultation with the metabolic specialists in the province, the task force presented its findings to the NSO-AC with the recommendation to remove C5OH-related disorders from the provincial screening panel. The Advisory Council voted unanimously to accept the recommendation of the task force. This recommendation was submitted to the Ontario Ministry of Health and Long Term Care and was accepted in the Fall of 2017.

To ensure sufficient time for all documentation and procedures to be updated to reflect this change, **Newborn Screening Ontario will no longer be screening for C5OH related disorders as of December 4th, 2017.**

The amount of blood required to perform the screen will not change, and there is no impact on the turn-around times for screening results. The NSO report will show "No longer tested" for C5OH related disorders results for samples received on or after December 4th, 2017.

In light of this change, and with the advice of the Advisory Council, NSO is reviewing the criteria used to count the number of diseases targeted by newborn screening. For example, Sickle Cell Disease is counted as one disease in some jurisdictions and three diseases in others. We have therefore removed a total number of diseases from NSO educational materials and emphasized the categories of disease and specific disease names instead.

While several disorders have been reviewed and some added to the NSO panel in the last few years, including Severe Combined Immuno-Deficiency (SCID) and Critical Congenital Heart Disease (CCHD), this is the first instance of a condition or set of conditions being removed. Critical review of the efficacy of the newborn screening panel is an important part of the ongoing quality management of the NSO system, and is vital to ensuring that the program is performing optimally and is providing high quality care for the newborns of Ontario.

Please do not hesitate to contact us with any questions.

