Newborn Screening Bulletin 2022-3

September 1, 2022

Screening for Guanidinoacetate Methyltransferase (GAMT) Deficiency

We are pleased to announce Guanidinoacetate Methyltransferase (GAMT) Deficiency will soon be added to Ontario's newborn screening panel. The projected **launch is Fall 2022.** Another communication will be sent when a formal start date is established. NSO will be the second newborn screening program in Canada to screen for this disease.

Specimen collection

- GAMT deficiency screening will be added onto the same testing platform used for the currently targeted organic acidemias and no additional blood is required.
- 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 the Newborn Screening Manual for a refresher of best practices in newborn screening specimen collection.

Reporting changes

- Once screening begins, GAMT deficiency 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 Code "NBS-GAMT" be used.

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

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NEWBORN SCREENING **ONTARIO**
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August 30, 2022

Information about GAMT Deficiency

The information below about GAMT deficiency should equip you to answer any questions you receive from parents about this disease. Babies with GAMT deficiency develop high levels of an organic acid called "guanidinoacetic acid" or GAA and so this disease is included in our report as an organic aciduria. 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 Guanidinoacetate Methyltransferase (GAMT) Deficiency

GAMT deficiency is a rare inherited (genetic) disease that affects how the body makes creatine. If the body does not have enough creatine, health problems like seizures can develop along with low muscle tone, muscle weakness, movement disorders, and intellectual disabilities. Babies identified through screening can be treated to prevent health and development problems.

GAMT deficiency happens when the enzyme guanidinoacetate methyltransferase is either missing or not working properly. The job of this enzyme is to create creatine from guanidinoacetate. When the guanidinoacetate methyltransferase enzyme is not working properly, creatine levels are low and guanidinoacetate levels are high. Newborn screening for GAMT deficiency measures the level of guanidinoacetate (GUAC) in the dried blood spot.

NSO will refer babies with positive GAMT deficiency newborn screening results to specialists at one of the Newborn Screening Regional Treatment Centres in the province for diagnostic testing and treatment, if indicated. GAMT deficiency is very rare. It is thought to affect between 1 in every 550,000 and 1 in every 2,500,000 babies born in Ontario.

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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