# Newborn Screening Bulletin 2022-4

September 28, 2022

## Screening for Guanidinoacetate Methyltransferase (GAMT) Deficiency

Guanidinoacetate Methyltransferase (GAMT) Deficiency will be added to Ontario's newborn screening panel **October 17, 2022.** Please refer to the <u>previous bulletin (2022-3)</u> for additional information on GAMT deficiency and changes to the newborn screening report.

### **Screening for Biliary Atresia**

Along with the addition of GAMT deficiency screening, NSO is working to implement a new screening initiative for biliary atresia scheduled for later this fall.

#### What is biliary atresia?

Biliary atresia is a rare but serious disease where bile cannot get from the liver to the stool, resulting in acholic (pale) stools. The accumulation of bile in the liver is damaging and without intervention can result in the need for transplant or eventually death. Ontario will be joining British Columbia as the second province in Canada to implement this type of screening.

#### How do you screen for biliary atresia?

This is the first of its kind in Ontario to involve parents as the primary screener with the help of an **infant stool colour card (ISCC)** to identify acholic stools which are often an early symptom of biliary atresia. Parents are simply asked to compare their child's stool colour to the images on the ISCC during regular diaper changes for the first month of life. If they detect acholic stool, they are asked to contact NSO using the contact methods shown on the card. When identification and treatment start early, babies with biliary atresia can have a better chance to grow and develop normally.

#### What does this mean for you?

- NSO will initially distribute the ISCCs to hospitals and midwifery clinics so they can be provided to all families
  with newborns in Ontario. ISCCs will then be made available, free of charge, using the same distributor used
  for newborn screening dried bloodspot collection cards.
- The ISCC itself is reasonably self-explanatory. A brief explanation to parents should be all that is required for its use.
- NSO will provide plenty of resources and educational support with materials soon to be published on our website, including translated copies of the card.
- We are currently targeting a timeline of early-mid November 2022 to start distribution of the ISCCs across the province.

Stay tuned – we will be sharing more information about screening for biliary atresia in the coming weeks. If you have any questions in the meantime, please don't hesitate to contact us at: <a href="mailto:nsoba@cheo.on.ca">nsoba@cheo.on.ca</a>

# 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.

www.newbornscreening.on.ca

