



Collection of Biliary Atresia (BA) Screening Information by Newborn Screening Ontario (NSO)

Newborn Screening Ontario (NSO), a provincial program located at the Children's Hospital of Eastern Ontario (CHEO), screens for over 30 rare, treatable diseases for all babies born in the province of Ontario. NSO added screening for biliary atresia (BA) to the newborn screening panel in January 2023.

In contrast to the standard screening via dried blood spot, this type of screening will typically be done at home, by the parent or guardian, for one month following the birth of the infant (or for one month post due date for pre-term infants). Parents/guardians will use an Infant Stool Colour Card (ISCC) as a reference to compare the colour of their infant's stool to 9 international reference stool colours depicted on the ISCC. In addition to the reference stool images, the card will contain information about BA and what to do if an infant is identified to have acholic (pale) stool (primary identification of BA).

If a parent/guardian suspects that their newborn has acholic stool, they should contact NSO's clinical team for decision support and next steps. NSO has three contact methods for parents to get in touch with the team by:

- Phone: Parents/guardians can contact NSO using the BA screening phone number 1-833-POOP-CHK (1-833-766-7245). This phone number is routed to the NSO clinical team directly for triaging cases.
- Email: Parents/guardians can email NSO using the BA screening email NSOBA@cheo.on.ca and may attach images of their child's stool if available. This email is monitored by the NSO clinical team for triaging cases.
- Webform: Parents/guardians can use the BA screening contact form on the NSO website to contact NSO and attach images of their child's stool if available. The form collects all the required information to match a patient to its existing newborn screening record, thereby enabling the NSO clinical team to open a clinical assessment case.

Through these methods for decision support, NSO collects personal information (PI) and personal health information (PHI) in the form of demographic information, qualitative stool colour information, and stool images. The collection of demographic information will be used to match the infant to an existing patient profile in NSO's Laboratory Information System and to contact the parent/caregiver about their concern. NSO may also use the PI and PHI collected for secondary uses as described in the section below.

NSO will use information collected through the different contact methods to:

Primary Uses



A. Ensure that systems are in place to support parents and families of newborns such that all infants have access to BA screening according to Ontario standards. Using the demographic and screening data submitted, NSO will identify and follow up on cases to:

- i. Interpret patient information to determine screening results for biliary atresia and make medical referrals to health care providers for diagnostic and follow up testing as needed.

NSO will identify cases where stool screening results may be indicative of BA (i.e., screen positive for BA). NSO will use this information to produce a referral package and refer the infant to one of five paediatric academic health sciences centres (PAHSC) in Ontario and notify the infant's primary health care provider (if applicable). Results from some diseases on the dried blood spot screening panel may be useful in the interpretation of results for second tier testing for BA. NSO will include these results in the referral package to the PAHSC. Upon receipt of a referral, specialists at the PAHSC will follow up with the family to coordinate care and follow up with a health care provider (if applicable).

NSO will request images of the stool to help determine whether a child screens positive for BA. Although providing stool photos is not mandatory, it is extremely helpful and is strongly encouraged to help ensure that our telephone triage team can make the most accurate assessment of cases where there is a concern about pale stool. The images collected will be linked to the infant's chart in NSO's Laboratory Information System and stored on NSO's secure servers.

In cases where the infant does not screen positive (i.e. the stool screening result is not indicative of an increased risk for BA), NSO will contact the child's primary health care provider (if applicable) to share details of the patient's interaction with NSO and the screening outcome. A letter will be sent to the primary health care provider and the parent/guardian, encouraging them to continue screening for 30 days (if applicable).

- ii. Reduce risk of interpretation

When the screener (parent/guardian) contacts NSO for assistance in screening their infant for BA, NSO will provide guidance to the parent to help them screen appropriately and to address their questions and/or concerns related to the stool screening. For instance, if a parent feels the colour of their infant's stool has become normal, but upon review of stool images NSO feels that the stool is still acholic, NSO would arrange for the referral of the child to a PAHSC and identify the child as a screen positive case.



iii. Reduce risk of missed cases

NSO will identify cases where infants were eventually diagnosed with BA, but screening may not have occurred and/or NSO was not contacted for clinical assessment with a possible screen positive result.

NSO has put in place channels to be alerted by paediatric gastroenterologists, and hepatologists in Ontario authorized to perform the Kasai procedure in case a patient is referred for the procedure, but screening and referral was not overseen by NSO. NSO will communicate with the hepatologist(s) and child's primary care provider and, once consent is obtained from the infant's parent/guardian to share PHI about the infant with NSO, record the reason(s) for which the child was not screened. NSO may follow up with the hospital, birthing centre, or midwifery practice that offered newborn screening to the child to reiterate the importance of educating parents about screening for BA in the first 30 days of life.

B. Assure the quality of and evaluate the provincial program.

- Evaluate the screening protocol and Infant Stool Colour Card (ISCC) screening tool.
- Evaluate the performance of the provincial BA screening program.
 - Assess the coverage and utilization of the screening tool
 - Minimize the number of missed screens
- Identify areas of improvement for the screening program such as:
 - The ISCC screening tool and the at-home screening process
 - Communications to parents, families, health-care providers
 - Time between screen positive and referral to PAHSC
 - Time between referral and treatment



Secondary Uses

- A. Develop and maintain BA screening standards, guidelines, screening tools, as well as educational materials.
- Continuously improve BA screening standards and guidelines.
 - Develop educational tools and communication strategies to increase awareness and support Ontario-wide BA screening.
 - Develop, improve, and maximize the utilization of screening tools and aids to enhance the parent-driven screening experience and accuracy of screening.
 - Use qualitative stool information (colour, texture, reference to abnormal stool colours on ISCC, and other information) to assess the effectiveness of the ISCC for the detection of acholic infant stools.
 - Collect samples of deidentified stool images for the purposes of developing technologies for the detection of acholic infant stools. These technologies may include:
 - i. artificial intelligence; and/or
 - ii. machine learning algorithms and models
- B. Conduct research following the rules set out in Ontario's Personal Health Information Protection Act, 2004 to contribute to further understanding of BA screening procedures, policies, and overall program outcomes. All research must be approved by the CHEO Research Ethics Board.