NEWBORN SCREENING
ONTARIO
DÉPISTAGE DES NOUVEAU-NÉS
DE L'ONTARIO

BULLETIN #62
January 24, 2013

1. Severe Combined Immune Deficiencies (SCID) screening to begin in April 2013

We are pleased to announce that the Government of Ontario has accepted the recommendation from the Newborn and Childhood Screening Subcommittee to add Severe Combined Immune Deficiencies to Ontario's newborn screening panel. This recommendation was endorsed by the Provincial Council on Maternal and Child Health (PCMCH) and Newborn Screening Ontario, and NSO is targeting April 22, 2013, to begin screening. Based on direct communications with other jurisdictions already screening for SCID, we estimate that approximately 50 infants will screen positive each year, and about 5-10 will have SCID.

SCID screening will be performed from the same samples currently being collected for newborn screening; no additional blood collection will be required.

Newborns with SCID appear healthy at birth, but are vulnerable to multiple life-threatening infections within a few months of life. Early diagnosis and treatment with stem cell transplant improves the long-term outcome of affected infants. Since SCID is not apparent at birth and early recognition is essential for lifesaving treatment, SCID is being added to our newborn screening panel.

NSO will use a real-time polymerase chain reaction assay to measure T-cell receptor excision circles (TRECs), which are formed in the maturation of normal T-cells. A lack or very low number of TRECs is consistent with T-cell deficiency. Infants who screen positive for SCID will be referred for diagnostic evaluation.

2. Recommendations for the collection of newborn screening samples from premature and/or low birth weight infants

Newborn Screening Ontario, with input from the provincial pediatric endocrinologists and neonatologists, has created recommendations surrounding premature and/or low birth weight infants and the timing of their newborn screen collection. In short, the policy recommends a first newborn screening specimen be collected between 24 and 72 hours of age and a second specimen collected at 3 weeks of age. Not all hospitals currently have this specimen collection algorithm in place and therefore, we appreciate that adapting these recommendations may take some time to implement.

As outlined above, NSO will be starting to screen for SCID in late April 2013. Premature infants have a higher false positive rate with SCID screening. Therefore, the first follow up investigation for screen positive premature infants is to retest a second newborn screening sample taken at a later age. The timeline for the repeat screening would be the same as the recommendations in this policy and so we would encourage adoption of these recommendations prior to the launch of SCID screening, if at all possible.

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In an effort to assist institutions that do not have tracking systems already in place, NSO can provide institutions with information regarding infants who require a repeat specimen collection. Please let us know if you have a system already in place and do not wish to receive these notifications. For institutions that have their own guidelines for internally screening infants by measuring FT4 and TSH, you may still receive requests for repeat NBS specimens for premature infants who screen positive for SCID. A separate system would be in place for infants who screen positive for SCID on their initial NBS sample and require a repeat sample.

More detailed information about SCID screening and recommendations for the collection of newborn screening samples for premature and/or low birth weight infants will be available soon on our website at www.newbornscreening.on.ca.

If you have any questions about this bulletin, please do not hesitate to contact NSO.

These bulletins are circulated to keep you abreast of changes to Newborn Screening Ontario. Please share this information with any relevant personnel in your hospital / practice. If there is someone who would like to be added to this list, contact NSO at: 1-877-627-8330, option #0 or at NewbornScreening@cheo.on.ca.

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Recommendations for Premature or Low Birth Weight Infants

Premature or low birth weight babies may have a delayed rise in TSH even if they have Congenital Hypothyroidism (CH). As Newborn Screening Ontario (NSO) uses elevation of TSH as the screening marker for CH, there is an increased risk of a false negative result (missed case) if these babies are screened only once in the early neonatal period. As well, premature newborns are known to have a lower number of T-cell receptor excision circles (TRECs) and are more likely to falsely screen positive for Severe Combined Immune Deficiencies (SCID). Therefore, the first follow up investigation for screen positive premature infants is to retest a second newborn screening sample taken at a later age. It is therefore important that screening samples are taken as outlined below.

Premature (<33^0 WGA) or very low birth weight (<1500g) babies should have:

1. A first Newborn Screening specimen collected between 24 and 72 hours of age

2. A second specimen collected at 3 weeks of age or when the baby is being discharged home from the hospital, whichever comes first.

   2.1. If the baby is discharged home prior to 3 weeks of age from a hospital with a robust tracking system to ensure follow-up, an outpatient appointment between 3 and 4 weeks of age can be arranged without the need for a blood draw prior to discharge.

   2.2. If the baby is discharged home with the second specimen collected before 3 weeks of age consideration should be given to having a third specimen arranged as an outpatient between 3 and 4 weeks of age.

3. If the baby is being transferred to another hospital after 3 weeks of age, the hospital receiving the baby should confirm that the second sample was taken prior to transfer. If it was not taken, the receiving hospital should take the second sample as soon as possible after the baby arrives. The receiving hospital can also call NSO to inquire whether a second sample was received.

The following exceptions to the above policy apply:

1. A specimen should be collected prior to the baby receiving a blood transfusion, even if this is prior to 24 hours of age. This will ensure a satisfactory screen for:
   a. Galactosemia since the test relies on measurement of the Galactose-1-Phosphate Uridyltransferase enzyme activity in red blood cells.
   b. Hemoglobinopathies since the adult hemoglobin from the transfused blood may mask an abnormal hemoglobin pattern.
   c. If the first sample is taken at less than 24 hours of age, a second sample should be taken between 24 hours and 7 days of age.

2. Total Parenteral Nutrition (TPN). Amino acid solutions administered as part of TPN are a common reason for false positive screening results for amino acid diseases in premature / LBW babies. Ideally, a specimen should be collected at a time when the baby is receiving lower amounts of TPN. For example, if the baby is receiving 1.5 g/kg/d of amino acids at 24 hours of age, it is preferable to take the screening sample before this amount is increased. Please note that there is no increased risk of a false negative result if a sample is taken when the baby is receiving higher amounts of amino acid solutions; screening should not be delayed beyond 72 hours of age for this reason.

Premature infants ≥33^0 WGA and >1500g should be screened no differently than term babies.

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