Information for Primary Care Providers

Alberta's NMS Program Panel Expansion Overview

March 2019

What is changing?

Newborn blood spot screening in Alberta will be expanding in a staged approach, beginning April 1, 2019. Four new conditions will be added to the screening panel including: tyrosinemia type 1, classic galactosemia, severe combined immunodeficiency and sickle cell disease.

Where do I find more information about these conditions?

Condition specific information sheets can be found by visiting www.ahs.ca/newbornscreening and clicking on 'Information for Health Professionals' then 'Resources'.

The condition information sheets contain details regarding clinical presentation, pathogenicity and diagnostic testing for each of the conditions.

What happens if a baby in my care has an abnormal newborn screen?

The ordering healthcare provider will be notified by telephone of the result by the newborn screening laboratory. The appropriate on-call specialist, such as a hematologist, immunologist or metabolic specialist, will also be notified by the newborn screening laboratory. Follow-up diagnostic testing and, if needed, treatment will be managed through the relevant specialty clinic.

Are there any incidental findings?

Newborn screening for sickle cell disease will identify babies with sickle cell trait (SCT). An individual has SCT when they have only one sickle cell genetic variant. There are no immediate health concerns for a baby who carries SCT. However, there may be implications for other family members. Long term health implications have also been reported in some individuals with SCT. The ordering healthcare provider will receive a letter notifying them of the baby's SCT status as well as an information sheet. Ordering providers will be responsible for disclosing SCT results to the family and offering hemoglobinopathy investigations to the parents if the family is planning more pregnancies.

Can there be false positive and false negative results?

Because the newborn screen is a screening test and is not a diagnostic test, there is the possibility of false negative and false positive results. Any baby with clinical features suggestive of a screened conditions requires clinical and diagnostic follow-up regardless of whether or not the newborn screen result is normal.

What if I have more questions?

If you have questions about sample collection, please contact the Newborn Metabolic Screening Program at nmsprogram@ahs.ca

If you have questions about screened conditions, please contact the Genetic Counsellors at nmsgeneticcounsellors@ahs.ca

Alberta Health Services
Newborn Metabolic Screening