

NEWBORN SCREENING NEWSLETTER

JUNE 17, 2016



Krabbe Screening will begin on July 1, 2016

The newborn screening laboratory will begin screening babies for Krabbe disease. Krabbe disease is a progressive neurodegenerative disease with a variable age of onset and severity. In the most severe infantile form, symptoms begin in the first few months of life. Affected babies present with irritability, feeding problems, and muscle rigidity. Neurologic deterioration progresses rapidly and death usually occurs in the second year of life. There is no cure for Krabbe disease; however, a bone marrow stem cell transplant may slow the progression of the disease if done before symptoms present. It is estimated that one to two babies are born in Ohio each year with the severe infantile form of Krabbe disease. Without newborn screening, a diagnosis of Krabbe disease cannot be made in time for parents to consider treatment options.

The Ohio Department of Health is mandated by law to begin screening of Krabbe disease for specimens received by the laboratory on July 1, 2016. The law gives parents/guardians the right to refuse screening for Krabbe disease. (The law does not allow parents to refuse screening for any other conditions included on the Ohio Newborn Screening Panel.) Parents must be given information about their right to refuse this screening prior to the collection of the blood specimen for any NBS to be shipped to the Newborn Screening Laboratory on or after June 30, 2016. The ODH has developed an insert, "*Screening for Infantile Krabbe Disease*", for the "*Why Must My Baby Be Screened*" pamphlet to inform families about Krabbe disease screening and their right to refuse this screening. The NBS coordinator at each birth facility will be mailed inserts for the pamphlets. The insert will also be available to print from the Ohio Newborn Screening website. Each birth facility and midwife practice must decide how they will assess and document parental refusal of Krabbe screening in the patient record.

Procedure for sending specimen when parents have refused

In order for the newborn screening laboratory to process specimens in accordance with the parent/guardian's wishes, the laboratory must be notified that Krabbe screening has been refused at the time the specimen is received at the lab. When Krabbe screening has been refused, facilities are asked to apply a "Krabbe Refusal" sticker to the top front page of the NBS kit (see image at right) prior to shipping the specimen. The newborn screen coordinator at each birthing facility will be mailed a supply of bright green stickers for this purpose. The sticker should be placed on the NBS kit only when the parent/guardian refuses Krabbe screening. If no stickers are available, the person who is notified of the refusal should write "Krabbe Screening Refused" in large letters in the box at the top of the front page of the NBS kit. In the near future, kits will be printed with a check-box for recording refusal of Krabbe screening.



Follow-up of Elevated Risk Results

Any infant that has been identified as being at an "elevated risk" for Krabbe disease based on newborn screening results should immediately be referred to a Regional Genetics Center for evaluation.

Increase in Newborn Screening Fee

There will be an increase of \$11 per newborn screening kit to expand testing to include Krabbe disease in the Ohio Newborn Screening Panel. Please notify your accounting office of the upcoming price change to ensure that your orders are processed correctly. Newborn screening kits ordered (postmarked if mailing) on or after July 1, 2016 will cost \$74.61 per kit purchased. Updated order forms will be mailed to each birth facility and are available on the Ohio Newborn Screening web site.

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