

NEWBORN SCREENING NEWSLETTER

NOVEMBER 15, 2016



Krabbe Screening began on July 1, 2016

The newborn screening (NBS) laboratory now screens 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 was mandated by law to begin screening of Krabbe disease for specimens received by the laboratory starting 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. 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. Supplies of these inserts can be requested on the literature and forms requisition. The insert is also available to print from the Ohio Newborn Screening website. Each birth facility and midwife practice should have developed a process to 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 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. Additional stickers can be requested on the literature and forms requisition. The newest lot of NBS kits has a check-box for recording refusal of Krabbe screening on line 15. (15) CHECK IF PARENT / GUARDIAN HAS REFUSED KRABBE SCREENING This checkbox should be used **in addition to the green sticker.** If the family does not refuse Krabbe screening, please leave line 15 blank - no words or stray marks should be placed in this area of the kit.



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

Please notify your accounting office of the price change to ensure that your orders are processed correctly. The Newborn screening kit cost is \$74.61 per kit purchased. Updated order forms were mailed to each birth facility and are available on the Ohio Newborn Screening web site.

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