



MEMORANDUM

DATE: May 1, 2023

TO: South Carolina Newborn Screening Community

FROM: Nicolas Epie, PhD, HCLD, TS (ABB), MT(ASCP)
SC DHEC Public Health Laboratory (PHL) Director

CC: Kimberly N. Seals, MSPH, MPA
Director, Bureau of Maternal and Child Health

SUBJECT: Newborn Screening for Krabbe Disease

Beginning the week of May 15th, Krabbe screening will be added to the South Carolina Newborn Screening (NBS) Panel. The Health Resources and Services Administration (HRSA) estimates that approximately 40 babies are born with this rare condition each year in the United States. This screening will allow infants potentially affected with Krabbe disease to be identified and treated as soon as possible. Screening will consist of two tier testing.

First tier screening will be performed using a quantitative test for β -galactocerebrosidase (GALC), the enzyme that is deficient in newborns affected with Krabbe disease. The screening assessment will be based on the daily median of GALC results; the expected range will be ">15% of the daily median". Results will be reported as "*Within Acceptable Limits*" or "*Outside Acceptable Limits.*" Additionally, GALC screening results that are reported as "*Outside Acceptable Limits*" ($\leq 15\%$ of the daily median) will include their calculated percentage of the daily median.

Initial specimens reported as, "*Outside Acceptable Limits*" will be sent to a reference lab for 2nd tier psychosine testing. SC Maternal and Child Health (MCH) NBS staff will provide notification of abnormal psychosine results to the primary care provider and/or physician indicated on the NBS specimen collection form by phone, fax, and/or mail.

Note, decreased enzyme activity is suggestive of Krabbe disease. However, this result alone does not exclude pseudo deficiency, which causes decreased enzyme levels without disease. Combined evaluation of GALC enzyme activity and psychosine concentration predict the phenotype (unaffected vs. early vs. late onset Krabbe disease).

Please direct any questions to Beth Bair, MS, Chemistry Division Director at bairea@dhec.sc.gov; 803-896-0991 or Tanya Spells, MS, MLS(ASCP), NBS Follow up Program Director at spellsty@dhec.sc.gov; 803-898-0619.