

Department of Public Health Newborn Screening Update – Krabbe Disease Screening

TO: Birthing Facilities and Hospitals within the State of Georgia
FROM: DPH Newborn Screening Program
RE: Krabbe Disease Pilot Project Screening
DATE: November 30, 2021

Background: Krabbe disease is a very rare inherited disorder that can cause severe neurological problems or even death. In 2017, the Georgia General Assembly mandated parents be made aware of the option to have their newborns screened for Krabbe disease shortly after birth. In 2021, the passing of House Bill 567, authorized the Georgia Department of Public Health (DPH) to begin screening for Krabbe disease at the Georgia Public Health Laboratory.

Program Update: Krabbe disease was recently added to the state newborn screening panel as part of a three-year pilot project. The Georgia Newborn Screening (NBS) program now screens for more than 35 primary conditions, including Krabbe disease. Newborn screening bloodspot specimens that are collected in county health departments, birthing facilities, hospitals, or providers' offices will be tested for the full panel of NBS disorders, including Krabbe disease.

Follow-up and Support for Families: The NBS program has partnered with Emory University Department of Human Genetics to conduct follow-up for infants with presumptive positive newborn screens for Krabbe disease. If a newborn has a presumptive positive newborn screening result for Krabbe disease, the newborn's primary care physician and Emory University Department of Human Genetics will be notified to coordinate confirmatory testing and treatment following a positive diagnosis.

There will be no added burden placed on hospital staff from the inclusion of Krabbe disease on the state newborn screening panel. Birthing facility and hospital staff should continue to collect newborn screening specimens as outlined in the Georgia Newborn Screening Policy & Procedure Manual.

If you have additional questions or need more information, please contact Tina Turner, DPH Child Health Services Deputy Director at Tina.Turner1@dph.ga.gov