

EXECUTIVE SUMMARY

This summary reviews the information the federal advisory committee used when deciding whether to recommend adding Krabbe Disease to the Recommended Uniform Screening Panel (RUSP) in 2009.

About the condition

Krabbe Disease is a rare disorder that affects the nervous system. About 1 out of every 100,000 babies has Krabbe Disease. People with Krabbe Disease do not have enough of the GALC enzyme that helps the body break down fats in the nervous system. Babies with Krabbe Disease appear normal. There are 4 types of Krabbe Disease: early infantile, late infantile, juvenile, and adult. Most children have the infantile types. Krabbe Disease can cause problems with development, movement, and other symptoms that can worsen quickly and cause death by age 2.

Treatment for Krabbe Disease

There is no cure for Krabbe Disease. Early diagnosis allows early treatment, which could improve outcomes for some babies with the disease. Hematopoietic stem cell transplant, also called a “bone marrow transplant,” is a treatment that might stop symptoms from getting worse. Whether and when a baby needs this treatment depends on many things, like the type of Krabbe Disease.

Detecting Krabbe Disease in newborns

Newborn screening for Krabbe Disease can happen along with routine newborn screening for other conditions during the first few days of life. Newborn Krabbe Disease screening measures GALC enzyme levels and looks for changes in the GALC gene. This process uses the same dried blood spots already collected for screening of other disorders. Newborns with low GALC enzyme levels and changes in the GALC gene are at higher risk for Krabbe Disease. They need more testing to diagnose the condition.

Public health impact

Detailed data on how newborn Krabbe Disease screening would affect public health were not available at the time of the report. However, newborn screening for the disease allows diagnosis and monitoring early in life. Without screening, diagnosing the disease can take 5 months or longer after symptoms begin. Newborn screening allows diagnosis and treatment before severe symptoms start.

Committee decision

The Committee voted in 2009 to recommend not adding Krabbe Disease to the RUSP. To add Krabbe Disease to the RUSP, experts need to know more about how to define disease types, the best screening method, and how well treatment works.