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 2023.

About the condition

Krabbe disease is a rare condition that affects about 1 out of every 100,000 people. People with Krabbe disease have too little of an enzyme called GALC. This leads to death of nerve cells, including those in the brain. Krabbe disease causes health problems, like severe irritability and trouble moving. These problems can worsen very quickly without treatment, especially if they arise in the first 6 months of life. Babies affected by Krabbe disease in their first year usually die in early childhood.

Treatment for Krabbe disease

Doctors may recommend hematopoietic stem cell transplant as a treatment for Krabbe disease. This treatment can lower risk of death in early childhood and help with some symptoms but does not cure Krabbe disease. For babies with severe Krabbe disease, doctors recommend treatment by 6 weeks after birth. Children at risk for later-onset Krabbe disease may get treatment if symptoms arise.

Detecting Krabbe disease in newborns

Newborn screening for Krabbe disease can be included with routine newborn screening. There are a few steps in screening. The first step uses the same dried blood spots collected to screen for other conditions to check for low GALC enzyme levels. The second step checks the spots for high levels of psychosine, a substance that builds up in Krabbe disease. Sometimes, the second step checks the blood for gene changes linked to Krabbe disease. Newborns whose screening results reveal higher risk for Krabbe disease should see a specialist right away.

Public health impact

Experts think that screening all newborns in the US would find about 70 babies with or at risk for Krabbe disease each year. Not all of these would have Krabbe disease or need treatment. About 15 of these babies would have the most severe Krabbe disease type. Treatment would prevent about 10 babies from dying in early childhood, though many treated children would still have some level of disability.

Committee decision

The Committee voted in 2023 not to recommend adding Krabbe disease to the RUSP. The Committee noted gaps in what is known about how well HSCT works, the risks of HSCT in very young babies, and the outcomes of late infantile Krabbe disease found by screening babies. Krabbe disease may be reconsidered for the RUSP in the future.