

EXECUTIVE SUMMARY

This summary reviews the information the federal advisory committee used when deciding whether to recommend adding Infantile Krabbe disease to the Recommended Uniform Screening Panel (RUSP) in 2024.

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. Most babies with Krabbe disease have the infantile form, and are affected in the first 12 months after birth. Infantile Krabbe disease (IKD) is the most severe form, causing health problems that worsen quickly and lead to death in early childhood without treatment.

Treatment for Infantile Krabbe disease

Doctors may recommend hematopoietic stem cell transplant (HSCT) as a treatment for IKD. This treatment can lower risk of death in early childhood and help with some symptoms, but does not cure Krabbe disease. For babies with IKD, doctors recommend treatment by 6 weeks after birth before major signs of disease.

Detecting Infantile Krabbe disease in newborns

Newborn screening for IKD 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 IKD. Sometimes, the second step will also include a check for gene changes linked to Krabbe disease. Newborns whose screening results reveal higher risk for IKD should see a specialist right away.

Public health impact

Screening all newborns in the US is expected to find about 11 babies with Infantile Krabbe disease each year. About 10 of these babies with IKD would likely be treated with an HSCT. Of the babies who receive an HSCT, 1 baby would likely die within 100 days and the others would be alive at 2.5 years but have some level of disability.

Committee decision

The ACHDNC voted in 2024 to recommend adding Infantile Krabbe disease to the RUSP. The ACHDNC noted the benefit of early treatment with HSCT at increasing survival of babies with IKD, although other health problems may remain.