



**ACHDNC**

*Advisory Committee  
on Heritable Disorders in  
Newborns and Children*

# Krabbe disease Summary

Re-nomination package

# Chair Letter Summary

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- Requested additional published data on the following topics:
  - » Evidence regarding efficacy of HSCT for early infantile Krabbe disease that (1) systematically reports outcomes and (2) adequately classifies cases as early vs. late onset (e.g. psychosine levels or genetic markers of infants transplanted).
  - » Evidence regarding harms of HSCT for infants transplanted in the first two months of life, including possibility of transplant of infants not meeting criteria for early infantile disease.
  - » Evidence regarding outcomes of infants who are at risk of late infantile Krabbe disease, as these children may experience even greater benefits than early infantile Krabbe disease; such evidence should include number lost to follow-up and potential burden on families and infants of intensive follow-up visits and consequences of indeterminate diagnostic testing.

# Background: Material Change

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- Must be a change in scope of the condition nominated and/or substantial new evidence for the nominated condition
  - If there is a change in scope, it is preferable that there is also new evidence provided in support
- Nomination package resubmission should address Committee questions and comments described in the Chair letter to the nominators

# Background: Timeline

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- ACHDNC voted to not recommend Krabbe disease for inclusion into the RUSP at the February 2023 meeting
- ACHDNC Chair sent letter to Krabbe nominators in March 2023
- Krabbe disease nominators resubmitted the nomination package and provide a response to Chair letter in July 2023

# Does Krabbe disease meet criteria for Expedited Review?

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- Has it been less than one year since the ACHDNC vote? **Yes**
- Is there a material change in the new nomination? **Yes**
- Material Change:
  - » Change in the target of screening to early infantile Krabbe disease (onset within the 1<sup>st</sup> year of life) with reduced GALC enzyme activity and elevated psychosine in the newborn dried-blood spot
  - » There are no additional outcomes data, but we believe the change in target of screening may change the benefit/harm ratio

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# Discussion