

Newborn Screening ACT Sheet

Krabbe Disease

Condition Description: Krabbe disease is a lysosomal storage disorder (LSD) and neurodegenerative disease caused by a deficiency of lysosomal galactocerebrosidase due to a mutation in the corresponding gene (*GALC*). There is wide variability in severity and age of onset. Krabbe disease is an autosomal recessive disorder.

YOU SHOULD TAKE THE FOLLOWING ACTIONS IMMEDIATELY:

- Consult with genetic metabolic specialist.
 - Contact family to inform them of the newborn screening result.
 - Evaluate the newborn with particular attention to neurologic dysfunction.
 - Provide the family with basic information about Krabbe disease.
 - Report confirmatory findings to newborn screening program.
-

Diagnostic Evaluation: Follow-up testing should include galactocerebrosidase enzyme assay and *GALC* gene analysis, as well as immediate assessment for neurologic involvement, including brain MRI, lumbar puncture for cerebrospinal fluid evaluation and clinical neurologic examination.

Clinical Considerations: The clinical presentation of Krabbe disease ranges from a rapidly progressive infantile form, which is uniformly lethal if untreated, to a more slowly progressive late-onset form. All forms of the disorder are associated with leukodystrophy and neurologic deterioration, but the age of onset and rate of progression vary widely. Mutation analysis of the *GALC* gene may provide information on expected age of first symptoms. The only available therapy is early hematopoietic stem cell transplantation.

Additional Information:

[Genetics Home Reference](#)

[Gene Reviews](#)

[Hunter James Kelly Research Institute](#)

Referral (local, state, regional and national):

[Testing](#)

[Clinical Services](#)

[Find Genetic Services](#)

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

© American College of Medical Genetics and Genomics, 2012 (Funded in part through MCHB/HRSA/HHS grant #U22MC03957)

LOCAL RESOURCES: Insert State newborn screening program web site links

State Resource site (insert state newborn screening program website information)

Name	<input type="text"/>
URL	<input type="text"/>
Comments	<input type="text"/>

Local Resource Site (insert local and regional newborn screening website information)

Name	<input type="text"/>
URL	<input type="text"/>
Comments	<input type="text"/>

APPENDIX: Resources with Full URL Addresses

Additional Information:

Genetics Home Reference

<http://ghr.nlm.nih.gov/condition/krabbe-disease>

Gene Reviews

<http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gene&part=krabbe>

Hunter James Kelly Research Institute

http://www.huntershope.org/site/PageServer?pagename=hjkri_centerforkrabbedisease

Referral (local, state, regional and national):

Testing

http://www.ncbi.nlm.nih.gov/sites/GeneTests/lab/clinical_disease_id/2189?db=genetests&country=United%20States

Clinical Services

<http://www.genetests.org>

Find Genetic Services

<http://www.acmg.net/gis>

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

© American College of Medical Genetics and Genomics, 2012 (Funded in part through MCHB/HRSA/HHS grant #U22MC03957)