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

ACMG
American College of Medical Genetics and Genomics
Translating Genes Into Health®

LOCAL RESOURCES: Insert State newborn screening program web site links

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

Name	
URL	
Comments	

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

Name	
URL	
Comments	

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

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