

Newborn Screening ACT Sheet

Fabry Disease

Condition Description: Fabry disease is a lysosomal storage disorder (LSD) caused by a defect in the alpha galactosidase A (*GLA*) gene, resulting in progressive accumulation of globotriaosylceramide (GL-3) throughout the body. Although Fabry disease is an X-linked disorder, there can be significant morbidity and mortality in females as well as males.

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Consult with genetic metabolic specialist.
 - Contact family to inform them of the newborn screening result.
 - Elicit family history of kidney disease, heart disease, stroke or paresthesias.
 - Provide the family with basic information about Fabry disease.
 - Report confirmatory findings to newborn screening program.
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Diagnostic Evaluation: Confirmatory alpha-galactosidase A enzyme assay in males. Confirmatory testing in females is *GLA* gene sequencing. Males who are confirmed to have low alpha-galactosidase A enzyme activity should also have *GLA* gene sequencing.

Clinical Considerations: Most significant clinical manifestations of disease include kidney disease, hypertrophic cardiomyopathy and central nervous system (CNS) evidence of strokes. Males with Fabry disease typically present in childhood with acroparesthesia of the hands and feet when febrile or exposed to excess heat or cold. Untreated, death may occur in the fifth and sixth decade from renal or cardiac failure or secondary to CNS infarcts. Adult females often have acroparesthesia and may have organ involvement. Enzyme replacement therapy is available for patients affected with Fabry disease and may be most effective if initiated at the first sign of clinical symptoms. ERT is highly complicated and should be given only under the guidance of a specialist with expertise in lysosomal storage disorders. Family studies are appropriate if the newborn screen is confirmed.

Additional Information:

[Genetics Home Reference](#)

[OMIM](#)

[ClinGen Actionability Report](#)

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.

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/fabry-disease>

OMIM

<http://www.ncbi.nlm.nih.gov/omim/301500>

ClinGen Actionability Report

<https://actionability.clinicalgenome.org/ac/Adult/ui/stg2SummaryRpt?doc=AC047>

Referral (local, state, regional and national):

Testing

http://www.ncbi.nlm.nih.gov/sites/GeneTests/lab/clinical_disease_id/2775?db=genetests&country=United%20States

Clinical Services

<http://www.genetests.org>

Find Genetic Services

<https://clinics.acmg/net>

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