

## Newborn Screening ACT Sheet

# Gaucher Disease

**Condition Description:** Gaucher disease is a lysosomal storage disorder (LSD) caused by a defect in beta glucocerebrosidase, resulting in progressive accumulation of cerebrosides throughout the body. There is wide variability in severity and age of onset. Gaucher disease is an autosomal recessive disorder.

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### ***YOU SHOULD TAKE THE FOLLOWING ACTIONS:***

- Consult with genetic metabolic specialist.
- Contact family to inform them of the newborn screening result.
- Evaluate the newborn with attention to neurologic examination and to the presence of hepatosplenomegaly.
- Provide the family with basic information about Gaucher disease.
- Report confirmatory findings to newborn screening program.

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**Diagnostic Evaluation:** Confirmatory beta-glucocerebrosidase enzyme assay. Patients with low activity should have *GBA* gene analysis. The identification of specific mutations may be valuable in predicting the potential for neurologic abnormality.

**Clinical Considerations:** There are three major clinical subtypes, I, II and III, as well as two other subtypes, a cardiovascular and a perinatal lethal. Most patients with Gaucher disease have type I, which affects the spleen, liver and bone, but is not associated with abnormal neurologic findings. Effective treatment, including enzyme replacement therapy (ERT), is available for this form of the disorder. ERT is highly complicated and should be given only under the guidance of a specialist with expertise in lysosomal storage disorders. The rarer types II and III are associated with neurologic degeneration which may begin in the first year of life (type II) or later in childhood (type III). Gaucher types II and III have not shown improvement with ERT.

### **Additional Information:**

[Genetics Home Reference](#)

### **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.

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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/gaucher-disease>

Referral (local, state, regional and national):

Testing

[http://www.ncbi.nlm.nih.gov/sites/GeneTests/lab/clinical\\_disease\\_id/2232?db=genetests&country=United%20States](http://www.ncbi.nlm.nih.gov/sites/GeneTests/lab/clinical_disease_id/2232?db=genetests&country=United%20States)

Clinical Services

<http://www.genetests.org>

Find Genetic Services

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

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