

Newborn Screening ACT Sheet

[Decreased β -glucocerebrosidase]

Gaucher Disease

Differential Diagnosis: None.

Condition Description: Gaucher disease is a lysosomal disorder caused by a deficiency of β -glucocerebrosidase, resulting in the progressive accumulation of cerebroside. There are several different forms of Gaucher disease, with a wide variability in severity and age of onset.

You Should Take the Following Actions:

- Inform family of the newborn screening result.
 - Ascertain clinical status (seizures, unusual eye movements).
 - Consult with the pediatric metabolic specialist.
 - Evaluate the newborn (hepatosplenomegaly, anemia, bruises).
 - Initiate confirmatory/diagnostic testing and management, as recommended by the specialist.
 - Provide the family with basic information about Gaucher disease and its management.
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- Report final diagnostic outcome to newborn screening program.

Diagnostic Evaluation: [Leukocyte \$\beta\$ -glucocerebrosidase activity](#): Decreased enzyme activity is suggestive of Gaucher disease. [Plasma or DBS glucopsychosine \(glucosylsphingosine, lyso-GL1\)](#): typically is elevated. [Molecular genetic testing](#): can confirm the diagnosis and may aid in predicting the potential for neurologic involvement.

Clinical Considerations: Types I, II, and III are the three major subtypes of Gaucher disease; there is also a cardiovascular and a perinatal lethal form. Most patients with Gaucher disease have type I, which affects the spleen, liver and bone, with symptoms developing between childhood and adulthood. Effective treatments, including enzyme replacement therapy, are available for this form of the disorder. Types II and III are associated with neurologic deterioration which may begin within the first year of life (type II) or later in childhood (type III). These forms of Gaucher disease have not shown improvement with enzyme replacement therapy. Treatment decisions for Gaucher disease should be made under the guidance of a specialist.

Additional Information:

[How to Communicate Newborn Screening Results](#)

[Gene Reviews](#)

[Medline Plus](#)

[Condition Information for Families- HRSA Newborn Screening Clearinghouse](#)

[ClinGen Actionability Report](#)

Referral (local, state, regional, and national):

[Find a Genetics Clinic Directory](#)

[Genetic Testing Registry](#)

Local Resources (Insert Local Website Links)
State Resource Site (Insert Website Information)

Name	
URL	
Comments	

Local Resource Site (Insert Website Information)

Name	
URL	
Comments	

Appendix (Resources with Full URL Addresses)

Additional Information

How to Communicate Newborn Screening Results

- <https://www.hrsa.gov/sites/default/files/hrsa/advisory-committees/heritable-disorders/Resources/achdnc-communication-guide-newborn.pdf>

Gene Reviews

- <https://www.ncbi.nlm.nih.gov/books/NBK1269/>

Medline Plus

- <https://medlineplus.gov/genetics/condition/gaucher-disease/>

Condition Information for Families-HRSA Newborn Screening Clearinghouse

- <https://newbornscreening.hrsa.gov/conditions/gaucher-disease>

ClinGen Actionability Report

- <https://actionability.clinicalgenome.org/ac/Adult/ui/stg2SummaryRpt?doc=AC104>

Referral (local, state, regional and national)

Find a Genetics Clinic Directory

- <https://clinics.acmg.net>

Genetic Testing Registry

- <https://www.ncbi.nlm.nih.gov/gtr/>

This practice resource is designed primarily as an educational resource for medical geneticists and other clinicians to help them provide quality medical services. Adherence to this practice resource is completely voluntary and does not necessarily assure a successful medical outcome. This practice resource 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. 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 practice resource. Clinicians also are advised to take notice of the date this practice resource was adopted, and to consider other medical and scientific information that becomes available after that date. It also would be prudent to consider whether intellectual property interests may restrict the performance of certain tests and other procedures.