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

ACT Sheet

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 cerebrosides. 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.

- Report final diagnostic outcome to newborn screening program.

Diagnostic Evaluation: Leukocyte β-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

 (Funded in part through MCHB/HRSA/HHS grant #U22MC03957;#UH9MC30770 National Coordinating Center for the Regional Genetics Networks*)*

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/

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