MAYO CLINIC LABORATORIES

Newborn Screening Act Sheet

Gaucher Disease: Decreased Acid Beta-Glucosidase

Differential Diagnosis: Gaucher disease type I (non-neuronopathic), Gaucher disease type II (acute neuropathic/infantile), Gaucher disease type III (subacute neuropathic/juvenile), perinatal lethal, cardiovascular form

Condition Description: Gaucher disease is a lysosomal storage disorder (LSD) caused by a defect in acid beta-glucosidase (glucocerebrosidase: GBA). GBA deficiency results in cellular accumulation of glucocerebroside and glucopsychosine, resulting in organ dysfunction and organomegaly. There are 3 types and 2 subtypes of Gaucher disease, with variability in clinical presentation. Gaucher disease is an autosomal recessive disorder.

You should take the following actions:

- Contact family to inform them of the newborn screening result and ascertain clinical status.
- Consult with genetic or metabolic specialist.
- Evaluate the newborn. Infants with type I Gaucher disease are typically asymptomatic. The presence of neurologic signs (such as opisthotonus, spasticity, seizures or abnormal eye movements), hepatosplenomegaly or pancytopenia may be indicative of type II or III Gaucher.
- Initiate timely confirmatory/diagnostic testing and management, as recommended by specialist.
- Provide family with basic information about Gaucher disease.

Diagnostic Evaluation: Confirmatory beta-glucosidase enzyme assay and measurement of glucopsychosine. When patients have low enzyme activity, *GBA* gene analysis and other laboratory studies may be required in consultation with the pediatric metabolic specialist.

Clinical Expectations: The clinical presentation of Gaucher disease varies depending on the type. The most common form is type I and typically presents anytime between childhood and adulthood with hepatosplenomegaly, anemia, and thrombocytopenia. Types II and III have a primary neurologic component with an earlier age of onset (newborn period-2 years of age), with type II being uniformly lethal. There is limited genotype-phenotype correlation and beta-glucosidase levels cannot reliably predict disease type or heterozygosity. Glucopsychosine elevation supports the diagnosis of Gaucher disease. Gaucher disease is caused by mutations in the *GBA* gene and has an estimated incidence of approximately 1 in 50,000 live births. Enzyme replacement therapy (ERT) and substrate reduction therapy (SRT) are available treatment options for some of the types. Administration of ERT and SRT are highly complicated and should only be given under the guidance of a specialist with expertise in LSD.

Additional Information

Genetics Home Reference

Genetic Testing Registry

Baby's First Test

Mayo Clinic Laboratories Testing

GPSYW / Glucopsychosine, Blood

GBAW / Beta-Glucosidase, Leukocytes

GBAZ / Gaucher Disease, Full Gene Analysis, Varies