Newborn Screen Follow-up for Gaucher Disease

Decreased beta-glucosidase* (GBA) with or without abnormal 2nd-tier testing

- BGL / Beta-Glucosidase, Leukocytes
- GPSY / Glucopsychosine, Blood Spot

GBA activity – decreased
Glucopsychosine – elevated
Gaucher disease confirmed

Optional
Referral to Genetics Specialist

GBA activity – decreased
Glucopsychosine – normal
Consider

GBA activity – normal
Glucopsychosine – normal
Gaucher disease confirmed

Optional
GBAZ / Gaucher Disease, Full Gene Analysis**
If the patient is of Ashkenazi Jewish descent, consider targeted mutation analysis (GAUP / Gaucher Disease, Mutation Analysis, GBA)

None or 1 mutation identified***
Not Gaucher disease

GBA=beta-glucosidase

*Beta-glucosidase is also known as glucocerebrosidase
**GBAZ deletion/duplication testing should be considered if sequencing is not informative
***Consult with Genetics Specialist if clinical suspicion for Gaucher disease is high