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

Referral to Genetics Specialist

GBA activity – decreased

Glucopsychosine – normal

Consider

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

Gaucher disease confirmed

None or 1 mutation identified***

Not Gaucher disease

Optional

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