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

If GBA activity is decreased and Glucopsychosine is elevated:
- Gaucher disease confirmed
- Referral to Genetics Specialist

If GBA activity is decreased and Glucopsychosine is 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)

If GBA activity is normal and Glucopsychosine is normal:
- Not Gaucher disease

GBA activity – decreased
Glucopsychosine – normal

Optional

Consider Gaucher disease

None or 1 mutation identified***

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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.