Newborn screen result: decreased beta-glucosidase* (GBA) with or without abnormal 2nd-tier testing

- **GBAW / Beta-Glucosidase, Leukocytes**
- **GPSY / Glucopsychosine, Blood Spot**
- **GPSYW / Glucopsychosine, Blood**

**GBA activity – decreased**
- **Glucopsychosine – elevated**
  - Gaucher disease confirmed
  - Referral to Genetics Specialist

**GBA activity – decreased**
- **Glucopsychosine – normal**
  - Consider
  - **GBAZ / Gaucher Disease, Full Gene Analysis, Varies**
    - If the patient is of Ashkenazi Jewish descent, consider targeted mutation analysis (GAUP / Gaucher Disease, Mutation Analysis, GBA, Varies)
    - Gaucher disease confirmed
    - None or 1 mutation identified

**GBA activity – normal**
- **Glucopsychosine – normal**
  - Not Gaucher disease

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*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