Newborn Screen Follow-up for Gaucher Disease

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***

Not Gaucher disease

Optional

*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