Newborn screen result: decreased acid alpha-glucosidase (GAA)

Assess clinically (including cardiac)
Perform:
- Creatine kinase
- Lactate dehydrogenase
- Aspartate aminotransferase
- Alanine aminotransferase

Order:
- PD2T / Pompe Disease Second-Tier Newborn Screening, Blood Spot - if patient is >6 weeks old, order PDBS / Pompe Disease, Blood Spot
- HEX4 / Glucotetrasaccharides, Random, Urine

■ PD2T/PDBS positive
■ HEX4 ± elevated
■ Evidence of cardiomyopathy

Pompe disease

GAAZ / Pompe Disease, Full Gene Analysis, Varies
Consider GAA deletion/duplication analysis if sequencing is uninformative (CGPH: GeneList ID: IEMCP-N3S8CX)

Genotype consistent with Pompe disease
Referral to Genetics Specialist

Testing negative or consistent with carrier status
If genotype is not informative of cross-reactive immunologic material (CRIM) status, consider PDCRW / Pompe Disease Cross-Reactive Immunological Material Status, Leukocytes

■ PD2T negative
■ HEX4 normal

Infantile/Early Onset Pompe disease unlikely, however Late Onset cannot be ruled out.
Consider

Infantile/Early Onset Pompe disease unlikely
Pompe disease unlikely

Referral to Genetics Specialist