Newborn Screen Follow-up for Pompe Disease

GAA=acid alpha-glucosidase
Glc4=glucose tetrasaccharide
CRIM=Cross-reactive immunologic material

* GAA deletion/duplication testing should be considered if sequencing is not informative
** Refer to Genetics Specialist if clinical suspicion is high

Decreased acid alpha-glucosidase and abnormal 2nd-tier test abnormal

- GAABS / Acid Alpha-Glucosidase, Blood Spot
- HEX4 / Glucotetrasaccharides, Urine

Assess clinically (including cardiac)
Routine labs: creatine kinase, lactate dehydrogenase, aspartate aminotransferase, alanine aminotransferase

GAA activity – deficient
Elevated urine glucotetrasaccharides
Evidence of cardiomyopathy and myopathy

Pompe disease confirmed

Referral to Genetics Specialist

GAA activity – deficient
Normal glucotetrasaccharides
No evidence of cardiomyopathy or myopathy

GAAZ / Pompe Disease, Full Gene Analysis*

Testing negative or consistent with carrier status

Not Pompe disease**

GAA activity – normal
Normal glucotetrasaccharides

Genotype consistent with Pompe disease

If genotype is not informative of CRIM status

CRIM western blot