Newborn Screen Follow-up for Pompe Disease

- Decreased acid alpha-glucosidase and abnormal 2nd-tier test abnormal
  - GAABS / Acid Alpha-Glucosidase, Blood Spot
  - HEX4 / Glucotetrasaccharides, Urine

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

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

- GAA activity – normal
  - Normal glucotetrasaccharides

- Pompe disease confirmed

- Referral to Genetics Specialist

- GAAZ / Pompe Disease, Full Gene Analysis

- Testing negative or consistent with carrier status

- Not Pompe disease

- CRIM western blot

- Genotype consistent with Pompe disease

- If genotype is not informative of CRIM status

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

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