Fabry Disease: Newborn Screen-Positive Follow-up

- Decreased alpha-galactosidase enzyme on newborn screen
  - MALE
  - FEMALE

AGAW / Alpha-Galactosidase, Leukocytes
OR
AGAS / Alpha-Galactosidase, Serum

- NORMAL
- LOW

- False-positive newborn screen
- FABRZ / Fabry Disease, Full Gene Analysis, Varies

- No disease-causing mutation identified
  - MALE
  - FEMALE

- Reduced enzyme result suggests Fabry disease
- Suggests the presence of a mutation that is undetectable by this method

- Disease-causing mutation identified
  - Refer for genetic counseling and comprehensive family history discussion.

- Pseudodeficiency allele identified
  - False-positive newborn screen

- Fabry disease unlikely
- Presence of a mutation that is undetectable by this method is not ruled out.

- Consider performing FMTT / Familial Mutation, Targeted Testing, Varies for at-risk family members