Fabry Disease: Newborn Screen-Positive Follow-up

- Decreased alpha-galactosidase enzyme on newborn screen

**MALE**
- AGA / Alpha-Galactosidase, Leukocytes
- OR
- AGAS / Alpha-Galactosidase, Serum

- NORMAL
  - False-positive newborn screen
  - No disease-causing mutation identified
  - **MALE**
    - Reduced enzyme result suggests Fabry disease
    - Suggests the presence of a mutation that is undetectable by this method

- LOW
  - **MALE**
    - False-positive newborn screen
  - **FEMALE**
    - Disease-causing mutation identified
    - Refer for genetic counseling and comprehensive family history discussion.

**FEMALE**
- FABRZ / Fabry Disease, Full Gene Analysis

- Pseudodeficiency allele identified
  - False-positive newborn screen

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

- **FEMALE**
  - No disease-causing mutation identified
  - **MALE**
    - Reduced enzyme result suggests Fabry disease
    - Suggests the presence of a mutation that is undetectable by this method
  - **FEMALE**
    - Fabry disease unlikely
    - Presence of a mutation that is undetectable by this method is not ruled out.

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