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

  **LOW**

  FABRZ / Fabry Disease, Full Gene Analysis

  **MALE**

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

  **FEMALE**

  No disease-causing mutation identified

  **NORMAL**

  Disease-causing mutation identified

  Refer for genetic counseling and comprehensive family history discussion.

  **LOW**

  Pseudodeficiency allele identified

  **FAMILY**

  False-positive newborn screen

  **MALE**

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

  **FEMALE**

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

  **NORMAL**

  **LOW**

  **FAMILY**

  **FEMALE**

  **NORMAL**

  **LOW**

  **FAMILY**

  **FEMALE**