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

**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.

Disease-causing mutation identified

Refer for genetic counseling and comprehensive family history discussion.

Pseudodeficiency allele identified

False-positive newborn screen

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

Normal

FEMALE

False-positive newborn screen

MALE

No disease-causing mutation identified

Reduced enzyme result suggests Fabry disease

Suggests the presence of a mutation that is undetectable by this method

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