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

Disease-causing mutation identified

Pseudodeficiency allele identified

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

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

Refer for genetic counseling and comprehensive family history discussion.

False-positive newborn screen

NORMAL

LOW

FEMALE

MAYO CLINIC LABORATORIES

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