Clinical suspicion or family history of Fabry disease

FMTT / Familial Mutation, Targeted Testing

YES

Positive family history with mutation identified

Females

Analysis of alpha-galactosidase enzyme activity is not sensitive for carrier detection in all females. It is recommended that molecular testing be performed for diagnosis in females.

Decreased alpha-galactosidase enzyme activity

FABRZ / Fabry Disease, Full Gene Analysis, Varies

Variant of unknown significance identified

No variant or pseudodeficiency allele identified

NO

Males

Recommended initial evaluation (select 1)
- AGAW / Alpha-Galactosidase, Leukocytes and/or
- AGAS / Alpha-Galactosidase, Serum

Normal alpha-galactosidase enzyme activity

Fabry disease highly unlikely (<1% of affected males have leukocyte enzyme values within the normal range)

NO

YES

Disease-causing variant identified

Fabry disease confirmed

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

If not already performed, consider:
- LGB3S / Globotriaosylsphingosine, Serum
- CTSA / Ceramide Trihexosides and Sulfatides, Urine

Females

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Decreased alpha-galactosidase enzyme activity

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