Clinical suspicion or family history of Fabry disease

↓

Positive family history with mutation identified

— YES → **FMTT / Familial Mutation, Targeted Testing, Varies**

↓ NO

Males | **Females**

Males:

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

↓

Normal alpha-galactosidase enzyme activity | Decreased alpha-galactosidase enzyme activity

↓ (Normal)

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

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.

↓

CGPH / Custom Gene Panel, Hereditary, Next-Generation Sequencing, Varies (*GLA* Gene List ID: IEMCP-SRU6J3)

Disease-causing variant identified | Variant of unknown significance identified | No variant or pseudodeficiency allele identified

↓ (Disease-causing variant identified)

Fabry disease confirmed

↓

Consider: **FMTT** for at-risk family members

↓ (Variant of unknown significance identified)

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

↓ (No variant or pseudodeficiency allele identified)

Fabry disease unlikely. Results must be interpreted in the context of an individual's clinical and/or biochemical profile.

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