



Discover Fabry Disease: THE ROLE OF THE CARDIOLOGIST

FABRY DISEASE CAN RESULT IN SUBSTANTIAL HEART DAMAGE¹

Fabry disease is a rare, progressive, and potentially life-threatening disorder that affects men and women.¹⁻³

As an X-linked lysosomal storage disorder that is multisystemic, Fabry disease is caused by complete or partial deficiency of the lysosomal enzyme α -GAL A, leading to GL-3 and lyso-GL-3 accumulation that can result in damage to the heart, kidneys, and cerebrovascular system.^{2,3}

EARLY DIAGNOSIS AND MANAGEMENT ARE CRITICAL AS FABRY DISEASE CAN DECREASE LIFE EXPECTANCY DUE TO HEART AND ORGAN DAMAGE^{2,4-6}



ROUTINELY MONITOR YOUR PATIENTS WITH FABRY DISEASE

Despite normal α -GAL A activity, females are not "just carriers." 69% of females manifest symptoms of Fabry disease and often experience life-altering symptoms and organ damage.⁷

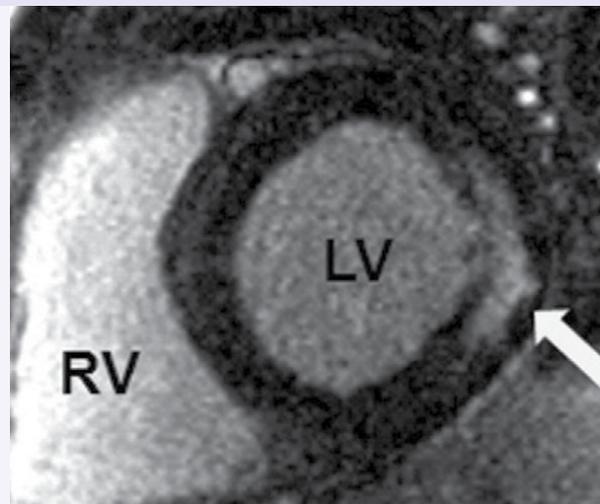
For each patient diagnosed, and average of 5 additional affected family members may be identified through pedigree review.^{9,10*}

Late-onset or non-classic Fabry disease can be just as severe in the heart, even in the absence of multisystem involvement.^{1,11}

IDENTIFY FREQUENT CARDIAC SIGNS AND SYMPTOMS OF FABRY DISEASE

Patients may experience a myriad of symptoms.¹²

- Fabry disease mimics HCM—however, distinct from HCM and other causes of hypertrophy, patients with Fabry disease show low native T1 values, independent of sex^{13,14}
- Progressive LVH, angina, and fibrosis, which lead to increased risk of cardiac arrhythmias^{1,12}
- Early ischemic stroke^{8,12}
- Progressive renal insufficiency¹²
- Peripheral neuropathy of extremities/episodic pain crises¹²
- Hypohidrosis¹²
- Gastrointestinal manifestations¹²
- Angiokeratomas¹²



MRI of typical Fabry fibrosis in the lateral wall of the left ventricle (shown by arrow). Reprinted with permission from Krämer J, et al. *Am J Cardiol*. 2014;114(6):895-900.

α -GAL A, α -galactosidase A; GL-3, globotriaosylceramide; HCM, hypertrophic cardiomyopathy; LV, left ventricle; LVH, left ventricular hypertrophy; lyso-GL-3, globotriaosylsphingosine; MRI, magnetic resonance imaging; RV, right ventricle.

*Pedigree review from the lysosomal storage disease centers and contributing family members found that, on average, there are at least 5 family members who are diagnosed with Fabry disease following the diagnosis of a proband.¹⁰

CARDIAC DISEASE IS THE LEADING CAUSE OF DEATH IN PATIENTS WITH FABRY DISEASE¹

CONSIDER FABRY DISEASE IN YOUR DIFFERENTIAL DIAGNOSIS WHEN¹⁵:

Presentation:

Unexplained HCM and/or LVH, fibrosis in the lateral wall of the left ventricle, low native T1 values, elevated hs-troponin in the absence of acute event^{13,16}

Family history:

Cardiomyopathy, premature stroke, sudden cardiac death, and renal failure^{15,17}

ECG:

Bradycardia, short PR interval (young age), AV block (cardiac clinical manifestations in the 3rd decade of life onward), voltage criteria for LVH, ST segment changes, T-wave inversion^{12,15}

● ● ● IF YOU SEE UNEXPLAINED CARDIOMYOPATHY, CONSIDER FABRY DISEASE.¹⁸ ● ● ●

Cardiologists have the tools to screen for, diagnose, and manage Fabry disease.

TESTING IS STRAIGHTFORWARD^{12,19,20}

Males	 <p>α-GAL A enzyme assay and/or GLA gene sequencing</p>
Females	 <p>GLA gene sequencing Affected females may have normal to low enzyme activity in plasma or leukocytes; therefore, <i>GLA</i> gene sequencing is required to confirm diagnosis in females.</p>

In patients with unexplained HCM, conduct genetic testing with an HCM panel that includes the *GLA* gene.

TREATMENT IS AVAILABLE FOR YOUR PATIENTS WITH FABRY DISEASE

Stabilizing disease progression is an important goal.¹²

Damage to cardiac, renal, and cerebrovascular systems can have life-threatening impact. Monitor patients in accordance with published management guidelines and clinicians' medical expertise.¹²

Laboratories across the United States offer diagnostic tests for Fabry disease. Visit www.discoverfabry.com/hcp.



α-GAL A, α-galactosidase A; AV, atrioventricular; CNS, central nervous system; ECG, electrocardiogram; *GLA*, galactosidase-alfa; HCM, hypertrophic cardiomyopathy; hs, high-sensitivity; LVH, left ventricular hypertrophy.

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