

CARDIAC AND EXTRA-CARDIAC RED FLAGS*

- The Fabry disease 'red flags' listed in both tables below identify what to look out for when diagnosing patients with idiopathic left ventricular hypertrophy (LVH) and/or HCM

Cardiac		
Diagnostic tool	History	Family history of LVH, particularly no evidence of male-to-male transmission
	Electrocardiography	Short PQ interval [†]
		Bradycardia
		Chronotropic incompetence
		Atrioventricular blocks [†]
	2D-echocardiography	LVH with normal systolic function
		Reduced global longitudinal strain
		Mild-to-moderate aortic root dilation
		Mitral and aortic valve thickening with mild-to-moderate regurgitation
	Cardiac magnetic resonance	Hypertrophy of papillary muscles
		Mid-layer posterolateral late gadolinium enhancement
		Low native T1

Extra-cardiac		Presenting decades of age
Family history of renal failure and/or stroke	Any time	
Neuropathic pain	1-2	
Gastrointestinal symptoms		
Angiokeratomas		
Cornea verticillata*		
Hypohidrosis, heat/cold and exercise intolerance		
Albuminuria	3-4	
Juvenile and/or cryptogenic TIA/stroke		
Hearing loss (either progressive or sudden)		
Dolichoectasia of the basilar artery, chronic white matter hyperintensities at brain MRI		
Proteinuria		
Renal failure		
Lymphoedema		

*In the absence of iatrogenic causes (chloroquine/amiodarone). †Short PQ interval in early stages; atrioventricular and bundle branch blocks are more common in advanced disease. 2D-echo = 2-dimensional echocardiography; MRI = magnetic resonance imaging; TIA = transient ischemic attack.