

CHECKLIST: FABRY DISEASE?

FOCUS ON CARDIOLOGY

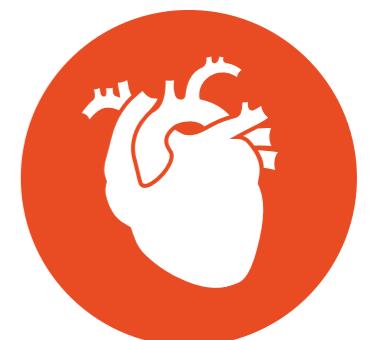
COULD IT BE FABRY DISEASE? Focus on Cardiology

Symptoms/Findings	Diagnostics	Patient's Medical History	Fabry Diagnosis
<p><input checked="" type="checkbox"/> Cardiomyopathy/ left ventricular hypertrophy (LVH) (without/mild hypertension)</p> <p><input type="checkbox"/> Prominent papillary muscle</p> <p><input checked="" type="checkbox"/> Cardiac arrhythmia (paroxysmal or permanent atrial fibrillation, bradycardia, malignant arrhythmia/ ventricular tachycardia)</p> <p><input type="checkbox"/> Myocardial infarction</p> <p><input type="checkbox"/> Congestive heart failure</p> <p><input type="checkbox"/> Intramyocardial fibrosis</p> <p><input type="checkbox"/> Valvular disorder (mitral valve, aortic valve)</p> <p><input type="checkbox"/> Dyspnoea</p>	<p><input type="checkbox"/> ECG (changes with short P wave and short PR interval, increase in the QRS width and repolarisation disturbances)</p> <p><input type="checkbox"/> Stress ECG</p> <p><input type="checkbox"/> Spiroergometry</p> <p><input type="checkbox"/> Holter ECG (24 hours)</p> <p><input checked="" type="checkbox"/> Echocardiography: LVH (end diastolic wall thickness >12 mm), prominent papillary muscle, myocardial replacement fibrosis</p> <p><input type="checkbox"/> Blood: hs-troponin, NT-proBNP</p>	<p>Fabry disease possible →</p> <p>Any signs of kidney failure such as uraemia or hypocalcaemia?</p> <p>Family Medical History</p> <p>Any signs of kidney failure such as uraemia or hypocalcaemia in relatives? Unexplained incidents of death in the family?</p>	<p>Fabry disease probable →</p> <p>Dry blood test determining:</p> <ul style="list-style-type: none"> • The genetic mutation • The level of α-galactosidase A (α-Gal A) activity • The biomarker lyso-Gb₃ <p>and/or</p> <p>Molecular genetic/ cytogenetic Fabry diagnosis from:</p> <ul style="list-style-type: none"> • EDTA blood test • Serum • Buccal swab

ECG, electrocardiogram; EDTA, ethylenediaminetetraacetic acid; hs, high sensitivity; lyso-Gb₃, globotriaosylsphingosine; NT-proBNP, N-terminal pro-brain natriuretic peptide.

Engagement of an expert centre for conclusive diagnosis.
In the event of confirmed Fabry disease diagnosis, further monitoring of progress and organ screening is recommended.

Note: this listing of symptoms/ findings and diagnostic tests may not be all-inclusive.



SIGNS AND SYMPTOMS OF FABRY DISEASE ACROSS ORGAN SYSTEMS

Kidneys	Eyes	Skin	Nervous System	Differential Diagnosis
<ul style="list-style-type: none"> • (Micro-) albuminuria • Proteinuria (>300 mg/day) • GFR decrease (GFR <60 mL/min/1.73 m²) • Oedemas in the hands and feet → Note relationship between leg oedema and protein excretion • Parapelvic renal cysts • Progressive renal failure • Dialysis • Rare: renal tubular acidosis and nephrogenic diabetes insipidus 	<ul style="list-style-type: none"> • Cornea verticillata • Fabry cataract • Corneal opacity • Vascular anomalies of the conjunctiva • Vascular tortuosity of the retina • Optic neuropathy with visual field loss 	<ul style="list-style-type: none"> • Angiokeratomas • Dyshidrosis • Telangiectasias <p>Quality of Life</p> <ul style="list-style-type: none"> • Reduced physical capacity • Psychological problems • Fatigue • Pain <p>Ears</p> <ul style="list-style-type: none"> • Hearing loss (often high frequencies) • Tinnitus • Dizziness 	<ul style="list-style-type: none"> • TIA, stroke • Acroparaesthesia → Burning pain in hands and feet • Small fibre neuropathy • Pain crises • Dyshidrosis • Temperature intolerance • Gastrointestinal discomfort • Basilar ectasia/ectatic vessels (vertebrobasilar) • White matter lesions • Depression/fatigue • Headaches • Reduced lacrimation and salivation 	<ul style="list-style-type: none"> • Atherosclerosis • Hyperthyroidism • Mitochondriopathy • Long QT syndrome • Myocarditis • Systemic lupus erythematosus • Rheumatoid arthritis • Dermatomyositis • Endocarditis • Diabetes mellitus • Arterial hypertension • Sjögren's syndrome • Cardiac amyloidosis • Cardiomyopathy in Friedreich's ataxia • Obstructive sleep apnea

GFR, glomerular filtration rate; TIA, transient ischaemic attack.

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