

**CHECKLIST: FABRY DISEASE?**

# **FOCUS ON NEPHROLOGY**

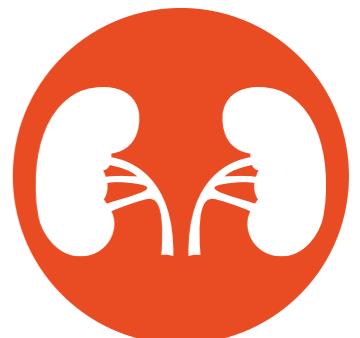
# COULD IT BE FABRY DISEASE? Focus on Nephrology

Symptoms/Findings	Diagnostics	Patient's Medical History	Fabry Diagnosis
<p><input type="checkbox"/> Microalbuminuria/  albuminuria</p> <p><input type="checkbox"/> Podocyturia  → Proteinuria  (&gt;300 mg/day)</p> <p><input type="checkbox"/> GFR decrease  (GFR &lt;60 mL/  min/1.73 m<sup>2</sup>)</p> <p><input type="checkbox"/> Oedemas in the  hands and feet  → Note relationship  between leg oedema  and protein excretion</p> <p><input type="checkbox"/> Parapelvic  renal cysts</p> <p><input type="checkbox"/> Progressive  renal failure</p> <p><input type="checkbox"/> Dialysis</p> <p><input type="checkbox"/> Rare: renal tubular  acidosis and  nephrogenic diabetes  insipidus</p>	<p><input type="checkbox"/> Renal function  test: GFR from  creatinine and urea  clearance, serum  creatinine (mg/dL),  GFR (mL/min/1.73 m<sup>2</sup>)  → Serum analysis,  semi-quantitative  protein or albumin  using test strips,  UPC, UAC  → Spontaneous urine  total protein, albumin,  GFR from creatinine  and urea clearance  → 24-hour urine</p> <p><input type="checkbox"/> Renal ultrasound  (morphology, vascular  lesions, cysts)</p> <p><input type="checkbox"/> 24-hour blood pressure  measurement</p> <p><input type="checkbox"/> Renal biopsy if  necessary (evaluation  of GL-3 deposits,  fibrosis, and sclerosis)  → Discuss light  microscopy using  toluidine blue stain  with Pathology</p>	<p>Any signs of kidney  failure such as uraemia  or hypocalcaemia?</p> <p><b>Family Medical History</b></p> <p>Any signs of kidney  failure such as uraemia  or hypocalcaemia in  relatives?  Unexplained incidents  of death in the family?</p>	<p><b>Fabry  disease  possible</b></p> <p><b>Fabry  disease  probable</b></p> <p>Dry blood test  determining:</p> <ul style="list-style-type: none"> <li>• The genetic  mutation</li> <li>• The level of  <math>\alpha</math>-galactosidase A  (<math>\alpha</math>-Gal A) activity</li> <li>• The biomarker  lyso-Gb<sub>3</sub></li> </ul> <p><b>and/or</b></p> <p>Molecular genetic/  cytogenetic Fabry  diagnosis from:</p> <ul style="list-style-type: none"> <li>• EDTA blood test</li> <li>• Serum</li> <li>• Buccal swab</li> </ul>

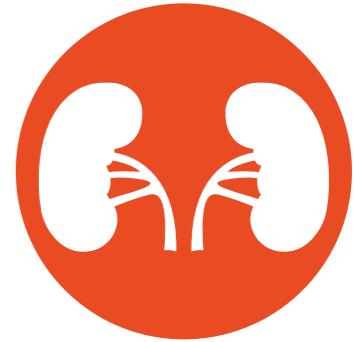
EDTA, ethylenediaminetetraacetic acid; GFR, glomerular filtration rate; GL-3, globotriaosylceramide;  
 lyso-Gb<sub>3</sub>, globotriaosylsphingosine; UAC, urine albumin to creatinine ratio; UPC, urine protein to creatinine ratio.

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

Nervous System	Eyes	Heart	Skin	Differential Diagnosis
<ul style="list-style-type: none"> <li><b>TIA, stroke</b></li> <li><b>Acroparaesthesia</b> → Burning pain in hands and feet</li> <li><b>Small fibre neuropathy</b></li> <li><b>Pain crises</b></li> <li><b>Dyshidrosis</b></li> <li><b>Temperature intolerance</b></li> <li><b>Gastrointestinal discomfort</b></li> <li>Basilar ectasia/ectatic vessels (vertebrobasilar)</li> <li>White matter lesions</li> <li>Depression/fatigue</li> <li>Headaches</li> <li>Reduced lacrimation and salivation</li> </ul>	<ul style="list-style-type: none"> <li><b>Cornea verticillata</b></li> <li><b>Fabry cataract</b></li> <li>Corneal opacity</li> <li>Vascular anomalies of the conjunctiva</li> <li>Vascular tortuosity of the retina</li> <li>Optic neuropathy with visual field loss</li> </ul> <p><b>Ears</b></p> <ul style="list-style-type: none"> <li>Hearing loss (often high frequencies)</li> <li>Tinnitus</li> <li>Dizziness</li> </ul>	<ul style="list-style-type: none"> <li><b>Cardiomyopathy/ left ventricular hypertrophy</b> (without/mild hypertension)</li> <li>Myocardial infarction</li> <li>Congestive heart failure</li> <li>Intramycocardial fibrosis</li> <li>Cardiac arrhythmia</li> <li>Valvular disorder (mitral valve, aortic valve)</li> <li>Dyspnoea</li> </ul>	<ul style="list-style-type: none"> <li><b>Angiokeratomas</b></li> <li><b>Dyshidrosis</b></li> <li>Telangiectasias</li> </ul> <p><b>Quality of Life</b></p> <ul style="list-style-type: none"> <li><b>Reduced physical capacity</b></li> <li>Psychological problems</li> <li>Fatigue</li> <li>Pain</li> </ul>	<ul style="list-style-type: none"> <li>Diabetes mellitus</li> <li>Hypertension</li> <li>Glomerulonephritis</li> <li>Systemic lupus erythematosus</li> <li>Haemolytic-uraemic syndrome (HUS)</li> <li>Gout</li> <li>Amyloidosis</li> <li>Henoch-Schönlein nephritis</li> </ul> 

TIA, transient ischaemic attack.

## References

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