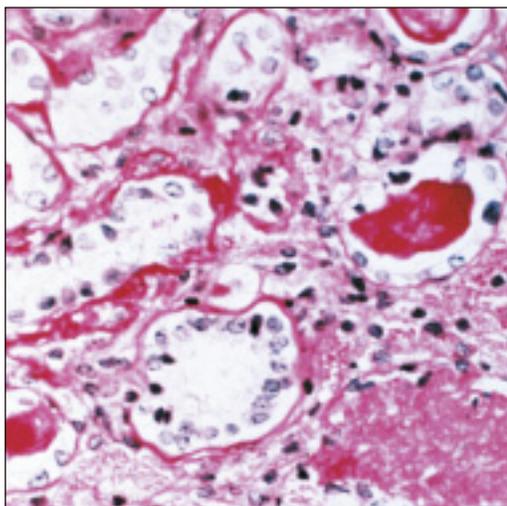
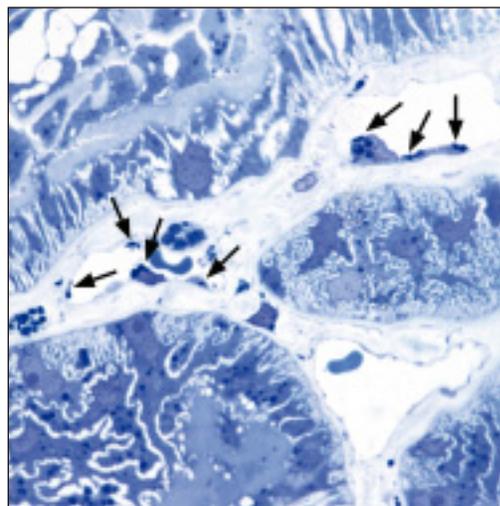


If you see a patient with a family history of chronic kidney disease It could be Fabry disease



Light micrograph of end-stage renal disease in Fabry patient. Note that the glomeruli are atrophic and hyalinized, typical of end-stage disease.



Light micrograph of Fabry renal capillary endothelium. Arrows indicate areas of globotriaosylceramide (GL-3) lipid substrate deposition, which may play a role in renal failure.

Nephrologists have the opportunity to identify patients with this progressive, often life-threatening disease.

In addition to a family history of chronic kidney disease, patients with Fabry disease may present at an early age with:

- Progressive and/or unexplained chronic kidney disease
- Proteinuria/isosthenuria
- Tubular dysfunction (polyuria, polydipsia)
- Symptoms suggesting Fanconi's syndrome
- Elevated serum creatinine

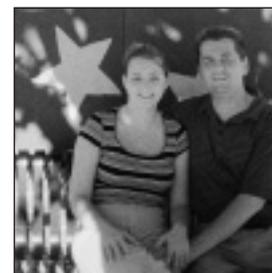
Other manifestations include:

- Premature cardiac disease
- Corneal and lenticular abnormalities (seen through slit lamp—generally does not affect vision)
- Premature stroke
- “Burning” pain in the hands and feet
- Heat/cold and exercise intolerance
- Impaired sweating
- Angiokeratomas (reddish-purple skin lesions that do not blanch with pressure)

While Fabry disease is rare, it may be more common within a Fabry family.

Fabry disease

Progressive. Destructive. Life-threatening.



FABRY DISEASE PROFILE

Fabry disease is an often life-threatening, panethnic, and heterogeneous inherited disorder caused by a lysosomal enzyme (alpha-galactosidase A) deficiency. The resulting progressive accumulation of globotriaosylceramide (GL-3) in the vasculature and other cell types or tissues eventually leads to major organ system damage including renal insufficiency, cardiac disease, and premature stroke.

DISEASE RISK IN FAMILIES

- An X-linked disorder, Fabry disease is carried on the X chromosome.
- Males with the disease pass the defective gene on to all of their daughters and none of their sons.
- Females have a 50% chance with each pregnancy of passing the defective gene to both their sons and daughters.
- Unlike many other X-linked diseases, females can have varying degrees of disease manifestations.

DIAGNOSIS

- Although Fabry disease usually presents in childhood, the disease often goes unrecognized by physicians until adulthood, when the underlying pathology is advanced.
- Delayed diagnosis may be the result of disease under-recognition and/or symptoms being mistaken for those of other disorders, such as rheumatoid or juvenile arthritis, rheumatic fever, erythromelalgia, multiple sclerosis, or lupus.
- Clinical diagnosis is based upon presentation of signs and symptoms.
- Diagnosis is confirmed in males by enzyme assay (blood test) detecting low or absent levels of alpha-galactosidase A (alpha-GAL), or in females by mutation linkage analysis (blood test).

TREATMENT

Treatment is available for Fabry disease. Patients should be referred to a geneticist for testing and further intervention.

LEARN MORE

Visit www.fabrycommunity.com for more information on Fabry disease or call Genzyme Medical Information at 800-745-4447 or 617-768-9000.

Early diagnosis and intervention are key—nephrologists can play a role.

genzyme