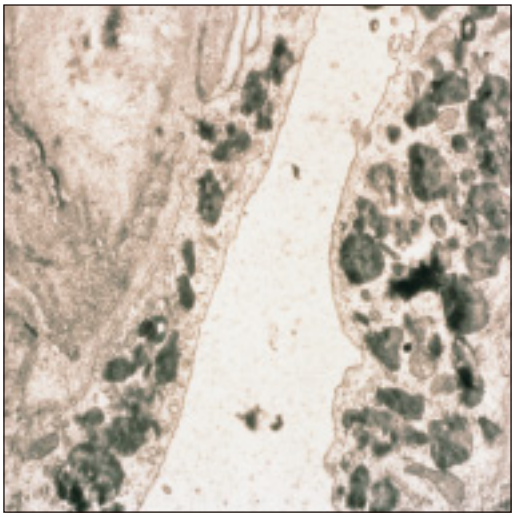
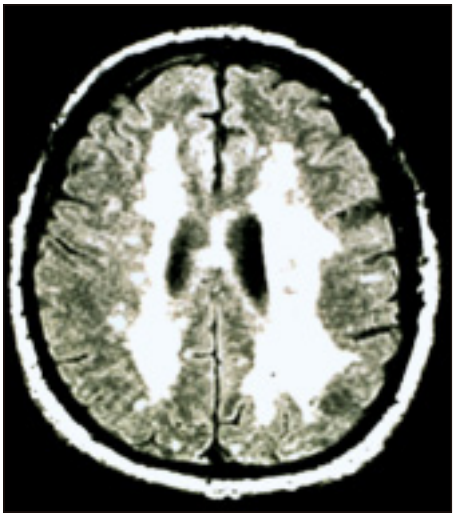
If you see premature stroke in a patient

# It could be Fabry disease



Progressive accumulation of substrate in the vascular endothelium leads to ischemia and infarction of these vessels.



White matter lesions on MRI, demonstrating evidence of cerebrovascular infarct. Image courtesy of Edward M. Kaye, MD.

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

**In addition to premature stroke, patients with Fabry disease may present with:**

- Transient ischemic attacks
- Acroparesthesias ("burning" pain in the hands and feet)
- Hypohidrosis
- Heat/cold and exercise intolerance
- Hearing loss, tinnitus
- Vertigo/dizziness
- Nystagmus

**Other manifestations include:**

- Progressive and/or unexplained chronic kidney disease
- Premature cardiac disease
- Corneal and lenticular abnormalities (seen through slit lamp—generally does not affect vision)
- 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.**

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Information for Neurologists

# 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—neurologists can play a role.**

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