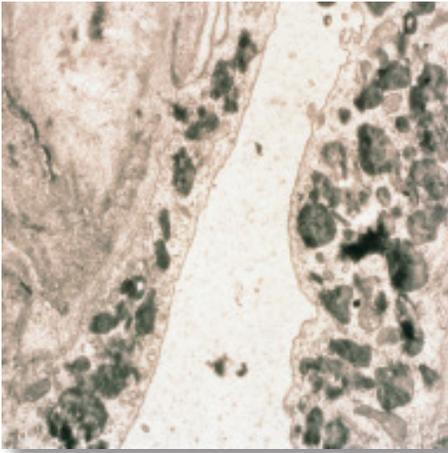
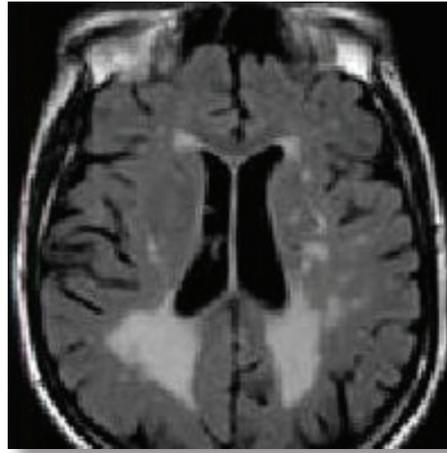


# If you see premature stroke in a patient It could be **FABRY DISEASE**

Information for  
Neurologists



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



Fluid-attenuated inversion recovery (FLAIR)-weighted axial MRI section showing multiple white matter lesions in the cerebral hemispheres

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

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

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

- Transient ischemic attacks
- Neuropathic pain (“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)
- Gastrointestinal problems

## **FABRY DISEASE**

**Silently Progressive.** Increasingly Debilitating. Often Life-Threatening.

## FABRY DISEASE PROFILE

Fabry disease is an inherited disorder that affects men, women, and children of all ethnicities. It is a multisystemic disorder that can result in irreversible, potentially life-threatening disease of the kidney, heart, and brain. The disease is characterized by the progressive cellular accumulation of a lipid substrate called globotriaosylceramide (or GL-3). Ongoing build-up of this substance is caused by deficiency of the lysosomal enzyme alpha galactosidase A (or  $\alpha$ -GAL), which usually metabolizes GL-3 and keeps it from accumulating. Without enough of this essential enzyme, GL-3 accumulates in the lysosomes of most cell types over the course of a lifetime, often causing debilitating symptoms in childhood and adolescence and potentially irreversible tissue damage by adulthood.

### DISEASE RISK IN FAMILIES

- Unlike many other X-linked disorders, females with the mutated gene are affected to varying degrees due to random X inactivation.
- Males with the disease pass the mutated gene on to all of their daughters and none of their sons.
- Females have a 50% chance with each pregnancy of passing the mutated gene to both their sons and daughters.
- If you identify a patient with Fabry disease, family testing should be considered.

### 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.
- Diagnosis is confirmed in males by enzyme assay (blood test) detecting low or absent levels of alpha-galactosidase A ( $\alpha$ -GAL), or in females through genetic testing to detect a mutation.

### LEARN MORE

Visit [www.fabrycommunity.com](http://www.fabrycommunity.com) for more information on Fabry disease or call Sanofi Genzyme Medical Information at 800-745-4447, option 2.

*Neurologists are in a unique position to identify patients and families at risk for Fabry disease.*

**SANOFI GENZYME** 