If you see a patient with corneal opacities
It could be FABRY DISEASE

Panels A and B: Distinctive corneal opacity (corneal verticillata or vortex keratopathy) in Fabry disease. Note the whorl-like corneal rays emanating from a single vertex like the spokes of a wheel. Panel B image used with permission, RL Abbott, MD.

In addition to corneal opacities, patients with Fabry disease may present with:

- Posterior capsular cataracts with whitish spokelike deposits of granular material (Fabry cataract)
- Aneurysmal dilatation of thin-walled venules on the bulbar conjunctiva
- Mild-to-marked tortuosity and angulation of the retinal vessels

Other manifestations include:

- Progressive and/or unexplained chronic kidney disease
- Premature cardiac disease and/or 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)
- Gastrointestinal problems

FABRY DISEASE
**FABRY DISEASE PROFILE**

Fabry disease is an inherited disorder that affects men, women, and children of all ethnicities. It is a multisystemic disorder that ultimately results in irreversible, potentially life-threatening disease of the kidney, heart, and brain. The disease is characterized by the progressive and unrelenting 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 α-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 defective gene are affected to varying degrees due to random X inactivation.
- 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.
- 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 (α-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.

Early diagnosis and intervention are key.
Eye care professionals can play a role.