

WHEN YOU SEE CORNEAL WHORLING, REFER TO A GENETICIST

You Can Play a Key Role in Diagnosing a Potentially Life-threatening Genetic Disorder

- Corneal verticillata is seen in approximately **80%** of patients with Fabry disease¹
- Fabry Disease is a progressive, often life-threatening genetic disorder that can result in renal failure, heart disease, and stroke
- Diagnosis can be delayed by an average of 15 years from symptom onset²
- A routine slit lamp exam can reveal corneal verticillata, one of the early signs of the disease
- Eye care professionals can be the first to recognize Fabry disease
- Earlier diagnosis can help patients receive disease management sooner

If you suspect Fabry disease, refer to a geneticist and help reduce diagnostic delays.

For more information

Contact Sanofi Genzyme Medical Information at 800-745-4447, option 2
www.fabrycommunity.com



Corneal verticillata is seen almost universally in patients with Fabry disease. Note the rays emanating from a single vortex. A number of medications, such as amiodarone or chloroquine, can also cause this phenomenon, and should be ruled out.³

- Undiagnosed and unmanaged, Fabry disease reduces life expectancy by 15-20 years, according to natural history data^{4,5}
- Early signs may include pain in the hands and feet, impaired sweating, heat/cold intolerance, and angiokeratomas
- For every index patient diagnosed, an average of 5 additional affected family members may be identified⁶



*After an index patient was identified during a routine slit lamp exam, 41 of 99 members of this extended family were diagnosed with Fabry disease.**

References

- ¹ Samiy N. *Surv Ophthalmol* 2008;53:416-423. ² Wilcox WR et al. *Mol Genet Metab*. 2008;93(2):112-128. ³ Mehta A. *Q J Med* 2002;95:647-653. ⁴ MacDermot KD et al. *J Med Genet*. 2001;38(11):750-760. ⁵ MacDermot KD et al. *J Med Genet*. 2001;38(11):769-775. ⁶ Laney D et al. *J Genet Couns* 2008;17:79-83.

*Prevalence of the disease may vary from family to family.

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