

## Fabry Disease

Category(ies): [Cornea](#)

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Fabry disease is an X-linked dominant sphingolipidosis caused by a deficiency in alpha-galactosidase A which results in accumulation of ceramide trihexoside in select body tissues. Ocular findings include cornea verticillata, conjunctival aneurysms, cataracts, retinal vascular changes and optic atrophy.

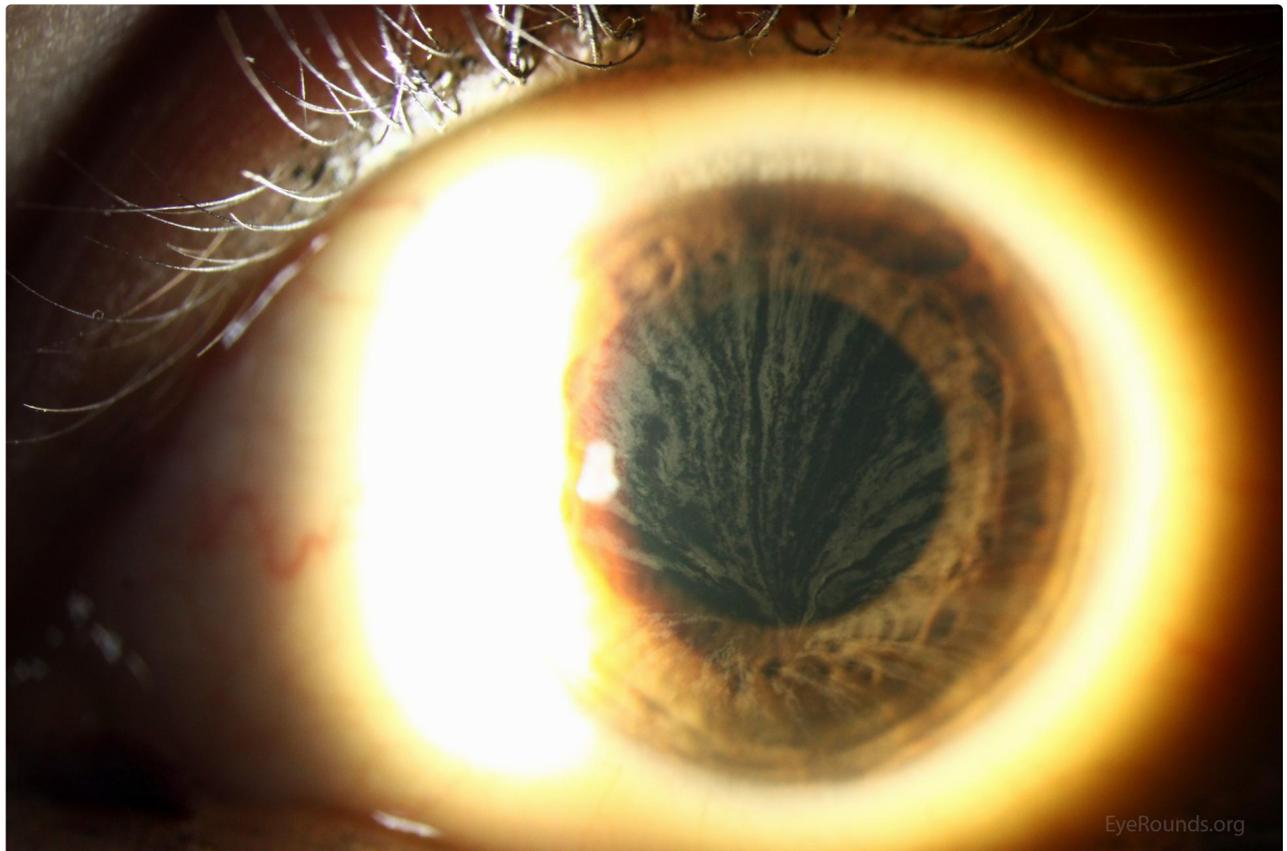


Figure 1: Cornea verticillata in a young female which led to the diagnosis of Fabry disease.

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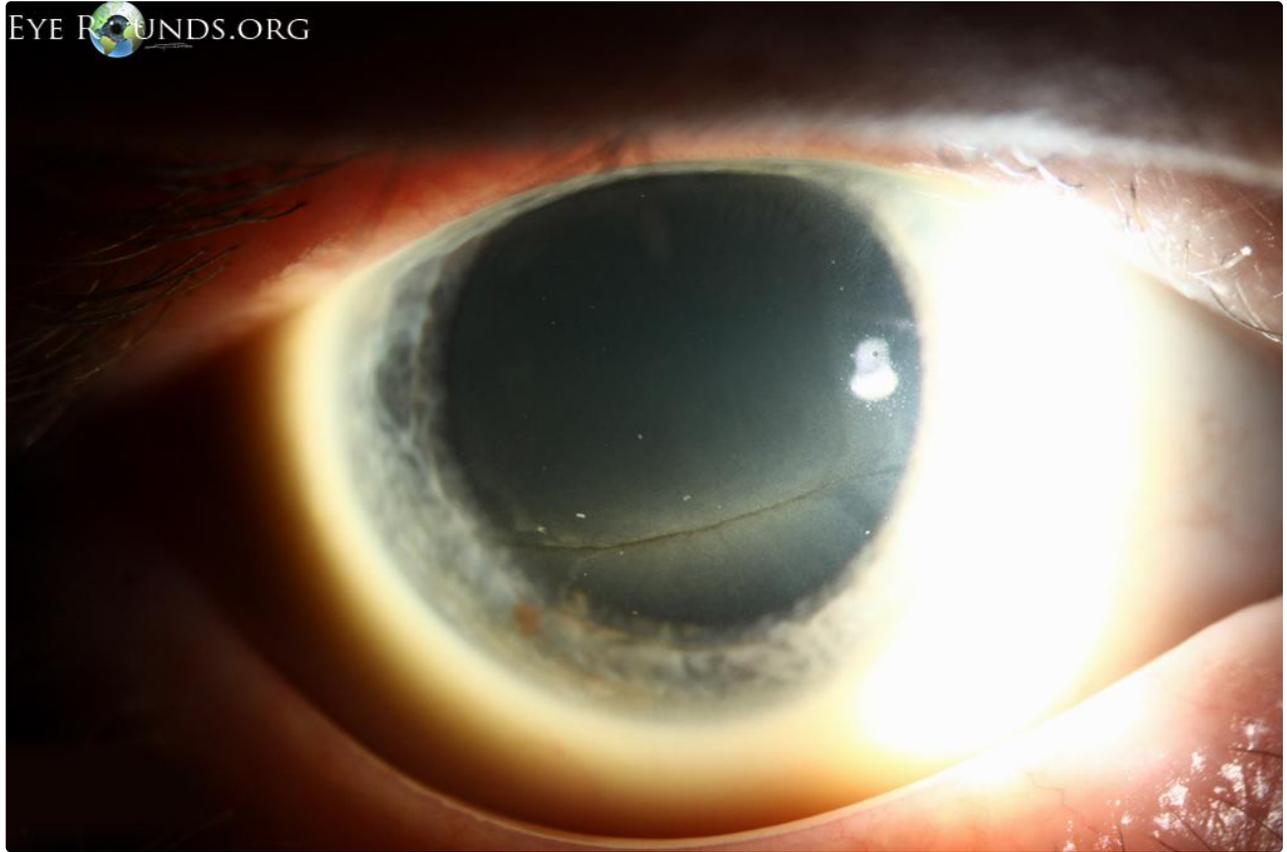
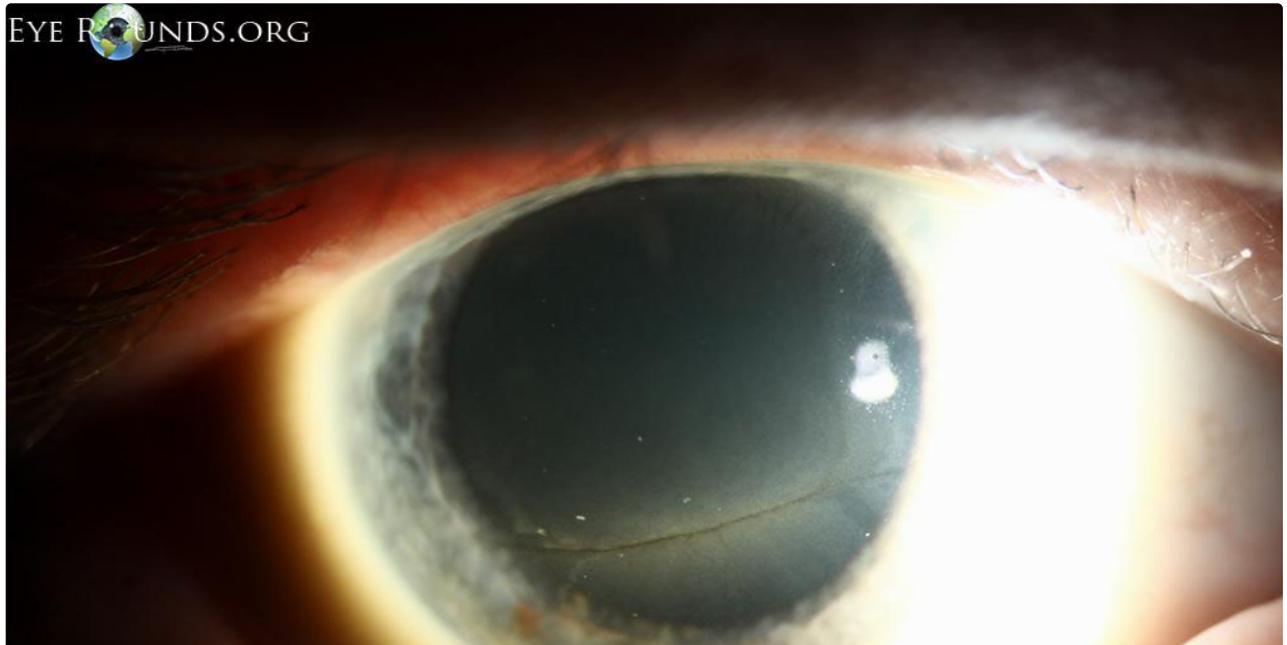


Figure 2: Cornea verticillata in a patient with known Fabry disease.  
Right Eye

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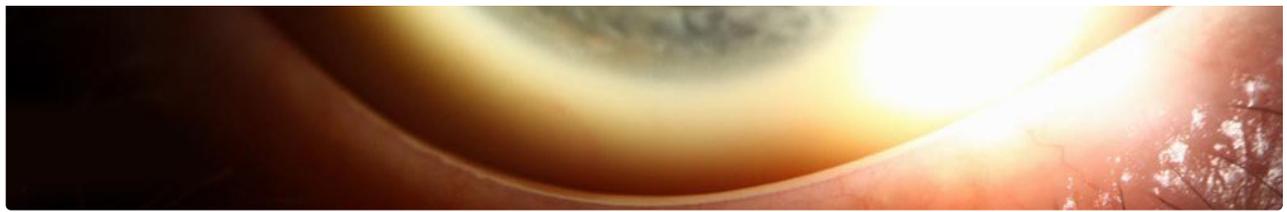


Figure 2: Cornea verticillata in a patient with known Fabry disease.  
Left Eye

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Figure 3: Skin angiokeratoma in the patient with corneal findings shown in Figure 2.  
Appearance of skin

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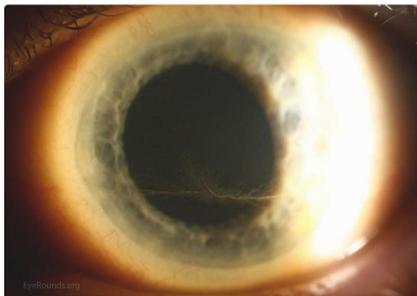
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