What is *Fabry Disease?*

Fabry disease is a form of lysosomal storage disease caused by mutations to the GLA gene.[1,2] It is a rare hereditary condition brought on by the accumulation of globotriaosylceramide, a type of fat in the body cells.[2]

The early detection of Fabry disease is difficult and its symptoms are frequently misdiagnosed.[3]

Signs and Symptoms[2]

- **Acroparesthesias** — Pain in the hands or feet
- **Angiokeratomas** — Small, dark red spots on the skin
- **Hypohidrosis** — Sweating difficulties
- **Corneal verticillata** — Opacity of the cornea
- **Tinnitus** — Ringing in one or both ears
- **Loss of hearing**
- **Heart attacks**
- **Kidney damage**
- **Gastrointestinal problems**

Onset of Symptoms[1,4]

Prenatal	Newborn	Infant	Child	Adolescent	Adult	Older adult
Before birth	Birth–4 weeks	1–23 months	2–11 years	12–18 years	19–65 years	65+ years

The typical ages for symptoms of the disease to appear

Potential Complications[2]

- Stroke
- Heart failure
- Progressive kidney failure

Stay *Aware of The Rare.* Get Screened For *Fabry Disease* Now!

References

1. Genetic and Rare Diseases Information Center. Fabry disease. Available at https://rarediseases.info.nih.gov/diseases/6400/fabry-disease. Accessed on 12 September 2022. **2.** National Library of Medicine. Fabry disease. Available at https://medlineplus.gov/genetics/condition/fabry-disease/#causes. Accessed on 12 September 2022. **3.** Sunder-Plassmann G, Födinger M. Diagnosis of Fabry disease: the role of screening and case-finding studies. Fabry Disease: Perspectives from 5 Years of FOS. 2006. **4.** Cleveland Clinic. Fabry disease. Available at https://my.clevelandclinic.org/health/diseases/16235-fabry-disease#. Accessed on 4 October 2022.

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