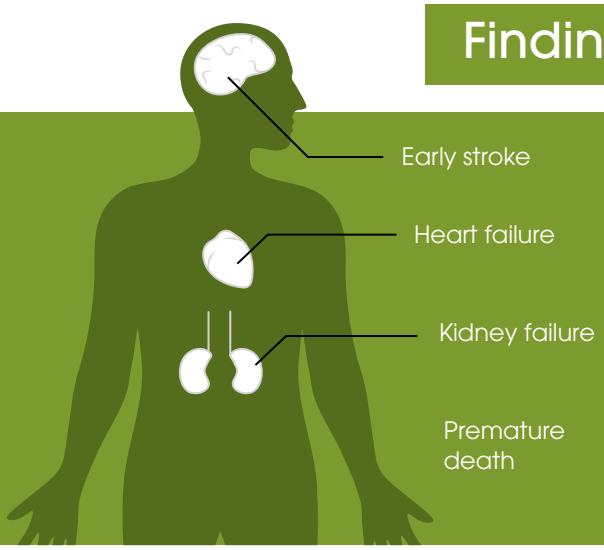
The Life-changing Role of the Eye Care Professional

Finding early ocular signs of Fabry disease

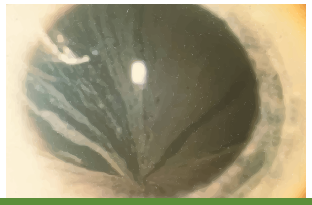


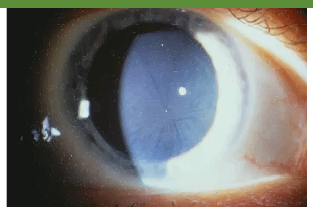
Fabry disease is a progressive and often life-threatening disorder[1,2]

- It is multisystemic and impacts essential organs such as the kidney, heart, and brain[1,2]
- It affects men, women, and children[1,2]
- Undiagnosed and unmanaged, Fabry disease can reduce life expectancy by **15 - 20 years**[3,4]

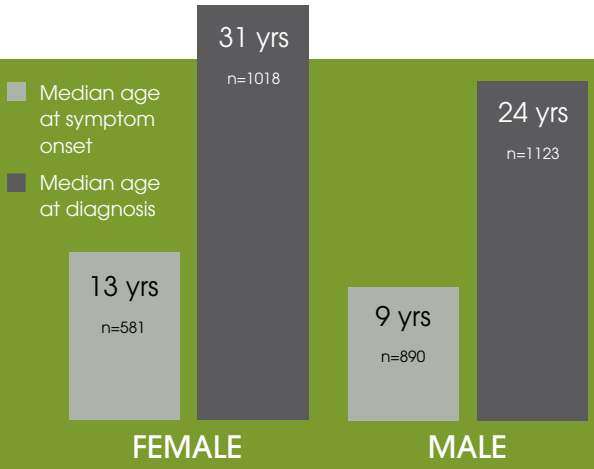
Corneal whorling (verticillata) is one of the earliest and most common signs of Fabry disease[1,5,6]

- Bilateral, whorl-like pattern of powdery, white, yellow, or cream-colored corneal epithelial deposits emanating from a single vortex[6,7]
- Be sure to rule out medications that can cause corneal whorling with long-term use, including amiodarone and chloroquine[1,6]





Seen in ~80% of patients with Fabry disease[5]



15-year diagnostic delay from symptom onset[8]

Identification of early ocular signs can help lead to timely diagnosis and management[6]

- Corneal whorling can be detected with a routine slit lamp exam[6]
- Diagnosis of Fabry disease is confirmed by a simple enzyme assay for males and genetic testing for females[5]
- Fabry disease affects families: For every index patient diagnosed, an average of **5 additional family members may be identified**[9]

Wilcox WR, et al; Fabry Registry. Mol Genet Metab. 2008;93:112-128

For more information: https://www.discoverfabry.com/hcp#fabry-disease-in-your-practice

Eye care professionals can play a critical role in the timely diagnosis of Fabry disease

Detect. Suspect. Refer.

If a patient presents with corneal whorling, suspect Fabry disease and promptly refer to a geneticist for testing

References. 1. Germain GP. *Orphanet Journal of Rare Diseases.* 2010;5:30. **2.** Krasnewich DM and Sidransky E. *Chapter 208: Lysosomal Storage Diseases.* In: Goldman L, Schafer AL, eds. Goldman-Cecil Medicine. 25th ed. Philadelphia, PA: Elsevier Saunders; 2016: 1399-1403. **3.** MacDermot KD, Holmes A, Miners AH. *J Med Genet.* 2001;38(11):750-760. **4.** MacDermot KD, Holmes A, Miners AH. *J Med Genet.* 2001;38(11):769-775. **5.** Mehta A, Hughes DA. *Fabry Disease.* 2002 Aug 5 (Updated 2017 Jan 5). In: Adam MP, Ardinger HH, Pagon RA, et al., editors. GeneReviews® (Internet). Seattle (WA): University of Washington, Seattle; 1993-2018. Available from: https://www.ncbi.nlm.nih.gov/books/NBK1292/. Accessed January 6, 2018. **6.** Morier AM, et al. *Optometry.* 2010;81(9):437-449. **7.** Ikegawa Y, et al. *J Ophthalmology.* 2018; doi: 10.1155/2018/5315137. **8.** Wilcox WR, et al; Fabry Registry. *Mol Genet Metab.* 2008;93:112-128. **9.** Laney D, Fernhoff PM. *J Genet Couns.* 2008;17:79-83.

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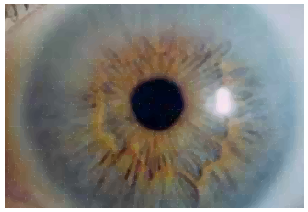
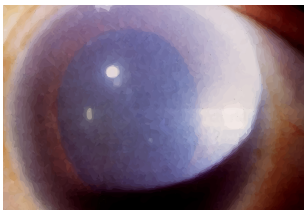
The Life-changing Role of the Eye Care Professional

Finding early ocular signs of MPS I

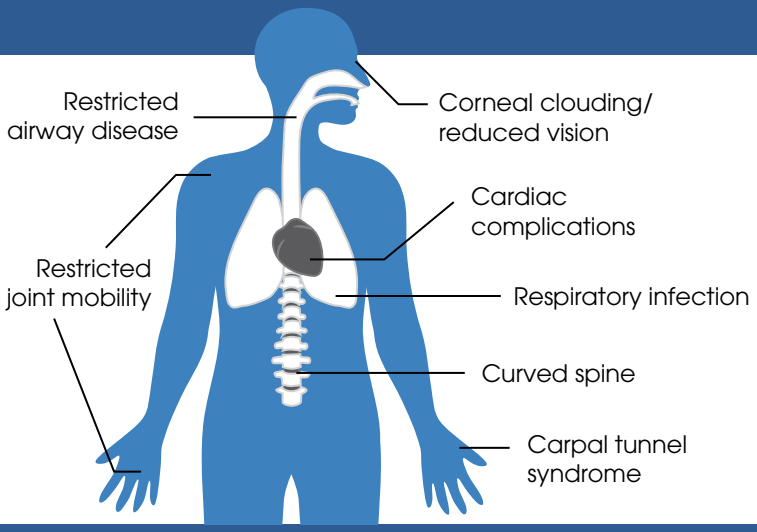
Corneal clouding is among one of the most identifiable early signs of MPS I[1-3]

- Corneal clouding can lead to significant visual disability and blindness, and can begin as early as the first year of life[2,4]
- "Ground-glass" appearance is typically more pronounced in the cornea periphery[4]





Seen in ~70% of all patients with MPS I[3]

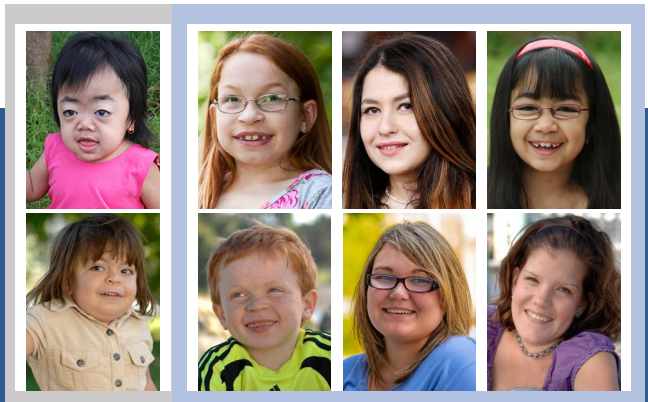


MPS I is a progressive and often life-threatening disease[3,5-7]

- Progressive organ damage and failure[3,5,6]
- Severe and debilitating morbidities[3,6,7]
- Reduced life expectancy[3,6]

Identification of early ocular signs can help lead to timely diagnosis[1]

- Attenuated MPS I can often go misdiagnosed for 5+ years[6]
- **Corneal clouding is an important identifier of MPS I since virtually all patients develop this abnormality** (as early as 1 year of age in Hurler patients and by 4 to 10 years in attenuated patients)[3,8]



MPS I Spectrum[6,7]

For more information: https://www.mps1disease.com/healthcare/about-mps1/symptoms-and-management/ocular-system

Eye care professionals can play a critical role in the timely diagnosis of MPS I

Detect. Suspect. Refer.

If a patient presents with corneal clouding, suspect MPS I and promptly refer to a geneticist for testing

References. 1. Fahnehjelm K, et al. *Acta Ophthalmol.* 2012;90:595-602. **2.** Clarke LA. *Mucopolysaccharidosis Type I.* In: Adam MP, Ardinger HH, Pagon RA, et al., eds. GeneReviews® (Internet). Seattle, WA: University of Washington, Seattle; 1993-2018. https://www.ncbi.nlm.nih.gov/books/NBK1162/?report=printable. Accessed February 1, 2018. **3.** Beck M, Arn P, Giugliani R, et al. *Genet Med.* 2014;16(10):759-765. **4.** Summers CG, et al. *Rheumatology.* 2011;50:v34-v40. **5.** Coutinho MF, et al. *Biochem Res Int.* 2012; 2012:471325. **6.** Bruni S, Lavery C, Broomfeld A. *Molec Genet Metab Reports.* 2016;8:67-73. **7.** Muenzer J. *Rheumatology.* 2011;50:v4-v12. **8.** Ashworth J, et al. *Eye.* 2006; 20:553-563.

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