Ocular Manifestations of Lysosomal Storage Disorders

Genetic disorders that an ophthalmologist, optometrist or corneal specialist can help identify

Corneal Whorling or Opacities
Seen in:
- Fabry disease
- MPS VI
- Niemann-Pick disease
- Sandhoff disease
- Sialidosis II

Lens Opacities
Seen in:
- Fabry disease
- Mannosidosis
- Sialidosis I

Cherry-Red Spot
Seen in:
- Farber disease
- Galactosialidosis
- GM1 Gangliosidosis
- Mucopolysaccharidosis (MPS) I, IV, VI, VII

Retinosis Pigmentosis/Retinal Dystrophy
Seen in:
- Neuronal Ceroid Lipofuscinoses
- Tay-Sachs disease

Strabismus
Seen in:
- Gaucher disease types II and III
- GM1 Gangliosidosis
- Infantile sialic acid storage disease (ISSD)
- Metachromatic leukodystrophy III
- Salla disease
- Sialidosis I

Retinal infiltrates
Strabismus

Patients with these manifestations should be referred to a geneticist for testing and further intervention. Lysosomal storage disorders are progressive and often life-threatening. Early diagnosis and intervention are important.

For more information, visit www.lysosomallearning.com or call Genzyme Medical Information at 800-745-4447 (option 2).