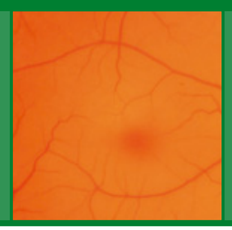
Ocular Manifestations of Lysosomal Storage Disorders

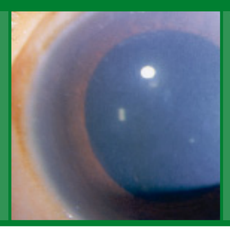
Genetic disorders that an ophthalmologist, optometrist or corneal specialist can help identify


1

Cherry-Red Spot

Seen in:

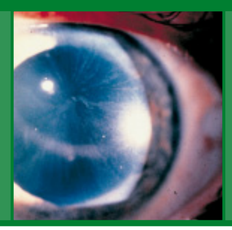
- Farber disease
- Galactosialidosis
- GM1 Gangliosidosis I
- Niemann-Pick disease
- Sandhoff disease
- Sialidosis
- Tay-Sachs disease I


2

Corneal Clouding

Seen in:

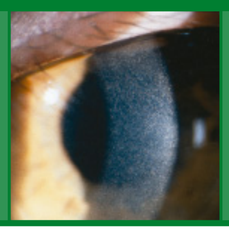
- Fucosidosis (slight)
- GM1 Gangliosidosis
- Mucolipidosis II, III, IV
- Mucopolysaccharidosis (MPS) I, IV, VI, VII


3

Corneal Whorling or Opacities

Seen in:

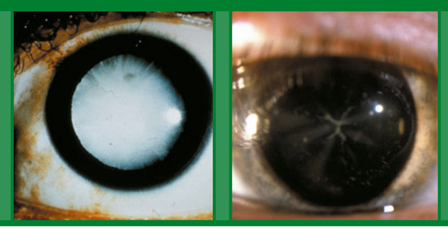
- Fabry disease
- MPS VI
- Niemann-Pick
- Sialidosis II


4

Cystine Crystal Deposits

Seen in:

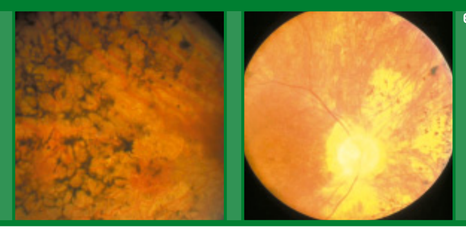
- Cystinosis


5

Lens Opacities

Seen in:

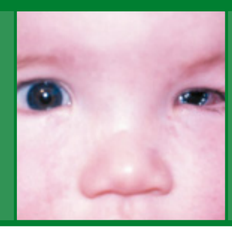
- Fabry disease
- Mannosidosis
- Sialidosis I


6

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

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6. Courtesy of John A. Moran Eye Center, University of Utah.

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).

Lysosomal Storage Disorders with Ocular Manifestations

Aspartylglycosaminuria
Eye deformities

Cystinosis
Refractile polychromatic crystals
Peripheral retinopathy
Hypopigmented fundi
Photophobia

Fabry Disease
Corneal whorling or opacities
Lenticular opacities
Retinal and lenticular abnormalities
Cataracts

Farber Disease
Cherry-red spot on retina
Pigmentary mottling
Granulomas

Fucosidosis type I
Tortuous conjunctival vessels
Corneal clouding (slight)
Pigmentary retinopathy

Galactosialidosis
Macular cherry-red spot
Gradual vision loss

Gaucher Disease type II
Strabismus

Gaucher Disease type III
Horizontal saccadic abnormalities
Retinal infiltrates
Strabismus

GM1 Gangliosidoses
Diffuse corneal clouding (type I)
Macular cherry-red spot (type I)
Retinal vascular tortuosity and hemorrhage (type I)
Optic atrophy (types I and II)
Blindness (types I and II)
Nystagmus (types I and II)
Strabismus (types I and II)

Infantile Sialic Acid Storage Disease (ISSD)
Clear cornea
Epicanthal folds
Ptosis
Nystagmus
Strabismus

Krabbe Disease
Vision loss leading to blindness

Mannosidosis
Lens opacities
Prognathism

Metachromatic Leukodystrophy
Nystagmus (types I and III)
Grayish discoloration of the macula (type I)
Optic atrophy leading to blindness (types I, II, and III)
Hypotonia (type I)
Strabismus (type III)

Mucolipidosis
Corneal clouding (types II, III, and IV)
Mild retinopathy (type III)
Hyperopic astigmatism
Pigmentary changes (type IV)
Optic atrophy (type IV)

Mucopolysaccharidoses
Corneal clouding (MPS I, IV, VI, and VII)
Retinal dysfunction/degeneration (MPS II)
Corneal opacities (MPS VI)

Neuronal Ceroid Lipofuscinoses
Retinitis pigmentosa
Visual loss
Optic atrophy leading to blindness

Niemann-Pick Disease
Macular cherry-red spot
Corneal opacification
Brown discoloration of the anterior lens capsule

Salla Disease
Nystagmus
Exotropia
Strabismus

Sandhoff Disease
Macular cherry-red spot
Visual inattention (progressive)
Blindness

Sialidosis
Lens opacities (type 1)
Impaired color vision (type I)
Night blindness (type I)
Nystagmus (type I)
Strabismus (type I)
Corneal opacities (type II)
Macular cherry-red spot (types I and II)
Loss of visual acuity (types I and II)

Tay-Sachs Disease
Macular cherry-red spot (type I)
Progressive visual inattention leading to blindness (type I)
Retinitis pigmentosis (type II)

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