

Teaching Video NeuroImage: Clues in Myoclonus Evaluation

When to Consider Sialidosis

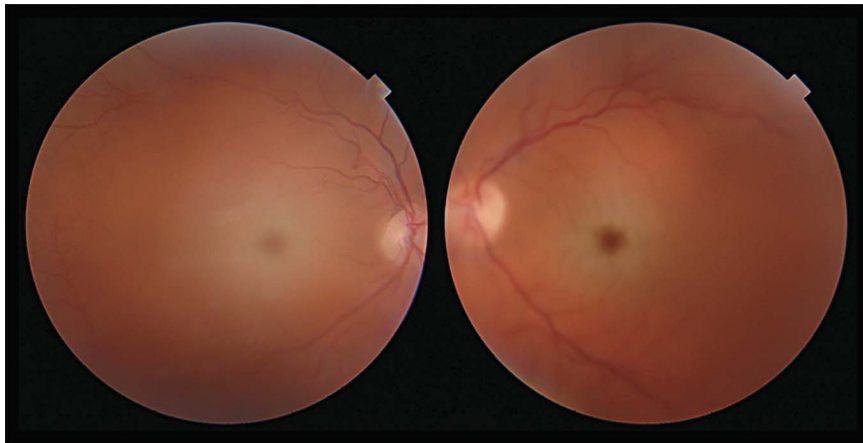
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Figure Ophthalmic Examination: Cherry-Red Spot



The dilated fundus examination showed bilateral cherry-red spots on the retina. This is a red zone at the center of the macula surrounded by retinal opacification. It is due to the accumulation of different lipids, sphingolipid, or oligo-saccharide material in the ganglion cells of the retina.

A 53-year-old woman was presented for the evaluation of visual disturbances, generalized and multifocal myoclonus, and progressive ataxia that began at age 30 years (cf. Video 1). Bilateral cherry-red spots in the macula (figure) and a cortical origin in the EEG-EMG coregistration with back-averaging were observed. Reduced neuraminidase activity in fibroblasts and the homozygous mutation c.403G>A in the *NEU1* gene confirmed the diagnosis of sialidosis type I.

Sialidosis or cherry-red spot–myoclonus syndrome is classified into normomorph or type I, beginning usually after 20 years old, whereas dysmorphic or type II begins at birth or in early childhood. In both, generalized myoclonus and ataxia can be found. EEG-EMG coregistration may show cortical potential followed by the myoclonus, proving a cortical origin. Differential diagnosis is necessary for other inherited metabolic disorders such as Tay-Sachs disease or Unverricht-Lundborg disease. Cherry-red spots on the retina, cortical myoclonus, and progressive ataxia are essential keys to suspicion.^{1,2}

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Disclosure

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Name	Location	Contribution
Beatriz Vélez Gómez, MD	Hospital Universitario Virgen del Rocío, Seville, Spain	Examined the patient, drafted the manuscript, and videotaped
Silvia Jesús, MD, PhD	Hospital Universitario Virgen del Rocío, Seville, Spain	Examined the patient, videotaped, and provided critical review of the manuscript
Daniel Macías-García, MD	Hospital Universitario Virgen del Rocío, Seville, Spain	Examined the patient and provided critical review of the manuscript
Beatriz Lechon, MD	Hospital Universitario Virgen del Rocío, Seville, Spain	Performed ophthalmologic examination
Pablo Mir, MD, PhD	Hospital Universitario Virgen del Rocío, Seville, Spain	Provided critical review of the manuscript and contributed to data acquisition

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