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Teaching Video NeuroImages: Clues in Myoclonus Evaluation: When to Consider Sialidosis

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Contributions:
Beatriz Vélez Gómez: Drafting/revision of the manuscript for content, including medical writing for content; Additional contributions: Dr Vélez Gómez examined the patient, drafted the manuscript and videotaped.
Silvia Jesús: Drafting/revision of the manuscript for content, including medical writing for content; Additional contributions: Dr. Jesús examined the patient, videotaped and provided critical review of the manuscript.
Daniel Macias-Garcia: Drafting/revision of the manuscript for content, including medical writing for content; Additional contributions: Dr. Macias-Garcia examined the patient and provided critical review of the manuscript.
Beatriz Lechon: Drafting/revision of the manuscript for content, including medical writing for content; Additional contributions: Dr. Lechon performed ophtalmological examination
Pablo Mir: Drafting/revision of the manuscript for content, including medical writing for content; Major role in the acquisition of data; Additional contributions: Dr. Mir provided critical review of the manuscript and contributed to data acquisition.

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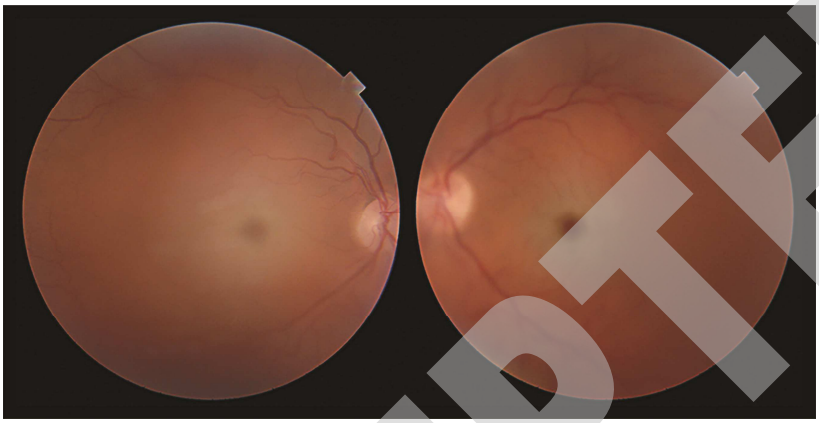
A 53-year-old woman was presented for evaluation of visual disturbances, generalized and multifocal myoclonus, and progressive ataxia that began at age 30 (cf. Video). Bilateral cherry-red spots in the macula (Figure 1) and a cortical origin in the EEG-EMG co-registration with back-averaging were observed. 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 normomorphic or type I, beginning usually after twenty-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 co-registration 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[12].

Figure 1. Ophthalmic examination: Cherry-red spot. The dilated fundus examination showed bilateral cherry-red spots on the retina. This is a red zone at the centre of the macula surrounded by retinal opacification. It is due to the accumulation of different lipid, sphingolipid, or oligosaccharide material in the ganglion cells of the retina.



Video. Cortical Myoclonus.

We observed generalized and multifocal myoclonus at rest (segment 1). It worsens during a sustained posture, such as holding arms or legs out against gravity. The myoclonus activity also increases with tactile sensory stimulation (segment 2). It worsens with activities such as finger-nose-finger test (segment 3). The patient shows dysarthria. The speech facility deteriorates with myoclonus (segment 4).

Appendix 1: Authors

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

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169826 Teaching Slides [KES 06.17.21] - USE THIS VERSION -- http://links.lww.com/WNL/B466

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