

Neurology Publish Ahead of Print
DOI: 10.1212/WNL.0000000000012464

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

Author(s):

Beatriz Vélez Gómez, MD¹; Silvia Jesús, MD, PhD^{1,2}; Daniel Macias-Garcia, MD^{1,2}; Beatriz Lechon, MD³; Pablo Mir, MD, PhD^{1,2}

Corresponding Author:

Pablo Mir
pmir@us.es

Neurology® Published Ahead of Print articles have been peer reviewed and accepted for publication. This manuscript will be published in its final form after copyediting, page composition, and review of proofs. Errors that could affect the content may be corrected during these processes.

Affiliation Information for All Authors: 1 Unidad de Trastornos del Movimiento. Servicio de Neurología y Neurofisiología Clínica. Hospital Universitario Virgen del Rocío/Instituto de Biomedicina de Sevilla/ CSIC/Universidad de Sevilla, Seville, Spain. 2 Centro de Investigación Biomédica en Red sobre Enfermedades Neurodegenerativas (CIBERNED), Madrid, Spain. 3 Servicio de Oftalmología. Hospital Universitario Virgen del Rocio, Seville, Spain.

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 ophthalmological 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.

Number of characters in title: 76

Abstract Word count: 0

Word count of main text: 146

References: 2

Figures: 1

Tables: 0

Supplemental: Figure_1, teaching_slide_deck, manuscript, Before_After, Response_to_reviewers.

Search Terms: [64] Epilepsy semiology, [296] Myoclonus; see Movement Disorders/myoclonus, [159] Peroxisomes, [173] Myoclonus, [191] Retina

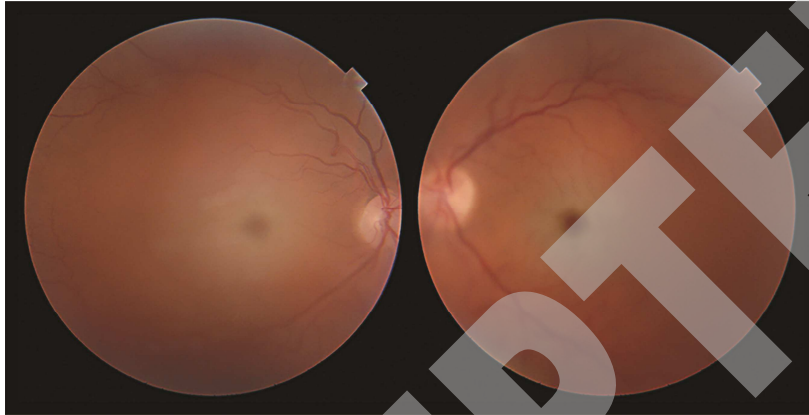
Study Funding: This research was supported by grants from the Spanish Ministry of Science and Innovation [RTC2019-007150-1], the Instituto de Salud Carlos III-Fondo Europeo de Desarrollo Regional (ISCIII-FEDER) [PI14/01823, PI16/01575, PI18/01898, PI19/01576], the Consejería de Economía, Innovación, Ciencia y Empleo de la Junta de Andalucía [CVI-02526, CTS-7685], the Consejería de Salud y Bienestar Social de la Junta de Andalucía [PI-0471-2013, PE-0210-2018, PI-0459-2018, PE-0186-2019], and the Fundación Alicia Koplowitz. Silvia Jesús has a contract Acción B Clínicos-Investigadores (Action B Clinicians-Researchers) contract [B-0007-2019] funded by the Consejería de Salud y Familiar , and Daniel Macías-García has a Río Hortega contract [CM18/00142] funded by the Health Institute Carlos III.

Disclosures: *B. Vélez Gómez has received honoraria from Sanofi, *S. Jesús has received honoraria from Abbvie, Bial, Merz, UCB, Italfarmaco and Zambon, *D. Macías-García has received honoraria from Abbvie, Teva and Zambon, *B. Lechon reports no disclosures relevant to the manuscript, *P. Mir has served in the Advisory Boards or received honorarium for lecturing from Abbott, Allergan, Abbvie, Bial, Britannia, Italfarmaco, Merz, UCB, Teva and Zambon.

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 suspicion¹².

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 ophthalmological examination
Pablo Mir, MD, PhD	Hospital Universitario Virgen del Rocio, Seville	Provided critical review of the manuscript and contributed to data acquisition

169826 Video [KES 06.15.21] ---<http://links.lww.com/WNL/B465>

169826 Teaching Slides [KES 06.17.21] - USE THIS VERSION --

<http://links.lww.com/WNL/B466>

REFERENCES

1. Caciotti A, Melani F, Tonin R, et al. Type I sialidosis, a normosomatic lysosomal disease, in the differential diagnosis of late-onset ataxia and myoclonus: An overview. *Mol Genet Metab.* 2020;129(2):47-58. doi:10.1016/j.ymgme.2019.09.005
2. Orsini A, Valetto A, Bertini V, et al. The best evidence for progressive myoclonic epilepsy: A pathway to precision therapy. *Seizure.* 2019;71:247-257. doi:10.1016/j.seizure.2019.08.012

Neurology®

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

Beatriz Vélez Gómez, Silvia Jesús, Daniel Macias-Garcia, et al.

Neurology published online July 14, 2021

DOI 10.1212/WNL.0000000000012464

This information is current as of July 14, 2021

Updated Information & Services	including high resolution figures, can be found at: http://n.neurology.org/content/early/2021/07/14/WNL.0000000000012464.citation.full
Subspecialty Collections	This article, along with others on similar topics, appears in the following collection(s): Epilepsy semiology http://n.neurology.org/cgi/collection/epilepsy_semiology Myoclonus http://n.neurology.org/cgi/collection/myoclonus Myoclonus; see Movement Disorders/myoclonus http://n.neurology.org/cgi/collection/myoclonus_see_movement_disorders-myoclonus Peroxisomes http://n.neurology.org/cgi/collection/peroxisomes Retina http://n.neurology.org/cgi/collection/retina
Permissions & Licensing	Information about reproducing this article in parts (figures, tables) or in its entirety can be found online at: http://www.neurology.org/about/about_the_journal#permissions
Reprints	Information about ordering reprints can be found online: http://n.neurology.org/subscribers/advertise

Neurology® is the official journal of the American Academy of Neurology. Published continuously since 1951, it is now a weekly with 48 issues per year. Copyright © 2021 American Academy of Neurology. All rights reserved. Print ISSN: 0028-3878. Online ISSN: 1526-632X.

