Teaching NeuroImages: Kearns-Sayre syndrome

Michael T.B. Nguyen, MD, Jonathan Micieli, MD, FRCSC, and Edward Margolin, MD, FRCSC, DABO

Neurology® 2019;92:e519-e520. doi:10.1212/WNL.0000000000006861

Correspondence

Dr. Margolin
edmargin@gmail.com

Figure 1 Ptosis and ophthalmoplegia

Photographs of eye movement demonstrate severe bilateral ptosis and mild diffuse ophthalmoplegia in all directions of attempted gaze.

A 19-year-old man presented 6 months postimplantation of permanent pacemaker for complete heart block with bilateral nonfatigable symmetric ptosis, diminished levator superioris function, and symmetric ophthalmoplegia (figure 1). Funduscopy revealed bilateral pigmentary retinopathy (figure 2). Skeletal muscle biopsy revealed presence of ragged-red fibers, consistent with Kearns-Sayre syndrome. This mitochondrial disorder is characterized by the triad of onset before age 20, chronic progressive external ophthalmoplegia, and pigmentary retinopathy. Other findings can include complete heart block, cerebellar ataxia, deafness, and endocrinopathies. CSF folate levels should be measured and supplemented if low. There is no definitive treatment but annual surveillance for comorbidities is required.^{1,2}

Study funding

No targeted funding reported.

Disclosure

The authors report no disclosures relevant to the manuscript. Go to Neurology.org/N for full disclosures.

MORE ONLINE

→Teaching slides

links.lww.com/WNL/A796

Figure 2 Pigmentary retinopathy

Fundus examination of right (A) and left eye (B) shows bilateral pigmentary retinopathy. There is diffuse depigmentation of the retinal pigment epithelium in a salt-and-pepper pattern of pigment clumping and involvement of the peripapillary zone.

Appendix Authors

Name	Location	Role	Contribution
Michael T.B. Nguyen, MD	University of Toronto	Author	Drafted manuscript
Jonathan A. Micieli, MD, FRCS	University of Toronto	Author	Revised manuscript
Edward Margolin, MD, FRCS, DABO	University of Toronto	Author	Conceptualization of case report and revised manuscript

References

1. Kearns TP, Sayre GP. Retinitis pigmentosa, external ophthalmoplegia, and complete heart block. *Arch Ophthalmol* 1958;60:280–289.
2. Shemesh A, Margolin E. Kearns Sayre syndrome. In: StatPearls [Internet]. Treasure Island, FL: StatPearls Publishing; 2018. Available at: ncbi.nlm.nih.gov/books/NBK482341/. Accessed June 9, 2018.

Neurology®

Teaching NeuroImages: Kearns-Sayre syndrome
Michael T.B. Nguyen, Jonathan Micieli and Edward Margolin
Neurology 2019;92:e519-e520
DOI 10.1212/WNL.0000000000006861

This information is current as of January 28, 2019

Updated Information & Services	including high resolution figures, can be found at: http://n.neurology.org/content/92/5/e519.full
References	This article cites 1 articles, 0 of which you can access for free at: http://n.neurology.org/content/92/5/e519.full#ref-list-1
Subspecialty Collections	This article, along with others on similar topics, appears in the following collection(s): All Neuro-ophthalmology http://n.neurology.org/cgi/collection/all_neuroophthalmology Eyelids http://n.neurology.org/cgi/collection/eyelids Mitochondrial disorders http://n.neurology.org/cgi/collection/mitochondrial_disorders Ocular motility http://n.neurology.org/cgi/collection/ocular_motility 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 © 2019 American Academy of Neurology. All rights reserved. Print ISSN: 0028-3878. Online ISSN: 1526-632X.