

Teaching NeuroImages: Kearns-Sayre syndrome

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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

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Disclosure

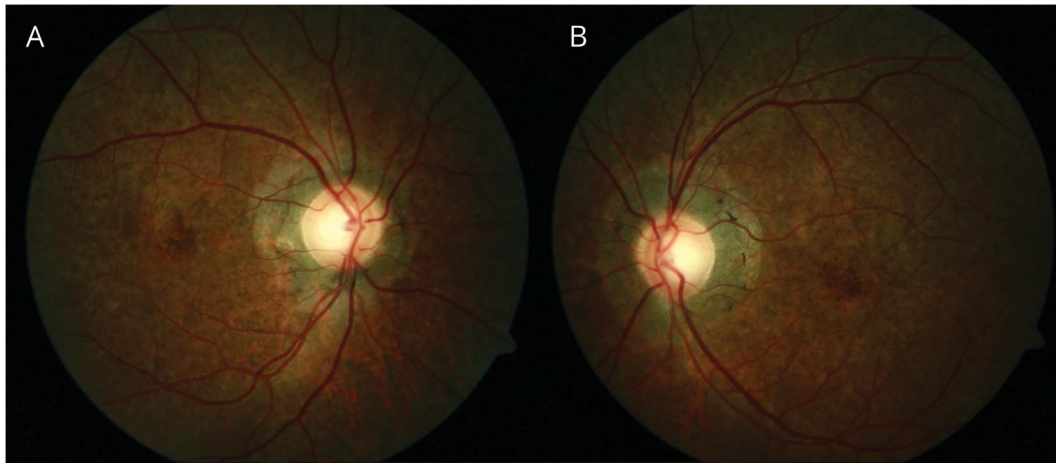
The authors report no disclosures relevant to the manuscript. Go to Neurology.org/N for full disclosures.

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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

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