Teaching NeuroImages: Retinopathy in spinocerebellar ataxia type 3

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Figure 1 Retinography, left eye

Peripapillary chorioretinal atrophy (red arrow), macular atrophy (white arrow), and extensive loss of retinal pigment epithelium in the lower retinal periphery (star).

A 17-year-old African Brazilian woman presented with a 1-year history of progressive ataxia, dysarthria, and decreased visual acuity. Family history was remarkable for multiple relatives with similar motor symptoms but no visual complaints. Examination showed upward gaze palsy, dysarthria, ataxia, and increased tone and reflexes. Fundoscopic examination and subsequent optical coherence tomography revealed bilateral atrophic maculopathy (figures 1 and 2). Genetic testing confirmed the diagnosis of spinocerebellar ataxia type 3 (SCA3) by revealing abnormal CAG repeats in the *ATXN3* gene—the pathologic allele had 68 repeats and the normal allele 14 repeats.

Retinopathy, a typical finding in spinocerebellar ataxia type 7,¹ has rarely been associated with SCA3.^{2–4} Further studies should elucidate this association, thereby possibly expanding the phenotypic spectrum of SCA3.

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Figure 2 Optical coherence tomography, right eye

(A) Near-infrared light shows macular atrophy (red arrow) and peripapillary chorioretinal atrophy (yellow arrow). (B) Complete foveal atrophy, loss of deep macular layers within the atrophic topography, and loss of peripapillary retinal pigment epithelium (star).

Disclosure

F.A. Nascimento is a member of the *Neurology*[®] Resident & Fellow Section Editorial Team. M.G. Ferreira, N. Shiokawa, M.T. Sato, and H.A.G. Teive report no relevant disclosures. Go to Neurology.org/N for full disclosures.

Appendix Authors

Name	Location	Contribution
Fábio A. Nascimento, MD	Baylor College of Medicine, Houston, TX	Designed study, analyzed and interpreted the data, drafted the manuscript
Matheus G. Ferreira, MD	Universidade Federal do Paraná, Curitiba, Brazil	Designed study, analyzed and interpreted the data, drafted the manuscript
Naoye Shiokawa, MD	Universidade Federal do Paraná, Curitiba, Brazil	Analyzed and interpreted the data, revised the manuscript

Appendix (continued)

Name	Location	Contribution
Mario T. Sato, MD, PhD	Universidade Federal do Paraná, Curitiba, Brazil	Analyzed and interpreted the data, revised the manuscript
Hélio A.G. Teive, MD, PhD	Universidade Federal do Paraná, Curitiba, Brazil	Designed and conceptualized study, analyzed and interpreted the data, revised the manuscript, supervised study, final approval

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