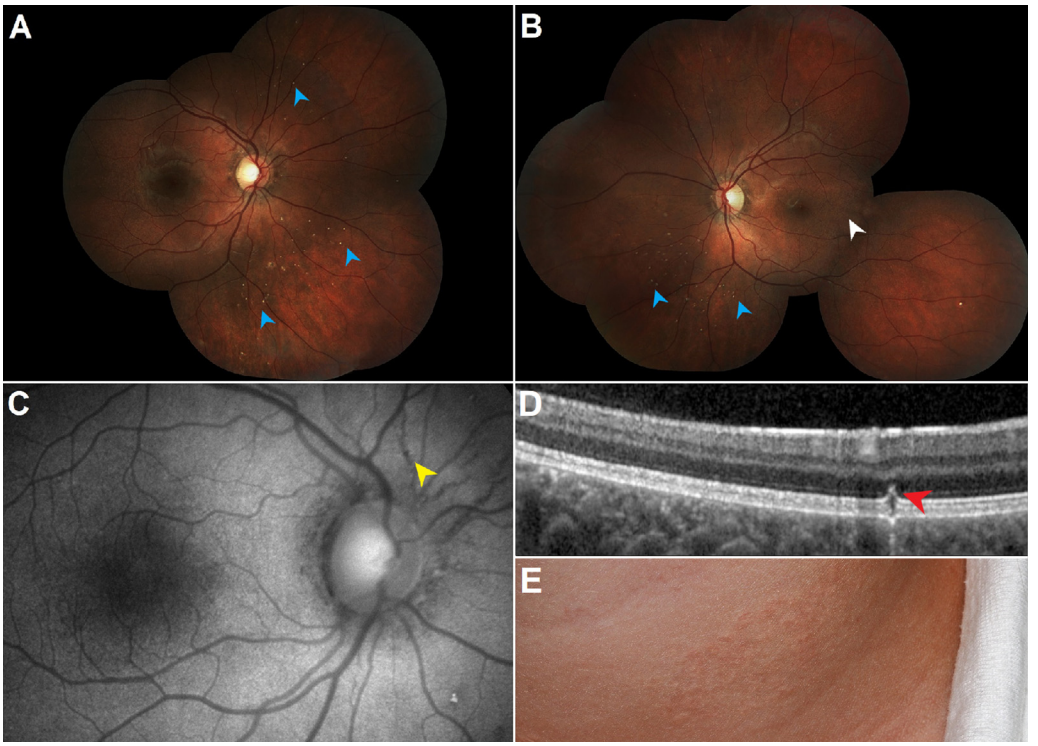

Retinal crystalline lesions in pseudoxanthoma elasticum

An 11-year-old healthy boy was evaluated for incidental retinal crystalline lesions. Visual acuity was 20/20 in both eyes. Fundus examination of the right (A) and left (B) eyes demonstrated multiple crystalline bodies (blue arrows) scattered throughout the midperipheral and peripheral retina, a peau d'orange pattern of retinal pigment epithelial (RPE), alterations in the temporal retina (white arrow), and angioid streaks (yellow arrow) around the optic nerve on fundus autofluorescence imaging (C). Optical coherence tomography (D) was performed through a retinal crystalline lesion, demonstrating a hyper-reflective lesion (red arrow) in the outer retinal layers with alterations in the RPE/Bruch membrane complex. Given the constellation of ocular findings, a diagnosis of pseudoxanthoma elasticum (PXE) was suspected. A dermatologic examination was subsequently performed demonstrating yellowish papules around the neck in a cobblestone pattern (E). Genetic testing revealed homozygous pathogenic variants in the gene ABCC6, a mutation associated with PXE. Retinal crystalline lesions can be a remarkable ocular manifestation of PXE and may represent degenerated photoreceptors overlying an area of RPE/Bruch membrane disruption.

Jason M. Kwok, MD, Cynthia VandenHoven, BAA, CRA, Rajeev H. Muni, MD, FRCSC
Department of Ophthalmology and Vision Sciences, University of Toronto, Toronto, Ont.

Originally received Dec. 13, 2020. Final revision Mar. 10, 2021. Accepted Apr. 7, 2021.

Correspondence to Rajeev H. Muni, MD, MSc, FRCSC, Department of Ophthalmology, St. Michael's Hospital, Unity Health Toronto, 8th floor, Donnelly Wing, 30 Bond St., Toronto, ON, M5B 1W8.; rajeev.muni@gmail.com.

Footnotes and Disclosure

The authors have no proprietary or commercial interest in any materials discussed in this article.

Supported by: University of Toronto Department of Ophthalmology and Vision Sciences Pilot Research Grant.