Discussion: Although he had features suggestive of a central retinal vein occlusion (CRVO), these could not explain the visual acuity of 3MCF and relative afferent pupillary defect, as the features of CRVO were mild. Retrobulbar neuritis was the main cause of reduced vision because of the visual acuity, RAPD, markedly reduced light appreciation and color desaturation, leakages around the optic nerve head seen in the late stages of FFA and the great improvement on commencing intravenous methylprednisolone.

Conclusion: A high index of suspicion is required to rule out retrobulbar neuritis when it co-exists with a retinal vein occlusion that cannot account for the clinical features seen.

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Hemiretinal cone-rod dystrophy in two male siblings: an unusual presentation

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Introduction: Cone-rod dystrophies are inherited retinal disorders occurring in the first three decades of life and rarely, the fifth decade.[1,2] The prevalence is 1 in 40,000. The ABCA4 gene is the most prominent causal gene known.[3] Symptoms and signs include decreased vision, central scotomas, colour vision loss, photophobia, bone-spicule pigmentation, macular and retinal atrophy.[2-5] Typical findings include bone-spicule pigmentation, macular and retinal atrophy.[5] On multimodal imaging, characteristic electroretinography and visual field abnormalities are seen. [2,3] Hemiretinal variants of rod-cone dystrophies are relatively rare.[6]

Methods: Case reports of two male siblings with hemiretinal cone-rod dystrophy by multimodal imaging. Informed consent was obtained from the patients for this report.

Case Presentation: The first patient is an 18-year-old male who presented with diminished vision since childhood. He had used spectacles for two years with little improvement. Best corrected visual acuity for distance and near was 6/36 and N12 in both eyes. Pendular nystagmus was present. The intraocular pressure was 10 mmHg bilaterally. He had disc pallor and atrophic macula with bull's eye maculopathy bilaterally. Hyperpigmented bone-spicule changes and attenuated vessels were restricted to the inferior and nasal retina bilaterally. (Figure 1a). Fundus autofluorescence showed hypoautofluorescent patches in the inferior and nasal hemiretina and alternating hyper- and hypo-fluorescent pattern at the macula, in a bull's eye pattern (Figure 1b). Optical coherence tomography (OCT) scan revealed retinal thinning with disruption of the ellipsoid layer, typifying photoreceptor loss (Figure 1c). Central visual field showed early ring scotoma pattern, and electroretinography showed reduced amplitudes in the photopic phase, reduced extinguished response in the scotopic phase across the whole retina.

The second patient is a 16-year-old male presenting with defective vision since childhood. Best corrected visual acuity was 6/36 in both eyes. Pendular nystagmus was present. The intraocular pressure was 12 mmHg bilaterally. Pale discs, attenuated vessels, symmetrical retinal pigment epithelium atrophic changes

and pigmentation in the inferior and nasal retina were present. Atrophic elliptical macula lesions were seen (Figure 2a). Hypoautofluorescence was seen in the inferior & and nasal retina (Figure 2b). OCT revealed distorted architecture of the retinal layers, altered foveal contour, atrophy and corrugations (Figure 2c). Central visual field showed peripherally constricted fields. Electroretinography showed extinguished waves with diminished amplitudes in the photopic phase involving the whole retina. Findings were in keeping with atypical hemiretinal cone-rod dystrophy in both patients.

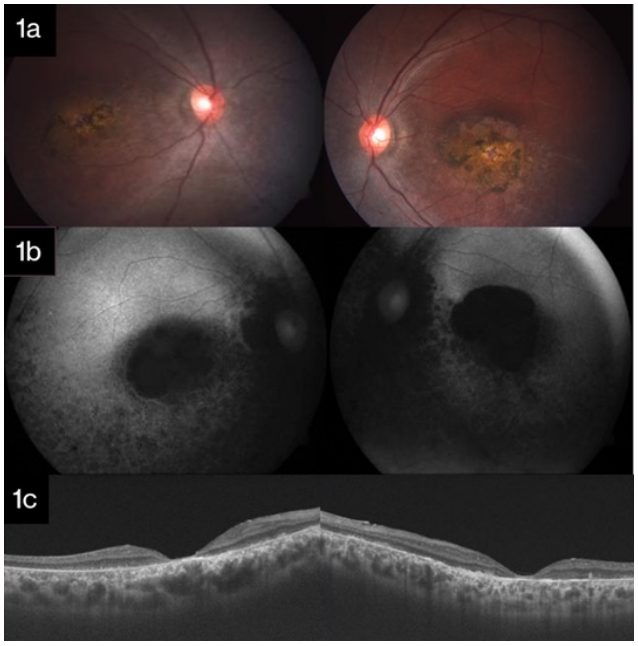

Figure 1a *shows bone spicule pigmentation in the inferior retina and atrophic maculopathy, while* ***Figure 1b*** *shows hypoautofluoresence of bone spicules and* ***Figure 1c*** *shows thinning at the fovea with loss of ellipsoid zone in the first sibling.*

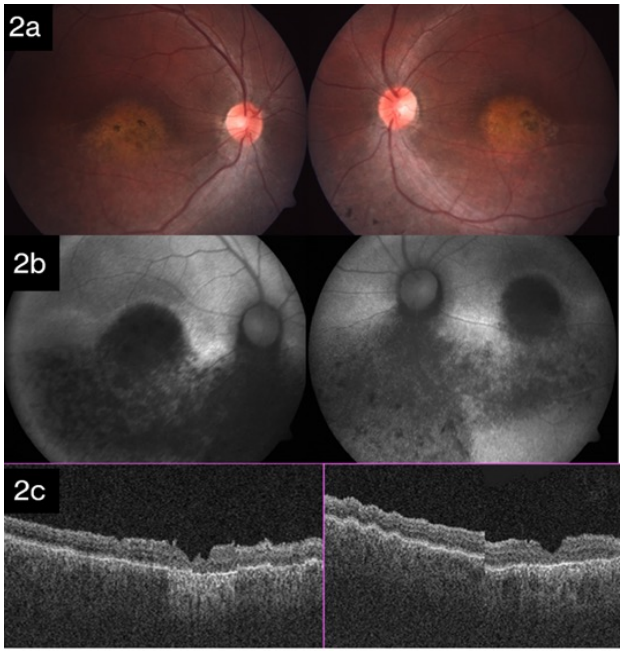

Figure 2a *shows bone spicule pigmentation in inferior retina with atrophic maculopathy, while* ***Figure 2b*** *shows hypoautofluoresence of the bone spicules in inferior retina, and* ***Figure 2c*** *shows thinning at the fovea with loss of the ellipsoid zone in the second sibling.*

Discussion: Hemiretinal cone-rod dystrophy is a rare variant of cone-rod dystrophy with few cases reported in literature. It has been reported in a seven-year-old female with a mutation at C1490Y of the ABCA4 gene.[6] Amelogenesis imperfecta with hemiretinal and bone spicule pigmentation is described in 3 families due to a mutation in CNNM 4.[7,8] The autosomal recessive form of amelanogenesis imperfecta is linked with hemiretinal cone dystrophy.[9]

Our patients had characteristic cone-rod dystrophy symptoms such as decreased vision and colour vision loss and bull's-eye maculopathy.[3,4] Electroretinography findings of cone-rod dystrophy involving the whole retina, despite signs only in the inferior and temporal retina, may be an indication of evolving or early-stage typical cone-rod dystrophy.

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