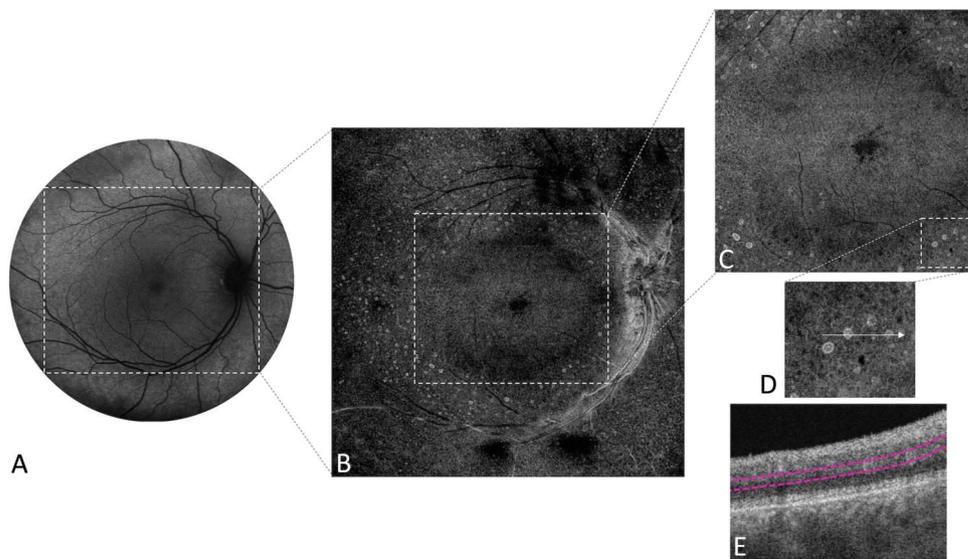


En Face Optical Coherence Tomography Imaging in Enhanced S-Cone Syndrome

Fig. 1. En face OCT imaging in a patient with enhanced S-Cone syndrome. **A.** Fundus autofluorescence shows a slightly hyperautofluorescent ring in midperiphery. **B.** 12- × 12-mm en face OCT imaging of the right eye, at the deep vascular complex level, shows numerous lesions surrounding the vascular arcades. **C.** 6 × 6 mm en face OCT imaging of the right eye, at the deep vascular complex level, shows numerous target-like lesions. **D.** Zoomed en face image of a target-like deposit near the inferior temporal vascular arcade and the embedded B-scan (**E**) demonstrate the correspondence between the target-like lesions seen on the en face imaging and the rosette-like lesions visualized on structural OCT.



Enanced S-Cone syndrome (ESCS) is a retinal dystrophy secondary to an autosomal recessive inheritance on NR2E3 gene. We report the peculiar aspect of en face optical coherence tomography (OCT) in a case of ESCS.

A 26-year-old woman without any past medical history and no family history of consanguinity was referred to the Creteil University Eye Department, Creteil, France. She presented with severe hemeralopia, associated with regressive bilateral macular edema under acetazolamide treatment. Best-corrected visual acuity was 20/32 on both eyes.

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Fundus examination showed white dots around the vascular arcades and no peripheral pigmentary changes.¹

Fundus autofluorescence (Spectralis, Heidelberg Engineering, Heidelberg, Germany) revealed a ring of hyperautofluorescence² (Figure 1).

Full-field International Society for Clinical Electrophysiology of Vision (ISCEV) Standard electroretinograms (ERG) (MonColor Metrovision, Pérénchies, France) (Supplemental Figure, <http://links.lww.com/IAE/B238>), had a typical aspect of ESCS.³

Genetic testing confirmed the diagnosis of ESCS with two compound heterozygous mutations in the NR2E3 gene: c.131C > A p.Ser 44* inherited from the healthy father and rs1484573200 c.639_640insT inherited from the healthy mother.⁴

Spectral domain OCT (Figure 1) showed hyperreflective, target-shaped lesions around the arcades, located in the outer nuclear layer, corresponding to rosette-like lesions, with loss of normal retinal lamination.⁵

En face 12- × 12-mm images (Plexelite 9000; Carl Zeiss Mediatec, Dublin, CA) showed numerous deposits surrounding the vascular arcades, well seen at the segmentation corresponding to the deep vascular complex (Figure 1), which encompasses the inner nuclear layer and outer nuclear layer. Using the embedded B-

scan, the lesions seen on the zoomed en face images (Figure 1) correspond in shape and location to the previously described OCT rosette-like lesions.⁵ Unlike the nummular pigment clumping, described at the level of the retinal pigment epithelium,⁶ the rosette-like lesions are located in the outer nuclear layer, with loss of normal retinal lamination.

Minnella et al⁷ described the en face OCT aspect of an ESCS patient with schisis. However, the 6- × 6-mm scanning area used by the authors did not capture the paramacular vascular arcades, where the rosette-like lesions are located. Hence, to the best of our knowledge, this is the first report of en face OCT imaging of ESCS with rosette-like lesions. This pattern of hyper-reflective lesions surrounding the paramacular vascular arcades in en face OCT may be a new diagnostic indicator of ESCS.

Key words: enhanced S-cone syndrome, en face OCT, NR2E3.

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