Enhanced Depth Imaging Spectral-Domain Optical Coherence Tomography Findings in Choroidal Neurofibromatosis

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ABSTRACT: The authors report multimodal imaging findings, including enhanced depth imaging-optical coherence tomography, in an affected child with choroidal neurofibromatosis. Novel features such as choroidal vessel compression from choroidal nodules related to neurofibromatosis type 1 and an increased subfoveal choroidal thickness are identified. This is the first report to use EDI-OCT to analyze choroidal features in neurofibromatosis type 1.

heads). However, no overlying retinal abnormalities were observed. The subfoveal choroidal thickness was 403 and 492 µm in the right and left eyes, respectively (Figures 2C-D).

DISCUSSION

The patchy NIR changes and corresponding ICG hypofluorescence — in conjunction with less obvious or negative findings with dilated fundus examination, FA, FAF, and red-free imaging — appear sensitive and specific for choroidal neurofibromatosis. For this reason, these multimodal imaging findings have been suggested as new diagnostic criteria for NF1 diagnosis. Our report confirms these features in an affected child. When analyzed with EDI-OCT, the high-signal NIR patches correspond to choroidal nodules that compress overlying choroidal vessels. The compression, while not significant enough to cause outer retinal thinning, may indicate ischemic choroidal foci visible with ICG angiography as punctate hypofluorescent regions. These nodules may correspond to ovoid bodies of proliferating, neoplastic Schwann cells arranged in concentric rings around axons or choroidal melanocytic proliferations. Interestingly, the subfo-
veal choroidal thickness in this patient is greater than the reported mean choroidal thickness in healthy children. Perhaps the posterior pole choroidal nodules contribute to this apparent difference. However, a larger EDI-OCT study comparing subfoveal and eccentric choroidal thicknesses of NF1-affected children to healthy controls would be needed to confirm whether there is an actual difference.

Combining EDI-OCT, ICG angiography, and microperimetry would provide a unique structural and functional tool to follow these nodules as they increase in size or number with time and potentially impair choroidal blood flow and compromise overlying retinal sensitivity. Together, these modalities pave the way toward a new understanding of the natural history of retinochoroidal neurofibromatosis. To our knowledge, this is the first report to use EDI-OCT to describe choroidal changes and subfoveal choroidal thickness in NF1.

REFERENCES