Neurofibromatosis type-1 with retinal microvascular corkscrew tortuosity

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Key words: Corkscrew vessels, neurofibromatosis type-1, retina, vascular tortuosity

A 40-year-old Caucasian woman presented with the history of neurofibromatosis type-1 (NF-1), diagnosed at age 5 years, with cutaneous neurofibromas on the face, back, and chest, and arachnoid cyst. Best-corrected visual acuity was 20/40 in the right eye and 20/20 in the left eye. Anterior segment examination revealed multifocal iris Lisch nodules in both eyes. Fundoscopic examination in the right eye revealed [Fig. 1a] 3 areas of retinal microvascular tortuosity, including superonasal to the optic disc (white arrows, 6.0 mm by 3.0 mm), in the papillomacular bundle (yellow arrows 1.5 mm by 1.0 mm), and inferior to the macula (blue arrows, 2.0 by 2.0 mm). In the

Figure 1: Fundoscopic examination in the right eye revealed (a) 3 areas of retinal microvascular tortuosity, including superonasal to the optic disc (white arrows, 6.0 mm by 3.0 mm), in the papillomacular bundle (yellow arrows 1.5 mm by 1.0 mm), and inferior to the macula (blue arrows, 2.0 by 2.0 mm). In the left eye, (b) there was 1 area of microvascular tortuosity in the papillomacular bundle (yellow arrows, 2.0 × 2.0 mm). Retinal microvascular tortuosity was confirmed on fundus autofluorescence imaging in the right (c) and left (d) eyes.

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Figure 3: Underlying the microvascular abnormalities were multiple hypopigmented choroidal lesions, suggestive of hamartoma, nevus, or neurofibroma. On OCT (vertical cut), these corresponded to [Figure 3] areas of increased choroidal density in the outer choroid (white arrow).

demonstrated retinal microvascular abnormalities (6/34) and choroidal nodules (23/34), while a larger cohort found no correlation of microvascular changes (18/294) with choroidal nodules (186/294) ($P = 0.48$). Our case exemplifies the subtle nature of microvascular abnormalities with underlying choroidal nodules in NF-1.

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Declaration of patient consent
The authors certify that they have obtained all appropriate patient consent forms. In the form the patient has given his consent for his images and other clinical information to be reported in the journal. The patient understands that his name and initials will not be published and due efforts will be made to conceal his identity, but anonymity cannot be guaranteed.

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Conflicts of interest
There are no conflicts of interest.

References