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

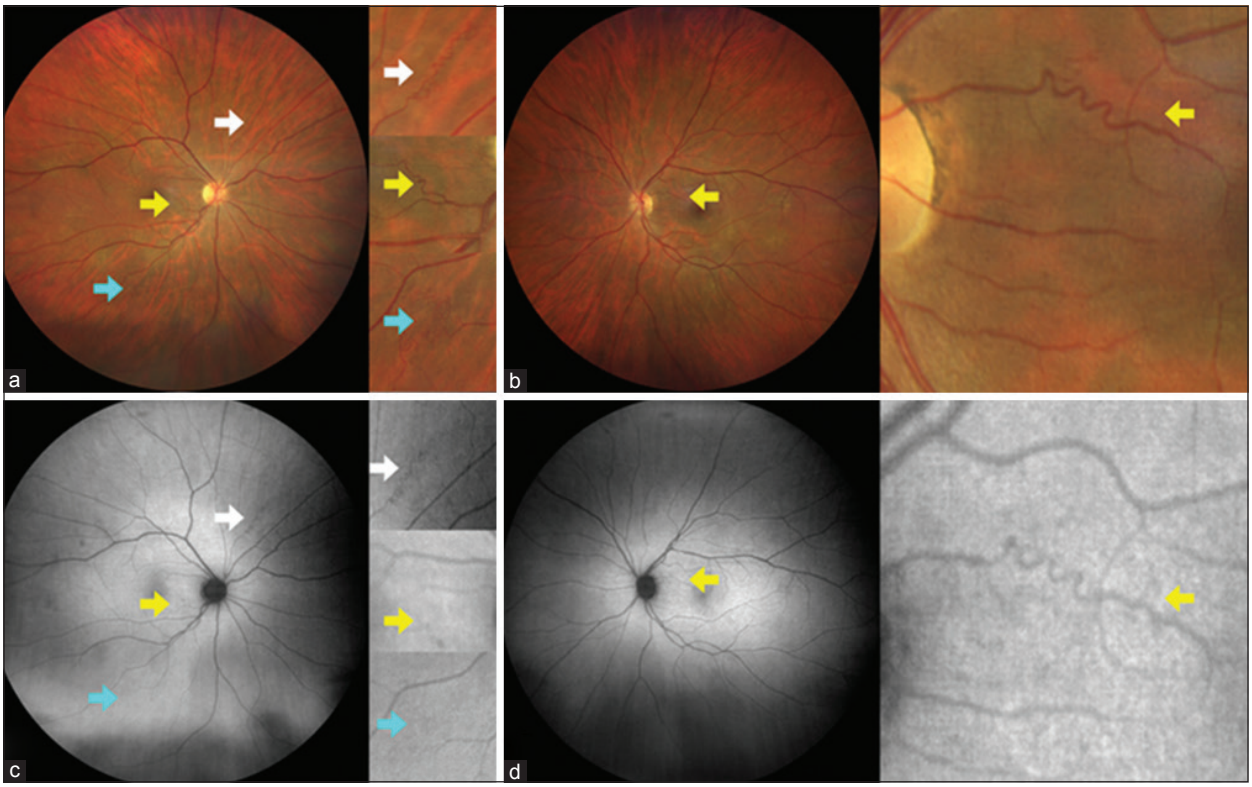

Figure 1: Fundoscopic examination in the right eye revealed (a) 3 areas of retinal microvascular tortuosity, including superonasal to the optic disc (white arrows, 6.0 mm by 3.0 mm), in the papillomacular bundle (yellow arrows 1.5 mm by 1.0 mm), and inferior to the macula (blue arrows, 2.0 by 2.0 mm). 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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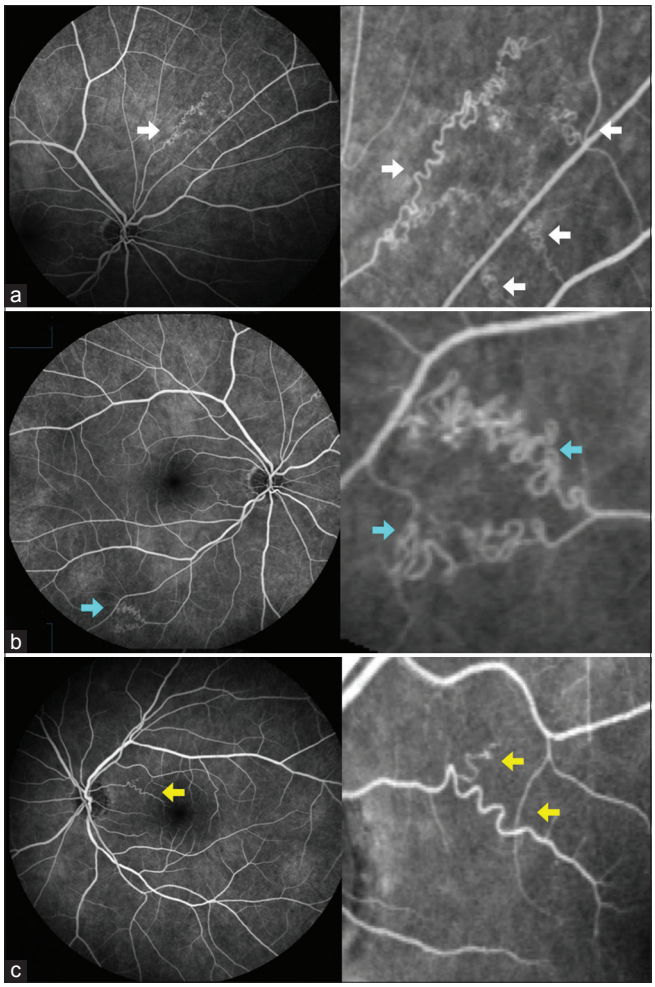

Figure 2: In the right eye, areas of retinal microvascular tortuosity were visible (A) superonasal to the optic disc (white arrows) and (B) inferior to the macula (blue arrows). (C) In the left eye, retinal microvascular tortuosity was observed in the papillomacular bundle (yellow arrows)

left eye [Fig. 1b], 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 [Fig. 1c] and left [Fig. 1d] eyes.

By fluorescein angiography, areas of retinal microvascular tortuosity were visible [Fig. 2] in each eye, without evidence of leakage, staining, or neovascularization.

Underlying the microvascular abnormalities were multiple hypopigmented choroidal lesions, suggestive of hamartoma, nevus, or neurofibroma. On OCT (vertical cut), these corresponded to [Fig. 3] areas of increased choroidal density in the outer choroid (white arrow). Based on the benign retinal and choroidal findings, observation was advised.

Retinal microvascular findings in NF-1 are likely congenital, representing defective migration of vasomotor nerve cells from the neural crest.[1-3] These benign abnormalities remain stable over time without retinal ischemia, exudation, hemorrhage, vascular leakage, or visual acuity loss.[1-4] An analysis of 34 NF-1 patients

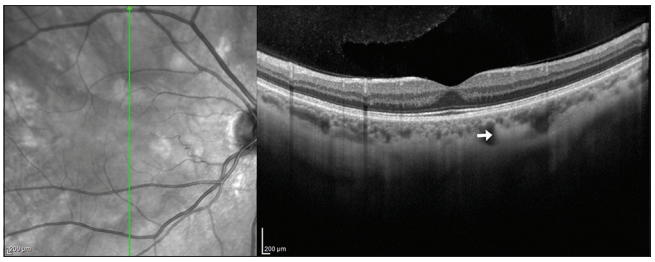

Figure 3: Underlying the microvascular abnormalities were multiple hypopigmented choroidal lesions, suggestive of hamartoma, nevus, or neurofibroma. On OCT (vertical cut), these corresponded to [Figure 3] areas of increased choroidal density in the outer choroid (white arrow)

demonstrated retinal microvasculature abnormalities (6/34) and choroidal nodules (23/34),[4] while a larger cohort found no correlation of microvascular changes (18/294) with choroidal nodules (186/294) (P = 0.48).[3] 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.

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