

Corkscrew Vessels and Hidden Clues: Retinal and Choroidal Manifestations of NF1

A 56-year-old woman with neurofibromatosis type 1 (NF1) was referred for possible uveitis regarding retinal abnormalities. Her visual acuity

was 20/20 bilaterally, with normal intraocular pressures. On slit-lamp examination, iris Lisch nodules were observed bilaterally, with no anterior chamber

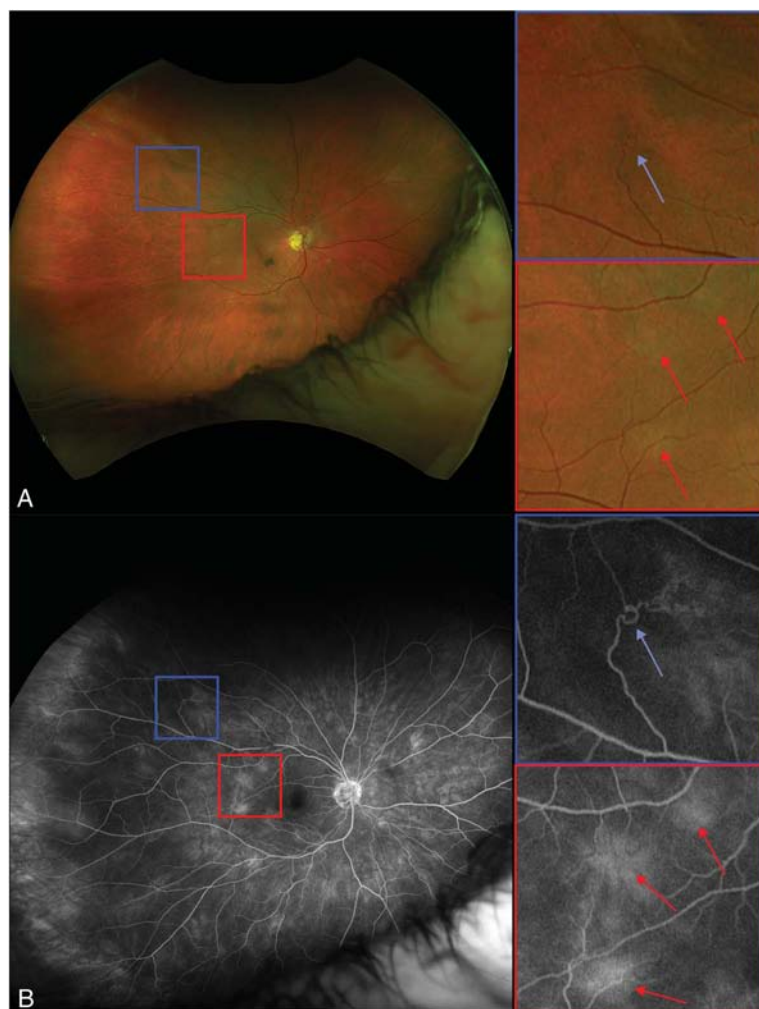


Fig. 1. A. Optos color fundus photography of the right eye (OD) shows a RVA inferiorly (indicated by the blue arrow) and yellow, nummular deep choroidal abnormalities (CA) (indicated by red arrows). B. Late-phase intravenous fluorescein angiography (IVFA, 4:07 minutes) OD reveals a corkscrew-shaped RVA superiorly (indicated by a blue arrow) and densely concentrated deep yellow nummular lesions in the posterior pole, with scattered anterior lesions (examples indicated by red arrows). Fundus autofluorescence and Optos Silverstone OCT revealed similar findings; however, because they were not particularly striking and would not add significant value, they were not included in this photo essay.

From the *Cumming School of Medicine, University of Calgary, Calgary, Alberta, Canada; and †Department of Surgery, Section of Ophthalmology, University of Calgary, Calgary, Alberta, Canada.

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Reprint requests: Monique Munro, MD, FRCSC, Department of Surgery, Section of Ophthalmology, University of Calgary, Calgary, Alberta, T2N1P5 Canada; e-mail: moniquepmunro@gmail.com

cells or vitritis. Fundoscopy revealed small, nummular, creamy white–yellow lesions in both eyes, suspected to be deep retinal or choroidal. Intravenous fluorescein angiography showed scattered nummular hyperfluorescent staining corresponding to the lesions on funduscopy. In addition, a well-defined, tortuous corkscrew retinal vascular abnormality

(RVA) was noted in the superior temporal and inferior retina of her right eye (Figure 1). Choroidal abnormalities (CAs) with pigmentation were noted in her left eye, although optical coherence tomography (OCT) cross-sections showed no obvious abnormalities.

This patient presented with several findings typical of NF1, although CAs were only recently documented in the literature.^{1,2} Histologically, CAs are multifocal aggregates of melanocyte clusters in the choroid, representing choroidal hamartomas, and analogous to Lisch nodules¹ which are long-recognized findings in NF1.² These regions of choroidal proliferation result in increased fluorescein uptake and hyperfluorescence appearing as bright, patchy nodules on spectral domain optical coherence tomography (SD-OCT) in near-infrared reflectance (NIR) due to hyperactive melanosomes or increased melanocyte density.² Although asymptomatic and usually undetectable by slit-lamp, the most pigmented and prominent CAs manifest as hyperpigmented spots on fundus examination.² CAs have recently been incorporated into the revised criteria for NF1.³ For individuals without a parent diagnosed with NF1, two or more Lisch nodules or CAs on OCT or NIR now fulfill part of the diagnostic criteria.³

Another retinal finding in NF1 is RVAs; small, well-defined, tortuous retinal vessels categorized as simple, corkscrew, and moyamoya-like arrangements.⁴ A potential link between CAs and RVAs may involve blood flow redistribution due to CA compression.²

Key words: choroidal abnormalities, color fundus photography, corkscrew retinal vascular anomaly, fundus autofluorescence, hyperpigmented spots, intravenous fluorescein angiography, neurofibromatosis.

LUCY YANG, BHSc*
ABDULLAH AL-ANI, MD, PhD†
MONIQUE MUNRO, MD, FRCSC†

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