A Case of a 65-Year-Old Female Complaining of a "Smudge" in her Left Eye

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Case Presentation:

A 65-year-old female presented with complaint of gradual development of a central "smudge" in the visual field of her left eye. Her past medical history included medically managed hypertension and hyperlipidemia. Her past ocular history included mild cataracts and she reported no family ocular history.

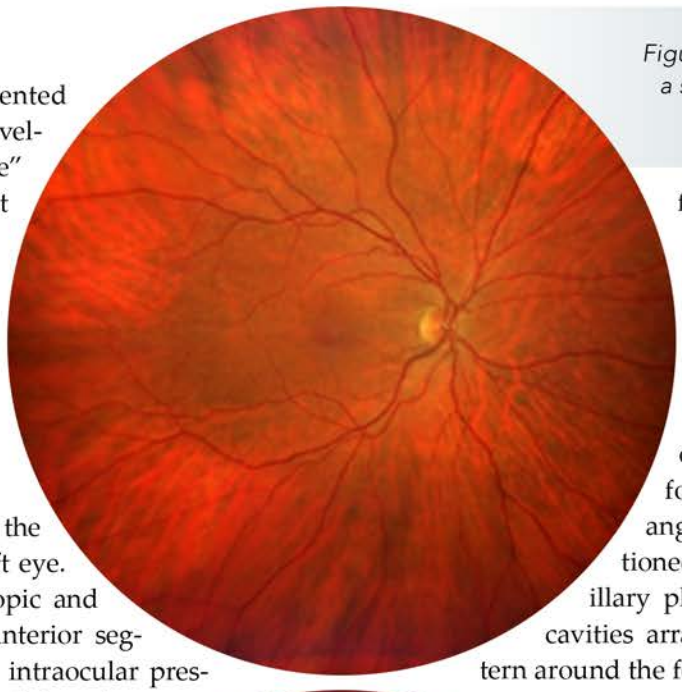
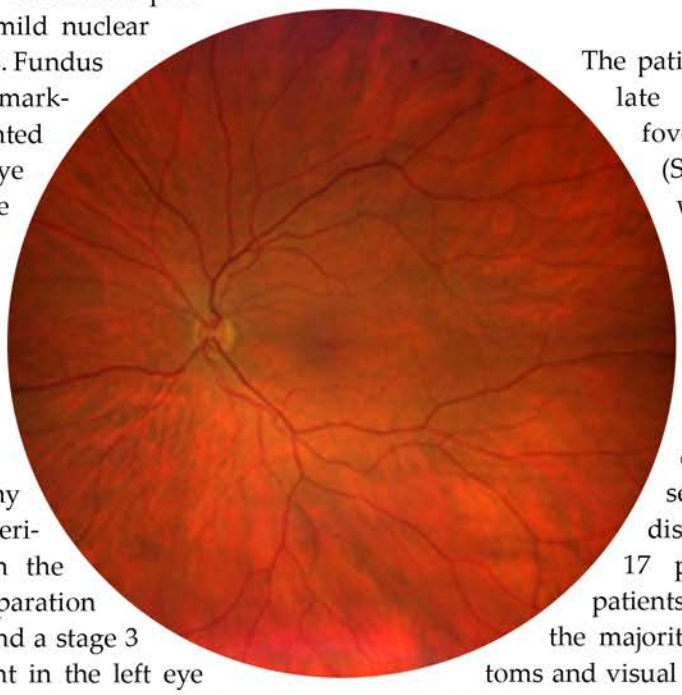


Figure 1: Fundus photos reveal a slight blunting of the foveal reflex in the left eye.

Visual acuity was 20/20 in the right eye and 20/30 in the left eye. She was essentially emmetropic and wore no corrective lenses. Anterior segment examination, including intraocular pressure, was notable only for mild nuclear sclerotic cataracts in both eyes. Fundus examination was largely unremarkable, with a moderately blunted foveal reflex in the left eye (Figure 1). Pertinent negative findings on fundus examination included a lack of vitritis, optic nerve cupping, vitreoretinal interface anomalies, pigmentary changes, and peripheral retinoschisis.



Optical coherence tomography (OCT) revealed a stage 1 posterior vitreous detachments in the right eye (vitreous face separation from the temporal macula) and a stage 3 posterior vitreous detachment in the left eye (vitreous face separation from the entire macula with persistent adhesion to the optic nerve head).[1] The foveal contour of the right eye was unremarkable but OCT of the left macula displayed notable retinoschisis (Figure 2). The retinoschisis localized to the outer plexiform layer and presented in a circular pattern involving the fovea and parafovea. OCT angiography of the left eye, sectioned at the level of the deep capillary plexus, highlighted the schisis cavities arranged in radial spoking pattern around the fovea (Figure 3).

The patient was diagnosed with stellate nonhereditary idiopathic foveomacular retinoschisis (SNIFR) and close observation was planned.

Discussion:

SNIFR was first described by Ober et al. in 2014.[2] Twenty-two eyes of 17 patients were described in the initial case series establishing SNIFR as a distinct clinical entity. Sixteen of 17 patients were female, five patients had bilateral findings, and the majority had minimal or no symptoms and visual acuity of 20/40 or better. Two patients in the series demonstrated concurrent peripheral retinoschisis.

To arrive at a diagnosis of SNIFR a host of other etiologies must be considered, rendering SNIFR a proverbial diagnosis of exclusion. Common etiologies to exclude include edema secondary to vasculopathy or inflammation as well as tractional forces secondary to epiretinal membrane or pathologic myopia.

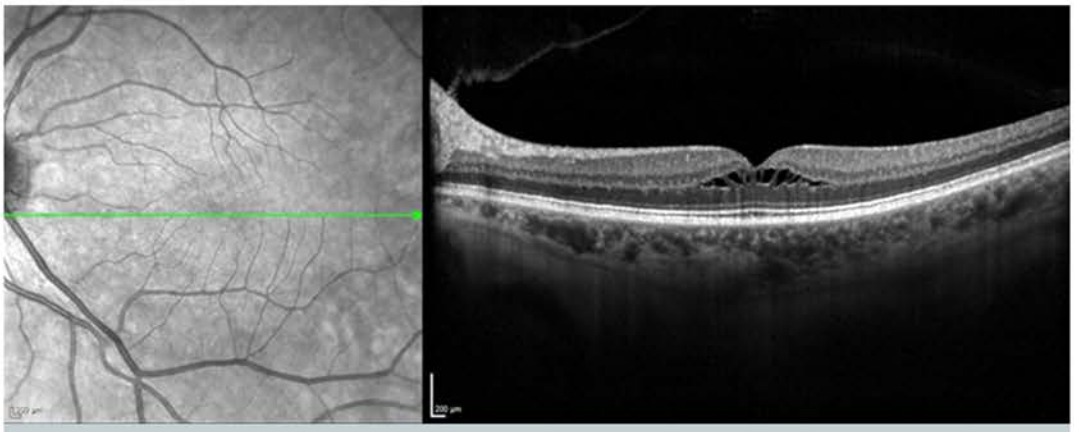


Figure 2: OCT of the left eye demonstrates foveomacular retinoschisis involving the outer plexiform layer.

However, other less common etiologies of foveomacular retinoschisis must also be considered in a patient with potential SNIFR. Juvenile X-linked retinoschisis is classically linked to foveomacular retinoschisis by way of a mutation of the RS1 gene linked to the Xp22.1-3 locus. Given its genetic locus X-linked retinoschisis is found almost exclusively in males, as opposed to the strong female predilection heretofore reported in SNIFR. No patients in the initial SNIFR case series tested positive for known X-linked retinoschisis mutations.[2] A recent OCTA study comparing SNIFR to X-linked retinoschisis noted that the schisis cavity in SNIFR is avascular (Figure 3) whereas vascular structures—with flow on OCTA—are present in X-linked retinoschisis.[3]

Optic pic maculopathy is another condition that may result in foveomacular retinoschisis. Optic pits, either congenital or acquired due to glaucomatous nerve damage, are usually visible on careful biomicroscopy or OCT. Enhanced S-cone syndrome can also exhibit as schisis pattern similar to SNIFR, but the concomitant ocular findings in enhanced S-cone syndrome—night blindness, optically empty vitreous, and peripheral pigmentary changes—can often starkly distinguish the condition from SNIFR.[2]

Medication review is also critically important for patients presenting with foveomacular retinoschisis. Niacin and taxanes often result in cystic changes in the outer plexiform layer as in SNIFR. These changes are often reversible with discontinuation of these medications. Familial internal limited membrane (ILM) dystrophy, also known as Muller cell sheen dystrophy, is another consideration but is often apparent by ILM appearance on exam and strong family history.

Most patients with SNIFR enjoy good outcomes, though currently no long-term outcome studies exist. In the initial series one patient developed poor vision secondary to development of subfoveal fluid; the rest retained good visual acuity.[2]

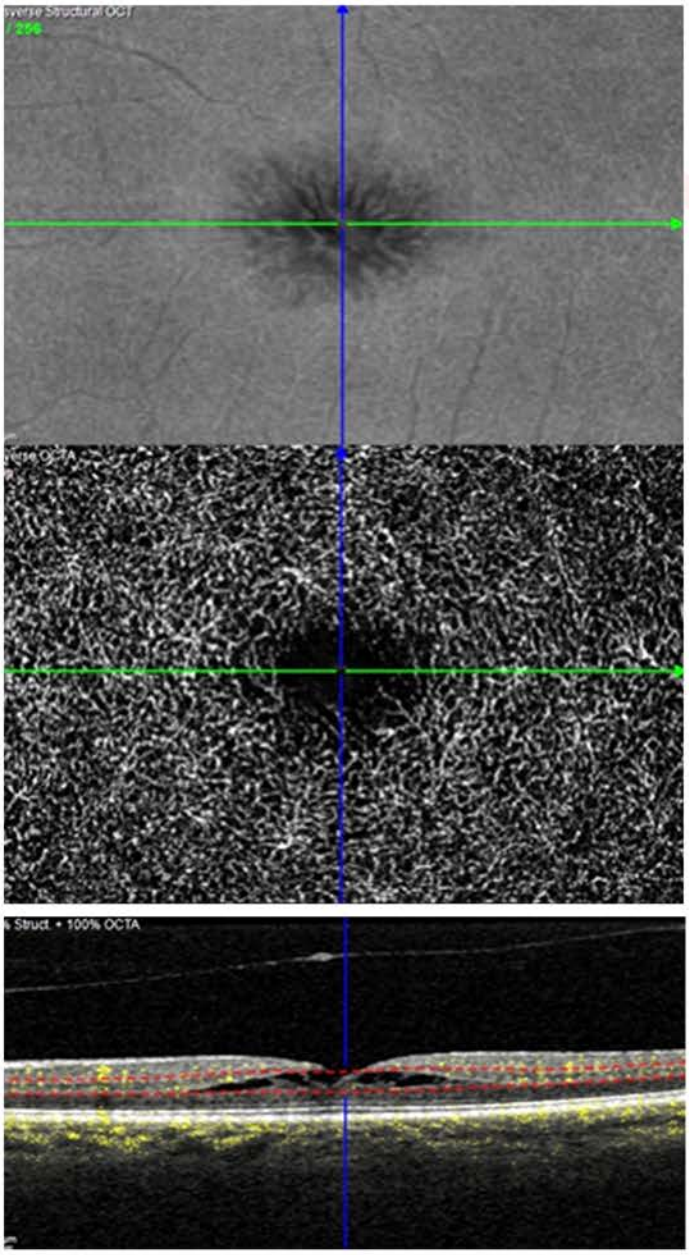
In the emerging literature on SNIFR case reports topical dorzolamide therapy are promising. Ajlan and Hammamji reported visual and anatomic improvement in a case a SNIFR.[4] On discontinuation of dorzolamide the benefits regressed, only to return when dorzolamide was restarted. Moraes et al. reported a positive outcome in a SNIFR patient that developed an outer retinal hole and underwent vitrectomy with ILM peeling.[5] Anatomy and vision improved post-operatively.

SNIFR will no doubt attract further attention as we seek to better understand this recently described clinical entity. In the meantime, it behooves all clinicians to know well the common and uncommon conditions on the differential diagnosis for foveomacular retinoschisis.

References:

1. Uchino E, Uemura A, Ohba N. Initial stages of posterior vitreous detachment in healthy eyes of older persons evaluated by optical coherence tomography. Arch Ophthalmol. 2001. 119:1475-1479.

2. Ober MD, Freund KB, Shah M, et al. Stellate nonhereditary idiopathic foveomacular retinoschisis. 2. Ophthalmology. 2014. 121:1406-1413.



References (cont.):

3. Fragiotta S, Leong BCS, Kaden TR et al. A proposed mechanism influencing structural patterns in X-linked retinoschisis and stellate nonhereditary idiopathic foveomacular retinoschisis. Eye. 2019. 33:724-728.

4. Ajlan RS, Hammamji KS. Stellate nonhereditary idiopathic foveomacular retinoschisis: response to topical dorzolamide therapy. Retinal Cases & Brief Reports. 2017. 0:1-3.

5. Moraes BR, Ferreira BF, Nogueira TM. Vitrectomy for stellate nonhereditary idiopathic foveomacular retinoschisis associated with outer retinal layer defect. Retinal Cases & Brief Reports. 2020. 0:1-4.

Figure 3: OCTA of the left eye at the level of the deep capillary plexus. A radial pattern of cystic spaces centered upon the fovea is evident, along with an absence of flow in the area of retinoschisis.

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