Multimodal imaging in a case of stellate nonhereditary idiopathic foveomacular retinoschisis

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Key words: Cystoid macular dystrophy, foveal schisis, outer plexiform layer splitting, retinal schisis, stellate foveomacular schisis

Stellate nonhereditary idiopathic foveomacular retinoschisis (SNIFR) is an uncommon cause of foveomacular retinoschisis with patients being asymptomatic or minimally symptomatic having good visual acuity. Most of the patients have unilateral disease without any genetic predisposition to retinoschisis, and they exhibit some degree of myopia. With characteristic stellate appearing macula, optical coherence tomography (OCT) shows splitting of outer plexiform layer (OPL).[1] It is imperative to rule out acquired and genetic causes of splitting like myopic traction maculopathy, epiretinal membrane, vitreoretinal traction, optic or scleral pit, or glaucomatous optic nerve changes.

A 64-year-old female was referred to retina clinic for internal limiting membrane (ILM) folds. She was a known diabetic, on hydrochloroquine (HCQ) 200 mg/day for 3 years for rheumatoid arthritis and post COVID-19 vaccination 1 month prior with no family history of retinal pathology. Visual acuity was 6/6 with refraction of +0.75D × 165 degrees cylinder in the right eye and 6/9 with -0.5D × 105 degrees in the left eye. Anterior segment was normal except for early cataractous changes. Fundus examination showed fine ILM wrinkling at fovea. [Fig. 1a and b] Both eyes OCT showed splitting at the level of OPL at fovea, rest of the retinal layers were intact. [Fig. 1e and f] Fundus fluorescein angiography in both eyes was normal with no foveal, vascular, or disc leak. [Fig. 1c and d] Three-dimensional topographic density plots on multifocal electroretinographic (ERG) showed blunted foveal peaks, and trace arrays showed diminished amplitudes in the central area in both eyes. [Fig. 2a and b] Full-field ERG showed reduced amplitude of scotopic b wave with a b/a ratio of 1.2 in the right eye and 1.1 in the left eye [Fig. 2c and d]. Photopic ERG was unaffected [Fig. 2e and f]. A thorough clinical examination, Optos and wide field OCT, did not reveal any peripheral retinoschisis. OCT-angiography of the right eye showed the presence of flow signals in the bridging septae of the split layers; however, flow signals were scarce in the bridging tissues in the left eye. [Fig. 1g and h] Pedigree analysis revealed that she was the sole affected member. She was born out of a consanguineous marriage, and her father is deceased. Detailed fundus examination and OCT of her son did not show any signs of foveal or peripheral retinoschisis. A diagnosis of SNIFR was made.

Discussion

Ober et al.[1] reported the largest known series of SNIFR, which included 22 eyes from 16 female and 1 male patient with foveal retinoschisis without a known hereditary or acquired predisposition. Gene testing was done in eight patients and was negative for defects in RS1 gene. In total, 12 cases were unilateral and 5 were bilateral. Out of the total eyes, 16 eyes were myopic, 3 plano, and 2 hyperopic. Three eyes showed bullous peripheral schisis, while 3 had subclinical peripheral retinoschisis.

Javaheri et al.[2] reported a case of SNIFR with atypical peripapillary multilayered retinoschisis in a woman. Mandell et al.[3] described a patient whose extensive peripheral disease and diagnosis of SNIFR were fully revealed only through widefield OCT and scotopic ERG, which showed significantly reduced b/a wave ratio. Ahmed et al.[4] reported a case of a woman with stellate foveal splitting of OPL in the right eye and bilateral peripheral OPL splitting demonstrated on widefield OCT. Genetic testing for X-linked retinoschisis (XLRS1) was negative.

Serena Fragiotto et al.[5] compared structural patterns on OCT-angiography in XLRS with SNIFR. In XLRS patient, it showed flow signal representing bridging vessels between intermediate and deep capillary plexuses in the connecting tissue within the inner nuclear layer. In SNIFR, the flow signal superimposed on the structural slab demonstrated the absence of flow signals within retinal tissue separating cystic spaces.

There is low-level evidence of response to dorzolamide in SNIFR. Ajlan et al. reported good response to topical dorzolamide in a 27-year-old male.[6] Machado Nogueira et al.[7] reported complete resolution of SNIFR cavities with spontaneous vitreomacular adhesion release.

XLRS, being a close differential, where all affected individuals manifest foveoschisis with approximately half of them also develop peripheral schisis. Typically, first identified in school-aged boys, the characteristic “spoke-wheel” appearance seen at fovea is caused by cystic changes, which is commonly replaced with nonspecific macular atrophy in middle age with an electronegative ERG. XLRS in females has.

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Figure 1: (a and b) Fundus photo showing normal fundi except subtle ILM wrinkling at fovea (yellow circles). (c and d) Fundus fluorescein angiography showed no leakage at fovea. (e and f) OCT B scan revealing splitting at OPL. Rest layers are intact. (g) OCT-angiography showing the presence of flow signals in bridging septae in the right eye, while panel (h) shows scarcity of the same in the left eye.

Figure 2: (a and b) Multifocal ERG shows diminished amplitudes on trace array and blunted foveal peaks on topographic density plots. (c and d) Scotopic full-field ERG shows reduced amplitude of b wave in both eyes. (e and f) Photopic ERG was unaffected.
been reported to be rare. Although we had least suspicion of XLRS, we suggested the patient to have genetic testing for RS-1 gene mutations which she refused as she did not have any visual complaints. Another close differential is familial foveal retinoschisis (FFR), an autosomal recessive disease presenting in first two decades of life with mild–moderate visual loss. It shows characteristic fovea-centered cart-wheel pattern with no peripheral involvement. OCT shows splitting at outer and inner nuclear layers. Vincent et al. reported CRB1 gene mutation as an underlying cause of FFR and suggested that this forms the mildest end of the spectrum of CRB-1-related diseases with possibility of some of SNIFR cases harboring mutations in CRB1. Kabanarou et al. introduced the term isolated foveal retinoschisis to accommodate both sporadic and familial cases of the disorder.

Earlier stages of dominant cystoid macular dystrophy may mimic SNIFR, which is diagnosed in the first or second decade of life followed by collapse of cystic spaces and chorioretinal atrophy in later stages. Development of cystic-appearing macular changes in SNIFR is postulated to be related to disruption of retinal architecture by defects in cell-to-cell adhesions. The absence of macular leakage on fluorescein angiography suggests that vascular leakage plays a minor role, if any, in its development. Drug toxicities with niacin or taxane can present with foveal splitting as in our case, and they are usually reversible. Cystoid macular edema with HCQ has rarely been described with both leaking and nonleaking types on angiography. To conclude, we describe utility of multimodal imaging to diagnose and characterize SNIFR features.

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

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Conflicts of interest
There are no conflicts of interest.

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