Goldmann-Favre syndrome is an autosomal recessive hereditary vitreo-retinal degeneration characterized by impaired visual acuity, nyctalopia, vitreous degeneration, atypical peripheral pigmentary dystrophy, and peripheral and macular retinoschisis [1-3]. Typically, macular retinoschisis has a characteristic microcystic appearance on ophthalmoscopy, even without fluorescein staining on angiography [4]. Macular retinoschisis is allelic with enhanced S cone syndrome. Both conditions are caused by mutations in the nuclear receptor gene NR2E3 on chromosome 15 [5]. Manifestation of the mutations, however, may not be detected in all cases.

**Case Reports**

**Case 1**

A 21-year-old male presented with a progressive decrease in vision that had begun at the age of 8 years. His brother was known to have the same problem. The patient’s best corrected visual acuity, with +4.00 DS / -0.50 DC × 180, was counting fingers at 50 cm in the right eye, and, with +1.00 DS / -1.50 DC × 20, was 6 / 24 in the left eye. The anterior segment examination was unremarkable. Indirect ophthalmoscopy revealed multiple vitreous membranes, peripheral vitreous detachment, peripheral retinal epithelial alterations, pigment clumping, equatorial chorioretinal atrophy inferior to the macula, lamellar macular holes, and foveal microcystic spaces in the patient’s right eye. Lattice degeneration at the 6 o’clock position was also observed in the left eye. A diagnosis of Goldmann-Favre syndrome was made.

Time domain (Stratus) optical coherence tomography demonstrated confluent macular cystoid changes, as well as foveal retinoschisis in both eyes, with macular holes in the right eye. During electroretinogram evaluation, isolated rod responses were unrecordable for the right eye, and showed delayed implicit time with grossly reduced amplitude in the left eye. Maximal combined responses showed a negative b-wave. Oscillatory potentials were flat in both eyes. Isolated cone responses showed delayed implicit time with reduced amplitudes in both eyes. There

**Morphological and Functional Correlates in Goldmann-Favre Syndrome: A Case Series**

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The purpose of this study is to describe the correlation of findings between results from spectral domain optical coherence tomography (SD-OCT) and microperimetry in a case series regarding patients with Goldmann-Favre syndrome. Goldmann-Favre syndrome is a rare autosomal recessive hereditary vitreo-retinal degeneration that impacts the functionality of vision in subjects. Three men with this condition were assessed and subjected to microperimetry and SD-OCT. Two of the men were brothers. This study finds that the retinoschisis and macular cystoid changes noted in the SD-OCT matched the scotomas revealed by the microperimetry. The findings of each of the individual cases are reported herein.

**Key Words:** Cystoid macular edema, Microperimeter, Retinoschisis, Spectral domain optical coherence tomography

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was a gross delay in implicit time in both eyes with 30 Hz flicker. Fundus fluorescein angiography (FFA) revealed window defects corresponding to areas of atrophy of the retinal pigment epithelium (RPE). The patient underwent laser photoagulation to the lattice in his left eye.

The patient returned for a follow-up visit 18 months later. His visual acuity was stable. Cataract formation was noted in his right eye. He had developed a bicycle wheel pattern of foveal schisis in his left eye. Imaging with spectral domain optical coherence tomography (SD-OCT; Copernicus, Optopol Technologies, Zawierci, Poland) of the right eye revealed lamellar macular holes with macular schisis, microcystic spaces, and vitreomacular traction. SD-OCT imaging of the left eye revealed cystoid macular edema with inner layer schisis. Foveal thickness was 77 microns in the right eye and 672 microns in the left eye, with a central dense scotoma in both eyes (Fig. 1).

Pedigree construction and venous blood sampling was done for cytogenetic analysis. The chromosomes were stained by Giemsa-trypsin banding and scanned using IKAROS software (MetaSystems, Altltussheim, Germany). The pedigree showed an autosomal recessive inheritance pattern with one additional affected male in the family. Twenty-five plates, which were screened for the proband, revealed normal karyotypes. However, genetic analysis could not be performed.

Case 2

A 41-year-old male presented with decreased vision, haloes, and visual distortion that he had been experiencing for the past 13 years. His brother (case 3) was known to have the same problems. The patient’s best corrected visual acuity, with +0.50 DS / -4.50 DC ×100, was 6 / 36 in the right eye, and, with +0.50 DS / -4.00 DC ×80, was 6 / 15 in the left eye. Anterior segment examination was normal. Indirect ophthalmoscopy revealed vitreous floaters, macular schisis, and diffuse RPE alterations in both eyes. Also revealed was a peripheral hole in the inferotemporal quadrant in the left eye.

The patient’s color vision was analyzed with the Farnsworth D-15 test [6]. The total error score for the Farnsworth test was 36 in the right eye and 19 in the left eye. The test results revealed tritanomaly in the right eye and diffuse color defect in the left eye. SD-OCT imaging of both eyes showed incomplete posterior vitreous detachment, elevated foveal contours, and foveal schisis. It also showed alteration of the photoreceptor layer. Foveal thickness was 292 microns in the right eye and 490 microns in the left eye. Microperimetry showed a reduced mean retinal sensitivity of 7.7 db in the right eye and 8.2 db in the left eye, with a central dense scotoma in both eyes (Fig. 2).

Case 3

A 36-year-old male presented with decreased vision, haloes, and visual distortion that he had been experienc-