European Journal of Ophthalmology / Vol. 13 no. 3, 2003 / pp. 307-310

SHORT COMMUNICATION

Case report

Goldmann-Favre vitreoretinal degeneration

F. BATIOĞLU

Department of Vitreoretinal, Eye Clinic, Ankara University, Faculty of Medicine, Ankara - Turkey

PURPOSE. *To describe a case of Goldmann-Favre vitreoretinal degeneration with typical clinical findings.*

METHODS. *The case report of a healthy 47-year-old woman with typical clinical findings of Goldmann-Favre vitreoretinal degeneration is presented. She had complaints of reduced visual acuity and night blindness. Her parents were first cousins.*

RESULTS. *Fundus examination revealed annular pigmentary degenerative changes, macular edema, and peripheral retinoschisis in both eyes. Electroretinography results were abnormal. Optical coherence tomography scans revealed hyporeflective spaces in the macular area and irregularities of the chorioretinal complex in the degenerative pigmentary areas.*

CONCLUSIONS. *Typical fundus findings combined with night blindness and electroretinogram abnormalities permited the diagnosis of Goldmann-Favre vitreoretinal degeneration. (Eur J Ophthalmol 2003; 13: 307-10)*

KEY WORDS. *Goldmann-Favre syndrome, Vitreoretinal degeneration, Night blindness, Optical coherence tomography*

Accepted: October 7, 2002

INTRODUCTION

Goldmann-Favre syndrome is a rare autosomal recessive hereditary vitreoretinal degeneration (1) characterized by pigmentary degeneration, macular and peripheral retinoschisis, posterior subcapsular cataract, and degenerative vitreous changes (2). Reduced or nonrecordable electroretinogram (ERG), abnormal dark adaptation, and progressive loss of peripheral visual field are associated findings.

We describe a case with typical clinical features of Goldmann-Favre syndrome and report the findings of different diagnostic tests.

Case report

A healthy 47-year-old woman complained of progressive visual loss since 1990 and night blindness since childhood. Her mother and father were first cousins. Visual acuity of the right eye was counting fingers at 2 m. She had had cataract surgery in this eye in 1999. Visual acuity of the left eye was 0.2 with +1.50 diopter correction. Intraocular pressure was 16 mm Hg in both eyes. Extraocular motility and anterior segment including conjunctiva, cornea, anterior chamber, and iris were normal. She had a posterior chamber intraocular lens in the right eye. In her left eye there was a slight degree of posterior subcapsular cataract.

Fundus examination revealed annular pigment epithelial changes in the midperiphery with pigment clumping and yellow-white areas more peripherally and edematous changes in both maculas (Fig. 1). Peripheral retinoschisis was observed in the inferotemporal quadrant of both eyes. There was a large retinal hole in the right eye (Fig. 2). Optic discs were slightly pale, retinal vessels were thin, and areas of vascular occlusion were observed especially in the left peripheral retina (Fig. 3). Fluorescein angiography detected an annular area of mild retinal staining surrounding the center of the fovea in both eyes. Because of the pig-

 1120-6721/307-04$02.00/0

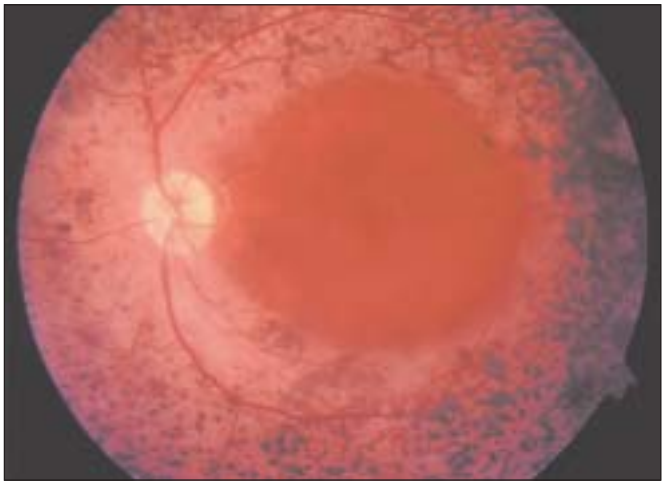

Fig. 1 - *Macular edema and annular pigmentary degeneration in the left eye.*

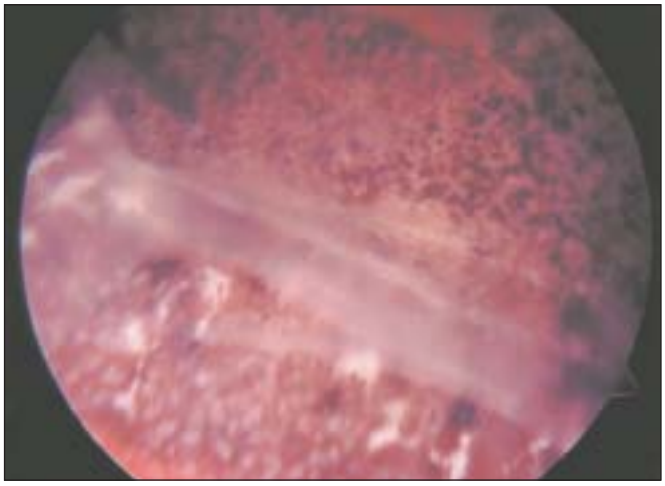

Fig. 2 - *Peripheral retinoschisis and a large retinal hole in the inferotemporal quadrant of the right eye.*

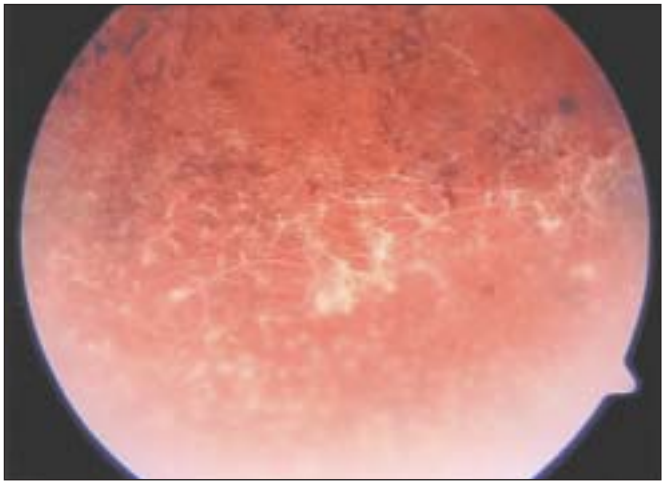

Fig. 3 - *Dendritic appearance of inferior peripheral vessels in the left eye.*

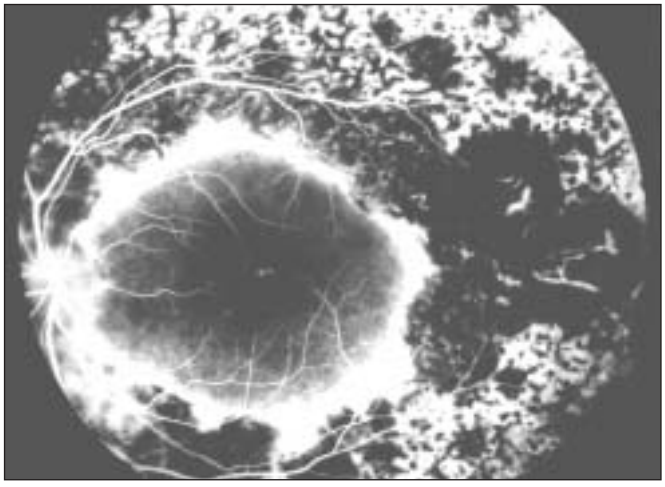

Fig. 4 - *Annular choroidal transmission defects with hypofluorescence of hyperpigmentation and mild macular staining on fluorescein angiogram.*

mentary retinopathy, there were irregular choroidal transmission defects in the midperiphery (Fig. 4).

Optical coherence tomography (OCT) was done through both macula and temporal periphery, horizontal and vertical cross-section images were taken. Tomograms through the fovea of both eyes showed hyporeflective areas. Vertical cross-sections from degenerative pigmentary areas showed irregularities in the chorioretinal layers (Figs. 5a and b).

The ERG gave cone responses that were both reduced in amplitude and prolonged in implicit time. After a 20-minute period of dark adaptation there was little change from photopic values in either amplitudes or implicit times in either eye, reflecting an apparent decrease in rod activity.

Visual field examination with the central 30-2 threshold test on a Humphrey field analyzer showed annular scotoma in the left eye. The patient was unable to give reliable answers in the right eye.

DISCUSSION

Goldmann-Favre syndrome was first reported in two siblings with a vitreoretinal degenerative disorder by Goldmann in 1957 and Favre in 1958 (1). Ricci later

described a similar case in a 41-year-old woman, and suggested an autosomal recessive pattern of inheritance (3). In later reports, various clinical features of Goldmann-Favre syndrome were described (1, 4, 5).

The classical symptoms are progressively declining visual acuity and night blindness since childhood (6). Our patient had noticed a gradual loss of vision for ten years, and had had night blindness since she was ten years old. She had been operated for cataract in her right eye 2.5 years before presentation and had a posterior chamber intraocular lens.

The clinical features of Goldmann-Favre syndrome are typically bilateral and symmetric. Vitreous changes included liquefaction and fibrillary degeneration. Retinal features are peripheral pigmentary changes including round dots and clumps of hyperpigmentation. Other fundus features are macular and peripheral retinoschisis, cystoid macular edema and, in the later stage, vascular abnormalities with optic disc pallor (1). In our case, typical fundus findings with slightly pale optic discs were present.

Both central and peripheral retinoschisis are present in Goldmann-Favre syndrome. Although foveal retinoschisis is thought to be pathognomonic for idiopathic juvenile X-linked retinoschisis, it may be seen as a manifestation of Goldmann-Favre syndrome (7). In a case with juvenile X-linked retinoschisis, histopathologic study showed a splitting of the nerve fiber layer (8). This may explain the normal results on fluorescein angiography of the macula usually seen in these individuals (7). However, some authors have reported fluorescein leakage in the macula (4). Our case had macular edema with mild staining on fluorescein angiography.

Peripheral retinoschisis in Goldmann-Favre syndrome is most common in the temporal periphery, especially the inferotemporal quadrant. There may be large retinal breaks in the inner wall of the schisis cavity. In our case, peripheral schisis was observed in the inferotemporal quadrant of both eyes, with a large retinal hole in the right eye. Recently, Theodossiadis et al reported an OCT study of Goldmann-Favre syndrome (9) where confluent macular cystoid changes and retinoschisis were found in both eyes. In the left eye there was a lamellar macular hole and retinoschisis, with inner retinal layer and outer retinal layer holes. In our case, hyporeflective areas were evident on macular OCT scans. Cross-sections obtained from the periphery of the retinoschisis showed irregularities in the chorioretinal layers. The retina was thinner at the far periphery.

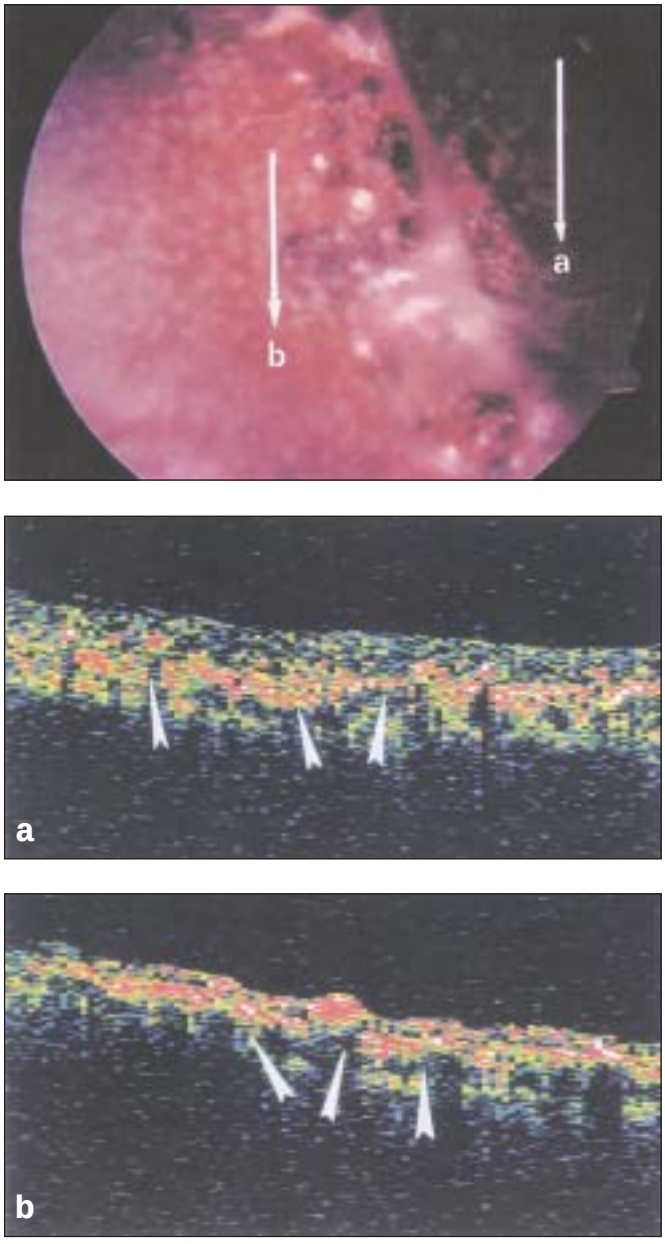

Fig. 5 - *The fundus photograph shows peripheral pigmentary degeneration in the right eye. The vertical white arrows indicate the scanning line of optical coherence tomography (OCT).*
a, b - *OCT scans through degenerative areas show irregularities in the chorioretinal layers (white arrowheads). The retina is thinner in the cross-sectional image taken from the periphery of retinoschisis (**b**).*

Retinal vascular changes range from attenuation of vessels in normal-appearing retina to obliteration of vessels in the area of peripheral retinoschisis.

Opaque, dendritic appearance of peripheral vessels was obvious, especially in the left eye of our case. Although previous reports of fluorescein angiographic findings in a patient with Goldmann-Favre syndrome did not mention leakage from or non-perfusion of retinal vessels (10), Fishman et al found diffuse vascular leakage in their patients, which was unique among retinal dystrophies (4).

Several examinations, including ERG, dark adaptation, visual fields, and color vision, show abnormalities in Goldmann-Favre syndrome (1, 4). The ERG is either reduced or nonrecordable with rod function affected earlier than cone function. Dark adaptation is always abnormal. The visual fields present central and peripheral defects depending on the fundus abnormalities. In our patient these tests gave abnormal findings. There was extensive rod and cone dysfunction as well as visual field constriction, in accordance with degenerative pigmentary changes.

A diagnosis of Goldmann-Favre syndrome should be considered in patients with an early history of poor night vision, atypical peripheral pigmentary changes, central and peripheral retinoschisis, complicated cataract at an early age, degenerative changes in the vitreous, and a markedly abnormal (often nonrecordable) ERG. Other tests, such as visual field examination, fluorescein angiography, and OCT, add some information which may usefully be assessed with the fundus findings.

Reprint requests to:
Prof. Figen Batıoğlu
Güzaltan sok. 22/6
06570 Maltepe
Ankara, Turkey
fbatioglu@hotmail.com

REFERENCES

1. Nasr YG, Cherfan GM, Michels RG, Wilkinson CP. Goldmann-Favre maculopathy. Retina 1990; 10: 178-80.
2. Gass JDM. Stereoscopic atlas of macular diseases diagnosis and treatment. 4th ed. St Louis: CV Mosby, 1997: 372-6.
3. Ricci A. Clinique et transmission génétique des differentes formes de degénerescences vitreo-rétiniennes. Ophthalmologica 1960; 139: 338-43.
4. Fishman GA, Jampol LM, Goldberg MF. Diagnostic features of the Favre-Goldmann syndrome. Br J Ophthalmol 1976; 60: 345-53.
5. Peyman GA, Fishman GA, Sanders DR, Vichek J. Histopathology of Goldmann-Favre syndrome obtained by full-thickness eye-wall biopsy. Ann Ophthalmol 1977; 9: 479-84.
6. Green JL Jr, Jampol LM. Vascular opacification and leakage in X-linked (juvenile) retinoschisis. Br J Ophthalmol 1979; 63: 368-73.
7. Noble KG, Carr RE, Siegel IM. Familial foveal retinoschisis associated with a rod-cone dystrophy. Am J Ophthalmol 1978; 85: 551-7.
8. Yanoff M, Kertesz Rahn E, Zimmerman LE. Histopathology of juvenile retinoschisis. Arch Ophthalmol 1968; 79: 49-53.
9. Theodossiadis PG, Koutsandrea C, Kollia AC. Optical coherence tomography in the study of the Goldmann-Favre Syndrome. Am J Ophthalmol 2000; 129: 542-4.
10. MacVicar J, Wilbrandt H. Hereditary retinoschisis and early hemeralopia. A report of two cases. Arch Ophthalmol 1970; 83: 629-36.