Case report

Bilateral giant macular schisis in a case of Goldmann-Favre syndrome

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SUMMARY

We describe a case of a 23-year-old man with giant macular schisis which can be seen in Goldmann-Favre syndrome. Associated history of decreased night vision and an enhanced S cone electroretinogram helps in confirming the diagnosis. Swept source optical coherence tomography of the same helps in confirming the schisis and delineates the extent and level of schisis. Other differential diagnosis of bilateral macular schisis in a young man could be juvenile X-linked retinoschisis.

BACKGROUND

Goldmann-Favre syndrome is a rare disease characterised by early onset night blindness, reduced visual acuity and bilateral retinoschisis. Although genetic analysis is confirmatory, but the use of swept source optical coherence tomography (OCT) in association with S cone electroretinogram (ERG) can help us to diagnose Goldmann-Favre syndrome. Proper genetic counselling and follow-up is of utmost importance in these patients as there is no permanent cure for this disease.

We present a case of Goldmann-Favre syndrome so that we can always have a differential diagnosis of it in our mind with bilateral macular schisis in a young patient and suggest appropriate investigations to confirm the diagnosis.

CASE PRESENTATION

A 23-year-old male patient presented with chief complaints of decreased vision and diminution of night vision in both eyes since last 12 years. Patient was systemically healthy and no known history of eye problems or similar symptoms was present in his family.

His best corrected visual acuity was 6/24 in right eye (RE) with Snellen chart and finger counting at 1 m in left eye (LE). Ocular examination revealed a normal anterior segment with bilateral schisis of the macula. There were some associated flecks around the area of the schisis and arterial attenuation (figure 1 A, B).

INVESTIGATIONS

OCT revealed the split to be primarily in the outer plexiform layer (figure 2A, B). Height of the split was 670 µm in RE and 1760 µm in LE. Photopic and scotopic ERG revealed a mildly reduced photopic response with a marked reduction in scotopic values (photopic response—RE: 30 µV, LE: 41 µV; scotopic response—RE: 24 µV, LE: 32 µV). S cone

ERG revealed an enhanced response of RE 8.1 µV and LE 11.6 µV. Genetic testing could not be performed due to cost issues.

DIFFERENTIAL DIAGNOSIS

The closest differential of this case is X-linked retinoschisis (XLRs). Presence of nyctalopia with presence of bilateral giant macular schisis with surrounding pigment clumps and attenuated vessels with split primarily in the outer plexiform layer and an abnormal ERG response favours the diagnosis of Goldmann-Favre syndrome rather than XLRs where macular schisis is less coarse with usually visible spokewheel pattern radiating from the fovea and a split at the nerve fibre layer.¹ Associated pigment clumps and nyctalopia are usually not present in XLRs, though abnormal ERG response is also noted in XLRs.

TREATMENT

Patient was started on oral dorzolamide 250 mg two times per day.

OUTCOME AND FOLLOW-UP

Patient was explained about the genetic association of the disease and poor visual prognosis and was started on oral dorzolamide. At 3-month follow-up, patient had no further deterioration in visual acuity with no further increase in the height of the schisis.

DISCUSSION

Goldmann-Favre syndrome is an autosomal recessive disorder resulting from a mutation in the gene NR2E3 on chromosome 15q23.² Prominent features include night blindness, increased sensitivity to blue light, unusual ERG abnormalities, varying degrees of visual field loss, macular schisis.³ OCT documents the splitting within the retinal layers, and ERG demonstrates the diminished activity of the rods and cones. The diagnosis can be confirmed by genetic analysis. Based on the clinical, OCT and electrophysiology findings, a diagnosis of Goldmann-Favre syndrome was made. Normal S cone value reported in literature is 0.54–2.46 µV (Shuichi *et al*)⁴ and our laboratory value is around 1.8 µV. Jacobson *et al*⁵ studied few cases of Goldmann-Favre syndrome with relatively enhanced S cone function electrophysiologically which was identical to that found in enhanced S cone syndrome and concluded that patients with Goldmann-Favre syndrome and enhanced S cone syndrome (with no macular schisis) are not distinct

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Figure 1 (A,B) Fundus photograph of right eye and left eye, respectively, showing the macular schisis with associated flecks.

identities, but simply two identifiable phenotypes of clinical expression of a retinal degenerative disease with a single pattern of retinal dysfunction. Histopathological examination of biopsy specimen of eye wall in Goldmann-Favre syndrome has demonstrated non-specific degeneration of sensory retinal layers with thickened retinal vessel basement membrane and choroidal vascular changes.⁶

Figure 2 (A,B) Swept source OCT images of right eye and left eye, respectively, showing the giant schisis at the macula. OCT, optical coherence tomography.

Learning points

- ▶ Goldmann-Favre syndrome and X-linked retinoschisis are the two main differential diagnosis to be considered in a case of bilateral macular schisis in a young patient.
- ▶ S cone electroretinogram plays an important role in establishing the diagnosis of Goldmann-Favre syndrome.
- ▶ Optical coherence tomography helps in confirming the level of the schisis and its extent and can be a useful tool to monitor the response to therapies under investigation for such genetic disorders.
- ▶ Patient should be counselled properly regarding the course of the disease and its genetic association.

Contributors RC had diagnosed the case with bilateral giant macular schisis and the necessary investigations to diagnose Goldmann-Favre syndrome was done by MB. The draft of the case was prepared by MB and the final editing was done by RC.

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