Case Report

Outcome of Macular Hole Surgery in Bietti Crystalline Dystrophy

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Abstract

Purpose: To describe a 42-year-old man, a known case of Bietti crystalline dystrophy who underwent surgery for unilateral full thickness macular hole.
Case Report: Clinical features, color fundus photographs, and optical coherence tomography, electroretinography, and electrooculography findings of the patient are reported. His visual acuity improved from counting fingers to 20/50 after pars plana deep vitrectomy with internal limiting membrane (ILM) peeling and gas injection.
Conclusion: Macular hole can occur in Bietti crystalline dystrophy and the post-surgical outcome is good.

Keywords: Bietti Crystalline Dystrophy; Crystalline Retinopathy; Macular Hole

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INTRODUCTION

Bietti crystalline dystrophy (BCD) is a rare progressive chorioretinal degenerative disease with an autosomal recessive inheritance pattern. It is characterized by numerous yellow-white dot-like crystalline deposits scattered over the fundus and is associated with atrophy of the retinal pigment epithelium (RPE) and choriocapillaris, pigment clumping, and choroidal sclerosis, with or without the presence of limbal corneal crystals.[1,2] In this paper, a known case of BCD who underwent surgery for unilateral full thickness macular hole is presented.

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CASE REPORT

A 42-year-old man with the diagnosis of BCD and a proven genetic mutation in *CYP4V2*, who was

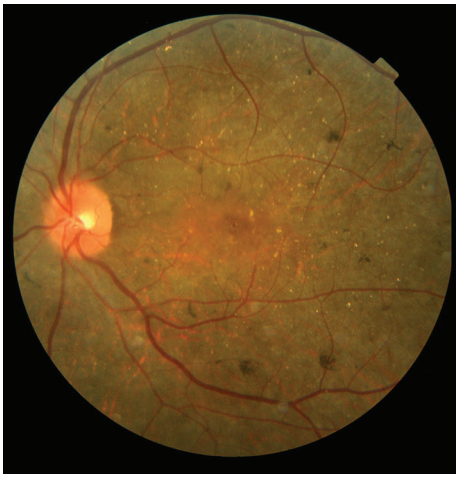

Figure 1. Color fundus photograph of patient's left eye with macular hole and characteristic crystalline deposits in the posterior pole.

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followed up for more than 10 years in our center, was admitted to the emergency ward with the complain of progressive visual loss in his left eye for three weeks. The best corrected visual acuity was 20/30 in his right eye (−1.75 − 1.25 × 180°) and counting fingers at 2 m in his left eye, with the same refraction. There was no relative afferent pupillary defect (RAPD). Slit lamp examination revealed clear cornea and lens, and normal intraocular pressure in both eyes. Funduscopic examination showed numerous bilateral, tiny, yellow crystalline deposits throughout the posterior pole and the midperiphery with diffuse RPE and choriocapillaris atrophy in both eyes and a full thickness macular hole in the left eye [Figure 1]. Optical coherence tomography (OCT) confirmed the presence of full thickness macular hole in the left eye [Figure 2]. There was no history of

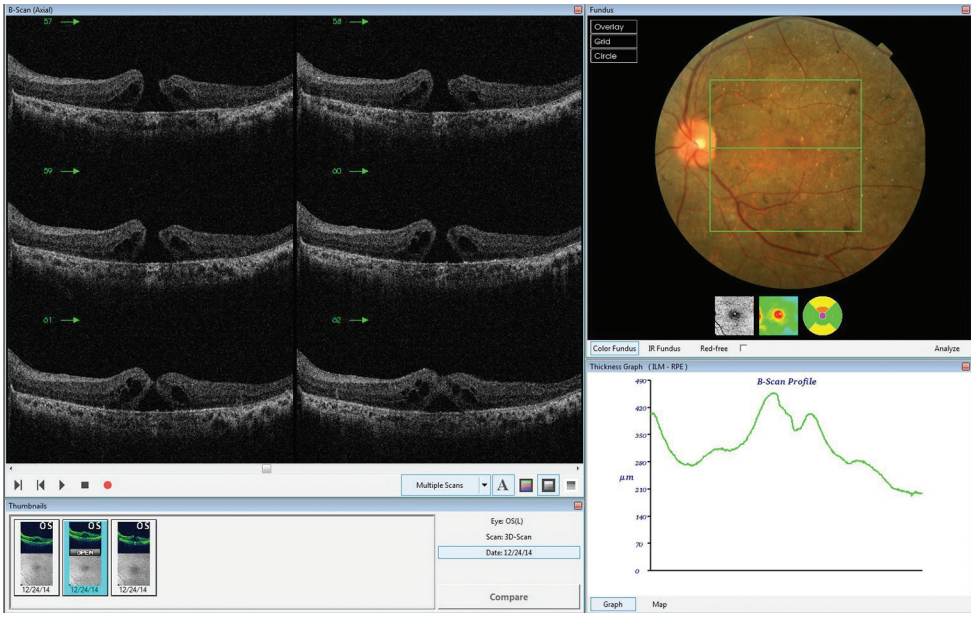

Figure 2. Optical coherence tomography of patient's left eye shows full thickness macular hole before surgery.

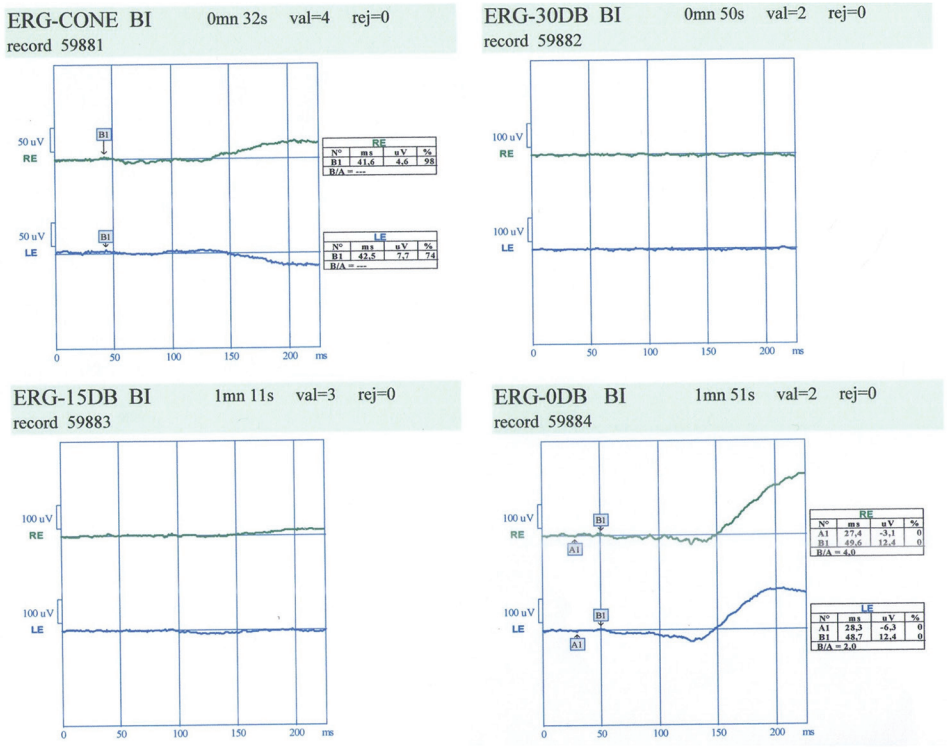

Figure 3. Electroretinography responses have become undetectable, which are characteristic of severe chorioretinal degeneration.

drug use or trauma. Electroretinography (ERG) and electrooculography (EOG) changes were compatible with the underlying disease [Figures 3 and 4]. He had visual acuity of 20/30 in both eyes in the last follow-up exam, 2 month prior to development of the new complaint. Pars plana deep vitrectomy with internal limiting membrane (ILM) peeling and gas injection was performed for the left eye. One month after surgery, the macular hole in the left eye was closed [Figure 5] and the visual acuity was improved to 20/50.

DISCUSSION

Bietti Crystalline Dystrophy accounts for 10% of autosomal recessive retinitis pigmentosa cases,[3] and is caused by mutations in the *CYP4V2* gene in chromosome 4q35.1.[1,4] Marked visual impairment, visual field constriction, and legal blindness by the fifth or sixth decades of life are the usual final outcomes.[5,6] Choroidal neovascularization,[7] cystoid macular edema,[8] and macular hole[9,10] have been reported in BCD patients but no causative mutations in *CYP4V2* or other genes were found associated with these complications. Trauma, progressive high myopia, and vitreomacular traction are some known causes of macular hole but we did not find any of these predisposing factors in this patient. Previous studies[9,10] reported both unilateral and bilateral macular holes in BCD patients without any clear predisposing factors. Unlike BCD, there is more experience on the pathogenesis of macular hole and its management in retinitis pigmentosa (RP) patients. Giusti et al[11] concluded that pathogenesis of macular hole in RP was strictly correlated to the presence of vitreoretinal interface abnormality, cellophane maculopathy, and cystic foveal degeneration with cystoid macular edema (CME). Jin et al[12] and Ratra et al[13] showed that despite variable results, it is worth attempting surgery for macular hole in the presence of other ocular co-morbidities such as RP. Successful restoration of the remaining central visual acuity not only would improve their vision, but also the quality of life. In the present case, pars plana deep vitrectomy with ILM peeling and gas injection was performed as the routine procedure for treating macular holes with good result.

In conclusion, macular hole may occur in BCD and can be successfully managed by routine macular hole surgery.

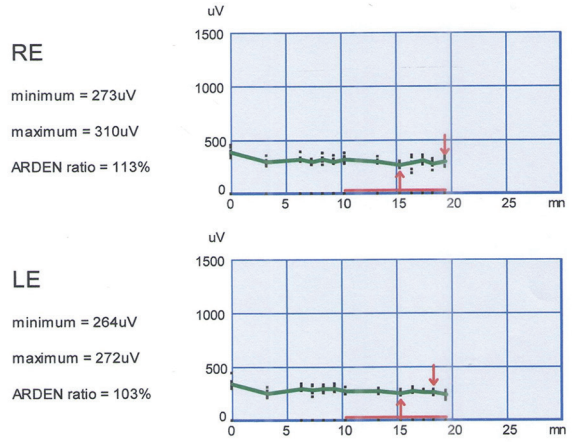

Figure 4. Electrooculography changes are characteristic of severe chorioretinal degeneration.

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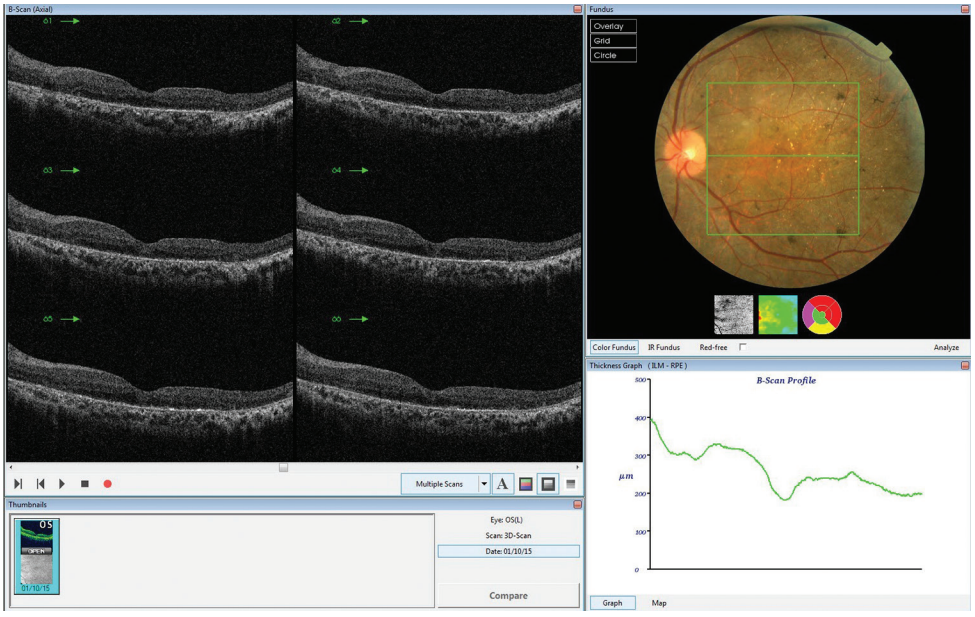

Figure 5. Closed macular hole one month after surgery.

Conflicts of Interest

There are no conflicts of interest.

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