Retinal Detachment Due To Gyrate Atrophy

Gyrate Atrofiye Bağlı Gelişen Retina Dekolmanı

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ABSTRACT

In this case study we report a 7-year old child who was first diagnosed with total retinal detachment and later with Gyrate Atrophy. The patient applied to our clinic with visual loss on his right eye without any history of trauma. The biomicroscobic evaluation fundoscopy showed a total retinal detachment on the right eye. A retinal detachment surgery was performed on the right eye. After the surgery re-detachment due to a retinal hole occurred, and the patient lost the light sense. The patient was considered inoperable.

Key Words: Gyrate Atrophy, Retinal Detachment

ÖZET

Bu olguda daha önce Gyrate atrofi tanısı almış 7 yaşında bir çocuk hastada gelişen total retina dekolmanı sunulmuştur. Herhangi bir travma öyküsü olmayan hasta sağ gözde görme kaybı ile kliniğe başvurdu. Yapılan muayenesinde sağ gözde retina dekolmanı tespit edildi. Sağ göze retina dekolman cerrahisi uygulandı. Sonrasında retinal delik nedeni ile tekrar dekole olan hastada ışık görme hissi kaybolduğu için tekrar cerrahi düşünülmedi.

Anahtar Sözcükler: Gyrate Atrophy, retina dekolmanı

INTRODUCTION

Gyrate atrophy (GA) is a rare, progressive metabolic choroidal and retinal degeneration resulting from the inherited deficiency of the pyridoxal phosphate-dependent mitochondrial matrix enzyme ornithine- δ-aminotransferase (OAT) mapped on chromosome 10q26. OAT deficiency causes hyperornithinemia, which results in progressive chorioretinal atrophy (1).

CASE REPORT

A 7 year-old child presented with decreased vision in his right eye. There was no history of trauma. The patient was diagnosed with GA with decreased night vision, high myopia and characteristic chorioretinal lesions. At presentation he was just perceiving light on right eye, and the best corrected visual acuity was 20/30 with a highly myopic correction (-12,75 D) on left eye. Biomicroscopic examination revealed an absence of red reflex on the right eye.

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Fundoscopy showed total retinal detachment on the right eye and severe chorioretinal atrophy with characteristic punched-out lesions in the peripheral retina confirming the diagnosis of GA (Figure 1). The plasma ornitin level was 580,73 µmol/L (10-163). The patient had no family history. Genetic testing was not performed in this case but we think the clinical appearance was sufficient for the diagnosis. The patient has undergone vitreoretinal surgery including parsplana vitrectomy, endolaser photocoagulation and intravitreal silicon tamponade. On the follow up examination, re-detachment due to a retinal hole occurred, and the patient lost the light sense. Cataract formation has occurred after the surgery (Figure 2). Afterwards, the patient was considered inoperable.

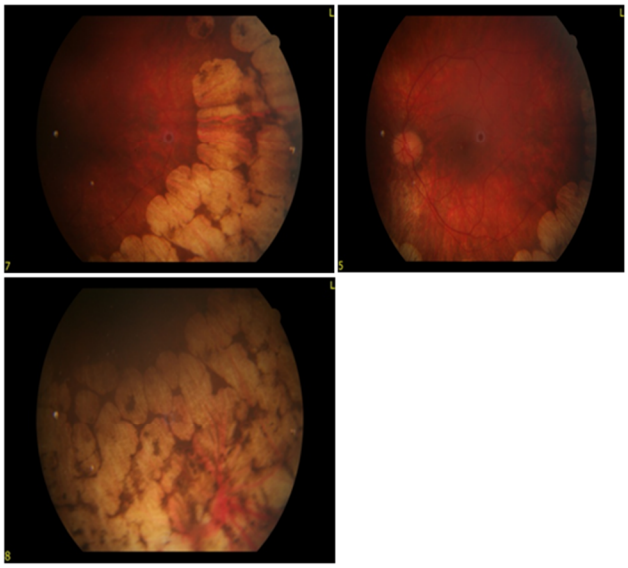

Figure 1. Fundus photo of the left eye showing chorioretinal atrophy with characteristic lesions in the peripheral retina.

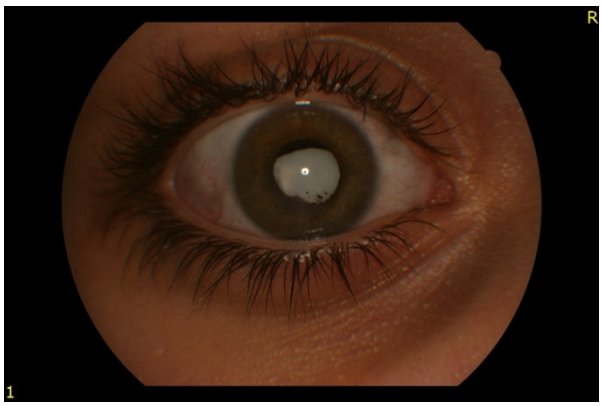

Figure 2. Cataract formation in the right eye after pars plana vitrectomy.

DISCUSSION

This case was reported because the retinal detachment due to Gyrate Atrophy is a rare condition among young children. GA is characterized by hyperornithinemia, typical retinal and choroidal lesions, and the early cataract formation. Patients with GA generally present with decreased night vision and high myopia during adolescence. Early diagnosis in these patients allows for early treatment (1,2). OAT gene studies in children with a high degree of myopia, especially in cases in which there is a complaint of nyctalopia, would be helpful for early identification (3). Retinal detachment occurs infrequently, with only isolated cases in older patients (4,5). In the treatment of GA, a low-protein diet is recommended. An arginine-free diet, may decrease ornithine to normal levels. Lysine supplement may improve the effect of the arginine- free diet (6). However, the long-term effects of this treatment approach have not been completely evaluated.

Conflict of Interest

No conflict of interest was declared by the authors.

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