Rieger Anomaly-Aniridia and Cataract Association: Asymmetric Phenotypic Presentation

Rieger Anomalisi-Aniridi ve Katarakt Birlikteliği: Asimetrik Fenotipik Prezentasyon

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ABSTRACT

We report a case of 34 years old male who had poor vision since his birth and could not sustain his life without help in recent years. We detected Rieger anomaly and cataract in the right eye, partial aniridia and cataract in the left eye. There was no posterior embryotoxon in both eyes and intraocular pressures were normal. Bilateral phacoemulsification and intraocular lens implantation were performed (the capsular tension ring support in the left eye). Postoperatively, bilateral functional visual acuity was obtained.

Key words: Axenfeld-Rieger syndrome, aniridia, cataract, phacoemulsification.

ÖZ

Bu olgu sunumunda doğumundan itibaren görme azlığı olan ve son yıllarda yardımsız hayatını devam ettiremeyen 34 yaşında bir erkek hasta rapor edilmektedir. Sol gözde Rieger anomalisi ve katarakt, sağ gözde parsiyel aniridi ve katarakt tespit edilen hastanın, her iki gözünde de posterior embriyotokson yoktu ve göz içi basınçları normaldi. Bilateral fakoemülsifikasyon ve göz içi lens implantasyonu yapıldı (sol gözde kapsül germe halkası desteği). Postoperatif olarak iki taraflı fonksiyonel görme keskinliği elde edildi.

Anahtar sözcükler: Axenfeld-Rieger sendromu, aniridi, katarakt, fakoemülsifikasyon.

INTRODUCTION

Axenfeld-Rieger Syndrome (ARS) refers to a rare autosomal dominant genetic condition characterized by anterior segment dysgenesis and systemic abnormalities. ARS characterized by posterior embryotoxon, iris hypoplasia, correctopia, and polycoria. The possible systemic findings include dental, facial bone defects, maxillary hypoplasia, umbilical abnormalities and/or pituitary involvement.[1] We aimed to report a case with asymmetric phenotypic presentation of ARS.

CASE REPORT

Thirty-four-year-old male who had poor vision since his birth and could not sustain his life without help in the last 5 years. He had no systemic disease and family history. Patient was referred to several hospitals because of cataract associated poor vision; he was undecided due to the gloomy predictions about the operation. Therefore, he was depressed. When he admitted to our hospital, we suggested a cataract surgery to him after his detailed clinical examinations. Additionally, he was consulted to the psychiatry department for his depressive states. Three weeks later, he was readmitted to our hospital for operation in a decisive way. On his ophthalmological evaluation, he had nystagmus and his visual acuities were hand motion in both eyes. Intraocular pressure was 18 mmHg on the right eye, 17 mmHg on the left eye. In the anterior segment examination, both eyes had cataract and narrow anterior chamber. There were corectopia, ectro-

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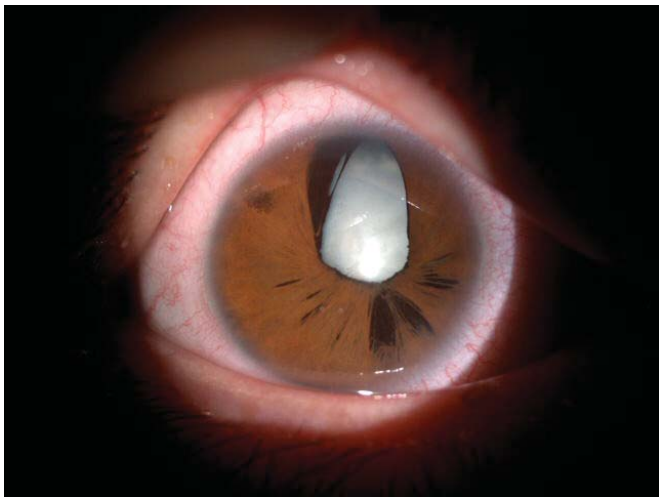

Figure 1. *Slit-lamp examination of the left eye before phaco+IOL surgery; corectopia, ectropion uvea, iris hypoplasia and cataract.*

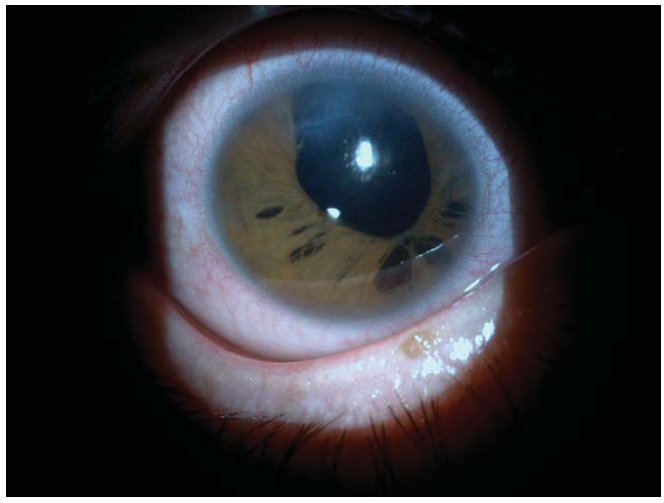

Figure 2. *Slit-lamp examination of the left eye after phaco+IOL surgery.*

pion uvea and iris hypoplasia on the right eye, partial aniridia and ectropion uvea on the left eye. There was no posterior embryotoxon in the both eyes. In gonioscopic evaluation in the right eye open angle with fibrillary structures extending from iris periphery, in left eye aniridia with rudimentary iris tissue and ciliary processes were observed (Figure 1). Posterior segment examinations of both eyes were failed due to the cataracts. B-mode ultrasonography of bilateral retina was normal. Endothelial cells were quantitatively normal and no disfigurement was reported on the specular microscopy evaluation. With these findings, the patient was diagnosed as iridogonyodisgenezis-aniridia cataract and both eyes were planned phacoemulsification and intraocular lens implantation surgery (phaco+IOL). First the left eye and then the right eye were performed phaco + IOL surgery under the local anesthesia. Operations were performed similar surgical technique. During the surgery, zonular weakness was identified in the aniridia area of the left eye, capsular tension ring (CTR) was applied (Figure 2-3). The surgeries were completed without complications by the careful surgical technique. Patient was followed for 6 months period after the surgery. The visual acuity in both eyes was 0.3 on the Snellen chart. Bilateral diffuse chorioretinal atrophy was observed in the posterior segment examination. Optic discs were normal and c / d ratios were 0.3 bilaterally.

SURGICAL TECHNIQUE

MVR knife was used to fashion a small paracentesis at the peripheral cornea, Intracameral injections of tripan blue dye (with the air support) was performed after the side ports were made and capsule had been visible. Anterior chamber was washed with balanced salt solution (BSS) and dispersive viscoelastic (Viscoat) was injected. Hydrodissection was performed carefully. Phacoemulsification was performed by stop and chop technique. We detected zonular weakness at

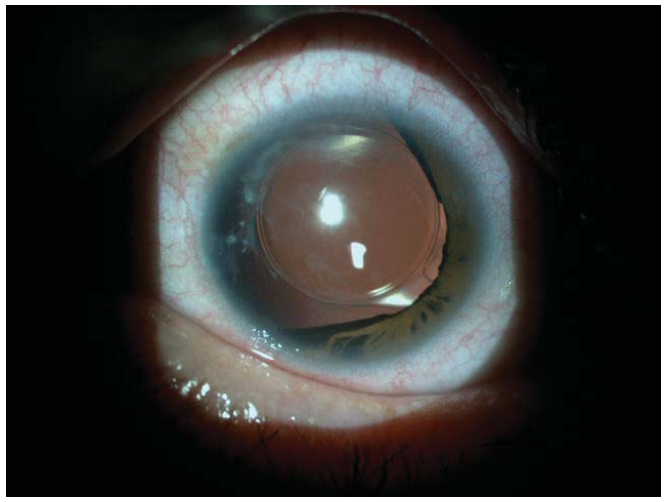

Figure 3. *Slit-lamp examination of the right eye after phaco+IOL surgery; partial aniridia and ectropion uvea. IOL is centralized.*

the aniridia part of the left eye and 12 mm CTR was placed into bag. After that procedure, uncomplicated phacoemulsification was completed. In both eyes, residual cortex material was removed by bimanual irrigation/aspiration. Foldable IOL was placed into bag after viscoelastic application. Intracameral cefuroxime application was made. Corneal inlet and side ports were closed by stromal hydration and operation was completed.

DISCUSSION

ARS frequently shows autosomal dominant pattern (70%), but sporadic cases have also been reported.[1] In our case, there was no family history. Various anterior segment findings can be seen in the spectrum of ARS. These are posterior embryotoxon, corectopia, ectropion uvea, pseudopolycoria, iris hypoplasia, mild to severe goniodysgenesis (abnormal

angle tissue and extending fibrillar structures from peripheral iris to angle). Systemic conditions associated with ARS are dental and facial abnormalities, umbilical hernia, cardiac problems and sensorineural hearing loss. Approximately half of the cases have glaucoma.[1] Most of the cases present glaucoma symetrically in both eyes. There are few reports similar to our case who presented with asymmetric phenotype.[2-4] To our knowledge, ın 1883 Vossius described a nine-year- old girl with Rieger anomaly in one eye including an ectopic pupil with full thickness iris stromal defects, while the fellow eye had a near-total absence of iris tissue (aniridia).[2]

In the new literature, eight patients with ARS were identified by Law et al. All patients had symmetric Axenfeld anomaly and / or Rieger anomaly in both eyes, except for one patient who presented with an asymmetric phenotype of the anterior segment with Axenfeld –Rieger anomaly in one eye, but aniridia in the other eye.[3] In our case, we detected Rieger anomaly and cataract in the right eye, partial aniridia and cataract in the left eye. Parveen et al. reported 76 patients with anterior segment dysgenesis in different forms. In this article, gross iris hypoplasia in an initial diagnosis of aniridia reported in one affected individual.[4] Asymmetric phenotypic presentation in ARS cases may be considered as the result of severe iris hypoplasia. However, the iris findings does not progress over the time in all cases.

Posterior embryotoxon can be helpful in establishing the diagnosis of ARS, the unexpected finding of posterior embryotoxon on a routine examination does not necessarily make a diagnosis of ARS as this finding occurs in 8% to %15 of the normal population.[1]

Unlike these reports, our patient had cataract and in the posterior segment examination after the surgery no foveal hypoplasia and / or optic disc hypoplasia were observed which may be associated with aniridia. But there were bilateral diffuse chorioretinal atrophy.

Progressive iris atrophy, one of the iridocorneal endothelial syndromes, is characterized by marked iris atrophy and hole(s) formation. In this disease, abnormal endothelial cells migration occurs. It is usually diagnosed in young adults and most often females. The endothelialization of the angle and peripheral anterior synechiae causes development of secondary glaucoma. In these cases, abnormal endothelial cells may be seen in the specular microscopy.[5] Our case is a man and the endothelial cells have normal number and shape in the specular microscopy.

In molecular genetic studies, two major genes have been described in ARS patients. These are pituitary homeobox 2 (PITX2) and forkhead box C1 (FOXC1).[6-8] The paired box 6 (PAX6) mutations have been identified in the aniridia.[9] Lawler et al. and Peer et et al. detected PITX2 mutations in their cases[4,5] Lawler et al. emphasized importance of the molecular genetic research in the diagnosis of diffucult case. In our case, no molecular genetic research was made.[3]

Glaucoma is seen in 50 % of ARS cases. In cases of early-onset, infantile glaucoma and buphthalmos may be seen. In our case, intraocular pressures were normal in both eyes. However, this patients should have life-long periodic glaucoma controls.

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