Rare association of aniridia with congenital aphakia and secondary glaucoma

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Abstract:
Congenital aniridia is a rare ocular disorder characterized by iris malformation. We present a 3-year-old boy with bilateral anterior-segment dysgenesis, congenital aniridia, congenital aphakia, secondary glaucoma, limbal stem cell deficiency, and band keratopathy. As the intraocular pressure was uncontrolled with antiglaucoma medications, the patient underwent multiple bilateral traditional cyclophotocoagulation (CPC), in addition to micropulse CPC. To the best of our knowledge, aniridia association with congenital aphakia and congenital glaucoma has been very rarely reported.

Keywords:
Anterior-segment dysgenesis, congenital aniridia, congenital aphakia, secondary glaucoma

Introduction
Congenital aniridia is a rare ocular disorder characterized by iris malformation, transmitted as an autosomal dominant trait in two-thirds of the cases; one-third of the cases are sporadic.[1,2] The incidence of aniridia is estimated to be 1 in 40,000–100,000 live births.[2-4] It can be partial or complete iris hypoplasia. In majority of cases, aniridia occurs isolated without systemic involvement, but it can also be associated with systemic disorders such as Wilm’s tumor, bilateral aniridia, genitourinary abnormalities, and mental retardation (WAGR). Visual function usually affected not due to aniridia itself but due to the associated ocular abnormalities mainly foveal hypoplasia, keratopathy, and glaucoma.[4] The common ocular manifestations of congenital aniridia are meibomian gland dysfunction, limbal stem cell deficiency, keratopathy, microcornea, lens subluxation, cataract, secondary angle closure glaucoma, and foveal hypoplasia.

To the best of our knowledge, the association of aniridia and cataract was reported in the literature, but association of aniridia with congenital aphakia and secondary glaucoma has been very rarely reported.[3]

We plan to highlight this rare association of aniridia with congenital aphakia and congenital glaucoma.

Case Report
A 3-year-old boy diagnosed with bilateral congenital glaucoma outside King Khaled Eye Specialist Hospital at 1 month of age as the mother noticed corneal opacity of both eyes since birth. There was no history of intraocular surgery in either eye. The baby was started already on dorzolamide eye drops. He is the product of normal delivery, normal pre- and post-natal history with normal birth weight. The parents are first cousins, and he is the second child (his older sister is normal). There was no family history of glaucoma or aniridia or infections during pregnancy. Parents denied any other congenital abnormalities including genitourinary dysgenesis.

Examination under sedation of both eyes revealed diffuse corneal opacity and sclerocornea with iridocorneal adhesion. Both eyes had aniridia and were aphakic clinically and confirmed with ultrasonography.

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Intraocular pressure (IOP) measured by a pneumotonometer was 24 mmHg and 30 mmHg in the right and left eye, respectively. The horizontal corneal diameter was 11.5 mm for the right eye and 10.5 mm for the left eye. Pachymetry was performed, and the central corneal thickness was more than 900 μm in both eyes. Fundus details were not clear, and the cup-to-disc ratio was 0.6 and 0.4 in the right and left eye, respectively.

A-scan ultrasound biometry revealed an axial length was 18.8 mm and 17.8 mm in the right eye and left eye, respectively. B-scan ultrasonography showed mild staphyloma of the right eye, mild vitreous opacities with posterior vitreous detachment of both eyes. There was no dropped lens material or retinal detachment detected.

A Flash VEP of the right eye showed P1 implicit time (ms): 60, P1 amplitude (μV): 3, and P2 implicit time and amplitude: 0, and all other values of the left eye are zero.

At the age of 5 months, examination performed under anesthesia. Unfortunately, the IOP was uncontrolled in his right eye despite maximum therapy. The patient underwent gentle cyclophotocoagulation (CPC) in the right eye. Afterward, he underwent bilateral micropulse CPC multiple times. First external photograph of the eyes at the age of 1 year [Figure 1]. Last Examination under anesthesia (EUA), at the age of 3 years, band keratopathy started to invade both corneas and affected the vision. Last external photograph of the eyes at the age of 3 years [Figure 2]. Right eye showing the extent of aniridia and the remaining iris tissue inferiorly is extremely thin and completely adherent to the cornea [Figure 3]. Therefore, ethylene diamine tetra acetic acid chelation with amniotic membrane transplantation was done in both eyes after one session of micropulse treatment bilaterally. Currently, IOP was controlled with only one medication for the right eye and without medication for the left eye. Last refraction was roughly +13.5 for both eyes.

**Discussion**

Aniridia is a rare congenital malformation of the iris. It can present isolated or associated with other systemic disorders such as WAGR. The incidence of glaucoma in cases of congenital aniridia has been reported to range from 6% to 75%.[4]

To the best of our knowledge, there is only one case was reported in the literature of aniridia associated with congenital aphakia.[3]

We reported this case as a case of rare association of aniridia with congenital aphakia and congenital glaucoma.

**Declaration of patient consent**

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initial s will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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**Conflicts of interest**

There are no conflicts of interest.

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