

Aniridia Associated with Lens Coloboma and Secondary Glaucoma Treated with Transcorneal Argon Laser Ciliary Body Photocoagulation: A Case Report

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Key Words

Aniridia · Glaucoma · Limbal stem cell deficiency · Congenital aniridia · Lens coloboma · Transcorneal ciliary body photocoagulation

Abstract

Purpose: The aim of this study is to describe the management of cataract and refractory glaucoma in a case of congenital aniridia (AN)-1. **Methods:** In an 18-year-old female patient affected by congenital AN, bilateral coloboma of the zonula and lens, cataract and glaucoma, pars plana vitrectomy, cataract extraction and subsequent transcorneal ciliary body argon laser photocoagulation were performed. **Results:** Five years after laser treatment, the best-corrected visual acuity was 20/50 and the intraocular pressure was stable at 18 mm Hg with topical pharmacological management consisting of timolol 0.5% and latanoprost 0.005%. **Conclusions:** From our experience, transcorneal ciliary body photocoagulation after cataract extraction and vitrectomy could be a useful technique to manage high ocular pressure in eyes affected by congenital AN.

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Introduction

Aniridia (AN) is a haploinsufficiency of the PAX6 gene (11p13) [1], presenting as total or partial absence of the iris. Multisystem diseases could be present, as this gene is also

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involved in the correct development of many systemic structures [2]. From an ophthalmological point of view, congenital AN is a complex embryologic malformation involving the iris, trabecula, cornea and lens. Increased central corneal thickness, cataracts, glaucoma, limbal stem cell deficiency, ptosis, nystagmus and foveal hypoplasia or foveal aplasia have been described [3, 4]. The incidence of glaucoma associated with AN has been reported to vary from 6 to 75% [5]. Glaucoma associated with AN typically develops during the first 2 decades of life and is attributed to progressive anterior rotation of the rudimentary iris, leading to angle closure. It is typically resistant to medical therapy, and glaucoma management for this condition is generally surgical, but there is a lack of consensus among professionals regarding which surgical approach is best [4–6]. We present the case of a patient affected by congenital AN associated with cataract, lens coloboma and refractory glaucoma, who was treated with pars plana cataract extraction using vitrectomy and transcorneal argon laser photocoagulation of the ciliary body in the right eye. The clinical condition of the left eye only required treatment with timolol 0.5% twice daily to control the intraocular pressure (IOP).

Case Presentation

Our patient was an 18-year-old female affected by congenital AN associated with palpebral ptosis and lens coloboma with an existing cataract (fig. 1a, b), but without systemic association (AN-1) [2]. The absence of a macular reflex (foveal hypoplasia) with yellowish deposits in the retinal periphery was also observed. Since the age of 5, the patient had been treated with timolol 0.05% to control the IOP of both eyes. Her best-corrected visual acuity was 20/400 in both eyes. The IOP, determined by Goldmann tonometry, was 33 mm Hg OD and 23 mm Hg OS, and corneal central thickness was 674 μm OD and 637 μm OS (Orbscan II; Bausch & Lomb, Rochester, N.Y., USA). The patient underwent pars plana cataract extraction, with both lens and capsule removed, and anterior 20G vitrectomy without intraocular lens (IOL) implantation in the right eye. After surgery, the IOP was well controlled by timolol 0.5%. However, 20 days later, the IOP began to exceed 26 mm Hg, although it was not appreciable at the slit lamp evaluation. We therefore decided to add latanoprost 0.005%. Forty-five days after surgery, the IOP continued to increase, reaching values up to 35 mm Hg. These IOP values were constant, although maximal ocular hypotensive therapy was added (brimonidine tartrate 0.2%, dorzolamide hydrochloride 2% drops and 250 mg acetazolamide tablets, twice daily). For this reason, we decided to perform transcorneal ciliary body photocoagulation. The procedure was performed with a solid-state 532-nm laser (Lumenis Novus® Spectra™). The inferior temporal quadrant was treated first (80 spots, diameter 100 μm , 250 mW, 0.3 s), and after 1 month, the inferior nasal quadrant was treated (103 spots, diameter 100 μm , 250 mW, 0.3 s). The power of the laser was regulated to obtain whitening at the head of the ciliary processes. Microbubbles and pigment release were observed at times during treatment.

Results

One day after laser treatment, the IOP was 8 mm Hg. During the successive months, it slowly increased, and 6 months after surgery, it was 28 mm Hg without medical therapy. We decided to start with topical treatment (timolol 0.5% and latanoprost 0.005%). One and 6 months after initiation of the therapy, the IOP was stable at 18 mm Hg. Moreover, after the

first month, at biomicroscopy, the ocular surface evaluation performed with dye tests (lissamine and fluorescein) did not show a significant change from the preoperative results. In our case, pars plana cataract extraction did not cause peripheral retinal tears, although the eye of the patient was affected by high myopia (10 diopters) and no acute ocular inflammation was found. After 2, 4 and 5 years, the IOP was still stable at 18 mm Hg with topical timolol 0.5% and latanoprost 0.005%, and the best-corrected visual acuity scores were 20/63, 20/50 and 20/50, respectively.

Discussion

Congenital AN is a rare pan-ocular disorder, characterized by partial or total absence of the iris and associated with cataracts, glaucoma, limbal stem cell deficiency and foveal hypoplasia or aplasia. In the aforementioned case, the patient underwent pars plana cataract extraction without IOL implantation. Surgical glaucoma management in congenital AN is a controversial topic. Filtering or drainage implant, goniosurgery or cyclodestructive procedures can be effective but are controversial [6]. The choice to perform pars plana cataract extraction without IOL implantation was made because the classical sclerocorneal approach, which is routine in cosmetic IOL implantation, could have further damaged the limbal stem cells and produced a permanent deficiency thereof [7]. Moreover, according to Lee et al. [8], congenital AN is a profibrotic disease, which could have negative implications for wound healing after surgery. This information supported our surgical plan (fig. 1c). The choice to avoid IOL implantation was further justified by a better exploration of the retinal periphery and ciliary body (fig. 1d) as well as the presence of high myopia (10 diopters) in the patient's eye. A cosmetic IOL implant, in fact, would not have allowed us to treat the ciliary body with transcorneal photocoagulation, and traditional, surgical glaucoma procedures, such as trabeculectomy or implantation of a drainage device, would probably have led to failure, as has been described in the literature [9, 10]. In conclusion, pars plana cataract extraction using vitrectomy, associated with transcorneal argon laser photocoagulation of the ciliary body, could be an alternative and valid way to treat cataract and refractory glaucoma in eyes affected by congenital AN. However, it is necessary to study and document a large number of cases to better understand this and similar pathological conditions as well as their ideal treatment.

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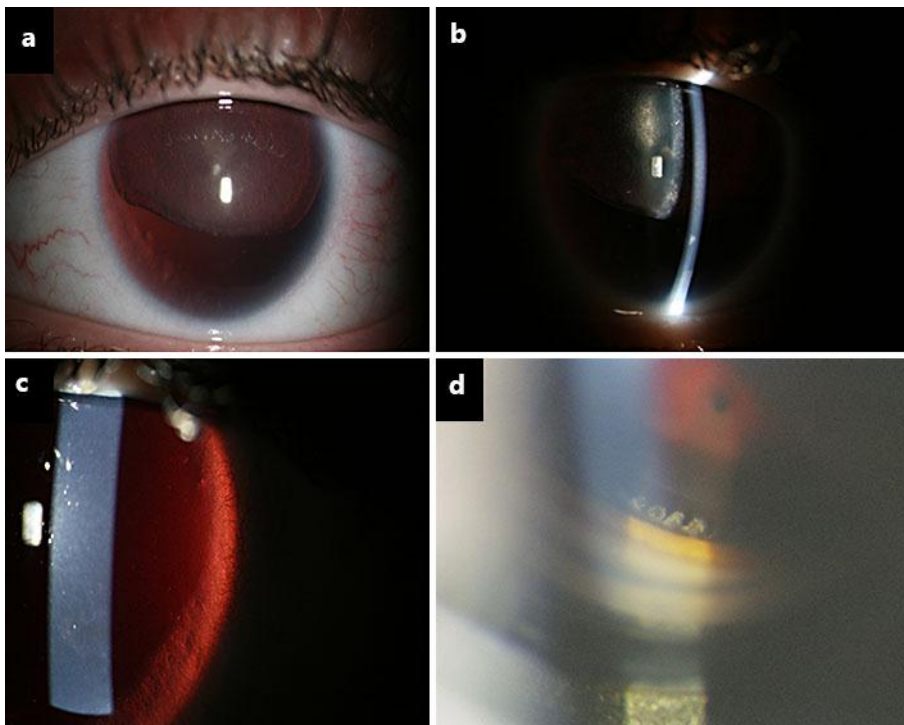


Fig. 1. **a, b** Congenital AN-1 associated with cataract and coloboma lens. **c** Post-surgical aphakia and limbal stem cell alterations. **d** Transcorneal argon laser photocoagulation of the ciliary body. The power of the laser was regulated to obtain a whitening at the head of the ciliary processes.