CASE REPORT

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Treatment of Congenital Aniridia Associated with Subluxated Infantile Cataract

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ABSTRACT

A 5 year old boy was presented at Eye clinic University clinical center Tuzla with congenital aniridia in both eyes. Clinical examination revealed visual acuity of 0,08 without correction in right and 0.7 with -5.0 Dsph and -1.0 Dcyl Axx 109° in left eye. Opthalmologic examination showed bilateral aniridia associated with moderate cataract in the right and incipient cataract in the left eye. In the right eye, zonular weakness with incipient capsular displacement and esotropia of Δ6°, were noted. The patient underwent phacoemulsification, implantation of capsular tension ring and Artificial Iris implant in the capsular bag. Phacoemulsification went uneventful and early postoperative recovery was successful with no signs of aniridia-associated keratopathy development and normal values of intra ocular pressure. Patient was not motivated for operation of the left eye and it was corrected with soft contact lens. Six month after the operation visual acuity in the right eye improved to 0.9 with +1.25Dsph and maintained stable in left eye, with complete elimination of esotropia and signs of binocular vision restoration. Small incision cataract extraction with IOL and Artificial Iris implantation in one procedure can be used to correct congenital aniridia and cataract with significant visual function improvement.

Key words: congenital aniridia, infantile cataract, Artificial Iris.

1. INTRODUCTION

Congenital aniridia is a rare bilateral pan ocular disorder characterized by complete or partial absence of the iris (1, 2). Although there are some reported cases with no mutations (3), the paired box gene 6 (PAX6) located at 11p13 is major gene associated with aniridia (1, 2, 4, 5). Reported prevalence of congenital aniridia in general population is between 1 : 64 000 and 1 : 100 000 (1, 6, 7). Two thirds of all cases are familial and one third is sporadic (1, 7).

Aniridia is associated with a wide range of other ocular abnormalities including aniridia-associated keratopathy (AAK), ectopia lentis, cataract, glaucoma, nystagmus, foveal and optic nerve hypoplasia (1, 2, 4, 8). It may occur as isolated disorder or be associated with a number of syndromes such as WAGR (Wilm's tumor, bilateral sporadic aniridia, genitourinary abnormalities and mental retardation) and Gillespie's syndrome (aniridia, mental retardation and ataxia) (1, 7).

Purpose of this report is to present a case of familial congenital aniridia associated with infantile subluxated cataract, successfully treated with phacoemulsification and Artificial Iris implantation. This report shows different opportunities in management of aniridia with therapeutic challenges for such complicated cases.

2. CASE REPORT

A 5 year old male presented at Eye clinic University clinical center Tuzla. Boy's mother complained that her son has some problems since birth; she noticed boy has wide pupils and he always walks with his head down. She noticed that in last couple of months he is unable to see well and his right eye is starting to look toward the nose. Boy's grandmother was treated at other clinic because of the aniridia, glaucoma, keratopathy and she is blind for the last 6 years. Mother also has diagnosed aniridia and regularly attends ophthalmologists' controls.

Complete opthalmologic examination revealed following: best corrected visual acuity (BCVA): 0,08 without any correction in the right eye and 0.7 with -5.0 diopters of sphere (Dsph) and -1.0 diopters of cylinder (Dcyl) axis (Axx) 109° in the left eye. Anterior segment examination revealed incipient degeneration of the peripheral cornea (grade 1 of AAK), and bilateral aniridia. Right eye had partially subluxated posterior polar cataract with zonular weakness in meridians 6-10 o'clock and incipient capsular displacement while left eye had incipient posterior polar cataract. Right eye also showed signs of nasal deviation–esotropia of Δ6°. Intra ocular pressure (IOP) measured with Goldmann applanation tonometry was 14 and 15 mm Hg in the right and left eye respectively. Fundus details were not clear in right eye and left eye showed normal fundus finding. Ocular Coherent Tomography (OCT)

Figure 1. Anterior segment of right and left eye. Posterior polar cataracts more pronounced in the right eye. Note the right eye has zonular dehiscence in meridians 6-10 o'clock.

Figure 2. Immediate postoperative finding and clinical appearance 1 month postoperatively

analysis revealed no pathological changes in both eyes. Ultrasonography examination showed opacification of posterior lens capsule, measured axial length (LAX) 22.58 mm in right eye and 23.03 mm in left eye and anterior chamber depth (ACD) of 3.03 mm and 3.09 mm in right and left eye respectively. Measured horizontal white to white distance (WTW) was 11.5 mm and diameter of the capsular bag was 9.0 mm in both eyes. Keratometry manual and automated revealed following values K_1 = 42.50 D and K_2= 44.00 D with astigmatism of -1.50Dcyl Axx 108° in right and K_1 = 42.00 D and K_2= 43.00 D with astigmatism of -1.0Dcyl Axx 109° in left eye. Corneal topography showed symmetric bow tie pattern with regular with the rule (WTR) astigmatism in both eyes. Systemic pediatric and ultrasonography examination did not find any systemic abnormalities.

The patient underwent phacoemulsification in general anesthesia. Clear cornea cut incision at the steepest meridian (108°) was made with Clear Cut HP Dual Bevel Slit Knife 2.8 mm. 5 mm capsulorrhexis was performed without capsular staining. After capsulorrhexis (approximately 6 mm wide), capsular bag was stabilized with implantation of capsular tension ring (CTR Morcher® type 1C, 9.00 mm). Upon completing the emulsification and aspiration (Infinity Vision System, Alcon Laboratories, Inc., measured phaco-time 10 seconds) intra ocular lens (IOL) Alcon Acrysof MA 60BM 24D was implanted in the capsular bag through clear cornea cut using Monarch III IOL delivery system cartridge B (Alcon Laboratories, Inc.). Upon IOL implantation viscoelastic was reinstilled in the anterior chamber to maintain space. The Artificial Iris implant (Dr. Schmidt Intraocularlinsen GmbH) was resized with 8 mm trephine. Resized Artificial Iris was gently manually folded with two forceps and inserted into the anterior chamber through the 2.8mm main incision with implantation forceps. After the implantation, Artificial Iris spontaneously opened in the anterior chamber. Smooth rotation with micro manipulator introduced implant into the capsular bag over the IOL. Viscoelastic was manually removed and wounds hydrated.

Postoperatively, moderate signs of ocular inflammation were noted. Patient was treated with topical corticosteroid eye drops for four weeks and continued topical therapy with lubricants. IOP was regularly monitored with no signs of raised IOP. 3 months postoperatively BCVA improved to 0.9 with +1.25 Dsph. Patient was not motivated for operation of left eye and soft contact lens is applied. 6 months after the operation BCVA remained the same in both eyes and complete elimination of esotropia was noted with signs of binocular vision restoration.

3. DISCUSSION

Aniridia is a complex disorder that usually results in severely impaired vision from multiple causes including aniridia associated keratopathy (AAK), cataract, glaucoma, foveal hypoplasia, nystagmus and retinal detachment (1, 7, 9).

Iris deficiency associated with aniridia is connected with decreased visual acuity, glare and photophobia (1). Aniridia associated with cataract develops in 50–85% of patients (7). Most of these patients come to operation as adults (10). These eyes can have zonular weaknesses and the anterior capsule can be altered and become very fragile which makes cataract extraction very difficult (11, 12). Anterior capsule staining may be necessary because of the poor visualization, although not in this particular case. Eyes with stem cell deficiency are sensitive, wounds heal slower and it is important to perform surgery with as little trauma as possible. Although it is imperative to make clear cornea cut smaller, it has to be large enough to allow manual implantation of the Artificial Iris.

Cataract surgery in aniridic eyes is also opportunity to make a refractive surgery. Phacoemulsification incision placement on the steep corneal axis corrects small amounts of astigmatism (13). Postoperative astigmatism is important for children because of adverse effects on vision development and the risk of amblyopia (14). Preoperative corneal astigmatism of -1.5 Dcyl was resolved with properly calculated surgically induced astigmatism (SIA) (15). For IOL power calculation SRK-T formula was used (16). Posterior capsulotomy although indicated (14, 16, 17) was not performed due to instability of the capsular bag. Anterior vitrectomy through pars plana approach was not indicated since the patient is 5.5 years old and because of potential adverse effects of additional trauma to the eye (14, 17). Due to instability of the capsular bag, CTR was introduced and three-piece mono focal acrylic IOL was implanted. Hydrophobic acrylic IOL are reported to have lower incidence of posterior capsule opacification (PCO) (14, 16, 17). Instead of calculated 23D IOL, hyperopic 24D IOL was used due to expected later myopic shift (14, 16, 17). Because of patient's age, questionable stability of the capsular bag and constant pupil diameter of the Artificial Iris implant, mono focal and not multifocal IOL was used.

Cataract extraction in aniridia patients is challenging task by itself and after cataract extraction there is still aniridia to be solved. Implantation of prosthetic iris devices improved postoperative outcomes by reducing glare disability (18). One of the options for treatment of both cataract and aniridia is use of the black iris diaphragm IOL (19). Latest innovation is the use of Artificial Iris implant which is fully customized with handmade color composition, structurally matches the appearance of natural iris and restores the aesthetic appearance (20). This prosthe-

sis can be injected via a clear cornea incision as part of a planned combined procedure (21) which allows minimal invasive surgical procedure. On the other hand, glaucoma has been found to be the most important complication, though implantation of the iris prosthesis in the capsular bag may reduce this complication. Implantation in the capsular bag requires a larger capsulorrhexis which can present a surgical challenge in eyes with changed anterior capsule (9).

AAK is thought to have an incidence of 20% of all aniridia cases (22). It is caused by a combination of factors: an abnormally differentiated epithelium, abnormal cell adhesion, impaired healing response, limbal stem-cell deficiency and the infiltration of conjunctival cells on the cornea (1, 8). There is also a large increase in central corneal thickness (23). Management of mild AAK includes preservative-free lubricants and dark glasses aid against photophobia. In moderate keratopathy, serum drops and amniotic membrane transplants may be a useful measure to enhance the survival and expansion of surviving limbal stem cells (24). In severe disease, a limbal cell transplant is recommended (25). Penetrating keratoplasty has a poor prognosis because of the frequent recurrence of the same pre-graft corneal changes (26). Patient's grandmother has grade 3 and mother has grade 1 AKK (27). Patient has signs of grade 1 AKK and minimally invasive surgical intervention is needed with reduced phaco time and continuous treatment with lubricants is indicated.

Glaucoma in aniridia occurs in 6–75% (15% of patients per age decade) (10) of all cases due to developmental abnormalities in the drainage angle of the eye (7, 28). Aniridia associated glaucoma requires medical therapy with topical and systemic agents and surgical treatment. Surgical treatment is challenging and includes prophylactic goniotomy, therapeutic goniotomy, trabeculectomy with and without antimetabolites, cyclocryotherapy and guarded filtration surgery (GFS) (1). GFS using Ahmed Molteno and Baervaldt implants have been found to be very effective in obtaining control in aniridic glaucoma. Although having some complications, success rate of GFS ranges from 66% to 100% (29). Implantation of Artificial Iris in the capsular bag should not increase IOP and if needed, further GFS surgery can be performed.

Optic nerve hypoplasia and macular hypoplasia are common findings in congenital aniridia and can lead to severe visual impairment (1, 7). Optic nerve hypoplasia is found in approximately 10% of all cases (30). Performed posterior segment OCT examination did not find any changes in either eye, which allowed significant visual improvement.

Aniridia is associated with several systemic disorders. WAGR syndrome is the most common and most important of the aniridia syndromes (31). Detailed systemic pediatric examination, ultrasonography and laboratory results and did not find any signs of systemic complications. Further pediatric controls are planned.

4. CONCLUSION

Aniridia patients require systemic ophthalmic evaluation before any surgery is performed. Small incision cataract extraction with IOL and Artificial Iris implantation in one procedure can be used to correct congenital aniridia and cataract with significant visual function improvement. Aesthetic results are satisfactory and photophobia is reduced to minimum. Amblyopia can be prevented with appropriate cataract extraction timing and correct IOL calculation.

CONFLICT OF INTEREST: NONE DECLARED

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