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Bilateral morganian cataract in patient with congenital aniridia - case report

Catarata morganiana bilateral em paciente com aniridia congênita - relato de caso

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ABSTRACT: Decreased vision due to important medium opacity is common in aniridia, however there are few reports of combined surgery in this condition. Cataract surgery with polymethylmethacrylate intraocular lens implant remains a challenge for surgeons, due to the greater weight of this material and the zonular dialysis presented by these patients. The present study aims to report a case of aniridia in a young patient, with hypermature cataract.

Keywords: Aniridia; Cataract; Cataract/genetics; Surgery.

RESUMO: A diminuição da visão devido à importante opacidade de meios é comum em aniridia, porém existem poucos relatos de cirurgia combinada nesta condição. A cirurgia de catarata com implante de lente intra-ocular de polimetilmetacrilato continua sendo um desafio para os cirurgiões, devido ao maior peso desse material e à diálise zonular apresentada por esses pacientes. O presente estudo, tem como objetivo relatar um caso de aniridia em uma paciente jovem, apresentando catarata hipermadura.

Descritores: Aniridia; Catarata; Catarata/genética; Cirurgia.

INTRODUCTION

Congenital aniridia is a rare condition, with an incidence between 1: 64000 to 1: 100000. It typically affects both eyes, with two thirds of the cases coming from autosomal dominant inheritance and the rest being sporadic cases[1,2]

It is associated with mutations in the PAX6 gene, located in the short arm of chromosome 11 (11p13)[3,4,5]. This gene has a role in ocular development, mainly in the cornea, iris, lens, camerular angle and ciliary body[1]. In some cases, an association with other anomalies may occur, characterizing WARG syndrome (Wilms' tumor, aniridia, genital abnormalities and intellectual disability)[1].

The present study aims to report a case of aniridia in a young patient, with hypermature cataract. Evidence that the reported association leads to an increased risk of subluxation of the intraocular lens (IOL) due to zonular fragility, making combined surgery for aniridia and cataract a challenge for the surgeon.

CASE REPORT

A 36-year-old, female black patient, was seen at the ophthalmology service of Santa Casa de Vitória. Referred for evaluation of low visual acuity and presence

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of leukocoria in both eyes (OU). On physical examination, she presented visual acuity uncorrected (UCVA) of hand motion (HM) at 1 meter in the right eye (OD) and HM at 6 meters in the left eye (OS). The ectoscopy showed hypertelorism, bilateral proptosis, horizontal nystagmus and exotropy in OU. In Biomicroscopy (BIO) it presented in OD: calm conjunctiva, clear cornea with peripheral neovascularization, aniridia and morganian cataract, in OS: calm conjunctiva, clear cornea with peripheral neovascularization, aniridia and total white cataract. The initial intraocular pressure (IOP) measured by a Goldmann tonometer was 26 mmHg in OU, with the use of 0.5% timolol 12/12h in OU. The dilated-pupil fundus examination (DFE) was impraticable in OU. Bilateral ultrasound showed posterior pole staphyloma with applied retina and posterior vitreous detachment.

Brimonidine at 12/12 hours and latanoprost 1 x at night was used to control IOP, which decreased to 18 mmHg in OU. The possibility of an IOL implant with a specific diaphragm was suggested for cases of aniridia, but it is not marketed in Brazil 7,8. In 2017, a 3-piece capsular tension ring and IOL was requested for right eye surgery, but due to operational and administrative difficulties inherent to the public health service, the material was not purchased, and the option was to proceed with the surgery. , without IOL implantation, due to the significant visual impairment and presence of myopic cone. The patient underwent extracapsular facectomy in OD (latanoprost was suspended during the period of the surgical procedure), without IOL implantation. After the OD surgery, the patient showed improvement of the UCVA to 20/200 and the visual acuity with correction (BCVA) was 20/80 with changes, with the refraction +8.00 spherical / -2.00 cylinder at 180º.

After the first surgery, the patient was absent from the service, returning later in 2018. The left eye developed from white cataract to morganian cataract. UCVA was 20/200 in OD and HM at 1 meter in OS. In BIO OD: clear cornea, full facectomy sutures, aniridia and in OS morganian cataract. Again, a three-piece capsular tension ring and IOL was requested, but administrative difficulties prevented the acquisition of the material. It was decided to perform an extracapsular facectomy with a rigid one-piece IOL implant, without a capsular tension ring, aiming to remain with a monovision far away in the left eye.

In the immediate postoperative period, the lens was centered and inside the capsular bag, with intact sutures, transparent cornea and a calm eye. On the seventh postoperative day, subluxation of the lens occurred, with the lower haptic in the anterior chamber and the upper dislocated posteriorly, with the patient presenting UCVA of 20/200. The intraocular lens was explanted, with subsequent follow-up the patient's BCVA progressed to 20/100 with correction of +9.00 spherical / -1.00 cylinder at 180º.

IOP remained 18 mmHg in OU, using brimonidine, timolol and latanoprost. The DFE showed increased optic disc excavation, foveal hypoplasia, significant atrophy of pigmented retinal epithelium and staphyloma of the posterior pole. In the Figures 1, 2, 3 below, the pre and postoperative phases are presented.

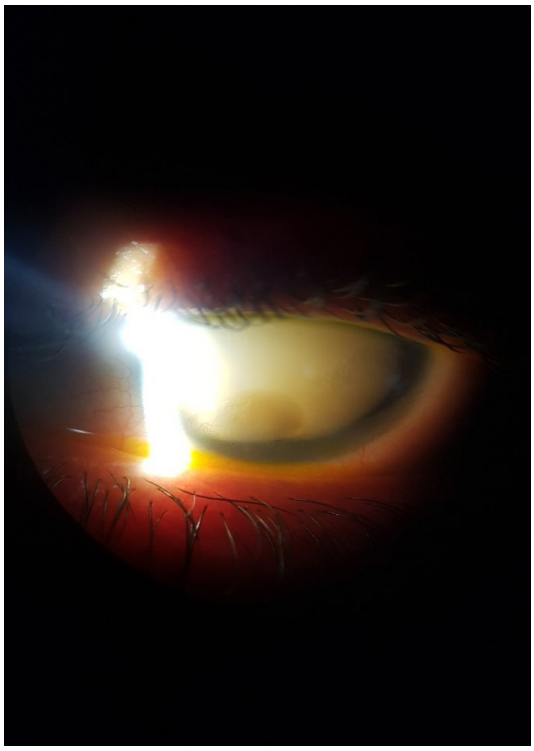

Figure 1 - Morganian cataract with liquefied cortex and detached nucleus (preoperactive)

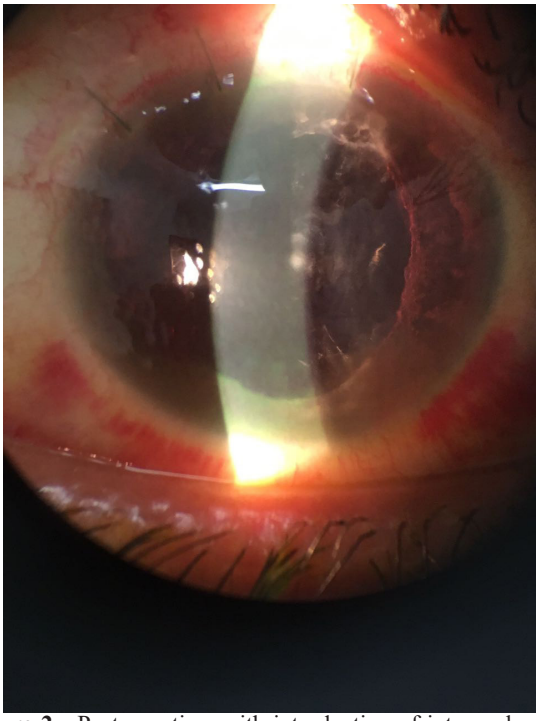

Figure 2 - Postoperative, with introduction of intraocular lens in the capsular bag

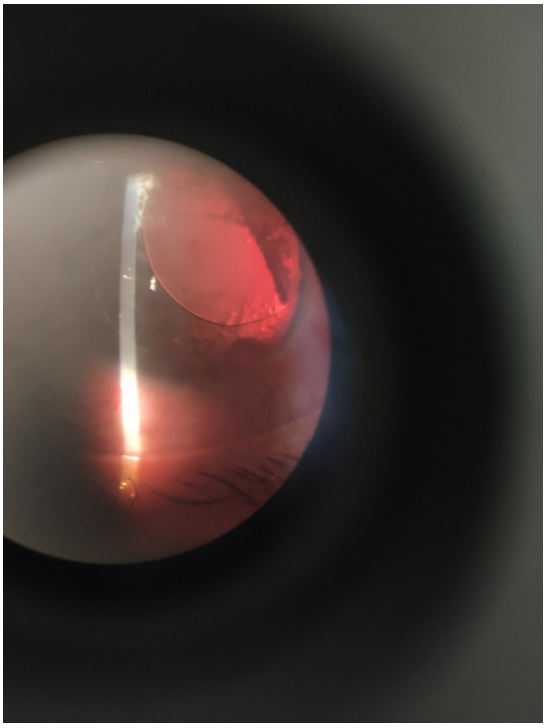

Figure 3 - Sublocation of the intraocular lens

DISCUSSION

Aniridia is a panocular disorder that affects the cornea, iris, camerular angle, crystalline, fovea and the optic disc. Individuals with aniridia usually have nystagmus, reduced visual acuity (usually 20/100 to 20/200), and phenotypes generally do not differ between the eyes[1-6].

The reduction in visual acuity is mainly caused by foveal hypoplasia. Cataract, glaucoma and corneal opacifications are responsible for progressive vision reduction. Most children have aniridia at birth, and the association with congenital glaucoma is rare in these cases. Despite the various eye problems, these patients still maintain a functional part of their vision with adequate eye care[1,3,6].

The most obvious clinical alteration of the disease is hypoplasia of the iris. The severity varies from almost normal to its complete absence. In milder cases, the pupillary diameter may be normal, but there is an absence of the surface architecture of the iris or the presence of transillumination. It may also present partial iris defects similar to colobomas or eccentric pupils[1,6].

In Cristalino, congenital opacities are common, and there may be persistence of vascularization of the anterior capsule (tunica vasculosalentis). Opacities are rarely dense to require surgery in childhood, but they develop in 50-85% of those affected in adolescence and early adulthood. Subluxation of the lens may also occur, which is a rare condition[1,3,6].

Eye pressure is commonly elevated in patients with aniridia, occurring in up to two-thirds of individuals. The elevation, associated with loss of ganglion cells, alteration of the visual field and increased excavation of the optic nerve constitutes the diagnosis of glaucoma. The onset of glaucoma usually occurs in adolescence, being rare in childhood[1,3,6].

Corneal degenerations appear as late manifestations and of multifactorial cause, which include abnormalities of limb cells and abnormal wound regeneration. The changes include peripheral neovascularization (which can evolve to corneal vascularization), opacification and keratinization. Tear film production is inadequate and exacerbates surface problems. The central corneal thickness is increased (which can result in an overestimated IOP)[1,3,6].

In the fundus of the eye, foveal hypoplasia is usually present. Findings include reduced foveal reflex, macular hypopigmentation and loss of foveal avascular zone boundary. Optic nerve hypoplasia (abnormally small nerve head) can occur in 10% of patients with or without the presence of nerve colobomas. In the retina, ruptures and detachments may appear due to high myopia or previous eye surgery. These patients have reduced smell and cognition, behavioral problems and developmental delay[1,3,6].

The treatment most often consists of simple but important measures. Patients should undergo regular examinations to correct refractive errors. Although low VA is not related to the absence of iris, 64% of these patients have myopia and 37% have amblyopia6, which should be treated early with occlusive therapy. The absence of iris is correlated with photophobia that can be treated with the use of photochromic or dark lenses to reduce sensitivity.

Patients with corneal pathology should be treated according to the severity of the involvement, in mild keratopathies, eye lubricants without preservatives should be used. In moderates, autologous serum and transplantation of amniotic membranes can be used. In severe keratopathies, there is usually a failure of the limbus and penetrating transplantation is usually not efficient and, as the disease affects both eyes, the transplantation of autologous limbar cells is also not efficient. Some centers have been using cultures of limbar cells in amniotic membranes and keratoprosthesis implantation successfully[1,3,6].

Intraocular pressure should initially be treated with topical hypotensive agents. Trabeculectomy or insertion of drainage tubes is reserved only for patients who do not respond to treatment .

When opacity of the lens occurs, cataract extraction must occur in important opacities, remembering the partial improvement in vision due to foveal hypoplasia. At the time of surgery, lenses with an artificial iris, capsular rings with specific opaque regions, or artificial

hydrophobic silicone irises, tailor-made for the patient, can be used[7,8,9,10,11].

In more recent studies conducted by Trindade et al.[9], the development of opaque implants using the pinhole technique has shown promising results. Although opaque, this implant has the advantage of allowing the monitoring of the patient's fundoscopy, it allows visualization through examination with infrared lamps. It also has the advantage of reducing interference caused by peripheral corneal astigmatisms.

In our case report, due to the presence of myopic cone and the administrative difficulties of a service of the Unified Health System, it was decided in the end to remain with the aphakic patient with the use of corrective lenses. We also decided not to request a specific IOL for aniridia, because it is not sold in Brazil (Morcher®, Stuttgart, Germany)[7,8], making the order unfeasible. Despite the difficulties inherent to the public network, such patients need follow-up, seeking to minimize suffering and preserve visual acuity and quality of life.

CONCLUSION

Aniridia is a complex disease that affects several eye structures. The visual prognosis is poor in these patients due to foveal involvement and commonly to the optic nerve. Despite this, regular monitoring is important to prevent the worsening of visual acuity that can occur due to amblyopia due to refractive errors, the presence of lens opacifications, corneal changes and the appearance of glaucoma. Aniridia is a challenging disease that requires the doctor's attention to preserve the patient's visual acuity in the best possible way.

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