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Case Report

Bilateral, Congenital Isolated Aniridia in a Child: A Case Report

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Abstract

Congenital aniridia is a rare genetic disorder due to PAX6 gene mutation primarily characterized by partial or complete absence of iris, associated with panocular defects. Here, we report a case of congenital, isolated bilateral aniridia with an affected parent. Our index case was a 7-year-old girl having diminution of vision since birth. We performed a through clinical examination; slit lamp biomicroscopy, gonioscopy, and fundus examination, apart from screening of her parents which revealed similar disease pattern in her father. The patient had no ocular and systemic complications; henceforth, she was treated conservatively for visual rehabilitation with best corrected visual acuity of 6/24 in both eyes.

Key words: Aniridia-associated keratopathy, Congenital aniridia, Ultrasound B scan

INTRODUCTION

Aniridia is a rare bilateral genetic disorder having a spectrum of ophthalmologic abnormalities.[4] Aniridia may further be subdivided into two broad categories congenital or acquired (post trauma and surgery). Majority (around 2/3rd) of the cases of congenital aniridia are familial with autosomal dominant pattern of inheritance with complete penetrance but variable severity.[1,5] People with familial aniridia, mainly display isolated ocular defects. Rest of the cases (1/3rd of the cases) are sporadic with ocular as well as systemic findings.[1] Visual acuity is highly variable in these patients, but the presence of nystagmus generally worsen the quality of vision.[5] Patients with aniridia mainly complaints of photophobia and glare.[6] Mainstay of treatment is conservative management, that is, maintenance of best corrected visual acuity by correction of refractive error.[5]

CASE REPORT

A 7-year-old girl presented in the eye clinic with chief complaints of painless diminution of vision associated with photophobia and glare since birth. There was no history of trauma, drug intake, and no significant past medical history. Her father had severe visual impairment in both eyes, mother had no complaints regarding her vision. Our case of interest had no siblings. Her uncorrected Snellen visual acuity was 4/60 in both eyes, with jerky horizontal nystagmus. Slit lamp examination revealed bilateral total aniridia (360°) with normal anterior chamber depth. In both eyes, there was subluxation of clear crystalline lens upward, and no zonules inferiorly lead to straightening of inferior lens margin mimicking lens coloboma [Figure 1]. Intraocular pressure (IOP) measured by applanation tonometry was 14 mm Hg and 16 mm Hg for the right and left eyes, respectively. Gonioscopy examination revealed open angles with rudimentary iris frill present in 360° [Figure 1]. Bilateral fundus evaluation done by indirect ophthalmoscopy showed mild optic disc hypoplasia at inferior disc margin.

Biometry

Parameter	Right eye	Left eye
Axial length	20.78 mm	21.00 mm
AC depth	2.19 mm	2.47 mm
Lens thickness	3.90 mm	3.85 mm
K1	42.25 D	41.00 D
K2	44.75 D	43.00 D
Axis	9°	168°

A thorough pediatric evaluation was done to rule out any systemic associations and the findings are listed below:

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Parameter	Finding in our subject	Expected at this age of 7 years
Height	110 cm	119 cm
Weight	16 kg	20 kg
Head circumference	46.5 cm	51 cm
Chest circumference	52 cm	66.71 cm
Upper segment: Lower segment (US: LS ratio)	0.8: 1 (56/63)	1.1: 1

Arm span–111 cm, body mass index–13 (underweight). Therefore, she had stunted growth [Figure 2].

No other systemic abnormalities were detected. Since she had subluxation of lens, Marfan's syndrome was ruled out by lower arm span: Height ratio = 1.009 compared to >1.5 seen in Marfan's syndrome. Thumb sign, wrist sign was negative, with no pectus excavatum or scoliosis.

Systemic examination including hemogram, blood coagulation profile, and serum calcium was within normal limits. Serum homocysteine level was normal and urine sodium nitroprusside test was negative. Electrocardiography and 2-D echocardiogram were unremarkable.

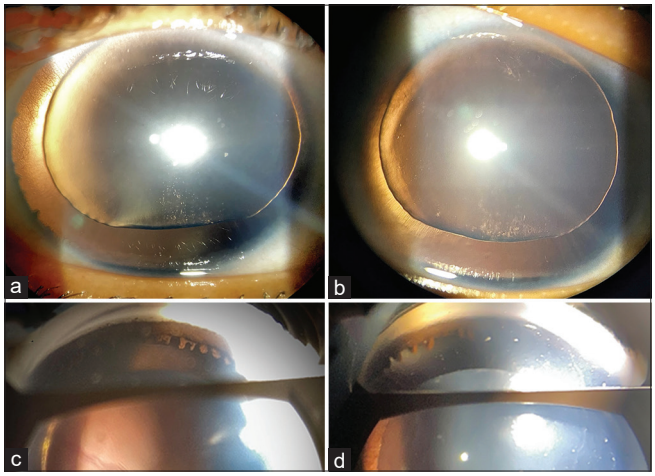

Figure 1: Slit lamp photography of the right (a) and left eye (b) showing aniridia with straightening of inferior lens margin due to absence of zonules inferiorly. Gonioscopy showing rudimentary iris frill in the right (c) and left eye (d), respectively

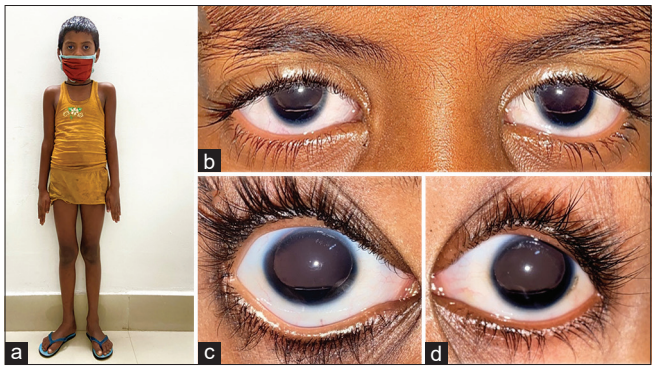

Figure 2: Clinical photograph (a) and facial profile (b) of a 7-year-old girl with clinical aniridia in the right (c) and left eye (d), respectively

Screening of parents was also done, keeping in mind the autosomal inheritance pattern of isolated aniridia. Subject's father had gross visual impairment in both eyes with no perception of light in the right eye and perception of light with accurate projection of rays in the left eye associated with bilateral horizontal jerky nystagmus, and conjunctival xerosis. In the right eye, he had history of vegetative matter trauma 2 years back leading to fungal corneal ulcer, which lead to central leukoma formation with peripheral scarring and corneal vascularization, iris details could not be seen [Figure 3]. In the left eye, there was diffuse corneal opacity associated with scarring of central and inferior cornea suggestive of Aniridia-associated keratopathy (AAK) due to limbal stem cell deficiency, and superiorly rudimentary iris was visible. The presence of ciliary staphyloma was also noted in the left eye [Figure 3]. IOP measurement revealed hypotony in the right eye, and high IOP 24 mm hg in the left eye. Ultrasound B scan showed choroid-scleral thickening, reduced axial length leading to pre-phthisical state in the right eye and posterior vitreous detachment with the low reflectivity vitreous echoes in the left eye [Figure 4]. He was under treatment for bilateral AAK and secondary glaucoma in the left eye for the past 2 years.

Our patient was prescribed glasses (right eye - 2.5 dS/-4.25 dC × 2°, left eye -1.75 dS/-5.25dC × 167°) with best corrected visual acuity of 6/60 in both eyes and advised to use preservative free lubricating eye drop, tinted glasses when exposed to sunlight to reduce photophobia and glare. Since she had no other ocular or systemic complications, she was asked to come for periodic follow-up in eye clinic mainly focusing on her visual impairment caused by any lenticular changes, and IOP measurement to prevent advancement of any glaucomatous optic neuropathy changes.

DISCUSSION

Aniridia is a rare genetic disease which can have ocular as well as systemic associations. Here, we screened three family members, among which daughter and father were found to be affected. Genetic analysis forms the backbone to identify this disease, but due to financial constraint, in this case diagnosis was solely based on thorough clinical ocular and systemic examination. This family presented with a plethora of ocular manifestations associated with congenital aniridia and its complications having a dreadful impact on the vision which can adversely affect the patient's day to day life. The girl had congenital isolated aniridia with no other complications, and these types of cases can

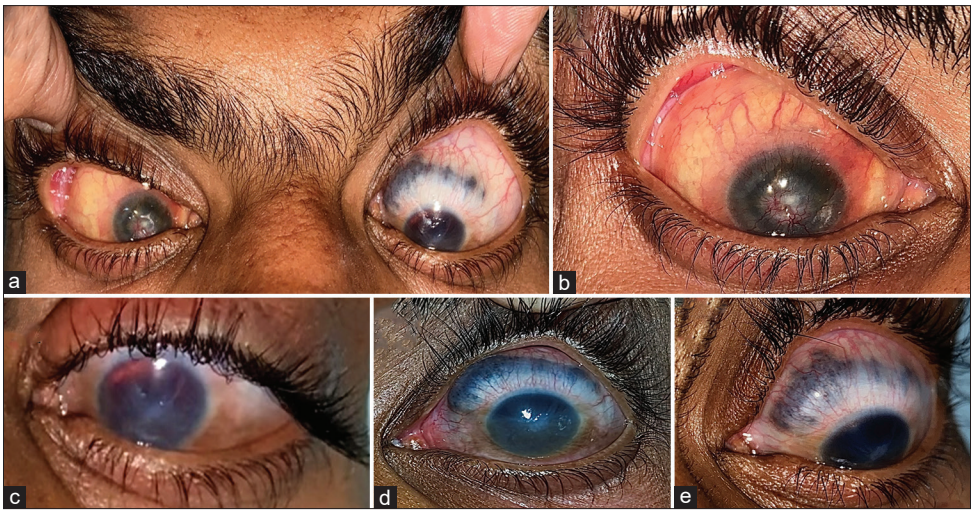

Figure 3: Facial profile of the father of index case (a) with right eye showing central leukoma with peripheral scarring and corneal vascularization (b). Left eye (c) showing diffuse corneal opacity and scarring involving central and inferior cornea suggestive of aniridia-associated keratopathy with rudimentary iris seen superiorly along with ciliary staphyloma in superiorly (d and e).

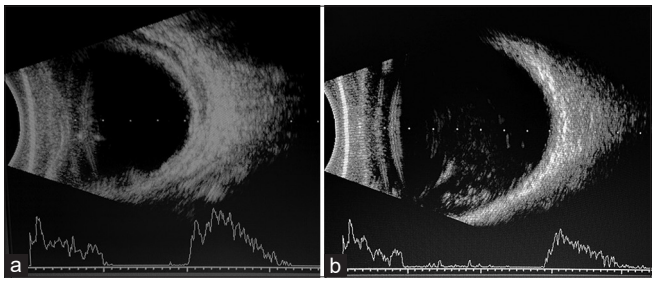

Figure 4: Ultrasound B scan of both eyes of father showing reduced axial length with thickened ocular coats in the right eye suggestive of pre-phthisical state (a). Left eye showing posterior vitreous detachment, low reflectivity vitreous echoes with attached retina (b)

be treated by correction of refractive error, tear substitute and periodic follow-up. However, her father was a case of aniridia having other ocular complications as well. These cases need to be treated for glaucoma, cataract extraction, tear substitute, and keratoplasty as per the need of the complication present in the affected individual.

CONCLUSION

Aniridia even being rare can have impact on visual outcome to a great extent and it may vary in severity from one person to another. However, proper diagnosis, timely intervention, and regular periodic follow-up are necessary for early visual rehabilitation. Apart from that, family screening is essential, along with genetic testing and counseling to raise more awareness.

AUTHORS CONTRIBUTIONS

The manuscript has been read and approved by all the authors that the requirements for authorship as stated earlier in this document have been met and that each author believes that the manuscript represents honest work.

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