Congenital aniridia with ectopia lentis and unilateral buphthalmos: an unusual presentation

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DESCRIPTION

A 7-year-old boy presented with poor vision in the right eye (RE) since infancy and diminution of vision in the left eye (LE) since 2 years. His best-corrected visual acuity at presentation was no light perception in RE and 20/80 in LE. Intraocular pressures (IOP) with Goldmann applanation tonometer were 34 mm Hg and 26 mm Hg in RE and LE, respectively. Anterior segment evaluation of RE revealed an enlarged eyeball with axial length of 28 mm and megalocornea with horizontal and vertical diameters of 14 mm and 13 mm, respectively, (figure 1A). The cornea also showed changes of aniridia-associated keratopathy (AAK).[1] There was complete absence of iris tissue in RE that was verified on gonioscopy and lens was subluxated superiorly (figure 1B). The LE had a normal corneal diameter with rudimentary iris stumps temporally and superior lens subluxation with broken zonules inferiorly (figure 1C,D). Fundus evaluation (figure 1E,F) revealed a total glaucomatous optic atrophy (GOA) in the RE and a vertical cup–disc ratio of 0.5:1 in LE. There was also absence of a foveal reflex in both eyes (BE). Optical coherence tomography revealed grade 4 foveal hypoplasia in BE (figure 1G).[2] Detailed systemic evaluation of the child was unremarkable.

Based on the examination findings, a diagnosis of congenital aniridia with bilateral ectopia lentis and right buphthalmos was made. The child was started on a combination of topical dorzolamide (2%) and timolol (0.5%) two times per day in LE. Poor prognosis was explained in RE. At 6-month follow-up, IOP in LE was 14 mm Hg on two topical antiglaucoma medications. He was advised 4-weekly IOP monitoring and periodic systemic evaluation.

Congenital aniridia is a panocular developmental disorder characterised by either partial or complete hypoplasia of iris. The genetic basis in 90% cases is a haploinsufficiency of the PAX6 gene at 11p13 by intragenic mutation or chromosomal rearrangement.[1] Aniridia patients warrant detailed systemic evaluation as they are associated with syndromes such as Wilms tumor–aniridia–genitourinary abnormalities–retardation) at 2.4% and Gillespie syndrome.[2 3] Glaucoma usually develops in late childhood with a reported incidence of 6%–75%. It may be due to progressive anatomical changes in

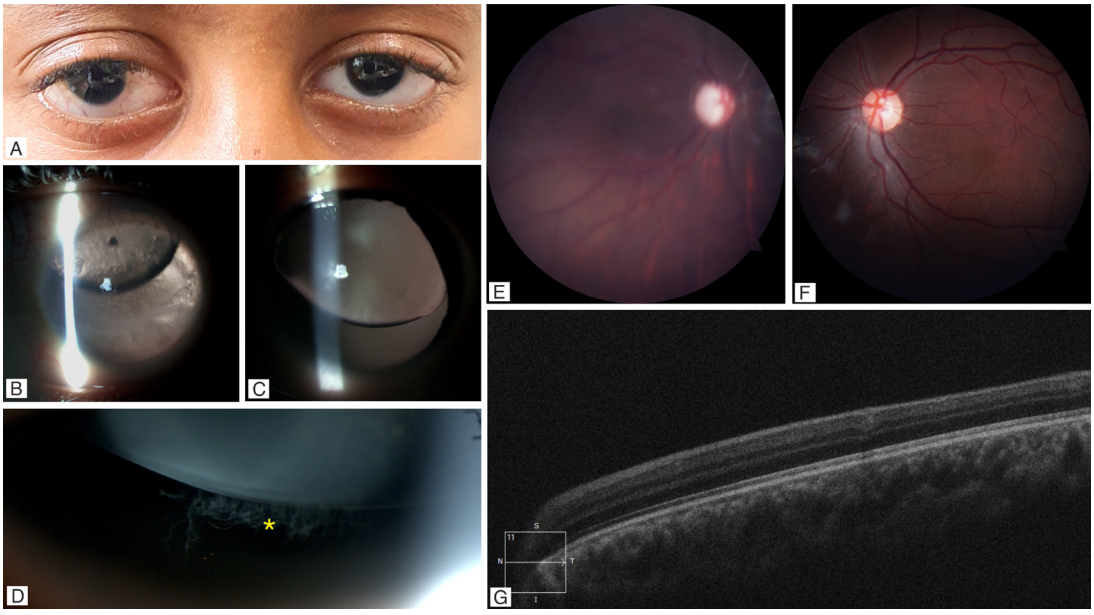

Figure 1 Diffuse light photography (A) showing enlarged right eyeball with excess inferior scleral show in the right eye (RE) and normal-sized left eye (LE). Slit lamp photography with retroillumination (B and C) showing bilateral superior subluxation of lens and aniridia-associated keratopathy. (D) High magnification (×40) photograph visualising the broken inferior zonules (yellow star) of LE. (E and F) Fundus photograph of both eyes revealing absent foveal reflexes with total glaucomatous optic atrophy in RE and a normal cup-disc ratio in LE. (G) Optical coherence tomography of LE showing grade 4 foveal hypoplasia.

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the drainage angles, due to absence of Schlemm's canal or due to the abnormality of the trabecular meshwork. Other recognised mechanisms are progressive angle closure with peripheral anterior synechiae and aniridia fibrosis syndrome secondary to intraocular surgeries.[4–8] Congenital glaucoma associated with or without buphthalmos is rare in aniridia.[5–7] The ocular phenotype can vary between and within families in aniridia, but affected individuals usually have similarity between the two eyes.[1] Asymmetry of ocular phenotype is not a rarity in syndromic cases and has been reported in the literature.[9 10] Our patient had an asymmetric ocular phenotype with buphthalmos and loss of vision secondary to total GOA seen only in the RE.

AAK vary from mild peripheral to pancorneal vascularisation, opacification and/or keratinisation due to limbal stem deficiency.[5 11] Ectopia lentis has been reported variably in literature in cases of aniridia with a reported prevalence of 0%–56%.[5 12 13] It usually occurs in the superior direction. Foveal hypoplasia is a characteristic fundus finding in aniridia patients.[12 13] The initial treatment of glaucoma associated with aniridia is usually medical therapy, even though majority may eventually require surgical intervention for uncontrolled glaucoma.[1] Most aniridia patients can preserve useful vision with appropriate ophthalmic care, in spite of ocular abnormalities.

Patient's perspective

My son has no vision in his right eye since infancy. I brought my son to get his left eye evaluated. Eye doctors examined both of his eyes and informed me that my son's right eye has irreversible damage. I have also been explained the need for regular eye pressure check-up. They also apprised me that my son might need glaucoma surgery in left eye in future if his eye pressures are not controlled with medication.

Learning points

- Congenital aniridia may present with asymmetric ocular phenotype.
- Buphthalmos can occur in congenital aniridia leading to loss of vision due to glaucomatous optic atrophy if left untreated.
- Rigorous systemic evaluation is of utmost importance in cases of congenital aniridia to detect coexisting systemic abnormalities.

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