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Case report

Bilateral spontaneous anterior lens capsule ruptures in a child: A rare presentation of Alport syndrome

Dian Petrus van der Westhuizen *, Kelsey Vernon Stuart

Department of Ophthalmology, Kimberley Hospital Complex, South Africa

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ABSTRACT

Purpose: This report describes the rare case of a child with bilateral spontaneous anterior lens capsule ruptures as the presenting feature of Alport syndrome.

Observations: The clinical presentation, special investigations and surgical management of the child are described, accompanied by a brief discussion of the genetic basis for the ocular and systemic manifestations of Alport syndrome.

Conclusions: Bilateral spontaneous anterior lens capsule ruptures as the presenting feature of Alport syndrome has not been described before.

1. Introduction

Alport syndrome (AS) is a rare basement membrane disorder characterized by hematuria with progressive renal failure; sensorineural deafness; and ocular abnormalities of the cornea, lens and retina.[1] AS is estimated to occur with a frequency of 1 per 50 000 live births.[2]

The disease occurs as a result of various mutations in genes coding for type IV collagen – the major protein constituent of basement membranes. The type IV collagen α3α4α5 network is found in the basement membranes of the glomerulus, cochlea, cornea (Descemet's and Bowman's membranes), lens capsule and retina (Bruch's membrane). Defective synthesis of any of these α-subunits results in structurally unsound basement membranes and accounts for the clinical manifestations of the disease.[1]

Inheritance of AS is X-linked in 85% of cases and occurs due to mutations in the *COL4A5* gene which codes for the α5 subunit of type IV collagen.[1,3] Autosomal recessive inheritance accounts for the remaining 15% and occurs as a result of mutations in the *COL4A3* and *COL4A4* genes, coding for the α3 and α4 subunits, respectively.[1,4] Autosomal dominant AS is very rare and remains a poorly defined clinical entity.[5]

The characteristic ocular manifestations of AS are anterior lenticonus, a peri-macular and/or peripheral fleck retinopathy, and temporal macular thinning.[1] Less commonly described features include recurrent corneal erosions, posterior polymorphous corneal dystrophy, posterior lenticonus, cataract, macular holes and other maculopathies.[1] Keratoconus has recently been described to occur in association with AS.[6]

Anterior lenticonus results in progressive lenticular myopia and, rarely, may result in spontaneous anterior lens capsule rupture, with or without rapid cataract formation.[7] The clinical presentation and surgical management of a case of bilateral spontaneous anterior lens capsule ruptures is described.

2. Case report

An eight-year-old male presented to the Ophthalmology outpatient department with the complaint of a progressive decrease in visual acuity (VA) and discomfort in his left eye over the preceding three months. There was no history of trauma. He had no significant medical history and a systemic examination was unremarkable. His mother described a family history of an unknown kidney disease in three of her maternal male cousins, all of whom had died before their third decade.

On examination, uncorrected distance VA (UDVA) was 20/30 and best-corrected VA (BCVA) was 20/20 in the right eye (manifest refraction −0.75/-0.75 × 34°). The left eye could only distinguish hand movements (refraction not possible). The left eye was mildly injected with an obvious leukocoria.

Slit-lamp biomicroscopy revealed a ruptured left anterior lens capsule with lens material protruding into the anterior chamber (see Fig. 1). The lens appeared hydrated and swollen, with cataract formation that precluded examination of the posterior segment. The intraocular pressure (IOP) was 38 mmHg.

Examination of the right eye demonstrated anterior lenticonus and a

* Corresponding author. Department of Ophthalmology, Kimberley Hospital Complex, Du Toitspan Road, Kimberley, 8301, Northern Cape, South Africa.
E-mail address: dianvdw@gmail.com (D.P. van der Westhuizen).

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central circular anterior lens capsule rupture, associated with early anterior polar cataract formation (see Fig. 1). Despite the clinically-evident capsular defect, lens material remained contained within the capsule. The cornea was clear and there was no peri-macular or peripheral fleck retinopathy.

Examination of the child's biological mother revealed only microscopic hematuria and no ocular abnormalities.

The child was subsequently taken to theatre where phacoemulsification of lens material and primary insertion of a +20.0D three-piece, acrylic intra-ocular lens (IOL) was performed under general anesthesia (based on the SRK-T formula with a 2.0D under-correction). The anterior lens capsule defect was of sufficient size to allow for complete extraction of lens material and placement of an IOL in the capsular bag, without the need for capsulorhexis. Post-operative UDVA was 20/30 and BCVA 20/20 (manifest refraction +2.50/-1.75 × 155°) in the left eye the day after surgery.

Audiological testing demonstrated bilateral moderate-to-severe high frequency sensorineural hearing loss. Urinalysis revealed proteinuria and microscopic hematuria with red blood cell casts. Electron microscopic analysis of a renal biopsy showed the characteristic basement membrane changes associated with AS and confirmed the diagnosis. The child was referred to the appropriate pediatric and renal services for long-term management. Both he and his mother were referred to the regional genetics service for further testing and counseling, with subsequent confirmation of X-linked AS.

Six weeks later, vision in the right eye had worsened to an UDVA of 20/60 with BCVA of 20/30. Examination revealed supero-nasal extension of the anterior capsular defect with bulging and opacification of the underlying lens cortex (see Fig. 2). Phacoemulsification with primary insertion of a +20.0D three-piece, acrylic IOL was again performed under general anesthesia (based on the same IOL calculation as used in the left eye). At surgery the lens capsule was found to be excessively elastic with a central anterior capsular defect. A capsulorhexis was successfully fashioned by extension and incorporation of the pre-existing defect, and an IOL was placed in the capsular bag. Post-operative UDVA was 20/30 and BCVA 20/20 in the right eye the day after surgery. Post-operative refraction and long-term visual outcomes were not available as the child and his mother were lost to further follow-up.

3. Discussion

The characteristic ocular manifestations of AS (anterior lenticonus, fleck retinopathy and/or temporal macular thinning) occur in >50% of males with X-linked AS and in >75% of individuals with autosomal recessive AS.[1] These ocular features are often not present in childhood and tend to become more prevalent with time and disease progression.[1] Due to the effects of lyonization, ocular manifestations are far less prevalent in females with X-linked AS.[1] Many of the ocular features of AS have no effect on vision, but are useful diagnostically and to determine mode of inheritance.[1] It is estimated that 40% of children with X-linked AS may be diagnosed based on ocular features alone.[8] Anterior lenticonus is pathognomic for AS and is associated with early-onset renal failure.[1]

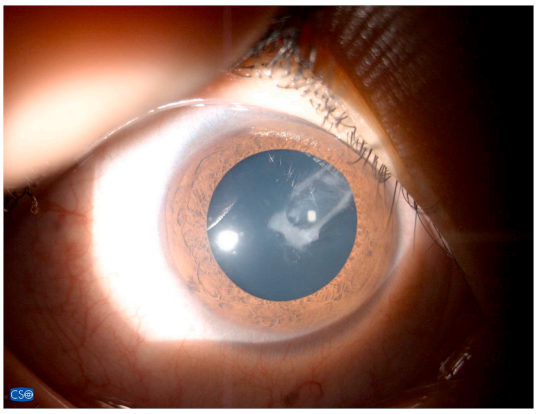

Fig. 2. Follow up six weeks later. Slit lamp biomicroscope picture of right eye demonstrating supero-nasal extension and opacification of anterior capsule defect.

Anterior lenticonus leads to visual disturbance as a result of progressive lenticular myopia. Very rarely, the anterior lens capsule may rupture – either spontaneously or following minor trauma. Subsequent cataract formation may be rapid, with release of lens matter into the anterior chamber, or may result from progressive opacification of lens material underlying the capsular defect. To the best of our knowledge only fourteen cases of anterior lens capsule rupture in AS have been reported in the past fifty years (see Table 1). Although a few of these cases went on to develop subsequent spontaneous anterior lens capsule rupture in the fellow eye,[9] bilateral spontaneous ruptures as the presenting feature of AS has not been described before.

Surgical management of anterior lenticonus is usually indicated when lenticular myopia or anterior subcapsular cataract result in significant visual impairment. Clear lens phacoemulsification with IOL implantation has been shown to be a safe and effective option in such cases with excellent visual outcomes.[7,10] The procedure does, however, tend to be technically more difficult, primarily as a result of lens capsule fragility. Electron microscopic analysis has shown the lens capsule in AS

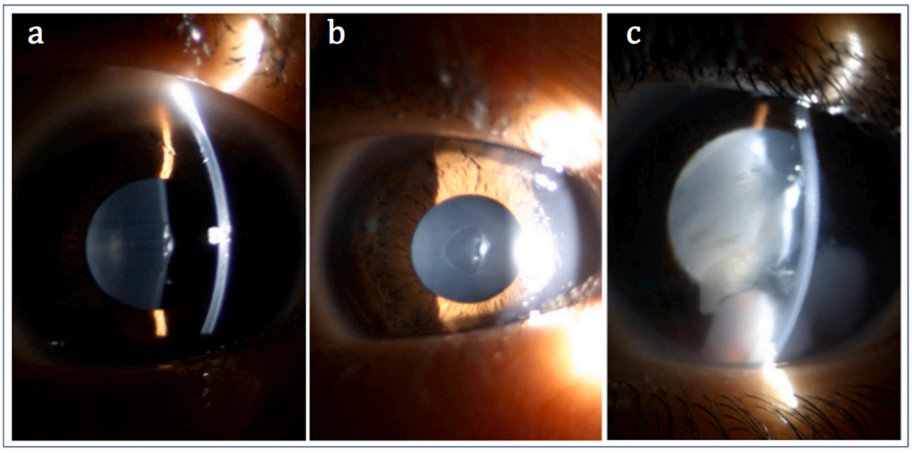

Fig. 1. Initial presentation. Slit lamp biomicroscope pictures of right eye demonstrating a) anterior lenticonus and b) central anterior capsule defect, and c) left eye demonstrating anterior capsule rupture with release of lens material.

Table 1
Previously published reports of anterior lens capsule rupture (spontaneous and traumatic) in Alport syndrome (since 1969).

Year	Author	Country	Number of cases
1980	Ignat'ev	USSR	1
1991	Teekhanaesee et al.	Thailand	1
1998	Oto & Aydin	Turkey	1
1999	Olitsky et al.	USA	2
2001	Sathish et al.	India	3
2004	Pelit et al.	Turkey	1
2006	Wilson et al.	USA	1
2007	Zare et al.	Iran	1
2011	Gupta et al.	India	1
2015	Agrawal et al.	India	1
2017	Trifonova et al.	Bulgaria	1

to be significantly thinned, with numerous internal dehiscences containing vacuoles and fibrillar material, especially at the poles.[10]

Various recommendations for a surgical approach in such cases have been suggested, and include: performing continuous curvilinear capsulorhexis (CCC) after aspiration of cortical lens material; beginning the CCC in the mid-periphery of the lens capsule rather than in the center; gentle but thorough hydrodissection of the lens nucleus; and avoiding polishing of the posterior lens capsule.[10,11]

In cases of anterior lens capsule rupture, surgical approach depends on the size and extent of the capsular rupture. A CCC may be fashioned around, or incorporated into, a small central defect; whereas larger defects may provide adequate access to allow phacoemulsification and IOL insertion without capsulorhexis.

AS should be considered in any patient presenting with anterior lenticonus or rupture of the anterior lens capsule, even if other features of the syndrome are not present.

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Authorship

All authors attest that they meet the current ICMJE criteria for Authorship.

Patient Consent

Written informed consent to publish this case was obtained from the patient's biological legal parent. This report does not contain any personal identifying information.

Declaration of competing interest

The following authors have no financial disclosures: DPvdW, KS

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