
Surgical management of bilateral anterior lenticonus and Giant macular hole in Alport syndrome

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Abstract: To report a rare case 35 yr old female of Alport syndrome presenting with bilateral anterior lenticonus associated with cataract and giant macular hole, managed surgically with successful outcome.

Keywords: Alport syndrome, anterior lenticonus, giant macular hole.

INTRODUCTION

Alport syndrome is an inherited disease characterized by progressive renal failure, hearing loss, and ocular abnormalities. It occurs in 1 in 5000 to 50,000 live births, can affect both sexes; however, severe disease and increased rate of progression are more common in males. The inheritance is predominantly X-linked (85%), although it can be autosomal recessive (10%) or autosomal dominant (5%). Mutations in the COL4A5 (X-linked), or COL4A3 and COL4A4 (autosomal recessive) genes result in absence of the collagen IV $\alpha3\alpha4\alpha5$ network from the basement membranes of the cornea, lens capsule, and retina and are associated with corneal opacities, anterior lenticonus, fleck retinopathy, and temporal retinal thinning. The rarer ophthalmic complications are posterior polymorphous corneal dystrophy and giant macular hole.

Authors' hereby describe a rare presentation of Alport syndrome, with bilateral anterior lenticonus and giant macular holes; managed surgically, with a successful outcome.

CASE REPORT

A 35-yr-old female presented with complaints of gradual progressive, painless, decrease in vision in both the eyes since 1 year. She had no significant past ocular history or history of trauma. Her best corrected visual acuity (BCVA), both eyes was counting fingers at 1 meter for distance and near vision <N36. Slit lamp examination showed bilateral anterior lenticonus with anterior sub capsular cataract. (Figure 1 and Figure 2). Fundus examination, both eyes revealed bilateral giant macular holes measuring about 2 disc diameters in size. There was an area of surrounding retinal pigment atrophy around the hole. Optical coherence tomography (OCT) showed bilateral large macular holes with elevated edges, measuring about 3010 × 1960 μ in the

right eye and 4250 × 2250 μ in the left eye (Figure 3 and 4). In addition she also had bilateral sensorineural deafness. Nephrology consultation was also sought

In view of significant cataract and long standing macular hole she underwent combined phacoemulsification with intraocular lens implantation with pars plana vitrectomy, internal limiting membrane peeling and fluid gas exchange in both the eyes. A standard 2.8 mm phacoemulsification through clear corneal incision was performed. Anterior capsulorhexis was made with rhexis holding forceps as the capsule was elastic. The rhexis extended for 2-3 clock hours but was continuous and completed meticulously. Stop and Chop technique was used to remove the nucleus. Automated bimanual aspiration was performed for cortex removal. In the bag fixation of foldable, hydrophilic, acrylic, single piece intraocular lens (RYCFLENS, Biovision limited) implantation was done. A standard 23G three port pars plana vitrectomy was done. After core vitrectomy, triamcinolone assisted posterior vitreous detachment was carried using microvitreous cutter. The internal limiting membrane was stained using brilliant blue dye and removed subsequently using ILM peeling forceps. At 1 month follow up, early posterior capsular opacification (PCO) was noticed for which she underwent ND: YAG laser capsulotomy (Figure 5). On fundus examination the size of macular hole was small and it showed type 2 closures (figure 6a and 6b) which were confirmed by OCT (Figure 7). The final visual outcome in the right eye was 20/320, N 18 at the end of 1 month. Similar surgery was repeated in the left eye. Capsulorhexis was difficult but was continuous. In the bag fixation of foldable, hydrophilic, acrylic, single piece intraocular lens implantation was performed. ILM peeling posed difficulty in this eye, since it was sticky and came out piece meal. Post-operatively, there was persistence of macular hole with type 2 closure (figure 8a and 8b),

confirmed by OCT (figure 9). The BCVA, left eye, 1 month post-operatively was 20/320, N18 and the IOL was in place with trace PCO (figure 10).

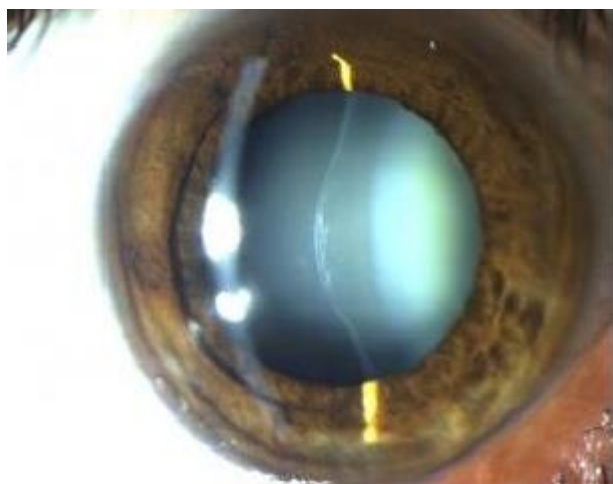


Fig 1: Pre-operative slit lamp photograph of right eye showing anterior lenticonus



Fig 2: Pre-operative slit lamp photograph of left eye showing anterior lenticonus



Fig 3: Pre-operative OCT of the Right eye

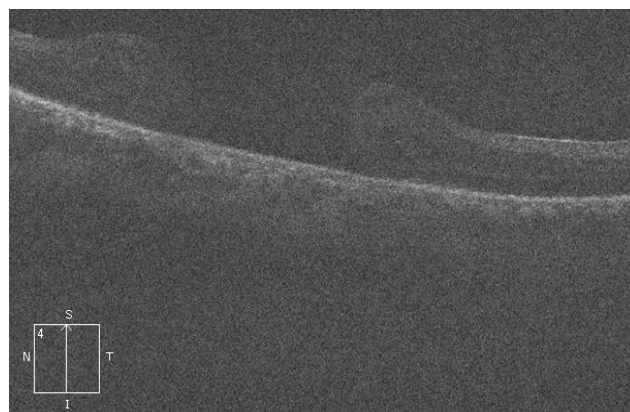


Fig 4: Pre-operative OCT of the left eye

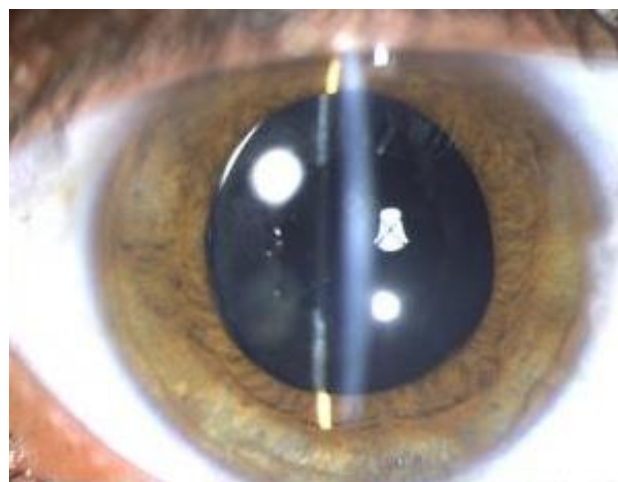


Fig 5: Post-operative slit lamp photograph of Right eye showing Yag opening



Fig 6a: Post-operative fundus photo of Right eye showing giant macular hole

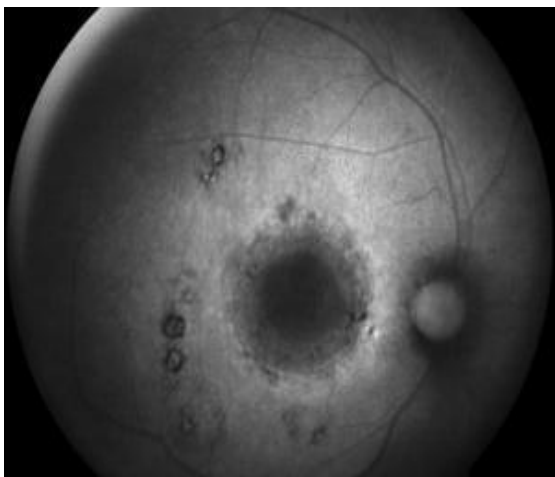


Fig 6b: Post-operative fundus auto fluorescence photo of Right eye showing giant macular hole

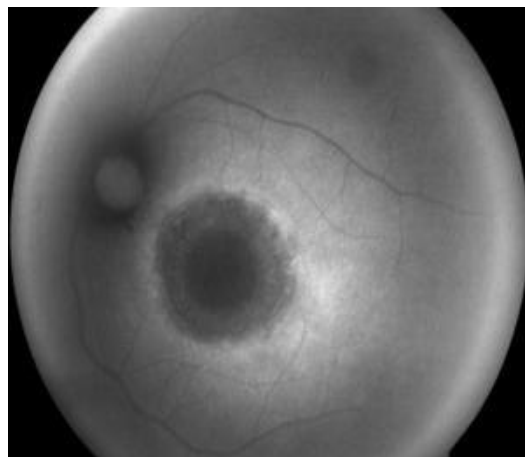


Fig 8b: Post-operative fundus auto fluorescence photo of left eye giant macular hole

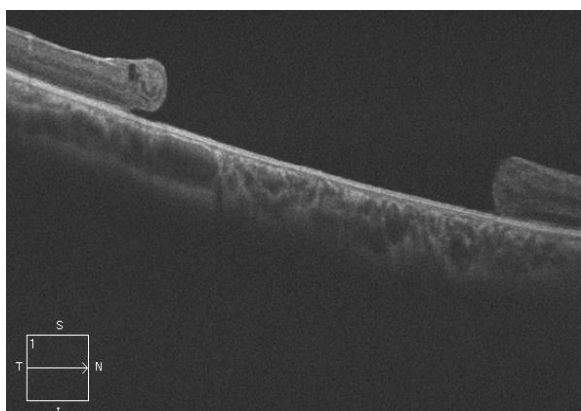


Fig 7: Post-operative OCT of the Right eye showing full thickness giant macular hole

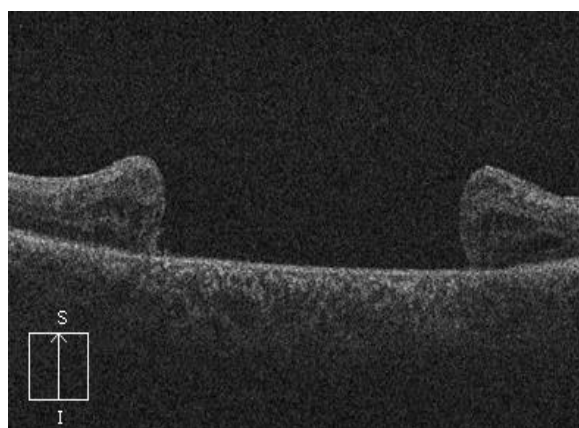


Fig 9: Post-operative OCT of the left eye showing full thickness giant macular hole

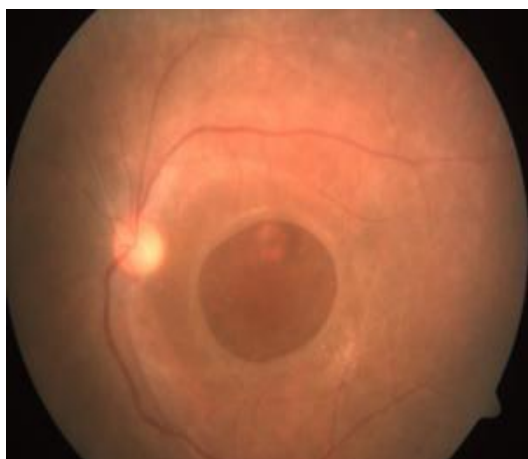


Fig 8a: Post-operative fundus photo of left eye giant macular hole

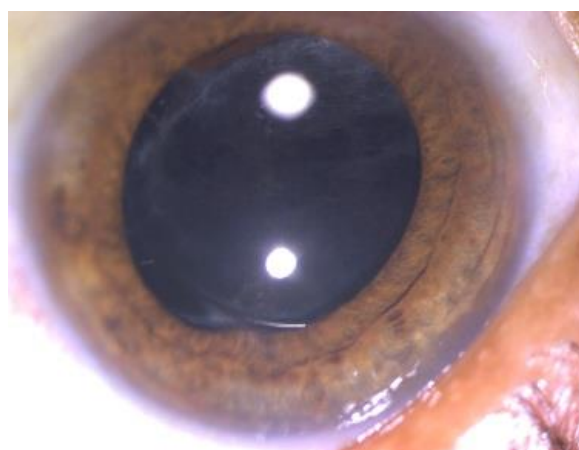


Fig 10: Post-operative slit lamp photograph of left eye

DISCUSSION

Alports syndrome is characterized by renal failure, hearing loss, lenticonus and retinopathy. The common ocular manifestations are anterior lenticonus and dot fleck retinopathy. Less common manifestations are posterior lenticonus, posterior polymorphous dystrophy and cataract. The formation of lenticonus is due to the defect in type IV collagen which cause partial

dehiscence's in the capsule with fibrillar material. This makes the anterior capsule fragile with progressive thinning and protrusion [1]. The anterior capsule can also be at stress during accommodation and becomes more convex and it may sometimes lead to rupture [2]. In our case, a rare presentation of bilateral giant macular holes is seen. Presence of giant macular holes was previously reported in very few studies [3-5]. The retinal abnormalities result from thinning of the ILM, nerve fiber layer and the RPE basement membrane of Bruch's membrane. This thinning affects the nutrition of the overlying retinal layers and cause overall thinning and development of macular hole [6]. Some authors thought that passage of fluid through abnormal Bruch's membrane and atrophic RPE which led to formation of microcystic centres in the retinal layers. Further disruption of these confluent cysts led to full thickness macular hole [4].

In view of significant lens abnormalities and poor vision early cataract surgery is contemplated for early rehabilitation. A standard phacoemulsification with IOL implantation is considered to be safe in treating lenticonus with cataract [7, 8]. However, in our case, anterior lenticonus, cataract and giant macular hole were simultaneously addressed. Phacoemulsification with IOL implantation along with pars plana vitrectomy and ILM peeling with fluid gas exchange was done. Capsulorhexis was challenging because there was every chance of extension due to thin anterior capsule [9]. Posterior capsular rupture was also common in Alport syndrome [10]. In our case, conventional rhexis starting at the centre of the lens was done. There was enlargement of the rhexis than intended since the capsule was elastic and fragile. We used foldable acrylic IOL to reduce the chance of posterior capsular opacification. Acrylic IOL's were recommended in order to retard PCO formation [7]. But there was early PCO formation in the right eye for which she had to undergo ND: YAG capsulotomy. Macular hole surgery in Alport syndrome is technically difficult and challenging because they are very much different from the idiopathic holes. This is due to the large size of the hole and abnormal behavior of the ILM. The peeling of the ILM in our case was done with difficulty in the right eye and in piece meal in the left eye since it was sticky. In a case report by Miller JJ *et al.*; surgical difficulty is attributed to abnormal vitreoretinal interface and weakness of basement membrane but they reported a successful closure [11]. In our case the macular hole was large and long standing as the surrounding retina showed atrophic area. The ILM was sticky and difficult to peel. This might be the reason for type 2 closure. We can explain the post-operative prognosis of the macular surgery by the chronicity of the macular hole.

CONCLUSION

Our case is a rare presentation of Alport syndrome with bilateral giant macular holes and

anterior lenticonus with cataract. It was managed surgically by combined approach addressing the lens and retina with favourable visual outcome. Patients with Alport syndrome and lenticonus should be examined for macular hole and if detected early can be managed timely with good visual outcome.

Disclosure Statement

The authors declare that there is no conflict of interest regarding the publication of this paper.

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