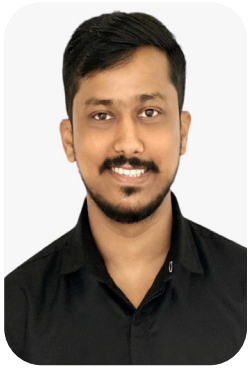
Article • Role of the Optometrist in the Management of Bilateral Anterior Lenticonus: A Case of Alport Syndrome

Krishna Shah, M. Optom • Sankara Nethralaya • Chennai, India
Janani Balaji, M. Optom • Sankara Nethralaya • Chennai, India
Madhumathi Subramanian, B.S. Optom • Sankara Nethralaya • Chennai, India



Krishna Shah, M. Optom
Chennai, India

Consultant Optometrist, Department of contact lens, Sankara Nethralaya, Chennai, India

Faculty at The Sankara Nethralaya Academy, Chennai, India

B.Optom, Bharati Vidyapeeth (Deemed to be University), Pune, India

M.Optom, The Sankara Nethralaya Academy, Chennai, Tamilnadu

ABSTRACT

Background: Alport Syndrome is a rare hereditary clinical condition that causes a disruption in collagen synthesis. It is associated with ocular findings most commonly involving the cornea, lens, and retina.

Case Report: An 18-year-old male presented with progressive bilateral vision loss and a history of hypertension and childhood-onset hearing loss. Retinoscopy revealed two distinct reflexes; further evaluation using slit lamp examination and anterior segment optical coherence tomography (AS-OCT) revealed anterior lenticonus. Corneal topography was also performed to rule out keratoconus or other forms of irregular astigmatism; the results were within normal limits, effectively ruling out corneal ectatic disorders. Genetic testing (COL4A5 mutation) was conducted and confirmed the diagnosis of X-linked Alport syndrome. For ocular management, a soft contact lens trial was performed, in which significant improvement in visual acuity was achieved by neutralizing the high myopic reflex from the central lenticonus.

Conclusion: This case highlights the importance of detailed retinoscopic assessment and optometric management in Alport syndrome.

Keywords: Alport syndrome, AS-OCT, contact lens, lenticonus, retinoscopy reflex

Introduction

Alport Syndrome is a rare hereditary clinical condition caused by a genetic type IV collagen disorder that disrupts collagen synthesis and is associated with renal failure (99%), hearing loss (82.5%), and ocular findings (44-81%), most commonly involving the cornea, lens, and retina.[1] Inheritance is predominantly X-linked (85%), with a prevalence of 1/5000; autosomal recessive in 10%; or autosomal dominant in 5%. Affected males typically experience renal failure and high-tone sensorineural deafness by the age of 20, and 85% of affected adult males have dot-and-fleck retinopathy.[1-4]

Retinoscopy provides valuable information regarding refractive errors by assessing the reflexes generated from the retinal plane. This tool has been in use for more than 100 years in the field of eye care, yet little has been published on its usefulness in determining appropriate values of objective refraction in cases of Alport syndrome.

This case report emphasises the clinician's expertise in using retinoscopy to assess and manage complex refractive errors. It also underscores the critical role of optometrists in the visual rehabilitation of patients with conditions like Alport syndrome and anterior lenticonus.

Case Report

An 18-year-old male presented to a tertiary eye care hospital with a gradual, progressive, painless, bilateral vision loss for the past ten years, with the right eye being highly symptomatic for the past two years. He had a medical history of hypertension for the past three years. The patient sought a second opinion and further management due to discomfort with the previously prescribed glasses. The manifest refraction was -0.75-0.50x180 (2/60) in the OD and -1.00-0.75x180 (6/60) in the OS. During retinoscopy, two different reflexes were observed: one from the central protruded area of the lens and another a peri-central reflex. Upon performing retinoscopy, the peri-central reflex was neutralised, the patient's manifest refraction was -0.75-0.50x180 OD and -1.00-0.75x180

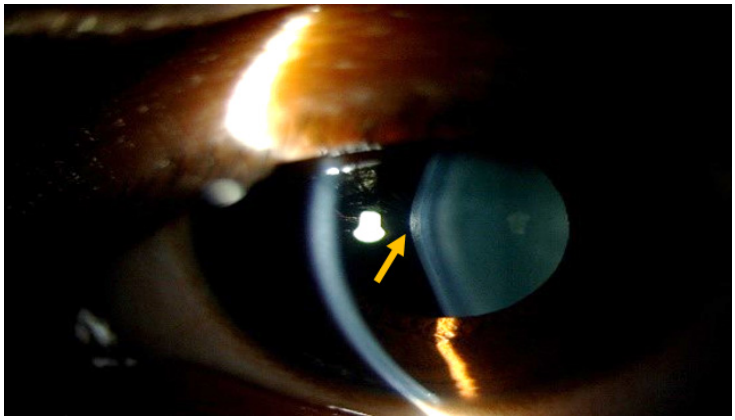


Figure 1. Optic section in slit lamp showing anterior lenticonus

OS with best-corrected visual acuity (BCVA) improved to 3/60 and 6/60, OD/OS respectively. A very small central area showing a very high myopic reflex was detected. On neutralising the reflex of the central protruded area with retinoscopy, the objective refraction manifested -12.00-3.00x20 OD and -8.00-2.00x160 OS, with which the patient was able to comfortably read 6/7.5 and 6/6 OD/OS, respectively. On slit lamp examination, a conical protrusion was observed in the pupillary axis of the anterior lens capsule OU, indicating a possible case of lenticonus with clear underlying lens details (Figure 1). Corneal topography was performed to rule out keratoconus or irregular astigmatism, and the results were within normal limits. Suspicion of Alport syndrome was made based on the clinical features. The patient was asked about the history of haematuria and deafness. Upon history-taking, the patient revealed decreased hearing since childhood but no macroscopic haematuria. The patient was referred for nephrology evaluation, which revealed proteinuria and mildly elevated creatinine, suggesting early renal involvement. Anterior segment optical coherence tomography (AS-OCT) revealed

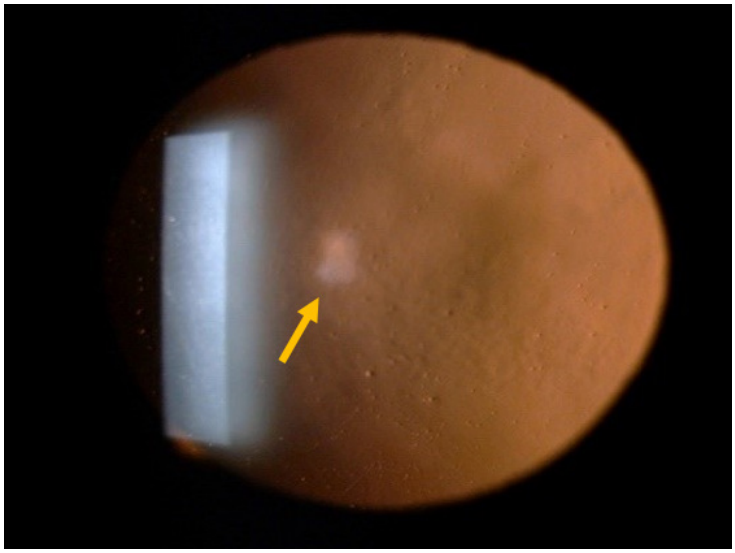


Figure 3. Oil drop reflex

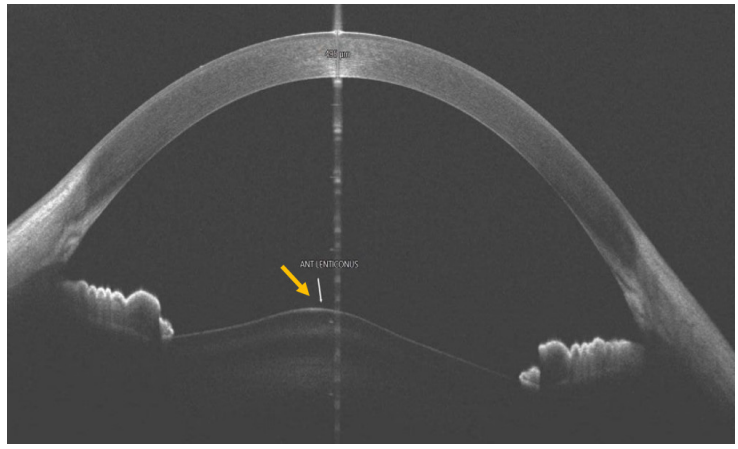


Figure 2. AS-OCT revealed the presence of anterior lenticonus

the presence of anterior lenticonus (Figure 2). An oil droplet reflex was observed (Figure 3). Dilated fundus examination revealed vitreous condensation and dot-and-fleck retinopathy. OCT revealed temporal retinal thinning in the right eye, while the left eye revealed vitreous condensation, along with mild dot-and-fleck retinopathy (Figure 4). The patient was referred for genetic testing (COL4A5 mutation), and it confirmed the diagnosis of X-linked Alport syndrome. In an effort to improve the patient's vision, a contact lens trial was performed in view of high myopic anisometropia. The details of the trial are outlined in Table 1. A spherical power monthly disposable silicon hydrogel contact lens with the above-mentioned parameters was dispensed, and the cylindrical power was given in spectacles over the contact lenses.

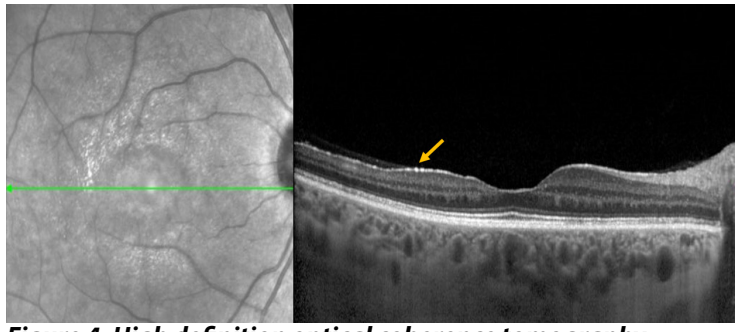


Figure 4. High definition optical coherence tomography

Table 1. Final Prescribed Contact Lens Parameters

Eye	Base	Power	Diam-eter	Material	Fit Assess-ment	VA (CLs with cyl power in spectacle)
OD	8.6	-10.50 D	14.0	Comfilcon A/	Optimal	6/7.5, N6
OS	8.6	-7.00 D	14.0	Monthly Disposable	Optimal	6/6, N6
				Comfilcon A/ Monthly Disposable		

Discussion

Alport syndrome is also known as hereditary nephritis or collagen IV-related nephropathies. It is associated with high-tone sensorineural deafness and eye abnormalities, including anterior lenticonus.[5] Lenticonus refers to an irregular curvature at the center of the lens, where the lens capsule appears cone-shaped.[6] This condition can occur either at the anterior or posterior portion of the lens. While posterior lenticonus is linked to Lowe's syndrome, anterior lenticonus combined with familial hemorrhagic nephritis is linked to Alport syndrome.[6,7]

In this case report, we observed central protrusion of the crystalline lens, lenticonus, which was confirmed through slit lamp examination and AS-OCT and which are consistent with the ocular manifestations of Alport syndrome.[8] These structural abnormalities can induce irregular astigmatism and high myopic refractive errors, leading to suboptimal visual outcomes.[9] We observed two different reflexes in retinoscopy: one for the central protruded area and one from the periphery. Significant improvement in vision was observed upon neutralising the high myopic reflex arising from the central small protruded area of the crystalline lens. Kurt et al. reported the association of high-degree myopia, corneal dystrophy, and deafness as an individual hereditary disease, which is consistent with the present case finding.[10]

Retinoscopy enables measurement of the refractive state. In keratoconus or highly astigmatic eyes, a non-homogeneous light-shadow movement is visible. At the corneal centre, the reflex will show slightly more myopia in comparison to the periphery. This opening and closing of the light reflex is called the scissors reflex. The scissoring reflex is more prominently noted in those with advanced disease like keratoconus and pellucid marginal degeneration. Additionally, numerous studies have documented the coexistence of posterior polymorphous corneal dystrophy (PPMD) and non-keratoconic astigmatism in conjunction with high-degree myopia. These ocular problems highlight the complexity of refractive assessments in patients with such conditions. Literature on retinoscopy, especially in Alport syndrome, is limited.[11]

The primary focus of this discussion is the successful improvement in visual acuity achieved through precise refractive correction. This was accomplished by neutralizing the retinoscopic reflex arising from the small central protrusion of the lenticonus crystalline lens observed during retinoscopy. Thus, in cases of anterior lenticonus, as seen in Alport syndrome, the role of the optometrist is crucial.

Conclusion

It is essential for clinicians/optometrists to neutralise the retinoscopic reflex in the central as well as the peripheral zone in cases of Alport syndrome. A precise refractive correction plays a key role in achieving better visual rehabilitation and also managing the high anisometropia with the help of contact lenses.

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Correspondence regarding this article should be emailed to Krishna Shah, M. Optom at krishna.researchid@gmail.com Shah K, Balaji J, Subramanian M. Role of the optometrist in the management of bilateral anterior lenticonus: A case of Alport syndrome. Optom Vis Perf 2026;14(1):32-34.