Case Report

Bilateral anterior lenticonus associated with Alport syndrome

Naurin Memon*, Divyang Patel, Kavita Shah

Department of Ophthalmology, Gujarat Adani Institute of Medical Sciences and GK General Hospital, Bhuj, Kachchh, Gujarat-370001
* Correspondence: Dr Naurin Memon (naurinmn@gmail.com)

ABSTRACT

Anterior lenticonus is a rare congenital anomaly of the eye characterized by a conical protrusion of the crystalline lens capsule and the underlying cortex in anterior chamber leading to high axial myopia; most cases are associated with Alport syndrome. Alport syndrome (AS) is an inherited disorder of many forms, most commonly X-linked. The main abnormality is deficient synthesis of type IV collagen, the main component of basement membranes. It typically presents with the classic triad of progressive glomerulonephritis, progressive high tone hearing loss, and several ocular signs; the most pathognomonic of which is the presence of anterior lenticonus. We report a case of a 21-year-old female with Alport syndrome presenting with gradual, progressive diminished vision in both eyes associated with anterior lenticonus and dot and fleck retinopathy with progressive sensorineural hearing loss.

Key words: Alport syndrome, Anterior Lenticonus, Dot and Fleck retinopathy, Glomerulonephritis, Sensorineural hearing loss, Type IV collagen

INTRODUCTION

Alport syndrome is a rare disorder of the basement membrane due to mutations involving the coding for type IV collagen resulting in defective synthesis; characterized by progressive hereditary nephritis, sensorineural hearing loss and ocular abnormalities.[1] Biochemical properties of normal capsule depend primarily on type IV collagen and other extracellular matrix constituents, such as collagen type I and III, laminin and fibronectin.[2] Defective type IV collagen eventually results in increased capsular fragility, thus leading to progressive forward bowing of capsule at its weakest point. It manifests as a slowly progressive deterioration of vision, resulting from high degree lenticular myopia.[3] Estimated prevalence of Alport syndrome is 1 case per 5000 people and 85% of patients have the X linked inheritance form, although there can be autosomal recessive (10%) or autosomal dominant (5%) inheritance. Men are affected more commonly and more severely. Nephritis with haematuria secondary to basement membrane disease of glomeruli is the most life-threatening aspect of this disorder. There is also progressive sensorineural hearing loss beginning with high frequencies. The most frequent ocular finding is progressive bilateral lenticonus and it occurs in approximately 25% of patients with X linked Alport syndrome, and when present, it may be a pathognomonic feature.[1-3]

CASE HISTORY

A 21-year-old female, consulted at Ophthalmology Department of a teaching hospital of western Gujarat with complaints of gradual, painless, diminished vision in both eyes for the past 10 years, with hearing impairment in both the ears for 8-9 years. On examination, the patient had best corrected visual acuity (BCVA) of 3/60 in right eye and 1/60 in left eye with refraction -8.00sph/-6.00cyl at 20-degree axis and -10.50sph/-3.0cyl at 180 degrees respectively. Pupils measured 4-5mm in size and reacted sluggishly to light. Both eyes showed nystagmus with alternate esotropia. Direct fundoscopy showed red-orange reflex with the characteristic appearance of an oil-droplet reflex in the centre of the pupils (Figure-1). This was also seen on slit-lamp examination through pharmacologically dilated pupils via retro-illumination. Both lenses were free of any opacity; anterior budging of the lenses was prominent, highly suggestive of anterior lenticonus (Figure-2). On indirect ophthalmoscopy, the optic disc in both eyes showed temporal pallor with

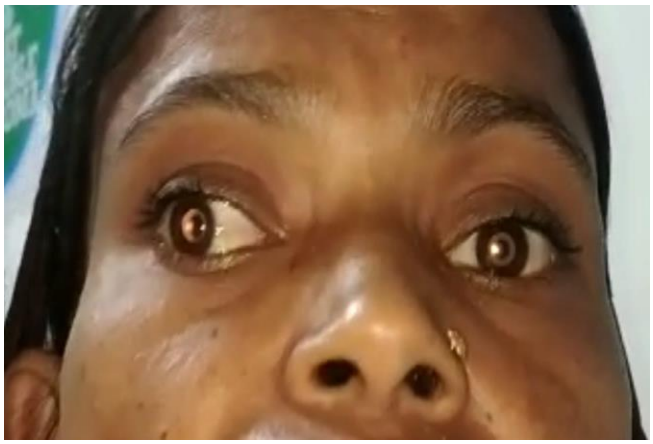

Figure-1: Oil droplet sign

peri-macular dot and fleck retinopathy (Figure-3). Pure tone audiometry (PTA) with speech testing confirmed bilateral sensorineural hearing loss. Complete blood count showed normal values. Serum blood urea nitrogen (BUN) and creatinine levels were elevated. BUN: creatinine ratio was 6, indicating an intrinsic renal disease. Urine examination showed marked haematuria and proteinuria. Chest X-ray and ECG were normal. Patient was advised to undergo clear lens extraction with intra-ocular lens (IOL) implantation in both the eyes and was referred to nephrology and otorhinolaryngology department for further management. Patient did not give any family history except for her first-degree aunt who was suffering from renal failure.

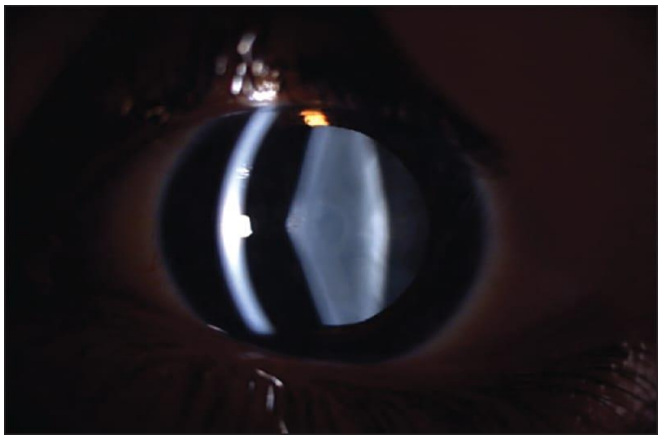

Figure-2: Slit lamp examination showing anterior protrusion of lens

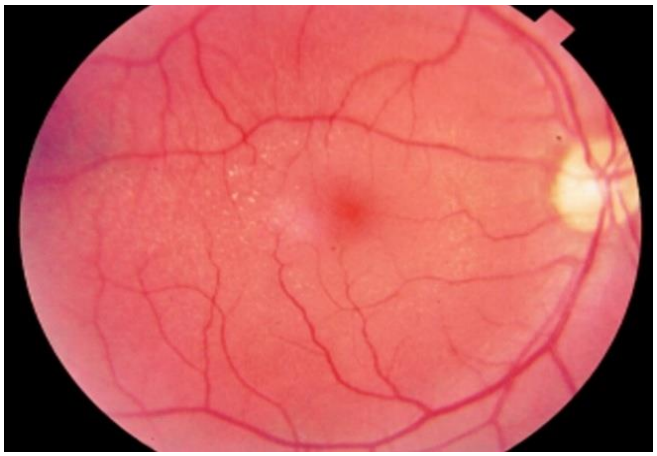

Figure-3: Fundus picture showing peri-macular dot and fleck retinopathy

DISCUSSION

Alport syndrome is an uncommon disorder characterized with ocular anomalies, progressive nephritis and sensorineural hearing difficulties. Development and progression of myopia and astigmatism are secondary to structural changes in one or both lenticular surfaces.[4,5] In the present case, there is high axial myopia with high lenticular astigmatism in both eyes, our case had findings of both eye temporal disc pallor with both eyes' nystagmus, which is a marker of poor visual prognosis. The patient was advised to go under clear lens extraction with IOL implantation.

CONCLUSION

There should be a high degree of suspicion for Alport syndrome in any patient who presents with anterior lenticonus, and thorough history and physical examination is necessary to establish the diagnosis. Slit lamp examination through a dilated pupil should be performed. There is no specific treatment for Alport syndrome. The treatment of choice for anterior lenticonus is clear lens extraction with implantation of IOL; a foldable, hydrophobic, acrylic IOL can be inserted into the capsular bag.

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