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CASE REPORT

A case of Alport syndrome presented with bilateral anterior lenticonus

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Purpose: The aim of this study was to report a rare case of Alport syndrome presented with bilateral anterior lenticonus in a 16-year-old boy.
Case report: A 16-year-old boy presented with decreased vision, a hearing defect, anemia, and proteinuria. His best corrected visual acuity was 6/18 in both eyes. Slit lamp biomicroscope showed anterior lenticonus in both eyes. He was managed by correction of refractive error and urgent referral to a nephrologist.
Conclusion: It is easy to diagnose Alport syndrome clinically, and close communication among ophthalmologists, otorhinolaryngologists, and nephrologists is crucial for effective management of this syndrome.

Keywords: Alport syndrome, anterior lenticonus, collagen type-IV, X-linked inheritance

Introduction

An geneticcondition of the basement membrane is known as Alport syndrome. Clinically, this syndrome is characterized by visual disturbance, hemorrhagic nephritis, and sensory neural deafness. In 1927, A. Cecil Alport reported that defective formation of type IV collagen is the main cause of this syndrome, and this type of collagen is present in the kidneys, inner ears, and eyes. Males are usually more affected by Alport syndrome in an X-linked inheritance, but in autosomal inheritance (both recessive and dominant), both sexes are equally affected. Symptoms of this syndrome include hematuria; proteinuria; hypertension; swelling of the legs, ankles, abdomen, and around the eyes; hearing loss; skin problems; nerve problems such as polyneuropathy; ocular lesions; and low blood platelet counts that compromise blood clotting. Bilateral anterior lenticonus is a hallmark of ocular manifestations. Other ocular changes include cataracts, central and midperipheral retinal flecks, corneal arcus, posterior lenticonus, recurrent corneal erosion, posterior polymorphous dystrophy, involuntary eye movements, and macular degeneration. Here, we present the detailed ocular findings and systemic problems of a 16-year-old boy with Alport syndrome.

Case report

A 16-year-old boy presented with decreased vision, a hearing defect, anemia, hematuria, and proteinuria (**Figure 1**). His highest corrected visual acuity in both of his eyes was 6/18. In the right eye, cycloplegic refraction revealed + 2.5 DS, and in the left eye, + 1.25 DS/ + 1.00 Cyl 1300. The Bruckner's test showed an oil droplet signs in both eyes. Slit lamp biomicroscopy showed anterior lenticonus in both eyes (**Figures 2**, **3**). A fundoscopy revealed no abnormalities. A routine blood examination showed Hb 9.5 gm/dl and ESR 27 mm in the first hour. Serum urea was 58 mg/dl and creatinine was 2.3 mg/dl. A routine urine examination showed 200 erythrocytes per high power field. The total urine protein was 1568 mg and 24-h urine volume was 2300 ml. The blood pressure was 100/70 mm Hg. An ultrasonogram of the kidney, ureter, and bladder (KUB) region was normal. An audiogram showed moderate mixed hearing loss in both

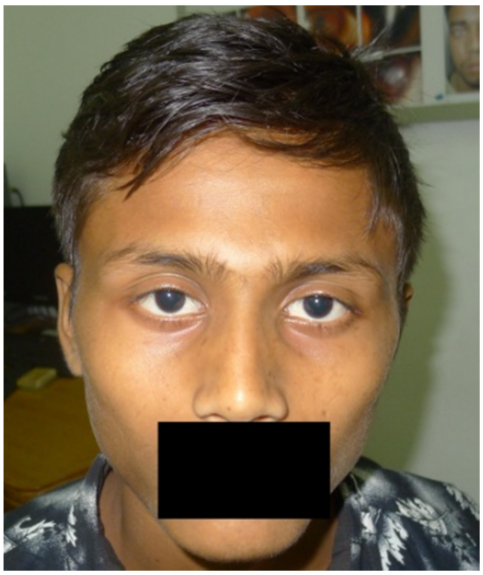

FIGURE 1 | Patient with Alport syndrome.

ears. He was given optical correction for vision improvement and referred to a nephrologist for further management.

Discussion

An uncommon genetic condition of the basement membrane is known as Alport syndrome. More than 80% of Alport syndrome cases are X-linked recessive where males are mostly affected and the remaining 10–15% are of autosomal inheritance where both males and females are equally affected (1, 2). Mutation in the *COL4A5* gene on X-chromosome encoded for type IV collagen causes defective formation of this collagen in the eye, inner ear, and glomerular basement membrane of the kidney (3). The *COL4A3* and *COL4A4* gene mutations that cause autosomal Alport syndrome (both recessive and dominant) are located on chromosome number 2 (4). The prevalence of Alport syndrome has been estimated at 1:10,000 in live birth for X-linked and 1:50,000 in live birth for autosomal inheritance (5).

Type IV collagen is found in the cochlea of the inner ear, the lens capsule, the Descemet's and Bowman's membranes of the cornea, the internal limiting membrane of the retina, and the basement membrane of kidney glomeruli. Hematuria and proteinuria were caused by an uneven thickening of the basal lamina, which was seen in the glomerular basement membrane's ultrastructure (6). Recurrent corneal erosion and posterior polymorphous corneal dystrophy are caused by defective Descemet's membrane and the corneal epithelium's basement membrane (7). Weakness of the anterior lens capsule and Bruch's membrane causes anterior lenticonus and retinal flecks, which are characteristic features of Alport syndrome. Hematuria, proteinuria, hearing defects, anterior lenticonus, and chronic kidney disease were noted in this case. Although

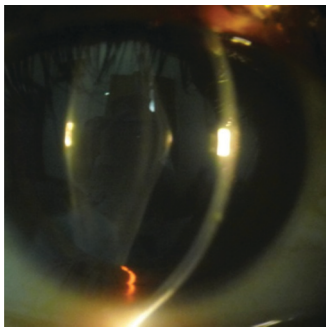

FIGURE 2 | Right anterior lenticonus.

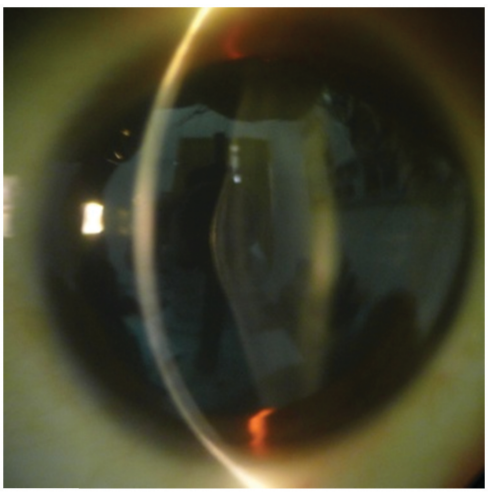

FIGURE 3 | Left anterior lenticonus.

retinal flecks in the retina or macula are the commonest finding of Alport syndrome, fundoscopy revealed normal findings. Aldosterone inhibitors, angiotensin converting enzyme inhibitors, and angiotensin receptor blockers can lower proteinuria (8). To enhance vision, anterior lenticonus coupled with cataracts requires cataract extraction with an intraocular lens (3). Hearing aids are required to deal with hearing loss, although fleck retinopathy does not require therapy. Alport syndrome patients must have ongoing therapy for renal failure (hypertension, proteinuria, hematuria, etc.), and some require dialysis or a kidney transplant. Alport syndrome does not recur in the transplanted kidney but can be damaged by antibodies attacking the normal collagen present in the glomeruli (9).

Conclusion

The ophthalmologist's role is the early detection of Alport syndrome. Any young patient with chronic renal failure should have a careful ophthalmologic evaluation.

References

1. Heidet L, Knebelmann B, Gubler M. Alport syndrome: overview. In: Turner N, Lameire N, Goldsmith D, Winearls C, Himmelfarb J, Remuzzi G editors. *Oxford Textbook of Clinical Nephrology*. Oxford: Oxford University Press (2015). p. 2695.
2. Feingold J, Bois E, Chompert A, Broyer M, Gubler M, Grünfeld J. Genetic heterogeneity of Alport syndrome. *Kidney Int.* (1985) 27:672–7.
3. Savige J, Gregory M, Gross O, Kashtan C, Ding J, Flinter F. Expert guidelines for the management of Alport syndrome and thin basement membrane nephropathy. *J Am Soc Nephrol.* (2013) 24:364–75. doi: 10.1681/ASN.2012020148
4. Arnott E, Crawfurd M, Toghill P. Anterior lenticonus and Alport's syndrome. *Br J Ophthalmol.* (1966) 50:390.
5. Ghosh S, Singh M, Sahoo R, Rao S. Alport syndrome: a rare cause of uraemia. *BMJ Case Rep.* (2014). doi: 10.1136/bcr-2013-201731
6. Jayaprasad B, Sathish K, Chandrasekhar N, Upadhyaya N, Mehta S. Alport's syndrome: a case report. *Indian J Ophthalmol.* (1994) 42:211–2.
7. Savige J, Sheth S, Leys A, Nicholson A, Mack H, Colville D. Ocular features in Alport syndrome: pathogenesis and clinical significance. *Clin J Am Soc Nephrol.* (2015) 10:703–9. doi: 10.2215/CJN.10581014
8. Zhang Y, Ding J. Renal, auricular, and ocular outcomes of Alport syndrome and their current management. *Pediatr Nephrol.* (2018) 33:1309–16. doi: 10.1007/s00467-017-3784-3
9. Browne G, Brown P, Tomson C, Fleming S, Allen A, Herriot R, et al. Retransplantation in Alport post-transplant anti-GBM disease. *Kidney Int.* (2004) 65:675–81. doi: 10.1111/j.1523-1755.2004.00428.x