A Case of Alport Syndrome Presented with Bilateral Anterior Lenticonus

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Abstract.
Purpose: To report a rare case of Alport syndrome presented with bilateral anterior lenticonus in a 16 years old boy.
Case report: A 16 years old boy presented with decrease vision, hearing defect, anaemia 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 nephrologist.
Conclusion: It is easy to diagnose Alport syndrome clinically and close communication between ophthalmologists, otorhinolaryngologists and nephrologists is crucial for effective management of this syndrome.
Keywords: Alport syndrome, anterior lenticonus, collagen type-IV, X-linked inheritance.

INTRODUCTION

A uncommon genetic condition of the basement membrane is known as Alport syndrome. Clinically this syndrome characterized by visual disturbance, hemorrhagic nephritis and sensory neural deafness. In 1927 A. Cecil Alport reported that defective formation of collagen type-IV is the main cause of this syndrome and this type of collagen is present in kidneys, inner ears and eyes. Male are usually more affected with 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 leg, ankle, abdomen and around the eyes; hearing loss; skin problem; nerve problems such as polyneuropathy; ocular lesions and low blood platelet counts that compromise blood clotting etc. Bilateral anterior lenticonus is a hallmark of ocular manifestations. Other ocular changes include cataract, 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 years old boy with Alport syndrome.

CASE REPORT

A 16 years old boy presented with decreased vision, hearing defect, anemia, hematuria and proteinuria (Figure 1). His greatest corrected visual acuity in both of his eyes was 6/18. In the right eye, cycloplegic refraction revealed +2.5DS, and in the left eye, +1.25 DS/+1.00 Cyl 1300.
Alport Syndrome: With Bilateral Anterior Lenticonus

Figure 1. Patient with Alport syndrome.

Figure 2. Right anterior lenticonus.

Figure 3. Left anterior lenticonus.

Bruckner’s test showed oil droplet sign in both eyes. Slit lamp biomicroscopy showed anterior lenticonus in both eyes (Figures 2 and 3). Fundoscopy revealed no abnormalities. Routine blood examination showed Hb 9.5 gm/dl and ESR 27 mm in first hour. Serum urea was 58 mg/dl and creatinine was 2.3 mg/dl. Routine urine examination showed 200 erythrocytes per high power field. Twenty four hours urine volume was 2300 ml and total urine protein was 1568 mg. Blood pressure was 100/70 mm of Hg. Ultrasonogram of kidney, ureter and bladder (KUB) region was normal. Audiogram showed moderate mixed hearing loss in both ears. He was given optical correction for vision improvement and referred to nephrologist for further management.

DISCUSSION

A uncommon genetic condition of the basement membrane is known as Alport syndrome. More than 80% Alport syndrome are X-linked recessive where male are mostly affected and remaining 10%–15% are of autosomal inheritance where both male and female 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 eye, inner ear and glomerular basement membrane of kidney [3]. The COL4A3 and COL4A4 gene mutations that cause autosomal Alport’s syndrome (both recessive and dominant) are located on chromosomal 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 anterior lens capsule and Bruch’s membrane causes anterior lenticonus and retinal flecks which are characteristics feature of Alport syndrome. Hematuria, proteinuria, hearing defects, anterior lenticonus and chronic kidney disease (CKD) were noted in this case. Though retinal flecks in the retina or macula are commonest finding of Alport syndrome, fundoscopy revealed normal findings here. Aldosterone inhibitors, angiotensin converting enzyme inhibitors, and angiotensin receptor blockers can all lower proteinuria [8]. To enhance vision, anterior lenticonus coupled with cataracts requires cataract extraction with intraocular lens [3]. Hearing aides 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 antibody attacking the normal collagen present in the glomeruli [9].

**CONCLUSION**

The ophthalmologists’ role is the early detection of this syndrome. Any young patient with a chronic renal failure should have a careful ophthalmologic evaluation.

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**REFERENCES**


