International Journal of Contemporary Pediatrics
Gande D et al. Int J Contemp Pediatr. 2026 Oct;13(10):2147-2150
https://www.ijpediatrics.com
pISSN 2349-3283 | eISSN 2349-3291

DOI: https://dx.doi.org/10.18203/2349-3291.ijcp20263325

Case Report

The eye, ear and the kidney: a diagnostic triad in a child with progressive sensory deficits

Deepak Gande*, Jahnavi Konkana, Sravanthi Kasireddy, Venkataramana Reddy Koluguri, Lakshmi Aparna Devi Velicheti

Department of Pediatrics, Mamata Academy of Medical Sciences, Hyderabad, Telangana, India

Received: 23 July 2026
Accepted: 03 September 2026

***Correspondence:**
Dr. Deepak Gande,
E-mail: gandedeepak20031999@gmail.com

ABSTRACT

Progressive hearing loss and visual impairment in childhood are often evaluated as isolated sensory disorders; however, their coexistence may indicate an underlying multisystem disease. We report a 13-year-old boy who presented with a three- to four-year history of gradually progressive bilateral hearing impairment and diminution of vision. Audiological assessment demonstrated bilateral moderate sensorineural hearing loss, while ophthalmological examination revealed bilateral anterior lenticonus with associated refractive abnormalities. Further evaluation uncovered persistent microscopic hematuria and proteinuria of glomerular origin. The combination of renal, auditory, and ocular abnormalities pointed to a hereditary basement membrane disorder affecting multiple organ systems. Based on the characteristic clinical triad, a diagnosis of Alport syndrome was established. Molecular confirmation was advised but could not be performed because of financial constraints. The patient was initiated on enalapril for renoprotection, fitted with bilateral hearing aids, and referred for ophthalmological surgical evaluation. This case highlights the importance of considering an underlying systemic disorder in children presenting with concurrent sensory deficits. It also emphasizes the value of simple urinalysis as an inexpensive screening tool capable of revealing occult renal involvement and facilitating early diagnosis before the onset of advanced kidney disease.

Keywords: Sensorineural hearing loss, Anterior lenticonus, Microscopic hematuria, Hereditary nephropathy, Type IV collagen disorder, Enalapril

INTRODUCTION

Inherited disorders affecting the glomerular basement membrane can present with a characteristic combination of renal, auditory, and ocular abnormalities. These conditions often manifest in childhood with persistent microscopic hematuria, progressive sensorineural hearing loss, and distinctive ocular findings, including anterior lenticonus and retinal abnormalities. Recognition of this constellation of features is important because renal involvement may initially be asymptomatic despite ongoing disease progression. Alport syndrome, also known as hereditary nephritis, is an inherited disorder of type IV collagen that primarily affects the glomerular basement membrane, inner ear, and ocular structures. It is characterized by progressive kidney disease, often beginning with persistent hematuria, frequently associated with sensorineural hearing loss and distinctive ocular abnormalities such as anterior lenticonus and retinal flecks.[1] The condition results from pathogenic variants in genes encoding the alpha chains of type IV collagen, most commonly COL4A5, which accounts for approximately 80-85% of cases and causes the X-linked form of the disease.[2] Less commonly, mutations in COL4A3 or COL4A4 lead to autosomal recessive or autosomal dominant forms.[3] Since its first description by Cecil Alport in 1927 in a family presenting with hematuria and deafness, the disease has been recognized

as a critical cause of progressive renal impairment, with affected males often exhibiting a severe clinical course.[4] We describe a patient in whom a parallel pattern of progressive auditory and visual impairment led to the clinical identification of Alport syndrome through targeted urinalysis, audiological testing, and ophthalmological evaluation.

CASE REPORT

A 13-year-old boy, born to non-consanguineous parents, presented with a four-year history of progressive bilateral hearing impairment and diminution of vision. He had been entirely asymptomatic until 9 years of age, when his parents first noticed inappropriate responses to verbal commands and a frequent need for repetition during conversation. The hearing loss was insidious in onset and gradually progressive, eventually interfering with classroom participation and daily communication. Around the same age, the patient developed difficulty reading and identifying distant objects, consistent with progressive visual impairment. Both auditory and visual symptoms worsened gradually over time, resulting in a noticeable decline in scholastic performance. His mother also reported intermittently foul-smelling urine from approximately 9 years of age, though there was no history of gross hematuria, dysuria, fever, flank pain, oliguria, facial puffiness, or pedal edema.

The patient was born at term via normal vaginal delivery following an uncomplicated antenatal, natal, and postnatal course. All developmental milestones were attained appropriately for age. There was no history of recurrent ear infections, ear discharge, ototoxic drug exposure, excessive noise exposure, head trauma, ocular trauma, headache, vomiting, vertigo, gait disturbance, ataxia, seizures, developmental regression, or focal neurological deficits. Family history was non-contributory; both parents were asymptomatic, with no personal or family history of hearing impairment, visual abnormalities, hematuria, proteinuria, chronic kidney disease, dialysis, or renal transplantation.

On physical examination, the patient was normotensive and systemic examination was entirely unremarkable, with no dysmorphic features, external ear anomalies, branchial cleft abnormalities, or peripheral edema. Diagnostic workup was initiated to explore the concurrent sensory deficits. Audiological evaluation with pure-tone audiometry confirmed bilateral, symmetrical sensorineural hearing loss, with pure-tone averages recorded at 58 dB HL in the right ear and 48 dB HL in the left ear, indicating moderate impairment (Figure 1). Detailed ophthalmological evaluation revealed a reduced visual acuity of 6/12 in both eyes, accompanied by high myopic astigmatism. Slit-lamp examination demonstrated bilateral anterior lenticonus with mild cortical lens changes, and corneal topography via Pentacam evaluation confirmed these intrinsic lenticular abnormalities (Figure 2). Targeted renal evaluation via urinalysis demonstrated persistent microscopic hematuria with 2+ proteinuria. Urine microscopy revealed 20-25 red blood cells per high-power field with a high percentage of dysmorphic red cells, indicating a true glomerular source of bleeding.

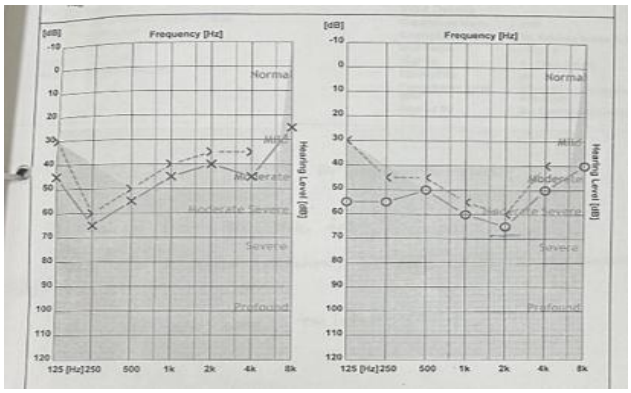

Figure 1: Pure tone audiometry showing bilateral moderate hearing deficit.

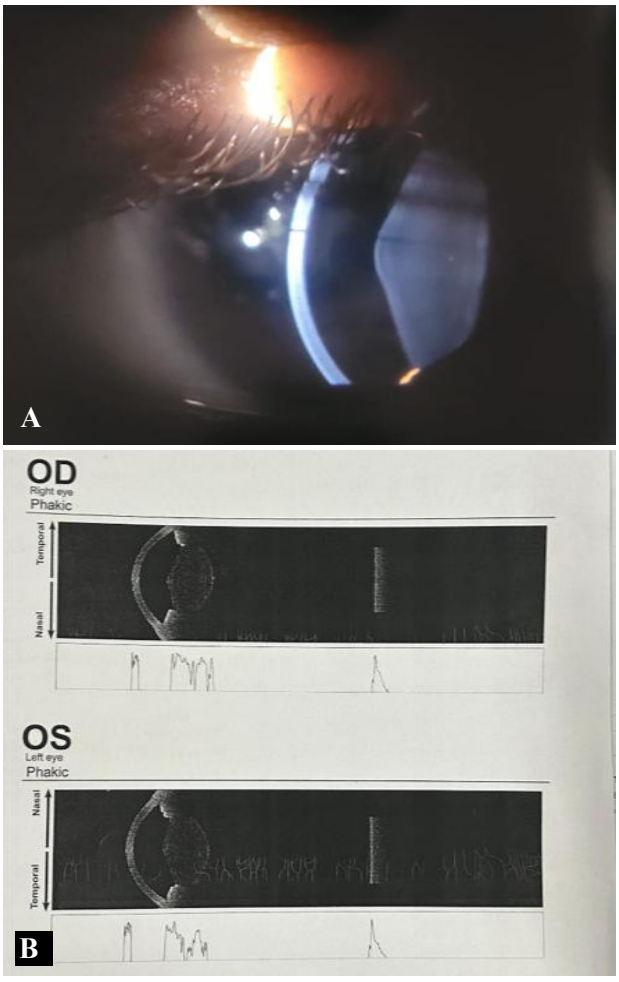

Figure 2: (A) Slit lamp image showing conical projection on the anterior surface of the lens suggestive of lenticonus and (B) anterior segment optical coherence tomography (AS-OCT) of both eyes showing anterior lenticonus.

The coexistence of progressive auditory and visual impairment in a young male raised suspicion for an inherited multisystem disorder. The primary differential diagnosis considered was a hereditary nephropathy, specifically Alport syndrome, given the classic combination of ocular, auditory, and renal manifestations. Other syndromic conditions were also evaluated as secondary possibilities, including branchio-oto-renal spectrum disorders, mitochondrial cytopathies, Fabry disease, and MYH9-related disorders. However, the absence of branchial cleft anomalies, external ear malformations, neuropathic pain, hypohidrosis, characteristic skin lesions, myopathy, seizures, abnormal platelet morphology, or bleeding manifestations effectively ruled out these alternative syndromes. The clinical triad of persistent glomerular hematuria with proteinuria, bilateral sensorineural hearing loss, and characteristic bilateral anterior lenticonus was strongly diagnostic of Alport syndrome. Genetic testing for molecular confirmation and precise characterization of the underlying type IV collagen defect was recommended but could not be performed due to family financial constraints.

Management was initiated using a coordinated, multidisciplinary approach. For renoprotection, the patient was started on an oral angiotensin-converting enzyme inhibitor, enalapril, to lower intraglomerular pressure, reduce proteinuria, and delay the progression of renal disease. Aural rehabilitation was provided by fitting the patient with bilateral digital hearing aids to support academic and social communication. The patient was also referred to a tertiary ophthalmology service for surgical consultation regarding the progressive anterior lenticonus and associated visual deficit. A structured follow-up schedule was established to monitor blood pressure, serum creatinine, and quantification of hematuria and proteinuria every three to six months. The family received extensive counseling regarding the progressive nature of the condition and the importance of regular long-term surveillance.

DISCUSSION

This case highlights that Alport syndrome should be considered in any child or adolescent presenting with progressive sensorineural hearing loss and visual impairment, particularly when urinalysis reveals persistent microscopic hematuria or proteinuria. Renal symptoms are frequently absent in the early stages of the disease, making simple urinalysis a valuable diagnostic clue even before overt renal disease becomes apparent.

Alport syndrome is a clinically important inherited disorder because it may present initially with subtle renal findings, while progressive hearing and visual impairment bring the child to medical attention. It results from pathogenic mutations affecting the alpha-3, alpha-4, or alpha-5 chains of type IV collagen, leading to defective formation of the collagen network required for the glomerular basement membrane, cochlea, lens capsule, and retina.[5] This explains the classic triad of renal involvement, sensorineural hearing loss, and ocular abnormalities. The X-linked form is the most common, accounting for 80-85% of cases, while autosomal recessive inheritance accounts for about 15% of cases. Renal disease usually begins with persistent microscopic hematuria, followed by proteinuria as the disease progresses. Hearing loss is typically sensorineural, and nearly 50% of males with X-linked disease develop hearing impairment by around the age of 15 years. Ocular findings provide important diagnostic clues; anterior lenticonus, seen in about 15% of affected males, is highly characteristic of Alport syndrome, while maculopathy with yellowish or whitish retinal flecks may occur in approximately 50-60% of males with X-linked disease.[3,6] In the present cases, the combination of persistent glomerular hematuria, trace proteinuria, bilateral sensorineural hearing loss, and bilateral anterior lenticonus strongly supported the diagnosis. The occurrence of similar manifestations in two male siblings with asymptomatic parents and negative maternal history of hearing or visual impairment suggests an inherited disorder with possible X-linked or autosomal recessive transmission. Although X-linked Alport syndrome is the most common form, the absence of hearing or visual symptoms in the mother does not exclude carrier status, as females may be asymptomatic or mildly affected.[7] Important differentials include branchio-oto-renal syndrome, mitochondrial cytopathies, Fabry disease, MYH9-related disorders, and other syndromic causes of hearing and visual impairment; however, the absence of branchial anomalies, external ear malformations, neuropathic pain, skin lesions, bleeding manifestations, myopathy, seizures, or developmental regression made these less likely.[8-11] Management is mainly supportive and renoprotective, with ACE inhibitors such as enalapril used to reduce proteinuria and delay progression of renal disease. Hearing aids, ophthalmological management, and regular monitoring of blood pressure, renal function, hematuria, and proteinuria are essential. Prognosis depends on genotype, sex, degree of proteinuria, and timing of treatment, with males affected by X-linked disease generally having a more progressive course. Early recognition is therefore crucial, as timely treatment, family screening, and genetic counselling can help reduce complications and identify at-risk relatives.

CONCLUSION

Alport syndrome should be suspected in pediatric patients presenting with progressive hearing loss and visual disturbances, even when overt signs of renal disease are absent. Finding persistent microscopic hematuria, proteinuria, and anterior lenticonus provides a secure clinical diagnosis. While molecular confirmation remains ideal, a clinical diagnosis is entirely sufficient to immediately launch renoprotective therapies and provide essential sensory rehabilitation, significantly optimising the patient's long-term quality of life.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

REFERENCES

1. Kaluvai SI, Rajalakshmi KV, Dayanandan Y. A case of Alport syndrome. Int J Adv Med. 2023;10(3):242-4.
2. Hartopo AB, Kuswadi I, Hartono. Two male siblings with Alport syndrome. World J Nephrol Urol. 2012;1(4-5):127-9.
3. Okikiade A, Abdulrahman N, Tikanide M, Ndu J, Akoth A, Akinyode E, et al. Alport syndrome: an overview of the pathology. Asian J Res Nephrol. 2022;5(1):121-30.
4. Gregorio V, Caparali EB, Shojaei A, Ricardo S, Barua M. Alport syndrome: clinical spectrum and therapeutic advances. Kidney Med. 2023;5(5):100631.
5. Pan S, Yu R, Liang S. Case report: a case report of Alport syndrome caused by a novel mutation of COL4A5. Front Genet. 2023;14:1216809.
6. Kang E, Park BH, Lee H, Kang HG, Kim JH, Kim YN, et al. A comprehensive review of Alport syndrome: definition, pathophysiology, clinical manifestations, and diagnostic considerations. Kidney Res Clin Pract. 2025;44(4):566-75.
7. Haldar I, Jeloka T. Alport's syndrome: a rare clinical presentation with crescents. Indian J Nephrol. 2020;30(2):129-31.
8. Kumarasamy G, Park SJ, Park PG, Moon IS, Shin JI. Branchio-oto-renal syndrome: a narrative review. Child Kidney Dis. 2026;30(2):86-96.
9. Ireland J, Rossetti S, Haugen E, Ireland J, Michels V, Harris P. Mitochondrial causes of renal insufficiency and hearing loss. Kidney Int. 2004;65(6):2444-5.
10. Köping M, Shehata-Dieler W, Cebulla M, Rak K, Oder D, Müntze J, et al. Cardiac and renal dysfunction is associated with progressive hearing loss in patients with Fabry disease. PLoS One. 2017;12(11):e0188103.
11. Ryżewska W, Chojnowski K, Treliński J. Clinical presentation and management of MYH9-related disease - a single-center experience short communication. Acta Haematol Pol. 2025;56(5):366-70.

Cite this article as: Gande D, Konkana J, Kasireddy S, Koluguri VR, Velicheti LAD. The eye, ear and the kidney: a diagnostic triad in a child with progressive sensory deficits. Int J Contemp Pediatr 2026;13:2147-50.