Diagnosis of Alport Syndrome by Kidney Biopsy and Electron Microscopy in a Resource-Limited Setting: A Case Report.

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Case Report

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Background: Alport syndrome is an inherited disorder caused by pathogenic variants in the COL4A3, COL4A4, and COL4A5 genes, resulting in structural abnormalities of the glomerular basement membrane (GBM) and progressive kidney disease. Although molecular genetic testing is considered the diagnostic reference standard, kidney biopsy with electron microscopy remains an essential diagnostic tool when genetic analysis is unavailable or inconclusive.

We report the case of a 17-year-old female who presented with systemic arterial hypertension, persistent microscopic hematuria, and proteinuria during the evaluation of secondary hypertension. Kidney biopsy complemented by electron microscopy demonstrated marked variability in GBM thickness (135–551 nm; mean 389.56 nm), with laminated, thickened, woven, bead-like, and electron-lucent resorptive segments, in addition to diffuse podocyte foot process effacement affecting approximately 40–50% of the capillary surface, without evidence of immune-complex deposition. Owing to financial limitations, molecular genetic testing could not be performed; therefore, the diagnosis was established through the integration of clinical, laboratory, histopathological, and ultrastructural findings.

Nephroprotective treatment with telmisartan was initiated, and the patient was referred for nephrology follow-up. This case highlights the continued relevance of kidney biopsy with electron microscopy within contemporary diagnostic algorithms and emphasizes its indispensable role in establishing an accurate diagnosis and facilitating early nephroprotective management when molecular testing is inaccessible.

Keywords: Alport Syndrome, Hereditary Nephropathy.

Alport syndrome is an inherited nephropathy caused by pathogenic variants in the COL4A3, COL4A4, and COL4A5 genes, which encode the α3, α4, and α5 chains of type IV collagen, an essential component of the glomerular basement membrane. Alterations in the synthesis of this collagen network result in progressive structural and functional changes of the basement membrane, clinically manifested by persistent hematuria, proteinuria, hypertension, and progressive deterioration of renal function, eventually leading to advanced chronic kidney disease or end-stage kidney disease. Currently, Alport syndrome represents one of the main hereditary causes of kidney failure in pediatric populations and young adults.[1,2,4,5,6,10]

The disease exhibits heterogeneous clinical expression depending on the inheritance pattern, the affected gene, and the underlying genetic variant. The X-linked form accounts for approximately 80–85% of cases, whereas autosomal recessive and autosomal dominant forms are less common. In addition to renal involvement, extrarenal manifestations such as sensorineural hearing loss and ocular abnormalities may develop, resulting from the distribution of type IV collagen in the cochlear and ocular basement membranes. Phenotypic variability, even among members of the same family, may complicate early recognition of the disease and delay the initiation of therapeutic measures aimed at preserving renal function.[2,4,5,6,10]

Currently, molecular analysis is considered the reference standard for confirming the diagnosis of Alport syndrome. However, contemporary diagnostic algorithms continue to recognize an important role for kidney biopsy, particularly when genetic testing is unavailable, inaccessible, or yields inconclusive results. In this context, electron microscopy allows the identification of characteristic ultrastructural abnormalities of the glomerular basement membrane, including irregular thinning and thickening, lamellation, disorganization of the lamina densa, and the classic “basket-weave” pattern. These findings constitute highly suggestive features of Alport syndrome and, in the appropriate clinical context, allow diagnosis to be established when molecular analysis is not available.[3,4,5,9,10]

Despite advances in molecular diagnosis, diagnostic delay remains a significant challenge, particularly in settings where access to genetic testing is limited due to availability or cost. Under these circumstances, the integration of clinical, laboratory, histopathological, and ultrastructural findings remains essential to achieve timely diagnosis and initiate early nephroprotective strategies aimed at delaying renal disease progression and improving patient outcomes.[4,7,8,9,10]

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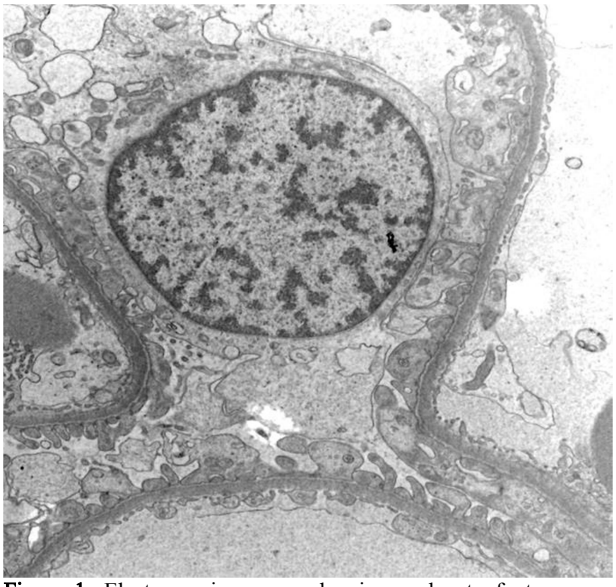

Figure 1. Electron microscopy showing podocyte foot process edema and focal effacement of the foot processes.

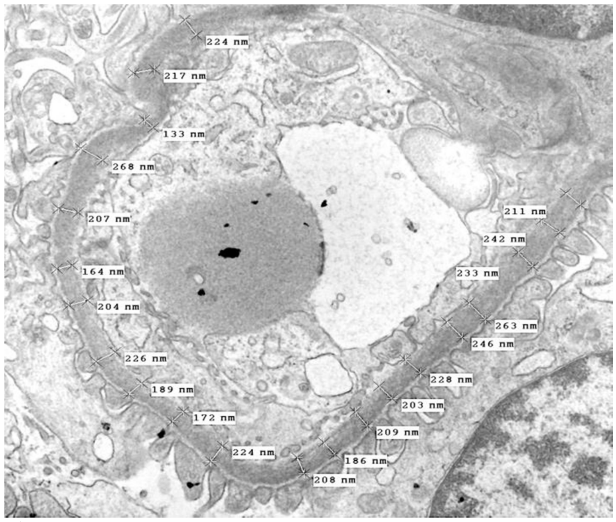

Figure 2. Ultrastructural measurement of the glomerular basement membrane demonstrating marked variability in thickness (135–551 nm), with alternating areas of thinning and irregular thickening.

We present the case of an adolescent female patient with Alport syndrome whose diagnosis was established through kidney biopsy and electron microscopy, given the inability to perform molecular confirmation due to financial limitations. This case highlights the continued relevance of kidney biopsy complemented by electron microscopy as a diagnostic tool in patients with suspected Alport syndrome when genetic analysis cannot be performed, emphasizing the importance of clinicopathological correlation in the evaluation of this condition.[3,4,5,9,10]

Case report

A 17-year-old female with a family history of systemic arterial hypertension and type 2 diabetes mellitus in both parents presented for evaluation. Her past medical history was significant for atopic dermatitis, asthma, and allergic rhinitis since childhood, for which she was receiving levocetirizine 5 mg as needed and inhaled salbutamol/ipratropium for respiratory symptoms. She denied smoking, alcohol consumption, or illicit drug use.

One year before presentation, she experienced an episode of anaphylaxis secondary to ketorolac ingestion. Subsequently, she developed persistent systemic arterial hypertension and was referred for evaluation of secondary hypertension. On admission, her blood pressure was 150/100 mmHg.

As part of the diagnostic workup, plasma renin concentration was elevated at 213 mIU/L, whereas plasma metanephrine levels were within normal limits. Immunological testing revealed positive antinuclear antibodies with a coarse speckled pattern (1:160), normal serum C3 and C4 complement levels, and an erythrocyte sedimentation rate of 34 mm/h. Urinalysis demonstrated microscopic hematuria and 2+ proteinuria. As part of the evaluation for extrarenal manifestations, pure-tone audiometry was performed and showed no evidence of hearing impairment.

Given the suspicion of glomerular disease, a percutaneous kidney biopsy was performed. Histopathological examination, complemented by electron microscopy, demonstrated structural abnormalities of the glomerular basement membrane. A total of 92 measurements were obtained from 14 glomerular capillary loops, revealing a mean glomerular basement membrane thickness of 389.56 nm (expected range for age and sex: 310–330 nm), with marked variability ranging from 135 to 551 nm. In addition, laminated, thickened, woven, bead-like, and electron-lucent resorptive segments of the glomerular basement membrane were identified. No electron-dense deposits suggestive of immune complexes were observed within the mesangium, subendothelial space, or glomerular basement membrane. Furthermore, diffuse edema and effacement involving approximately 40–50% of podocyte foot processes were documented. Collectively, these findings were consistent with structural abnormalities of the glomerular basement membrane suggestive of Alport syndrome (Figure 1) (Figure 2) (Figure 3). Owing to financial constraints that precluded molecular genetic testing, the diagnosis was established based on the clinical, histopathological, and ultrastructural findings obtained by electron microscopy.

Following the diagnosis, telmisartan 80 mg orally once daily was initiated as nephroprotective therapy, and the patient was referred for continued follow-up by the Nephrology Department.

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Discussion

Alport syndrome is a hereditary disorder of the glomerular basement membrane (GBM) caused by pathogenic variants in the COL4A3, COL4A4, and COL4A5 genes, which encode the α3, α4, and α5 chains of type IV collagen. These collagen chains constitute the structural framework of the mature GBM and are also present in the basement membranes of the cochlea, lens capsule, cornea, retina, and other specialized tissues. Defects in the assembly of the collagen IV α3α4α5 network result in progressive instability of the GBM, leading to alterations in glomerular filtration barrier integrity, progressive glomerulosclerosis, tubulointerstitial fibrosis, and ultimately chronic kidney disease. This molecular mechanism also explains the development of extra-renal manifestations, particularly sensorineural hearing loss and characteristic ocular abnormalities, which reflect the systemic distribution of type IV collagen.[1,2,4,5,6,10]

Although Alport syndrome has long been considered an uncommon inherited nephropathy, recent evidence suggests that it is substantially underrecognized. Improvements in molecular diagnostics have demonstrated that pathogenic variants in type IV collagen genes are more prevalent than previously estimated and account for a considerable proportion of hereditary kidney disease worldwide. X-linked Alport syndrome, resulting from COL4A5 variants, remains the predominant inheritance pattern, representing approximately 80–85% of affected individuals, whereas autosomal recessive disease caused by biallelic COL4A3 or COL4A4 variants and autosomal dominant forms associated with heterozygous variants in these genes occur less frequently. Nevertheless, considerable phenotypic heterogeneity exists both among and within families, reflecting the influence of the underlying genotype, inheritance pattern, and additional modifying factors that contribute to disease severity and progression.[2,4,5,6,10]

The clinical presentation of Alport syndrome is equally heterogeneous, often posing significant diagnostic challenges during the early stages of disease. Persistent microscopic hematuria is generally the earliest and most consistent manifestation, frequently preceding proteinuria, hypertension, and decline in renal function by several years. As structural damage to the GBM progresses, increasing proteinuria, progressive loss of glomerular filtration rate, and eventual kidney failure develop, although the age at onset and rate of progression vary considerably according to the causative mutation and mode of inheritance. Extra-renal manifestations, particularly bilateral high-frequency sensorineural hearing loss and characteristic ocular abnormalities such as anterior lenticonus and retinal changes, provide important diagnostic clues but are not universally present, especially during childhood or adolescence. Consequently, the absence of these manifestations should not exclude the diagnosis when compatible renal findings and family history are present.[1,2,4,5,6,9,10]

The present case illustrates several of these diagnostic challenges. Although the patient presented with persistent hypertension, microscopic hematuria, and proteinuria—findings highly suggestive of glomerular disease—she did not exhibit hearing impairment on audiometric evaluation, nor were ocular abnormalities documented at the time of diagnosis. This clinical presentation is consistent with the well-recognized age-dependent penetrance and phenotypic variability described in Alport syndrome, in which extra-renal manifestations may develop later during the disease course or remain absent depending on the underlying genotype. Therefore, reliance solely on the presence of the classic clinical triad may delay diagnosis, particularly in adolescents with isolated renal manifestations.[2,4,5,9,10]

During the past decade, the diagnostic approach to Alport syndrome has undergone substantial evolution owing to the increasing availability of molecular genetic testing. Current recommendations recognize molecular analysis as the reference standard for confirming the diagnosis because it permits identification of the causative pathogenic variant, establishes the mode of inheritance, facilitates accurate genetic counseling, enables family screening, and provides valuable prognostic information. Moreover, the incorporation of next-generation sequencing into routine nephrological practice has significantly improved diagnostic yield, particularly among patients with persistent hematuria or familial kidney disease of previously unknown etiology. Consequently, several contemporary diagnostic algorithms advocate molecular testing as the preferred initial confirmatory investigation whenever clinical suspicion is sufficiently high and genetic testing is readily accessible.[3,4,5,9,10]

However, despite these advances, current diagnostic algorithms do not consider kidney biopsy obsolete. Instead, recent reviews and consensus recommendations emphasize that histopathological evaluation continues to occupy an important role in carefully selected clinical scenarios. Kidney biopsy remains particularly valuable in patients without access to genetic testing, when molecular studies are unavailable because of financial or logistical limitations, when genetic results are inconclusive or reveal variants of uncertain significance, or when alternative glomerular diseases must be excluded. Under these circumstances, integration of clinical findings with light microscopy, immunopathology

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when indicated, and particularly electron microscopy remains essential for establishing a reliable diagnosis and guiding subsequent management.[3,4,5,9,10]

In the present patient, financial limitations precluded molecular confirmation, making kidney biopsy the cornerstone of the diagnostic evaluation. Rather than representing an alternative of lesser value, this approach is fully supported by contemporary diagnostic recommendations, which acknowledge that clinicopathological correlation continues to provide robust diagnostic evidence when molecular studies cannot be obtained. Accordingly, the diagnosis in this case was established through the integration of compatible clinical manifestations with characteristic ultrastructural abnormalities identified on electron microscopy, highlighting the continued relevance of renal pathology in modern nephrology despite the increasing availability of genetic technologies.[3,4,5,9,10]

Among the histopathological techniques currently available, electron microscopy (EM) remains the most valuable morphologic tool for the diagnosis of Alport syndrome when molecular confirmation cannot be obtained. Although light microscopy may demonstrate nonspecific findings, particularly during the early stages of disease, ultrastructural examination enables direct visualization of the characteristic alterations of the glomerular basement membrane (GBM) that reflect the underlying collagen IV defect. Contemporary reviews emphasize that the sensitivity of electron microscopy is greatest when interpreted within the appropriate clinical context and in conjunction with family history and laboratory findings, underscoring the importance of an integrated clinicopathological approach rather than reliance on a single diagnostic modality.[1,3,4,5,9,10]

The ultrastructural abnormalities described in Alport syndrome evolve progressively throughout the natural history of the disease. Early lesions may consist predominantly of diffuse thinning of the GBM, whereas more advanced stages demonstrate alternating areas of thinning and thickening, irregular variation in membrane thickness, lamellation and splitting of the lamina densa, multilayering, and the characteristic "basket-weave" appearance that has long been considered a hallmark of the disease. Additional findings may include irregular electron-lucent areas, disruption of the normal trilaminar architecture, and progressive remodeling of the basement membrane secondary to abnormal collagen IV assembly. Importantly, these structural abnormalities may coexist within the same glomerulus, reflecting the dynamic nature of disease progression and explaining the marked variability observed in morphometric measurements.[3,4,5,6,9,10]

The findings observed in our patient closely parallel these characteristic ultrastructural features. Electron microscopy demonstrated marked variability in GBM thickness, with measurements ranging from 135 to 551 nm and a mean thickness exceeding the expected values for age and sex. Furthermore, laminated, thickened, woven, bead-like, and electron-lucent resorptive segments were identified, together with diffuse podocyte foot process effacement involving approximately 40–50% of the capillary surface. Equally relevant was the absence of electron-dense immune deposits within the mesangial, subendothelial, or glomerular basement membrane compartments, a finding that supports a structural hereditary nephropathy rather than an immune complex-mediated glomerulopathy. Taken together, these ultrastructural abnormalities are highly consistent with the morphologic spectrum of Alport syndrome described in contemporary pathological series and recent diagnostic reviews.[3,4,5,9,10]

Recent diagnostic algorithms have progressively shifted toward a genetics-first approach, recommending molecular testing whenever clinical suspicion is supported by persistent glomerular hematuria, familial kidney disease, unexplained chronic kidney disease, or compatible extra-renal manifestations. Nevertheless, these same algorithms continue to recognize kidney biopsy as an essential complementary investigation in selected circumstances. Specifically, renal biopsy with electron microscopy remains recommended when molecular testing is unavailable, financially inaccessible, technically limited, delayed, or fails to identify a pathogenic variant despite persistent clinical suspicion. Likewise, histopathological evaluation is valuable when differential diagnoses remain under consideration or when additional prognostic information regarding renal injury is required. Consequently, current recommendations do not establish a competition between molecular diagnosis and renal biopsy but rather propose both strategies as complementary components of an individualized diagnostic pathway.[1,2,3,4,5,9,10]

This concept is particularly relevant in many healthcare systems where comprehensive genetic testing remains inaccessible because of economic constraints. Although advances in next-generation sequencing have considerably improved diagnostic accuracy, disparities in availability continue to limit its routine implementation in numerous regions. Under these circumstances, delaying diagnosis until molecular confirmation becomes available may postpone the initiation of nephroprotective interventions during a critical period of disease progression. Our patient exemplifies this scenario, as financial limitations precluded genetic analysis despite a strong clinical suspicion of hereditary nephropathy. The diagnosis therefore relied on the integration of clinical findings with characteristic ultrastructural abnormalities identified by electron microscopy,

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illustrating the continued practical importance of renal pathology in resource-limited settings.[3,4,5,9,10]

Another important aspect highlighted by this case is the value of clinicopathological correlation. Neither persistent microscopic hematuria nor proteinuria is specific for Alport syndrome, and hypertension in adolescents encompasses a broad differential diagnosis. Similarly, the absence of sensorineural hearing loss, which has traditionally been regarded as a classical feature of the disease, could potentially reduce clinical suspicion if interpreted in isolation. However, when these findings were evaluated together with the characteristic ultrastructural alterations of the GBM, the diagnostic probability increased substantially. Contemporary literature consistently emphasizes that Alport syndrome should be suspected in patients with persistent glomerular hematuria even in the absence of extra-renal manifestations, particularly because hearing and ocular abnormalities may appear later in the disease course or may vary according to the underlying genotype and inheritance pattern.[2,4,5,9,10]

Beyond establishing the diagnosis, early recognition has important therapeutic implications. Progressive renal fibrosis begins long before the development of advanced chronic kidney disease, making timely diagnosis essential to preserve renal function. Current evidence supports early initiation of renin–angiotensin–aldosterone system (RAAS) blockade, even before substantial deterioration in kidney function occurs, as this strategy has consistently been associated with delayed progression of proteinuria, slower decline in glomerular filtration rate, and postponement of kidney failure. Recent systematic reviews and contemporary clinical recommendations therefore advocate RAAS inhibition as the cornerstone of nephroprotective management in patients with Alport syndrome, emphasizing that therapeutic benefit is greatest when treatment is initiated during the earliest clinically detectable stages of disease.[4,7,8,10]

Accordingly, initiation of telmisartan in the present patient was consistent with current evidence-based recommendations. Although RAAS blockade does not correct the underlying genetic defect, it remains the intervention with the strongest clinical evidence for slowing disease progression. In parallel, ongoing research into novel therapeutic strategies—including molecular and gene-targeted approaches—offers promising future perspectives; however, these therapies remain under investigation, reinforcing the importance of establishing an early diagnosis and implementing currently available nephroprotective measures without delay.[7,8,10]

Although molecular diagnosis has transformed the evaluation of hereditary nephropathies, the present case underscores that advances in genetic technologies should not diminish the diagnostic value of renal pathology. Instead, contemporary evidence supports a complementary approach in which molecular testing and kidney biopsy are integrated according to the clinical scenario and resource availability. While identification of pathogenic variants in COL4A3, COL4A4, or COL4A5 establishes the diagnosis with certainty and provides important information regarding inheritance patterns, prognosis, and family counseling, characteristic ultrastructural alterations identified by electron microscopy remain highly reliable diagnostic findings when interpreted alongside compatible clinical manifestations. Consequently, kidney biopsy continues to represent an indispensable diagnostic resource in patients for whom molecular confirmation cannot be achieved.[3,4,5,9,10]

The present patient exemplifies this diagnostic paradigm. Persistent microscopic hematuria, proteinuria, and systemic arterial hypertension strongly suggested an underlying glomerular disorder; however, the absence of hearing impairment and the inability to perform molecular testing could potentially have delayed or obscured the diagnosis. Rather than limiting the diagnostic evaluation, renal biopsy with electron microscopy provided decisive evidence by demonstrating the characteristic spectrum of GBM abnormalities associated with Alport syndrome, including marked variation in membrane thickness, lamellation, woven architecture, bead-like segments, and electron-lucent resorptive areas in the absence of immune-complex deposition. These findings, interpreted together with the clinical presentation, permitted a confident diagnosis despite the lack of genetic confirmation and illustrate the continued importance of clinicopathological correlation in everyday nephrological practice.[3,4,5,9,10]

An additional strength of this case lies in the detailed ultrastructural assessment performed by electron microscopy. Morphometric evaluation of multiple glomerular capillary loops documented considerable variability in GBM thickness rather than uniform thickening or thinning, emphasizing one of the characteristic pathological features of Alport syndrome. Contemporary pathological reviews highlight that careful quantitative and qualitative assessment of the GBM substantially increases diagnostic confidence, particularly when characteristic structural abnormalities coexist within the same biopsy specimen. This reinforces the importance of adequate tissue processing, systematic ultrastructural evaluation, and interpretation by experienced renal pathologists when hereditary nephropathies are suspected.[3,5,9]

From a clinical perspective, establishing the diagnosis at an early stage has implications that extend beyond diagnostic certainty. Recognition of Alport syndrome allows prompt initiation of nephrop otective therapy, appropriate longitudinal surveillance of renal

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function and proteinuria, monitoring for extra-renal manifestations, and evaluation of potentially affected family members. Furthermore, confirmation of the diagnosis avoids unnecessary investigations for alternative causes of persistent hematuria and proteinuria and facilitates individualized counseling regarding disease progression and inheritance. Contemporary recommendations consistently emphasize that delaying diagnosis may postpone interventions capable of slowing renal deterioration, thereby reinforcing the importance of recognizing the disease before irreversible structural damage develops.[2,4,7,8,10]

The present report also highlights an important reality encountered in many healthcare settings, where access to comprehensive genetic testing remains limited by cost or availability. Although international recommendations increasingly favor molecular confirmation, they also acknowledge that diagnostic strategies must be adapted to local resources without compromising diagnostic accuracy. In this context, kidney biopsy complemented by electron microscopy remains a highly valuable and clinically applicable diagnostic approach, particularly in resource-limited environments where reliance on clinical and pathological correlation continues to play a central role.[3,4,5,9,10]

Overall, this case reinforces the concept that the diagnosis of Alport syndrome should be based on the integration of clinical, laboratory, histopathological, and ultrastructural findings rather than on a single diagnostic modality. While molecular analysis currently represents the diagnostic reference standard, kidney biopsy with electron microscopy continues to provide essential diagnostic information when genetic testing is unavailable, inaccessible, or inconclusive. The characteristic GBM alterations identified in our patient allowed timely diagnosis and initiation of nephroprotective therapy, highlighting that renal pathology remains an indispensable component of contemporary diagnostic algorithms. Continued recognition of the complementary roles of molecular genetics and renal biopsy is fundamental to achieving early diagnosis, optimizing patient management, and improving long-term renal outcomes in individuals with suspected Alport syndrome.[3,4,5,7,8,9,10]

Conclusion

Alport syndrome remains a diagnostic challenge because of its marked clinical and genetic heterogeneity. Although molecular genetic testing is currently considered the reference standard for diagnosis, kidney biopsy complemented by electron microscopy continues to play a pivotal role when genetic analysis is unavailable, inaccessible, or inconclusive. The present case demonstrates that integration of clinical findings with characteristic ultrastructural alterations of the glomerular basement membrane can establish a reliable diagnosis, enabling timely initiation of nephroprotective therapy. This report reinforces the continued relevance of clinicopathological correlation in contemporary diagnostic algorithms and highlights the indispensable value of electron microscopy in resource-limited settings, where it remains an essential tool for the diagnosis and management of patients with suspected Alport syndrome.[1,3,4,5,7,8,9,10]

Conflicts of interests

None declared by the authors

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