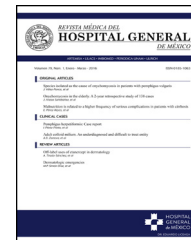




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CLINICAL CASE

3 **Nephropathies with pattern of structural alterations of**  
4 **the glomerular basement membrane: Case study**

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12 **KEYWORDS**

13 Structural alteration  
14 of the glomerular  
15 basement membrane;  
16 Alport syndrome;  
17 Thin basement  
18 membrane  
19 nephropathy;  
20 Collagen type IV  
21 nephropathies

**Abstract**

*Background:* The pattern of structural alterations of the glomerular basement membrane comprises two hereditary glomerular diseases: Alport syndrome (AS) and thin basement membrane nephropathy (TBMN); both are rare entities.

*Material and methods:* A retrospective, descriptive and observational study was carried out. A sample of 90 cases from Hospital General de México "Eduardo Liceaga" and the Instituto Nacional de Cardiología "Ignacio Chávez" were obtained over a period of 5 years (2011–2016). The diagnoses provided in all cases were reviewed and the clinical and histological spectrum was described. Data were analysed using descriptive statistics.

*Results:* Structural alterations of the basement membrane were found to be more frequent in women (60%), and occurred most frequently in the second decade of life. We found 38 cases (42.22%) compatible with TBMN and 52 cases (57.77%) with ultrastructural changes suggestive of Alport syndrome. Of the cases with characteristics compatible with AS, the majority were men (55.76%) with an average age of 14 years (3–39 years), starting with haematuria (42.30%); in the electron microscopy (EM), the glomerular basement membranes (GBM) measured on average 245.27 nm, with very irregular ranges. The majority of patients with TBMN were women (81.57%) with an average age of 29 years (6–66 years) and with persistent microscopic haematuria at the time of diagnosis (47.36%); in the EM, the GBM averaged 170.63 nm in thickness.

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*Conclusions:* We report one of the largest case series in Latin America with respect to entities that share a morphological pattern via the study of optical microscopy, that of structural alterations of the glomerular basement membrane. With the EM study, both entities can be suggested.

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**PALABRAS CLAVE**  
Alteración estructural de la membrana basal glomerular;  
Síndrome de Alport;  
Enfermedad de membranas basales delgadas;  
Nefropatías del colágeno tipo IV

## Nefropatías con patrón de alteraciones estructurales de la membrana basal glomerular: estudio de casos

### Resumen

*Antecedentes:* El patrón de alteraciones estructurales de la membrana basal glomerular comprende a dos enfermedades hereditarias del glomérulo: el síndrome de Alport (SA) y la enfermedad de membranas basales delgadas (EMBD); ambas son entidades poco frecuentes.

*Material y métodos:* Se hizo un estudio retrospectivo, descriptivo y observacional. Se obtuvo una muestra de 90 casos del Hospital General de México "Eduardo Liceaga" y del Instituto Nacional de Cardiología "Ignacio Chávez" de un periodo de 5 años (2011 a 2016). Se revisaron los diagnósticos prestados en todos los casos y se describió el espectro clínico e histológico. Los datos fueron analizados utilizando estadística descriptiva.

*Resultados:* Se encontró que las alteraciones estructurales de la membrana basal eran más frecuentes en las mujeres (60%), y la mayor parte se presentaron en la segunda década de la vida. Se encontraron 38 casos (42.22%) compatibles con enfermedad de membranas basales delgadas (EMBD) y 52 casos (57.77%) con cambios ultraestructurales sugerentes de síndrome de Alport. De los casos con características compatibles con SA, la mayoría eran hombres (55.76%) con una edad promedio de 14 años (3-39 años), debutando con hematuria (42.30%); en el estudio de microscopía electrónica de transmisión (MET) las membranas basales glomerulares (MBG) midieron en promedio 245.27 nm, con rangos muy irregulares. La mayoría de los pacientes con EMBD eran mujeres (81.57%) en una edad promedio de 29 años (6-66 años), con hematuria microscópica persistente al momento de su diagnóstico (47.36%); en el estudio de MET las MBG en promedio midieron 170.63 nm de grosor.

*Conclusiones:* Se informa una de las casuísticas más grandes de Latinoamérica respecto a entidades que comparten un patrón morfológico por el estudio de microscopía óptica, el de alteraciones estructurales de la membrana basal glomerular. Con el estudio de MET se pueden sugerir ambas entidades.

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## Introduction

Alport syndrome (AS) and thin basement membrane disease (TBMD) are genetically heterogeneous diseases characterised by structural abnormalities in the glomerular basement membrane (GBM).<sup>1</sup> They are entities that do not show specific findings in the optical microscopy study for their diagnosis, so it can be difficult to differentiate between them. The current recommendations suggest the genetic study as the gold standard for the diagnosis of AS and to exclude it in a case of TBMD<sup>2</sup>; however, by using extension studies such as direct immunofluorescence and transmission electron microscopy (TEM), it is possible to diagnose, or at least to suggest, these disorders in most cases.<sup>3</sup>

AS is currently considered a spectrum of pathologies secondary to mutations in the  $\alpha 3$ ,  $\alpha 4$  and  $\alpha 5$  chains of type IV collagen, usually with auditory and ocular

abnormalities. Its incidence in the US is 1:5000-1:10,000, according to the database of the different mutations.<sup>4</sup> Approximately 85% of cases are due to several mutations in the COL4A5 gene, located on chromosome Xq26-48, which encodes the  $\alpha 5$  chain of type IV collagen, presenting progressive kidney disease until terminal stages in affected men but only with urinary disorders (haematuria with or without mild proteinuria, which may increase with age) in women.<sup>5</sup> Although most patients with X-linked AS will have a family history of kidney disease, 10%-15% of cases represent *de novo* mutations in the COL4A5 gene. The remaining AS cases result from mutations in the COL4A3/COL4A4 locus on chromosome 2q35-37, which encode the  $\alpha 3$  and  $\alpha 4$  chains of type IV collagen with autosomal recessive inheritance; autosomal dominant inheritance is less common. The phenotypic expression of these patients is quite variable, ranging from benign urinary disorders to progressive

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