KSN SEOUL, KOREA 2023
April 27(Thu) - 30(Sun), 2023
Coex, Seoul, Korea

Abstract Type : Poster
Abstract Submission No. : 1524

Indolent course of Alport syndrome in 74 years old male

Jin Seon Jeong[1], Hyun Soon Lee[2], Hajeong Lee[3], Sang Hee Lee[1], Dong-Young Lee[1], Beom Kim[1], Kyoung Hyoub Moon[1]
[1]Department of Internal Medicine-Nephrology, Seoul Veterans Hospital, Korea, Republic of
[2]Department of Pathology, Hankook Renal Pathology Lab, Korea, Republic of
[3]Department of Internal Medicine-Nephrology, Seoul National University Hospital, Korea, Republic of

Case Study: Alport syndrome is a genetic condition characterized by kidney disease, loss of hearing and eye abnormalities. Kidney manifestations include proteinuria, hypertension and loss of kidney function and ESRD. It is hereditary disorder caused by variants of the type IV collagen gene alfa chain involving the basement membrane of the glomeruli, cochlea and eye. We report a case of a 73-year-old male with slowly increased serum creatinine (SCr 3.19mg/dL) for 7 months. His previous kidney biopsy was performed ten years ago for proteinuria (2,650mg/day) with normal renal function (SCr 1.01mg/dL). Pathologic finding was GBM abnormalities but he had no family history, biopsy was inconclusive in making some decision. He had hypertension and no diabetes. He had a history of cataract, but no hearing problem. His family members had no kidney disease. He took angiotension receptor blocker and proteinuria was improved. Recently, his kidney function has been slowly deteriorating over several months, he had second kidney biopsy. A full serology and urology workup was negative for any abnormality. A kidney biopsy for the patient revealed a suspicion of Alport syndrome presenting as the glomerular basement membrane shows diffuse severe abnormalities with splitting, reticulation and thickening of lamina densa with basket-weave transformation. Patient was refered genetic analysis for correct diagnosis. Gene study revealed variant of uncertain significance COL4A5 and further study and familial counseling are processing. His daughter and grandchild had same variant. As inherited disease, genetic testing is essential for correct diagnosis and familial counseling of Alport syndrome.

Figure 1. Kidney pathology on biopsy (EM)