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Glomerular disease

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Immunohistochemical study can contribute to the diagnosis of Alport syndrome in the early stage

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Background: The most common presenting sign of Alport syndrome (AS) is hematuria with/without proteinuria. Therefore, this disorder should be distinguished from IgA nephropathy (IgAN) and thin glomerular basement membrane nephropathy (TBMN). It may be difficult to detect AS on a purely morphologic basis especially in the early stage of the disease. We report two cases of AS that was misinterpreted other nephropathy on renal biopsy through conventional method.

Methods: Fluorochrome-conjugated monoclonal antibodies for alpha5(IV) chain [FITC-conjugated-anti α5(IV)] and alpha2(IV) chain [Texas Red-anti α2(IV)] were used for staining the renal basement membrane structure. We performed staining in 17 patients diagnosed with TBMN and 1 IgAN patient who was clinically suspected as AS between 2011 and 2015 in a single center.

Results: We discovered two AS patients, one male and one female, who had been misdiagnosed as IgAN and TBMN at the initial pathology review. Male patient initially presented as asymptomatic proteinuria in school urinary screening and showed mild bilateral sensorineural hearing loss without a family history. Female patient initially presented as asymptomatic microscopic hematuria without hearing impairments and ocular defects with a family history of ESRD. In the immunohistochemical stain, no FITC fluorescence was observed, and only Texas Red fluorescence was present in the male patient with AS. Meanwhile, in the female patient, discontinuous FITC fluorescence was observed. Then, X-linked dominant inheritance was confirmed by detection of COL4A5 gene mutation in these patients.

Conclusion: Even though there were no characteristic clinical findings of AS, careful examination of electron microscopy and immunohistochemical staining for type IV collagen alpha chains could be useful modalities to discover AS in the early stage. Also, close cooperation between nephrologists and pathologists is important for the suspected cases.

Keywords: Alport syndrome, Collagen type IV, Hereditary nephritis, Immunohistochemistry