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**Clinical characteristics and long-term prognosis of Alport syndrome: single center study**

**HEAMIN JANG**, HEESUN BAEK, JEONGHYUN SUN, JINWOO KIM, SUNHEE PARK, YONGLIM KIM, CHANDUCK KIM, HEEYEON JUNG, JANGHEE CHO, MINHYUN CHO  
Department of Pediatrics-Nephrology, Kyungpook National University Hospital, Korea, Republic of

**Objectives:** Alport syndrome (AS) is one of most common inherited renal diseases caused by mutations of genes encoding specific proteins of the type IV collagen family and the major features are progressive renal failure, sensorineural deafness and ocular abnormalities. The purpose of this study was to analyze the clinical characteristics and long-term prognosis of AS in single-center Korean pediatric and adult population.

**Methods:** We have retrospectively reviewed medical records of 33 patients who had been diagnosed or treated with AS at the department of Pediatrics and Internal Medicine, Kyungpook National University Hospital from 1985 to 2019.

**Results:** The mean age of 33 patients who had been diagnosed with AS was  $16.2 \pm 13.6$  years old and male to female ratio was 2:1. At the first visit, recurrent gross hematuria was the most common symptom. Of the 25 children, 14 were diagnosed by renal biopsy, and 5 were diagnosed by gene study. 3 patients were diagnosed by renal biopsy and gene study together. 18 patients had a family history of kidney disease. However, only 9 cases were able to identify specific genetic patterns based on the actual family history and genetic test results. 7 of them have X-linked inheritance and 2 have autosomal recessive inheritance. 18 of 33 patients underwent gene study. 14 patients performed conventional sanger sequencing on COL4A5, and the 4 patients performed Next Generation Sequencing. As a result, 7 cases of COL4A5 gene mutation were identified, accounting for 39%. One person with COL4A4 gene mutation was identified through NGS. 11 patients progressed to end stage renal disease. During follow-up period, SNHL was observed in 30.3% and ophthalmic complications were not observed.

**Conclusions:** Genetic analysis may provide information for studying about genetic etiology of Alport syndrome. Therefore, additional prospective cohort study is needed to investigate with Alport syndrome in Korea.