국내 Alport 증후군의 경험

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Alport Syndrome in Korea

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Previously we had reported the mutational analysis of X-linked Alport syndrome (AS) in Korean patients with mutation detection rate of 40% in patients with pathologically proven AS. Ten years later, we report our experience of sixty three patients (M:F=39:24) with AS, diagnosed by pathology (n=29), mutational analysis (n=9), or both (n=25). Patients presented with gross hematuria (52%), nephrotic syndrome (15%) or asymptomatic urinary abnormality at average age of 6.5 year. Sensory-neural hearing loss was found in 15 patients and lens abnormality in 5. At the last follow up (range 0-21 years, average 9 years after presentation), 8 patients had lost their kidney function 13 years later than their presentation, while the other patients maintained their kidney function normally (n=48) or insufficiently (n=7). Mutational analysis of 39 patients revealed COL4A5 mutation in 34 patients (detection rate 87%); 12 missense mutations, 3 nonsense mutations, 6 frame-shifting deletions, 11 intronal mutations and in 2 cases large deletions encompassing more than 3 exons. Among the 22 patients in whom auditory function was tested, all of the 5 patients with truncation mutation of COL4A5 (frame-shifting or nonsense mutation or large deletion) showed sensory neural hearing loss, while patients with non-truncating mutation (n=17), only 35% had hearing loss.

Key Words : Alport 증후군, 유전자 진단
Alport Syndrome, Genetic Diagnosis