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Kidney Manifestations and Clinical Features of Nail-Patella Syndrome: A Retrospective Study

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Objectives : Nail-patella syndrome (NPS) is an autosomal dominant genetic disorder caused by pathogenic variants in LMX1B gene. It is characterized by classic tetrad of dysplastic nail, hypoplastic or absent patella, iliac horns, and elbow deformities, but other organs can also be involved. Kidney involvement, which typically present initially as proteinuria, is found in 30~50% of the patients. This study aimed to describe the clinical features and prevalence of kidney manifestations in patients with NPS.
Methods : A retrospective analysis was conducted on 35 patients who underwent genetic testing between 2001 and 2024 at Seoul National University Children's Hospital. These patients were part of a cohort of 65, with data on kidney involvement, including chronic kidney disease (CKD), hematuria, and proteinuria.
Results : Out of the 35 patients, 19 were boys. The patients were followed for mean 8.3 years until average age of 21.6 years. Kidney involvement was identified in 15 (42.9%) patients, diagnosed at mean age of 13.9 years. They included 5 patients with CKD, 4 patients with hematuria, 4 patients with proteinuria, and 2 patients with hematuria and proteinuria. Proteinuria was detected in patients aged mean 10.0 years, and CKD was diagnosed at mean 8.2 years. One patient developed nephrotic syndrome at age 4, and kidney biopsy revealed focal segmental glomerulosclerosis. She started peritoneal dialysis at age 4, underwent allograft nephrectomy at age 7 due to chronic rejection, and is now on hemodialysis. Pathogenic variants in LMX1B were found in 29 (82.9%) patients; 22 (62.9%) had variants in the LMX1 homeodomain, 3 (8.6%) in LIM-A, and 4 (11.4%) in LIM-B. No significant correlation was observed between the location of the pathogenic variant and the kidney involvement.
Conclusions : Patients with NPS are at risk of kidney diseases including proteinuria and CKD, some progressing to end stage kidney disease. Risk factors for nephropathy should be further studied.