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Clinical Characteristics and Long-term Renal Outcomes of Cystinuria in Korean Patients: A 30-year Single-center Experience

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Objectives : Cystinuria is a hereditary renal tubulopathy characterized by impaired cystine reabsorption in the proximal tubule. Due to the extremely low solubility of cystine, its elevated urinary excretion leads to recurrent nephrolithiasis. This study aims to analyze the clinical characteristics and long-term outcomes of cystinuria in Korean patients.
Methods : We conducted a retrospective review of patients diagnosed with cystinuria and managed at Samsung Medical Center between November 1994 and December 2025. Demographic data, clinical presentations, treatment history, and genetic test results (SLC3A1 and SLC7A9) were analyzed.
Results : A total of 12 patients (8 males, 66.7%) were included. The mean age at first symptom onset was 17.3 years, while the mean age at genetic evaluation was 34.6 years. Genetic analysis identified SLC3A1 mutations in 5 cases and SLC7A9 in 5 cases; 2 patients did not undergo genetic testing. The mean follow-up duration was 13.8 years (median 9.1 years). The overall mean eGFR was 78.6 mL/min/1.73m² at the initial visit and 88.3 mL/min/1.73m² at the last follow-up. Notably, the final mean eGFR was lower in the SLC3A1 group (67.8 mL/min/1.73m²) compared to the SLC7A9 group (116.6 mL/min/1.73m²). All patients required repeated surgical interventions and extracorporeal shock wave lithotripsy (ESWL), and two patients underwent unilateral nephrectomy.
Conclusions : Korean patients with cystinuria face a high burden of recurrent nephrolithiasis and repeated surgical interventions. Although overall renal function remained relatively stable during long-term follow-up, genotype-specific differences in eGFR suggest that patients with SLC3A1 mutations may require more intensive monitoring. Early genetic diagnosis and proactive stone management are essential to preserve long-term renal function.