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Clinical and genetic characterization of Chinese pediatric cystine stone patients

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Summary *Introduction:* Cystine stone is the only clinical manifestation in patients with cystinuria, which is an autosomal recessive inheritable disease. However, clinical and genetic data vary among patients in different countries.

Objective: To investigate the characteristics of Chinese pediatric cystine stone patients.

Patients and methods: Thirteen pediatric patients with cystine stones were evaluated in our clinic between 2012 and 2015. Gene mutations in SLC3A1 and SLC7A9 were investigated. Metabolic evaluation was also performed. Thirteen pediatric patients with calcium oxalate stones were selected as controls.

Results: Of these patients, eight were males and five were females. Average age at detection of the first stone was 6.8±5.2 years. Urinary stones in three of the 13 cystine patients were composed of cystine and calcium oxalate. The 63.6% of patients with upper urinary stones had bilateral stones. A total of 17 different missense mutations were identified, and 12 of these mutations were first reported in this study. Metabolic abnormalities could be detected in 77% of cystine stone patients. The most common metabolic abnormality was hyperoxaluria, followed by hypercalciuria and hypocitraturia. Compared with calcium stone patients, our cystine stone patients had a higher rate of bilateral stones, larger stone size, higher levels of