Genetics and Molecular Research

Clinical and genetic analyses of Chinese patients with Gitelman syndrome

M. Miao, C.Q. Zhao, X.L. Wang and Z.Y. Shan

Key Laboratory of Endocrine Diseases of Liaoning Province,
Department of Endocrinology and Metabolism, Institute of Endocrinology,
The First Affiliated Hospital, China Medical University, Shenyang, China

Corresponding author: X.L. Wang
E-mail: wlittlepear@163.com

Genet. Mol. Res. 15 (2): gmr.15027859
Received October 20, 2015
Accepted January 18, 2016
Published May 6, 2016
DOI http://dx.doi.org/10.4238/gmr.15027859

ABSTRACT. To evaluate the genotype-phenotype relationship of Gitelman syndrome in Chinese patients. We selected patients with Gitelman syndrome presenting hypokalemia. Medical history, clinical manifestations, laboratory test results, and imaging data of these patients were collected for analysis. Target gene sequencing was performed to evaluate the genotype-phenotype relationship. Gitelman syndrome was diagnosed based on medical history, clinical manifestations, laboratory test results, and imaging data. The causative gene for Gitelman syndrome, *SLC12A3*, and the causative gene for the classic Bartter syndrome, *CLCNKB*, were screened for disease-causing mutations by direct sequencing. Clinical diagnoses of ten patients were consistent with Gitelman syndrome. Disease-causing mutations in the *SLC12A3* gene were found in six patients. Among the variants, T60M in exon 1 was the hot spot in Chinese patients. Additionally, we found a small deletion of ACGG in exon 3 and L671P in exon 16; these have not been reported in previous studies. No disease-causing mutations were observed in the other four patients. Since mutations in the *SLC12A3* and *CLCNKB* genes are not present in all patients with

clinical manifestations of Gitelman syndrome, genetic screening after clinical diagnosis is essential.

Key words: Gitelman syndrome; *SLC12A3*; *CLCNKB*; Gene mutation

