Genetics and Molecular Research

Mutations in *WT1* in boys with sporadic isolated steroid-resistant nephrotic syndrome

Y. Yang[1,2,3]*, F. Zhao[1,2,3]*, X. Tu[4] and Z. Yu[1,2,3]

[1]Department of Pediatrics, Fuzhou Dongfang Hospital, Fuzhou, Fujian, China
[2]Department of Pediatrics, Fuzhou Clinical Medical College, Fujian Medical University, Fuzhou, Fujian, China
[3]Department of Pediatrics, Affiliated Dongfang Hospital, Xiamen University, Fuzhou Fujian, China
[4]Research Center for Molecular Diagnosis of Genetic Diseases, Fuzhou Dongfang Hospital, Fuzhou, Fujian, China

*These authors contributed equally to this study.
Corresponding author: Z. Yu
E-mail: zihuayu@vip.sina.com

Genet. Mol. Res. 15 (1): gmr.15017559
Received September 1, 2015
Accepted November 17, 2015
Published March 11, 2016
DOI http://dx.doi.org/10.4238/gmr.15017559

ABSTRACT. Mutations in the Wilms' tumor gene, *WT1,* can lead to syndromic steroid-resistant nephrotic syndrome and isolated steroid-resistant nephrotic syndrome. *WT1* mutations have been identified in the majority of children with Denys-Drash or Frasier syndrome. *WT1* mutations have not previously been identified in boys with sporadic isolated steroid-resistant nephrotic syndrome, but, recently, four boys with isolated nephrotic syndrome were identified to have *WT1* mutations. However, whether boys with sporadic isolated steroid-resistant nephrotic syndrome should be routinely subjected to mutation analysis of *WT1* has not been established. We examined 35 boys with sporadic isolated steroid-resistant nephrotic syndrome for mutations in *WT1*. Mutation analysis of all 10 exons of *WT1* was performed by polymerase chain reaction and direct sequencing. Karyotype analysis or Y chromosome identification was performed for all patients. A Y chromosome or a 46, XY karyotype was demonstrated for

all 35 patients. No causative *WT1* mutation was identified in any of the patients. The *WT1* mutation, IVS4+14T>C, which is not predicted to affect splicing, was identified in one patient who achieved complete remission after 8 weeks of oral prednisone treatment, indicating that IVS4+14T>C is not a causative mutation. Five *WT1* polymorphisms were also identified in some patients and controls. Our results suggest that mutation analysis of *WT1* should not be routinely performed for genetically defined boys with sporadic isolated steroid-resistant nephrotic syndrome.

Key words: Male; Mutation; Polymerase chain reaction; *WT1;* Steroid-resistant nephrotic syndrome

