Wilms’ tumor suppressor gene mutations in girls with sporadic isolated steroid-resistant nephrotic syndrome

Y.H. Yang1, F. Zhao1*, D.N. Feng1, J.J. Wang1, C.F. Wang1, J. Huang1, X.J. Nie1, G.Z. Xia1, G.M. Chen1 and Z.H. Yu1,2,3

1Department of Pediatrics, Fuzhou Dongfang Hospital, Fuzhou, Fujian, China
2Department of Pediatrics, Fuzhou Clinical Medical College, Fujian Medical University, Fuzhou, Fujian, China
3Department of Pediatrics, Affiliated Dongfang Hospital, Xiamen University, Fuzhou, Fujian, China

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

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ABSTRACT. Mutations in the Wilms’ tumor suppressor gene (WT1) can lead to syndromic forms of steroid-resistant nephrotic syndrome (SRNS) such as Denys-Drash or Frasier syndrome and can cause isolated SRNS. A mutation within WT1 is a frequent cause of sporadic isolated SRNS in girls. In a worldwide cohort of girls, the rate of occurrence was 10.8%. Previous reports have indicated that in Chinese girls, the detection rate of WT1 mutations is 16.7% for early onset isolated nephrotic syndrome. The detection rate of WT1 mutations in Chinese girls with sporadic isolated SRNS is unknown. We examined WT1 mutations in 14 Chinese girls with sporadic isolated SRNS using polymerase chain reaction and direct sequencing and studied a control group of 38 boys with sporadic isolated SRNS. We identified a WT1 mutation in 1 of 14 (7.1% detection rate) Chinese girls with sporadic...
isolated SRNS. No mutations occurred in WT1 in the remaining 13 girls or the control group. Our investigation supports the necessity of genetic examination for mutations in WT1 in girls with sporadic isolated SRNS.

**Key words:** Steroid-resistant nephrotic syndrome; Mutation; Genetics; Wilms’ tumor suppressor gene