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

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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

