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Mutational analysis of podocyte genes in children with sporadic steroid-resistant nephrotic syndrome

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ABSTRACT. Recent studies have demonstrated that mutations in 4 podocyte genes, *NPHS1*, *NPHS2*, *CD2AP*, and *WT1*, are associated with the pathogenesis of steroid-resistant nephrotic syndrome (SRNS). Systematic investigation of all 4 genes for sporadic SRNS in China has not been performed. We examined 10 Chinese children with sporadic SRNS who showed no response to immunosuppressive agents and 20 SRNS controls who exhibited a response to prolonged steroid or immunosuppressive treatment and achieved complete remission. We analyzed mutations in the 4 podocyte genes, *NPHS1*, *NPHS2*, *CD2AP*, and *WT1*. Mutational analysis was performed using polymerase chain reaction and direct sequencing. Of the 10 SRNS children who showed no response to immunosuppressive agents, the compound heterozygous *NPHS1* mutations 2677A>G (T893A)

and *142T>C were identified in 1 patient, while a heterozygous mutation in *WT1*, 1180C>T (R394W), was found in another patient. Of the 20 SRNS children showing complete remission who responded to prolonged steroid therapy or immunosuppressive agents, 4 heterozygous *NPHS1* mutations, 928G>A, IVS8+30C>T, IVS21+14G>A, and IVS25-23C>T, were identified in 4 patients and a heterozygous *CD2AP* mutation, IVS7-135G>A, was identified in 1 patient. Our results indicate the necessity of genetic examination for mutations in podocyte genes in Chinese SRNS children who show no response to immunosuppressive agents.

Key words: Immunosuppressive agents; Mutation; Podocyte genes; Steroid-resistant nephrotic syndrome; Steroid treatment