



## Novel *NPHS1* splice site mutations in a Chinese child with congenital nephrotic syndrome

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**ABSTRACT.** Congenital nephrotic syndrome (CNS) is defined as heavy proteinuria or nephrotic syndrome occurring before 3 months of age. It is characterized by early onset and progresses to end-stage renal disease. Recently, several genes associated with CNS have been identified, including *NPHS1* and *NPHS2*. Mutations in the *NPHS1* gene have been identified in patients with CNS in Finland with relatively high frequency. Thus far, only a few case reports about CNS have described an *NPHS1* mutation in China. In this study, mutational analyses of *NPHS1* and *NPHS2* were performed in a Chinese child with CNS. Mutations were analyzed in all exons and exon/intron boundaries of *NPHS1* and *NPHS2* in the patient and his parents as well as in 50 unrelated controls using polymerase chain reaction and

direct sequencing techniques. No mutations were detected in *NPHS2*. A novel splice site mutation (IVS11+1G>A) within intron 11 and a missense mutation within exon 8 (c.928G>A) in the *NPHS1* gene were detected in the child. The child's mother had normal urinalysis and a c.928G>A (D310N) heterozygous mutation, and his father had normal urinalysis and IVS11+1G>A. These were not identified in the 50 unrelated controls. The novel splice site mutation of IVS11+1G>A and a missense mutation at c.928G>A in *NPHS1* were found to cause CNS in this Chinese child.

**Key words:** Chinese; Congenital nephrotic syndrome; *NPHS1*