Two novel \textit{NPHS1} mutations in a Chinese family with congenital nephrotic syndrome

L.Q. Wu*, J.J. Hu*, J.J. Xue and D.S. Liang

State Key Laboratory of Medical Genetics, Xiangya Hospital, Central South University, Changsha, Hunan, China

*These authors contributed equally to this study.
Corresponding author: D.S. Liang
E-mail: liangdesheng@sklmg.edu.cn

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\textbf{ABSTRACT.} Congenital nephrotic syndrome of the Finnish type (CNF) is a lethal, autosomal recessive disorder mainly caused by mutations in the \textit{NPHS1} gene; it is found at a relatively high frequency in Finns. We investigated the disease-causing mutations in a Chinese family with CNF and developed a prenatal genetic diagnosis for their latest pregnancy. Mutation analysis was made of all exons and exon/intron boundaries of \textit{NPHS1} in the fetus, parents and 50 unrelated controls using PCR and direct sequencing. A heterozygous nonsense mutation within exon 20 (c.2783C>A) and a missense mutation within exon 17 (c.2225T>C) in \textit{NPHS1} were detected in the proband’s father and mother, respectively, but were not found in the fetus or in 50 unrelated controls. Two novel mutations of c.2783C>A and c.2225T>C in \textit{NPHS1} were found to be causative in this Chinese CNF family with no known Finnish ancestry. The most recent sibling did not inherit these two mutations and hence was unaffected with CNF. Determining the cumulative number and ethnic distribution of known mutations can help expedite further study of the pathogenesis of CNF.

\textbf{Key words:} Congenital nephrotic syndrome of Finnish type; \textit{NPHS1}; Mutation analysis; Prenatal genetic diagnosis; Chinese family