Mutations in \textit{NPHS1} in a Chinese child with congenital nephrotic syndrome

Z.H. Yu$^{1,2,3}$, D.J. Wang$^1$, D.C. Meng$^1$, J. Huang$^1$ and X.J. Nie$^1$

$^1$Department of Pediatrics, Fuzhou Dongfang Hospital, Fuzhou, Fujian, P.R. China  
$^2$Department of Pediatrics, Fuzhou Clinical Medical College, Fujian Medical University, Fuzhou, Fujian, P.R. China  
$^3$Department of Pediatrics, Affiliated Dongfang Hospital, Xiamen University, Fuzhou, Fujian, P.R. China

Corresponding author: Z.H. Yu  
E-mail: zihuayu@vip.sina.com

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\textbf{ABSTRACT.} Since the identification of the \textit{NPHS1} gene, which encodes nephrin, various investigators have demonstrated that the \textit{NPHS1} mutation is a frequent cause of congenital nephrotic syndrome (CNS); it is found in 98\% of Finnish children with this syndrome and in 39-80\% of non-Finnish cases. In China, compound heterozygous mutations in the \textit{NPHS1} gene have been identified in two Chinese families with CNS. To our knowledge, however, whether or not \textit{NPHS1} is the causative gene in sporadic Chinese CNS cases has not been established. We identified a homozygous mutation of \textit{NPHS1}, 3250insG (V1084fsX1095), in a Chinese child with sporadic CNS. This finding leads us to suggest that \textit{NPHS1} mutations are also present in sporadic Chinese CNS cases. This gives additional support for the necessity for genetic examination of mutations in the \textit{NPHS1} gene in Chinese children with sporadic CNS.

\textbf{Key words:} Congenital nephrotic syndrome; \textit{NPHS1}; Chinese; Nephrin