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Molecular genetic analysis of steroid resistant nephrotic syndrome: Detection of a novel mutation

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Background: Nephrotic syndrome is one of the most common kidney diseases in childhood. About 20% of children are steroid-resistant NS (SRNS) which progress to end-stage renal disease (ESRD). More than 53 genes are associated with SRNS which represent the genetic heterogeneity of SRNS. This study was aimed to screen disease causing mutations within NPHS1 and *NPHS2* and evaluate new potential variants in other genes.

Method: In first phase of study, 25 patients with SRNS were analyzed for NPHS1 (exon 2, 26) and all exons of *NPHS2* genes by Sanger sequencing. In the second phase, whole exome sequencing was performed on 10 patients with no mutations in NPHS1 and NPHS2.

Result: WES analysis revealed a novel mutation in *FAT1* (c.10570C>A; Q3524K). We identified 4 pathogenic mutations, located in exon 4 and 5 of *NPHS2* gene in 20% of patients (V180M, P118L, R168C and Leu156Phe). Also our study has contributed to the descriptions of previously known pathogenic mutations across WT1 (R205C) and SMARCAL1 (R764Q) and a novel polymorphism in CRB2.

Conclusion: Our study concludes that mutations of exon 4 and 5 *NPHS2* gene are common in Iranian and some other ethnic groups. We suggest conducting WES after *NPHS2* screening and further comprehensive studies to identify the most common genes in the development of SRNS, which might help in clinical impact on management in patients with SRNS.

Biography

Azadeh Shojaei is working in Department of Medical Genetics and Molecular Biology. She is a Faculty of Medicine in Iran University of Medical Sciences, Tehran, Iran. She has published 5-7 papers in publication.

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