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Genetic mutation in Egyptian children with steroid-resistant nephrotic syndrome

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Nephrotic syndrome is the commonest etiology of proteinuria in children. Steroid-resistant nephrotic syndrome (SRNS) is defined by resistance to standard steroid therapy, and it continues to be one of the most intractable etiologies of renal failure. Molecular studies discovered specialized molecules in podocytes that play a role in proteinuria. Mutations in NPHS2 that encodes for podocin constitute a frequent cause of SRNS worldwide. This study aimed to screen for podocin mutations in SRNS Egyptian children and their parents. Our study included patients from 10 unrelated Egyptian families diagnosed with SRNS. Mutational analysis of the *NPHS2* gene was performed by polymerase chain reaction amplification of the whole coding region of the gene and direct sequencing. Positive consanguinity was detected in five cases, and four of them had a positive family history of SRNS in a family member. Mutational analysis of NPHS2 revealed pathogenic mutations in four cases (40%) including a novel missense in one patient (c.1A>T; p.M1L). Our study concluded that mutations of *NPHS2* gene are common among Egyptian children with SRNS. We support a model where ethnicity plays an important role in specific NPHS2 mutations, since a novel mutation was found in one patient in this study. Future study on a large number of Egyptian patients with SRNS is warranted to identify the actual genetic contribution of this gene in the development of SRNS in our population, which might help in patients' prognosis and management.

Biography

Manal M Thomas is an Assistant Professor in Clinical Genetics department - Human Genetics and Genome Research division - National Research Centre, Cairo, Egypt. She has completed Master degree in Pediatrics from Medical School of Cairo University and PhD degree from Ain Shams University. She is a member of National Society of Human Genetics. She has many publications in medical genetics.

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