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Autor	WILLIAM ISRAEL CARDOSO DA SILVA
Orientador	ELVINO JOSE GUARDAO BARROS

NPHS2 Gene Polymorphisms in Sporadic and Familial Focal Segmental Glomerulosclerosis

William Israel Cardoso da Silva[1]

Elvino José Guardão Barros[2,3]

[1]Graduating in Medicine, Federal University of Rio Grande do Sul, Porto Alegre, RS, Brazil; [2]Teacher, Department of Internal Medicine, Faculty of Medicine, Federal University of Rio Grande do Sul, Porto Alegre, RS, Brazil; [3]Department of Nephrology, Hospital de Clínicas, Porto Alegre, RS, Brazil.

Introduction: NPHS2 gene variants are found in 5% to 30% of patients with Focal Segmental Glomerulosclerosis (FSGS), and are associated with corticosteroid-resistant nephrotic syndrome (SRNS).

Objectives: The objective of this study was to determine the prevalence of NPHS2 variants in patients with FSGS in their familial and sporadic forms.

Methods: The sample consisted of 40 children and 70 adults diagnosed with FSGS confirmed by renal biopsy. Age at disease onset, presenting renal syndrome, renal function, steroid resistance, and clinical outcomes were evaluated. Genotyping for the three single nucleotide polymorphisms (SNPs) was performed by real-time polymerase chain reaction (RT-PCR). Two polymorphisms were found in exon 5 - p.R229Q (rs61747728) and p.A242V (rs61747727), and one in exon 3 - p.R138Q (rs74315342). Gene variants were correlated with ethnicity, clinical presentation, treatment response, and renal outcomes.

Results: Among the 40 children analyzed, 8 (20%) had familial FSGS and 32 (80%) had sporadic FSGS. Only 3 (4.3%) had familial FSGS; 67 (95.7%) had the sporadic form. Overall, 49 adults (70%) and 36 children (90%) had SRNS. Among children, variants were detected in only 2 (5%) with sporadic FSGS: p.R229Q in one and p.A242V in another. Among adults, the analyzed polymorphisms were present in 9 patients (12.9%), all with sporadic FSGS: 4 had p.R229Q and 5 had p.A242V. No patient had the p.R138Q polymorphism. All patients carrying the p.R229Q variant were white, while 67% of carriers of the p.A242V variant were black. When ethnicity, clinical and renal outcomes were correlated with the variants, there was no significant difference for both children and adults; only a trend of higher proteinuria at the end of follow-up (p=0.06) in cases carrying a variant was found. Comparing prolonged cyclosporine use (> 3 years) between patients with variants (n=4, 36.4%) and those without variants (n=17, 17.2%), there was no statistical difference between the groups (p=0.124). In relation to ethnicity, 17% of African descendants had variants against 8% of Caucasian patients (p=0.184).

Conclusion: In these patients with familial or sporadic FSGS, the prevalence of p.R229Q and p.A242V variants in children was 5% and in adults 12.9%; no patient presented the p.R138Q variant. There was no association between the presence of NPHS2 variants with ethnicity or dependence on immunosuppressive treatment with cyclosporine.