عنوان مقاله:

NPHS۲ gene in steroid-resistant nephrotic syndrome: prevalence,clinical course,and mutational spectrum in South-West Iranian children

محل انتشار:

ششمین کنگره بین المللی انجمن نفرولوژی کودکان ایران (سال: ۱۳۹۷)

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خلاصه مقاله:

Introduction: Mutations in podocin (NPHS۲) gene have the key role in the pathogenesis of steroid-resistant nephrotic syndrome (SRNS) in children, but data is scarce regarding their prevalence and natural course among different all ethnic groups. This study was aimed to demonstrate the spectrum of NPHS۲ mutations in children with SRNS and to compare the clinical course of disease in patients with and without mutation.Materials and Methods: All ۸ exons of NPHS۲ were sequenced in ۹۹ children, including ۴۹ with SRNS and ۵۰ with steroid-sensitive nephrotic syndrome (control group) by DNA sequencing.Results:The prevalence rates of NPHS۲ gene mutation among children with SRNS and SSNS were ۳۱% and ۴%, respectively. The prevalence rates of mutation among familial and sporadic forms were ۵۷% and ۲۶%, respectively. Thirty-three percent of the children experienced recurrence of primary disease after kidney transplantation, none of whom had a mutation. The clinical response to treatment was poorer in children with mutation in comparison with patients without mutation (۱۲% versus ۳۲%, respectively; odds ratio, ۳.۲۹, ۹۵% confidence interval, ۰.۴۰ to ۲۵.۶۴). Patients with and without mutation could not be differentiated by demographic and histological features, glomerular filtration rate at onset, hypertension, progression to end-stage renal disease, and proteinuria.Conclusions: Mutations of NPHS۲ gene are frequent among Iranian children with SRNS. Regarding similar clinical features in patients with and without mutation and poor response to pharmacotherapy in patients with mutation, a molecular approach might be necessary for different treatment plans and prediction of prognosis

کلمات کلیدی:

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