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Evaluation of association between UMOD gene variants and renal outcomes in children with nephrotic syndrome

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Background and Aims: In recent genome-wide association studies have identified common variants in the uromodulin (UMOD) gene relating to eGFR and risk of chronic kidney disease. In this study, we aimed to explore associations of UMOD gene variants with renal outcome and clinical findings in children with nephrotic syndrome (NS).

Method: The children with NS were enrolled in this study. Demographic characteristics, clinical and laboratory data, and presence of UMOD rs12917707 and rs11864909 variants of children were evaluated.

Results: A total of 120 children, 73 boys (60.8%), median age at diagnosis 65.88 ± 45.06 months, median follow-up 82.93 ± 54.97 months, were included in the study. A total of 36, 15 and 69 children were in groups I, II and III, respectively. In the group 1 (ESRD), it was found that median age at diagnosis was higher, the follow-up period was longer, the rate of consanguineous marriage between the parents was higher, and the rate of presence of NS in the family was more frequent ($P < 0.05$). The pathological allelic distributions of the UMOD rs12917707 and rs11864909 variants were found to be similar in the patient with steroid-resistant, steroid-sensitive, steroid-dependent, frequently relapsing and complete remission NS subgroups. UMOD gene rs11864909 pathological variant distribution was found to be higher in group 1 (ESRD) than the others ($P = 0.04$). Similarly gene rs11864909 pathological variant distribution was higher in patients with steroid resistant NS and ESRD than the others. And also gene rs11864909 pathological variant distribution was higher in patients with hematuria at diagnosis than the others ($P = 0.003$)

Conclusion: Our results suggest that UMOD gene rs11864909 variant associated with the renal outcomes in children with NS