GENOTYPING FOR PERSONALIZED MEDICINE IN ADULT PATIENTS WITH PRIMARY FOCAL SEGMENTAL GLOMERULOSCLEROSIS

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INTRODUCTION AND AIMS

In the search of genetic disorders associated with the development of primary glomerulopathies, multiple mutations and variations have been identified so far.[1] The aim of this study is to investigate the genetic alterations that may be responsible for the development of primary focal segmental glomerulosclerosis (FSGS) in adult patients.

METHODS

In this single-center Turkish cohort, high-throughput sequencing using custom-designed multi-gene next generation sequencing panel for podocyte disorders was used to investigate 31 genes in 30 adult patients presented with a family history of kidney disease and biopsy-confirmed global or segmental sclerosis.

RESULTS

Of these 30 patients, 16 (53.3%) were male, and mean age was 42±10 years. Renal replacement therapies were initiated in 20 (66.6%) patients, 17 of them underwent a renal transplantation. Two transplant recipients experienced recurrent disease. In analyses, 20 (66.6%) patients harbored mutations and variations in 12 genes, most frequently in NPHS2 [nephrosis 2, idiopathic, steroid-resistant (podocin)] (6 patients) and TTC21B (tetratricopeptide repeat domain-containing protein 21B) (3 patients). The other susceptible genes were as follows: COQ2 (coenzyme Q2, polyprenyltransferase), COQ6 (coenzyme Q6, monooxygenase), ADCK4 (AARF domain-containing kinase 4), LMX1B (LIM homeobox transcription factor 1 beta), WT1 (Wilms tumor 1), MYH9 (myosin heavy chain 9), CD2AP (CD2 associated protein), PLCE1 (phospholipase C epsilon 1), KANK2 (KN motif and ankyrin repeat domains 2), MAGI2 (membrane-associated guanylate kinase inverted 2). One of the transplant recipients who experienced recurrent disease harbored both NPHS2 and TTC21B mutations.

CONCLUSIONS

Gene-testing may be useful as a non-invasive diagnostic tool for adults in order to identify the susceptible genes for FSGS. Further studies with larger cohorts are needed.

REFERENCE

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