NPHS2, R229Q gene polymorphism in Iranians with late onset steroid resistance nephritic syndrome

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- **Introduction:** Nephritic syndrome (NS) is defined by proteinuria, peripheral edema and hypoalbuminemia . Depending on the response to standard steroid therapy it is sub-classified to steroid-sensitive NS (SSNS) and steroid-resistance NS (SRNS). Mutations in several genes including; NPHS2, have been implicated in SRNS. NPHS2 genes R229Q polymorphism (p.R229Q) is associated with adolescent- or adult onset SRNS in European and South American population. Here we investigated this polymorphism among a group of Iranian-Azeri patients with primary SRNS.
- **Materials and methods**: All patients in our cohort had the late adolescence or adult onset disease. They had the pathologic feature of FSGS and clinical feature of steroid unresponsiveness. All individuals had a primary FSGS without any obvious secondary causes. We also studied a group of age and sex matched individuals without any renal disease as controls
- **Results**:A total number of 25 patients (M/F 15/10, age; 26.6+/- 8.0 years) with primary FSGS and 35 controls (M/F 17/13, age 26+/- 8.7) were studied. Serum creatinin levels of patients and their 24 hours protein excretion at the time of study were; 2.4+/- 1.94 mg/dl and 2830+/- 981 mg/dl respectively. Molecular study showed no R229Q polymorphism either in patients or in controls.
- **Discussion:** NPHS2 gene,s R229Q polymorphism is not important in Iranian –Azeri population with SRNS and other mutated genes should be considered in these patients.

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