

MEFV mutation analysis in patients with idiopathic mesangioproliferative glomerulonephritis and its relation to changes in renal function parameters

**Background and aim:** Familial Mediterranean fever (FMF) is an autosomal recessive autoinflammatory disorder characterized by self-limited recurrent bouts of fever and painful episodes of sterile serositis. A less frequent but important complication of FMF is glomerulonephritis (GN) including mesangioproliferative subtype. Since it has been found that the responsible gene in the pathogenesis of FMF is MEFV, it might be thought that there could be an association between mutations of this gene and the pathogenesis of idiopathic mesangioproliferative GN. The present study aimed to investigate this possibility.

**Methods & Materials:** A total of 39 patients with pathologically proven mesangioproliferative GN and 156 healthy subjects were recruited from Imam Reza teaching Hospital during an 18-month period of time. MEFV mutations including M694V, M680I, E148Q, R761H, R408Q, A744S, F479L, M694I, V726A, P369S and E167D were documented in both case and control groups. Changes in serum creatinine, as well as the response to treatment on the basis of 24-h proteinuria after 6 months were outcome measures.

**Results:** Patients were 22 males and 17 females with a mean age of  $40.13 \pm 14.42$  years (range: 16-57). Mutations of MEFV gene was more common among patients than in controls (28.2% vs. 17.3%), but the difference did not reach a statistically significant level ( $p=0.12$ ). Changes in serum creatinine within 6 months after treatment was different between patients with and without MEFV gene mutations, significantly better in those without such mutations ( $p=0.04$ ). Complete response to treatment at the endpoint was also significantly more frequent in the group of patients without MEFV mutations (18.2% vs. 67.9%,  $p=0.01$ ).

**Conclusion:** Although MEFV mutations may be associated with idiopathic mesangioproliferative GN, they definitely influence outcome of treatment in such patients.

**Keywords:** MEFV Gene, Idiopathic Mesangioproliferative Glomerulonephritis, Prognosis, Familial Mediterranean Fever