

Familial Mediterranean Fever E148Q Mutation, Episodic Fever and Kidney Allograft Dysfunction

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Dear Editor,

Familial Mediterranean fever (FMF) is a recessively inherited disease associated with mutations in the FMF gene (*MEFV*), which encodes the pyrin protein. AA amyloidosis is the most devastating complication of FMF. Nonamyloid renal lesions also have been reported including vasculitis, focal diffuse glomerulonephritis, and immunoglobulin A nephropathy. *M694V* is the most common mutation of *MEFV* gene, but *E148Q* pyrin mutation is also common in some regions.¹

A 25-year-old man with a history of renal transplantation 11 years earlier was admitted to our center because of high fever and serum creatinine elevation. On admission, his temperature was 39°C and the blood laboratory findings were as follows: creatinine, 3.2 mg/dL; blood urea, 107 mg/dL; sodium, 139 mEq/L; potassium, 4 mEq/L; uric acid, 10 mg/dL; hemoglobin, 12 mg/dL; leukocyte count, $9 \times 10^9/L$; and platelet count, $12.5 \times 10^9/L$. Cytomegalovirus immunoglobulin M antibody was negative. Urinalysis was normal. Cyclosporine blood level was 126 ng/mL. During the past year, he had at least 5 similar episodes and 2 times of hospitalization. During each episode he had taken broad-spectrum antibiotics without any visible effect, and each time, episodes were remitted suddenly. His primary kidney disease was urinary calculus and obstruction and he had not have any history of such febrile episodes before transplantation or thereafter. These episodes had started since 2 years ago.

The FMF *MEFV* gene was studied for common mutations in exons 2, 3, 5, and 10. Heterozygote mutation of E148Q was detected, but the results were negative for M694V, M680I, R761H, R408Q,

A744S, F479L, M694I, V726A, P369S, and E167D mutations. We started colchicine during his last febrile episode, and the response was visible and febrile episode never happened during the 6 months of follow-up.

We have reported another patient with heterozygote E148Q mutation who presented with massive proteinuria and acute glomerulonephritis. His clinical condition improved after treatment with corticosteroid and colchicine.² It seems that E148Q mutations are heterogeneous and with unusual clinical presentations. They should be studied in patients with suspicious symptoms in endemic regions. Unknown genetic modifiers may have some role in E148Q-associated clinical symptoms.²⁻⁴

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