Dear Editor,

Protracted febrile myalgia syndrome (PFMS) is one of the rare vasculitic diseases that affect patients with Familial Mediterranean Fever (FMF). PFMS was described in patients with FMF by Langevitz et al in 1994 (1). Widespread myalgia, high fever, vasculitic rashes, arthritis or arthralgia, abdominal pain, similar symptoms can be seen. Higher erythrocyte sedimentation rate (ESR), hyperglobulinemia, normal serum levels of creatine phosphokinase can be seen and electromyographic (EMG) findings are non-specific (2). Homozygous mutation for M694V gene is often detected in PFMS patients (3). This syndrome may develop in patients under colchicine prophylaxis, without corticosteroid treatment and the duration of clinical findings may take up 4-6 weeks (2). We presented a patient who were admitted to our clinic with clinical manifestations of protracted febrile myalgia and diagnosis of FMF made after identifying compound heterozygocity for M694V/M680I mutation of the MEFV gene.

A six year-old girl presented with inability to walk, widespread myalgia and rash. She had intermittent abdominal pain and fever for three months. Four days before she had admitted to the hospital, widespread myalgia and rash began to manifest, and then tumescence on bilateral ankles occurred. She was a well-developed girl with normal blood pressure. Her body temperature was 39.5°C and there was a widespread rash on her body, tumescence and pain on both ankles. Laboratory analyses revealed normal urinary test results hemoglobin 10.8 g/dL, mean corpuscular volume 80 fl, leukocyte count 10.800/mm³, platelets 450.000/mm³; transaminases, creatine phosphokinase (CPK), blood urea nitrogen, and creatinine were within limits, c-reactive protein (CRP) (11.2 mg/dl) and ESR (93 mm/h) were elevated. ECG, EMG, chest x-ray, and abdominal ultrasonography were normal. Serology for brucellosis, salmonellosis, toxoplasmosis, hepatitis viruses, cytomegalovirus and Epstein-Barr virus were negative. Blood, urine, and throat cultures were negative for bacterial agents. Levels of the C3 and C4 complements were normal; rheumatoid factor, antinuclear antibody, antidsDNA, and anti-neutrophil cytoplasmic antibody were negative. Immunoglobulin values were normal. Compound heterozygocity for M694V/M680I mutation of the MEFV gene was detected.

Diagnosis of PFMS was considered based on the presence of fever, widespread muscle pain and rash, normal EMG, elevated CRP and ESR. Prednisolone (2 mg/kg/d) was started. Her symptoms resolved and
acute phase reactants declined in a week. FMF was diagnosed based on mutation of the MEFV gene and clinical findings. Colchicine at a dosage of 2 x 0.5 mg was started.

FMF is associated with several types of vasculitis, including polyarteritis nodosa, Henoch-Schönlein purpura (HSP) and Behçet’s disease. The clinical spectrum of FMF has recently been expanded and PFMS is now a frequently recognized component in these patients (4). PFMS is seen in only a small proportion of FMF patients, and M694V, V726A, and E148Q mutations are common in these patients (5). Kaplan et al reported PFMS clinics in 33% of the patients with FMF and detected M694V mutation in 93% of the PFMS patients (6). PFMS is characterized by severe debilitating myalgia and high fever, occasionally accompanied by abdominal pain, diarrhea, arthritis and transient vasculitic purpura mimicking HSP (7). Kavukcu et al. reported 4 patients with PFMS presenting with skin lesions, arthritis/arthralgia or recurrent myalgia (8). Our patient had intermittent abdominal pain and fever for 3 months and had rash on admission. She was compound heterozygote for M694V/M680I mutation of the MEFV gene.

A few patients have classic symptoms of FMF when they present with a clinical picture of PFMS (9). Introduction of genetic analysis for MEFV mutations would possibly change the diagnostic criteria for FMF and may be helpful in providing counselling service and optimum therapeutical approach for the patients and their families (10). Our patient presented without typical symptoms of FMF. Therefore, we started colchicine after detection of compound heterozygocity for M694V/M680I mutation of the MEFV gene.

REFERENCES