Frequency of common MEFV gene mutations in different phenotypes of patients with Behjat syndrome in Ardabil province

Abstract

Background: Behcet's disease is a rare severe recurrent inflammatory disorder that affects several organs of the body. Since FMF and BD affect almost a certain population, both diseases can mimic the other clinically, and these two diseases sometimes occur in the same family and in the same patient, also from Since Iran is among the countries with a relatively high prevalence of Behjat and a small number of studies have been performed to investigate MEFV gene mutations in Behjat patients, in this study we aimed to investigate the mutations of this gene and its relationship. Let's deal with the clinical signs of the disease in our region - Ardabil - which has a relatively high prevalence of Behcet.

Aim: To determine Frequency of common MEFV gene mutations in different phenotypes of patients with Behjat syndrome in Ardabil province

Methods and material: In this study, about 50 patients with Behcet's syndrome were diagnosed according to the ICBDR criteria (For oral pests, genital pests and ocular lesions, 2 points each, and for skin symptoms, vascular symptoms, positive pathogenesis test and neurological symptoms, 1 point each. A score of 4 or higher indicates disease.) were included in the study. Genomic DNA was extracted from peripheral blood leukocytes using standard protocols. The extraction of the gene were done by Boiling method and buffers and lubricants from Vienna Lab Diagnostics GmbH. PCR amplification were performed and MEFV sequences were evaluated and mutations detected. Also, for comparison, 224 healthy individuals were included in the study in terms of Behcet's syndrome.

Results: In the present study, 50 patients with Behcet's syndrome were included in the study. The mean age of patients was 38.68 years with a standard deviation of 11.004 years. Most of the patients were women with 56%. 66% (33 patients) mentioned that their parents had no family relationship. In the healthy group, out

of 224 people, 113 were men (50.4%) and 111 were women (49.6%). Among 50 patients with Behcet's syndrome, 38 (76%) had no mutations and 12 (24%) had mutations in this gene. 7 patients (14%) had E148Q mutation, 3 patients (6%) had V726A mutation, 1 patient (2%) had M694V mutation and 1 patient (2%) had A744S mutation.
Among healthy individuals, out of a total of 224 patients, 57 patients (25.4%) had mutations, of which 4 patients (1.5%) were homozygous and 53 patients (23.7%) were heterozygous. Also, 167 patients (74.6%) had no mutations. The most common type of mutation in both groups was E148Q.

Conclusion: In total, 24% of patients with Behcet's disease had mutations in MEFV gene, the most common case was E148Q mutation with 14%. These results were similar to the healthy population in terms of Behcet's syndrome.

Keywords: Behcet's syndrome, MEFV, mutation