Mutation of pyrin E148Q in patients with refractory rheumatoid arthritis

Ahmadi A. MD., Kolahi S. MD.

Department of internal medicine, Imam Reza Hospital, Faculty of medicine, Tabriz University of Medical Sciences, Iran

Abstract

Introduction: Rheumatoid Arthritis (RA) is among the most common chronic unknown diseases and cause the majority of adult disabilities. Due to the high prevalence of Familial Mediterranean fever in patients and overlap of autoimmune diseases, we decided to study the Mutation of pyrin E148Q in patients with refractory rheumatoid arthritis in the northwest of Iran. **Methods:** In a cross-sectional descriptive-analytic study carried out on patients with refractory rheumatoid arthritis, Mutation of pyrin E148Q in patients with refractory rheumatoid arthritis in northwest of Iran was studied. A total of 50 patients with refractory rheumatoid arthritis and 50 patients with rheumatoid arthritis in remission (control group) were selected and included in the study. Next, blood samples were taken from the participants and DNAs were extracted using the standard method. The samples were examined for mutation of pyrin E148Q of exon 2 of the MEFV gene using the sequencing method. **Results:** Mean age of patients with refractory rheumatoid arthritis was 43.2 ± 7.8 year and in patients of control was 38.6 ± 7.7 year. Mean of rheumatoid arthritis duration was 49.8 ± 36.7 month in patients with refractory R.A and 54.9 ± 42.6 month in patients of control. In patients suffering from refractory RA concerning exon 2 of the MEFV gene, mutation of pyrin E148Q was in 6 patients (17.6%) and in control group was in 5 patients (14.7%). **Conclusion:** Significant differences were not found between mutations of pyrin E148Q in patients' of both groups. In patients with refractory RA other mutations were observed in Exon 2 of MEFV gene include: K114E in 20.5%, A106S in 5.8% and S108C in 2.9%.

Key words: Refractory Rheumatoid Arthritis, MEFV gene, Iran, Exon 2.