Kawasaki disease and familial mediterranean fever gene mutations, is there any link?

Abstract

Background and objective: Kawasaki disease (KD) is an acute febrile, self-limiting, and

systemic vasculitis of unknown etiology. MEFV gene has a major role in autoinflammatory

disorders and innate immune reactions. Several reports revealed that MEFV gene mutations are

associated with systemic vasculitis. The aim of this study was to determine the association

between KD and MEFV gene mutations.

Methods: The peripheral blood of 30 patients who were diagnosed with KD based on ACC

criteria were collected and the samples screened for the 12 common pathogenic variants

according to manufacturer's instructions.

Results: The mean age of patients (13 females and 17 males) was 7.7 years. Ten percent of

patients showed a mutation, that was meaningfully (p<0.05%) lower than that of healthy controls

(25%). E148Q was shown in two patients and compound heterozygous (E148Q-M680I) was

detected in one of them with lack of FMF presentations. No significant and meaningful

associations were detected between the MEFV gene variant alleles and KD.

Conclusion: Unlike in other types of pediatric vasculitis, this study did not reveal any significant

association between the MEFV gene mutations and KD, moreover, because of the lower

frequency of mutations in these patients, it seems that this gene has a modifier and/or protective

role in KD.

Key Words: FMF, vasculitis, MEFV gene, Kawasaki disease