PEDIATRIC RHEUMATOLOGY

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MEFV and NLRP3 gene variants in children with pfapa syndrome in slovenia

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Introduction

PFAPA syndrome is the most common autoinflammatory fever disorder in childhood, characterized by recurrent fever, aphthous stomatitis, pharyngitis and adenitis. Mutations in the *MEFV* and *NLRP3* genes are known to cause syndromes with PFAPA overlapping symptoms (Familial Mediterranean Fever and Cryopyrin-Associated Periodic Syndrome), which are rarely reported in patients from Slovenia.

Objectives

The aim of the study was to assess the frequency of *MEFV* and *NLRP3* gene variants in pediatric patients with PFAPA syndrome from Slovenia in order to determine whether genes involved in other autoinflammatory diseases, might play a role in PFAPA pathogenesis.

Methods

We collected clinical and laboratory data of PFAPA patients under the age of 5, who were followed at the University Children's Hospital Ljubljana. All 10 exons of *MEFV* gene and 9 exons of *NLRP3* gene, including intron/exon regions of both genes were directly sequenced.

Results

In total, 30 PFAPA patients were tested for *MEFV* and *NLRP3* gene variants. Mean age at the syndrome onset was 2.1±1.3 and at diagnosis 4.2±1.8 years. 19(63%) patients were male and 11(37%) were female. Mean duration of episode was 3.5 days, mean interval between the episodes was 3.5 weeks. Most common symptoms beside fever were pharyngitis and cervical adenitis in 90% and aphtosis (always or sometimes) in 63%.

Overall, 10 patients (33%) were found to have 11 variants, all in heterozygous state. 6 patients have Q703K variant in *NLRP3*, one E148Q in *MEFV* and one combination of I591T in *MEFV* and Q703K in *NLRP3*. Novel variant in *NLRP3*, P200T, was identified in one patient. One girl was found to have known variant in *NLRP3* gene, S726G, which is associated with CINCA syndrome. This girl has had typical PFAPA symptoms, but she also has epilepsy and mild developmental delay.

Conclusion

Five different *MEFV* and *NLRP3* gene variants were identified in 10 of 30 PFAPA patients with *MEFV* variants found in 2 patients and *NLRP3* variants in 9. Our results indicate genetic heterogeneity of PFAPA population and possible overlap with other periodic fever syndromes.

Disclosure of interest

None declared.

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