



MEETING ABSTRACT

Open Access

PW01-020 – MEFV mutations carrier rate in Central Europe

M Debeljak^{1*}, N Toplak², N Abazi³, M Kolnik⁴, B Szabados⁵, V Mulaosmanovic⁶, J Radović⁷, J Vojnović⁷, T Constantin⁵, D Kuzmanovska³, T Avčin²

From 7th Congress of International Society of Systemic Auto-Inflammatory Diseases (ISSAID) Lausanne, Switzerland. 22-26 May 2013

Introduction

Familial Mediterranean fever (FMF) is an autosomal-recessive disorder characterized by recurrent attacks of fever and serositis common in eastern Mediterranean population. Over 160 mutations have been identified in MEFV gene responsible for FMF. The most common mutations in MEFV gene are E148Q, M694I, M694V, V726A and M680I. The distribution pattern of MEFV mutation along the Mediterranean Sea is not uniform; eastern populations have the highest number of carriers (20-39%), whereas western Mediterranean populations are practically unaffected.

Objectives

The aim of this study is to determine the carrier rate in healthy Macedonian, Serbian, Slovene, Bosnian and Hungarian population.

Methods

We screened 100 healthy subjects from all 5 populations. Exon 10 was PCR amplified and screening was performed with dHPLC. All amplicons with detected nucleotide changes were subsequently sequenced with ABI prism 310 genetic analyzer. Amplicons of exon 2 were directly sequenced.

Results

Heterozygous mutations were found in 4% of apparently healthy Hungarians, 7% of Slovenians, 8% of Bosnians, 11% of Serbians and in 16% of apparently healthy Macedonians. Mutations found in Hungarian population were as follows: V726A (1), K695R (3). Mutations found in

Slovenian population were: V726A (1), K695R (5) and E148Q (1). Mutations found in Bosnian population were: V726A (1), K695R (6) and F756C (1). Mutations found in Serbian population were: E148Q (6), K695R (5). Mutations found in Macedonian population were as follows: E148Q (8), K695R (7) and M694V (1).

Conclusion

We found higher than expected carrier rate in all populations, from 4% to 16%. It is interesting to note that more than half (60%) of detected carriers in all analyzed populations has K695R mutation.

Disclosure of interest

None declared.

Authors' details

¹Center for Medical Genetics, University Children's Hospital, University Medical Center, Ljubljana, Slovenia. ²Department of Allergology, Rheumatology and Clinical Immunology, University Children's Hospital, University Medical Center, Ljubljana, Slovenia. ³University Children Hospital, Medical Faculty, Ss. Cyril and Methodius University, Skopje, Macedonia, the Former Yugoslav Republic of. ⁴Department of Allergology, Rheumatology and Clinical Immunology, University Children's Hospital, University Medical Center, Ljubljana, Slovenia. ⁵Unit of Paediatric Rheumatology, 2nd Department of Pediatrics, Semmelweis University Budapest, Budapest, Hungary. ⁶Children's Hospital University, Clinical Center, Sarajevo, Bosnia and Herzegovina. ⁷Department of Pediatric Rheumatology, Faculty of Medicine, University Niš, Niš, Serbia.

Published: 8 November 2013

doi:10.1186/1546-0096-11-S1-A73

Cite this article as: Debeljak et al.: PW01-020 – MEFV mutations carrier rate in Central Europe. *Pediatric Rheumatology* 2013 **11**(Suppl 1):A73.

¹Center for Medical Genetics, University Children's Hospital, University Medical Center, Ljubljana, Slovenia
Full list of author information is available at the end of the article