

POSTER PRESENTATION

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Impact of MEFV genotype in Caucasian children with periodic fever

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Introduction

Despite FMF is considered an autosomal recessive disease caused by mutations of *MEFV*, one third of patients carries one mutation only.

Aim

To analyze the actual impact of MEFV mutations in children with periodic fever.

Methods

113 caucasian patients carrying MEFV mutations (46 with mutations in two alleles, 67 heterozygous) and 205 genetically negative patients for MEFV, TNFSF1A and MEFV (70% with a PFAPA phenotype) were analyzed. The fol-

lowing groups were considerd: patients with: i) 2 high penetrance mutations (M694V, M694I, M680I), ii) 1 high, 1 low penetrance mutation, iii) 2 low penetrance mutations, iv) 1 high penetrance mutation, v) one low penetrance mutation, vi) genetically negative.

Results

Patients with two mutations displayed a higher prevalence of chest pain (p = 0.001), pleurisy (p = 0.003) and severe abdominal pain (p = 0.002) in respect to heterozygous patients, which clinical phenotype was more similar to that presented by genetically negative patients, with an higher prevalence of erythematous (p = 0.01) and exudative (p = 0.009) pharyngitis, enlarged cervical

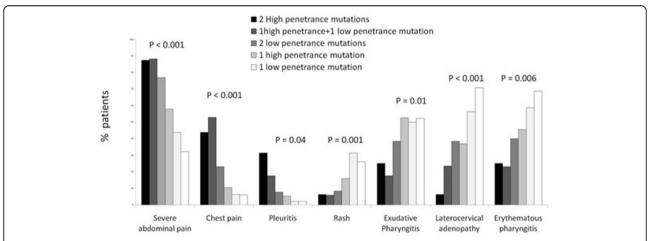


Figure 1 Prevalence of the clinical manifestations associated to fever attacks in patients with different MEFV genotypes (see text) and in patients with periodic fever negative for mutations of MEFV, MVK and TNFSRF1A genes. P values were assessed by a Chi-square test for trend.

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lymph nodes (p = 0.002). The frequency of "FMF-like symptoms" decreases from patients carrying two high penetrance mutations towards patients with a single low penetrance mutation with a specular increase of "PFAPA-like symptoms" (Figure 1).

Conclusions

The present study shows a dosage effect of MEFV mutations not consistent with a pure autosomal recessive disorder. A dominant negative or gain of function effects or variants of still unidentified modifier genes may influence the presence of a FMF phenotype in heterozygous patients.

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