Pediatric Rheumatology

Poster presentation

The clinical spectrum of 94 French patients carrying a single mutated MEFV allele

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Background

The reason why FMF carriers may develop the disease is still unclear. We assessed the clinical characteristics of French FMF patients carrying a single MEFV mutation

Methods

A retrospective chart review of patients referred to us with FMF symptoms. Systematic genetic screening of exon 2 and 10 was performed in MEFV gene. A subset of patients was also investigated for other hereditary recurrent fevers

Results

We analysed 94 patients (sex-ratio:1). 42% were Jews and 17% were Arabs. Familial history of FMF was found in 23%, MICI in 10%, amyloidosis in 3% and Behçet in 3%. Median age of onset was 2 y. Fever was >39°C in 80%, duration and frequency of an attack varied (<24 h: 8%, 1–3 d: 56%, >3 d: 36%; >2/m: 15%, 1–2 m: 48% <1 m: 37% respectively). Peritonitis occurred in 97%, pleuritis in 25%, arthralgia in 53%; arthritis in 4 cases; skin rashes in 20%, aphtosis in 18% and lymphadenopathy in 9%.

MEFV mutation were: M694V (60%), M694I (7%).The R92Q TRAPS mutation was retrieved in 3/21 patients tested and the V377I MKD mutation in 1/6. Colchicine treatment was required in 82% of them and was effective in >90% of them. Associated diseases in these patients were PFAPA (4), Ankylosing spondylitis (5), Crohn's disease (1) and Castleman disease (1).

Discussion

The clinical picture of MEFV heterozygotes resembles that of homozygote patients. This study displays a wide variety of associated diseases. Complete screening of both MEFV and other auto-inflammatory gene mutation may increase our understanding of disease expression.