Pediatric Rheumatology

Poster presentation

International PFAPA syndrome registry: cohort of 214 patients

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Background

PFAPA syndrome is a periodic fever syndrome with diagnostic criteria including unspecific symptoms and exclusion of other fever syndromes.

Aim and methods

To investigate the clinical spectre, clinical course and long-term follow-up of PFAPA, we established a web-based multicentric registry as an international collaboration within the working party "periodic fevers" of PReS. Patients with PFAPA were included according to previously published criteria.

Results

We included 214 patients from 14 centres: 122 males, 92 females, median age at onset 1.9 year. The main clinical manifestations were present in a majority of the patients: pharyngitis (94%), cervical adenitis (83%), aphtous stomatitis (59%); 48% of the patients presented all 3 clinical features. 170 patients presented additional symptoms (gastrointestinal symptoms 131, arthralgias and/or myalgias 86, arthritis 4, skin rash 36, neurological symptoms 8). In 79 patients a genetic testing was done for periodic fever syndromes (FMF 49, TRAPS 52, HIDS 46, CAPS 7) and was negative, except for 8 cases (polymorphisms: 3, carrier for MEFV mutation: 5) without known clinical significance. Improvement or remission was observed in 99/105 patients with steroids, in 28/35 patients with tonsillectomy and in 5/15 patients with cimetidine.

Discussion

We describe the largest cohort of PFAPA patients presented so far. We confirm that PFAPA syndrome may present with varied clinical manifestations and that the diagnostic criteria lack of precision. Based on detailed analysis of this cohort, a new definition of PFAPA with better-defined criteria should be discussed in an international consensus conference.