

Poster presentation

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Evidences for the need of new Diagnostic Criteria for PFAPA syndrome

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Objective

The clinical manifestations of PFAPA syndrome largely overlap with those of monogenic Autoinflammatory diseases: Familial Mediterranean Fever (FMF), tumor necrosis factor (TNF) receptor-associated periodic syndrome (TRAPS) and Mevalonate kinase deficiency (MKD). Aim of this study is to evaluate the specificity of the available diagnostic criteria for PFAPA.

Patients and methods

307 consecutive patients with a clinical history of periodic fever were screened for mutations of *MVK*, *TNFRSF1A* and *MEFV* genes and detailed clinical information was collected. PFAPA diagnostic criteria were applied in all these patients. The clinical parameters associated with an high risk to be affected by an Autoinflammatory disease were identified on the basis of a univariate and multivariate analysis in both genetically positive and negative patients complying PFAPA criteria.

Results

133 out of 307 patients satisfying PFAPA criteria. 33 carried relevant mutations on the screened genes (27 MKD, 3 TRAPS, 3 FMF), 28 were heterozygous for *MEFV* mutations, 7 carried R92Q mutation of *TNFRSF1A* gene, show-

ing the low specificity of current criteria. Rash (OR = 2.975, $p = 0.009$), abdominal pain (OR = 3.261, $p = 0.005$) and vomiting (OR = 2.445, $p = 0.3$) were the variables most correlated to the positivity at the genetic test.

Conclusion

Current PFAPA criteria display a low specificity. According to this study, the presence of gastrointestinal manifestations and skin rash in patients fulfilling the current PFAPA criteria should orientate towards the exclusion of monogenic periodic fevers by molecular analysis. Consistent modifications of ongoing clinical criteria are proposed.