Pediatric Rheumatology

BioMed Central

Poster presentation

Open Access

Evidences for the need of new Diagnostic Criteria for PFAPA syndrome

R Caorsi*[1], A Meini[2], MP Sormani[3], M Cattalini[2], MA Pelagatti[1], F Zulian[4], E Cortis[5], G Calcagno[6], A Tommasini[7], F Traverso[1], S Federici[1], J Frenkel[8], S Plebani[2], A Martini[1] and M Gattorno[1]

Address: [1]UO Pediatria II, G. Gaslini Institute and Department of Pediatrics, University of Genoa, Genova, Italy, [2]Dipartimento di Pediatria, Unità di Immunologia e Reumatologia Pediatrica, Spedali Civili e University of Brescia, Brescia, Italy, [3]Unità di Biostatistica, DISSAL, University of Genoa, Genova, Italy, [4]Dipartimento A.I. di Pediatria, University of Padua, Padova, Italy, [5]Ospedale Pediatrico Bambino Gesu', Roma, Roma, Italy, [6]Unità di Pediatria, Policlinico "G. Martino", Messina, Italy, [7]IRCCS Burlo Garofalo, Dipartimento di Pediatria, University of Trieste, Trieste, Italy and [8]Department of General Pediatrics, Wilhelmina Children's Hospital, University Medical Center Utrecht, Utrecht, Netherlands

* Corresponding author

from 15th Paediatric Rheumatology European Society (PreS) Congress
London, UK. 14–17 September 2008

Published: 15 September 2008

Pediatric Rheumatology 2008, **6**(Suppl 1):P181 doi:10.1186/1546-0096-6-S1-P181

This abstract is available from: http://www.ped-rheum.com/content/6/S1/P181

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, showing 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 PFAFA criteria should orientate towards the exclusion of monogenic periodic fevers by molecular analysis. Consistent modifications of ongoing clinical criteria are proposed.