mutations: 6 patients homozygotes (2: V250I, 4: V377I) and 5 patients double heterozygotes: 4(I268T, V377I), 1(N205D, R388X). The genetic study of the whole family was performed in 7 families. Corticoid treatment used for 100% of the patients. Anakinra in 45% (parcial-complete response), Canakinumab 82% (complete response), Etanercept 9% (no response) and adalimumab 9% (hydradenitis). Long-term manifestations: 2 patients with cutaneous abscess and 1 patient with hydradenitis suppurativa.
Conclusion: In this cohort the high response to canakinumab treatment is shown and late clinical manifestations are described, to our knowledge, for the first time in literature.

Disclosure of Interest
None Declared

P1013

Novel assay to diagnose and monitor cryopyrin associated periodic syndromes (CAPS)

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Introduction: Cryopyrin associated periodic syndromes (CAPS) are rare autoinflammatory disorders associated with dominantly gain-of-function mutations in the NLRP3 gene that result in overactivation of the inflammasome, increased secretion of interleukin (IL)-1beta and IL-18, and systemic inflammation. It has been reported that oligomeric particles of the adaptor ASC (apoptosis-associated Speck-like protein with a caspase-recruitment domain) are released together with IL-1beta and active caspase-1 subunits after activation of the inflammosome complex and that patients with CAPS show an increased serum concentration of ASC^+ particles.
Objectives: The diagnosis of CAPS is a critical factor due to both the lack of specific laboratory results and the sharing of similar clinical manifestations with other autoinflammatory diseases, our aim is to develop a simple assay to evaluate the levels of ASC particles in the serum of CAPS patients to provide novel biomarkers facilitating early disease diagnosis and able to monitor treatment responses.
Methods: We developed an ELISA for the quantification of ASC particles in serum and plasma of normal and pathological subjects. We analysed samples from CAPS patients and from patients with autoimmune disorders (Multiple Sclerosis (MS), Type 1 Diabetes (T1D) and juvenile idiopathic arthritis), to confirm that ASC presence in the serum is not due to other chronic inflammatory processes characterizing autoimmunity. In addition, we also evaluated the concentration of ASC in the sera of TNF receptor–associated periodic syndrome (TRAPS) patients to reinforce the concept of specificity of this biomarker in CAPS patients and not in individuals suffering from others inflammatory disorders.
Results: We observed that untreated CAPS patients are characterized by the presence of a significant higher amount of ASC particles when compared with healthy controls (HS) and with patients suffering from MS and T1D. This tendency was also evident in patients with arthritis and TRAPS even if the difference was not statistically significant due to the small number of samples. In addition there is a tendency through a reduction of ASC levels in CAPS patients after pharmacological treatment, which require future investigations.
Conclusion: These data suggest that ELISA quantitation of ASC protein could represent a novel and additional strategy for the diagnosis and monitoring of CAPS.

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An Italian family with FCAS

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Introduction: The prevalence of Cryopyrin-Associated Periodic Syndrome (CAPS) is estimated at about one per million. The Familial Cold Autoinflammatory Syndrome (FCAS) presents in about 95% of the patients by 6 months with cold-induced, urticaria-like rash, fever, and arthralgia. FCAS causes lifelong debilitating effects that restrict patients' daily activities. Diagnostic delay related to lack of knowledge in Autoinflammatory Diseases is still an important problem.
Objectives: To report an Italian family with FCAS successfully treated with anti-IL1.
Methods: Three subjects, members of the same family, were screened by an experienced doctor. Clinical and laboratory variables, Brain-CT scan, X-ray, audiometry and lung function tests were performed. A targeted review of clinical feature for Autoinflammatory diseases and standardized questioning for CAPS-associated symptoms was conduct. Genetic testing for the NLRP3 mutation was performed.