Background: Familial cold autoinflammatory syndrome 3 (FCAS3) is an autoinflammatory disease (AID) caused by mutation of the PLCG2 gene, which has not been reported in China. We will report 2 cases of Chinese FCAS3 patients with no claimed family history, but we found the same mutations in a patient during their genetic analysis. After further inquiry of the parent’s medical history, we confirmed that actually they were two FCAS3 families. Through a literature review, we found that the clinical features of Chinese patients are milder than foreign countries, and their symptoms are concealed and may be ignored, resulting in mistakes in family history collecting.

Methods: Two suspected AID children with recurrent fever and urticaria were enrolled in this study. Clinical data and family history were collected, and genetic analysis was performed by next-generation sequencing (PJD panel or WES) and Sanger-based validation. Literature was reviewed from PubMed, CNKI, and Wanfang Database.

Results: The two children were both diagnosed to be FCAS3 with PLCG2 mutation. The clinical manifestations of 2 children were recurrent fever, urticaria, and increased ESR and CRP. Case 1 has a paternal, and Case 2 has a maternal heterozygous mutation in the PLCG2 gene, while both had claimed without a family history. Further inquiry showed the two parents used to have a fever with urticaria. By comparing with foreign literature, we found our patients were milder than abroad patients. Large fragment deletions were relatively more common in foreign patients.

Conclusion: We reported the case of FCAS3 in China for the first time. Their genotype and phenotype were different from foreign patients. Their symptoms are mild, and heterozygous mutations are more common than foreign patients, which are the main differences. The difference in mutation type may be the reason for different clinical manifestations. Besides, both two families showed a trend of more severe clinical features in the next generation. As the symptoms of the children were not obvious and may be ignored, it causes troubles for the genetic diagnosis. Therefore, family history should be collected carefully. For rashes and fevers, which are not so severe in overall symptoms, care should be taken about the possibility of AIDs. Genetic testing can help to make a definite diagnosis.

References:

Disclosure of Interests: None declared

DOI: 10.1136/annrheumdis-2020-eular.862

AB1086

REGULATORY EFFECT OF SHORT-TERM LOW DOSE IL-2 RESTORES REGULATORY T CELLS IN IG4G-RELATED DISEASE

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Background: Little known about the roles of peripheral immune cell subsets in IgG4-related disease (IgG4-RD).

Objectives: The aim of our study was to analyze the role of low-dose interleukin-2 (IL-2) on these cells in IgG4-RD.

Methods: The percentage and absolute counts of lymphocyte subpopulations [CD3+ (T cells), CD4+, CD8+, CD19+ (B cells) and CD16+CD56+ (NK cells) and CD4+T cell subsets (Th1, Th2, Th17, regulatory T (Treg)) using single platform flow cytometry in 23 IgG4-RD patients who were admitted and treated, as well as 24 healthy controls (HCs). Among IgG4-RD patients, 19 patients given only conventional treatments while 5 patients were not only given conventional treatments but also received IL-2 (0.5 million IU/day) for 5 days.

Results: We found that the absolute counts of T, CD4+T and Th17 cells were increased in the peripheral immune cells of IgG4-RD patients when compared with HCs. Meanwhile, the percentage of B, Th2, Th17 and Treg cells demonstrated significantly decreased. The ratio of Th1/Th2 and Th1/Treg in IgG4-RD patients were higher than that in HCs. After IL-2 administration, the absolute numbers of Treg cells increased dramatically; furthermore, the proportion of Treg cells had a trend towards higher values compared with those before treatment. Conversely, the ratio of Th2/Treg was downward. There were no any significant differences in the above subsets between before and after conventional treatments.

Conclusion: Our findings support that the reduction of Treg cells in IgG4-RD patients, as well as IL-2 combined with conventional treatments were able to restore the Treg cells.

References:

Disclosure of Interests: None declared

DOI: 10.1136/annrheumdis-2020-eular.3649

AB1089

CONSIDERATION OF YAO SYNDROME AS A DIFFERENTIAL DIAGNOSIS FOR HEREDITARY PERIODIC FEVER SYNDROMES

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Background: Yao syndrome (YAOs, OMIM 617321), formerly termed nucleotide-binding oligomerization domain 2 (NOD2)-associated autoinflammatory disease, is characterized by periodic fever, dermatitis, arthralgia, and swelling of the distal extremities, as well as gastrointestinal and sicca-like symptoms. This disorder shares similar clinical phenotypes with hereditary periodic fever syndromes (HPFS) and thus can mimic one another.

Objectives: This study aimed to embody by a comparison of YAOs vs familial Mediterranean fever (FMF).

Methods: In this retrospective study, electronic medical records of a series of patients with YAOs were analyzed. All patients underwent genetic testing for periodic fever syndrome 6-gene panel (MEVF, TNFRSF1A, NLRP3, MVK, NLRP12 and NOD2).

Results: All patients were Caucasian and had recurrent fever, patchy erythema, arthralgia, and gastrointestinal symptoms (Table 1). With negative DNA sequencing for MEVF, these patients were treated with colchicine for presumed FMF, with a good response in patient 2 and minimal or transient response in other two patients. Further genetic testing identified the NOD2 variants. Unlike HPFS, YAOs is generally sporadic and is mostly reported in adults; spongiotic dermatitis is common; YAOs is associated with the NOD2 variants, IVS8 + 15B in nearly all patients, IVS8 + 15B/R02IV in up to 30%, and IVS8 + 15B/1007ts, G908R or other rarer NOD2 variants in some patients.

Conclusion: YAOs could masquerade HPFS like FMF. Molecular analysis should cover NOD2 whole gene sequencing to help distinguish these diseases.

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China


