

POSTER PRESENTATION

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Diagnosis of early-onset sarcoidosis with non-classical symptoms

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Results

A 15-month-old male patient was referred to pediatric rheumatology for evaluation of non-pruritic skin eruption most prominent on arm and leg, fever and lymphadenopathy. These symptoms began when the patient was four months old. Several antibiotics therapies had been examined. The maximal convalescence period was a month. There were no consanguinity and family history. Physical examination showed generalized, maculopapular eruption especially on arm and leg, lymph node on left cervical and axillary region, and persistent fever. In laboratory examinations, acute phase reactants were found to be high. The serology of EBV, CMV, toxoplasma, brucella, tularemia, bartonella henselae, and Quantiferon test results were negative. Immunologic evaluations contained hemogram, blood smear, immunoglobulins, fagotest, and CD panel were normal. Urine calcium/creatinin ratio, serum ACE levels were normal. A skin biopsy taken from a papule showed subepidermal non-langerhans histiocytes and a non-caseating granuloma. A lymph node biopsy showed granulomatous inflammation. Mediastinal lymph node was absent on thorax CT evaluation. His ophthalmological examination was normal. The granulomatous autoinflammatory disease was diagnosed. Prednisolone 2 mg/kg was administered which reduced symptoms and patient's complaints. Then, the steroid dose was dropped. The fever and rash were absent, but the acute phase reactants were still high. His blood samples were referred for genetic testing NOD2 mutations. Methotrexate was added to the treatment, but the flare occurred two months later. NOD2 mutations were detected to be P268S heterozygote and V955I heterozygote. Since the patient's response to anakinra was poor, TNF blocker was later added to the treatment.

Conclusion

Granulomatous autoinflammatory diseases include Blau syndrome and early-onset sarcoidosis, both are caused by mutations in the NOD2/CARD15 gene, inherited autosomal dominant and sporadic form of disease, respectively. Clinical triads are uveitis, arthritis and granulomatous dermatitis whereas the symptoms we observed were fever, granulomatous dermatitis, and lymphadenopathy.

Disclosure of interest

None declared.

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