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STAT3 mutation presenting with atopic dermatitis

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Introduction: Hyper immunoglobulin E syndrome (HIES) is a rare systemic disease characterized by recurrent skin and lung infections, eczema and serum total IgE elevation. It can easily be confused with atopic dermatitis and other causes of rare immune deficiencies in clinical practice. We presented a case which was followed with atopic dermatitis diagnosis for a long time and received the diagnosis of HIES associated with STAT3 mutation during the follow-up.

Case: A 10-year old girl was referred to our clinic for atopic dermatitis resistant to medical treatment and extremely high total IgE levels (18223 IU/L). In her past history, she had omphalitis during neonatal period; eczematous lesions developed in hairy and hairless skin, white plaques developed in the mucosa of mouth and tongue continuously since the infancy period, recurrent purulent otitis. She did not have a family history of immunodeficiency. She was begun to be followed up with the diagnosis of atopic dermatitis when she was a 16-month-old baby and atopic dermatitis symptoms did not respond to treatment sufficiently. Asthma and allergic rhinitis symptoms developed during the follow-up period. On admission, physical examination revealed prevalent eczematous eruptions, oropharyngeal candidiasis, changes in the nails suggesting mycotic infection. In the laboratory analysis, IgA, M and IgG levels, C3 and C4 levels of the patient were determined to be normal and tetanus, diphtheria and hepatitis B vaccine responses of the case emerged positive. In lymphocyte subgroups, total lymphocyte and CD8+ cells had 5-10 percentile value numerically in terms of age; CD19+ and NK cells were detected below the value of 5 percentile. *Candida albicans* multiplied in a culture medium received from oral mucosa. With possible diagnosis of HIES, the patient was started to be administered IV immunoglobulin replacement, trimethoprim-sulfamethoxazole and flukonazole prophylaxis. Genetic analysis performed at CeMM-Research Centre for Molecular Medicine of the Austrian Academy of Sciences revealed STAT3 mutation.

Conclusion: In such cases with atopic dermatitis symptoms and elevated total IgE levels, who do not adequately respond to the treatment, HIES associated with STAT3 mutation should be remembered during differential diagnosis.

Biography

Ferah Genel is currently an Associate Professor in Dr. Behçet Uz Children's Hospital, Department of Pediatrics, Division of Pediatric Immunology. He/She is interested in Congenital Immune Deficiencies, Flow cytometry and he/she has 52 papers published in reputed journals to his/her credit.

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