Investigation of skin barrier functions and allergic sensitization in patients with Hyper-IgE syndrome

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Hyper-IgE syndrome (HIES) is a rare, but severe primary immunodeficiency, characterized by increased serum IgE levels, recurrent infections and atopic dermatitis (AD)-like skin lesions. STAT3 mutations are detected in the most patients, which cause impaired Th17 development. AD is a chronic inflammatory skin disease with immunologic alterations and skin barrier dysfunctions. Our aim was to investigate and compare the skin barrier alterations and allergic sensitization (AS) in HIES and AD patients and to find similar or different pathogenetic events in the development of skin lesions. Analyses of STAT3 and filaggrin (FLG) mutations were performed in 6 HIES and 30 AD patients as controls. Laboratory parameters (LDH level and eosinophil count), immunologic alterations (intracellular cytokine staining), AS (total and specific IgE levels, medical history), and skin barrier changes [transepidermal water loss (TEWL), serum thymic stromal lymphopoietin (TSLP) levels] were also examined. Mutation analysis of STAT3 showed 100% positivity in HIES patients, although all of them had FLG wild-type concerning R501X and 2282del4 mutations, which were found in 31% of our AD patients in heterozygous form. No differences were found between the two diseases regarding LDH and IgE levels or eosinophil counts. Impaired Th17 cell numbers were detected in T cells of HIES patients. No altered barrier functions were found in HIES patients which were significantly impaired in AD patients. AS was more frequent in AD. On the basis of these results barrier alterations probably are not the main pathogenetic events in the development of skin lesions in HIES. Despite of the high IgE levels, AS is not a characteristic feature in these patients, which can be the consequence of their normal skin barrier functions, since outside-inside barrier impairment seems to be necessary for the development of AS.