Hyperimmunoglobulin E syndrome in three siblings of non-consanguineous healthy Egyptian family. Case report

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**Background:** Hyperimmunoglobulin E syndrome (HIES) is a rare primary immunodeficiency disorder characterize by elevated IgE level, eczema, recurrent staphylococcal skin abscesses, and pulmonary infections. Autosomal dominant and autosomal recessive forms of this disorder had been described. Most autosomal dominant HIES (AD-HIES) have been found to be due to mutations in STAT3 (Signal transducer and activator of transcription 3); whereas DOCK8 (dedicator of cytokinesis 8) mutations have been described in patients with autosomal recessive HIES (AR-HIES).

**Case report:** We describe here non-consanguineous health Egyptian family with three affected siblings with hyper IgE syndrome (two females, 9 and 5.5 years, and one male 9-months), but the second girl died after diagnosis because of trauma at age of 1.8 years. The clinical presentations of all siblings are neonatal pustular rash, eczema, recurrent skin abscesses that need surgical drainage, lymphadenitis, recurrent oral and napkins severe fungal infections, recurrent chest infections with formation of pneumatoceles and complications with pneumothorax that required chest tube drainage and retention of primary teeth in the oldest girl. Regarding the laboratory tests high eosinophilia > 15 %, IgE level > 2500 IU/mL, normal other Igs, CD4 lymphopenia and coarse facial features. Their HIES scores were 65, 52 and 51, respectively.

**Conclusions:** Our three patients had the typical features of HIES (neonatal pustular rash, eczema, recurrent skin abscesses, pulmonary manifestations, dental characteristics, skeletal and coarse facial features) although their parents are completely health with normal level of IgE.

**Keywords:** Hyperimmunoglobulin E syndrome; Primary immunodeficiency