

Facts on Hyper Immunoglobulin E Syndrome (HIES)

- **HIES is a primary immunodeficiency disorder characterized by the diagnostic triad of elevated serum IgE levels, recurrent skin infections, and recurrent pneumonias, often accompanied by skeletal and connective tissue abnormalities.**

[Paulson ML, et al. \(2008\) Curr Opin Allergy Clin Immunol 8:527-533](#)

- **Diagnosis of HIES is complicated by the non-specificity of some symptoms, such as elevated serum IgE levels and skin rashes, and the atypical presentation of others, such as HIES-related pneumonia often presenting without fever and cough.**

[DeWitt CA, et al. \(2006\) J Am Acad Dermatol 54: 855-865](#)

- **HIES is caused by mutations in the *STAT3* gene, which codes for a protein important for normal immune system function.**
 - Mutations in *STAT3* account for up to 22% of familial HIES.

[Holland SM, et al. \(2007\) NEJM 357:1608-1619](#)

[Ma CS, et al. \(2008\) J Exp Med 205:1551-1557](#)

- **Age of onset of HIES is usually in the newborn period, but diagnosis may be delayed into the adolescent period.**

[Freeman AF, et al. \(2008\) Immunol Allergy Clin North Am 28:277-291](#)

- **Early diagnosis and treatment of HIES patients with aggressive antibiotics is crucial, since severe infections are a major cause of mortality.**

[Freeman AF, et al. \(2007\) J Allergy Clin Immunol 119: 1234-1240](#)

- **Genetic testing can confirm a diagnosis of familial HIES in the index patient for a family and identify family members with a predisposition for HIES.**

[Holland SM, et al. \(2007\) NEJM 357:1608-1619](#)