

Using Genetic Testing To Diagnose Hyper Immunoglobulin E Syndrome (HIES)

Indications:

- Clinical diagnosis of HIES
- Elevated serum IgE levels and at least one other characteristic manifestation, such as a *Staphylococcus aureus*-positive lung infection or skin infection
- Family history of HIES

Benefits:

Genetic testing for HIES can:

- confirm a clinical diagnosis of HIES.
- help to recognize HIES-related pneumonias and facilitate prophylactic treatment with antibiotics.
- allow detection of at-risk individuals in family members of HIES patients.

Background:

- Hyper Immunoglobulin E (IgE) syndrome (HIES) is a rare primary immunodeficiency syndrome characterized clinically by the triad of elevated serum IgE levels, recurrent skin infections, and recurrent pneumonia, as well as by eczema and connective tissue and skeletal abnormalities.¹
- HIES is likely to be under-diagnosed, since 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 or cough.²
- Up to 22% cases of familial HIES show an autosomal dominant mode of inheritance due to mutations in the *STAT3* gene,³ which codes for a protein important for normal immune system function.
- Early diagnosis and treatment of HIES patients with aggressive antibiotics is crucial, to prevent bacterial superinfections accompanying recurrent pneumonias that are a major cause of mortality.⁴

References: 1. Paulson ML, et al. (2008) *Curr Opin Allergy Clin Immunol* 8:527-533. 2. DeWitt CA, et al. (2006) *J Am Acad Dermatol* 54: 855-865. 3. Holland SM, et al. (2007) *NEJM* 357:1608-1619. 4. Freeman AF, et al. (2007) *J Allergy Clin Immunol* 119: 1234-1240.

Ordering Information: Please see other side.



Ordering Information for Hyper Immunoglobulin E Syndrome (HIES) Testing

Indications for Testing

- Clinical diagnosis of Hyper Immunoglobulin E Syndrome (HIES)
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- Family history of HIES

Ordering Information for Single Gene Tests

Gene(s)	CPT Codes	Test Code
STAT3	83891(1) 83892(1) 83898(21) 83904(42) 83909(42) 83912(1)	120201

Family Testing (single amplicon)

STAT3	83891(1) 83892(1) 83898(1) 83904(2) 83909(2) 83912(1)	120201
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Test Methodology and Sample Requirements

- Amplification by polymerase chain reaction (PCR); sequencing of entire protein-coding region
- For blood samples:
 - 2 mL whole blood in EDTA tube (lavender top)
 - Samples can be stored briefly at 4°C, but should be shipped on day of collection.
- For buccal swab samples: (only accepted for family testing)
 - Please contact client services at 1-866-647-0735 for instructions.
- All sample types should be shipped overnight at room temperature.
- To request a sample shipping kit, please call 1-866-647-0735.

For more information, please contact Correlagen Diagnostics, Inc., at 1-866-647-0735 or visit us on the web at www.correlagen.com.