

## 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.<sup>1</sup>
- 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.<sup>2</sup>
- Up to 22% cases of familial HIES show an autosomal dominant mode of inheritance due to mutations in the *STAT3* gene,<sup>3</sup> 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.<sup>4</sup>

**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)
- 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

### Ordering Information for Single Gene Tests

Gene(s)	Test Code
<b>STAT3</b>	120201

### Family Testing (single amplicon)

Family Testing is available. Please contact Client Services at 1-866-647-0735 for requirements.

### Test Methodology

- Amplification by polymerase chain reaction (PCR); sequencing of entire protein-coding region

NOTE: Specimens must be accompanied by a completed consent form. In the case of family tests (ie, known mutations), a copy of the result of the first patient tested in the family (the index case) must be submitted unless that test was performed at Correlagen. Other family members are subsequently tested for the specific mutation found in the first patient tested.

**For test information, sample requirements, or to request a sample shipping kit, please contact Client Services at 1-866-647-0735 or visit us on the web at [www.correlagen.com](http://www.correlagen.com).**