

## Using Genetic Testing to Diagnose Hypohidrotic Ectodermal Dysplasia with Immunodeficiency (HED-ID)

### Indications:

- Recurrent bacterial, viral, and/or mycobacterial infections, usually accompanied by ectodermal dysplasia (ED), in male infants and young children
- Impaired generation of specific antibodies in response to polysaccharide antigens
- Reduced NK-cell cytotoxicity
- Male-limited family history of ED and recurrent, severe, or atypical infections, or a family history of *IKBKG*-related HED-ID

### Benefits:

- Early diagnosis of HED-ID is important since timely initiation of treatment can prevent complications and improve prognosis.<sup>1</sup>
- Genetic testing can confirm or establish a diagnosis of HED-ID in infants before the symptoms of ED become obvious.<sup>1</sup>
- Genetic testing facilitates carrier detection and genetic counseling.

### Background:

- HED-ID is a rare primary immunodeficiency, with an estimated incidence of 1 in 250,000 live male births.<sup>1</sup>
- HED-ID is associated with X-linked recessive mutations in *IKBKG* that impair, but do not completely abolish activity of NEMO, the *IKBKG* gene product.<sup>2,3,4</sup>
- Classic HED-ID is a heterogeneous disorder, characterized by increased susceptibility to severe, recurrent, and atypical infections and abnormal development of ectoderm-derived skin structures, including teeth, hair, nails, and eccrine sweat glands.<sup>5,6</sup>
- HED-ID is typically treated with intravenous immunoglobulin replacement therapy (IVIG) and prophylactic antibiotics. In one case, a patient was successfully treated with hematopoietic stem cell transplantation.<sup>1</sup>

**References:** 1. Orange JS et al (2004) *J Allergy Clin Immunol* 113:725-733. 2. Zonana J et al (2000) *Am J Hum Genet* 67:1555-1563. 3. Doffinger R et al (2001) *Nat Genet* 27:277-285. 4. Orange JS et al (2005) *Immunol Rev* 203:21-37. 5. Uzel, G (2005) *Curr Opin Allergy Clin Immunol* 5:513-518

**Ordering Information:** please see other side

## Ordering Information for Hypohidrotic Ectodermal Dysplasia with Immunodeficiency (HED-ID) Testing

### Indications for Testing

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### Ordering Information

Gene	CPT Codes	Test Code
<i>IKBKG</i> ( <i>NEMO</i> )	83891(1) 83892(1) 83898(9) 83904(18) 83909(18) 83912(1)	100601

### Family Testing (single amplicon)

<i>IKBKG</i> ( <i>NEMO</i> )	83891(1) 83892(1) 83898(1) 83904(2) 83909(2) 83912(1)	100601
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### Test Methodology

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

### Sample Requirements

- 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:
  - 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.

### Turn-around Times

Turn-around times typically range from 7 to 21 days of receipt of sample and all required forms, but may vary depending on test volume and test-specific technical difficulties. Current TATs are posted on our website. Please schedule patient follow-up appointments for discussion of test results conservatively at 6 weeks.

**For more information, please contact Correlagen Diagnostics, Inc., at 1-866-647-0735 or visit us on the web at [www.correlagen.com](http://www.correlagen.com).**