

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.¹
- Genetic testing can confirm or establish a diagnosis of HED-ID in infants before the symptoms of ED become obvious.¹
- 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.¹
- 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.^{2,3,4}
- 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.^{5,6}
- 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.¹

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 Hypohydrotic Ectodermal Dysplasia with Immunodeficiency (HED-ID) Testing

Indications for Testing

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Ordering Information

Gene	Test Code
<i>IKBKG</i> (NEMO)	100601

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.