

Using Genetic Testing to Identify Mutations in *AIRE* as the Cause of CMC and/or Multiple Autoimmune Manifestations

Indications:

- Chronic mucocutaneous candidiasis (CMC)
- Two or more autoimmune manifestations characteristic for APS1/APECED
- Family history of CMC or APS1/APECED

Benefits:

- Genetic testing for mutations in the gene *AIRE* can:
 - identify APS1/APECED as the cause of autoimmunities and/or CMC.
 - alert patients and physicians to the risk of other autoimmune or immune-mediated manifestations associated with APS1/APECED.
 - aid in genetic counseling for APS1/APECED.

Background:

- Autoimmune Polyglandular Syndrome type 1 (APS1), also known as Autoimmune Polyendocrinopathy-Candidiasis-Ectodermal Dystrophy (APECED), is characterized by CMC, autoimmune hypoparathyroidism, and autoimmune adrenal insufficiency.¹
- CMC affects almost all patients with APS1/APECED and is typically the presenting symptom.¹
- Additional autoimmunities and immune-mediated diseases may develop throughout a patient's lifetime.²
- Early diagnosis of APS1/APECED can facilitate prompt intervention as new autoimmunities develop and help to prevent severe complications.^{3,4}
- APS1/APECED is inherited in an autosomal recessive manner.^{5,6}
- APS1/APECED is due to loss-of-function mutations in the gene *AIRE*.^{5,6}

References: 1. Betterle C, et al (2002) *Endocrine Reviews* 23:327-64. 2. Ahonen P, et al (1990) *N Engl J Med* 322:1829-36. 3. Myhre AG, et al (2001) *Clin Endocrinol* 54:211-7. 4. Eisenbarth GS, Gottlieb PA (2004) *N Engl J Med* 350:2068-79. 5. Nagamine K, et al (1997) *Nat Genet* 17:393-8. 6. The Finnish-German APECED Consortium (1997) *Nat Genet* 17:399-403.

Ordering Information: please see other side



Ordering Information for the *AIRE* DNA Sequencing Test

Indications for Testing

- Chronic mucocutaneous candidiasis
- Two or more autoimmune manifestations characteristic for APS1/APECED
- Family history of chronic mucocutaneous candidiasis or APS1/APECED

Ordering Information

Gene(s)	Test Code
<i>AIRE</i>	110101

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.