

*Identify patients with APS1  
before severe complications develop.*

# Autoimmune Polyglandular Syndrome (AIRE) Evaluation

## *What is autoimmune polyglandular syndrome type 1 (APS1)?*

- APS1 is characterized by the triad of chronic mucocutaneous candidiasis, autoimmune hypoparathyroidism, and autoimmune primary adrenal insufficiency.<sup>1,2</sup>
- APS1 is also known as autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy (APECED), since ectodermal dystrophies such as dental enamel hypoplasia and keratopathy are common.
- Clinical diagnosis of APS1 is based on the presence of at least two of the three primary component diseases and is often delayed until severe complications associated with an endocrinopathy have developed.<sup>3</sup>
- APS1 has been associated with autosomal recessive loss-of-function mutations in the gene AIRE.<sup>4,5</sup>

## *Why genetic testing?*

### **Genetic testing for mutations in AIRE associated with autoimmune polyglandular syndrome type 1**

- Permits fast and accurate diagnosis of APS1 after only one of the primary component diseases of APS1 has developed.
- Can identify APS1-associated mutations in relatives of APS1 patients before any symptoms develop.
- Diagnosis of APS1 permits increased vigilance for other APS1-associated phenotypes, allowing early intervention and helping to prevent potentially fatal complications from untreated endocrine or metabolic insufficiencies.

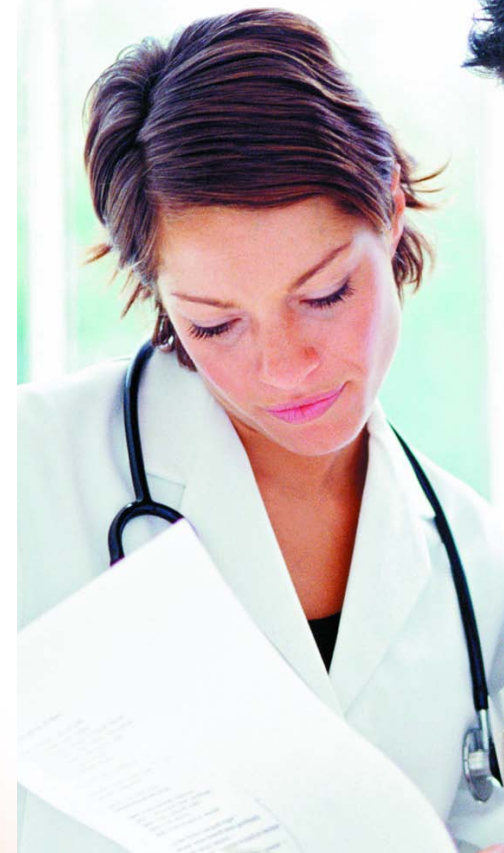
## *Indications for testing:*

- Presence of at least one of the three characteristic component diseases of autoimmune polyglandular syndrome type 1 (APS1) in children age ten or younger
- Suspicion of APS1 in individuals of any age
- Family history of APS1

*For complete ordering information,  
please see the reverse side.*

**References:** 1. Pitkanen J & Peterson P (2003) *Genes and Immunity* 4:12-21. 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. Nagamine K et al (1997) *Nat Genet* 17:393-8. 5. The Finnish-German APECED Consortium (1997) *Nat Genet* 17:399-403.

**For a brief review on APS, please visit [www.athenadiagnostics.com/DR](http://www.athenadiagnostics.com/DR)**



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# Autoimmune Polyglandular Syndrome (AIRE) Evaluation

- Typical Presentation:** Chronic mucocutaneous candidiasis, hypoparathyroidism, primary adrenal insufficiency
- Synonyms:** Autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy (APECED)
- Indications for Testing:**
- Presence of at least one of the three characteristic component diseases of autoimmune polyglandular syndrome type 1 (APS1) in children age ten or younger
  - Suspicion of APS1 in individuals of any age
  - Family history of APS1

## TEST DETAILS

- Test Code:** 812
- Test Turnaround:** 14-21 days

## TECHNICAL INFORMATION

- Methodology:** Polymerase Chain Reaction (PCR), DNA sequencing of entire protein coding region of gene

## SHIPPING CONSIDERATIONS

- Specimen Type:** Whole blood
- Volume:** 10 mL (pediatric minimum: 2 mL)
- Collection Tube:** Yellow or lavender top
- Stability:** Hemolysis may compromise DNA recovery and integrity after 48 hrs
- Storage Conditions:** For short periods (until shipped) at 4°C
- Shipping Conditions:** Overnight at room temperature (specimen arrival must be less than 24 hrs after collection); ship Monday through Thursday only

www.athenadiagnostics.com

Call Athena Diagnostics' Customer Service Representatives to order the Autoimmune Polyglandular Syndrome (AIRE) Evaluation (Test #812) at:

**800-394-4493 x2**



*Testing You Can Count On.*

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