



Genetics Uncoded:



Facts about

Autoimmune Polyglandular Syndrome Type 1



What Your Test Results Mean

Carriers typically show no symptoms of autoimmune polyglandular syndrome type 1. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

● Autoimmune Polyglandular Syndrome Type 1 Explained

Autoimmune polyglandular syndrome type 1 is an inherited disorder that affects many of the body's organs. The *AIRE* gene is involved in regulating certain aspects of immune system function. Variations in the *AIRE* gene result in autoimmunity, which can cause inflammation and damage to healthy cells and tissues, specifically the adrenal glands and parathyroid glands. Symptoms usually appear in early childhood or adolescence and include hypoparathyroidism, mucocutaneous candidiasis, and Addison's disease. Affected individuals usually have at least two of these features, and many have all three. Mucocutaneous candidiasis is a chronic fungal infection that affects the skin and mucous membranes. Hypoparathyroidism and Addison's disease can cause muscle pain, fatigue, weight loss, and low blood pressure.

Treatment for autoimmune polyglandular syndrome type 1 includes hormone replacement therapy to replace the insufficient amounts being made by the adrenal glands. Treatment of mucocutaneous candidiasis can be challenging but success has been found with ketoconazole. Oral calcium and vitamin D are usually adequate therapy for hypoparathyroidism.

● How the Genetics Work

Autoimmune polyglandular syndrome type 1 is an autosomal recessive disorder caused by variants in the *AIRE* gene. In general, individuals have two copies of the *AIRE* gene. Carriers of autoimmune polyglandular syndrome type 1 have a variant in one copy of the *AIRE* gene while individuals with autoimmune polyglandular syndrome type 1 have variants in both copies of *AIRE*, one inherited from each parent. Risk for two carriers to have a child with the disorder is 25%.

Questions? Contact us at **1-866-661-7966** to set up an appointment to discuss your results in more detail with a Clarity Genetics genetic counselor.