

# Autoimmune Polyendocrinopathy with Candidiasis and Ectodermal Dystrophy

## What Your Results Mean

Test results indicate that you are a carrier of autoimmune polyendocrinopathy with candidiasis and ectodermal dystrophy. Carriers typically show no symptoms. You and your partner would both have to be carriers of autoimmune polyendocrinopathy with candidiasis and ectodermal dystrophy for there to be an increased chance to have a child with symptoms; this is known as autosomal recessive inheritance. Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment.

Since this is an inherited gene change, this information may be helpful to share with family members as it may impact their family planning.



## Recommended Next Steps

Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment. If both you and your partner are carriers for autoimmune polyendocrinopathy with candidiasis and ectodermal dystrophy, each of your children has a 1 in 4 (25%) chance to have the condition.

## Autoimmune Polyendocrinopathy with Candidiasis and Ectodermal Dystrophy Explained

### What is Autoimmune Polyendocrinopathy with Candidiasis and Ectodermal Dystrophy?

Autoimmune polyendocrinopathy with candidiasis and ectodermal dystrophy 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.



## Prognosis

Prognosis depends on the severity of symptoms. Early detection and treatment of the disease are important for achieving the optimum quality of life. Life expectancy may be decreased due to complications from symptoms.

## Treatment

Prognosis depends on the severity of symptoms. Early detection and treatment of the disease are important for achieving the optimum quality of life. Life expectancy may be decreased due to complications from symptoms.



### Resources

#### National Organization for Rare Disorders

<https://rarediseases.org/rare-diseases/autoimmune-polyglandular-syndrome-type-1/>

#### American Autoimmune Related Diseases Association, Inc.

<https://www.aarda.org/>

#### National Society of Genetic Counselors

<https://www.nsgc.org/>