Autoimmune Polyglandular Syndrome Type 1 (APS Type 1)

APS 1
The APS Type 1 Foundation Inc.

Autoimmune Polyglandular Syndrome Type 1 (APS Type 1), also known as Autoimmune Polyendocrinopathy-Candidiasis-Ectodermal Dystrophy (APECED), is a rare genetic disorder that is often misdiagnosed. Mutations in the *AIRE* gene lead to three classic manifestations along with other common symptoms. This resource aims to assist you with diagnosis and referring a patient to a genetics professional. For additional information, visit the APS Type 1 Foundation website or review this research article.

Classic Manifestations

1. **Chronic Mucocutaneous Candidiasis**
Patients may experience recurring and long-lasting fungal infections that affect the skin and mucous membranes.

2. **Hypoparathyroidism**
When the parathyroid glands malfunction, low serum calcium may lead to patients experiencing seizures, fatigue, muscle pain and cramping, and a tingling sensation in lips, fingers, and toes.

3. **Adrenal Insufficiency (Addison's Disease)**
Malfunction of the adrenal glands leads to loss of appetite, weight loss, low blood pressure, salt cravings, muscle weakness, and changes in skin coloring.

Adjunct Triad

1. **Urticarial Eruption (hives)**
Patients often experience a recurrent maculopapular rash that typically resolves on its own and is not usually itchy but can be.

2. **Enamel Hypoplasia**
Patients may have a deficiency in enamel production, making the teeth appear yellow or spotted, with or without cavities.

3. **Intestinal Malabsorption**
When nutrients are not properly absorbed by the small intestine, patients experience increased fecal fat and disruption of the gut microbiome.

Does your patient have...

symptoms for one classical manifestation

+

symptoms for a manifestation within the adjunct triad?

OR

symptoms for two of the classic manifestations?

→

It is recommended that your patient be referred to a genetics professional for genetic testing of the *AIRE* gene.