

APS1 DISEASE SPECIFIC FORM

Participant Ancestry

Family Ancestry

- German
- British
- French
- Italian
- Irish
- Scandinavian
- Other Ancestry _____

Diagnostic Criteria

Select All that Apply

- Disease-causing mutations in both copies of the AIRE genes
- Sibling is affected by APECED
- Presence of autoantibodies

Which Autoantibodies?

- Interferon alpha
- Interferon omega
- 21- hydroxylase
- GAD65
- NALP5
- TPH

Major Manifestations
(select all that apply)

- Chronic mucocutaneous candidiasis
- Hypoparathyroidism
- Adrenal insufficiency

Minor Manifestations
(select all that apply)

- Alopecia
- Autoimmune hepatitis
- Chronic diarrhea
- Enamel hypoplasia
- Keratitis
- Periodic rash with fever
- Severe chronic constipation
- Vitiligo

DEFINITE = 2+ major manifestations
1 major manifestation & Sibling is affected by APECED
Disease-causing mutations in both copies of AIRE genes

PROBABLE = 1 major manifestation & 1 minor manifestation
Any manifestation & presence of autoantibodies