### APS1 DISEASE SPECIFIC FORM

#### Participant Ancestry

<table>
<thead>
<tr>
<th>Family Ancestry</th>
<th>□ German</th>
<th>□ British</th>
<th>□ French</th>
<th>□ Italian</th>
<th>□ Irish</th>
<th>□ Scandinavian</th>
<th>□ Other Ancestry ________________</th>
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</thead>
</table>

#### 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

<table>
<thead>
<tr>
<th>DEFINITE</th>
<th>= 2+ major manifestations</th>
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</thead>
<tbody>
<tr>
<td></td>
<td>1 major manifestation &amp; Sibling is affected by APECED</td>
</tr>
<tr>
<td></td>
<td>Disease-causing mutations in both copies of AIRE genes</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>PROBABLE</th>
<th>= 1 major manifestation &amp; 1 minor manifestation</th>
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</thead>
<tbody>
<tr>
<td></td>
<td>Any manifestation &amp; presence of autoantibodies</td>
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</tbody>
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