

APECED Surveillance Recommendations

This handout contains recommendations from the NIH APECED team for the diagnostic surveillance of patients with APECED. These surveillance recommendations are intended to provide a uniform platform for testing of patients. However, any specific hormonal insufficiency state will be managed by local endocrinologists, and the frequency of lab testing should be individualized according to the needs of each patient.

Please fax results to 301-480-5787.

Proposed Yearly Laboratory Work-Up for Patients With APECED

Blood Tests

- CBC with differential
- Peripheral blood smear (looking for Howell-Jolly/Pappenheimer bodies in those who do not have asplenia)
- Comprehensive metabolic panel
- Liver function tests (every 6 months)
- Magnesium
- Phosphorus
- Prealbumin
- Lipid panel
- Immunoglobulin levels (IgG, IgA, IgM)
- ESR and CRP
- TSH and free T4
- HgA1c
- Vitamin B12 and folate
- Iron, transferrin, and ferritin
- Cystatin C
- Alkaline phosphatase, bone-specific
- Vitamin C
- Estradiol for females or testosterone for males
- FSH and LH
- Anti-Müllerian hormone in females only
- ACTH

- ACTH stimulation test (only in patients without adrenal insufficiency)
- Plasma renin activity
- Vitamin D, 25-hydroxy, total
- Vitamin D, 1,25-dihydroxy
- Zinc
- Intrinsic factor antibody, blocking (if not already positive)
- 21-Hydroxylase antibody (if patient does not have adrenal insufficiency)

Radiology and Imaging

- Abdominal ultrasound to evaluate liver, kidneys, and spleen
- DEXA (bone density) scan once every 2 years to include (1) whole body composition and (2) radius, femur, spine-AP

Urine Studies (at least once per year)

- 24-hr urine testing: calcium, magnesium, phosphorus, creatinine clearance, protein
- Spot urine testing: urinalysis, protein/creatinine ratio, albumin/creatinine ratio, beta-2 microglobulin

