

APECED Surveillance Recommendations

This handout contains recommendations from the National Institutes of Health (NIH) APECED team for the diagnostic surveillance of APECED patients. 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 (attention: Elise Ferré). Please address questions to Elise Ferré (elise.ferre@nih.gov) or Michail Lionakis (lionakism@mail.nih.gov).

Yearly Laboratory Work-Up for APECED Patients

Blood Tests

- CBC with differential
- Comprehensive metabolic panel
- Liver function tests (**every 6 months**)
- Magnesium
- Phosphorus
- PT/PTT
- Prealbumin
- Lipid panel
- Immunoglobulin levels (IgG, IgA, IgM, IgE)
- ESR and CRP
- TSH and free T4
- Anti-thyroglobulin and thyroperoxidase antibodies
- HgA1c
- Vitamin B12 and folate
 - If B12 is less than 400 pg/mL, methylmalonic acid, homocysteine, and B12 levels should be checked every 6 months.
- Iron, transferrin, and ferritin
- Cystatin C
- Alkaline phosphatase, bone-specific
- Vitamin C
- Estradiol (females) or testosterone (males)
- FSH and LH

- ACTH
- ACTH stimulation test (**only in patients without adrenal insufficiency**)
- Plasma renin activity
- Insulin-like growth factor-1
- Vitamin D, 25-hydroxy, total
- Vitamin D, 1,25-dihydroxy
- Zinc
- Intrinsic factor antibody, blocking
- 21-Hydroxylase antibody
- GAD65 antibody assay

Radiology and Imaging

- Abdominal ultrasound to evaluate liver and kidneys
- 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-hour urine testing: calcium, magnesium, phosphorus, creatinine clearance, protein
- Spot urine testing: urinalysis, protein/creatinine ratio, albumin/creatinine ratio, beta-2 microglobulin

