

## 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 & Thyroperoxidase antibodies
- HgA1c
- Vitamin B12 and Folate levels
- Iron, Transferrin, and Ferritin
- Cystatin C
- Alkaline Phosphatase, Bone Specific
- Vitamin C level
- Estradiol or Testosterone, for females or males, respectively
- FSH and LH
- Antimullerian Hormone in females only
- 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 / Imaging:**

- Liver Ultrasound once yearly
- Renal Ultrasound once yearly
- DEXA (bone density) scan **once every 2 years**. The DEXA scan should include the following studies:
  - Whole Body
  - 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

## Recommended Surveillance of Potential APECED Manifestations That Have Not Yet Developed

**ENDOCRINE:** All hospital or doctor's visits should include blood pressure and blood draw for serum electrolytes, calcium, and glucose levels. It should also include assessment for presence of fatigue, neuromuscular irritability and anthropometric measures (height and weight).

- If NO hypoparathyroidism: Calcium every 12 months *or when symptomatic*
- If NO Adrenal Insufficiency: ACTH level and **ACTH stimulation test** every 12 months *or when symptomatic*
- If NO hypogonadism: FSH/LH every 12 months *or when symptomatic*
- If NO hypothyroidism: Thyroid Antibody and TSH every 12 months
- If NO Growth Hormone Deficiency: Check height / weight velocity in children and adolescents every 6 months
- If NO Diabetes: GAD65, HgA1c, fasting glucose, urinalysis once a year.

### **GASTROENTEROLOGY/HEPATOLOGY:**

- If NO Hepatitis: Check liver function tests **every 6 months** and obtain liver ultrasound every 12 months *or when symptoms of nausea/vomiting/new abdominal pain.*
- If NO B12 deficiency: Check B12 level every 12 months
  - **If B12 is less than 400: Methylmalonic acid, Homocysteine, and B12 levels every 6 months**