

# Cobalamin Disorder Monitoring Recommendations

## Genetic or Metabolic

### Clinic Visits

Sources : Huemer et al. 2016 <https://doi.org/10.1007/s10545-016-9991-4>  
Sloan et al. 2018 <https://www.ncbi.nlm.nih.gov/books/NBK1328/>

First Year of Life

1-2 times/month  
as needed until metabolic  
control and growth stabilize



Toddler  
School Age  
2-4x/year



Teen  
Adult  
1-2x/year

## Monitor at Every Clinic Visit



### Biochemical/Metabolic Control: Area to Monitor: Recommended Values

**Plasma Total Homocysteine:** <5y/o <40 umol/L <12y/o <60 umol/L Adults <100 umol/L Sick <100 umol/L

**Methionine:** 20-50 umol/L

**Serum Methylmalonic Acid:** <10 umol/L

### Anthropometry

**Height:** Poor Growth

**Weight:** Malnutrition, Poor Weight Gain

**Head Circumference:** Microcephaly

### Diet

**Food Diary:** Poor feeding, Meet caloric and protein goals

## Monitor Yearly

### Area to Monitor: Associated Issues

### Nutritional/Metabolic Labs

**Vitamin B12**

**Plasma Amino Acids:** Track methionine to adjust Betaine and OHCBL

**Complete Blood Count:** Cytopenia, Megaloblastic Anemia

**Prealbumin:** Malnutrition, Liver Function

**Albumin/Total protein:** Malnutrition, Liver Function

**Urine Organic Acids**

**ALT/AST**

**Electrolytes**

**Renal Function**

**Carnitine plasma, total/free**

### Ophthalmological

**Eye Exam:** Maculopathy, Retinopathy, Strabismus detection and management

(retinal photography, optical coherence tomography, ERG (every 6 months first 2 years):

**Ophthalmologic Assessment:** Referral to resources/low vision services for Visually Impaired individuals

### Kidney

**Blood tests:** Creatinine, Blood Urea Nitrogen, Cystatin C: Kidney disease, Thrombotic Microangiopathy, Hemolytic Uremic Syndrome

**Urine tests:** Urinalysis

### Neurological and Neurodevelopment/Neuropsychological

**Clinical Exam:** Hypotonia, Seizures, Peripheral Neuropathy, Abnormal Movements, Developmental milestones, Need for services, IEP

### Cardiovascular

**Cardiovascular Risk Factor Review:** Thromboembolic complications

**Echocardiogram:** Left Ventricular Non-Compaction, Other congenital heart disease

**Blood pressure:** Hypertension

## Monitor As Needed

### Area to Monitor: Associated Issues

### Nutritional/Metabolic Labs

**Ferritin/Iron:** Periodic Nutritional Assessment

**25-Hydroxyvitamin D**

**Essential Fatty Acids**

### Neurology

**Brain and Spine MRI:** Hydrocephalus, Seizures, Movement Disorders. Subacute Combined Degeneration of the spinal cord

**Electroencephalogram (EEG):** Seizures

**Electromyogram/Nerve Conduction Studies:** Neuropathy

### Neurodevelopment /Neuropsychological

**IQ Testing:** Cognitive Impairment, Autism Evaluation

### Psychological

**Clinical Psychology or Psychiatric Assessment:**

Behavioral & Mental Disorders

### Liver Health

**Abdominal Ultrasound:** Fatty Liver

Every 3-5 years from adolescence

Unless clinically indicated earlier

### Rehabilitation Medicine

Physical Medicine and Rehab, Physical, Occupational and Speech therapy

### Bone Health

**DEXA Scan:** Every 5 years from adolescence

Unless clinically indicated earlier: Bone health