

THIS IS NOT A TEST REQUEST FORM.
The information below is required to perform Vitamin B12 panel genetic testing.
Please fill out this form and submit it with the test request form or electronic packing list.

PATIENT HISTORY FOR COBALAMIN/PROPIONATE/HOMOCYSTEINE METABOLISM RELATED DISORDERS GENETIC TESTING

Patient Name _____ **Date of Birth** ____/____/____ **Gender** F M

Physician _____ **Physician Phone** (____) _____ **Practice Specialty** _____

Genetic Counselor _____ **Counselor Phone** (____) _____

PATIENT'S ETHNICITY (check all that apply)

- African-American Ashkenazi Jewish Asian Caucasian
 Hispanic Middle Eastern Native American Other _____

CLINICAL DIAGNOSIS/Reason for referral:

- Methylmalonic aciduria Homocystinuria Propionic acidemia Other _____

SYMPTOMS? No Yes, check all that apply

- | | | |
|---|---|--|
| <input type="checkbox"/> Failure to thrive | <input type="checkbox"/> Craniofacial _____ | <input type="checkbox"/> Renal _____ |
| <input type="checkbox"/> Neurological _____ | <input type="checkbox"/> Gastrointestinal _____ | <input type="checkbox"/> Cutaneous _____ |
| <input type="checkbox"/> Neuromuscular _____ | <input type="checkbox"/> Hematologic _____ | <input type="checkbox"/> Respiratory _____ |
| <input type="checkbox"/> Skeletal _____ | <input type="checkbox"/> Immunologic _____ | <input type="checkbox"/> Other _____ |
| <input type="checkbox"/> Cardiovascular _____ | <input type="checkbox"/> Ocular _____ | |

LABORATORY FINDINGS

- Acidosis Hypoglycemia
- | | | | | |
|---|---------------------------------|---|--|----------------------------------|
| Plasma Acylcarnitine Profile | <input type="checkbox"/> Normal | <input type="checkbox"/> Abnormal (result: _____) | <input type="checkbox"/> Not performed | <input type="checkbox"/> Unknown |
| Plasma Amino Acids | <input type="checkbox"/> Normal | <input type="checkbox"/> Abnormal (result: _____) | <input type="checkbox"/> Not performed | <input type="checkbox"/> Unknown |
| Urine Organic acids | <input type="checkbox"/> Normal | <input type="checkbox"/> Abnormal (result: _____) | <input type="checkbox"/> Not performed | <input type="checkbox"/> Unknown |
| Vitamin B12 | <input type="checkbox"/> Normal | <input type="checkbox"/> Abnormal (result: _____) | <input type="checkbox"/> Not performed | <input type="checkbox"/> Unknown |
| Methylmalonic acid, serum /plasma/urine | <input type="checkbox"/> Normal | <input type="checkbox"/> Abnormal (result: _____) | <input type="checkbox"/> Not performed | <input type="checkbox"/> Unknown |
| Homocysteine, Total | <input type="checkbox"/> Normal | <input type="checkbox"/> Abnormal (result: _____) | <input type="checkbox"/> Not performed | <input type="checkbox"/> Unknown |

Enzyme analysis (specify) _____

Fibroblast complementation studies (specify) _____

Histochemistry _____

MRI _____

Other _____

Has the patient had an allogeneic bone marrow or umbilical cord blood transplant? No Yes Unknown

Has the patient undergone previous DNA testing? No Yes Unknown

If yes, please describe the gene/disorder, methodology, and results _____

FAMILY HISTORY of cobalamin/propionate/homocysteine metabolism related disorder? No Yes Unknown

If yes, attach a PEDIGREE or specify the relative's relationship to the patient, symptoms/clinical diagnosis and age of onset. _____

Has DNA testing been performed for these family member(s)? No Yes Unknown

Attach a copy of the relative's DNA laboratory result. REQUIRED for familial mutation testing.

Circle the test you intend to order.

Initial Test for Cobalamin/Propionate/Homocysteine Metabolism Related Disorder	
2011157	Cobalamin/Propionate/Homocysteine Metabolism Related Disorders Panel, Sequencing (25 Genes) and Deletion/Duplication (24 Genes)
Follow-up Testing for Family Members- A copy of a relative's DNA laboratory result is REQUIRED	
2001961	Familial Mutation, Targeted Sequencing: Tests for a specific sequence change previously identified in a family member.

For questions, contact a genetic counselor at (800) 242-2787, ext. 2141

Master Label