ARUP LABORATORIES | NATIONAL REFERENCE LABORATORY

A nonprofit enterprise of the University of Utah and its Department of Pathology

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PATIENT HISTORY FOR COBALAMIN/PROPIONATE/HOMOCYSTEINE RELATED DISORDERS GENE PANEL

Patient Name _____ **Date of Birth** _____ **Sex** ☐ **F** ☐ **M**

Physician _____ **Physician Phone** _____

Practice Specialty _____ **Physician Fax** _____

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 _____

Patient's Symptoms: Check all that apply.

☐ Failure to thrive ☐ Craniofacial _____ ☐ Renal _____
☐ Neurological _____ ☐ Gastrointestinal _____ ☐ Cutaneous _____
☐ Neuromuscular _____ ☐ Hematologic _____ ☐ Respiratory _____
☐ Skeletal _____ ☐ Immunologic _____ ☐ Other _____
☐ Cardiovascular _____ ☐ Ocular _____ _____

Laboratory Findings

***Vitamin B12**	☐ Normal	☐ Abnormal (result: _____)	☐ Not performed	☐ Unknown
***Methylmalonic acid before vitamin B12 therapy**	☐ Normal	☐ Abnormal (result: _____)	☐ Not performed	☐ Unknown
***Methylmalonic acid after vitamin B12 therapy**	☐ Normal	☐ Abnormal (result: _____)	☐ Not performed	☐ Unknown
Homocysteine, Total	☐ Normal	☐ Abnormal (result: _____)	☐ Not performed	☐ Unknown
Plasma Acylcarnitine Profile	☐ Normal	☐ Abnormal (result: _____)	☐ Not performed	☐ Unknown
Plasma Amino Acids	☐ Normal	☐ Abnormal (result: _____)	☐ Not performed	☐ Unknown
Urine Organic Acids	☐ Normal	☐ Abnormal (result: _____)	☐ Not performed	☐ Unknown
Acidosis	☐			
Hypoglycemia	☐			

***If the patient is a breast fed baby, was MATERNAL vitamin B12 deficiency excluded?** _____

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, specify the affected relative's relationship to the patient, symptoms/clinical diagnosis, and age of onset. _____

_____

Has DNA testing been performed for these family members? ☐ No ☐ Yes ☐ Unknown
Attach a copy of the relative's DNA laboratory result. **REQUIRED for familial mutation testing.**

Check the test you intend to order:

☐ **Initial Test for Cobalamin/Propionate/Homocysteine Metabolism Related Disorder**
2011157 Cobalamin/Propionate/Homocysteine 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 an ARUP genetic counselor at (800) 242-2787, ext. 2141

Master Label