ARUP LABORATORIES

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

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THIS IS NOT A TEST REQUEST FORM. Please complete and submit with the test request form or electronic packing list.

COBALAMIN/PROPIONATE/HOMOCYSTEINE-RELATED DISORDERS GENE PANEL PATIENT HISTORY FORM

Patient Name: _____ **Date of Birth:** _____ **Sex:** ☐ Female ☐ Male

Ordering Provider: _____ **Provider's Phone:** _____

Practice Specialty: _____ **Provider's Fax:** _____

Genetic Counselor: _____ **Counselor Phone:** _____

Patient's Ethnicity/Ancestry (check all that apply)

☐ African American/Black ☐ Asian ☐ Hispanic ☐ White ☐ Other: _____

List country of origin (if known): _____

Clinical Diagnosis/Reason for Referral:

☐ Methylmalonic aciduria ☐ Homocystinuria ☐ Propionic acidemia ☐ Other _____

Patient's Symptoms (check all that apply and describe):

☐ Failure to thrive	☐ Craniofacial_____	☐ Renal_____
☐ Neurological_____	☐ Gastrointestinal_____	☐ Cutaneous_____
☐ Neuromuscular_____	☐ Hematologic_____	☐ Respiratory_____
☐ Skeletal_____	☐ Immunologic_____	☐ Other_____
☐ Cardiovascular_____	☐ Ocular_____	_____

Laboratory Findings

*Vitamin B12	☐ Normal (result: _____)	☐ 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
***If the patient is a breastfed baby, was MATERNAL vitamin B12 deficiency excluded?**	☐ No	☐ Yes		☐ 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

Has the patient undergone previous DNA testing? ☐ No ☐ Yes ☐ Unknown

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

Is there any relevant <u>family history</u> of cobalamin/propionate/homocysteine metabolism related disorder? ☐ No ☐ Yes ☐ Unknown

If yes, attach a pedigree or specify the relative's <u>relationship</u> to the patient. List their <u>symptoms</u> and <u>age of onset</u>:

_____

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

If yes, attach a copy of the relative's DNA laboratory result (REQUIRED for familial mutation testing).

Check the test you intend to order.

<u>Initial Test for cobalamin/propionate/homocysteine metabolism related disorder</u>

☐ **2011157 Cobalamin/Propionate/Homocysteine Metabolism Related Disorders Panel, Sequencing and Deletion/Duplication**

<u>Follow-up testing for family members</u>

☐ **2001961 Familial Mutation, Targeted Sequencing:** Tests for a specific sequence change previously identified in a family member; a copy of the relative's laboratory result is REQUIRED.

Master Label

For questions, contact an ARUP genetic counselor at 800-242-2787 ext. 2141.