ARUP LABORATORIES | NATIONAL REFERENCE LABORATORY

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 fill out this form and submit it with the test request form or electronic packing list.

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 ☐ Asian ☐ Hispanic ☐ Native American
☐ Ashkenazi Jewish ☐ Caucasian ☐ Middle Eastern ☐ Other: _____

Clinical Diagnosis /Reason for Referral:

☐ Methylmalonic aciduria ☐ Homocystinuria ☐ Propionic acidemia ☐ Other: _____

Patient's Symptoms (check all that apply)

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

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 ☐ N/A

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, 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

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