ARUP LABORATORIES | NATIONAL REFERENCE LABORATORY

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

500 Chipeta Way
Salt Lake City, Utah 84108-1221
phone: (801) 583-2787 | toll free: (800) 242-2787
fax: (801) 584-5249 | www.aruplab.com

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 CREATINE DEFICIENCY SYNDROMES TESTING

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: _____

Does the patient have symptoms? ☐ No ☐ Yes (check all that apply)

☐ Autistic behaviors
☐ Intellectual disability
☐ Movement disorder
☐ Seizures
☐ Self injury
☐ Speech/language delay
☐ Other symptom(s): _____

Laboratory Findings:

Brain creatine level (by MRS)	☐ Normal	☐ Low	☐ High	☐ Not performed	☐ Unknown
Guanidinoacetate (plasma)	☐ Normal	☐ Low	☐ High	☐ Not performed	☐ Unknown
Guanidinoacetate (urine)	☐ Normal	☐ Low	☐ High	☐ Not performed	☐ Unknown
Creatine (plasma)	☐ Normal	☐ Low	☐ High	☐ Not performed	☐ Unknown
Creatine (urine)	☐ Normal	☐ Low	☐ High	☐ Not performed	☐ Unknown
Creatine: creatinine ratio (urine)	☐ Normal	☐ Low	☐ High	☐ Not performed	☐ Unknown
Creatine transport (fibroblasts)	☐ Normal	☐ Abnormal (activity: _____)		☐ Not performed	☐ Unknown

Is there any relevant family history? ☐ No ☐ Yes ☐ Unknown

If yes, attach a pedigree or specify the relative's relationship to the patient. List their symptoms:

_____

_____

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.

Biochemical Testing

☐ **2002333 Creatine Disorders Panel, Urine:** Order as initial test with plasma panel in individuals with symptoms or abnormal MRS.
☐ **2002328 Creatine Disorders Panel, Plasma:** Order as initial test with urine panel in individuals with symptoms or abnormal MRS.

Molecular Testing

☐ **2008610 Creatine Transport Deficiency (*SLC6A8*) Sequencing and Deletion/Duplication:** Sequencing of *SLC6A8* coding regions and intron/exon boundaries, MLPA to detect deletions/duplications. Clinical sensitivity 99%.
☐ **2008615 Creatine Transport Deficiency (*SLC6A8*) Sequencing:** Sequencing of *SLC6A8* coding regions and intron/exon boundaries. Clinical sensitivity predicted to be up to 99%.
☐ **2008606 Creatine Transport Deficiency (*SLC6A8*) Deletion/Duplication:** Deletion/duplication analysis of all *SLC6A8* exons. Clinical sensitivity is unknown, possibly 1–2%.
☐ **2011144 Arginine: Glycine Amidinotransferase (*GATM*) Deficiency Sequencing:** Sequencing of the *GATM* coding regions and intron/exon boundaries. Clinical sensitivity predicted to be up to 99%.
☐ **2011140 Guanidinoacetate Methyltransferase (*GAMT*) Deficiency Sequencing:** Sequencing of the *GAMT* coding regions and intron/exon boundaries. Clinical sensitivity predicted to be up to 99%.
☐ **2001961 Familial Mutation, Targeted Sequencing:** Tests for a mutation previously identified in a family member; a copy of relative's lab result is REQUIRED.

Master Label

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