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 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:** ☐ Female ☐ Male

Physician: _____ **Physician Phone:** _____

Practice Specialty: _____ **Physician Fax:** _____

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

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

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

List countries of origin (if known):_____

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

☐ Autistic behaviors ☐ Movement disorder ☐ Self-injury ☐ Other symptom(s):

☐ Intellectual disability ☐ Seizures ☐ Speech/language delay _____

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 each 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, Serum or Plasma:**
Order as initial test with urine panel in individuals with symptoms or abnormal MRS.

Molecular Testing

☐ **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.

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

Master Label