

THIS IS NOT A TEST REQUEST FORM
The information below is required to perform creatine deficiency testing.
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 INFORMATION

Patient's Last Name: _____ First Name: _____ MI: _____
 Birth Date: _____ Gender: Male Female
 Referring Physician (last, first): _____
 Referring Physician Phone: (_____) _____ Practice Specialty: _____
 Genetic Counselor (last, first): _____
 Genetic 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 OF A CREATINE DEFICIENCY SYNDROME? No Yes Unknown

If Yes, check all that apply:

- Intellectual disability Autistic behaviors Speech/Language delay Other _____
 Movement disorder Self injury Seizures

LABORATORY FINDINGS

- | | | | | | |
|-----------------------------------|---------------------------------|------------------------------|-------------------------------|--|----------------------------------|
| Brain creatine level (by MRS) | <input type="checkbox"/> Normal | <input type="checkbox"/> Low | <input type="checkbox"/> High | <input type="checkbox"/> Not performed | <input type="checkbox"/> Unknown |
| Guanidinoacetate (plasma) | <input type="checkbox"/> Normal | <input type="checkbox"/> Low | <input type="checkbox"/> High | <input type="checkbox"/> Not performed | <input type="checkbox"/> Unknown |
| Guanidinoacetate (urine) | <input type="checkbox"/> Normal | <input type="checkbox"/> Low | <input type="checkbox"/> High | <input type="checkbox"/> Not performed | <input type="checkbox"/> Unknown |
| Creatine (plasma) | <input type="checkbox"/> Normal | <input type="checkbox"/> Low | <input type="checkbox"/> High | <input type="checkbox"/> Not performed | <input type="checkbox"/> Unknown |
| Creatine (urine) | <input type="checkbox"/> Normal | <input type="checkbox"/> Low | <input type="checkbox"/> High | <input type="checkbox"/> Not performed | <input type="checkbox"/> Unknown |
| Creatine:creatinine ratio (urine) | <input type="checkbox"/> Normal | <input type="checkbox"/> Low | <input type="checkbox"/> High | <input type="checkbox"/> Not performed | <input type="checkbox"/> Unknown |
| Creatine transport (fibroblasts) | <input type="checkbox"/> Normal | <input type="checkbox"/> Low | <input type="checkbox"/> High | <input type="checkbox"/> Not performed | <input type="checkbox"/> Unknown |

FAMILY HISTORY OF ANY OF THE ABOVE SYMPTOMS? No Yes Unknown

If Yes, please specify the relationship of the family member(s) to the patient and describe the symptoms in each symptomatic relative:

Has DNA testing for creatine deficiency syndromes been performed for these family members? No Yes Unknown

If Yes, please attach a copy of the laboratory result (REQUIRED for familial mutation testing).

DESCRIPTION OF CREATINE DEFICIENCY SYNDROMES TESTING

- UGUANI Creatine Disorders Panel, Urine:** Order as initial test with plasma in individuals with symptoms or abnormal MRS.
 GUANID Creatine Disorders Panel, Blood: Order as initial test with urine panel in individuals with symptoms or abnormal MRS.