

Overview

Useful For

Evaluation of patients with a clinical suspicion of arginine:glycine amidinotransferase deficiency and guanidinoacetate methyltransferase deficiency.

This test is **not useful for** the diagnosis of creatine transporter deficiency.

Genetics Test Information

Depletion of cerebral creatine occurs in all 3 types of creatine deficiency syndromes (CDS): arginine:glycine amidinotransferase deficiency, guanidinoacetate methyltransferase deficiency, and creatine transporter deficiency. Measurement of guanidinoacetate, creatine, and creatinine along with associated analyte ratios in serum and urine distinguishes among the types of creatine deficiency syndromes.

Treatment with oral creatine supplementation is effective in some types of CDS.

Creatine supplementation may impact reliability of test results.

Testing Algorithm

For more information, see [Newborn Screening Act Sheet Guanidinoacetate Methyltransferase Deficiency: Increased Guanidinoacetate](#) in Special Instructions.

The following algorithms are available in Special Instructions:

- [Newborn Screen Follow-up for Guanidinoacetate Methyltransferase Deficiency \(GAMT\)](#)
- [Epilepsy: Unexplained Refractory and/or Familial Testing Algorithm](#)

Special Instructions

- [Newborn Screening Act Sheet Guanidinoacetate Methyltransferase Deficiency: Increased Guanidinoacetate](#)
- [Newborn Screen Follow-up for Guanidinoacetate Methyltransferase Deficiency \(GAMT\)](#)
- [Epilepsy: Unexplained Refractory and/or Familial Testing Algorithm](#)

Method Name

Liquid Chromatography-Tandem Mass Spectrometry (LC-MS/MS)

NY State Available

Yes

Specimen

Specimen Type

Serum

Ordering Guidance

For diagnosis of creatine transporter deficiency, order CRDPU / Creatine Disorders Panel, Random, Urine.

Additional Testing Requirements

To diagnose all creatine deficiency syndromes, order CRDPU / Creatine Disorders Panel, Random, Urine in addition to this test.

Necessary Information

Patient's age and sex are required.

Specimen Required

Collection Container/Tube: Red top (serum gel/SST are **not acceptable**)

Submission Container/Tube: Plastic vial

Specimen Volume: 0.5 mL

Collection Instructions: Centrifuge and aliquot serum into plastic vial. Send serum frozen.

Reject Due To

Gross hemolysis OK

Gross lipemia OK

Gross icterus OK

Specimen Minimum Volume

0.1 mL

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Serum	Frozen (preferred)	14 days	
	Refrigerated	7 days	
	Ambient		

Clinical & Interpretive
Clinical Information

Disorders of creatine synthesis (guanidinoacetate methyltransferase: GAMT, L-arginine:glycine amidinotransferases: AGAT, and creatine transporter deficiency: CTD) are collectively described as creatine deficiency syndromes (CDS). GAMT and AGAT deficiencies are inherited in an autosomal recessive manner, while CTD is X-linked. All 3 disorders result in a depletion of cerebral creatine and typically present with global developmental delays, intellectual disability, and severe speech delay. Affected patients may have abnormal magnetic resonance imaging findings and exhibit cerebral creatine deficiency in brain magnetic resonance spectroscopy. Patients with GAMT and male patients with CTD develop seizures, behavioral problems, and autistic features and may have abnormal movements. Female carriers for CTD can be asymptomatic or exhibit features similar to affected male patients such as intellectual disability, behavioral problems, and seizures.

Diagnosis of creatine synthesis disorders relies on measurement of guanidinoacetate (GAA), creatine (Cr), and creatinine (Crn) in serum and urine. The profiles are specific for each clinical entity. In serum, patients with GAMT deficiency typically exhibit very elevated GAA, low Cr, and normal to low Crn. Patients with AGAT deficiency typically exhibit low to normal GAA, low Cr, and normal to low Crn. In comparison, patients with CTD typically have normal serum levels of GAA, Cr and Crn, and measurement of these analytes in urine is required for diagnosis in male patients (characteristic findings are elevated Cr, normal to low Crn, and an elevated Cr:Crn ratio in urine). The only reliable method for diagnosis of CTD in female patients is molecular analysis of the *SLC6A8* gene. The diagnosis of GAMT and AGAT can be confirmed by molecular analysis of *GAMT* and *GATM*.

Treatment with oral supplementation of creatine monohydrate is available and effective for the AGAT and GAMT deficiencies. Patients with GAMT deficiency may also be treated with supplemental ornithine and dietary arginine restriction.

Early treatment has been reported to prevent disease manifestations in affected but presymptomatic newborn siblings of individuals with GAMT or AGAT deficiencies.

Reference Values

Creatine Disorders Panel Reference Values (creatinine, creatinine, and guanidinoacetate results reported as nmol/mL)	< or =11 Months		12-23 Months		24-35 Months	Female
	Male	Female	Male	Female	Male	Creatine
38.6-96.8	39.0-97.0	38.2-96.5	38.6-96.5	37.7-96.0	38.2-96.0	
Creatinine	27.6-35.9	27.6-35.2	27.6-36.5	27.6-35.5	27.6-37.1	
27.6-36.0	Guanidinoacetate	0.7-2.0	0.7-2.1	0.7-2.0	0.7-2.1	
0.7-2.0		0.7-2.1	Creatine/creatinine	< or =3.07	< or =3.60	< or =3.02
< or =3.54	< or =2.96	< or =3.48	Guanidinoacetate/ creatine	< or =0.040	< or =0.040	
< or =0.042	< or =0.040	< or =0.043		< or =0.042	Guanidinoacetate/ creatinine	< or =0.051
< or =0.081	< or =0.051	< or =0.080	< or =0.051	< or =0.079		
	3 Years	4 Years	5 Years	Female	Male	
Female	Male	Female	Male	Creatine	37.1-95.5	
37.7-95.3	36.0-94.4	36.8-94.1	34.6-93.2	35.6-92.7	Creatinine	
27.6-37.9	27.7-36.9	27.6-39.3	27.7-38.2	27.6-40.9	27.8-39.9	
Guanidinoacetate	0.7-2.1	0.7-2.2	0.7-2.1	0.7-2.2	0.7-2.1	
0.7-2.2	Creatine/creatinine	< or =2.89	< or =3.40	< or =2.77	< or =3.26	
< or =2.64	< or =3.09	Guanidinoacetate	< or =0.045	< or =0.043	< or =0.049	

< or =0.045	< or =0.053	te/ creatine < or =0.049		< or =0.050	< or =0.077
			Guanidinoaceta te/ creatinine		
< or =0.050	< or =0.075	< or =0.049	< or =0.072		
6 Years	7 Years	8 Years	Female	Male	Female
Male	Female	Male	Creatine	33.0-91.7	34.3-91.0
31.2-90.0	32.7-89.2	29.2-88.1	31.0-87.3	Creatinine	27.6-42.8
28.0-41.9	27.7-44.9	28.3-44.3	27.8-47.0	28.8-47.1	Guanidinoaceta te
0.7-2.1	0.7-2.3	0.7-2.1	0.8-2.3	0.8-2.1	0.8-2.4
Creatine/ creatinine	< or =2.49	< or =2.91	< or =2.33	< or =2.70	< or =2.17
< or =2.49	Guanidinoaceta te/ creatine	< or =0.058	< or =0.053	< or =0.063	< or =0.058
< or =0.069	< or =0.064	Guanidinoaceta te/ creatinine	< or =0.049	< or =0.069	< or =0.048
< or =0.066	< or =0.047	< or =0.063		9 Years	10 Years
11 Years	Female	Male	Female	Male	Female
Male	Creatine	27.2-85.9	29.3-85.2	25.2-83.7	27.4-83.1
23.4-81.3	25.7-80.9	Creatinine	28.0-49.3	29.5-50.1	28.2-51.5
30.6-53.6	28.4-53.6	32.0-57.2	Guanidinoaceta te	0.8-2.2	0.8-2.5
0.9-2.2	0.9-2.6	0.9-2.2	1.0-2.6	Creatine/ creatinine	< or =2.02
< or =2.28	< or =1.86	< or =2.07	< or =1.72	< or =1.87	Guanidinoaceta te/ creatine
< or =0.075	< or =0.070	< or =0.081	< or =0.078	< or =0.087	< or =0.085
Guanidinoaceta te/ creatinine	< or =0.047	< or =0.060	< or =0.046	< or =0.057	< or =0.045
< or =0.055		12 Years	13 Years	14 Years	Female
Male	Female	Male	Female	Male	Creatine
21.7-78.7	23.9-78.6	20.3-76.2	22.3-76.2	19.0-73.6	20.8-73.8
Creatinine	28.7-55.7	33.8-61.0	29.1-57.7	35.9-64.8	29.5-59.5
38.1-68.5	Guanidinoaceta	0.9-2.2	1.0-2.7	1.0-2.3	1.1-2.8

	te				
1.0-2.3	1.1-2.9	Creatine/ creatinine	< or =1.58	< or =1.68	< or =1.45
< or =1.50	< or =1.33	< or =1.34	Guanidinoaceta te/ creatinine	< or =0.092	< or =0.093
< or =0.097	< or =0.101	< or =0.101	< or =0.109	Guanidinoaceta te/ creatinine	< or =0.044
< or =0.053	< or =0.043	< or =0.051	< or =0.042	< or =0.050	
15 Years	16 Years	17 Years	Female	Male	Female
Male	Female	Male	Creatine	18.1-71.1	19.5-71.2
17.4-68.7	18.4-68.6	16.9-66.5	17.4-65.9	Creatinine	29.9-61.3
40.4-71.9	30.4-62.9	42.4-75.0	30.9-64.4	44.2-77.6	Guanidinoaceta te
1.0-2.3	1.2-2.9	1.1-2.3	1.3-3.0	1.1-2.3	1.3-3.1
Creatine/ creatinine	< or =1.22	< or =1.20	< or =1.12	< or =1.07	< or =1.04
< or =0.97	Guanidinoaceta te/ creatinine	< or =0.104	< or =0.117	< or =0.107	< or =0.125
< or =0.109	< or =0.132	Guanidinoaceta te/ creatinine	< or =0.041	< or =0.049	< or =0.040
< or =0.048	< or =0.040	< or =0.048		18 Years	19 Years
20 Years	Female	Male	Female	Male	Female
Male	Creatine	16.7-64.4	16.6-63.3	16.6-62.7	15.8-60.7
16.5-61.1	15.2-58.3	Creatinine	31.3-65.8	45.6-80.0	31.8-67.0
46.7-82.0	32.2-68.2	47.4-83.9	Guanidinoaceta te	1.1-2.4	1.4-3.1
1.1-2.4	1.4-3.2	1.1-2.4	1.5-3.2	Creatine/ creatinine	< or =0.98
< or =0.87	< or =0.93	< or =0.80	< or =0.89	< or =0.73	Guanidinoaceta te/ creatinine
< or =0.111	< or =0.139	< or =0.112	< or =0.145	< or =0.113	< or =0.150
Guanidinoaceta te/ creatinine	< or =0.039	< or =0.047	< or =0.038	< or =0.047	< or =0.038
< or =0.046		21 Years	22 Years	23 Years	Female
Male	Female	Male	Female	Male	Creatine
16.6-59.8	14.7-56.0	16.6-58.8	14.2-54.0	16.7-57.9	13.7-52.2

Creatinine	32.5-69.2	47.9-85.6	32.8-70.2	48.2-87.2	33.1-71.1
48.4-88.8	Guanidinoacetate	1.1-2.4	1.5-3.2	1.1-2.5	1.5-3.3
1.1-2.5	1.6-3.3	Creatine/creatinine	< or =0.87	< or =0.68	< or =0.85
< or =0.64	< or =0.84	< or =0.61	Guanidinoacetate/ creatine	< or =0.114	< or =0.156
< or =0.115	< or =0.161	< or =0.116	< or =0.165	Guanidinoacetate/ creatinine	< or =0.037
< or =0.045	< or =0.037	< or =0.045	< or =0.037	< or =0.044	
24 Years	25 Years	26 Years	Female	Male	Female
Male	Female	Male	Creatine	16.7-57.2	13.3-50.6
16.7-56.5	12.9-49.3	16.7-56.0	12.5-48.1	Creatinine	33.3-71.9
48.6-90.2	33.6-72.8	48.7-91.5	33.7-73.6	48.9-92.7	Guanidinoacetate
1.1-2.5	1.6-3.3	1.1-2.5	1.6-3.3	1.1-2.6	1.6-3.4
Creatine/creatinine	< or =0.84	< or =0.58	< or =0.84	< or =0.56	< or =0.84
< or =0.54	Guanidinoacetate/ creatine	< or =0.116	< or =0.170	< or =0.117	< or =0.174
< or =0.118	< or =0.179	Guanidinoacetate/ creatinine	< or =0.036	< or =0.043	< or =0.036
< or =0.043	< or =0.036	< or =0.042		27 Years	28 Years
29 Years	Female	Male	Female	Male	Female
Male	Creatine	16.7-55.5	12.1-47.1	16.6-55.1	11.8-46.3
16.5-54.7	11.5-45.4	Creatinine	33.9-74.4	49.0-93.7	34.1-75.2
49.1-94.5	34.2-76.0	49.2-95.3	Guanidinoacetate	1.1-2.6	1.6-3.4
1.1-2.6	1.6-3.4	1.1-2.6	1.6-3.4	Creatine/creatinine	< or =0.84
< or =0.52	< or =0.84	< or =0.51	< or =0.84	< or =0.49	Guanidinoacetate/ creatine
< or =0.118	< or =0.182	< or =0.119	< or =0.186	< or =0.119	< or =0.188
Guanidinoacetate/ creatinine	< or =0.036	< or =0.042	< or =0.036	< or =0.041	< or =0.036

< or =0.041		30 Years	31 Years	32 Years	Female
Male	Female	Male	Female	Male	Creatine
16.4-54.2	11.3-44.7	16.4-53.8	11.1-43.9	16.3-53.4	11.0-43.2
Creatinine	34.4-76.8	49.3-96.0	34.6-77.5	49.3-96.7	34.7-78.2
49.4-97.4	Guanidinoacetate	1.2-2.7	1.6-3.5	1.2-2.7	1.6-3.5
1.2-2.7	1.6-3.5	Creatine/ creatinine	< or =0.84	< or =0.48	< or =0.83
< or =0.47	< or =0.83	< or =0.46	Guanidinoacetate/ creatinine	< or =0.120	< or =0.190
< or =0.120	< or =0.192	< or =0.119	< or =0.192	Guanidinoacetate/ creatinine	< or =0.036
< or =0.041	< or =0.036	< or =0.042	< or =0.037	< or =0.042	
33 Years	34 Years	35 Years	Female	Male	Female
Male	Female	Male	Creatine	16.3-53.0	10.9-42.5
16.4-52.7	10.8-41.7	16.4-52.3	10.7-41.0	Creatinine	34.9-78.8
49.4-98.0	35.1-79.4	49.5-98.6	35.3-79.9	49.5-99.2	Guanidinoacetate
1.2-2.8	1.6-3.5	1.2-2.8	1.6-3.5	1.2-2.8	1.6-3.4
Creatine/ creatinine	< or =0.82	< or =0.45	< or =0.82	< or =0.45	< or =0.82
< or =0.44	Guanidinoacetate/ creatinine	< or =0.119	< or =0.192	< or =0.118	< or =0.191
< or =0.118	< or =0.189	Guanidinoacetate/ creatinine	< or =0.037	< or =0.042	< or =0.037
< or =0.042	< or =0.037	< or =0.042		36 Years	37 Years
38 Years	Female	Male	Female	Male	Female
Male	Creatine	16.5-52.0	10.7-40.2	16.7-51.6	10.6-39.5
16.9-51.3	10.6-38.9	Creatinine	35.4-80.3	49.5-99.8	35.6-80.7
49.5-100.3	35.8-81.0	49.6-100.8	Guanidinoacetate	1.2-2.8	1.6-3.4
1.2-2.8	1.6-3.4	1.2-2.9	1.6-3.4	Creatine/ creatinine	< or =0.82
< or =0.44	< or =0.82	< or =0.44	< or =0.83	< or =0.44	Guanidinoacetate/ creatinine
< or =0.117	< or =0.187	< or =0.115	< or =0.184	< or =0.114	< or =0.182

Guanidinoacetate/ creatinine	< or =0.037	< or =0.042	< or =0.037	< or =0.042	< or =0.036
< or =0.042		39 Years	40 Years	41 Years	Female
Male	Female	Male	Female	Male	Creatine
17.1-51.1	10.6-38.2	17.3-50.9	10.7-37.7	17.5-50.8	10.7-37.2
Creatinine	35.9-81.4	49.6-101.3	36.0-81.6	49.6-101.7	36.1-81.9
49.7-102.1	Guanidinoacetate	1.2-2.9	1.6-3.4	1.2-2.9	1.6-3.4
1.2-2.9	1.6-3.4	Creatine/ creatinine	< or =0.83	< or =0.44	< or =0.83
< or =0.44	< or =0.84	< or =0.44	Guanidinoacetate/ creatine	< or =0.113	< or =0.179
< or =0.111	< or =0.176	< or =0.110	< or =0.174	Guanidinoacetate/ creatinine	< or =0.036
< or =0.041	< or =0.036	< or =0.041	< or =0.036	< or =0.040	
42 Years	43 Years	44 Years	Female	Male	Female
Male	Female	Male	Creatine	17.7-50.8	10.8-36.7
17.8-50.8	10.9-36.3	17.8-50.9	11.0-36.0	Creatinine	36.2-82.1
49.7-102.5	36.3-82.4	49.8-102.8	36.4-82.6	49.8-103.1	Guanidinoacetate
1.2-3.0	1.6-3.3	1.2-3.0	1.6-3.3	1.2-3.0	1.6-3.3
Creatine/ creatinine	< or =0.84	< or =0.44	< or =0.84	< or =0.43	< or =0.84
< or =0.43	Guanidinoacetate/ creatine	< or =0.109	< or =0.172	< or =0.108	< or =0.171
< or =0.107	< or =0.170	Guanidinoacetate/ creatinine	< or =0.036	< or =0.039	< or =0.036
< or =0.039	< or =0.036	< or =0.038		45 Years	46 Years
47 Years	Female	Male	Female	Male	Female
Male	Creatine	17.7-51.0	11.1-35.6	17.6-51.2	11.2-35.3
17.4-51.4	11.3-35.1	Creatinine	36.4-82.8	49.9-103.4	36.5-83.0
49.9-103.6	36.5-83.2	49.9-103.9	Guanidinoacetate	1.2-3.0	1.7-3.3
1.2-3.1	1.7-3.3	1.2-3.1	1.7-3.3	Creatine/ creatinine	< or =0.84
< or =0.42	< or =0.83	< or =0.41	< or =0.83	< or =0.40	

					Guanidinoaceta te/ creatine
< or =0.106	< or =0.169	< or =0.106	< or =0.168	< or =0.106	< or =0.167
Guanidinoaceta te/ creatinine	< or =0.037	< or =0.038	< or =0.037	< or =0.037	< or =0.037
< or =0.037		48 Years	49 Years	50 Years	Female
Male	Female	Male	Female	Male	Creatine
17.2-51.7	11.5-34.8	17.1-51.9	11.6-34.6	17.0-52.1	11.7-34.4
Creatinine	36.6-83.4	49.9-104.1	36.6-83.5	49.9-104.2	36.7-83.7
49.9-104.4	Guanidinoaceta te	1.2-3.1	1.7-3.3	1.2-3.1	1.7-3.3
1.2-3.1	1.7-3.3	Creatine/ creatinine	< or =0.82	< or =0.39	< or =0.82
< or =0.38	< or =0.82	< or =0.38	Guanidinoaceta te/ creatine	< or =0.106	< or =0.166
< or =0.106	< or =0.164	< or =0.105	< or =0.163	Guanidinoaceta te/ creatinine	< or =0.038
< or =0.036	< or =0.038	< or =0.036	< or =0.039	< or =0.036	
51 Years	52 Years	53 Years	Female	Male	Female
Male	Female	Male	Creatine	17.0-52.2	11.9-34.3
17.1-52.3	12.0-34.3	17.3-52.4	12.2-34.3	Creatinine	36.8-83.9
49.8-104.6	36.8-84.0	49.8-104.8	36.9-84.2	49.8-104.9	Guanidinoaceta te
1.2-3.1	1.7-3.3	1.2-3.1	1.7-3.2	1.2-3.1	1.6-3.2
Creatine/ creatinine	< or =0.82	< or =0.37	< or =0.82	< or =0.37	< or =0.82
< or =0.38	Guanidinoaceta te/ creatine	< or =0.105	< or =0.161	< or =0.104	< or =0.159
< or =0.103	< or =0.157	Guanidinoaceta te/ creatinine	< or =0.039	< or =0.036	< or =0.039
< or =0.036	< or =0.039	< or =0.036		54 Years	55 Years
56 Years	Female	Male	Female	Male	Female
Male	Creatine	17.6-52.5	12.4-34.3	18.1-52.6	12.5-34.4
18.6-52.7	12.6-34.4	Creatinine	37.0-84.4	49.8-105.0	37.1-84.5
49.8-105.2	37.2-84.7	49.8-105.3	Guanidinoaceta te	1.2-3.1	1.6-3.2

1.2-3.1	1.6-3.2	1.2-3.1	1.6-3.2	Creatine/ creatinine	< or =0.82
< or =0.38	< or =0.83	< or =0.39	< or =0.84	< or =0.40	Guanidinoaceta te/ creatinine
< or =0.102	< or =0.155	< or =0.100	< or =0.154	< or =0.099	< or =0.152
Guanidinoaceta te/ creatinine	< or =0.039	< or =0.036	< or =0.039	< or =0.036	< or =0.039
< or =0.036		57 Years	58 Years	59 Years	Female
Male	Female	Male	Female	Male	Creatine
19.2-52.9	12.7-34.4	19.9-53.1	12.8-34.3	20.5-53.3	12.8-34.2
Creatinine	37.3-84.9	49.8-105.4	37.4-85.1	49.8-105.5	37.6-85.2
49.8-105.6	Guanidinoaceta te	1.2-3.0	1.6-3.2	1.3-3.0	1.6-3.2
1.3-3.0	1.6-3.2	Creatine/ creatinine	< or =0.84	< or =0.40	< or =0.85
< or =0.41	< or =0.86	< or =0.42	Guanidinoaceta te/ creatinine	< or =0.098	< or =0.151
< or =0.096	< or =0.151	< or =0.095	< or =0.150	Guanidinoaceta te/ creatinine	< or =0.039
< or =0.036	< or =0.038	< or =0.036	< or =0.038	< or =0.036	
60 Years	61 Years	62 Years	Female	Male	Female
Male	Female	Male	Creatine	21.0-53.6	12.8-34.1
21.5-53.9	12.7-34.0	21.9-54.2	12.6-33.9	Creatinine	37.8-85.4
49.9-105.7	38.0-85.5	49.9-105.9	38.3-85.7	49.9-106.0	Guanidinoaceta te
1.3-2.9	1.6-3.2	1.3-2.9	1.6-3.1	1.3-2.9	1.6-3.1
Creatine/ creatinine	< or =0.87	< or =0.43	< or =0.87	< or =0.44	< or =0.88
< or =0.44	Guanidinoaceta te/ creatinine	< or =0.094	< or =0.150	< or =0.093	< or =0.150
< or =0.093	< or =0.150	Guanidinoaceta te/ creatinine	< or =0.037	< or =0.036	< or =0.036
< or =0.035	< or =0.036	< or =0.035		63 Years	64 Years
65 Years	Female	Male	Female	Male	Female
Male	Creatine	22.2-54.6	12.4-33.7	22.3-55.0	12.3-33.6
22.5-55.5	12.2-33.4	Creatinine	38.7-85.8	50.0-106.1	39.1-85.9

50.0-106.3	39.6-86.1	50.1-106.4	Guanidinoacetate	1.3-2.8	1.6-3.1
1.3-2.8	1.6-3.1	1.3-2.8	1.6-3.1	Creatine/creatinine	< or =0.88
< or =0.45	< or =0.89	< or =0.46	< or =0.89	< or =0.46	Guanidinoacetate/ creatine
< or =0.092	< or =0.150	< or =0.092	< or =0.149	< or =0.091	< or =0.149
Guanidinoacetate/ creatinine	< or =0.035	< or =0.035	< or =0.034	< or =0.034	< or =0.034
< or =0.034		66 Years	67 Years	68 Years	Female
Male	Female	Male	Female	Male	Creatine

Interpretation

Reports include concentrations of guanidinoacetate, creatine, and creatinine, and the calculated analyte ratios. When no significant abnormalities are detected, a simple descriptive interpretation is provided. When abnormal results are detected, a detailed interpretation is given. This interpretation includes an overview of the results and their significance, a correlation to available clinical information, elements of differential diagnosis, and recommendations for additional biochemical testing.

Cautions

Creatine supplementation will cause falsely elevated creatine results and falsely decreased guanidinoacetate results. Guanidinoacetate can be elevated in patients with urea cycle defects.

Clinical Reference

1. Clark JF, Cecil KM: Diagnostic methods and recommendations for the cerebral creatine deficiency syndromes. *Pediatr Res*; 2015 Mar;77(3):398-405
2. Mercimek-Mahmutoglu S, Salomons GS: Creatine deficiency syndromes. In: Adam MP, Ardinger HH, Pagon RA, et al. eds. *GeneReviews* [Internet]. University of Washington, Seattle; 2009. Updated December 10, 2015. Accessed December 4, 2019. Available at www.ncbi.nlm.nih.gov/books/NBK3794/
3. Stockler S, Schultz PW, Salomons GS: Cerebral creatine deficiency syndromes: clinical aspects, treatment and pathophysiology. *Subcell Biochem*. 2007;46:149-166
4. Longo N, Ardon O, Vanzo R, et al: Disorders of creatine transport and metabolism. *Am J Med Genet*. 2011;157:72-78. doi: 10.1002/ajmg.c.30292

Performance

Method Description

A serum sample is combined with stable isotope-labeled internal standards and acetonitrile. After centrifugation, an aliquot of this diluted sample is analyzed by injection onto liquid chromatography columns that separate the analytes from the bulk of the stable isotope dilution in the positive electrospray selected reaction monitoring mode using the Applied Biosystems API 3200 liquid chromatography-tandem mass spectrometry (LC-MS/MS) System with Analyst Software. (Bodamer OA, Bloesch SM, Gregg AR, et al: Analysis of guanidinoacetate and creatine by isotope dilution

electroscopy tandem mass spectrometry. Clin Chim Acta. 2001;308:173-178; Cognat S, Cheillan D, Piraud M, et al: Determination of guanidinoacetate and creatine in urine and plasma by liquid chromatography-tandem mass spectrometry. Clin Chem. 2004;50:1459-1461; Sharer JD, Bodamer O, Longo N, Tortorelli S, Wamelink M, Young S: Laboratory diagnosis of creatine deficiency syndromes: a technical standard and guideline of the American College of Medical Genetics and Genomics. Genet Med. 2017 Feb;19[2]:256-263)

PDF Report

No

Specimen Retention Time

1 month

Performing Laboratory Location

Rochester

Fees & Codes
Test Classification

This test was developed, and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the US Food and Drug Administration.

CPT Code Information

82540

82565

82542

LOINC® Information

Test ID	Test Order Name	Order LOINC Value
CRDPS	Creatine Disorders Panel, S	In Process

Result ID	Test Result Name	Result LOINC Value
608077	Interpretation	59462-2
608078	Creatine	15045-8
608079	Creatinine	14682-9
608080	Guanidinoacetate	33244-5
608081	Creatine/Creatinine	In Process
610857	Guanidinoacetate/Creatine	In Process
610858	Guanidinoacetate/Creatinine	In Process
608082	Reviewed By	18771-6