

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 plasma 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

Plasma

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:

Preferred: Lavender top (EDTA)

Acceptable: Green top (sodium heparin), yellow top (ACD)

Submission Container/Tube: Plastic vial

Specimen Volume: 0.5 mL

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

Forms

1. [Biochemical Genetics Patient Information](#) (T602) in Special Instructions
2. [If not ordering electronically, complete, print, and send a Biochemical Genetics Test Request](#) (T798) with the specimen.

Specimen Minimum Volume

0.1 mL

Reject Due To

Gross hemolysis	OK
Gross lipemia	OK
Gross icterus	OK

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Plasma	Frozen (preferred)	14 days	
	Refrigerated	7 days	
	Ambient	72 hours	

Clinical and 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
Creatinine/creatinine	< or =3.07	< or =3.60	< or =3.02	< or =3.54	< or =2.96	< or =3.48
Guanidinoacetate/creatinine	< or =0.040	< or =0.040	< or =0.042	< or =0.040	< or =0.043	< or =0.042



Guani dinoace tate/ creatin ine	< 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	37.1-9 5.5	37.7-95.3	36.0-9 4.4	36.8-94.1	34.6-9 3.2	35.6-92.7
Creatinine	27.6-3 7.9	27.7-36.9	27.6-3 9.3	27.7-38.2	27.6-4 0.9	27.8-39.9
Guanidinoaceta te	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
Guanidinoaceta te/ creatine	< or =0.045	< or =0.043	< or =0.049	< or =0.045	< or =0.053	< or =0.049
Guanidinoaceta te/ creatinine	< or =0.050	< or =0.077	< 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
Guanidinoace tate	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
Guanidinoace tate/ creatine	< or =0.058	< or =0.053	< or =0.063	< or =0.058	< or =0.069	< or =0.064
Guanidinoace tate/ 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
Guanidinoace tate	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
Guanidinoace tate/ creatinine	< or =0.075	< or =0.070	< or =0.081	< or =0.078	< or =0.087	< or =0.085
Guanidinoace tate/ 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
Guanidinoace tate	0.9-2.2	1.0-2.7	1.0-2.3	1.1-2.8	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
Guanidinoace tate/ creatinine	< or =0.092	< or =0.093	< or =0.097	< or =0.101	< or =0.101	< or =0.109
Guanidinoace tate/ 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
Guanidinoace tate	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
Guanidinoace tate/ creatinine	< or =0.104	< or =0.117	< or =0.107	< or =0.125	< or =0.109	< or =0.132
Guanidinoace tate/ 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
Guanidinoace tate	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

Guanidinoacetate/creatinine	< or =0.111	< or =0.139	< or =0.112	< or =0.145	< or =0.113	< or =0.150
Guanidinoacetate/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	Â 32.8-70.2	48.2-87.2	Â 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/creatinine	< 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/creatinine	< 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/creatinine	< 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/creatinine	< 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/creatinine	< 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



Guanidinoacetate/creatinine	< or =0.106	< or =0.169	< or =0.106	< or =0.168	< or =0.106	< or =0.167
Guanidinoacetate/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
Guanidinoacetate	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
Guanidinoacetate/creatinine	< or =0.106	< or =0.166	< or =0.106	< or =0.164	< or =0.105	< or =0.163
Guanidinoacetate/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
Guanidinoacetate	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
Guanidinoacetate/creatinine	< or =0.105	< or =0.161	< or =0.104	< or =0.159	< or =0.103	< or =0.157
Guanidinoacetate/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
Guanidinoacetate	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



Guanidinoacetate/creatinine	< or =0.102	< or =0.155	< or =0.100	< or =0.154	< or =0.099	< or =0.152
Guanidinoacetate/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
Guanidinoacetate	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
Guanidinoacetate/creatinine	< or =0.098	< or =0.151	< or =0.096	< or =0.151	< or =0.095	< or =0.150
Guanidinoacetate/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
Guanidinoacetate	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
Guanidinoacetate/creatinine	< or =0.094	< or =0.150	< or =0.093	< or =0.150	< or =0.093	< or =0.150
Guanidinoacetate/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/creatinine	< 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	22.6-55.9	12.3-33.1	22.7-56.3	12.3-32.8	22.9-56.7	12.5-32.4
Creatinine	40.2-86.2	50.2-106.6	40.7-86.3	50.3-106.7	41.3-86.5	50.4-106.9
Guanidinoacetate	1.3-2.7	1.6-3.1	1.3-2.7	1.6-3.0	1.4-2.7	1.6-3.0
Creatine/creatinine	< or =0.90	< or =0.47	< or =0.90	< or =0.48	< or =0.90	< or =0.48
Guanidinoacetate/creatinine	< or =0.091	< or =0.149	< or =0.090	< or =0.148	< or =0.090	< or =0.148
Guanidinoacetate/creatinine	< or =0.034	< or =0.034	< or =0.033	< or =0.034	< or =0.033	< or =0.034
	69 Years		70 Years		> or = 71 Years	
	Female	Male	Female	Male	Female	Male
Creatine	23.2-57.0	12.8-32.0	23.6-57.2	13.1-31.4	24.1-57.3	13.6-30.8
Creatinine	41.9-86.6	50.5-107.1	42.4-86.8	50.7-107.4	42.8-86.9	50.8-107.6
Guanidinoacetate	1.4-2.6	1.6-3.0	1.4-2.6	1.6-3.0	1.4-2.5	1.5-3.0
Creatine/creatinine	< or =0.90	< or =0.48	< or =0.90	< or =0.48	< or =0.90	< or =0.48
Guanidinoacetate/creatinine	< or =0.090	< or =0.148	< or =0.090	< or =0.148	< or =0.090	< or =0.148
Guanidinoacetate/creatinine	< or =0.033	< or =0.034	< or =0.033	< or =0.034	< or =0.033	< or =0.034

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: Pagon RA, Adam MP, Ardinger HH, et al. eds. *GeneReviews* [Internet]. University of Washington, Seattle; Jan 15 2009. Updated 2015 Dec 10. Accessed 2019 Dec 4. 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 plasma 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 electrospray 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

Day(s) Performed

Tuesday; 12 p.m.

Report Available

2 to 9 days

Specimen Retention Time

1 month

Performing Laboratory Location

Rochester

Fees and Codes

Fees

- Authorized users can sign in to [Test Prices](#) for detailed fee information.
- Clients without access to Test Prices can contact [Customer Service](#) 24 hours a day, seven days a week.

- Prospective clients should contact their Regional Manager. For assistance, contact [Customer Service](#).

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
CRDPP	Creatine Disorders Panel, P	In Process

Result ID	Test Result Name	Result LOINC Value
608071	Interpretation	59462-2
608072	Creatine	15045-8
608073	Creatinine	14682-9
608074	Guanidinoacetate	33244-5
608075	Creatine/Creatinine	In Process
610624	Guanidinoacetate/Creatine	In Process
610856	Guanidinoacetate/Creatinine	In Process
608076	Reviewed By	18771-6