MAYO CLINIC LABORATORIES

Test Definition: CRDPS

Creatine Disorders Panel, Serum

Overview

Useful For

Evaluating patients with a clinical suspicion of arginine:glycine amidinotransferase deficiency,guanidinoacetate methyltransferase deficiency, and creatine transporter deficiency using serum specimens

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, aids in distinguishing 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 Screen Follow-up for Guanidinoacetate Methyltransferase Deficiency

-Epilepsy: Unexplained Refractory and/or Familial Testing Algorithm

If the patient has abnormal newborn screening results for guanidinoacetate methyltransferase deficiency, refer to the appropriate ACMG Newborn Screening ACT Sheet.(1)

Special Instructions

- Newborn Screen Follow-up for Guanidinoacetate Methyltransferase Deficiency
- 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 additional diagnostic testing, consider ordering 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

Supplies: Sarstedt Aliquot Tube 5 mL (T914)
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.

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
Serum	Frozen (preferred)	14 days	
	Ambient	72 hours	
	Refrigerated	7 days	

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, especially expressive speech and language delay and intellectual disability. 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 may develop seizures, autistic-like behaviors, and 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. Patients with CTD may have normal or abnormal serum levels of GAA, Cr and Crn, and measurement of these analytes in urine is also useful for diagnosis in male patients (characteristic findings are elevated Cr, normal to low Crn, and an elevated Cr:Crn ratio in urine). The only consistently reliable method for diagnosis of CTD in female patients is molecular analysis of the *SLC6A8* gene. The diagnosis of GAMT,AGAT, and CTD can be confirmed by molecular analysis of *GAMT,GATM,* and *SLC6A8* respectively.

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. CTD is treated with oral creatine monohydrate and arginine and glycine supplementation.

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 (creatine, 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 / creatine	< or =0.045	< or =0.043	< or =0.049	< or =0.045	< or =0.053	< or =0.049

Guanidinoacetate / 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
Guanidinoacetate	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
Guanidinoacetate / creatine	< or =0.058	< or =0.053	< or =0.063	< or =0.058	< or =0.069	< or =0.064
Guanidinoacetate / 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
Guanidinoacetate	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
Guanidinoacetate / creatine	< or =0.075	< or =0.070	< or =0.081	< or =0.078	< or =0.087	< or =0.085
Guanidinoacetate / 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
Guanidinoacetate	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
Guanidinoacetate / creatine	< or =0.092	< or =0.093	< or =0.097	< or =0.101	< or =0.101	< or =0.109
Guanidinoacetate /	< or =0.044	< or =0.053	< or =0.043	< or =0.051	< or =0.042	< or =0.050

creatinine						

	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
Guanidinoacetate	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
Guanidinoacetate / creatine	< or =0.104	< or =0.117	< or =0.107	< or =0.125	< or =0.109	< or =0.132
Guanidinoacetate / 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
Guanidinoacetate	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 / creatine	< 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	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 / creatine	< 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 / creatine	< 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 / creatine	< 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
Guanidinoacetate / creatine	< 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 / creatine	< 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/	< or =0.82	< or =0.37	< or =0.82	< or =0.37	< or =0.82	< or =0.38

creatinine						
Guanidinoacetate / creatine	< 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 / creatine	< 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 / creatine	< 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	< or =0.094	< or =0.150	< or =0.093	< or =0.150	< or =0.093	< or =0.150

/ creatine						
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 / 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	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 / creatine	< 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 / creatine	< 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. ACMG Newborn Screening ACT Sheets. Accessed December 16, 2024. Available at www.acmg.net/ACMG/Medical-Genetics-Practice-Resources/ACT_Sheets_and_Algorithms/ACMG/Medical-Genetics-Practice-Resources/ACT_Sheets_and_Algorithms.aspx?hkey=9d6bce5a-182e-42a6-84a5-b2d88240c508
2. Sanders K, Peck D, Bentz Pino G, et al. SLC6A8 creatine transporter deficiency can be detected by plasma creatine and creatinine concentrations. Mol Genet Metab. 2024;142(1):108455. doi:10.1016/j.ymgme.2024.108455
3.Clark JF, Cecil KM. Diagnostic methods and recommendations for the cerebral creatine deficiency syndromes. Pediatr Res. 2015;77(3):398-405
4. Mercimek-Mahmutoglu S, Salomons GS. Creatine deficiency syndromes. In: Adam MP, Mirzaa GM, Pagon RA, et al. eds. GeneReviews [Internet]. University of Washington, Seattle; 2009. Updated February 10, 2022. Accessed December 16, 2024. Available at www.ncbi.nlm.nih.gov/books/NBK3794/
5. Stockler S, Schultz PW, Salomons GS. Cerebral creatine deficiency syndromes: clinical aspects, treatment, and pathophysiology. Subcell Biochem. 2007;46:149-166
6. Longo N, Ardon O, Vanzo R, Schwartz E, Pasquali M. Disorders of creatine transport and metabolism. Am J Med Genet. 2011;157:72-78. doi:10.1002/ajmg.c.30292
7. Fernandes-Pires G, Braissant O. Current and potential new treatment strategies for creatine deficiency syndromes. Mol Genet Metab. 2022;135(1):15-26. doi:10.1016/j.ymgme.2021.12.005

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 system with Analyst software.(Bodamer OA, Bloesch SM, Gregg AR, Stockler-Ipsiroglue S, O'Brien WEO. 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,

Roos B, Jakobs C, Vianey-Saban C. Determination of guanidinoacetate and creatine in urine and plasma by liquid chromatography-tandem mass spectrometry. Clin Chem. 2004;50[8]: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;19[2]:256-263)

PDF Report
No

Day(s) Performed
Tuesday

Report Available
3 to 9 days

Specimen Retention Time
1 month

Performing Laboratory Location
Mayo Clinic Laboratories - Rochester Main Campus

Fees & 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 account representative. 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. It 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