

Disease

Cerebral creatine deficiency syndrome 2¹

Gene

GAMT

Inheritance	AR
Current Treatments	creatine monohydrate and ornithine supplementation. Arginine restriction.
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	guanidinoacetate, creatine, and creatinine levels in urine and plasma ¹
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=creatine+deficiency&term=&cntry=&state=&city=&dist=

References

¹ <https://www.ncbi.nlm.nih.gov/books/NBK3794/>

2