

Variant: *NM_000156.6(GAMT):c.279C>T (p.Asp93=)*

Version: 1.0

CA291015 [↗](#)

137434 (ClinVar) [↗](#)

Gene: GAMT ([HGNC:2593](#))

Condition: guanidinoacetate methyltransferase deficiency
([MONDO:0012999](#))

Inheritance Mode: Autosomal recessive inheritance

UUID: 3ce763f1-6ff6-44c9-8764-99f198f14dc1

Approved on: 2022/6/6

Published on: 2022/10/7

HGVS expressions

NM_000156.6(GAMT):c.279C>T (p.Asp93=)

NM_000156.6:c.279C>T

NC_000019.10:g.1399841G>A

CM000681.2:g.1399841G>A

NC_000019.9:g.1399840G>A

CM000681.1:g.1399840G>A

NC_000019.8:g.1350840G>A

NG_009785.1:g.6713C>T

ENST00000252288.8:c.279C>T

ENST00000447102.8:c.279C>T

ENST00000640762.1:c.210C>T

ENST00000252288.6:c.279C>T

ENST00000447102.7:c.279C>T

NM_000156.5:c.279C>T

NM_138924.2:c.279C>T

NM_138924.3:c.279C>T

Benign

Met criteria codes **3**

BP7

BP4

BA1

Evidence Links **0**

Expert Panel

Cerebral Creatine Deficiency Syndromes VCEP [↗](#)

Criteria Specification Information

[↗](#) **Criteria Specification:** ClinGen Cerebral Creatine Deficiency Syndromes Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for GAMT Version 1

[↗](#) **Criteria Specification Approval History**

[↗](#) **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

Cerebral Creatine Deficiency Syndromes VCEP

The NM_000156.6:c.279C>T(p.Asp93=) is a synonymous variant in GAMT that is predicted to not impact splicing by SpliceAI and VarSeak, and the nucleotide is not highly conserved (BP4, BP7). The highest population minor allele frequency in gnomAD v2.1.1 is 0.00484 (87/17976 alleles) in the East Asian population, which is higher than the ClinGen CCDS VCEP’s threshold for BA1 (>0.003), and therefore meets this criterion (BA1). This variant does not appear to have been previously reported in the published literature. It is noted in ClinVar (Variation ID 137434). In summary, this variant meets the criteria to be classified as benign for GAMT deficiency. GAMT-specific ACMG/AMP codes met, as specified by the ClinGen CCDS VCEP (Specifications Version 1.1.0): BA1, BP4, BP7. (Classification approved by the ClinGen CCDS VCEP on June 6, 2022).

Met criteria codes

BP7			It is predicted to not impact splicing by SpliceAI and VarSeak, and the nucleotide is not highly conserved (GERP rejected substitutions (RS) score -7.09) (BP7).
BP4			This variant is predicted to not impact splicing by SpliceAI and VarSeak BP4).
BA1			The highest population minor allele frequency in gnomAD v2.1.1 is 0.00484 (87/17976 alleles) in the East Asian population, which is higher than the ClinGen CCDS VCEP’s threshold for BA1 (>0.003), and therefore meets this criterion (BA1).

Curation History



 

 

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