

Variant: *NM_000152.5(GAA):c.1332T>C (p.Pro444=)*

Version: 1.1

CA8815288 [↗](#)

286018 (ClinVar) [↗](#)

Gene: GAA ([HGNC:2548](#))

Condition: glycogen storage disease II ([MONDO:0009290](#))

Inheritance Mode: Autosomal recessive inheritance

UUID: 6677c5dc-80b2-4ac1-b197-6c8a76d3a6a9

Approved on: 2023/3/13

Published on: 2023/3/13

HGVS expressions

NM_000152.5:c.1332T>C

NM_000152.5(GAA):c.1332T>C (p.Pro444=)

NC_000017.11:g.80109950T>C

CM000679.2:g.80109950T>C

NC_000017.10:g.78083749T>C

CM000679.1:g.78083749T>C

NC_000017.9:g.75698344T>C

NG_009822.1:g.13395T>C

ENST00000570803.6:c.1332T>C

ENST00000572080.2:c.1332T>C

ENST00000577106.6:c.1332T>C

ENST00000302262.8:c.1332T>C

ENST00000302262.7:c.1332T>C

ENST00000390015.7:c.1332T>C

NM_000152.3:c.1332T>C

NM_001079803.1:c.1332T>C

NM_001079804.1:c.1332T>C

NM_000152.4:c.1332T>C

NM_001079803.2:c.1332T>C

NM_001079804.2:c.1332T>C

NM_001079803.3:c.1332T>C

NM_001079804.3:c.1332T>C

Likely Benign

Met criteria codes **3**

BS1 BP7 BP4

Evidence Links **0**

Expert Panel

[Lysosomal Diseases VCEP](#) [↗](#)

Criteria Specification Information

[↗](#) **Criteria Specification:** *ClinGen Lysosomal Storage Disorders Variant Curation Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines Version 2*

[↗](#) PDF

[↗](#) Criteria Specification Approval History

[↗](#) Criteria Specifications for this VCEP

Lysosomal Diseases VCEP

The NM_000152.5:c.1332T>C (p.Pro444=) variant in GAA is a synonymous (silent) variant that is not predicted to impact splicing; the nucleotide is not highly conserved (PhyloP 100 way score is -2.4 (BP4, BP7). There is a ClinVar entry for this variant (Variation ID: 286018). In summary, this variant meets the criteria to be classified as likely benign for Pompe disease. GAA-specific ACMG/AMP criteria applied, as specified by the ClinGen Lysosomal Diseases VCEP (Specifications Version 2.0): BS1, BP4, BP7. (Classification approved by the ClinGen Lysosomal Diseases VCEP, March 13, 2023).

Met criteria codes

BS1			The highest population minor allele frequency in gnomAD v2.1.1 is 0.00859 (171/19912 alleles; including one homozygote) in the East Asian population, which is higher than the ClinGen Lysosomal Diseases VCEP threshold (>0.005) for BS1, and meets this criterion.
BP7			The NM_000152.5:c.1332T>C (p.Pro444=) variant in GAA is a synonymous (silent) variant that is not predicted to impact splicing; the nucleotide is not highly conserved (PhyloP 100 way score is -2.4 (BP7).
BP4			The computational splicing predictor SpliceAI suggests that the variant has no impact on splicing (BP4).

Curation History



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