

Variant: *NM_000152.5(GAA):c.852G>A (p.Ala284=)*

Version: 1.2

CA198776 [↗](#)

188477 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: d791b5fc-8883-4686-9ee0-24599175ad75

Approved on: 2020/1/23

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HGVS expressions

NM_000152.5:c.852G>A

NM_000152.5(GAA):c.852G>A (p.Ala284=)

NC_000017.11:g.80107716G>A

CM000679.2:g.80107716G>A

NC_000017.10:g.78081515G>A

CM000679.1:g.78081515G>A

NC_000017.9:g.75696110G>A

NG_009822.1:g.11161G>A

ENST00000570803.6:c.852G>A

ENST00000572080.2:c.852G>A

ENST00000577106.6:c.852G>A

ENST00000302262.8:c.852G>A

ENST00000302262.7:c.852G>A

ENST00000390015.7:c.852G>A

ENST00000570803.5:c.852G>A

NM_000152.3:c.852G>A

NM_001079803.1:c.852G>A

NM_001079804.1:c.852G>A

NM_000152.4:c.852G>A

NM_001079803.2:c.852G>A

NM_001079804.2:c.852G>A

NM_001079803.3:c.852G>A

NM_001079804.3:c.852G>A

Benign

Met criteria codes **1**

BA1

Evidence Links **0**

Expert Panel

[Lysosomal Diseases VCEP](#) [↗](#)

Criteria Specification Information **!**

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

Evidence submitted by expert panel

Lysosomal Diseases VCEP

The highest continental population minor allele frequency for c.852G>A (p.Ala284=) in gnomAD v2.1.1 is 0.01017 in the European non-Finnish population. This is higher than the ClinGen LSD VCEP’s BA1 threshold (>0.01), meeting this criterion. There is a ClinVar entry for this variant (Variation ID: 188477, two star review status), with 5 submitters classifying the variant as benign. In summary, this variant meets the criteria to be classified as benign for Pompe disease. GAA-specific ACMG/AMP criteria applied, as specified by the ClinGen LSD VCEP: BA1.

Met criteria codes

BA1			The highest population minor allele frequency in gnomAD v2.1.1 is 0.01017 (European non-Finnish). This is higher than the ClinGen LSD VCEP threshold (>0.01) for BA1. Therefore, the allele frequency data for this variant meet the BA1 criterion.
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Curation History



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