

Variant: NM_000152.5(GAA):c.1100G>A (p.Trp367Ter)

Version: 1.0

CA401364953 [↗](#)

972773 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 86a773b1-d072-4569-8580-1ef8dd204b12

Approved on: 2026/4/6

Published on: 2026/5/14

HGVS expressions

NM_000152.5:c.1100G>A

NM_000152.5(GAA):c.1100G>A (p.Trp367Ter)

NC_000017.11:g.80108513G>A

CM000679.2:g.80108513G>A

NC_000017.10:g.78082312G>A

CM000679.1:g.78082312G>A

NC_000017.9:g.75696907G>A

NG_009822.1:g.11958G>A

ENST00000570803.6:c.1100G>A

ENST00000572080.2:c.1100G>A

ENST00000577106.6:c.1100G>A

ENST00000302262.8:c.1100G>A

ENST00000302262.7:c.1100G>A

ENST00000390015.7:c.1100G>A

NM_000152.3:c.1100G>A

NM_001079803.1:c.1100G>A

NM_001079804.1:c.1100G>A

NM_000152.4:c.1100G>A

NM_001079803.2:c.1100G>A

NM_001079804.2:c.1100G>A

NM_001079803.3:c.1100G>A

NM_001079804.3:c.1100G>A

Pathogenic

Met criteria codes **3**

PVS1 **PM3_Supporting**

PM2_Supporting

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.1100G>A (p. Trp367Ter) variant in GAA is a nonsense variant predicted to cause a premature stop codon in biologically-relevant-exon 7/20, leading to nonsense mediated decay in a gene in which loss-of-function is an established disease mechanism (PVS1). One patient with this variant has been reported with childhood onset Pompe disease. GAA enzyme activity level for this patient was reported as 8.9% of normal, but the tissue source was not stated. This patient is compound heterozygous for the c.1100G>A (p. Trp367Ter) variant and a variant in GAA that has been classified as pathogenic by the ClinGen Lysosomal Diseases VCEP, c.-32-13T>G. The phase of the variants is unknown (PM3_Supporting). While this patient was diagnosed with childhood onset Pompe disease with a stated GAA enzyme activity of 8.9%, the tissue source was not indicated. As a result, PP4 was not applied (PMID: 17723315). The variant is absent from gnomAD v4.1.0. There is a ClinVar entry for this variant (Variation ID: 972773; 2 star review status) with two submitters classifying the variant as pathogenic. In summary, the variant meets the criteria to be classified as pathogenic for Pompe disease by the ClinGen Lysosomal Diseases Variant Curation Expert Panel. GAA-specific ACMG/AMP criteria applied (Specifications Version 2.0.0): PVS1, PM2_Supporting, PM3_Supporting. (Classification approved by the ClinGen Lysosomal Diseases Variant Curation Expert Panel on April 6, 2026)

Met criteria codes

PVS1			The NM_000152.5:c.1100G>A (p. Trp367Ter) variant in GAA is a nonsense variant predicted to cause a premature stop codon in biologically-relevant-exon 7 of 20, leading to nonsense mediated decay in a gene in which loss-of-function is an established disease mechanism (PVS1).
PM3_Supporting			One patient is compound heterozygous for the variant and a pathogenic variant (c.-32-13T>G PMID: 17723315), phase unconfirmed (0.5 points)(PM3_supporting)
PM2_Supporting			This variant is absent in gnomAD v4.1.0 (PM2_Supporting).

Curation History

▼

▼

Showing 1 to 1 of 1 rows

--

