The ClinGen Evidence Repository is an **FDA-recognized human genetic variant database** containing expert-curated assertions regarding variants' pathogenicity and supporting evidence summaries. [Disclaimer]

Variant: NM_000152.5(GAA):c.2061del (p.Phe687fs)

Version: 1.2

CA16620648

423932 (ClinVar)

Gene: GAA (HGNC:2548)
Condition: glycogen storage disease II (MONDO:0009290)
Inheritance Mode: Autosomal recessive inheritance
UUID: 85b16715-d707-4bdf-b668-afbb2ad54177
Approved on: 2026/4/9
Published on: 2026/4/9

HGVS expressions

NM_000152.5:c.2061del
NM_000152.5(GAA):c.2061del (p.Phe687fs)
NC_000017.11:g.80113238del
CM000679.2:g.80113238del
NC_000017.10:g.78087037del
CM000679.1:g.78087037del
NC_000017.9:g.75701632del
NG_009822.1:g.16683del
ENST00000570803.6:c.2061del
ENST00000572080.2:c.*199del
ENST00000577106.6:c.2061del
ENST00000302262.8:c.2061del
ENST00000302262.7:c.2061del
ENST00000390015.7:c.2061del
ENST00000570716.1:n.501del
ENST00000572080.1:c.480del
NM_000152.3:c.2061del
NM_001079803.1:c.2061del
NM_001079804.1:c.2061del
NM_000152.4:c.2061del
NM_001079803.2:c.2061del
NM_001079804.2:c.2061del
NM_001079803.3:c.2061del
NM_001079804.3:c.2061del

Likely Pathogenic

Met criteria codes 2

PVS1 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

Evidence submitted by expert panel

Lysosomal Diseases VCEP

The NM_000152.5:c.2061del (p.Phe687LeufsTer9) variant in GAA is a frameshift variant predicted to cause a premature stop codon in exon 15 out of a total of 20 exons, leading to nonsense mediated decay in a gene in which loss-of-function is an established disease mechanism (PVS1). The variant is absent in gnomAD v4.1.0. (PM2_Supporting). To our knowledge, this variant has not been reported in the literature in individuals with Pompe disease, and results of experimental studies are not available. There is a ClinVar entry for this variant (Variation ID: 423932). The classification of this variant has been upgraded from Variant of Uncertain Significance to Likely Pathogenic based on the recommendations of the ClinGen Sequence Variant Interpretation Working Group, that a variant meeting PVS1 and PM2_Supporting is classified as Likely Pathogenic (https://clinicalgenome.org/site/assets/files/5182/pm2_-_svi_recommendation_-_approved_sept2020.pdf). In summary, this variant meets the criteria to be classified as Likely Pathogenic for Pompe disease. GAA-specific ACMG/AMP criteria met, based on the specifications of the ClinGen Lysosomal Diseases VCEP (Specifications Version 2.0): PVS1, PM2_Supporting. (Classification approved by the ClinGen Lysosomal Diseases VCEP, April 9, 2026).

Met criteria codes

PVS1	✔	The NM_000152.5:c.2061del (p.Phe687LeufsTer9) variant in GAA is a frameshift variant predicted to cause a premature stop codon in exon 15 out of a total of 20 exons, leading to nonsense mediated decay in a gene in which loss-of-function is an established disease mechanism (PVS1).
PM2_Supporting	✔	This variant is absent in gnomAD v4.1.0. (PM2_Supporting).

Curation History

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