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.55del (p.Val19fs)

Version: 1.2

CA658824772

555040 (ClinVar)

Gene: GAA (HGNC:2548)
Condition: glycogen storage disease II (MONDO:0009290)
Inheritance Mode: Autosomal recessive inheritance
UUID: 7be90f98-3deb-4709-a72e-f2671b28e7ea
Approved on: 2026/7/21
Published on: 2026/7/21

HGVS expressions

NM_000152.5(GAA):c.55del (p.Val19fs)
NM_000152.5:c.55del
NC_000017.11:g.80104641del
CM000679.2:g.80104641del
NC_000017.10:g.78078440del
CM000679.1:g.78078440del
NC_000017.9:g.75693035del
NG_009822.1:g.8086del
ENST00000570803.6:c.55del
ENST00000572080.2:c.55del
ENST00000577106.6:c.55del
ENST00000302262.8:c.55del
ENST00000302262.7:c.55del
ENST00000390015.7:c.55del
ENST00000570803.5:c.55del
ENST00000577106.5:c.55del
NM_000152.3:c.55del
NM_001079803.1:c.55del
NM_001079804.1:c.55del
NM_000152.4:c.55del
NM_001079803.2:c.55del
NM_001079804.2:c.55del
NM_001079803.3:c.55del
NM_001079804.3:c.55del

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 2.0*
PDF

Criteria Specification Approval History
Criteria Specifications for this VCEP

Evidence submitted by expert panel

Lysosomal Diseases VCEP

The NM_000152.5:c.55del (p.Val19CysfsTer24) variant in GAA is a frameshift variant predicted to cause a premature stop codon, leading to nonsense mediated decay in a gene in which loss-of-function is an established disease mechanism (PVS1). This 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: 555040. 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, July 21, 2026).

Met criteria codes

PVS1	✔	The NM_000152.5:c.55del (p.Val19CysfsTer24) variant in GAA is a frameshift variant predicted to cause a premature stop codon, 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

Showing 1 to 3 of 3 rows

The information on this website is not intended for direct diagnostic use or medical decision-making without review by a genetics professional. Individuals should not change their health behavior solely on the basis of information contained on this website. If you have questions about the information contained on this website, please see a health care professional.

ClinGen Terms of Use.

¤ Powered by BCM's Genboree.