

Variant: NM\_000152.5(GAA):c.2400C>T (p.Ser800=)

Version: 1.0

CA180077 [↗](#)

167115 (ClinVar) [↗](#)

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

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

**Inheritance Mode:** Autosomal recessive inheritance

**UUID:** f1b0a8d8-3cc5-4dc5-9a2a-21e4715f92e1

**Approved on:** 2024/8/6

**Published on:** 2024/9/3

## HGVS expressions

**NM\_000152.5(GAA):c.2400C>T (p.Ser800=)**

**NM\_000152.5:c.2400C>T**

NC\_000017.11:g.80117668C>T

CM000679.2:g.80117668C>T

NC\_000017.10:g.78091467C>T

CM000679.1:g.78091467C>T

NC\_000017.9:g.75706062C>T

NG\_009822.1:g.21113C>T

ENST00000570803.6:c.2400C>T

ENST00000572080.2:c.\*538C>T

ENST00000577106.6:c.2400C>T

ENST00000302262.8:c.2400C>T

ENST00000302262.7:c.2400C>T

ENST00000390015.7:c.2400C>T

ENST00000573556.1:n.353C>T

NM\_000152.3:c.2400C>T

NM\_001079803.1:c.2400C>T

NM\_001079804.1:c.2400C>T

NM\_000152.4:c.2400C>T

NM\_001079803.2:c.2400C>T

NM\_001079804.2:c.2400C>T

NM\_001079803.3:c.2400C>T

NM\_001079804.3:c.2400C>T

**Benign**

**Met criteria codes** 1

**BA1**

**Not Met criteria codes** 1

**PM2**

**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.2400C>T variant in GAA is a synonymous (silent) variant (p.Ser800=) that is not predicted to impact splicing. The highest population minor allele frequency in gnomAD v2.1.1. is 0.02504 (617/24640 alleles; 9 homozygotes) in the African/African American population, which is higher than the ClinGen Lysosomal Diseases VCEP’s threshold for BA1 (>0.01), and therefore meets this criterion (BA1). There is a ClinVar entry for this variant (Variation ID: 167115. 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 Lysosomal Diseases Variant Curation Expert Panel (specifications Version 2.0): BA1. (Classification approved by the ClinGen Lysosomal Diseases Variant Curation Expert Panel on August 6, 2024).

**Met criteria codes**

|            |                                                                                                                                                                     |                                                                                                                                                                                                                                                                                           |
|------------|---------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>BA1</b> |   | The highest population minor allele frequency in gnomAD v2.1.1. is 0.02504 (617/24640 alleles; 9 homozygotes) in the African/African American population, which is higher than the ClinGen Lysosomal Diseases VCEP’s threshold for BA1 (>0.01), and therefore meets this criterion (BA1). |
|------------|---------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|

**Not Met criteria codes**

|            |                                                                                                                                                                       |                                                                                                                            |
|------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------------------------------------------------------------------------------------------------------------------|
| <b>PM2</b> |   | No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline |
|------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------------------------------------------------------------------------------------------------------------------|

Curation History 



 

 

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