

Variant: *NM_000337.6(SGCD):c.91C>T (p.Arg31Trp)*

Version: 1.0

CA3530486 [↗](#)

652862 (ClinVar) [↗](#)

Gene: SGCD (HGNC:6444)

Condition: autosomal recessive limb-girdle muscular dystrophy (MONDO:0015152)

Inheritance Mode: Autosomal recessive inheritance

UUID: 6d79dee2-9e4a-45cc-b64b-3a04b1690f9f

Approved on: 2025-01-08

Published on: 2025-01-08

HGVS expressions

NM_000337.6:c.91C>T

NM_000337.6(SGCD):c.91C>T (p.Arg31Trp)

NC_000005.10:g.156344576C>T

CM000667.2:g.156344576C>T

NC_000005.9:g.155771586C>T

CM000667.1:g.155771586C>T

NC_000005.8:g.155704164C>T

NG_008693.2:g.479233C>T

ENST00000337851.9:c.91C>T

ENST00000337851.8:c.91C>T

ENST00000435422.7:c.88C>T

ENST00000517913.5:c.91C>T

ENST00000524347.2:c.91C>T

NM_000337.5:c.91C>T

NM_001128209.1:c.88C>T

NM_172244.2:c.91C>T

NM_001128209.2:c.88C>T

NM_172244.3:c.91C>T

Uncertain Significance

Met criteria codes **1**

PP3

Not Met criteria codes **25**

PS1 PS2 PS3 PS4 PP1 PP2

PP4 PVS1 PM1 PM3 PM5

PM4 PM6 PM2 BA1 BS1

BS4 BS3 BS2 BP5 BP7 BP4

BP3 BP1 BP2

Evidence Links **0**

Expert Panel

Limb Girdle Muscular Dystrophy VCEP [↗](#)

Criteria Specification Information

[↗](#) **Criteria Specification:** ClinGen Limb Girdle Muscular Dystrophy Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for SGCD Version 1.0.0

[↗](#) **Criteria Specification Approval History**



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

Evidence submitted by expert panel



Limb Girdle Muscular Dystrophy VCEP


The NM_000337.6: c.91C>T variant in SGCD is a missense variant predicted to cause substitution of arginine by tryptophan at amino acid 31 (p.Arg31Trp). The highest minor allele frequency for this variant is 0.00009898 in the South Asian population of gnomAD v2.1.1 (3/30310 exome chromosomes), which is higher than the LGMD VCEP threshold for PM2_Supporting (0.00009) (criterion not met). The computational predictor REVEL gives a score of 0.835, which is above the threshold of ≥ 0.70 , evidence that correlates with impact to SGCD function (PP3). In summary, at this time there is insufficient evidence to classify this variant as pathogenic or benign, and it remains a variant of uncertain significance for autosomal recessive limb girdle muscular dystrophy based on the ACMG/AMP criteria applied, as specified by the ClinGen LGMD VCEP (LGMD VCEP specifications version 1.0.0; 01/08/2025): PP3.



Met criteria codes



PP3			The computational predictor REVEL gives a score of 0.835, which is above the threshold of ≥ 0.70 , evidence that correlates with impact to SGCD function (PP3).
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Not Met criteria codes


PS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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PS2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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PS3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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PS4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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PP1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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PP2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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PP4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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PVS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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PM1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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PM3			
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No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

PM5			c.91C>G (p.Arg31Gly) = LB c.92G>A (p.Arg31Gln) = VUS
PM4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM6			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM2			The highest minor allele frequency for this variant is 0.00009898 in the South Asian population of gnomAD v2.1.1 (3/30310 exome chromosomes), which is higher than the LGMD VCEP threshold for PM2_Supporting (0.00009) (criterion not met).
BA1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP5			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP7			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

BP2



No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History [↗](#)

Showing 1 to 1 of 1 rows

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