

Variant: *NM_000337.6(SGCD):c.289C>T (p.Arg97Ter)*

Version: 1.0

CA3530549 [↗](#)

1677453 (ClinVar) [↗](#)

Gene: SGCD ([HGNC:6444](#))

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

Inheritance Mode: Autosomal recessive inheritance

UUID: ce1b25c2-4939-4be5-a164-23a81edf121e

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HGVS expressions

NM_000337.6(SGCD):c.289C>T (p.Arg97Ter)

NM_000337.6:c.289C>T

NC_000005.10:g.156508697C>T

CM000667.2:g.156508697C>T

NC_000005.9:g.155935707C>T

CM000667.1:g.155935707C>T

NC_000005.8:g.155868285C>T

NG_008693.2:g.643354C>T

ENST00000337851.9:c.289C>T

ENST00000337851.8:c.289C>T

ENST00000435422.7:c.286C>T

ENST00000517913.5:c.289C>T

ENST00000524347.2:c.*153C>T

NM_000337.5:c.289C>T

NM_001128209.1:c.286C>T

NM_172244.2:c.289C>T

NM_001128209.2:c.286C>T

NM_172244.3:c.289C>T

Pathogenic

Met criteria codes **4**

PM3_Supporting

PM2_Supporting

PP4

PVS1

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.289C>T p.(Arg97Ter) variant in SGCD is a nonsense variant predicted to cause a premature stop codon in biologically relevant exon 4/9, leading to nonsense mediated decay in a gene in which loss of function is an established disease mechanism (PVS1). This variant has been identified in a homozygous state in at least two patients with LGMD who were from consanguineous families (0.5 pts, PMID: 34515763, 30733730; PM3_Supporting). At least one of these patients displayed progressive limb girdle weakness observed over more than six months (PP4). The highest population minor allele frequency in gnomAD v4.1.0 is 0.00001146 (1/87260 alleles) in the South Asian population, which is lower than the ClinGen LGMD VCEP threshold (<0.00009) for PM2_Supporting, meeting this criterion (PM2_Supporting). In summary, this variant meets the criteria to be classified as Pathogenic 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): PVS1, PM3_Supporting, PP4, PM2_Supporting.

Met criteria codes

PM3_Supporting			This variant has been identified in a homozygous state in at least two patients with LGMD who were from consanguineous families (0.5 pts, PMID: 34515763, 30733730; PM3_Supporting).
PM2_Supporting			The highest population minor allele frequency in gnomAD v4.1.0 is 0.00001146 (1/87260 alleles) in the South Asian population, which is lower than the ClinGen LGMD VCEP threshold (<0.00009) for PM2_Supporting, meeting this criterion (PM2_Supporting).
PP4			At least one patient with this variant and a second variant in SGCD displayed progressive limb girdle weakness observed over more than six months (PP4; PMID: 34515763, 30733730).
PVS1			The NM_000337.6: c.289C>T p.(Arg97Ter) variant in SGCD is a nonsense variant predicted to cause a premature stop codon in biologically relevant exon 4/9, leading to nonsense mediated decay in a gene in which loss of function is an established disease mechanism (PVS1).

Curation History



 

 

Showing 1 to 1 of 1 rows

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