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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
Approved on: 2025/1/8
Published on: 2025/1/8

HGVS expressions

NM_000337.6:c.289C>T
NM_000337.6(SGCD):c.289C>T (p.Arg97Ter)
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 | PP4 | PVS1 | PM2_Supporting

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).
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).
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).

Curation History

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