

Variant: NM_001130987.2(DYSF):c.5210C>G (p.Pro1737Arg)

Version: 1.0

CA1707292 [↗](#)

567124 (ClinVar) [↗](#)

Gene: DYSF ([HGNC:8291](#))

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

Inheritance Mode: Autosomal recessive inheritance

UUID: d8445323-1604-42aa-a076-e8e266797546

Approved on: 2026/1/23

Published on: 2026/2/17

HGVS expressions

NM_001130987.2(DYSF):c.5210C>G (p.Pro1737Arg)

NM_001130987.2:c.5210C>G

NC_000002.12:g.71665197C>G

CM000664.2:g.71665197C>G

NC_000002.11:g.71892327C>G

CM000664.1:g.71892327C>G

NC_000002.10:g.71745835C>G

NG_008694.1:g.216575C>G

ENST00000698057.1:c.2624C>G

ENST00000698058.1:c.1841C>G

ENST00000698059.1:c.1949C>G

ENST00000258104.8:c.5093C>G

ENST00000410020.8:c.5210C>G

ENST00000258104.7:c.5093C>G

ENST00000394120.6:c.5096C>G

ENST00000409366.5:c.5159C>G

ENST00000409582.7:c.5207C>G

ENST00000409651.5:c.5189C>G

ENST00000409744.5:c.5117C>G

ENST00000409762.5:c.5144C>G

ENST00000410020.7:c.5210C>G

ENST00000410041.1:c.5147C>G

ENST00000413539.6:c.5186C>G

ENST00000429174.6:c.5156C>G

ENST00000479049.6:n.1978C>G

NM_001130455.1:c.5096C>G

NM_001130976.1:c.5051C>G

NM_001130977.1:c.5114C>G

NM_001130978.1:c.5156C>G

NM_001130979.1:c.5186C>G

NM_001130980.1:c.5144C>G

NM_001130981.1:c.5207C>G

NM_001130982.1:c.5189C>G

NM_001130983.1:c.5159C>G

NM_001130984.1:c.5117C>G

NM_001130985.1:c.5147C>G

NM_001130986.1:c.5054C>G

NM_001130987.1:c.5210C>G
NM_003494.3:c.5093C>G
NM_001130455.2:c.5096C>G
NM_001130976.2:c.5051C>G
NM_001130977.2:c.5114C>G
NM_001130978.2:c.5156C>G
NM_001130979.2:c.5186C>G
NM_001130980.2:c.5144C>G
NM_001130981.2:c.5207C>G
NM_001130982.2:c.5189C>G
NM_001130983.2:c.5159C>G
NM_001130984.2:c.5117C>G
NM_001130985.2:c.5147C>G
NM_001130986.2:c.5054C>G
NM_003494.4:c.5093C>G

Likely Pathogenic

Met criteria codes 6

- PM3_Supporting
- PM2_Supporting
- PM5_Supporting
- PP1
- PP3
- PP4

Not Met criteria codes 1

PS3

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 DYSF Version 2.0.0
- [Criteria Specification Approval History](#)
- [Criteria Specifications for this VCEP](#)

Evidence submitted by expert panel

Limb Girdle Muscular Dystrophy VCEP

The NM_003494.4: c.5093C>G variant in DYSF, which is also known as NM_001130987.2: c.5210C>G p.(Pro1737Arg), is a missense variant predicted to cause substitution of proline to arginine at amino acid 1698, p.(Pro1698Arg). This variant has been observed in three individuals with LGMD, including two siblings in a homozygous state with reported consanguinity (0.25 pts, ClinVar SCV000814671.7 internal data communication; PP1) and one with a pathogenic variant in unknown phase (NM_003494.4: c.265C>T p.(Arg89Ter), 0.5 pts, PMID: 38582400) (PM3_Supporting; PP4). The highest allele frequency for this variant in gnomAD v4.1.0 is 0.00001098 (1/91080 South Asian chromosomes), which is less than the ClinGen LGMD VCEP threshold (<0.0001) (PM2_Supporting). The computational predictor REVEL gives a score of 0.91, which is above the LGMD VCEP threshold of ≥0.70 (PP3). In addition, another missense variant at the same codon, NM_003494.4: c.5093C>T p.(Pro1698Leu), has been classified as likely pathogenic by the ClinGen LGMD VCEP (PM5_Supporting). In summary, this variant is classified as Likely Pathogenic for autosomal recessive limb girdle muscular dystrophy based on the ACMG/AMP criteria applied, as specified by the ClinGen LGMD VCEP (specifications v2.0.0; 01/23/2026): PM3_Supporting, PP4, PP3, PM2_Supporting, PM5_Supporting, PP1.

Met criteria codes

PM3_Supporting  

This variant has been observed in three individuals with LGMD, including two siblings in a homozygous state with reported consanguinity (0.25 pts, ClinVar SCV000814671.7 internal data communication; PP1) and one with a pathogenic variant in unknown phase (c.265C>T p.(Arg89Ter), 0.5 pts, PMID: 38582400) (PM3_Supporting; PP4).

PM2_Supporting			The highest allele frequency for this variant in gnomAD v4.1.0 is 0.00001098 (1/91080 South Asian chromosomes), which is less than the ClinGen LGMD VCEP threshold (<0.0001) (PM2_Supporting).
PM5_Supporting			Another missense variant at the same codon, c.5093C>T p.(Pro1698Leu), has been classified as likely pathogenic by the ClinGen LGMD VCEP without the use of PM5 (PM5_Supporting). splice AI<0.5 for both variants In addition, there is another variant, p.(Pro1698Ser), that looks like it will not go above VUS (not curated).
PP1			Siblings diagnosed with LGMD. Both had this variant in a homozygous state (ClinVar SCV000814671.7 internal data communication).
PP3			The computational predictor REVEL gives a score of 0.91, which is above the LGMD VCEP threshold of ≥ 0.70 (PP3). Splice AI: 0.00
PP4			This variant has been observed in three individuals with LGMD with two presumably diagnostic variants (ClinVar SCV000814671.7 internal data communication; PMID: 38582400) (PP4).
Not Met criteria codes			
PS3			Not in Tominaga et al. study. No other functional studies.

Curation History



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