

Variant: NM_001130987.2(DYSF):c.5711G>A
(p.Gly1904Asp)

Version: 1.0

CA347223773 [↗](#)

617543 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 462a7dde-20d7-410e-b954-74e0d49325f6

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HGVS expressions

NM_001130987.2(DYSF):c.5711G>A (p.Gly1904Asp)
NM_001130987.2:c.5711G>A
NC_000002.12:g.71669673G>A
CM000664.2:g.71669673G>A
NC_000002.11:g.71896803G>A
CM000664.1:g.71896803G>A
NC_000002.10:g.71750311G>A
NG_008694.1:g.221051G>A
ENST00000698057.1:c.3125G>A
ENST00000698058.1:c.2342G>A
ENST00000698059.1:c.2450G>A
ENST00000258104.8:c.5594G>A
ENST00000410020.8:c.5711G>A
ENST00000258104.7:c.5594G>A
ENST00000394120.6:c.5597G>A
ENST00000409366.5:c.5660G>A
ENST00000409582.7:c.5708G>A
ENST00000409651.5:c.5690G>A
ENST00000409744.5:c.5618G>A
ENST00000409762.5:c.5645G>A
ENST00000410020.7:c.5711G>A
ENST00000410041.1:c.5648G>A
ENST00000413539.6:c.5687G>A
ENST00000429174.6:c.5657G>A
ENST00000479049.6:n.2479G>A
NM_001130455.1:c.5597G>A
NM_001130976.1:c.5552G>A
NM_001130977.1:c.5615G>A
NM_001130978.1:c.5657G>A
NM_001130979.1:c.5687G>A
NM_001130980.1:c.5645G>A
NM_001130981.1:c.5708G>A
NM_001130982.1:c.5690G>A
NM_001130983.1:c.5660G>A
NM_001130984.1:c.5618G>A

NM_001130985.1:c.5648G>A
NM_001130986.1:c.5555G>A
NM_001130987.1:c.5711G>A
NM_003494.3:c.5594G>A
NM_001130455.2:c.5597G>A
NM_001130976.2:c.5552G>A
NM_001130977.2:c.5615G>A
NM_001130978.2:c.5657G>A
NM_001130979.2:c.5687G>A
NM_001130980.2:c.5645G>A
NM_001130981.2:c.5708G>A
NM_001130982.2:c.5690G>A
NM_001130983.2:c.5660G>A
NM_001130984.2:c.5618G>A
NM_001130985.2:c.5648G>A
NM_001130986.2:c.5555G>A
NM_003494.4:c.5594G>A

Likely Pathogenic

Met criteria codes 4

- PM3_Strong
- PP3
- PP4
- 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 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.5594G>A variant in DYSF which is also known as NM_001130987.2: c.5711G>A p.(Gly1904Asp), is a missense variant predicted to cause substitution of glycine to aspartic acid at amino acid 1865, p.(Gly1865Asp). This variant has been reported in at least four unrelated individuals with features consistent with LGMD (PMID: 29382405, 34559919, 32140910; ClinVar SCV003831300.3 internal data communication), including confirmed in trans with a pathogenic variant in two patients (NM_003494.4: c.2779del p.(Ala927LeufsTer21), 1.0 pt, PMID: 32140910; c.5713C>T p.(Arg1905Ter), 1.0 pt, PMID: 34559919). It was also observed in unconfirmed phase with a pathogenic variant (c.2697+1G>A, 0.5 pts, PMID: 29382405) (PM3_Strong). At least one of the individuals with this variant and a second pathogenic DYSF variant displayed progressive limb girdle muscle weakness and had a clinical suspicion or diagnosis of LGMD (PMID: 34559919; PP4). The highest minor allele frequency of this variant is 0.00001333 (1/75032 chromosomes) for the African/African-American population in gnomAD v4.1.0, which is lower than the LGMD VCEP threshold (<0.0001) (PM2_Supporting). The computational predictor REVEL gives a score of 0.905, which is above the LGMD VCEP threshold of ≥0.70, (PP3). In summary, this variant meets the criteria to be 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; 04/29/2026): PM3_Strong, PP4, PM2_Supporting, PP3.

Met criteria codes

PM3_Strong			This variant has been reported in at least four unrelated individuals with features consistent with LGMD (PMID: 29382405, 34559919, 32140910; ClinVar SCV003831300.3 internal data communication), including confirmed in trans with a pathogenic variant in two patients (NM_003494.4: c.2779del p.(Ala927LeufsTer21), 1.0 pt, PMID: 32140910; c.5713C>T p.(Arg1905Ter), 1.0 pt, PMID: 34559919). It was also observed in unconfirmed phase with a pathogenic variant (c.2697+1G>A, 0.5 pts, PMID: 29382405) (PM3_Strong). Total points 2.5 This variant is absent from gnomAD v2, so co-occurrence can not be assessed 2/12/2026 Revvity unable to release the 2nd "pathogenic" variant.
PP3			The computational predictor REVEL gives a score of 0.905, which is above the LGMD VCEP threshold of ≥ 0.70 , (PP3). SpliceAI: no splice effect. (donor and acceptor gain 0.01)
PP4			At least one individual with this variant displayed progressive limb girdle muscle weakness and had a clinical suspicion of LGMD (PMID: 34559919; PP4).
PM2_Supporting			The highest minor allele frequency of this variant is 0.00001333 (1/75032 chromosomes) for the African/African-American population in gnomAD v4.1.0, which is lower than the LGMD VCEP threshold (< 0.0001) for PM2_Supporting.

Curation History 		
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