

Variant: NM_001130987.2(DYSF):c.1465G>A (p.Glu489Lys)

Version: 1.0

CA147721 [↗](#)

94270 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: efc07fa6-6c3d-4618-aa25-644f1e78cf54

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HGVS expressions

NM_001130987.2(DYSF):c.1465G>A (p.Glu489Lys)
NM_001130987.2:c.1465G>A
NC_000002.12:g.71535283G>A
CM000664.2:g.71535283G>A
NC_000002.11:g.71762413G>A
CM000664.1:g.71762413G>A
NC_000002.10:g.71615921G>A
NG_008694.1:g.86661G>A
ENST00000258104.8:c.1369G>A
ENST00000410020.8:c.1465G>A
ENST00000258104.7:c.1369G>A
ENST00000394120.6:c.1372G>A
ENST00000409366.5:c.1372G>A
ENST00000409582.7:c.1462G>A
ENST00000409651.5:c.1465G>A
ENST00000409744.5:c.1372G>A
ENST00000409762.5:c.1462G>A
ENST00000410020.7:c.1465G>A
ENST00000410041.1:c.1465G>A
ENST00000413539.6:c.1462G>A
ENST00000429174.6:c.1369G>A
NM_001130455.1:c.1372G>A
NM_001130976.1:c.1369G>A
NM_001130977.1:c.1369G>A
NM_001130978.1:c.1369G>A
NM_001130979.1:c.1462G>A
NM_001130980.1:c.1462G>A
NM_001130981.1:c.1462G>A
NM_001130982.1:c.1465G>A
NM_001130983.1:c.1372G>A
NM_001130984.1:c.1372G>A
NM_001130985.1:c.1465G>A
NM_001130986.1:c.1372G>A
NM_001130987.1:c.1465G>A
NM_003494.3:c.1369G>A
NM_001130455.2:c.1372G>A
NM_001130976.2:c.1369G>A

NM_001130977.2:c.1369G>A
NM_001130978.2:c.1369G>A
NM_001130979.2:c.1462G>A
NM_001130980.2:c.1462G>A
NM_001130981.2:c.1462G>A
NM_001130982.2:c.1465G>A
NM_001130983.2:c.1372G>A
NM_001130984.2:c.1372G>A
NM_001130985.2:c.1465G>A
NM_001130986.2:c.1372G>A
NM_003494.4:c.1369G>A

Benign

Met criteria codes 1

BA1

Not Met criteria codes 1

BP4

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 1.0.0

Criteria Specification Approval History

Criteria Specifications for this VCEP

Evidence submitted by expert panel

Limb Girdle Muscular Dystrophy VCEP

The NM_003494.4: c.1369G>A variant in DYSF, which is also known as NM_001130987.2: c.1465G>A (p.Glu489Lys), is a missense variant predicted to cause substitution of glutamic acid by lysine at amino acid 457 (p.Glu457Lys). The filtering allele frequency of the variant is 0.02368 for European (non-Finnish) exome chromosomes in gnomAD v2.1.1 (the lower threshold of the 95% CI of 4291/251472), which is higher than the VCEP threshold of 0.003 (BA1). The SpliceAI score for this variant is 0.28 and the computational predictor REVEL gives a score of 0.41. Both of these scores are neither above nor below the thresholds predicting a damaging or benign impact (BP4/PP3 not met). In summary, this variant meets the criteria to be classified as Benign 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): BA1.

Met criteria codes

BA1



The filtering allele frequency of the variant is 0.02368 for European (non-Finnish) exome chromosomes in gnomAD v2.1.1 (the lower threshold of the 95% CI of 4291/251472), which is higher than the VCEP threshold of 0.003 (BA1).

Not Met criteria codes

BP4



The SpliceAI score for this variant is 0.28 and the computational predictor REVEL gives a score of 0.41. Both of these scores are neither above nor below the thresholds predicting a damaging or benign impact (BP4/PP3 not met).



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