

Variant: NM_001130987.2(DYSF):c.2402C>T (p.Ala801Val)

Version: 1.0

CA1706139 [↗](#)

288008 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: b02c22a7-4e54-4755-aae6-73cce8a45c16

Approved on: 2026/1/23

Published on: 2026/2/17

HGVS expressions

NM_001130987.2(DYSF):c.2402C>T (p.Ala801Val)

NM_001130987.2:c.2402C>T

NC_000002.12:g.71561937C>T

CM000664.2:g.71561937C>T

NC_000002.11:g.71789067C>T

CM000664.1:g.71789067C>T

NC_000002.10:g.71642575C>T

NG_008694.1:g.113315C>T

ENST00000258104.8:c.2348C>T

ENST00000410020.8:c.2402C>T

ENST00000258104.7:c.2348C>T

ENST00000394120.6:c.2351C>T

ENST00000409366.5:c.2351C>T

ENST00000409582.7:c.2399C>T

ENST00000409651.5:c.2444C>T

ENST00000409744.5:c.2309C>T

ENST00000409762.5:c.2399C>T

ENST00000410020.7:c.2402C>T

ENST00000410041.1:c.2402C>T

ENST00000413539.6:c.2441C>T

ENST00000429174.6:c.2348C>T

NM_001130455.1:c.2351C>T

NM_001130976.1:c.2306C>T

NM_001130977.1:c.2306C>T

NM_001130978.1:c.2348C>T

NM_001130979.1:c.2441C>T

NM_001130980.1:c.2399C>T

NM_001130981.1:c.2399C>T

NM_001130982.1:c.2444C>T

NM_001130983.1:c.2351C>T

NM_001130984.1:c.2309C>T

NM_001130985.1:c.2402C>T

NM_001130986.1:c.2309C>T

NM_001130987.1:c.2402C>T

NM_003494.3:c.2348C>T

NM_001130455.2:c.2351C>T

NM_001130976.2:c.2306C>T

NM_001130977.2:c.2306C>T
NM_001130978.2:c.2348C>T
NM_001130979.2:c.2441C>T
NM_001130980.2:c.2399C>T
NM_001130981.2:c.2399C>T
NM_001130982.2:c.2444C>T
NM_001130983.2:c.2351C>T
NM_001130984.2:c.2309C>T
NM_001130985.2:c.2402C>T
NM_001130986.2:c.2309C>T
NM_003494.4:c.2348C>T

Likely Benign

Met criteria codes 1

BS1

Not Met criteria codes 4

PS3

PP3

PM3

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 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.2348C>T variant in DYSF, which is also known as NM_001130987.2: c.2402C>T p.(Ala801Val), is a missense variant predicted to cause substitution of alanine to valine at amino acid 801, p.(Ala801Val). This variant has been observed in at least three individuals with suspected LGMD in a heterozygous state, but none of them would be scoreable for PM3 application (PMID: 30564623, Jain Foundation Dysferlin Registry internal data communication; PM3_supporting not met). The filtering allele frequency of this variant is 0.001406 (the lower threshold of the 95% CI of 123/74924 African/African American chromosomes) in gnomAD v4.1.0, which is greater than the LGMD VCEP BS1 threshold (>0.001) (BS1 met). The computational predictor REVEL gives a score of 0.28 (PP3 and BP4 not met). In summary, this variant is classified as likely benign 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): BS1.

Met criteria codes

BS1



The filtering allele frequency of this variant is 0.001406 (the lower threshold of the 95% CI of 123/74924 African/African American chromosomes) in gnomAD v4.1.0, which is greater than the LGMD VCEP BS1 threshold (>0.001) (BS1 met).

Not Met criteria codes

PS3



Not in Tominaga et al. paper and no other functional studies.

PP3			The computational predictor REVEL gives a score of 0.28 (PP3 and BP4 not met). Splice AI; 0.05
PM3			This variant has been observed in at least three individuals with suspected LGMD in a heterozygous state, but none of them would be scoreable for PM3 application (PMID: 30564623, Jain Foundation Dysferlin Registry internal data communication; PM3_supporting not met).
BP4			The computational predictor REVEL gives a score of 0.28 (PP3 and BP4 not met).

Curation History 		
  ▾  ▾		
Showing 1 to 1 of 1 rows		

The information on this website is not intended for direct diagnostic use or medical decision-making without review by a genetics professional. Individuals should not change their health behavior solely on the basis of information contained on this website. If you have questions about the information contained on this website, please see a health care professional.