

Variant: NM_001130987.2(DYSF):c.2554A>G (p.Ile852Val)

Version: 1.0

CA147734 [↗](#)

94289 (ClinVar) [↗](#)

Gene: DYSF ([HGNC:8291](#))
Condition: autosomal recessive limb-girdle muscular dystrophy ([MONDO:0015152](#))
Inheritance Mode: Autosomal recessive inheritance
UUID: e670c716-71d2-4c5a-9f1c-dec8f61eb22b
Approved on: 2025/1/8
Published on: 2025/1/8

HGVS expressions

NM_001130987.2(DYSF):c.2554A>G (p.Ile852Val)
NM_001130987.2:c.2554A>G
NC_000002.12:g.71564202A>G
CM000664.2:g.71564202A>G
NC_000002.11:g.71791332A>G
CM000664.1:g.71791332A>G
NC_000002.10:g.71644840A>G
NG_008694.1:g.115580A>G
ENST00000258104.8:c.2500A>G
ENST00000410020.8:c.2554A>G
ENST00000258104.7:c.2500A>G
ENST00000394120.6:c.2503A>G
ENST00000409366.5:c.2503A>G
ENST00000409582.7:c.2551A>G
ENST00000409651.5:c.2596A>G
ENST00000409744.5:c.2461A>G
ENST00000409762.5:c.2551A>G
ENST00000410020.7:c.2554A>G
ENST00000410041.1:c.2554A>G
ENST00000413539.6:c.2593A>G
ENST00000429174.6:c.2500A>G
NM_001130455.1:c.2503A>G
NM_001130976.1:c.2458A>G
NM_001130977.1:c.2458A>G
NM_001130978.1:c.2500A>G
NM_001130979.1:c.2593A>G
NM_001130980.1:c.2551A>G
NM_001130981.1:c.2551A>G
NM_001130982.1:c.2596A>G
NM_001130983.1:c.2503A>G
NM_001130984.1:c.2461A>G
NM_001130985.1:c.2554A>G
NM_001130986.1:c.2461A>G
NM_001130987.1:c.2554A>G
NM_003494.3:c.2500A>G
NM_001130455.2:c.2503A>G
NM_001130976.2:c.2458A>G

NM_001130977.2:c.2458A>G
NM_001130978.2:c.2500A>G
NM_001130979.2:c.2593A>G
NM_001130980.2:c.2551A>G
NM_001130981.2:c.2551A>G
NM_001130982.2:c.2596A>G
NM_001130983.2:c.2503A>G
NM_001130984.2:c.2461A>G
NM_001130985.2:c.2554A>G
NM_001130986.2:c.2461A>G
NM_003494.4:c.2500A>G

Benign

Met criteria codes 2

BP4

BA1

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.2500A>G variant in DYSF, which is also known as NM_001130987.2: c.2554A>G p.(Ile852Val), is a missense variant predicted to cause substitution of isoleucine by valine at amino acid 834 (p.Ile834Val). The filtering allele frequency of the variant is 0.08367 for African/African American genome chromosomes in gnomAD v2.1.1 (the lower threshold of the 95% CI of 1541/31380), which is higher than the VCEP threshold of 0.003 (BA1). The SpliceAI score for this variant is 0, which suggests it does not impact splicing and is below the LGMD VCEP threshold of ≤0.05. The computational predictor REVEL gives a score of 0.08, which is below the LGMD VCEP threshold predicting a benign impact on DYSF function (≤0.1) (BP4). 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, BP4.

Met criteria codes

BP4



The SpliceAI score for this variant is 0, which suggests it does not impact splicing and is below the LGMD VCEP threshold of ≤0.05. The computational predictor REVEL gives a score of 0.08, which is below the LGMD VCEP threshold predicting a benign impact on DYSF function (≤0.1) (BP4).

BA1



The filtering allele frequency of the variant is 0.08367 for African/African American genome chromosomes in gnomAD v2.1.1 (the lower threshold of the 95% CI of 1541/31380), which is higher than the VCEP threshold of 0.003 (BA1).



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