

Variant: NM_001130987.2(DYSF):c.3946A>G (p.Ile1316Val)

Version: 1.0

CA179991 [↗](#)

6667 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: aca77cd8-4d85-454c-8411-cf7c9b4b8b2e

Approved on: 2025/1/8

Published on: 2025/1/8

HGVS expressions

NM_001130987.2:c.3946A>G
NM_001130987.2(DYSF):c.3946A>G (p.Ile1316Val)
NC_000002.12:g.71602794A>G
CM000664.2:g.71602794A>G
NC_000002.11:g.71829924A>G
CM000664.1:g.71829924A>G
NC_000002.10:g.71683432A>G
NG_008694.1:g.154172A>G
ENST00000698057.1:c.1318A>G
ENST00000698058.1:c.535A>G
ENST00000698059.1:c.535A>G
ENST00000258104.8:c.3892A>G
ENST00000410020.8:c.3946A>G
ENST00000258104.7:c.3892A>G
ENST00000394120.6:c.3895A>G
ENST00000409366.5:c.3895A>G
ENST00000409582.7:c.3943A>G
ENST00000409651.5:c.3988A>G
ENST00000409744.5:c.3853A>G
ENST00000409762.5:c.3943A>G
ENST00000410020.7:c.3946A>G
ENST00000410041.1:c.3946A>G
ENST00000413539.6:c.3985A>G
ENST00000429174.6:c.3892A>G
ENST00000472873.5:n.276A>G
ENST00000479049.6:n.777A>G
ENST00000487180.5:n.111A>G
ENST00000494501.5:n.252A>G
NM_001130455.1:c.3895A>G
NM_001130976.1:c.3850A>G
NM_001130977.1:c.3850A>G
NM_001130978.1:c.3892A>G
NM_001130979.1:c.3985A>G
NM_001130980.1:c.3943A>G
NM_001130981.1:c.3943A>G
NM_001130982.1:c.3988A>G
NM_001130983.1:c.3895A>G

	NM_001130984.1:c.3853A>G NM_001130985.1:c.3946A>G NM_001130986.1:c.3853A>G NM_001130987.1:c.3946A>G NM_003494.3:c.3892A>G NM_001130455.2:c.3895A>G NM_001130976.2:c.3850A>G NM_001130977.2:c.3850A>G NM_001130978.2:c.3892A>G NM_001130979.2:c.3985A>G NM_001130980.2:c.3943A>G NM_001130981.2:c.3943A>G NM_001130982.2:c.3988A>G NM_001130983.2:c.3895A>G NM_001130984.2:c.3853A>G NM_001130985.2:c.3946A>G NM_001130986.2:c.3853A>G NM_003494.4:c.3892A>G
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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

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Limb Girdle Muscular Dystrophy VCEP

The NM_003494.4: c.3892A>G variant in DYSF, which is also known as NM_001130987.2: c.3946A>G p.(Ile1316Val), is a missense variant predicted to cause substitution of isoleucine by valine at amino acid 1298 (p.Ile1298Val). The filtering allele frequency of the variant is 0.006916 for European (non-Finnish) genome chromosomes in gnomAD v3.1.2 (the lower threshold of the 95% CI of 695/152140), which is higher than the VCEP threshold of 0.003 (BA1). The SpliceAI score for this variant is 0.12 and the computational predictor REVEL gives a score of 0.42. 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 0.006916 for European (non-Finnish) genome chromosomes in gnomAD v3.1.2 (the lower threshold of the 95% CI of 695/152140), which is higher than the VCEP threshold of 0.003 (BA1).
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Not Met criteria codes

BP4			The SpliceAI score for this variant is 0.12 and the computational predictor REVEL gives a score of 0.42. 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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Curation History



 

 

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