

Variant: NM_001130987.2(DYSF):c.3381_3382del
(p.Phe1128fs)

Version: 1.0

CA222154 [↗](#)

94303 (ClinVar) [↗](#)

Gene: DYSF ([HGNC:8291](#))
Condition: limb-girdle muscular dystrophy ([MONDO:0016971](#))
Inheritance Mode: Autosomal recessive inheritance
UUID: f53de391-e73a-469c-928e-cec5a6ce81e4
Approved on: 2026/2/10
Published on: 2026/2/17

HGVS expressions

NM_001130987.2:c.3381_3382del
NM_001130987.2(DYSF):c.3381_3382del (p.Phe1128fs)
NC_000002.12:g.71574350_71574351del
CM000664.2:g.71574350_71574351del
NC_000002.11:g.71801480_71801481del
CM000664.1:g.71801480_71801481del
NC_000002.10:g.71654988_71654989del
NG_008694.1:g.125728_125729del
ENST00000698057.1:c.753_754del
ENST00000258104.8:c.3327_3328del
ENST00000410020.8:c.3381_3382del
ENST00000258104.7:c.3327_3328del
ENST00000394120.6:c.3330_3331del
ENST00000409366.5:c.3330_3331del
ENST00000409582.7:c.3378_3379del
ENST00000409651.5:c.3423_3424del
ENST00000409744.5:c.3288_3289del
ENST00000409762.5:c.3378_3379del
ENST00000410020.7:c.3381_3382del
ENST00000410041.1:c.3381_3382del
ENST00000413539.6:c.3420_3421del
ENST00000429174.6:c.3327_3328del
NM_001130455.1:c.3330_3331del
NM_001130976.1:c.3285_3286del
NM_001130977.1:c.3285_3286del
NM_001130978.1:c.3327_3328del
NM_001130979.1:c.3420_3421del
NM_001130980.1:c.3378_3379del
NM_001130981.1:c.3378_3379del
NM_001130982.1:c.3423_3424del
NM_001130983.1:c.3330_3331del
NM_001130984.1:c.3288_3289del
NM_001130985.1:c.3381_3382del
NM_001130986.1:c.3288_3289del
NM_001130987.1:c.3381_3382del

NM_003494.3:c.3327_3328del
NM_001130455.2:c.3330_3331del
NM_001130976.2:c.3285_3286del
NM_001130977.2:c.3285_3286del
NM_001130978.2:c.3327_3328del
NM_001130979.2:c.3420_3421del
NM_001130980.2:c.3378_3379del
NM_001130981.2:c.3378_3379del
NM_001130982.2:c.3423_3424del
NM_001130983.2:c.3330_3331del
NM_001130984.2:c.3288_3289del
NM_001130985.2:c.3381_3382del
NM_001130986.2:c.3288_3289del
NM_003494.4:c.3327_3328del

Pathogenic

Met criteria codes 4

- PM3_Supporting
- PVS1
- PM2_Supporting
- PP4

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.3327_3328del p.(Phe1110CysfsTer3) variant in DYSF, which is also known as NM_001130987.2: c.3381_3382del p.(Phe1128CysfsTer3), is a frameshift variant that is predicted to result in a premature stop codon in exon 30/55, leading to nonsense mediated decay in a gene in which loss of function is an established mechanism of disease (PVS1). This variant has been reported in at least two unrelated patients with clinical signs of LGMD (PMID: 35723113, 34559919), including in unconfirmed phase with a pathogenic variant (NM_003494.4: c.965T>C p.(Leu322Pro), PMID: 34559919, 0.5 pts) (PM3_Supporting). At least one patient with this variant and a second presumed diagnostic DYSF variant displayed progressive limb girdle muscle weakness (PMID: 34559919; PP4). This variant is also absent from gnomAD v4.1.0 (PM2_Supporting). In summary, this variant meets the criteria to be classified as Pathogenic for autosomal recessive limb girdle muscular dystrophy based on the ACMG/AMP criteria applied, as specified by the ClinGen LGMD VCEP specifications version 2.0.0; 02/10/2026): PVS1, PM3_Supporting, PP4, PM2_Supporting.

Met criteria codes

PM3_Supporting  

This variant has been reported in at least two unrelated patients with clinical signs of LGMD (PMID: 35723113, 34559919), including in unconfirmed phase with a pathogenic variant (NM_003494.4: c.965T>C p.(Leu322Pro), PMID: 34559919, 0.5 pts) (PM3_Supporting).

PVS1  

The NM_003494.4: c.3327_3328del p.(Phe1110CysfsTer3) variant in DYSF, which is also known as NM_001130987.2: c.3381_3382del p.(Phe1128CysfsTer3), is a frameshift variant that is predicted to result in a premature stop codon in

exon 30/55, leading to nonsense mediated decay in a gene in which loss of function is an established mechanism of disease (PVS1). SpliceAI 0.01

PM2_Supporting			Variant is absent from gnomAD v4.1.0 (PM2_Supporting).
PP4			At least one patient with this variant and a second presumed diagnostic DYSF variant displayed progressive limb girdle muscle weakness (PMID: 34559919; PP4).

Curation History



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