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Variant: NM_001130987.2(DYSF):c.1589_1592dup (p.Phe532fs)

Version: 1.0

CA913190077

596644 (ClinVar)

Gene: DYSF (HGNC:8291)
Condition: autosomal recessive limb-girdle muscular dystrophy (MONDO:0015152)
Inheritance Mode: Autosomal recessive inheritance
UUID: 34281398-243b-4022-a3bb-e7be1d31978d
Approved on: 2025/5/28
Published on: 2025/6/6

HGVS expressions

NM_001130987.2(DYSF):c.1589_1592dup (p.Phe532fs)
NM_001130987.2:c.1589_1592dup
NC_000002.12:g.71551053_71551056dup
CM000664.2:g.71551053_71551056dup
NC_000002.11:g.71778183_71778186dup
CM000664.1:g.71778183_71778186dup
NC_000002.10:g.71631691_71631694dup
NG_008694.1:g.102431_102434dup
ENST00000258104.8:c.1535_1538dup
ENST00000410020.8:c.1589_1592dup
ENST00000258104.7:c.1535_1538dup
ENST00000394120.6:c.1538_1541dup
ENST00000409366.5:c.1538_1541dup
ENST00000409582.7:c.1586_1589dup
ENST00000409651.5:c.1631_1634dup
ENST00000409744.5:c.1496_1499dup
ENST00000409762.5:c.1586_1589dup
ENST00000410020.7:c.1589_1592dup
ENST00000410041.1:c.1589_1592dup
ENST00000413539.6:c.1628_1631dup
ENST00000429174.6:c.1535_1538dup
NM_001130455.1:c.1538_1541dup
NM_001130976.1:c.1493_1496dup
NM_001130977.1:c.1493_1496dup
NM_001130978.1:c.1535_1538dup
NM_001130979.1:c.1628_1631dup
NM_001130980.1:c.1586_1589dup
NM_001130981.1:c.1586_1589dup
NM_001130982.1:c.1631_1634dup
NM_001130983.1:c.1538_1541dup
NM_001130984.1:c.1496_1499dup
NM_001130985.1:c.1589_1592dup
NM_001130986.1:c.1496_1499dup
NM_001130987.1:c.1589_1592dup
NM_003494.3:c.1535_1538dup

NM_001130455.2:c.1538_1541dup
NM_001130976.2:c.1493_1496dup
NM_001130977.2:c.1493_1496dup
NM_001130978.2:c.1535_1538dup
NM_001130979.2:c.1628_1631dup
NM_001130980.2:c.1586_1589dup
NM_001130981.2:c.1586_1589dup
NM_001130982.2:c.1631_1634dup
NM_001130983.2:c.1538_1541dup
NM_001130984.2:c.1496_1499dup
NM_001130985.2:c.1589_1592dup
NM_001130986.2:c.1496_1499dup
NM_003494.4:c.1535_1538dup

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 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.1535_1538dup p.(Phe514GlyfsTer57) variant in DYSF, which is also known as NM_001130987.2: c.1589_1592dup p.(Phe532GlyfsTer57), is a frameshift variant predicted to cause a premature stop codon in biologically relevant exon 19/55, leading to nonsense mediated decay in a gene in which loss of function is an established disease mechanism (PVS1). This variant has been identified in one individual with suspected LGMD in unknown phase with a pathogenic variant (NM_003494.4: c.3051dup p.(Ile1018HisfsTer14), 0.5 pts, GRASP-LGMD consortium internal data communication) (PM3_Supporting; PP4). This variant is absent from gnomAD v.4.1.0 (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 (LGMD VCEP specifications version 1.0.0; 05/28/2025): PVS1, PM2_Supporting, PM3_Supporting, PP4.

Met criteria codes

PM3_Supporting	✔	This variant has been identified in one individual with suspected LGMD in unknown phase with a pathogenic variant (NM_003494.4: c.3051dup p.(Ile1018HisfsTer14), 0.5 pts, GRASP-LGMD consortium internal data communication) (PM3_Supporting; PP4). cannot check co-occurrence since this variant is absent from gnomAD c.3051dup is classified as Pathogenic independent of this case
PVS1	✔	The NM_003494.4: c.1535_1538dup p.(Phe514GlyfsTer57) variant in DYSF, which is also known as NM_001130987.2: c.1589_1592dup p.(Phe532GlyfsTer57), is a frameshift variant predicted to cause a premature stop codon in

biologically relevant exon 19/55, leading to nonsense mediated decay in a gene in which loss of function is an established disease mechanism (PVS1).

PM2_Supporting	✔	This variant is absent from gnomAD v.4.1.0 (PM2_Supporting).
PP4	✔	Individual with suspected LGMD and two presumed diagnostic variants (GRASP-LGMD consortium internal data).

Curation History

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