

Variant: NM_000070.3(CAPN3):c.1743_1744del (p.Glu582fs)

Version: 1.0

CA10604416 [↗](#)

283179 (ClinVar) [↗](#)

Gene: CAPN3 ([HGNC:825](#))

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 52524095-2571-4b87-98cc-3e7ef324e320

Approved on: 2026/2/18

Published on: 2026/4/9

HGVS expressions

NM_000070.3(CAPN3):c.1743_1744del (p.Glu582fs)
NM_000070.3:c.1743_1744del
NC_000015.10:g.42403000_42403001del
CM000677.2:g.42403000_42403001del
NC_000015.9:g.42695198_42695199del
CM000677.1:g.42695198_42695199del
NC_000015.8:g.40482490_40482491del
NG_008660.1:g.59898_59899del
ENST00000349748.8:c.1599_1600del
ENST00000357568.8:c.1743_1744del
ENST00000397163.8:c.1743_1744del
ENST00000466369.5:n.2252_2253del
ENST00000483208.5:n.2632_2633del
ENST00000495723.1:n.2632_2633del
ENST00000549793.5:n.1974_1975del
ENST00000638141.2:n.1614_1615del
ENST00000673646.1:c.207_208del
ENST00000673705.1:c.309+3348_309+3349del
ENST00000673813.1:n.580+85_580+86del
ENST00000318023.11:c.1599_1600del
ENST00000349748.7:c.1599_1600del
ENST00000357568.7:c.1743_1744del
ENST00000397163.7:c.1743_1744del
ENST00000397200.8:c.207_208del
ENST00000567071.5:c.202_203del
ENST00000569827.5:c.207_208del
NM_000070.2:c.1743_1744del
NM_024344.1:c.1743_1744del
NM_173087.1:c.1599_1600del
NM_173088.1:c.207_208del
NM_024344.2:c.1743_1744del
NM_173087.2:c.1599_1600del
NM_173088.2:c.207_208del

Pathogenic

PP4PM2_SupportingPVS1

Not Met criteria codes1

PM3

Evidence Links0

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 CAPN3 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_000070.3: c.1743_1744del p.(Glu582GlyfsTer3) variant in CAPN3 is a frameshift variant predicted to cause a premature stop codon in biologically relevant exon 13/24, 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 at least two patients with features consistent with LGMD (PMID: 30564623, 15689361, 10330340), at least one of whom had a second presumed diagnostic CAPN3 variant as well as a clinical suspicion of LGMD (PMID: 15689361, LOVD Individual #00214348; PP4). This variant is 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 Limb Girdle Muscular Dystrophy Expert Panel (specifications version 2.0.0; 02/18/2026): PVS1, PM2_Supporting, PP4.

Met criteria codes

PP4

At least one individual with this variant and a second presumed diagnostic CAPN3 variant displayed progressive limb girdle muscle weakness or had a clinical suspicion of LGMD (PMID: 15689361, LOVD Individual #00214348; PP4).

PM2_Supporting

This variant is absent from gnomAD v4.1.0 (PM2_Supporting).

PVS1

The NM_000070.3: c.1743_1744del p.(Glu582GlyfsTer3) variant in CAPN3 is a frameshift variant predicted to cause a premature stop codon in biologically relevant exon 13/24, leading to nonsense-mediated decay in a gene in which loss of function is an established disease mechanism (PVS1).

Not Met criteria codes

PM3

This variant has been identified in at least two patients with features consistent with LGMD (PMID: 30564623, 15689361, 10330340).

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.