

Variant: *NM_213599.3(ANO5):c.108_109del (p.Glu36fs)*

Version: 1.0

CA10606240 [↗](#)

288833 (ClinVar) [↗](#)

Gene: ANO5 ([HGNC:203859](#))

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 6f0d42f5-587e-4b7c-a76d-9a83877e909a

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HGVS expressions

NM_213599.3:c.108_109del

NM_213599.3(ANO5):c.108_109del (p.Glu36fs)

NC_000011.10:g.22211284_22211285del

CM000673.2:g.22211284_22211285del

NC_000011.9:g.22232830_22232831del

CM000673.1:g.22232830_22232831del

NC_000011.8:g.22189406_22189407del

NG_015844.1:g.23109_23110del

ENST00000682084.1:n.3282_3283del

ENST00000682266.1:c.-301_-300del

ENST00000682341.1:c.108_109del

ENST00000682530.1:c.105_106del

ENST00000682684.1:n.529_530del

ENST00000683197.1:c.108_109del

ENST00000683411.1:c.-362_-361del

ENST00000683437.1:c.-270-9813_-270-9812del

ENST00000683834.1:n.350_351del

ENST00000683897.1:n.394_395del

ENST00000684365.1:n.519_520del

ENST00000684663.1:c.105_106del

ENST00000324559.9:c.108_109del

ENST00000648804.1:n.639_640del

ENST00000324559.8:c.108_109del

NM_001142649.1:c.105_106del

NM_213599.2:c.108_109del

NM_001142649.2:c.105_106del

Pathogenic

Met criteria codes **4**

PM2_Supporting

PVS1

PP4

PM3_Supporting

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 ANO5 Version 1.0.0

Evidence submitted by expert panel

Limb Girdle Muscular Dystrophy VCEP

The NM_213599.3: c.108_109del p.(Glu36AspfsTer7) variant in ANO5 is a frameshift variant predicted to cause a premature stop codon in biologically relevant exon 3/22, 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 ANO5-related muscle disease (PMID: 32367299, 39666917, 30564623; LOVD Individuals #00460288, #00221640), including in unknown phase with a pathogenic ANO5 variant in one patient with hyperCKemia, signs of myopathy on EMG, mitochondrial signs on muscle biopsy, and typical changes on muscle MRI (c.191dup p.(Asn64LysfsTer15), 0.5 pts, PMID: 32367299, 39666917; LOVD Individual #00460288) (PM3_Supporting). At least one patient reported with this variant and a second presumed diagnostic ANO5 variant had a clinical suspicion of LGMD (PMID: 30564623; LOVD Individual #00221640) (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 LGMD VCEP (LGMD VCEP specifications version 1.0.0; 06/24/2025): PVS1, PM3_Supporting, PP4, PM2_Supporting.

Met criteria codes

PM2_Supporting			This variant is absent from gnomAD v4.1.0 (PM2_Supporting).
PVS1			The NM_213599.3: c.108_109del p.(Glu36AspfsTer7) variant in ANO5 is a frameshift variant predicted to cause a premature stop codon in biologically relevant exon 3/22, leading to nonsense mediated decay in a gene in which loss of function is an established disease mechanism (PVS1).
PP4			At least one patient reported with this variant and a second presumed diagnostic ANO5 variant had a clinical suspicion of LGMD (PMID: 30564623; LOVD Individual #00221640) (PP4).
PM3_Supporting			This variant has been identified in at least two patients with features consistent with ANO5-related muscle disease (PMID: 32367299, 39666917, 30564623; LOVD Individuals #00460288, #00221640), including in unknown phase with a pathogenic ANO5 variant in one patient with hyperCKemia, signs of myopathy on EMG, mitochondrial signs on muscle biopsy, and typical changes on muscle MRI (c.191dup p.(Asn64LysfsTer15), 0.5 pts, PMID: 32367299, 39666917; LOVD Individual #00460288) (PM3_Supporting). Cannot check co-occurrence data since variant under curation is absent from gnomAD.

Curation History [↗](#)

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