

Variant: NM_001100.4(ACTA1):c.217dup (p.Ile73fs)

Version: 1.0

CA915942047 [↗](#)

817462 (ClinVar) [↗](#)

Gene: ACTA1 ([HGNC:58](#))

Condition: alpha-actinopathy ([MONDO:0100084](#))

Inheritance Mode: Autosomal recessive inheritance

UUID: e3f9ec67-adea-4393-8423-5bcc29d619bd

Approved on: 2024/8/7

Published on: 2024/12/19

HGVS expressions

NM_001100.4(ACTA1):c.217dup (p.Ile73fs)

NM_001100.4:c.217dup

NC_000001.11:g.229432793dup

CM000663.2:g.229432793dup

NC_000001.10:g.229568540dup

CM000663.1:g.229568540dup

NC_000001.9:g.227635163dup

NG_006672.1:g.6304dup

ENST00000366683.4:c.217dup

ENST00000684723.1:c.82dup

ENST00000366683.3:c.217dup

ENST00000366684.7:c.217dup

NM_001100.3:c.217dup

Pathogenic

Met criteria codes **3**

PM2_Supporting

PM3

PVS1

Not Met criteria codes **1**

PS4

Evidence Links **0**

Expert Panel

Congenital Myopathies VCEP [↗](#)

Criteria Specification Information

[↗](#) **Criteria Specification:** ClinGen Congenital Myopathies

Expert Panel Specifications to the ACMG/AMP Variant

Interpretation Guidelines for ACTA1 Version 1.0.0

[↗](#) **Criteria Specification Approval History**

[↗](#) **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

Congenital Myopathies VCEP

The NM_001100.4:c.217dup (p.Ile73fs*11) variant in ACTA1 is a frameshift variant that is expected to result in nonsense-mediated mRNA decay and is present in biologically-relevant transcripts (PVS1). The variant is absent from gnomAD v4.1.0 (PM2_Supporting). There is no published data on this variant, however Invitae has reported the variant in one proband, who presented with hypotonia (SCV001201868.3). In addition, this proband carried the variant of interest in trans with a second ACTA1 variant c.782A>T (p.Glu261Val) that has been classified as pathogenic at Invitae (PM3). In summary, this variant meets criteria to be classified as pathogenic for autosomal recessive

Met criteria codes

PM2_Supporting			The variant is absent from gnomAD v4.1.0 with adequate genome and exome coverage, meeting PM2_Supporting criteria.
PM3			The proband carried the variant of interest in trans with a second ACTA1 variant c.782A>T (p.Glu261Val) that has been classified as pathogenic at Invitae.
PVS1			The variant is a frameshift variant that is predicted to undergo nonsense-mediated mRNA decay and is present in biologically-relevant transcripts

Not Met criteria codes

PS4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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Curation History



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