The ClinGen Evidence Repository is an **FDA-recognized human genetic variant database** containing expert-curated assertions regarding variants' pathogenicity and supporting evidence summaries. [Disclaimer]

Variant: NM_000257.4(MYH7):c.5401G>A (p.Glu1801Lys)

CA016087

43069 (ClinVar)

Gene: MYH7 (HGNC:4625)
Condition: dilated cardiomyopathy (MONDO:0005021)
Inheritance Mode: Autosomal dominant inheritance
UUID: 7c729fe3-84ef-4174-a0d8-c152b6dc76d7
Approved on: 2016-12-15
Published on: 2018-11-16

HGVS expressions

NM_000257.4:c.5401G>A
NM_000257.4(MYH7):c.5401G>A (p.Glu1801Lys)
NC_000014.9:g.23415153C>T
CM000676.2:g.23415153C>T
NC_000014.8:g.23884362C>T
CM000676.1:g.23884362C>T
NC_000014.7:g.22954202C>T
NG_007884.1:g.25509G>A
ENST00000355349.4:c.5401G>A
ENST00000355349.3:c.5401G>A
NM_000257.3:c.5401G>A

Likely Pathogenic

Met criteria codes 4

PP3 PM6 PM2 PS4_Supporting

Evidence Links 1

Expert Panel

Cardiomyopathy VCEP

Criteria Specification Information

Criteria Specification: *ClinGen Cardiomyopathy Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for MYH7 Version 2.0.0*
Criteria Specification Approval History
Criteria Specifications for this VCEP

Evidence submitted by expert panel

Cardiomyopathy VCEP

The c.5401G>A (p.Glu1801Lys) variant in MYH7 has been reported in 4 individuals with dilated cardiomyopathy, one of whom had additional myopathy features (PS4_Supporting; PMID:19477645; Partners LMM ClinVar SCV000059616.5). Additionally, in two of the probands with dilated cardiomyopathy, the variant occurred de novo (PM6; Partners LMM ClinVar SCV000059616.5). This variant was absent from large population studies (PM2; http://exac.broadinstitute.org). Computational prediction tools and conservation analysis suggest that this variant may impact the protein (PP3). In summary, this variant meets criteria to be classified as likely pathogenic for dilated cardiomyopathy in an autosomal dominant manner. MYH7-specific ACMG/AMP criteria applied (PMID:29300372): PM2; PM6; PP3; PS4_ Supporting

Met criteria codes

PP3	✔	Tools predict damaging
PM6	✔	2 assumed de novo occurrences (from ClinVar SCV000059616.5)
PM2	✔	Absent from ExAC
PS4_Supporting	✔	4 cases with DCM identified, one of which has myopathy features (including ClinVar SCV000059616.5)

Variant identified in 1 case with DCM and myopathy phenotype **PubMed:19477645**

Curation History

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