

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_000257.3(MYH7):c.4377G>T (p.Lys1459Asn)*

CA014901 [↗](#)

43012 (ClinVar) [↗](#)

Gene: MYH7 (HGNC:4625)

Condition: cardiomyopathy (MONDO:0004994)

Inheritance Mode: Autosomal dominant inheritance

UID: dfd5170f-2105-4cc6-870a-2f7511d716cd

Approved on: 2016-12-15

Published on: 2018-11-16

HGVS expressions

NM_000257.3:c.4377G>T

NM_000257.3(MYH7):c.4377G>T (p.Lys1459Asn)

NC_000014.9:g.23417295C>A

CM000676.2:g.23417295C>A

NC_000014.8:g.23886504C>A

CM000676.1:g.23886504C>A

NC_000014.7:g.22956344C>A

NG_007884.1:g.23367G>T

NR_126491.1:n.735C>A

NM_000257.4:c.4377G>T

ENST00000355349.3:c.4377G>T

Likely Benign

The Expert Panel has overridden the computationally generated classification - "Uncertain Significance - Conflicting Evidence"

Met criteria codes **2**

PP3 BS1

Evidence Links **0**

Expert Panel

Cardiomyopathy VCEP [↗](#)

Criteria Specification Information **!**

[↗](#) Criteria Specifications for this VCEP

Evidence submitted by expert panel

Cardiomyopathy VCEP

The filtering allele frequency of the c.4377G>T (p.Lys1459Asn) variant in the MYH7 gene is 0.0375% (34/66566) of European chromosomes by the Exome Aggregation Consortium (<http://exac.broadinstitute.org>), which is a high enough frequency to be classified as likely benign based on thresholds defined by the ClinGen Inherited Cardiomyopathy Expert Panel (BS1; PMID:29300372). Additionally, while computational prediction tools and conservation analysis suggest that this variant may impact the protein (PP3), this pathogenic evidence code (PP3) was not considered to be in conflict with a likely benign conclusion given the accuracy of computation prediction tools. In summary, this variant meets criteria to be classified as likely benign for cardiomyopathy in an autosomal dominant manner. MYH7-specific ACMG/AMP criteria applied (PMID:29300372): PP3; BS1

Met criteria codes

PP3



Tools predict damaging

BS1



Filtering allele frequency of 0.0375% in Europeans in ExAC

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.