

Variant: NM_000257.3(MYH7):c.77C>T (p.Ala26Val)

CA016817 [↗](#)

37375 (ClinVar) [↗](#)

Gene: MYH7 ([HGNC:4625](#))

Condition: hypertrophic cardiomyopathy ([MONDO:0005045](#))

Inheritance Mode: Autosomal dominant inheritance

UUID: e26a6375-f03a-4542-9924-d09781723678

Approved on: 2016-12-15

Published on: 2018-11-16

HGVS expressions

NM_000257.3:c.77C>T

NM_000257.3(MYH7):c.77C>T (p.Ala26Val)

NC_000014.9:g.23433656G>A

CM000676.2:g.23433656G>A

NC_000014.8:g.23902865G>A

CM000676.1:g.23902865G>A

NC_000014.7:g.22972705G>A

NG_007884.1:g.7006C>T

ENST00000355349.4:c.77C>T

ENST00000355349.3:c.77C>T

NM_000257.4:c.77C>T

Benign

Met criteria codes **1**

BA1

Evidence Links **0**

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 filtering allele frequency of the c.77C>T (p.Ala26Val) variant in the MYH7 gene is 0.55% (60/8646) of East Asian chromosomes by the Exome Aggregation Consortium (<http://exac.broadinstitute.org>), which is a high enough frequency to be classified as benign based on thresholds defined by the ClinGen Inherited Cardiomyopathy Expert Panel (BA1; PMID:29300372).

Met criteria codes

BA1





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Curation History [↗](#)

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