

Variant: *NM_000257.3(MYH7):c.4974C>T (p.Asp1658=)*

[CA015510](#) 

[43048 \(ClinVar\)](#) 

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

Condition: cardiomyopathy ([MONDO:0004994](#))

Inheritance Mode: Autosomal dominant inheritance

UUID: 9d15308c-24d6-4612-83db-e16129e10e7f

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HGVS expressions

NM_000257.3:c.4974C>T

NM_000257.3(MYH7):c.4974C>T (p.Asp1658=)

NR_126491.1:n.244G>A

NM_000257.4:c.4974C>T

ENST00000355349.3:c.4974C>T

NC_000014.9:g.23415812G>A

CM000676.2:g.23415812G>A

NC_000014.8:g.23885021G>A

CM000676.1:g.23885021G>A

NC_000014.7:g.22954861G>A

NG_007884.1:g.24850C>T

Benign

Met criteria codes **1**

BA1

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.4974C>T (p.Asp1658=)* variant in the MYH7 gene is 0.36% (53/11558) of Latino 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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