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.4974C>T (p.Asp1658=)

Version: 2.0

CA015510

43048 (ClinVar)

Gene: MYH7 (HGNC:4625)
Condition: hypertrophic cardiomyopathy (MONDO:0005045)
Inheritance Mode: Autosomal dominant inheritance
UUID: 9d15308c-24d6-4612-83db-e16129e10e7f
Approved on: 2025-11-11
Published on: 2026-06-04

HGVS expressions

NM_000257.3:c.4974C>T
NM_000257.3(MYH7):c.4974C>T (p.Asp1658=)
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
ENST00000355349.4:c.4974C>T
ENST00000355349.3:c.4974C>T
NR_126491.1:n.244G>A
NM_000257.4:c.4974C>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.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	✔	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	

Curation History

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See Report	Preferred Variant Title	Classification	Condition	Published Date	Version	Criteria Specification	Gene
View	NM_000257.3(MYH7):c.4974C>T (p.As...	Benign	Hypertrophic Cardiomyopathy	2026-06-04	2.0	ClinGen Cardiomyopathy Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for MYH7 Version 2.0.0	MYH7
View	NM_000257.3(MYH7):c.4974C>T (p.As...	Benign	Cardiomyopathy	2018-11-16	1.0	-	MYH7

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