

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

**UID:** 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 [↗](#)

Showing 1 to 2 of 2 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.