

Variant: *NM_000257.3(MYH7):c.2717A>G (p.Asp906Gly)*

[CA012936](#)

[14125 \(ClinVar\)](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 6a11c78d-e05e-417f-8453-1e66356e09e9

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HGVS expressions

NM_000257.3:c.2717A>G

NM_000257.3(MYH7):c.2717A>G (p.Asp906Gly)

NM_000257.4:c.2717A>G

ENST00000355349.3:c.2717A>G

NC_000014.9:g.23424112T>C

CM000676.2:g.23424112T>C

NC_000014.8:g.23893321T>C

CM000676.1:g.23893321T>C

NC_000014.7:g.22963161T>C

NG_007884.1:g.16550A>G

Pathogenic

Met criteria codes **4**

PM2 **PM1** **PS4** **PP1_Strong**

Evidence Links **5**

Expert Panel

[Cardiomyopathy VCEP](#)

Criteria Specification Information

[Criteria Specifications for this VCEP](#)

Evidence submitted by expert panel

Cardiomyopathy VCEP

The c.2717A>G (p.Asp906Gly) variant in MYH7 has been reported in >20 individuals with hypertrophic cardiomyopathy (PS4; PMID:27532257; PMID:12081993; PMID:15528230; PMID:16267253; PMID:24510615; Partners LMM ClinVar SCV000059468.5). This variant segregated with disease in 10 affected individuals (PP1_Strong; PMID:12081993; PMID:15528230). This variant was absent from large population studies (PM2; <http://exac.broadinstitute.org>). This variant lies in the head region of the protein (aa 181-937) and missense variants in this region are statistically more likely to be disease-associated (PM1; PMID:27532257). In summary, this variant meets criteria to be classified as pathogenic for hypertrophic cardiomyopathy in an autosomal dominant manner. MYH7-specific ACMG/AMP criteria applied (PMID:29300372): PS4; PP1_Strong; PM1; PM2

Met criteria codes

PM2



Absent from ExAC

PM1



Variants in head region of the protein (aa 181-937) are statistically more likely to be disease-associated

Variants in head region of the protein (aa 181-937) are statistically more likely to be disease-associated

[PubMed:27532257](#)

PS4



Variant identified in >20 probands with HCM including ClinVar SCV000059468.5

Variant identified in 1 proband with HCM [PubMed:16267253](#)

Variant identified in 1 proband with HCM [PubMed:15528230](#)

Variant identified in 5 probands with HCM [PubMed:24510615](#)

Variant identified in 1 proband with HCM [PubMed:12081993](#)

Variant identified in 7 probands with HCM [PubMed:27532257](#)

PP1_Strong



10 segregations

2 segregations [PubMed:15528230](#)

10 segregations [PubMed:12081993](#)

Curation History [↗](#)

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