

Variant: NM_000527.5(LDLR):c.1167G>A (p.Thr389=)

Version: 1.0

CA032357 [↗](#)

256365 (ClinVar) [↗](#)

Gene: LDLR ([HGNC:3949](#))

Condition: hypercholesterolemia, familial ([MONDO:0007750](#))

Inheritance Mode: Semidominant inheritance

UUID: bda540d2-f8f0-4520-bf4a-f111b5cb9d1b

Approved on: 2023/3/20

Published on: 2023/3/31

HGVS expressions

NM_000527.5(LDLR):c.1167G>A (p.Thr389=)

NM_000527.5:c.1167G>A

NC_000019.10:g.11111620G>A

CM000681.2:g.11111620G>A

NC_000019.9:g.11222296G>A

CM000681.1:g.11222296G>A

NC_000019.8:g.11083296G>A

NG_009060.1:g.27240G>A

ENST00000252444.10:c.1425G>A

ENST00000559340.2:c.1167G>A

ENST00000560467.2:c.1047G>A

ENST00000558518.6:c.1167G>A

ENST00000252444.9:c.1421G>A

ENST00000455727.6:c.663G>A

ENST00000535915.5:c.1044G>A

ENST00000545707.5:c.786G>A

ENST00000557933.5:c.1167G>A

ENST00000558013.5:c.1167G>A

ENST00000558518.5:c.1167G>A

ENST00000560173.1:n.166G>A

ENST00000560467.1:c.647G>A

NM_000527.4:c.1167G>A

NM_001195798.1:c.1167G>A

NM_001195799.1:c.1044G>A

NM_001195800.1:c.663G>A

NM_001195803.1:c.786G>A

NM_001195798.2:c.1167G>A

NM_001195799.2:c.1044G>A

NM_001195800.2:c.663G>A

NM_001195803.2:c.786G>A

Likely Benign

Met criteria codes 2

BP4BP7

Not Met criteria codes 3

BS1BA1PM2

Evidence Links 0

Expert Panel

Familial Hypercholesterolemia VCEP

Criteria Specification Information

[Criteria Specification:](#) ClinGen Familial Hypercholesterolemia Expert Panel Specifications to the ACMG/AMP Variant Classification Guidelines Version 1.2

[PDF](#)

[Criteria Specification Approval History](#)

[Criteria Specifications for this VCEP](#)

Evidence submitted by expert panel

Familial Hypercholesterolemia VCEP

The NM_000527.5(LDLR):c.1167G>A (p.Thr389=) variant is classified as likely benign for Familial Hypercholesterolemia by applying evidence codes: BP4, BP7 as defined by the ClinGen Familial Hypercholesterolemia Expert Panel LDLR-specific variant curation guidelines (<https://doi.org/10.1016/j.gim.2021.09.012>). The supporting evidence is as follows: BP4 - No REVEL, splicing evaluation needed. Functional data on splicing not available. A) not on limits B) does not create GT Variant is not predicted to alter splicing BP7 - Variant is synonymous and meets BP4.

Met criteria codes

BP4			No REVEL, splicing evaluation needed. Functional data on splicing not available. A) not on limits B) does not create GT Variant is not predicted to alter splicing
BP7			Variant is synonymous and meets BP4.

Not Met criteria codes

BS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BA1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM2			African/African American MAF = 0.001324 (0.13%)

Curation History

Showing 1 to 1 of 1 rows

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