

Variant: NM_000527.5(LDLR):c.1441G>A (p.Val481Met)

Version: 1.0

CA10585453 [🔗](#)

251843 (ClinVar) [🔗](#)

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

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

Inheritance Mode: Semidominant inheritance

UUID: e9fae1f3-84f4-4a1e-9c2f-fd89c9805ae0

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HGVS expressions

NM_000527.5(LDLR):c.1441G>A (p.Val481Met)

NM_000527.5:c.1441G>A

NC_000019.10:g.11113617G>A

CM000681.2:g.11113617G>A

NC_000019.9:g.11224293G>A

CM000681.1:g.11224293G>A

NC_000019.8:g.11085293G>A

NG_009060.1:g.29237G>A

ENST00000252444.10:c.1699G>A

ENST00000559340.2:c.1441G>A

ENST00000560467.2:c.1321G>A

ENST00000558518.6:c.1441G>A

ENST00000252444.9:c.1695G>A

ENST00000455727.6:c.937G>A

ENST00000535915.5:c.1318G>A

ENST00000545707.5:c.1060G>A

ENST00000557933.5:c.1441G>A

ENST00000558013.5:c.1441G>A

ENST00000558518.5:c.1441G>A

ENST00000559340.1:c.162G>A

ENST00000560467.1:c.921G>A

NM_000527.4:c.1441G>A

NM_001195798.1:c.1441G>A

NM_001195799.1:c.1318G>A

NM_001195800.1:c.937G>A

NM_001195803.1:c.1060G>A

NM_001195798.2:c.1441G>A

NM_001195799.2:c.1318G>A

NM_001195800.2:c.937G>A

NM_001195803.2:c.1060G>A

Uncertain Significance

Met criteria codes 2

PP3PM2

Not Met criteria codes 5

PS1BA1PM5BS1BP4

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.1441G>A (p.Val481Met) variant is classified as Uncertain significance - insufficient evidence for Familial Hypercholesterolemia by applying ACMG/AMP evidence codes PM2 and PP3 as defined by the ClinGen Familial Hypercholesterolemia Expert Panel LDLR-specific variant curation guidelines (specification version 1.2) on February 28, 2025. The supporting evidence is as follows: PM2: PopMax MAF = 0.0000008475 (0.00008475%) in European (non-Finnish) exomes + genomes (gnomAD v4.1.0). PP3: REVEL = 0.83.

Met criteria codes

PP3

REVEL = 0.83, It is above 0.75

PM2

PopMax MAF = 0.0000008475 (0.00008475%) in European (non-Finnish) exomes (gnomAD v4.1.0)

Not Met criteria codes

PS1

There are no other missense variants in the same codon

BA1

Variant is absent in gnomAD v4.1.0

PM5

There are no other missense variants in the same codon

BS1

Variant is absent in gnomAD v4.1.0

BP4

REVEL = 0.83, It is not below 0.50

Curation History

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