

Variant: NM_000527.5(LDLR):c.1190C>T (p.Ser397Phe)

Version: 1.0

CA404084657 [↗](#)

925475 (ClinVar) [↗](#)

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

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

Inheritance Mode: Semidominant inheritance

UUID: 1461d879-5453-46e8-824d-6d7214d2b247

Approved on: 2023/6/23

Published on: 2025/9/16

HGVS expressions

NM_000527.5(LDLR):c.1190C>T (p.Ser397Phe)

NM_000527.5:c.1190C>T

NC_000019.10:g.11113281C>T

CM000681.2:g.11113281C>T

NC_000019.9:g.11223957C>T

CM000681.1:g.11223957C>T

NC_000019.8:g.11084957C>T

NG_009060.1:g.28901C>T

ENST00000252444.10:c.1448C>T

ENST00000559340.2:c.1190C>T

ENST00000560467.2:c.1070C>T

ENST00000558518.6:c.1190C>T

ENST00000252444.9:c.1444C>T

ENST00000455727.6:c.686C>T

ENST00000535915.5:c.1067C>T

ENST00000545707.5:c.809C>T

ENST00000557933.5:c.1190C>T

ENST00000558013.5:c.1190C>T

ENST00000558518.5:c.1190C>T

ENST00000560173.1:n.189C>T

ENST00000560467.1:c.670C>T

NM_000527.4:c.1190C>T

NM_001195798.1:c.1190C>T

NM_001195799.1:c.1067C>T

NM_001195800.1:c.686C>T

NM_001195803.1:c.809C>T

NM_001195798.2:c.1190C>T

NM_001195799.2:c.1067C>T

NM_001195800.2:c.686C>T

NM_001195803.2:c.809C>T

Uncertain Significance

Met criteria codes 1

PM2

Not Met criteria codes 6

PM5

BS1

PS1

BP4

BA1

PP3

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.1190C>T (p.Ser397Phe) variant is classified as **Uncertain significance - insufficient evidence for Familial Hypercholesterolemia** by applying ACMG/AMP evidence code PM2 as defined by the ClinGen Familial Hypercholesterolemia Expert Panel LDLR-specific variant curation guidelines (specification version 1.2) on 23 June 2023. The supporting evidence is as follows: PM2: This variant is absent from gnomAD (gnomAD v2.1.1).

Met criteria codes

PM2			This variant is absent from gnomAD (gnomAD v2.1.1).
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Not Met criteria codes

PM5			2 other missense variants in the same codon: - NM_000527.5(LDLR):c.1189T>A (p.Ser397Thr) (ClinVar ID 251725) - Uncertain significance by these guidelines - NM_000527.5(LDLR):c.1189T>C (p.Ser397Pro) (ClinVar ID 923326) - Uncertain significance by these guidelines There are no variants in the same codon classified as Pathogenic by these guidelines.
BS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS1			There are no variants causing the same amino acid change classified as Pathogenic by these guidelines.
BP4			REVEL = 0.619. It is not below 0.5.
BA1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP3			REVEL = 0.619. It is not above 0.75, splicing evaluation needed. Functional data on splicing not available. A) Variant not on limits. B) Variant does not create de novo AG/GT. C) There is no AG/GT nearby Variant is not predicted to alter splicing.

Curation History



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