

Variant: *NM_000527.5(LDLR):c.1577C>T (p.Pro526Leu)*

Version: 1.0

CA404086610 [↗](#)

431531 (ClinVar) [↗](#)

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

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

Inheritance Mode: Semidominant inheritance

UUID: 56a69abc-eb8b-47de-9473-4dd3b351cbcc

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HGVS expressions

NM_000527.5(LDLR):c.1577C>T (p.Pro526Leu)

NM_000527.5:c.1577C>T

NC_000019.10:g.11113753C>T

CM000681.2:g.11113753C>T

NC_000019.9:g.11224429C>T

CM000681.1:g.11224429C>T

NC_000019.8:g.11085429C>T

NG_009060.1:g.29373C>T

ENST00000252444.10:c.1835C>T

ENST00000559340.2:c.1577C>T

ENST00000560467.2:c.1457C>T

ENST00000558518.6:c.1577C>T

ENST00000252444.9:c.1831C>T

ENST00000455727.6:c.1073C>T

ENST00000535915.5:c.1454C>T

ENST00000545707.5:c.1196C>T

ENST00000557933.5:c.1577C>T

ENST00000558013.5:c.1577C>T

ENST00000558518.5:c.1577C>T

ENST00000559340.1:c.298C>T

NM_000527.4:c.1577C>T

NM_001195798.1:c.1577C>T

NM_001195799.1:c.1454C>T

NM_001195800.1:c.1073C>T

NM_001195803.1:c.1196C>T

NM_001195798.2:c.1577C>T

NM_001195799.2:c.1454C>T

NM_001195800.2:c.1073C>T

NM_001195803.2:c.1196C>T

Uncertain Significance

Met criteria codes **2**

PP3 PM2

Not Met criteria codes **24**

Expert Panel

Familial Hypercholesterolemia VCEP [↗](#)

Criteria Specification Information **!**

PS1

PS2

PS3

PS4

BA1

PP1

PP2

PP4

PM1

PM3

PM5

PM4

PM6

BS1

BS4

BS3

BS2

BP5

BP7

BP4

BP3

BP1

BP2

PVS1

[Criteria Specifications for this VCEP](#)

Evidence Links 0

Evidence submitted by expert panel

Familial Hypercholesterolemia VCEP

NM_000527.5(LDLR):c.1577C>T (p.Pro526Leu) variant is classified as variant of Uncertain significance for Familial Hypercholesterolemia by applying evidence codes (PM2 and PP3) 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: PM2 - This variant is absent from gnomAD (gnomAD v2.1.1). PP3 - REVEL = 0.951

Met criteria codes

PP3			REVEL = 0.951
PM2			This variant is absent from gnomAD (gnomAD v2.1.1).

Not Met criteria codes

PS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS4			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
PP1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP2			

No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline			
PP4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM5			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM6			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP5			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP7			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

BP1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PVS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History 	
  	
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