

Variant: *NM_000527.5(LDLR):c.590G>T (p.Cys197Phe)*

Version: 1.0

CA044039 [↗](#)

251309 (ClinVar) [↗](#)

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

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

Inheritance Mode: Semidominant inheritance

UUID: 80471e75-a7c0-43b1-adbb-8e157b402631

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HGVS expressions

NM_000527.5:c.590G>T

NM_000527.5(LDLR):c.590G>T (p.Cys197Phe)

NC_000019.10:g.11105496G>T

CM000681.2:g.11105496G>T

NC_000019.9:g.11216172G>T

CM000681.1:g.11216172G>T

NC_000019.8:g.11077172G>T

NG_009060.1:g.21116G>T

ENST00000252444.10:c.848G>T

ENST00000559340.2:c.590G>T

ENST00000560467.2:c.590G>T

ENST00000558518.6:c.590G>T

ENST00000252444.9:c.844G>T

ENST00000455727.6:c.314-1896G>T

ENST00000535915.5:c.467G>T

ENST00000545707.5:c.314-1069G>T

ENST00000557933.5:c.590G>T

ENST00000558013.5:c.590G>T

ENST00000558518.5:c.590G>T

ENST00000560467.1:c.190G>T

NM_000527.4:c.590G>T

NM_001195798.1:c.590G>T

NM_001195799.1:c.467G>T

NM_001195800.1:c.314-1896G>T

NM_001195803.1:c.314-1069G>T

NM_001195798.2:c.590G>T

NM_001195799.2:c.467G>T

NM_001195800.2:c.314-1896G>T

NM_001195803.2:c.314-1069G>T

Likely Pathogenic

Met criteria codes **5**

PP1_Moderate

PP3

PP4

PM2

PM1

Evidence Links **0**

Expert Panel

Familial Hypercholesterolemia VCEP [↗](#)

Criteria Specification Information **!**

Evidence submitted by expert panel

Familial Hypercholesterolemia VCEP

NM_000527.5(LDLR):c.590G>T (p.Cys197Phe) variant is classified as likely pathogenic for Familial Hypercholesterolemia by applying evidence code PM1, PM2 PP1, PP3_Supporting and PP4 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 - PopMax MAF = 0.00006 (0.006%) in African exomes (gnomAD v2.1.1). PP3 - REVEL = 0.945. It is above 0.75 PM1 - Variant meets PM2 and located in highly conserved Cystein 197. PP1_Moderate - Variant segregate with phenotype in 4 informative meioses in 1 family. PP4 - Variant meets PM2 and is identified in 1 index case who fulfil SB criteria for FH (Cardiovascular Research Group, Instituto Nacional de Saude Doutor Ricardo Jorge)

Met criteria codes

PP1_Moderate			Variant segregate with phenotype in 4 informative meioses in 1 family.
PP3			REVEL = 0.945. It is above 0.75
PP4			Variant meets PM2 and is identified in 1 index case who fulfil SB criteria for FH (Cardiovascular Research Group, Instituto Nacional de Saude Doutor Ricardo Jorge)
PM2			PopMax MAF = 0.00006 (0.006%) in African exomes (gnomAD v2.1.1).
PM1			Variant meets PM2 and located in highly conserved Cystein 197.

Curation History [↗](#)

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