

Variant: *NM_000527.5(LDLR):c.1221C>T (p.His407=)*

Version: 1.0

CA505743141 [↗](#)

430774 (ClinVar) [↗](#)

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

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

Inheritance Mode: Semidominant inheritance

UUID: 04e95969-168a-4ce5-a889-deeb413333ea

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HGVS expressions

NM_000527.5:c.1221C>T

NM_000527.5(LDLR):c.1221C>T (p.His407=)

NC_000019.10:g.11113312C>T

CM000681.2:g.11113312C>T

NC_000019.9:g.11223988C>T

CM000681.1:g.11223988C>T

NC_000019.8:g.11084988C>T

NG_009060.1:g.28932C>T

ENST00000252444.10:c.1479C>T

ENST00000559340.2:c.1221C>T

ENST00000560467.2:c.1101C>T

ENST00000558518.6:c.1221C>T

ENST00000252444.9:c.1475C>T

ENST00000455727.6:c.717C>T

ENST00000535915.5:c.1098C>T

ENST00000545707.5:c.840C>T

ENST00000557933.5:c.1221C>T

ENST00000558013.5:c.1221C>T

ENST00000558518.5:c.1221C>T

ENST00000560173.1:n.220C>T

ENST00000560467.1:c.701C>T

NM_000527.4:c.1221C>T

NM_001195798.1:c.1221C>T

NM_001195799.1:c.1098C>T

NM_001195800.1:c.717C>T

NM_001195803.1:c.840C>T

NM_001195798.2:c.1221C>T

NM_001195799.2:c.1098C>T

NM_001195800.2:c.717C>T

NM_001195803.2:c.840C>T

Uncertain Significance

Met criteria codes **3**

BP4

BP7

PM2

Evidence Links **0**

Expert Panel

Familial Hypercholesterolemia VCEP [↗](#)

Criteria Specification Information **!**

Evidence submitted by expert panel

Familial Hypercholesterolemia VCEP

The NM_000527.5(LDLR):c.1221C>T (p.His407=) variant is classified as Uncertain significance - insufficient evidence for Familial Hypercholesterolemia by applying evidence codes (PM2, BP4, BP7) as defined by the ClinGen Familial Hypercholesterolemia Expert Panel LDLR-specific variant curation guidelines (<https://doi.org/10.1101/2021.03.17.21252755>). The supporting evidence is as follows: PM2 - This variant is absent from gnomAD (gnomAD v2.1.1). BP4 - No REVEL, splicing evaluation required. Functional data not available A) not on limits B) does not create AG Variant not predicted to alter splicing BP7 - Variant is synonymous and meets BP4.

Met criteria codes

BP4			BP4 - No REVEL, splicing evaluation required. Functional data not available A) not on limits B) does not create AG Variant not predicted to alter splicing
BP7			BP7 - Variant is synonymous and meets BP4.
PM2			PM2 - This variant is absent from gnomAD (gnomAD v2.1.1).

Curation History [↗](#)







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