

Variant: NM_000527.5(LDLR):c.1210A>C (p.Thr404Pro)

Version: 1.0

CA10585359 [🔗](#)

251735 (ClinVar) [🔗](#)

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

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

Inheritance Mode: Semidominant inheritance

UUID: 893fa193-f63f-4fcd-b2be-1768f75bd99b

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HGVS expressions

NM_000527.5:c.1210A>C

NM_000527.5(LDLR):c.1210A>C (p.Thr404Pro)

NC_000019.10:g.11113301A>C

CM000681.2:g.11113301A>C

NC_000019.9:g.11223977A>C

CM000681.1:g.11223977A>C

NC_000019.8:g.11084977A>C

NG_009060.1:g.28921A>C

ENST00000252444.10:c.1468A>C

ENST00000559340.2:c.1210A>C

ENST00000560467.2:c.1090A>C

ENST00000558518.6:c.1210A>C

ENST00000252444.9:c.1464A>C

ENST00000455727.6:c.706A>C

ENST00000535915.5:c.1087A>C

ENST00000545707.5:c.829A>C

ENST00000557933.5:c.1210A>C

ENST00000558013.5:c.1210A>C

ENST00000558518.5:c.1210A>C

ENST00000560173.1:n.209A>C

ENST00000560467.1:c.690A>C

NM_000527.4:c.1210A>C

NM_001195798.1:c.1210A>C

NM_001195799.1:c.1087A>C

NM_001195800.1:c.706A>C

NM_001195803.1:c.829A>C

NM_001195798.2:c.1210A>C

NM_001195799.2:c.1087A>C

NM_001195800.2:c.706A>C

NM_001195803.2:c.829A>C

Uncertain Significance

Met criteria codes 3

PP3

PP4

PM2

Not Met criteria codes 19

PS1

PS2

PS3

PS4

BP7

BP4

BP2

PP1

PM1

PM3

PM5

PM4

BA1

PM6

BS1

BS4

BS3

BS2

PVS1

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.1210A>C (p.Thr404Pro) variant is classified as Uncertain significance - insufficient evidence for Familial Hypercholesterolemia by applying evidence codes PM2, PP3, 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: This variant is absent from gnomAD v2.1.1 PP3: REVEL= 0.93 PP4: Variant meets PM2 and is identified in at least 1 index case who fulfills SB definite criteria for FH from PMID 8697568, after alternative causes of high cholesterol were excluded.

Met criteria codes

PP3			REVEL= 0.93
PP4			Variant meets PM2 and is identified in at least 1 index case who fulfills SB definite criteria for FH from PMID 8697568, after alternative causes of high cholesterol were excluded.
PM2			This variant is absent from gnomAD v2.1.1

Not Met criteria codes

PS1			Not met.
PS2			Not met.
PS3			No Functional evidence.
PS4			Not met.
BP7			Not a synonymous variant.
BP4			REVEL= 0.93

BP2			Not met.
PP1			Not met.
PM1			Not located in exon 4 or at a cysteine residue.
PM3			Not met.
PM5			Not met.
PM4			Not an in-frame deletion or insertion.
BA1			This variant is absent from gnomAD v2.1.1
PM6			Not met.
BS1			This variant is absent from gnomAD v2.1.1
BS4			Not met.
BS3			No Functional evidence.
BS2			Not met.
PVS1			Not met.

Curation History 		
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