

Variant: NM_000527.5(LDLR):c.904T>C (p.Cys302Arg)

Version: 1.0

CA404081019 [↗](#)

440611 (ClinVar) [↗](#)

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

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

Inheritance Mode: Semidominant inheritance

UUID: bcab7d84-27ed-49e7-9cc2-1691afb8d508

Approved on: 2023/6/23

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HGVS expressions

NM_000527.5(LDLR):c.904T>C (p.Cys302Arg)

NM_000527.5:c.904T>C

NC_000019.10:g.11107478T>C

CM000681.2:g.11107478T>C

NC_000019.9:g.11218154T>C

CM000681.1:g.11218154T>C

NC_000019.8:g.11079154T>C

NG_009060.1:g.23098T>C

ENST00000252444.10:c.1162T>C

ENST00000559340.2:c.904T>C

ENST00000560467.2:c.904T>C

ENST00000558518.6:c.904T>C

ENST00000252444.9:c.1158T>C

ENST00000455727.6:c.400T>C

ENST00000535915.5:c.781T>C

ENST00000545707.5:c.523T>C

ENST00000557933.5:c.904T>C

ENST00000558013.5:c.904T>C

ENST00000558518.5:c.904T>C

ENST00000558528.1:n.419T>C

ENST00000560467.1:c.504T>C

NM_000527.4:c.904T>C

NM_001195798.1:c.904T>C

NM_001195799.1:c.781T>C

NM_001195800.1:c.400T>C

NM_001195803.1:c.523T>C

NM_001195798.2:c.904T>C

NM_001195799.2:c.781T>C

NM_001195800.2:c.400T>C

NM_001195803.2:c.523T>C

Uncertain Significance

Met criteria codes 3

PP3PM1PM2

Not Met criteria codes 19

BS2PVS1BS1BS4BS3BP4BP2BP7PS1PS2PS3PS4BA1PP1PP4PM3PM5PM4PM6

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.904T>C (p.Cys302Arg) variant is classified as Uncertain significance - insufficient evidence for Familial Hypercholesterolemia by applying ACMG/AMP evidence codes PM1, PM2 and PP3 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 version 2.1.1). PP3: REVEL = 0.972. PM1: Variant meets PM2 and alters Cys302, one of the cysteine residues listed.

Met criteria codes

PP3			REVEL = 0.972, this is greater than 0.75.
PM1			Variant meets PM2 and alters Cys302, one of the cysteine residues listed.
PM2			This variant is absent from gnomAD (gnomAD version 2.1.1).

Not Met criteria codes

BS2			No data.
PVS1			Not a null variant.
BS1			This variant is absent from gnomAD (gnomAD version 2.1.1).
BS4			No data.
BS3			No data.
BP4			REVEL = 0.972, this is greater than 0.50.

BP2			No data.
BP7			Not a synonymous variant.
PS1			Variant meets criteria for PM1.
PS2			Not met.
PS3			No data.
PS4			No data.
BA1			This variant is absent from gnomAD (gnomAD version 2.1.1).
PP1			No data.
PP4			No data.
PM3			No data.
PM5			Variant meets criteria for PM1.
PM4			Not an in-frame deletion/insertion.
PM6			Not met.

Curation History 		
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