

Variant: *NM_000527.5(LDLR):c.170A>C (p.Asp57Ala)*

Version: 1.0

CA404075004 [↗](#)

523722 (ClinVar) [↗](#)

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

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

Inheritance Mode: Semidominant inheritance

UUID: cf402cf5-6041-4c9f-8b0b-38094f351c3f

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HGVS expressions

NM_000527.5:c.170A>C

NM_000527.5(LDLR):c.170A>C (p.Asp57Ala)

NC_000019.10:g.11100325A>C

CM000681.2:g.11100325A>C

NC_000019.9:g.11211001A>C

CM000681.1:g.11211001A>C

NC_000019.8:g.11072001A>C

NG_009060.1:g.15945A>C

ENST00000252444.10:c.428A>C

ENST00000559340.2:c.170A>C

ENST00000560467.2:c.170A>C

ENST00000558518.6:c.170A>C

ENST00000252444.9:c.424A>C

ENST00000455727.6:c.170A>C

ENST00000535915.5:c.170A>C

ENST00000545707.5:c.170A>C

ENST00000557933.5:c.170A>C

ENST00000557958.1:n.256A>C

ENST00000558013.5:c.170A>C

ENST00000558518.5:c.170A>C

ENST00000560502.5:n.256A>C

NM_000527.4:c.170A>C

NM_001195798.1:c.170A>C

NM_001195799.1:c.170A>C

NM_001195800.1:c.170A>C

NM_001195803.1:c.170A>C

NM_001195798.2:c.170A>C

NM_001195799.2:c.170A>C

NM_001195800.2:c.170A>C

NM_001195803.2:c.170A>C

Uncertain Significance

Met criteria codes **2**

PP3

PM2

Not Met criteria codes **16**

Expert Panel

Familial Hypercholesterolemia VCEP [↗](#)

Criteria Specification Information **!**

BA1BP4BP2BS1BS4BS3

BS2PP1PP4PM6PM3

PM5PS1PS2PS3PS4

[Criteria Specifications for this VCEP](#)

Evidence Links 0

Evidence submitted by expert panel

Familial Hypercholesterolemia VCEP

The NM_000527.5(LDLR):c.170A>C (p.Asp57Ala) variant is classified as Uncertain significance - insufficient evidence 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.). So PM2 is met. PP3: REVEL= 0.959. It is above 0.75, so PP3 is met.

Met criteria codes

PP3			PP3: REVEL= 0.959. It is above 0.75, so PP3 is met.
PM2			This variant is absent from gnomAD (gnomAD v2.1.1.). So PM2 is met.

Not Met criteria codes

BA1			This variant is absent from gnomAD (gnomAD v2.1.1.).
BP4			PP3: REVEL= 0.959. It is above 0.5
BP2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS1			This variant is absent from gnomAD (gnomAD v2.1.1.).
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
PP1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

