

Variant: NM_000527.5(LDLR):c.1817C>A (p.Ala606Asp)

Version: 1.0

CA404089836 [↗](#)

440663 (ClinVar) [↗](#)

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

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

Inheritance Mode: Semidominant inheritance

UUID: f692c391-d1c9-4835-985a-a0b96c72e904

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HGVS expressions

NM_000527.5:c.1817C>A

NM_000527.5(LDLR):c.1817C>A (p.Ala606Asp)

NC_000019.10:g.11116970C>A

CM000681.2:g.11116970C>A

NC_000019.9:g.11227646C>A

CM000681.1:g.11227646C>A

NC_000019.8:g.11088646C>A

NG_009060.1:g.32590C>A

ENST00000252444.10:c.2075C>A

ENST00000559340.2:c.1705+758C>A

ENST00000560467.2:c.1697C>A

ENST00000558518.6:c.1817C>A

ENST00000252444.9:c.2071C>A

ENST00000455727.6:c.1313C>A

ENST00000535915.5:c.1694C>A

ENST00000545707.5:c.1436C>A

ENST00000557933.5:c.1817C>A

ENST00000558013.5:c.1817C>A

ENST00000558518.5:c.1817C>A

ENST00000559340.1:c.426+758C>A

NM_000527.4:c.1817C>A

NM_001195798.1:c.1817C>A

NM_001195799.1:c.1694C>A

NM_001195800.1:c.1313C>A

NM_001195803.1:c.1436C>A

NM_001195798.2:c.1817C>A

NM_001195799.2:c.1694C>A

NM_001195800.2:c.1313C>A

NM_001195803.2:c.1436C>A

Uncertain Significance

Met criteria codes **2**

PP3 PM2

Not Met criteria codes **1**

PM5

Expert Panel

Familial Hypercholesterolemia VCEP [↗](#)

Criteria Specification Information **!**

Evidence submitted by expert panel

Familial Hypercholesterolemia VCEP

The NM_000527.5 (LDLR): c.1817C>A (p.Ala606Asp) variant is classified as Uncertain significance for Familial Hypercholesterolemia by applying evidence codes (PM2, 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 Met: PopMax MAF = 0.00002637in European (non-Finnish) population in gnomAD (gnomAD v2.1.1). PP3 Met: REVEL = 0.756, which is above the threshold of 0.75. There are two other variants in same codon: LDLR: NM_000527:c.1816G>T (p.Ala606Ser), LDLR: NM_000527:c.1816G>A (p.Ala606Thr), which are classified as Uncertain significance by these guidelines. Therefore PM5 is not met.

Met criteria codes

PP3			REVEL = 0.756, which is above the threshold of 0.75.
PM2			PopMax MAF = 0.00002637in European (non-Finnish) population in gnomAD (gnomAD version 2.1.1).

Not Met criteria codes

PM5			There are two other variants in same codon: LDLR: NM_000527:c.1816G>T (p.Ala606Ser), LDLR: NM_000527:c.1816G>A (p.Ala606Thr), which are classified as Uncertain significance by these guidelines. Therefore PM5 is not met.
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Curation History



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