

Variant: *NM_000527.5(LDLR):c.2051C>T (p.Ala684Val)*

Version: 1.0

CA404093668 [↗](#)

1785078 (ClinVar) [↗](#)

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

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

Inheritance Mode: Semidominant inheritance

UUID: 6fb2c16d-3325-4eb5-a0ba-46d378052c62

Approved on: 2025/2/20

Published on: 2025/6/17

HGVS expressions

NM_000527.5(LDLR):c.2051C>T (p.Ala684Val)

NM_000527.5:c.2051C>T

NC_000019.10:g.11120433C>T

CM000681.2:g.11120433C>T

NC_000019.9:g.11231109C>T

CM000681.1:g.11231109C>T

NC_000019.8:g.11092109C>T

NG_009060.1:g.36053C>T

ENST00000252444.10:c.2309C>T

ENST00000559340.2:c.*120C>T

ENST00000560467.2:c.1931C>T

ENST00000558518.6:c.2051C>T

ENST00000252444.9:c.2305C>T

ENST00000455727.6:c.1547C>T

ENST00000535915.5:c.1928C>T

ENST00000545707.5:c.1606+200C>T

ENST00000557933.5:c.2051C>T

ENST00000558013.5:c.2051C>T

ENST00000558518.5:c.2051C>T

NM_000527.4:c.2051C>T

NM_001195798.1:c.2051C>T

NM_001195799.1:c.1928C>T

NM_001195800.1:c.1547C>T

NM_001195803.1:c.1606+200C>T

NM_001195798.2:c.2051C>T

NM_001195799.2:c.1928C>T

NM_001195800.2:c.1547C>T

NM_001195803.2:c.1606+200C>T

Uncertain Significance

Met criteria codes **2**

PP3

PM2

Not Met criteria codes **1**

PM5

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.2051C>T (p.Ala684Val) variant is classified as Uncertain Significance - insufficient evidence for Familial Hypercholesterolemia by applying ACMG/AMP evidence codes PM2 and PP3 as defined by the ClinGen Familial Hypercholesterolemia Expert Panel LDLR-specific variant curation guidelines (specification version 1.2) on 20 February 2025. The supporting evidence is as follows: **PM2: PopMax MAF=00001667 (0.001667%) in Admixed American exomes+genomes (gnomAD v4.1.0).** **PP3: REVEL score = 0.787.**

Met criteria codes

PP3			REVEL score = 0.787.
PM2			PopMax MAF=00001667 (0.001667%) in Admixed American exomes+genomes (gnomAD v4.1.0).

Not Met criteria codes

PM5			No missense variants at the same codon predict a different amino acid change that is classified as pathogenic.
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Curation History [↗](#)



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