

Variant: *NM_000527.5(LDLR):c.2388C>T (p.Ile796=)*

Version: 1.0

CA040070 [↗](#)

406165 (ClinVar) [↗](#)

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

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

Inheritance Mode: Semidominant inheritance

UUID: 03a6010c-ae44-4b8-8e4f-6b6060b33856

Approved on: 2025/3/28

Published on: 2025/6/30

HGVS expressions

NM_000527.5(LDLR):c.2388C>T (p.Ile796=)

NM_000527.5:c.2388C>T

NC_000019.10:g.11128084C>T

CM000681.2:g.11128084C>T

NC_000019.9:g.11238760C>T

CM000681.1:g.11238760C>T

NC_000019.8:g.11099760C>T

NG_009060.1:g.43704C>T

ENST00000252444.10:c.2646C>T

ENST00000559340.2:c.*457C>T

ENST00000560467.2:c.2268C>T

ENST00000558518.6:c.2388C>T

ENST00000252444.9:c.2642C>T

ENST00000455727.6:c.1884C>T

ENST00000535915.5:c.2265C>T

ENST00000545707.5:c.1854C>T

ENST00000557933.5:c.2388C>T

ENST00000558013.5:c.2388C>T

ENST00000558518.5:c.2388C>T

ENST00000560628.1:n.108+430C>T

NM_000527.4:c.2388C>T

NM_001195798.1:c.2388C>T

NM_001195799.1:c.2265C>T

NM_001195800.1:c.1884C>T

NM_001195803.1:c.1854C>T

NM_001195798.2:c.2388C>T

NM_001195799.2:c.2265C>T

NM_001195800.2:c.1884C>T

NM_001195803.2:c.1854C>T

Uncertain Significance

Met criteria codes **1**

PM2

Not Met criteria codes **5**

BS3

BP4

BP7

PS3

PP3

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.2388C>T (p.Ile796=) variant is classified as Uncertain significance - insufficient evidence for Familial Hypercholesterolemia by applying ACMG/AMP evidence code PM2 as defined by the ClinGen Familial Hypercholesterolemia Expert Panel LDLR-specific variant curation guidelines (specification version 1.2) on 28 March 2025. The supporting evidence is as follows: PM2: PopMax MAF= 0.00004238 (0.004238%) in European non-Finnish exomes + genomes (gnomAD v.4.1.0).

Met criteria codes

PM2			PopMax MAF= 0.00004238 (0.004238%) in European non-Finnish exomes + genomes (gnomAD v.4.1.0).
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Not Met criteria codes

BS3			No functional studies.
BP4			No REVEL, splicing evaluation required. A) located on limits Score in MES: Variant donor= 8.72, WT donor=9.86 Ratio variant / wt = 0.884 B) does not create GT C) not on limits
BP7			Does not meet BP4
PS3			No functional studies.
PP3			No REVEL, splicing evaluation required. A) located on limits Score in MES: Variant donor= 8.72, WT donor=9.86 Ratio variant / wt = 0.884 B) does not create GT C) not on limits

Curation History [↗](#)

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