

Variant: *NM_000527.5(LDLR):c.2397_2412del (p.Val800fs)*

Version: 1.0

CA10585851 [↗](#)

252321 (ClinVar) [↗](#)

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

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

Inheritance Mode: Semidominant inheritance

UUID: e9698f39-b579-4c41-860b-85f710734630

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HGVS expressions

NM_000527.5:c.2397_2412del

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NC_000019.10:g.11129520_11129535del

CM000681.2:g.11129520_11129535del

NC_000019.9:g.11240196_11240211del

CM000681.1:g.11240196_11240211del

NC_000019.8:g.11101196_11101211del

NG_009060.1:g.45140_45155del

ENST00000252444.10:c.2655_2670del

ENST00000559340.2:c.*466_*481del

ENST00000560467.2:c.2277_2292del

ENST00000558518.6:c.2397_2412del

ENST00000252444.9:c.2651_2666del

ENST00000455727.6:c.1893_1908del

ENST00000535915.5:c.2274_2289del

ENST00000545707.5:c.1863_1878del

ENST00000557933.5:c.2459_2474del

ENST00000558013.5:c.2397_2412del

ENST00000558518.5:c.2397_2412del

ENST00000560628.1:n.108+1866_108+1881del

NM_000527.4:c.2397_2412del

NM_001195798.1:c.2397_2412del

NM_001195799.1:c.2274_2289del

NM_001195800.1:c.1893_1908del

NM_001195803.1:c.1863_1878del

NM_001195798.2:c.2397_2412del

NM_001195799.2:c.2274_2289del

NM_001195800.2:c.1893_1908del

NM_001195803.2:c.1863_1878del

Likely Pathogenic

Met criteria codes **2**

PVS1 PM2

Not Met criteria codes **2**

BA1 BS1

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

NM_000527.5(LDLR):c.2397_2412del (p.Val800fs) variant is classified as Likely Pathogenic, for Familial Hypercholesterolemia by applying evidence code PM2 and PVS1 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). . PSV1: Deletion of 16 bp causing an out-of-frame consequence (Exon 17)

Met criteria codes

PVS1			Deletion of 16 bp causing an out-of-frame consequence (Exon 17)
PM2			This variant is absent from gnomAD (gnomAD v2.1.1).

Not Met criteria codes

BA1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History [↗](#)



 

 

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