The ClinGen Evidence Repository is an **FDA-recognized human genetic variant database** containing expert-curated assertions regarding variants' pathogenicity and supporting evidence summaries. [Disclaimer]

Variant: NM_000527.5(LDLR):c.2287dup (p.Glu763fs)

Version: 1.0

CA16602349

375836 (ClinVar)

Gene: LDLR (HGNC:3949)
Condition: hypercholesterolemia, familial (MONDO:0007750)
Inheritance Mode: Semidominant inheritance
UUID: faf42b59-11b0-4332-b9fd-3e72787070ba
Approved on: 2026/1/23
Published on: 2026/2/2

HGVS expressions

NM_000527.5(LDLR):c.2287dup (p.Glu763fs)
NM_000527.5:c.2287dup
NC_000019.10:g.11123320dup
CM000681.2:g.11123320dup
NC_000019.9:g.11233996dup
CM000681.1:g.11233996dup
NC_000019.8:g.11094996dup
NG_009060.1:g.38940dup
ENST00000252444.10:c.2545dup
ENST00000559340.2:c.*356dup
ENST00000560467.2:c.2167dup
ENST00000558518.6:c.2287dup
ENST00000252444.9:c.2541dup
ENST00000455727.6:c.1783dup
ENST00000535915.5:c.2164dup
ENST00000545707.5:c.1753dup
ENST00000557933.5:c.2287dup
ENST00000558013.5:c.2287dup
ENST00000558518.5:c.2287dup
NM_000527.4:c.2287dup
NM_001195798.1:c.2287dup
NM_001195799.1:c.2164dup
NM_001195800.1:c.1783dup
NM_001195803.1:c.1753dup
NM_001195798.2:c.2287dup
NM_001195799.2:c.2164dup
NM_001195800.2:c.1783dup
NM_001195803.2:c.1753dup

Likely Pathogenic

Met criteria codes 2
PVS1 PM2

Not Met criteria codes 3
BS2 BS1 BA1

Expert Panel

Familial Hypercholesterolemia VCEP

Criteria Specification Information

Evidence Links 0

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.2287dup (p.Glu763fs) variant is classified as Likely Pathogenic for Familial Hypercholesterolemia by applying ACMG/AMP evidence codes PVS1 and PM2 as defined by the ClinGen Familial Hypercholesterolemia Expert Panel LDLR-specific variant curation guidelines (specification version 1.2) on January 23, 2026. The supporting evidence is as follows: PM2: This variant is absent from gnomAD (gnomAD v4.1.0). PVS1: Variant is nonsense/frameshift causing a premature stop codon amino-terminal of amino acid 830.

Met criteria codes

PVS1	✔	Variant is nonsense/frameshift causing a premature stop codon amino-terminal of amino acid 830.
PM2	✔	This variant is absent from gnomAD (gnomAD v4.1).

Not Met criteria codes

BS2	✖	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
BA1	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History

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