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.1830_1839del (p.Ala612fs)

Version: 1.0

CA10588898

265904 (ClinVar)

Gene: LDLR (HGNC:3949)
Condition: hypercholesterolemia, familial (MONDO:0007750)
Inheritance Mode: Semidominant inheritance
UUID: cf82bec2-5833-4b2a-a05a-5caef8898450
Approved on: 2026/1/23
Published on: 2026/2/2

HGVS expressions

NM_000527.5(LDLR):c.1830_1839del (p.Ala612fs)
NM_000527.5:c.1830_1839del
NC_000019.10:g.11116983_11116992del
CM000681.2:g.11116983_11116992del
NC_000019.9:g.11227659_11227668del
CM000681.1:g.11227659_11227668del
NC_000019.8:g.11088659_11088668del
NG_009060.1:g.32603_32612del
ENST00000252444.10:c.2088_2097del
ENST00000559340.2:c.1705+771_1705+780del
ENST00000560467.2:c.1710_1719del
ENST00000558518.6:c.1830_1839del
ENST00000252444.9:c.2084_2093del
ENST00000455727.6:c.1326_1335del
ENST00000535915.5:c.1707_1716del
ENST00000545707.5:c.1449_1458del
ENST00000557933.5:c.1830_1839del
ENST00000558013.5:c.1830_1839del
ENST00000558518.5:c.1830_1839del
ENST00000559340.1:c.426+771_426+780del
NM_000527.4:c.1830_1839del
NM_001195798.1:c.1830_1839del
NM_001195799.1:c.1707_1716del
NM_001195800.1:c.1326_1335del
NM_001195803.1:c.1449_1458del
NM_001195798.2:c.1830_1839del
NM_001195799.2:c.1707_1716del
NM_001195800.2:c.1326_1335del
NM_001195803.2:c.1449_1458del

Likely Pathogenic

Met criteria codes 2
PM2 PVS1

Not Met criteria codes 3
PP4 BA1 BS1

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.1830_1839del (p.Ala612fs) 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 frameshift causing a premature stop codon amino-terminal of amino acid 830.

Met criteria codes

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

Not Met criteria codes

PP4	✖	No case/Segregation available
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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