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.2215C>T (p.Gln739Ter)

Version: 1.0

CA10585809

252258 (ClinVar)

Gene: LDLR (HGNC:3949)
Condition: hypercholesterolemia, familial (MONDO:0007750)
Inheritance Mode: Semidominant inheritance
UUID: c4ca2e41-d963-4724-bc04-e9367a9c883c
Approved on: 2022/8/29
Published on: 2022/12/23

HGVS expressions

NM_000527.5:c.2215C>T
NM_000527.5(LDLR):c.2215C>T (p.Gln739Ter)
NC_000019.10:g.11123248C>T
CM000681.2:g.11123248C>T
NC_000019.9:g.11233924C>T
CM000681.1:g.11233924C>T
NC_000019.8:g.11094924C>T
NG_009060.1:g.38868C>T
ENST00000252444.10:c.2473C>T
ENST00000559340.2:c.*284C>T
ENST00000560467.2:c.2095C>T
ENST00000558518.6:c.2215C>T
ENST00000252444.9:c.2469C>T
ENST00000455727.6:c.1711C>T
ENST00000535915.5:c.2092C>T
ENST00000545707.5:c.1681C>T
ENST00000557933.5:c.2215C>T
ENST00000558013.5:c.2215C>T
ENST00000558518.5:c.2215C>T
NM_000527.4:c.2215C>T
NM_001195798.1:c.2215C>T
NM_001195799.1:c.2092C>T
NM_001195800.1:c.1711C>T
NM_001195803.1:c.1681C>T
NM_001195798.2:c.2215C>T
NM_001195799.2:c.2092C>T
NM_001195800.2:c.1711C>T
NM_001195803.2:c.1681C>T

Pathogenic

Met criteria codes 3
PVS1 PM2 PP4

Not Met criteria codes 2
PS3 PS4

Expert Panel

Familial Hypercholesterolemia VCEP

Criteria Specification Information

Criteria Specifications for this VCEP

Evidence Links 0

Evidence submitted by expert panel

Familial Hypercholesterolemia VCEP

The NM_000527.5(LDLR) :c.2215C>T (p.Gln739Ter) variant is classified as Pathogenic for Familial Hypercholesterolemia by applying evidence code PVS1, PM2 and PP4 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). PVS1 - Stop in codon 739. It is upstream of amino acid 830. PP4 - Variant meets PM2 and is identified in one index case who fulfill FH/DLCN>=6 criteria for FH from Robarts Research Institute after alternative causes of high cholesterol were excluded.

Met criteria codes

PVS1	✔	Stop in codon 739. It is upstream of amino acid 830.
PM2	✔	This variant is absent from gnomAD (gnomAD v2.1.1).
PP4	✔	Variant meets PM2 and is identified in one index case who fulfill FH/DLCN>=6 criteria for FH from Robarts Research Institute after alternative causes of high cholesterol were excluded.

Not Met criteria codes

PS3	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS4	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History

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