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.1577C>A (p.Pro526His)

Version: 1.0

CA404086609

496019 (ClinVar)

Gene: LDLR (HGNC:3949)
Condition: hypercholesterolemia, familial (MONDO:0007750)
Inheritance Mode: Semidominant inheritance
UUID: e76857b0-1c5f-44bc-b84b-9aca1b9395c6
Approved on: 2022/1/18
Published on: 2022/6/30

HGVS expressions

NM_000527.5(LDLR):c.1577C>A (p.Pro526His)
NM_000527.5:c.1577C>A
NC_000019.10:g.11113753C>A
CM000681.2:g.11113753C>A
NC_000019.9:g.11224429C>A
CM000681.1:g.11224429C>A
NC_000019.8:g.11085429C>A
NG_009060.1:g.29373C>A
ENST00000252444.10:c.1835C>A
ENST00000559340.2:c.1577C>A
ENST00000560467.2:c.1457C>A
ENST00000558518.6:c.1577C>A
ENST00000252444.9:c.1831C>A
ENST00000455727.6:c.1073C>A
ENST00000535915.5:c.1454C>A
ENST00000545707.5:c.1196C>A
ENST00000557933.5:c.1577C>A
ENST00000558013.5:c.1577C>A
ENST00000558518.5:c.1577C>A
ENST00000559340.1:c.298C>A
NM_000527.4:c.1577C>A
NM_001195798.1:c.1577C>A
NM_001195799.1:c.1454C>A
NM_001195800.1:c.1073C>A
NM_001195803.1:c.1196C>A
NM_001195798.2:c.1577C>A
NM_001195799.2:c.1454C>A
NM_001195800.2:c.1073C>A
NM_001195803.2:c.1196C>A

Uncertain Significance

Met criteria codes 2

PP3 PM2

Not Met criteria codes 24

Expert Panel

Familial Hypercholesterolemia VCEP

Criteria Specification Information

PS1 PS2 PS3 PS4 BA1 PP1
PP2 PP4 PM1 PM3 PM5
PM4 PM6 BS1 BS4 BS3
BS2 BP5 BP7 BP4 BP3 BP1
BP2 PVS1

Evidence Links 0

Criteria Specifications for this VCEP

Evidence submitted by expert panel

Familial Hypercholesterolemia VCEP

NM_000527.5(LDLR):c.1577C>A (p.Pro526His) variant is classified as variant of Uncertain significance for Familial Hypercholesterolemia by applying evidence codes (PM2 and PP3) 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). PP3 - REVEL = 0.946.

Met criteria codes

PP3	✔	REVEL = 0.946
PM2	✔	This variant is absent from gnomAD (gnomAD v2.1.1).

Not Met criteria codes

PS1	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS2	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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
BA1	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP1	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP2	✖	

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

PP4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM5			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM6			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
BS4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP5			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP7			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

BP1		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP2		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PVS1		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History

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