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.797A>T (p.Asp266Val)

Version: 1.0

CA10585137

251458 (ClinVar)

Gene: LDLR (HGNC:3949)
Condition: hypercholesterolemia, familial (MONDO:0007750)
Inheritance Mode: Semidominant inheritance
UUID: 1170e266-2790-4fd8-9f99-c5add945f0d0
Approved on: 2021/11/14
Published on: 2022/6/30

HGVS expressions

NM_000527.5:c.797A>T
NM_000527.5(LDLR):c.797A>T (p.Asp266Val)
NC_000019.10:g.11106667A>T
CM000681.2:g.11106667A>T
NC_000019.9:g.11217343A>T
CM000681.1:g.11217343A>T
NC_000019.8:g.11078343A>T
NG_009060.1:g.22287A>T
ENST00000252444.10:c.1055A>T
ENST00000559340.2:c.797A>T
ENST00000560467.2:c.797A>T
ENST00000558518.6:c.797A>T
ENST00000252444.9:c.1051A>T
ENST00000455727.6:c.314-725A>T
ENST00000535915.5:c.674A>T
ENST00000545707.5:c.416A>T
ENST00000557933.5:c.797A>T
ENST00000558013.5:c.797A>T
ENST00000558518.5:c.797A>T
ENST00000558528.1:n.312A>T
ENST00000560467.1:c.397A>T
NM_000527.4:c.797A>T
NM_001195798.1:c.797A>T
NM_001195799.1:c.674A>T
NM_001195800.1:c.314-725A>T
NM_001195803.1:c.416A>T
NM_001195798.2:c.797A>T
NM_001195799.2:c.674A>T
NM_001195800.2:c.314-725A>T
NM_001195803.2:c.416A>T

Likely Pathogenic

Met criteria codes 3

PP3 PM2 PM5_Strong

Not Met criteria codes 23

Expert Panel

Familial Hypercholesterolemia VCEP

Criteria Specification Information

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

Evidence Links 0

Criteria Specifications for this VCEP

Evidence submitted by expert panel

Familial Hypercholesterolemia VCEP

The NM_000527.5(LDLR): c.797A>T (p.Asp266Val) variant is classified as Likely pathogenic for Familial Hypercholesterolemia by applying evidence codes PM5_Strong, PM2, and PP3 as defined by the ClinGen Familial Hypercholesterolemia Expert Panel LDLR-specific variant curation guidelines (https://doi.org/10.1101/2021.03.17.21252755). PM5_Strong - 4 other missense variants in the same codon: - NM_000527.5(LDLR): c.796G>T (p.Asp266Asn) (ClinVar ID 226334) - Pathogenic by these guidelines, - NM_000527.5(LDLR): c.797A>G (p.Asp266Gly) (ClinVar ID 251458) – Likely pathogenic by these guidelines, - NM_000527.5(LDLR): c.796A>T (p.Asp266Tyr) (ClinVar ID 251456) – Pathogenic by these guidelines, - NM_000527.5(LDLR): c.798T>A (p.Asp266Glu) - (ClinVar ID 161287) - Pathogenic by these guidelines. There are 3 variants in the same codon classified as Pathogenic by these guidelines; PM2 - This variant is absent from gnomAD (gnomAD v2.1.1); PP3 - REVEL = 0.97;

Met criteria codes

PP3	✔	REVEL = 0.97
PM2	✔	This variant is absent from gnomAD (gnomAD v2.1.1)
PM5_Strong	✔	4 other missense variants in the same codon: - NM_000527.5(LDLR): c.796G>T (p.Asp266Asn) (ClinVar ID 226334) - Pathogenic by these guidelines, - NM_000527.5(LDLR): c.797A>G (p.Asp266Gly) (ClinVar ID 251458) – Likely pathogenic by these guidelines, - NM_000527.5(LDLR): c.796A>T (p.Asp266Tyr) (ClinVar ID 251456) – Pathogenic by these guidelines, - NM_000527.5(LDLR): c.798T>A (p.Asp266Glu) - (ClinVar ID 161287) - Pathogenic by these guidelines. There are 3 variants in the same codon classified as Pathogenic by these guidelines;

Not Met criteria codes

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
BA1	✖	

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
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
PVS1		✖	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
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
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
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

Curation History

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