

Variant: NM_000527.5(LDLR):c.854A>G (p.His285Arg)

Version: 1.0

CA404080674 [↗](#)

450684 (ClinVar) [↗](#)

Gene: LDLR ([HGNC:3949](#))

Condition: hypercholesterolemia, familial ([MONDO:0007750](#))

Inheritance Mode: Semidominant inheritance

UUID: 1e59e697-8a4f-42d0-b9c5-16127233c7b4

Approved on: 2023/3/24

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HGVS expressions

NM_000527.5(LDLR):c.854A>G (p.His285Arg)

NM_000527.5:c.854A>G

NC_000019.10:g.11107428A>G

CM000681.2:g.11107428A>G

NC_000019.9:g.11218104A>G

CM000681.1:g.11218104A>G

NC_000019.8:g.11079104A>G

NG_009060.1:g.23048A>G

ENST00000252444.10:c.1112A>G

ENST00000559340.2:c.854A>G

ENST00000560467.2:c.854A>G

ENST00000558518.6:c.854A>G

ENST00000252444.9:c.1108A>G

ENST00000455727.6:c.350A>G

ENST00000535915.5:c.731A>G

ENST00000545707.5:c.473A>G

ENST00000557933.5:c.854A>G

ENST00000558013.5:c.854A>G

ENST00000558518.5:c.854A>G

ENST00000558528.1:n.369A>G

ENST00000560467.1:c.454A>G

NM_000527.4:c.854A>G

NM_001195798.1:c.854A>G

NM_001195799.1:c.731A>G

NM_001195800.1:c.350A>G

NM_001195803.1:c.473A>G

NM_001195798.2:c.854A>G

NM_001195799.2:c.731A>G

NM_001195800.2:c.350A>G

NM_001195803.2:c.473A>G

Uncertain Significance

Met criteria codes 2

BP4

PM2

Not Met criteria codes 23

BA1

BS2

BS1

BS4

BS3

BP3

BP1

BP2

PVS1

BP7

PS1

PS2

PS3

PS4

PP1

PP2

PP3

PP4

PM1

PM3

PM5

PM4

PM6

Evidence Links 0

Expert Panel

Familial Hypercholesterolemia VCEP

Criteria Specification Information

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.854A>G (p.His285Arg) variant is classified as Uncertain significance - insufficient evidence for Familial Hypercholesterolemia by applying ACMG/AMP evidence codes PM2 and BP4 as defined by the ClinGen Familial Hypercholesterolemia Expert Panel LDLR-specific variant curation guidelines (specification version 1.2) on 24 March 2023. The supporting evidence is as follows: PM2: This variant is absent from gnomAD (v2.1.1). BP4: REVEL = 0.419, splicing evaluation required. A.) Not on limits. B.) Does not create AG. C.) There is an AG nearby. Cryptic scores are negative, so cryptic site is not used - variant is not predicted to alter splicing. BP4 is met.

Met criteria codes

BP4

REVEL = 0.419, splicing evaluation required. The variant is not predicted to affect splicing.

PM2

This variant is absent from gnomAD (v2.1.1).

Not Met criteria codes

BA1

Not met.

BS2

Not met.

BS1

Not met.

BS4

Not met.

BS3

Not met.

BP3

Not applicable

BP1

Not applicable

BP2			Not met.
PVS1			Not a null variant.
BP7			Not a synonymous variant.
PS1			Not met.
PS2			Not met.
PS3			Not met.
PS4			Not met.
PP1			Not met.
PP2			Not applicable
PP3			REVEL = 0.419, splicing evaluation required. Neither scenario A, B or C is met. The variant is not predicted to affect splicing.
PP4			Not met.
PM1			Not located in exon 4 or at a cysteine residue.
PM3			Not met.
PM5			Not met.
PM4			Not an in-frame deletion/insertion.
PM6			Not met.

Curation History [↗](#)

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