

Variant: NM_000527.5(LDLR):c.*13A>G

Version: 1.0

CA032236 [↗](#)

265909 (ClinVar) [↗](#)

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

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

Inheritance Mode: Semidominant inheritance

UUID: ae223342-a204-4cd9-aa08-f14be46a3820

Approved on: 2024/10/28

Published on: 2025/1/19

HGVS expressions

NM_000527.5:c.*13A>G

NM_000527.5(LDLR):c.*13A>G

NC_000019.10:g.11131329A>G

CM000681.2:g.11131329A>G

NC_000019.9:g.11242005A>G

CM000681.1:g.11242005A>G

NC_000019.8:g.11103005A>G

NG_009060.1:g.46949A>G

ENST00000252444.10:c.*13A>G

ENST00000559340.2:c.*665A>G

ENST00000560467.2:c.*13A>G

ENST00000558518.6:c.*13A>G

ENST00000252444.9:c.2850A>G

ENST00000455727.6:c.*13A>G

ENST00000535915.5:c.*13A>G

ENST00000545707.5:c.*13A>G

ENST00000557933.5:c.2658A>G

ENST00000558013.5:c.*13A>G

ENST00000558518.5:c.*13A>G

ENST00000560628.1:n.109-1766A>G

NM_000527.4:c.*13A>G

NM_001195798.1:c.*13A>G

NM_001195799.1:c.*13A>G

NM_001195800.1:c.*13A>G

NM_001195803.1:c.*13A>G

NM_001195798.2:c.*13A>G

NM_001195799.2:c.*13A>G

NM_001195800.2:c.*13A>G

NM_001195803.2:c.*13A>G

Benign

Met criteria codes **1**

BA1

Not Met criteria codes **7**

Expert Panel

Familial Hypercholesterolemia VCEP [↗](#)

Criteria Specification Information

BP4

BS1

BS2

PP3

PP4

PS4

PM2

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.*13A>G variant is classified as Benign for Familial Hypercholesterolemia by applying ACMG/AMP evidence code BA1 as defined by the ClinGen Familial Hypercholesterolemia Expert Panel LDLR-specific variant curation guidelines (specification version 1.2) on 28 October 2024. The supporting evidence is as follows: BA1: FAF = 0.007871(0.7871%) in African/African American exomes (gnomAD v4.1.0).

Met criteria codes

BA1

FAF = 0.007871(0.7871%) in African/African American exomes (gnomAD v4.101).

Not Met criteria codes

BP4

Variant is not a Missense variant

BS1

FAF = 0.007871(0.7871%) in African/African American exomes (gnomAD v4.101).

BS2

Variant not identified in any normolipidemic individuals

PP3

Variant is not a Missense variant

PP4

Variant does not meet PM2

PS4

Variant does not meet PM2

PM2

PopMax MAF = 0.008567 (0.8567%) in African/African American exomes+genomes (gnomAD v4.1.0).

Curation History [↗](#)

Showing 1 to 1 of 1 rows

The information on this website is not intended for direct diagnostic use or medical decision-making without review by a genetics professional. Individuals should not change their health behavior solely on the basis of information contained on this website. If you have questions about the information contained on this website, please see a health care professional.