

Variant: NM_000527.5(LDLR):c.1194C>T (p.Ile398=)

Version: 1.2

CA033001 [↗](#)

224620 (ClinVar) [↗](#)

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

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

Inheritance Mode: Semidominant inheritance

UUID: 8ef9f316-9ab5-49b9-8eb2-cea7a1ccbdb5

Approved on: 2022/7/22

Published on: 2024/12/2

HGVS expressions

NM_000527.5:c.1194C>T

NM_000527.5(LDLR):c.1194C>T (p.Ile398=)

NC_000019.10:g.11113285C>T

CM000681.2:g.11113285C>T

NC_000019.9:g.11223961C>T

CM000681.1:g.11223961C>T

NC_000019.8:g.11084961C>T

NG_009060.1:g.28905C>T

ENST00000252444.10:c.1452C>T

ENST00000559340.2:c.1194C>T

ENST00000560467.2:c.1074C>T

ENST00000558518.6:c.1194C>T

ENST00000252444.9:c.1448C>T

ENST00000455727.6:c.690C>T

ENST00000535915.5:c.1071C>T

ENST00000545707.5:c.813C>T

ENST00000557933.5:c.1194C>T

ENST00000558013.5:c.1194C>T

ENST00000558518.5:c.1194C>T

ENST00000560173.1:n.193C>T

ENST00000560467.1:c.674C>T

NM_000527.4:c.1194C>T

NM_001195798.1:c.1194C>T

NM_001195799.1:c.1071C>T

NM_001195800.1:c.690C>T

NM_001195803.1:c.813C>T

NM_001195798.2:c.1194C>T

NM_001195799.2:c.1071C>T

NM_001195800.2:c.690C>T

NM_001195803.2:c.813C>T

Likely Benign

Met criteria codes 3

BS1BP4BP7

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.1194C>T (p.Ile398=) variant is classified as Likely Benign for Familial Hypercholesterolemia by applying ACMG/AMP evidence codes BS1, BP4 and BP7 as defined by the ClinGen Familial Hypercholesterolemia Expert Panel LDLR-specific variant curation guidelines (specification version 1.2) on 22 July 2022. The supporting evidence is as follows: BS1 - FAF = 0.00448 (0.448%) in East Asian exomes (gnomAD v2.1.1). BP4 - No REVEL, splicing evaluation required. Functional data on splicing not available. Variant is not located within the limits of canonical splicing sites Variant is exonic and at least 50bp upstream from canonical donor site but does not creates AG There is no AG Nearby Variant is not predicted to alter splicing. BP7 - Variant is synonymous and meets BP4

Met criteria codes

BS1			FAF = 0.00448 (0.448%) in East Asian exomes (gnomAD v2.1.1).
BP4			No REVEL, splicing evaluation required. Functional data on splicing not available. Variant is not located within the limits of canonical plicing sites Variant is exonic and at least 50bp upstream from canonical donor site but does not creates AG There is no AG Nearby Variant is not predicted to alter splicing.
BP7			Variant is synonymous and meets BP4.

Curation History

Showing 1 to 3 of 3 rows

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