

Variant: NM_000527.5(LDLR):c.1403T>A (p.Val468Asp)

Version: 1.0

CA10585439 [↗](#)

251828 (ClinVar) [↗](#)

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

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

Inheritance Mode: Semidominant inheritance

UUID: b3c1c7a2-7cb3-4ef1-a039-967c6020a365

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HGVS expressions

NM_000527.5(LDLR):c.1403T>A (p.Val468Asp)

NM_000527.5:c.1403T>A

NC_000019.10:g.11113579T>A

CM000681.2:g.11113579T>A

NC_000019.9:g.11224255T>A

CM000681.1:g.11224255T>A

NC_000019.8:g.11085255T>A

NG_009060.1:g.29199T>A

ENST00000252444.10:c.1661T>A

ENST00000559340.2:c.1403T>A

ENST00000560467.2:c.1283T>A

ENST00000558518.6:c.1403T>A

ENST00000252444.9:c.1657T>A

ENST00000455727.6:c.899T>A

ENST00000535915.5:c.1280T>A

ENST00000545707.5:c.1022T>A

ENST00000557933.5:c.1403T>A

ENST00000558013.5:c.1403T>A

ENST00000558518.5:c.1403T>A

ENST00000559340.1:c.124T>A

ENST00000560467.1:c.883T>A

NM_000527.4:c.1403T>A

NM_001195798.1:c.1403T>A

NM_001195799.1:c.1280T>A

NM_001195800.1:c.899T>A

NM_001195803.1:c.1022T>A

NM_001195798.2:c.1403T>A

NM_001195799.2:c.1280T>A

NM_001195800.2:c.899T>A

NM_001195803.2:c.1022T>A

Uncertain Significance

Met criteria codes 3

PP3PP4PM2

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.1403T>A (p.Val468Asp) variant is classified as Uncertain significance - insufficient evidence for Familial Hypercholesterolemia by applying evidence codes (PM2, PP3, PP4) as defined by the ClinGen Familial Hypercholesterolemia Expert Panel LDLR-specific variant curation guidelines (https://doi.org/10.1101/2021.03.17.21252755). The supporting evidence is as follows: PM2 - This variant is absent from gnomAD (gnomAD v2.1.1). PP3 - REVEL = 0.823 PP4 - Variant meet PM2. PMID: 20828696 (Medeiros et al., 2010), 1 case who fulfills Simon-Broome criteria for FH, after alternative causes of high cholesterol have been excluded.

Met criteria codes

PP3	i ✓	PP3 - REVEL = 0.823
PP4	i ✓	PP4 - Variant meet PM2. PMID: 20828696 (Medeiros et al., 2010), 1 case who fulfills Simon-Broome criteria for FH, after alternative causes of high cholesterol have been excluded.
PM2	i ✓	PM2 - This variant is absent from gnomAD (gnomAD v2.1.1).

Curation History

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