

Variant: *NM_000527.5(LDLR):c.298G>A (p.Asp100Asn)*

Version: 1.0

CA10584832 [↗](#)

251121 (ClinVar) [↗](#)

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

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

Inheritance Mode: Semidominant inheritance

UUID: 2a5e70a7-3328-461c-97ec-ad2a36bcd68d

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HGVS expressions

NM_000527.5(LDLR):c.298G>A (p.Asp100Asn)

NM_000527.5:c.298G>A

NC_000019.10:g.11102771G>A

CM000681.2:g.11102771G>A

NC_000019.9:g.11213447G>A

CM000681.1:g.11213447G>A

NC_000019.8:g.11074447G>A

NG_009060.1:g.18391G>A

ENST00000252444.10:c.556G>A

ENST00000559340.2:c.298G>A

ENST00000560467.2:c.298G>A

ENST00000558518.6:c.298G>A

ENST00000252444.9:c.552G>A

ENST00000455727.6:c.298G>A

ENST00000535915.5:c.190+2426G>A

ENST00000545707.5:c.298G>A

ENST00000557933.5:c.298G>A

ENST00000557958.1:n.384G>A

ENST00000558013.5:c.298G>A

ENST00000558518.5:c.298G>A

NM_000527.4:c.298G>A

NM_001195798.1:c.298G>A

NM_001195799.1:c.190+2426G>A

NM_001195800.1:c.298G>A

NM_001195803.1:c.298G>A

NM_001195798.2:c.298G>A

NM_001195799.2:c.190+2426G>A

NM_001195800.2:c.298G>A

NM_001195803.2:c.298G>A

Likely Pathogenic

Met criteria codes **6**

PP1

PP3

PP4

PM2

PM3

PS4_Supporting

Evidence Links **0**

Expert Panel

Familial Hypercholesterolemia VCEP [↗](#)

Criteria Specification Information

Evidence submitted by expert panel

Familial Hypercholesterolemia VCEP

The NM_000527.5(LDLR):c.298G>A (p.Asp100Asn) variant is classified as Likely Pathogenic for Familial Hypercholesterolemia by applying evidence codes PM2, PM3, PP1, PP3, PP4, PS4_supporting as defined by the ClinGen Familial Hypercholesterolemia Expert Panel LDLR-specific variant curation guidelines (<https://doi.org/10.1016/j.gim.2021.09.012>). The supporting evidence is as follows: - PM2: This variant is absent from gnomAD (gnomAD v2.1.1) - PM3: Variant meets PM2 and is identified in an index case with homozygous FH phenotype (LDL-C=10.25 mmol/L) as homozygous status, from PMID: 25936346. - PP1: Variant segregates with FH phenotype in at least 2 informative meioses from 1 family from PMID: 25936346 - PP3: REVEL = 0.93 - PP4: Variant meets PM2 and is identified in at least 1 index case who fulfills SB possible FH from Ambry Genetics, after alternative causes for high cholesterol were excluded. - PS4_supporting: Variant meets PM2 and is identified in 2 index cases (1 case with Simon-Broome criteria of possible FH from Ambry Genetics, 1 cases with Simon-Broome criteria of possible FH from PMID: 25936346)

Met criteria codes

PP1			Variant segregates with FH phenotype in at least 2 informative meioses from 1 family from PMID: 25936346
PP3			REVEL = 0.93
PP4			Variant meets PM2 and is identified in at least 1 index case who fulfills SB possible FH from Ambry Genetics, after alternative causes for high cholesterol were excluded.
PM2			This variant is absent from gnomAD (gnomAD v2.1.1)
PM3			Variant meets PM2 and is identified in an index case with homozygous FH phenotype (LDL-C=10.25 mmol/L) as homozygous status, from PMID: 25936346.
PS4_Supporting			Variant meets PM2 and is identified in 2 index cases (1 case with Simon-Broome criteria of possible FH from Ambry Genetics, 1 cases with Simon-Broome criteria of possible FH from PMID: 25936346)

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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.