The ClinGen Evidence Repository is an **FDA-recognized human genetic variant database** containing expert-curated assertions regarding variants' pathogenicity and supporting evidence summaries. [Disclaimer]

Variant: NM_000527.5(LDLR):c.239A>G (p.Asn80Ser)

Version: 1.0

CA404075595

923296 (ClinVar)

Gene: LDLR (HGNC:3949)
Condition: hypercholesterolemia, familial (MONDO:0007750)
Inheritance Mode: Semidominant inheritance
UUID: 2c91e453-6e50-4a68-b3e1-c61db58864f3
Approved on: 2022/8/29
Published on: 2022/12/23

HGVS expressions

NM_000527.5:c.239A>G
NM_000527.5(LDLR):c.239A>G (p.Asn80Ser)
NC_000019.10:g.11102712A>G
CM000681.2:g.11102712A>G
NC_000019.9:g.11213388A>G
CM000681.1:g.11213388A>G
NC_000019.8:g.11074388A>G
NG_009060.1:g.18332A>G
ENST00000252444.10:c.497A>G
ENST00000559340.2:c.239A>G
ENST00000560467.2:c.239A>G
ENST00000558518.6:c.239A>G
ENST00000252444.9:c.493A>G
ENST00000455727.6:c.239A>G
ENST00000535915.5:c.190+2367A>G
ENST00000545707.5:c.239A>G
ENST00000557933.5:c.239A>G
ENST00000557958.1:n.325A>G
ENST00000558013.5:c.239A>G
ENST00000558518.5:c.239A>G
NM_000527.4:c.239A>G
NM_001195798.1:c.239A>G
NM_001195799.1:c.190+2367A>G
NM_001195800.1:c.239A>G
NM_001195803.1:c.239A>G
NM_001195798.2:c.239A>G
NM_001195799.2:c.190+2367A>G
NM_001195800.2:c.239A>G
NM_001195803.2:c.239A>G

Uncertain Significance

Met criteria codes 2

PM2 BP4

Evidence Links 0

Expert Panel

Familial Hypercholesterolemia VCEP

Criteria Specification Information

Criteria Specifications for this VCEP

Evidence submitted by expert panel

Familial Hypercholesterolemia VCEP

The NM_000527.5(LDLR):c.239A>G (p.Asn80Ser)variant is classified as Uncertain significance - insufficient evidence for Familial Hypercholesterolemia by applying evidence code PM2, BP4 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) BP4 - REVEL = 0.176. It is below 0.50. splicing evaluation required. Functional data not available A) not on limits B) variant is exonic and at least 50bp upstream from canonical donor site and creates AG MES scores: de novo variant = -2.32; canonical acceptor = 7.07. Ratio de novo variant/canonical donor = -2.32/7.07 --- de novo score is negative, so it is not used. Variant is not predicted to alter splicing.

Met criteria codes

PM2	✔	PM2 - This variant is absent from gnomAD (gnomAD v2.1.1)
BP4	✔	BP4 - REVEL = 0.176. It is below 0.50. splicing evaluation required. Functional data not available A) not on limits B) variant is exonic and at least 50bp upstream from canonical donor site and creates AG MES scores: de novo variant = -2.32; canonical acceptor = 7.07. Ratio de novo variant/canonical donor = -2.32/7.07 --- de novo score is negative, so it is not used. Variant is not predicted to alter splicing.

Curation History

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