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_004360.5(CDH1):c.2311C>T (p.Gln771Ter)

Version: 3.0

CA396470822

428624 (ClinVar)

Gene: CDH1 (HGNC:999)
Condition: CDH1-related diffuse gastric and lobular breast cancer (MONDO:0100488)
Inheritance Mode: Autosomal dominant inheritance
UUID: a900acc8-7a69-4034-a12b-01fa5d64d142
Approved on: 2023/8/4
Published on: 2023/8/4

HGVS expressions

NM_004360.5:c.2311C>T
NM_004360.5(CDH1):c.2311C>T (p.Gln771Ter)
NC_000016.10:g.68829669C>T
CM000678.2:g.68829669C>T
NC_000016.9:g.68863572C>T
CM000678.1:g.68863572C>T
NC_000016.8:g.67421073C>T
NG_008021.1:g.97378C>T
ENST00000261769.10:c.2311C>T
ENST00000261769.9:c.2311C>T
ENST00000422392.6:c.2128C>T
ENST00000562118.1:n.529C>T
ENST00000562836.5:n.2382C>T
ENST00000566510.5:c.*977C>T
ENST00000566612.5:c.*551C>T
ENST00000611625.4:c.2374C>T
ENST00000612417.4:c.1853+3115C>T
ENST00000621016.4:c.1866-4534C>T
NM_004360.3:c.2311C>T
NM_001317184.1:c.2128C>T
NM_001317185.1:c.763C>T
NM_001317186.1:c.346C>T
NM_004360.4:c.2311C>T
NM_001317184.2:c.2128C>T
NM_001317185.2:c.763C>T
NM_001317186.2:c.346C>T

Pathogenic

Met criteria codes 3

PVS1 PM5_Supporting PM2_Supporting

Not Met criteria codes 23

PM1 PM3 PM4 PM6 BA1 BS2 BS1 BS4 BS3 BP5 BP7

Expert Panel

CDH1 VCEP

Criteria Specification Information

Criteria Specification: *ClinGen CDH1 Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines Version 3.1*

BP4 BP3 BP1 BP2 PS1 PS2
PS3 PS4 PP1 PP2 PP3 PP4

Criteria Specification Approval History
Criteria Specifications for this VCEP

Evidence Links 0

Evidence submitted by expert panel

CDH1 VCEP

The c.2311C>T (p.Gln771Ter) variant is predicted to result in a premature stop codon that leads to nonsense mediate decay (PVS1 and PM5_supporting). The variant is absent in the gnomAD cohort (PM2_supporting; http://gnomad.broadinstitute.org). Therefore, this variant meets criteria to be classified as pathogenic based on the ACMG/AMP criteria applied as specified by the CDH1 Variant Curation Expert Panel (CDH1 VCEP specifications version 3.1): PVS1, PM2_supporting, PM5_supporting.

Met criteria codes

PVS1	✔	Premature termination occurs upstream of the NMD boundary
PM5_Supporting	✔	Premature termination occurs upstream of the NMD boundary
PM2_Supporting	✔	Absent in gnomAD population database

Not Met criteria codes

PM1	✖	Do not use for this gene
PM3	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM4	✖	variant is predicted to result in a premature stop codon
PM6	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BA1	✖	Absent in gnomAD population database
BS2	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS1	✖	Absent in gnomAD population database
BS4	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

BS3	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP5	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP7	ℹ	✖	variant is predicted to result in a premature stop codon
BP4	ℹ	✖	variant is predicted to result in a premature stop codon
BP3		✖	variant is predicted to result in a premature stop codon
BP1		✖	variant is predicted to result in a premature stop codon
BP2	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS1		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS2	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS3	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS4	ℹ	✖	Ambry: Proband 1: F with gastric cancer in 30s. Mother with stomach cancer in 20s. MGM with colon cancer in 70s. (not meet due to lack of pathology information)
PP1	ℹ	✖	applied PS4
PP2		✖	variant is predicted to result in a premature stop codon
PP3	ℹ	✖	variant is predicted to result in a premature stop codon
PP4		✖	Use PS4 in place of PP4

Curation History

Showing 1 to 3 of 3 rows

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