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_001317186.2:c.-1818T>A

Version: 2.0

CA396451188

1292057 (ClinVar)

Gene: CDH1 (HGNC:999)
Condition: CDH1-related diffuse gastric and lobular breast cancer (MONDO:0100488)
Inheritance Mode: Autosomal dominant inheritance
UUID: 4298342a-f7ef-461f-bdcb-ce5a49048165
Approved on: 2023/8/25
Published on: 2023/8/25

HGVS expressions

NM_001317186.2:c.-1818T>A
NC_000016.10:g.68737417T>A
CM000678.2:g.68737417T>A
NC_000016.9:g.68771320T>A
CM000678.1:g.68771320T>A
NC_000016.8:g.67328821T>A
NG_008021.1:g.5126T>A
ENST00000261769.10:c.2T>A
ENST00000261769.9:c.2T>A
ENST00000422392.6:c.2T>A
ENST00000566510.5:c.2T>A
ENST00000566612.5:c.2T>A
ENST00000611625.4:c.2T>A
ENST00000612417.4:c.2T>A
ENST00000621016.4:c.2T>A
NM_004360.3:c.2T>A
NM_001317184.1:c.2T>A
NM_001317185.1:c.-1614T>A
NM_001317186.1:c.-1818T>A
NM_004360.4:c.2T>A
NM_004360.5:c.2T>A
NM_001317184.2:c.2T>A
NM_001317185.2:c.-1614T>A

Pathogenic

Met criteria codes 3

PVS1 PM2_Supporting PS4_Supporting

Not Met criteria codes 23

PM1 PM3 PM5 PM4 PM6 BA1 BS2 BS1 BS4 BS3 BP5 BP7 BP4 BP3 BP1 BP2 PS1 PS2 PS3 PP1 PP2 PP3 PP4

Evidence Links 0

Expert Panel

CDH1 VCEP

Criteria Specification Information

- **Criteria Specification:** *ClinGen CDH1 Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines Version 3.1*
- **Criteria Specification Approval History**
- **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

CDH1 VCEP

The c.2T>A (p.Met1?) variant alters the start codon of the CDH1 coding sequence and is predicted to lead to an absent protein (PVS1). This variant is absent in the gnomAD cohort (PM2_supporting; http://gnomad.broadinstitute.org). This variant has been reported in at least one proband meeting HDGC clinical criteria (PS4_supporting; Internal laboratory contributor). In summary, this variant meets criteria to be classified as pathogenic based on the ACMG/AMP Variant Interpretation Guidelines Version 3.1 as specified by the CDH1 Variant Curation Expert Panel: PVS1, PM2_supporting, PS4_supporting.

Met criteria codes

PVS1	✔	PVS1 applies to initiation codon variants
PM2_Supporting	✔	Absent from all population databases
PS4_Supporting	✔	One proband meet HDGC phenotype criteria (Internal laboratory contributor).

Not Met criteria codes

PM1	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM3	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM5	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM4	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM6	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BA1	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS2	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS1	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

BS4	ℹ	✖	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	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP4	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP3		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP1		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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
PP1	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP2		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP3	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP4		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History

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