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.1A>G (p.Met1Val)

Version: 2.0

CA396451176

532457 (ClinVar)

Gene: CDH1 (HGNC:999)
Condition: CDH1-related diffuse gastric and lobular breast cancer (MONDO:0100488)
Inheritance Mode: Autosomal dominant inheritance
UUID: 6d19d5b0-c34b-4794-89e5-d0173e561a83
Approved on: 2023/8/25
Published on: 2023/8/25

HGVS expressions

NM_004360.5:c.1A>G
NM_004360.5(CDH1):c.1A>G (p.Met1Val)
NC_000016.10:g.68737416A>G
CM000678.2:g.68737416A>G
NC_000016.9:g.68771319A>G
CM000678.1:g.68771319A>G
NC_000016.8:g.67328820A>G
NG_008021.1:g.5125A>G
ENST00000261769.10:c.1A>G
ENST00000261769.9:c.1A>G
ENST00000422392.6:c.1A>G
ENST00000566510.5:c.1A>G
ENST00000566612.5:c.1A>G
ENST00000611625.4:c.1A>G
ENST00000612417.4:c.1A>G
ENST00000621016.4:c.1A>G
NM_004360.3:c.1A>G
NM_001317184.1:c.1A>G
NM_001317185.1:c.-1615A>G
NM_001317186.1:c.-1819A>G
NM_004360.4:c.1A>G
NM_001317184.2:c.1A>G
NM_001317185.2:c.-1615A>G
NM_001317186.2:c.-1819A>G

Pathogenic

Met criteria codes 4

PVS1 PM2_Supporting PS4_Supporting PP1

Not Met criteria codes 22

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

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 Links 0

Evidence submitted by expert panel

CDH1 VCEP

The c.1A>G (p.Met1Val) 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; SCV000760810.2). This variant was also found to co-segregate with disease in multiple affected family members (PP1; SCV000760810.2). 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, PS4_supporting, PM2_supporting, PP1.

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 (SCV000760810.2).
PP1	✔	This variant has been observed to segregate with gastric cancer in a family (SCV000760810.2).

Not Met criteria codes

BS1	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS4	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS3	✖	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
BP5	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP7	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

BP4		✖	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		✖	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		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS3		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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
BA1		✖	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
PP2		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP3		✖	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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