

Variant: *NM_004360.5(CDH1):c.436_437TC[1] (p.Pro147fs)*

Version: 3.0

CA658683944 [↗](#)

491536 (ClinVar) [↗](#)

Gene: [CDH1](#)

Condition: CDH1-related diffuse gastric and lobular breast cancer
([MONDO:0100488](#))

Inheritance Mode: Autosomal dominant inheritance

UUID: b0e566e8-f394-4d9f-b6a9-3f29c1b5705c

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HGVS expressions

NM_004360.5:c.436_437TC[1]

NM_004360.5(CDH1):c.436_437TC[1] (p.Pro147fs)

NC_000016.10:g.68808474_68808475del

CM000678.2:g.68808474_68808475del

NC_000016.9:g.68842377_68842378del

CM000678.1:g.68842377_68842378del

NC_000016.8:g.67399878_67399879del

NG_008021.1:g.76183_76184del

ENST00000261769.10:c.438_439del

ENST00000261769.9:c.438_439del

ENST00000422392.6:c.438_439del

ENST00000561751.1:c.205_206del

ENST00000562836.5:n.509_510del

ENST00000564676.5:n.720_721del

ENST00000564745.1:n.433_434del

ENST00000566510.5:c.438_439del

ENST00000566612.5:c.438_439del

ENST00000611625.4:c.438_439del

ENST00000612417.4:c.438_439del

ENST00000621016.4:c.438_439del

NM_004360.3:c.438_439del

NM_001317184.1:c.438_439del

NM_001317185.1:c.-1178_-1177del

NM_001317186.1:c.-1382_-1381del

NM_004360.4:c.438_439del

NM_004360.5:c.438_439del

NM_001317184.2:c.438_439del

NM_001317185.2:c.-1178_-1177del

NM_001317186.2:c.-1382_-1381del

Pathogenic

Met criteria codes **3**

PM2_Supporting

PVS1

PM5_Supporting

Not Met criteria codes **23**

Expert Panel

CDH1 VCEP [↗](#)

Criteria Specification Information

PS1

PS2

PS3

PS4

BA1

PP1

PP2

PP3

PP4

PM1

PM3

PM4

PM6

BS1

BS4

BS3

BS2

BP5

BP7

BP4

BP3

BP1

BP2

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.438_439delTC p.(Pro147Trpfs*20) 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

PM2_Supporting		✓	Absent in gnomAD v2.1.1, in a region of sufficient coverage.
PVS1		✓	PTC created in exon 4 of 16, expected to undergo NMD.
PM5_Supporting		✓	PTC created in exon 4 of 16, expected to undergo NMD.

Not Met criteria codes

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
PS4		✗	One family that does not meet HDGC criteria: proband with gastric cancer <50 years with no pathology information available (SCV000689538.1).
BA1		✗	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP1		✗	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
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
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
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

Curation History 		
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