

Variant: *NM_004360.5(CDH1):c.1480G>T (p.Glu494Ter)*

Version: 3.0

CA396463169 [↗](#)

485481 (ClinVar) [↗](#)

Gene: CDH1 ([HGNC:999](#))

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

Inheritance Mode: Autosomal dominant inheritance

UUID: c488215c-7af9-4f59-98ec-ef006c6c4f83

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HGVS expressions

NM_004360.5(CDH1):c.1480G>T (p.Glu494Ter)

NM_004360.5:c.1480G>T

NC_000016.10:g.68815674G>T

CM000678.2:g.68815674G>T

NC_000016.9:g.68849577G>T

CM000678.1:g.68849577G>T

NC_000016.8:g.67407078G>T

NG_008021.1:g.83383G>T

ENST00000261769.10:c.1480G>T

ENST00000261769.9:c.1480G>T

ENST00000422392.6:c.1297G>T

ENST00000562836.5:n.1551G>T

ENST00000566510.5:c.*146G>T

ENST00000566612.5:c.1480G>T

ENST00000611625.4:c.1543G>T

ENST00000612417.4:c.1480G>T

ENST00000621016.4:c.1480G>T

NM_004360.3:c.1480G>T

NM_001317184.1:c.1297G>T

NM_001317185.1:c.-69G>T

NM_001317186.1:c.-340G>T

NM_004360.4:c.1480G>T

NM_001317184.2:c.1297G>T

NM_001317185.2:c.-69G>T

NM_001317186.2:c.-340G>T

Pathogenic

Met criteria codes 3

PVS1 PM5_Supporting

PM2_Supporting

Not Met criteria codes 23

BA1 BP5 BP7 BP4 BP3 BP1
BP2 BS1 BS4 BS3 BS2 PP1
PP2 PP3 PP4 PM6 PM1

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**

PM3

PM4

PS1

PS2

PS3

Criteria Specifications for this VCEP

Evidence Links

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Evidence submitted by expert panel			
CDH1 VCEP			
The c.1480G>T p.(Glu494Ter) 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		✓	Predicted NMD (exon 10 of 16).
PM5_Supporting		✓	Predicted NMD (exon 10 of 16).
PM2_Supporting		✓	Absent in population databases.
Not Met criteria codes			
BA1		✗	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

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
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
PM6			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
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



SCV000673183.2 - does not meet HDGC criteria.

Curation History [↗](#)

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