

Variant: *NM_004360.5(CDH1):c.467G>A (p.Trp156Ter)*

Version: 2.0

CA396457680 [↗](#)

463781 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 6cb9aca3-f806-41d5-9c8e-94aa0916aee1

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HGVS expressions

NM_004360.5(CDH1):c.467G>A (p.Trp156Ter)

NM_004360.5:c.467G>A

NC_000016.10:g.68808503G>A

CM000678.2:g.68808503G>A

NC_000016.9:g.68842406G>A

CM000678.1:g.68842406G>A

NC_000016.8:g.67399907G>A

NG_008021.1:g.76212G>A

ENST00000261769.10:c.467G>A

ENST00000261769.9:c.467G>A

ENST00000422392.6:c.467G>A

ENST00000561751.1:c.234G>A

ENST00000562836.5:n.538G>A

ENST00000564676.5:n.749G>A

ENST00000564745.1:n.462G>A

ENST00000566510.5:c.467G>A

ENST00000566612.5:c.467G>A

ENST00000611625.4:c.467G>A

ENST00000612417.4:c.467G>A

ENST00000621016.4:c.467G>A

NM_004360.3:c.467G>A

NM_001317184.1:c.467G>A

NM_001317185.1:c.-1149G>A

NM_001317186.1:c.-1353G>A

NM_004360.4:c.467G>A

NM_001317184.2:c.467G>A

NM_001317185.2:c.-1149G>A

NM_001317186.2:c.-1353G>A

Pathogenic

Met criteria codes **3**

PM5_Supporting

PVS1

PM2_Supporting

Not Met criteria codes **23**

Expert Panel

CDH1 VCEP [↗](#)

Criteria Specification Information

PS1

PP3

BS1

PM4

BP5

PS2

PP4

BS4

BP4

BP7

PS3

BA1

BS3

BP3

PS4

BS2

PM1

BP1

PP1

PM6

PM3

BP2

PP2

[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

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Evidence submitted by expert panel

CDH1 VCEP

The CDH1 c.467G>A (p.Trp156Ter) 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

PM5_Supporting	i	✓	This variant results in a nonsense codon predicted to under NMD.
PVS1	i	✓	This variant results in a nonsense codon predicted to under NMD.
PM2_Supporting	i	✓	This variant is absent from populations in gnomAD, ExAC, 1000 Genomes and ESP.

Not Met criteria codes

PS1		✗	PS1 does not apply to nonsense variants.
PS2	i	✗	This information is not available.
PS3	i	✗	PS3 does not apply to nonsense variants.
PS4	i	✗	SCV000637830.1 and GeneDx - these families do not meet IGCLC criteria for HDGC.
PP1	i	✗	This information is not available.
PP2		✗	PP2 does not apply to CDH1.
PP3	i	✗	PP3 does not apply to nonsense variants.
PP4		✗	PP4 does not apply to CDH1.

BA1			This variant is absent from populations in gnomAD, ExAC, 1000 Genomes and ESP.
BS2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM6			This information is not available.
BS1			This variant is absent from populations in gnomAD, ExAC, 1000 Genomes and ESP.
BS4			This information is not available.
BS3			To our knowledge, no evidence supporting BS3 has been reported for this variant.
PM1			PM1 does not apply to CDH1.
PM3			PM3 does not apply to CDH1.
PM4			PM4 does not apply to nonsense variants.
BP4			BP4 does not apply to nonsense variants.
BP3			BP3 does not apply to CDH1.
BP1			BP1 does not apply to CDH1.
BP2			To our knowledge, this variant has not been reported in cis or trans with a known pathogenic variant.
BP5			To our knowledge, this variant has not been reported in a case with an alternate molecular basis for disease.
BP7			BP7 does not apply to nonsense variants.

Curation History [↗](#)

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