

Variant: *NM_004360.4(CDH1):c.1460_1461del*
(*p.Val487Alafs*)

Version: 3.0

CA891843864 [↗](#)

567085 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 682240bb-090e-4b45-9fa1-c93c2e646a05

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HGVS expressions

NM_004360.4(CDH1):c.1460_1461del (p.Val487Alafs)

NM_004360.4:c.1460_1461del

NC_000016.10:g.68815654_68815655del

CM000678.2:g.68815654_68815655del

NC_000016.9:g.68849557_68849558del

CM000678.1:g.68849557_68849558del

NC_000016.8:g.67407058_67407059del

NG_008021.1:g.83363_83364del

ENST00000261769.10:c.1460_1461del

ENST00000261769.9:c.1460_1461del

ENST00000422392.6:c.1277_1278del

ENST00000562836.5:n.1531_1532del

ENST00000566510.5:c.*126_*127del

ENST00000566612.5:c.1460_1461del

ENST00000611625.4:c.1523_1524del

ENST00000612417.4:c.1460_1461del

ENST00000621016.4:c.1460_1461del

NM_004360.3:c.1460_1461del

NM_001317184.1:c.1277_1278del

NM_001317185.1:c.-89_-88del

NM_001317186.1:c.-360_-359del

NM_004360.5:c.1460_1461del

NM_001317184.2:c.1277_1278del

NM_001317185.2:c.-89_-88del

NM_001317186.2:c.-360_-359del

Pathogenic

Met criteria codes 3

PVS1

PM5_Supporting

PM2_Supporting

Not Met criteria codes 23

BA1

BS1

BS4

BS3

BS2

BP5

BP7

BP4

BP3

BP1

BP2

PS1

Expert Panel

CDH1 VCEP [↗](#)

Criteria Specification Information

[↗](#) **Criteria Specification:** ClinGen CDH1 Expert Panel
Specifications to the ACMG/AMP Variant Interpretation
Guidelines Version 3.1

Evidence submitted by expert panel

CDH1 VCEP

The c.1460_1461del (p.Val487Alafs*3) 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	i	✓	Deletion of 2 bp in exon 10 of 16, predicted to cause a frameshift leading to NMD.
PM5_Supporting	i	✓	Deletion of 2 bp in exon 10 of 16, predicted to cause a frameshift leading to NMD.
PM2_Supporting	i	✓	Absent in gnomAD cohort.

Not Met criteria codes

BA1	i	✗	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS1	i	✗	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
BS2	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			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
PS4			Proband doesn't meet IGCLC HDGC criteria. (SCV000814618.1).
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

Curation History [↗](#)

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