

Variant: *NM_004360.5(CDH1):c.1312del (p.Thr438fs)*

Version: 3.0

CA658824987 [↗](#)

545807 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: b5811c1f-e4fd-48a9-b336-2896da3fd5b9

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HGVS expressions

NM_004360.5:c.1312del

NM_004360.5(CDH1):c.1312del (p.Thr438fs)

NC_000016.10:g.68813487del

CM000678.2:g.68813487del

NC_000016.9:g.68847390del

CM000678.1:g.68847390del

NC_000016.8:g.67404891del

NG_008021.1:g.81196del

ENST00000261769.10:c.1312del

ENST00000261769.9:c.1312del

ENST00000422392.6:c.1137+1224del

ENST00000562836.5:n.1383del

ENST00000566510.5:c.1156del

ENST00000566612.5:c.1312del

ENST00000611625.4:c.1312del

ENST00000612417.4:c.1312del

ENST00000621016.4:c.1312del

NM_004360.3:c.1312del

NM_001317184.1:c.1137+1224del

NM_001317185.1:c.-304del

NM_001317186.1:c.-508del

NM_004360.4:c.1312del

NM_001317184.2:c.1137+1224del

NM_001317185.2:c.-304del

NM_001317186.2:c.-508del

Pathogenic

Met criteria codes 3

PVS1 PM5_Supporting

PM2_Supporting

Not Met criteria codes 23

PM6 PM1 PM3 PM4 BS1

BS4 BS3 BS2 PS1 PS2 PS3

PS4 BP5 BP7 BP4 BP3 BP1

BP2 BA1 PP1 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**

Evidence submitted by expert panel

CDH1 VCEP

The c.1312delA p.(Thr438fs) 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			Exon 9 of 16, predicted NMD.
PM5_Supporting			Exon 9 of 16, predicted NMD.
PM2_Supporting			Absent in population databases.

Not Met criteria codes

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
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			
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		✖	SCV000779091.2 - proband does not meet HDGC phenotype criteria.
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
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

Curation History [↗](#)

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