

Variant: *NM_004360.5(CDH1):c.1235_1236TA[3] (p.Ile415fs)*

Version: 2.0

CA645369678 [↗](#)

428622 (ClinVar) [↗](#)

Gene: [CDH1](#)

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 169b8ea6-d8f6-4bc7-8434-67e66ee59492

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HGVS expressions

NM_004360.5:c.1235_1236TA[3]
NM_004360.5(CDH1):c.1235_1236TA[3] (p.Ile415fs)
NC_000016.10:g.68813412_68813413dup
CM000678.2:g.68813412_68813413dup
NC_000016.9:g.68847315_68847316dup
CM000678.1:g.68847315_68847316dup
NC_000016.8:g.67404816_67404817dup
NG_008021.1:g.81121_81122dup
ENST00000261769.10:c.1237_1238dup
ENST00000261769.9:c.1237_1238dup
ENST00000422392.6:c.1137+1149_1137+1150dup
ENST00000562836.5:n.1308_1309dup
ENST00000566510.5:c.1081_1082dup
ENST00000566612.5:c.1237_1238dup
ENST00000611625.4:c.1237_1238dup
ENST00000612417.4:c.1237_1238dup
ENST00000621016.4:c.1237_1238dup
NM_004360.3:c.1237_1238dup
NM_001317184.1:c.1137+1149_1137+1150dup
NM_001317185.1:c.-379_-378dup
NM_001317186.1:c.-583_-582dup
NM_004360.4:c.1237_1238dup
NM_004360.5:c.1237_1238dup
NM_001317184.2:c.1137+1149_1137+1150dup
NM_001317185.2:c.-379_-378dup
NM_001317186.2:c.-583_-582dup

Pathogenic

Met criteria codes 4

PM2_Supporting PVS1
PM5_Supporting PS4_Supporting

Not Met criteria codes 22

PS1 PS2 PS3 BA1 PP1 PP2
PP3 PP4 PM1 PM3 PM4

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.1235_1236TA[3] (p.Ile415fs) variant is predicted to result in a premature stop codon that leads to a truncated or absent protein (PVS1, PM5_Supporting). The variant is absent in the gnomAD cohort (PM2_Supporting; <http://https://gnomad.broadinstitute.org/>). The variant has been reported in a family meeting HDGC clinical criteria (SCV000580699.3; PS4_Supporting). In summary, 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 (Variant Interpretation Guidelines Version 3.1): PVS1, PM2_Supporting, PS4_Supporting, PM5_Supporting.

Met criteria codes

PM2_Supporting		✓	Variant absent from gnomAD
PVS1		✓	Variant predicted to cause NMD with premature stop at amino acid 417
PM5_Supporting		✓	Apply PM5_Supporting to nonsense/frameshift variants that are predicted/proved to undergo NMD.
PS4_Supporting		✓	The variant has been reported in a family meeting HDGC clinical criteria (SCV000580699.3; PS4_Supporting).

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
BA1		✗	Variant absent from gnomAD
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			Variant absent from gnomAD
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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