

Variant: *NM_004360.4(CDH1):c.1612delG (p.Asp538Thrfs)*

Version: 2.0

CA645509536 [↗](#)

437928 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 47f620ff-80ee-453c-9f55-58567c1f14be

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HGVS expressions

NM_004360.4(CDH1):c.1612delG (p.Asp538Thrfs)

NM_004360.4:c.1612delG

NC_000016.10:g.68819326del

CM000678.2:g.68819326del

NC_000016.9:g.68853229del

CM000678.1:g.68853229del

NC_000016.8:g.67410730del

NG_008021.1:g.87035del

ENST00000261769.10:c.1612del

ENST00000261769.9:c.1612del

ENST00000422392.6:c.1429del

ENST00000562836.5:n.1683del

ENST00000566510.5:c.*278del

ENST00000566612.5:c.1566-2675del

ENST00000611625.4:c.1675del

ENST00000612417.4:c.1612del

ENST00000621016.4:c.1612del

NM_004360.3:c.1612del

NM_001317184.1:c.1429del

NM_001317185.1:c.64del

NM_001317186.1:c.-254-2675del

NM_004360.4:c.1612del

NM_004360.5:c.1612del

NM_001317184.2:c.1429del

NM_001317185.2:c.64del

NM_001317186.2:c.-254-2675del

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 Links

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Evidence submitted by expert panel

CDH1 VCEP

The c.1612delG p.(Asp538Thrfs) 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://gnomad.broadinstitute.org>). This variant has been reported in at least one family meeting HDGC clinical criteria (PS4_Supporting; PMID: 29131691). 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			Absent in population databases.
PVS1			Exon 11 of 16. Predicted NMD.
PM5_Supporting			Apply PM5_Supporting to nonsense/frameshift variants that are predicted/proved to undergo NMD.
PS4_Supporting			PMID: 29131691 - 41-year-old with metastatic GC characterized by the presence of signet ring cells. Same patient as in PMID: 29131691 PubMed:29295527 41-year-old with metastatic GC characterized by the presence of signet ring cells PubMed:29131691

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			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
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		✖	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
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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