

Variant: *NM_004360.4(CDH1):c.2293C>T (p.Gln765Ter)*

Version: 2.0

CA10580156 [↗](#)

230451 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: d4d80a05-4db4-4643-877f-10dbb705c844

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HGVS expressions

NM_004360.4(CDH1):c.2293C>T (p.Gln765Ter)

NM_004360.4:c.2293C>T

NC_000016.10:g.68828302C>T

CM000678.2:g.68828302C>T

NC_000016.9:g.68862205C>T

CM000678.1:g.68862205C>T

NC_000016.8:g.67419706C>T

NG_008021.1:g.96011C>T

ENST00000261769.10:c.2293C>T

ENST00000261769.9:c.2293C>T

ENST00000422392.6:c.2110C>T

ENST00000562118.1:n.511C>T

ENST00000562836.5:n.2364C>T

ENST00000566510.5:c.*959C>T

ENST00000566612.5:c.*533C>T

ENST00000611625.4:c.2356C>T

ENST00000612417.4:c.1853+1748C>T

ENST00000621016.4:c.1866-5901C>T

NM_004360.3:c.2293C>T

NM_001317184.1:c.2110C>T

NM_001317185.1:c.745C>T

NM_001317186.1:c.328C>T

NM_004360.5:c.2293C>T

NM_001317184.2:c.2110C>T

NM_001317185.2:c.745C>T

NM_001317186.2:c.328C>T

Pathogenic

Met criteria codes 4

PM2_Supporting

PS4_Supporting

PM5_Supporting

PVS1

Not Met criteria codes 22

BA1

BP4

BP3

BP1

BP2

BP5

BP7

BS2

BS1

BS4

BS3

PP1

Expert Panel

CDH1 VCEP [↗](#)

Criteria Specification Information

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

PP2PP3PP4PS1PS2PS3

PM6PM1PM3PM4

[Criteria Specification Approval History](#)

[Criteria Specifications for this VCEP](#)

Evidence Links

1

Evidence submitted by expert panel

CDH1 VCEP

The c.2293C>T (p.Gln765*) variant is predicted to result in a premature stop codon that leads to a truncated or absent protein (PVS1, PM5_Supporting). This variant is absent in the gnomAD cohort (PM2_Supporting; <http://gnomad.broadinstitute.org>). The variant has been reported in at least one family meeting HDGC clinical criteria (PS4_Supporting; SCV000839094.1). 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 gnomAD
PS4_Supporting			One family that meets HDGC criteria (SCV000839094.1). Proband with unknown whether family fulfills HDGC criteria (SCV000274004.4).
Somatic variant in a breast tumour from a white female PubMed:29156750 			
PM5_Supporting			Apply PM5_Supporting to nonsense/frameshift variants that are predicted/proved to undergo NMD.
PVS1			Stop gain introduced at position 765 in exon 14 of 16. Predicted to undergo NMD.

Not Met criteria codes

BA1			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
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
BS2			

			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
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
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
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

Curation History [↗](#)



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