

Variant: *NM\_004360.5(CDH1):c.48+5C>G*

Version: 2.0

[CA163551](#)

[140784 \(ClinVar\)](#)

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

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

**Inheritance Mode:** Autosomal dominant inheritance

**UUID:** 1f5a92b8-7747-42bb-83cd-f8c57088c41b

**Approved on:** 2023-08-10

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### *HGVS expressions*

**NM\_004360.5:c.48+5C>G**

NM\_004360.5(CDH1):c.48+5C>G

NC\_000016.10:g.68737468C>G

CM000678.2:g.68737468C>G

NC\_000016.9:g.68771371C>G

CM000678.1:g.68771371C>G

NC\_000016.8:g.67328872C>G

NG\_008021.1:g.5177C>G

ENST00000261769.10:c.48+5C>G

ENST00000261769.9:c.48+5C>G

ENST00000422392.6:c.48+5C>G

ENST00000566510.5:c.48+5C>G

ENST00000566612.5:c.48+5C>G

ENST00000611625.4:c.48+5C>G

ENST00000612417.4:c.48+5C>G

ENST00000621016.4:c.48+5C>G

NM\_004360.3:c.48+5C>G

NM\_001317184.1:c.48+5C>G

NM\_001317185.1:c.-1568+5C>G

NM\_001317186.1:c.-1772+5C>G

NM\_004360.4:c.48+5C>G

NM\_001317184.2:c.48+5C>G

NM\_001317185.2:c.-1568+5C>G

NM\_001317186.2:c.-1772+5C>G

**Benign**

Met criteria codes **2**

**BA1** **BP2**

Not Met criteria codes **24**

**PM6** **PM2** **PM1** **PM3** **PM5**  
**PM4** **BS2** **BS1** **BS4** **BS3**  
**BP5** **BP7** **BP4** **BP3** **BP1**  
**PVS1** **PS1** **PS2** **PS3** **PS4**  
**PP1** **PP2** **PP3** **PP4**

Expert Panel

Criteria Specification Information

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

[Criteria Specification Approval History](#)





[Criteria Specifications for this VCEP](#)

Evidence submitted by expert panel



















### CDH1 VCEP






















The NM\_004360.5(CDH1):c.48+5C>G variant has an allele frequency of 0.11584 (11.584%, 1670/14416 alleles, 98 homozygotes) in the African subpopulation of the gnomAD v2.1.1 cohort (BA1; BP2). Therefore, this variant meets criteria to be classified as benign. ACMG/AMP criteria applied, as specified by the CDH1 Variant Curation Expert Panel (Variant Interpretation Guidelines Version 3.1): BA1, BP2.

#### Met criteria codes

<b>BA1</b>			MAF > 0.2%: African AF is 0.11584 (1670/14416 alleles, 98 homozygotes in gnomAD v2.1.1)
<b>BP2</b>			Ninety-eight Africa homozygotes in the controls cohort in gnomAD v2.1.1

#### Not Met criteria codes

<b>PM6</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>PM2</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>PM1</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>PM3</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>PM5</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>PM4</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>BS2</b>			Not assessed, because BA1 met
<b>BS1</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>BS4</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>BS3</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>BP5</b>			Not assessed, because BA1 met

<b>BP7</b>			Not assessed, because BA1 met
<b>BP4</b>			Not assessed, because BA1 met
<b>BP3</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>BP1</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>PVS1</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>PS1</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>PS2</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>PS3</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>PS4</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>PP1</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>PP2</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>PP3</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>PP4</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History [↗](#)

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