

Variant: *NM_004360.5(CDH1):c.1937-13T>C*

Version: 2.0

[CA163616](#)

[140806 \(ClinVar\)](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 84f291c3-4e16-4968-9103-5a1c820e0bf5

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HGVS expressions

NM_004360.5:c.1937-13T>C

NM_004360.5(CDH1):c.1937-13T>C

NC_000016.10:g.68823386T>C

CM000678.2:g.68823386T>C

NC_000016.9:g.68857289T>C

CM000678.1:g.68857289T>C

NC_000016.8:g.67414790T>C

NG_008021.1:g.91095T>C

ENST00000261769.10:c.1937-13T>C

ENST00000261769.9:c.1937-13T>C

ENST00000422392.6:c.1754-13T>C

ENST00000562118.1:n.142T>C

ENST00000562836.5:n.2008-13T>C

ENST00000566510.5:c.*603-13T>C

ENST00000566612.5:c.*177-13T>C

ENST00000611625.4:c.2000-13T>C

ENST00000612417.4:c.1830+1267T>C

ENST00000621016.4:c.1865+1232T>C

NM_004360.3:c.1937-13T>C

NM_001317184.1:c.1754-13T>C

NM_001317185.1:c.389-13T>C

NM_001317186.1:c.-29-13T>C

NM_004360.4:c.1937-13T>C

NM_001317184.2:c.1754-13T>C

NM_001317185.2:c.389-13T>C

NM_001317186.2:c.-29-13T>C

Benign

Met criteria codes **2**

BP2 **BA1**

Not Met criteria codes **24**

BS1 **BS4** **BS3** **BS2** **BP5** **BP7**
BP4 **BP3** **BP1** **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](#)





Evidence Links 0

Evidence submitted by expert panel

















CDH1 VCEP

The NM_004360.5(CDH1):c.1937-13T>C variant has an allele frequency of 0.16607 (16.6%, 4107/24730 alleles, 330 homozygotes) in the European (Finnish) 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

BP2			330 European (Finnish) homozygotes in the controls cohort in gnomAD v2.1.1
BA1			MAF > 0.2%: European (Finnish) AF is 0.16607 (4107/24730 alleles, 330 homozygotes in gnomAD v2.1.1)

Not Met criteria codes

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			Not assessed, because BA1 met
BP5			Not assessed, because BA1 met
BP7			Not assessed, because BA1 met
BP4			Not assessed, because BA1 met
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

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		✘	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
PM5		✘	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
PM2		✘	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

PVS1



No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History [↗](#)

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