

Variant: *NM_004360.5(CDH1):c.866C>T (p.Ala289Val)*

Version: 2.0

CA336712 [↗](#)

216597 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 38e7c69b-3661-43f7-afea-1d95f740ec34

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HGVS expressions

NM_004360.5:c.866C>T

NM_004360.5(CDH1):c.866C>T (p.Ala289Val)

NC_000016.10:g.68811717C>T

CM000678.2:g.68811717C>T

NC_000016.9:g.68845620C>T

CM000678.1:g.68845620C>T

NC_000016.8:g.67403121C>T

NG_008021.1:g.79426C>T

ENST00000261769.10:c.866C>T

ENST00000261769.9:c.866C>T

ENST00000422392.6:c.866C>T

ENST00000561751.1:c.488C>T

ENST00000562836.5:n.937C>T

ENST00000566510.5:c.710C>T

ENST00000566612.5:c.866C>T

ENST00000611625.4:c.866C>T

ENST00000612417.4:c.866C>T

ENST00000621016.4:c.866C>T

NM_004360.3:c.866C>T

NM_001317184.1:c.866C>T

NM_001317185.1:c.-750C>T

NM_001317186.1:c.-954C>T

NM_004360.4:c.866C>T

NM_001317184.2:c.866C>T

NM_001317185.2:c.-750C>T

NM_001317186.2:c.-954C>T

Likely Benign

Met criteria codes **1**

BS2

Not Met criteria codes **25**

BS1

BS4

BS3

PVS1

BP5

BP7

BP4

BP3

BP1

BP2

BA1

PS1

PS2

PS3

PS4

PP1

PP2

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.866C>T (p.Ala289Val) missense variant has a frequency of 0.00002 (6 of 251,472) in gnomAD, with a maximum allele frequency of 0.00010 (3 of 30,616) in the South Asian subpopulation (<http://gnomad.broadinstitute.org>). This variant has been observed in ≥10 individuals without DGC, SRC tumours or LBC and whose families do not suggest HDGC (BS2; SCV000669016.2, SCV000254833.6). In summary, the clinical significance of this variant is classified as of likely benign based the ACMG/AMP criteria applied, as specified by the CDH1 Variant Curation Expert Panel (Variant Interpretation Guidelines Version 3.1): BS2.

Met criteria codes

BS2			This variant has been observed in ≥10 individuals without DGC, SRC tumours or LBC and whose families do not suggest HDGC (SCV000669016.2, SCV000254833.6)
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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
PVS1			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
BA1		✖	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		✖	3 probands with breast cancer (all reported IDC). One with family history of breast and endometrial cancer. Two with no family history of cancer.
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
PM6		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM2		✖	frequency of 0.00002 (6 of 251,472) in gnomAD, with a maximum allele frequency of 0.00010 (3 of 30,616) in the South Asian subpopulation and 0.00003 (3 of 113,754) in the European (Non-Finnish) subpopulation
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

Curation History 		
		
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