

Variant: NM_004360.4(CDH1):c.79C>T (p.Pro27Ser)

CA10583401 [↗](#)

239913 (ClinVar) [↗](#)

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

Condition: CDH1-related diffuse gastric and lobular breast cancer ([MONDO:0007648](#))

Inheritance Mode: Autosomal dominant inheritance

UUID: 9a9082a0-343e-4b16-a256-c64b923d37b2

Approved on: 2018-11-21

Published on: 2018-12-07

HGVS expressions

NM_004360.4:c.79C>T

NM_004360.4(CDH1):c.79C>T (p.Pro27Ser)

NC_000016.10:g.68738327C>T

CM000678.2:g.68738327C>T

NC_000016.9:g.68772230C>T

CM000678.1:g.68772230C>T

NC_000016.8:g.67329731C>T

NG_008021.1:g.6036C>T

ENST00000261769.10:c.79C>T

ENST00000261769.9:c.79C>T

ENST00000422392.6:c.79C>T

ENST00000566510.5:c.79C>T

ENST00000566612.5:c.79C>T

ENST00000611625.4:c.79C>T

ENST00000612417.4:c.79C>T

ENST00000621016.4:c.79C>T

NM_004360.3:c.79C>T

NM_001317184.1:c.79C>T

NM_001317185.1:c.-1537C>T

NM_001317186.1:c.-1741C>T

NM_004360.5:c.79C>T

NM_001317184.2:c.79C>T

NM_001317185.2:c.-1537C>T

NM_001317186.2:c.-1741C>T

NM_004360.5(CDH1):c.79C>T (p.Pro27Ser)

Uncertain Significance

Met criteria codes **1**

PM2_Supporting

Not Met criteria codes **25**

BA1	BP2	BP3	BP1	BP4	BP5
BP7	BS2	BS4	BS3	BS1	PP1
PP4	PP2	PP3	PS2	PS4	PS3
PS1	PM6	PM3	PM1	PM4	
PM5	PVS1				

Expert Panel

CDH1 VCEP [↗](#)

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 c.79C>T (p.Pro27Ser) variant has allele frequency <1 out of 100, 000 in the gnomAD cohort (1/156646 in gnomAD v2.1.1; PM2_Supporting). No additional evidence met criteria for consideration. In summary, the clinical significance of this variant is uncertain. ACMG/AMP criteria applied, as specified by the CDH1 Variant Curation Expert Panel (Variant Interpretation Guidelines Version 3.1): PM2_Supporting.

Met criteria codes

PM2_Supporting   Allele frequency <1 out of 100, 000 (1/156646 in gnomAD v2.1.1).

Not Met criteria codes

BA1			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
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
BP4			REVEL, SIFT, PolyPhen2-HVAR, MutationTaster, MutationAssessor, FATHMM, PROVEAN, MetaSVM, and CADD predict benign. P27S observed in Marmoset, Rat, and Mouse. No new splice site predicted by NNSPLICE, however HumanSplicingFinder predicts: "Alteration of an exonic ESE site. Potential alteration of splicing."
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
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
BS1			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
PP4			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			REVEL, SIFT, PolyPhen2-HVAR, MutationTaster, MutationAssessor, FATHMM, PROVEAN, MetaSVM, and CADD predict benign. P27S observed in Marmoset, Rat, and Mouse. No new splice site predicted by NNSPLICE, however HumanSplicingFinder predicts: "Alteration of an exonic ESE site. Potential alteration of splicing."
PS2			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
PS3			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
PM6			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
PM1			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
PM5			

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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