

Variant: *NM_004360.5(CDH1):c.7C>T (p.Pro3Ser)*

Version: 2.0

CA16620227 [↗](#)

418111 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 916169ad-d424-4f36-8530-42c87c14d450

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HGVS expressions

NM_004360.5:c.7C>T

NM_004360.5(CDH1):c.7C>T (p.Pro3Ser)

NC_000016.10:g.68737422C>T

CM000678.2:g.68737422C>T

NC_000016.9:g.68771325C>T

CM000678.1:g.68771325C>T

NC_000016.8:g.67328826C>T

NG_008021.1:g.5131C>T

ENST00000261769.10:c.7C>T

ENST00000261769.9:c.7C>T

ENST00000422392.6:c.7C>T

ENST00000566510.5:c.7C>T

ENST00000566612.5:c.7C>T

ENST00000611625.4:c.7C>T

ENST00000612417.4:c.7C>T

ENST00000621016.4:c.7C>T

NM_004360.3:c.7C>T

NM_001317184.1:c.7C>T

NM_001317185.1:c.-1609C>T

NM_001317186.1:c.-1813C>T

NM_004360.4:c.7C>T

NM_001317184.2:c.7C>T

NM_001317185.2:c.-1609C>T

NM_001317186.2:c.-1813C>T

Uncertain Significance

Met criteria codes **1**

PM2_Supporting

Not Met criteria codes **25**

BA1	BP4	BP3	BP1	BP2	BP5
BP7	BS2	BS1	BS4	BS3	PP1
PP2	PP3	PP4	PS1	PS2	PS3
PS4	PM6	PM1	PM3	PM5	
PM4	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.7C>T (p.Pro3Ser) is absent from gnomAD (PM2_Supporting; <http://gnomad.broadinstitute.org>). In summary, the clinical significance of this variant is classified as a variant of uncertain significance based on 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   This variant is absent from populations in gnomAD, ExAC, 1000 Genomes and ESP.

Not Met criteria codes

BA1   This variant is absent from populations in gnomAD, ExAC, 1000 Genomes and ESP.

BP4   This variant is not predicted to result in abnormal splicing by in silico splice site predictors.

BP3  BP3 does not apply to CDH1.

BP1  BP1 does not apply to CDH1.

BP2   To our knowledge, this variant has not been reported in cis or trans with a known pathogenic variant.

BP5   To our knowledge, this variant has not been reported in a case with an alternate molecular basis for disease.

BP7   BP7 does not apply to this variant.

BS2   This variant was observed in 2 individuals without DGC, SRC tumours or LBC and whose family history does not suggest HDGC (SCV000564833.3, SCV000957806.1).

BS1   This variant is absent from populations in gnomAD, ExAC, 1000 Genomes and ESP.

BS4   No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

BS3   BS3 does not apply to this variant.

PP1   No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

PP2		✖	PP2 does not apply to CDH1.
PP3		✖	This variant is not predicted to result in abnormal splicing by in silico splice site predictors.
PP4		✖	PP4 does not apply to CDH1.
PS1		✖	No pathogenic variants resulting in the same amino acid change have been described.
PS2		✖	To our knowledge, this variant has not been observed de novo.
PS3		✖	PS3 does not apply to this variant.
PS4		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM6		✖	To our knowledge, this variant has not been observed de novo.
PM1		✖	BP1 does not apply to CDH1.
PM3		✖	PM3 does not apply to CDH1.
PM5		✖	PM5 does not apply to CDH1.
PM4		✖	PM4 does not apply to this variant.
PVS1		✖	PVS1 does not apply to this variant.

Curation History 		
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Showing 1 to 2 of 2 rows		

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