

Variant: *NM_004360.3:c.614T>C*

CA396458013 [↗](#)

599654 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 36659513-f448-4457-900a-485812518ebb

Approved on: 2018-11-22

Published on: 2018-12-07

HGVS expressions

NM_004360.3:c.614T>C

NC_000016.10:g.68808775T>C

CM000678.2:g.68808775T>C

NC_000016.9:g.68842678T>C

CM000678.1:g.68842678T>C

NC_000016.8:g.67400179T>C

NG_008021.1:g.76484T>C

ENST00000261769.10:c.614T>C

ENST00000261769.9:c.614T>C

ENST00000422392.6:c.614T>C

ENST00000561751.1:n.381T>C

ENST00000562836.5:n.685T>C

ENST00000564676.5:n.896T>C

ENST00000566510.5:c.531+208T>C

ENST00000566612.5:c.614T>C

ENST00000567320.1:n.124T>C

ENST00000611625.4:c.614T>C

ENST00000612417.4:c.614T>C

ENST00000621016.4:c.614T>C

NM_001317184.1:c.614T>C

NM_001317185.1:c.-1002T>C

NM_001317186.1:c.-1206T>C

NM_004360.4:c.614T>C

NM_004360.5:c.614T>C

NM_001317184.2:c.614T>C

NM_001317185.2:c.-1002T>C

NM_001317186.2:c.-1206T>C

NM_004360.5(CDH1):c.614T>C (p.Phe205Ser)

Uncertain Significance

Met criteria codes **2**

PM2_Supporting

PS4_Supporting

Not Met criteria codes **24**

BA1

BP2

BP3

BP1

BP4

BP5

BP7

BS2

BS4

BS3

BS1

PP1

PP4

PP2

PP3

PS2

PS1

PS3

Expert Panel

CDH1 VCEP [↗](#)

Criteria Specification Information

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

Evidence submitted by expert panel

CDH1 VCEP

The c.614T>C (p. Phe205Ser) variant is absent in the gnomAD cohort (PM2_Supporting). This variant has been reported in at least one family meeting HDGC clinical criteria (PS4_Supporting; internal laboratory contributor). 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, PS4_Supporting.

Met criteria codes

PM2_Supporting			Absent from gnomAD databases.
PS4_Supporting			1 proband with DGC with age of dx at 35.

Not Met criteria codes

BA1			Absent from control databases.
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			Multiple computational tools predict a deleterious effect on protein function.
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			Data suggests that cells with this variant have reduced cell compaction, increased cell invasion and reduced E-cadherin at the cell membrane.
BS1			Absent from control databases.
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			Multiple computational tools predict a deleterious effect on protein function.
PS2			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
PS3			Data suggests that cells with this variant have reduced cell compaction, increased cell invasion and reduced E-cadherin at the cell membrane.
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
PM6			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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