

Variant: *NM_004360.5(CDH1):c.164T>G (p.Val55Gly)*

Version: 2.0

[CA157980](#)

[132769 \(ClinVar\)](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 87528509-8592-44e0-bf92-b276bdd488fa

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HGVS expressions

NM_004360.5:c.164T>G

NM_004360.5(CDH1):c.164T>G (p.Val55Gly)

NC_000016.10:g.68801670T>G

CM000678.2:g.68801670T>G

NC_000016.9:g.68835573T>G

CM000678.1:g.68835573T>G

NC_000016.8:g.67393074T>G

NG_008021.1:g.69379T>G

ENST00000261769.10:c.164T>G

ENST00000261769.9:c.164T>G

ENST00000422392.6:c.164T>G

ENST00000562836.5:n.235T>G

ENST00000564676.5:n.446T>G

ENST00000564745.1:n.159T>G

ENST00000566510.5:c.164T>G

ENST00000566612.5:c.164T>G

ENST00000611625.4:c.164T>G

ENST00000612417.4:c.164T>G

ENST00000621016.4:c.164T>G

NM_004360.3:c.164T>G

NM_001317184.1:c.164T>G

NM_001317185.1:c.-1452T>G

NM_001317186.1:c.-1656T>G

NM_004360.4:c.164T>G

NM_001317184.2:c.164T>G

NM_001317185.2:c.-1452T>G

NM_001317186.2:c.-1656T>G

Likely Benign

Met criteria codes **1**

BS2

Not Met criteria codes **25**

BS1 BS4 BS3 BP4 BP3 BP1

BP2 BP5 BP7 PVS1 PS1

PS2 PS3 PS4 BA1 PP1 PP2

Expert Panel

Criteria Specification Information

Criteria Specification: *ClinGen CDH1 Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines Version 3.1*

Evidence Links 0

Evidence submitted by expert panel















CDH1 VCEP



























The c.164T>G (p.Val55Gly) variant has been observed in >10 (113) individuals without a diagnosis of diffuse gastric cancer, signet ring tumor or lobular breast cancer and whose family histories do not suggest HDGC (BS2; SCV000321508.8, SCV000153979.11). In summary, the clinical significance of this variant is classified as likely benign based on BS2 alone, as specified by the CDH1 Variant Curation Expert Panel (Variant Interpretation Guidelines Version 3.1): BS2.

Met criteria codes

BS2	 	>10 (113) individuals w/o DCG, SRC tumors, or LBC & whose families do not suggest HDGC (SCV000321508.8, SCV000153979.11)
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Not Met criteria codes

BS1	 	MAF <0.1%: European (non-Finnish) AF is 0.005% (7/129,106 alleles, 0 homozygotes in gnomAD v2.1)
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
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
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
PVS1			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			7 probands with a personal history of LBC or family history of gastric cancer that do not meet HDGC clinical criteria (SCV000321508.8, SCV000153979.11)
BA1			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			Located in 1st bp of exon 3 in acceptor splice region. No predicted splice impact by MaxEntScan or Splice AI
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



>1:100,000 alleles in gnomAD v2.1: European (non-Finnish) AF is 0.005% (7/129,106 alleles, 0 homozygotes)

Curation History [↗](#)

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