

Variant: NM_004360.4(CDH1):c.286A>G (p.Ile96Val)

CA349520 [↗](#)

220611 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: d9039763-3145-4298-b468-da3967101df3

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HGVS expressions

NM_004360.4:c.286A>G

NM_004360.4(CDH1):c.286A>G (p.Ile96Val)

NC_000016.10:g.68801792A>G

CM000678.2:g.68801792A>G

NC_000016.9:g.68835695A>G

CM000678.1:g.68835695A>G

NC_000016.8:g.67393196A>G

NG_008021.1:g.69501A>G

ENST00000261769.10:c.286A>G

ENST00000261769.9:c.286A>G

ENST00000422392.6:c.286A>G

ENST00000561751.1:n.53A>G

ENST00000562836.5:n.357A>G

ENST00000564676.5:n.568A>G

ENST00000564745.1:n.281A>G

ENST00000566510.5:c.286A>G

ENST00000566612.5:c.286A>G

ENST00000611625.4:c.286A>G

ENST00000612417.4:c.286A>G

ENST00000621016.4:c.286A>G

NM_004360.3:c.286A>G

NM_001317184.1:c.286A>G

NM_001317185.1:c.-1330A>G

NM_001317186.1:c.-1534A>G

NM_004360.5:c.286A>G

NM_001317184.2:c.286A>G

NM_001317185.2:c.-1330A>G

NM_001317186.2:c.-1534A>G

NM_004360.5(CDH1):c.286A>G (p.Ile96Val)

Benign

Met criteria codes 2

BS1 BS2

Not Met criteria codes 24

PS2 PS4 PS3 PS1 BA1 PP4
PP1 PP2 PP3 PM4 PM1

Expert Panel

CDH1 VCEP [↗](#)

Criteria Specification Information

- PM3PM5PM6PM2BS4BS3BP7BP5BP3BP2BP1BP4PVS1

Evidence Links 1

- [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.286A>G (p.Ile96Val) variant has an allele frequency of 0.00122 (0.12%, 41/33,578 alleles) in the Latino subpopulation of the gnomAD cohort (BS1). This variant has also been observed in >10 without a diagnosis of diffuse gastric cancer, signet ring tumor or lobular breast cancer and whose family histories do not suggest HDGC (BS2; internal laboratory contributors). In summary, this variant meets criteria to be classified as benign. ACMG/AMP criteria applied, as specified by the CDH1 Variant Curation Expert Panel (Variant Interpretation Guidelines Version 3.1): BS1, BS2.

Met criteria codes

BS1			Frequency of 0.16% in ExAC, 0.12% in gnomAD
BS2			Observed in 42 individuals w/o a dx of HDGC.

Not Met criteria codes

PS2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS4			Reported in one Hispanic individual with HDGC
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
BA1			Frequency of 0.16% in ExAC, 0.12% in Gnomad
PP4			Reported in one Hispanic individual with HDGC
This variant was reported in one Hispanic proband with Diffuse gastric cancer at age 29 but no family history. Meets IGCLC criteria 3. PubMed:26182300 			
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
PM4			

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		✖	Other variant at this position in Clinvar is VUS
PM6		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM2		✖	Frequency of 0.16% in ExAC, 0.12% in gnomAD
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
BP7		✖	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
BP3		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP2		✖	1 homozygote in gnomAD
BP1		✖	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
PVS1		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline



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