

Variant: *NM_004360.5(CDH1):c.121G>T (p.Val41Leu)*

Version: 1.0

CA396452116 [↗](#)

481692 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: eaf93bd1-8295-4f6c-8bfb-f6a0520b948b

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HGVS expressions

NM_004360.5(CDH1):c.121G>T (p.Val41Leu)

NM_004360.5:c.121G>T

NC_000016.10:g.68738369G>T

CM000678.2:g.68738369G>T

NC_000016.9:g.68772272G>T

CM000678.1:g.68772272G>T

NC_000016.8:g.67329773G>T

NG_008021.1:g.6078G>T

ENST00000261769.10:c.121G>T

ENST00000261769.9:c.121G>T

ENST00000422392.6:c.121G>T

ENST00000566510.5:c.121G>T

ENST00000566612.5:c.121G>T

ENST00000611625.4:c.121G>T

ENST00000612417.4:c.121G>T

ENST00000621016.4:c.121G>T

NM_004360.3:c.121G>T

NM_001317184.1:c.121G>T

NM_001317185.1:c.-1495G>T

NM_001317186.1:c.-1699G>T

NM_004360.4:c.121G>T

NM_001317184.2:c.121G>T

NM_001317185.2:c.-1495G>T

NM_001317186.2:c.-1699G>T

Uncertain Significance

Met criteria codes **2**

PM2_Supporting

BS2_Supporting

Not Met criteria codes **24**

PS1

PS2

PS3

PS4

BA1

PP1

PP2

PP3

PP4

PM1

PM3

PM5

PM4

PM6

BS1

BS4

BS3

BP5

BP7

BP4

BP3

BP1

BP2

PVS1

Expert Panel

CDH1 VCEP [↗](#)

Criteria Specification Information **!**

[↗](#) Criteria Specifications for this VCEP

Evidence submitted by expert panel

CDH1 VCEP

The c.121G>T (NM_004360.5) variant in CDH1 is a missense variant predicted to predicted to cause substitution of Val by Leu at amino acid 41 (p.Val41Leu). This variant has been observed in Variant seen in >3 heterozygous individuals without GC, DGC, SRC tumours or LBC and whose families do not suggest HDGC (BS2_Supporting; Ambry, GeneDx, Invitae). This variant is absent from gnomAD 2.1.1 (PM2_Supporting). In summary, this variant is classified as uncertain significance for DGLBCS based on the ACMG/AMP criteria applied, as specified by the ClinGen CDH1 VCEP: PM2_Supporting, BS2_Supporting. (CDH1 VCEP specifications version 3.1; 04/24/2023)

Met criteria codes

PM2_Supporting			This variant is absent from populations in gnomAD 2.1.1 (PM2_Supporting).
BS2_Supporting			This variant has been observed in 7 heterozygous individuals without GC, DGC, SRC tumours or LBC and whose families do not suggest HDGC (BS2_Supporting; Ambry, GeneDx, Invitae).

Not Met criteria codes

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			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BA1			Variant absent from gnomAD
PP1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP2			NA
PP3			NA
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
BS1		✖	Variant absent from 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
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
BP4		✖	NA
BP3		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP1		✖	NA
BP2		✖	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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