

Variant: *NM_004360.5(CDH1):c.125C>T (p.Pro42Leu)*

Version: 1.0

CA10580073 [↗](#)

231745 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 769d2160-06b8-4eb7-894a-450600260917

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HGVS expressions

NM_004360.5:c.125C>T

NM_004360.5(CDH1):c.125C>T (p.Pro42Leu)

NC_000016.10:g.68738373C>T

CM000678.2:g.68738373C>T

NC_000016.9:g.68772276C>T

CM000678.1:g.68772276C>T

NC_000016.8:g.67329777C>T

NG_008021.1:g.6082C>T

ENST00000261769.10:c.125C>T

ENST00000261769.9:c.125C>T

ENST00000422392.6:c.125C>T

ENST00000566510.5:c.125C>T

ENST00000566612.5:c.125C>T

ENST00000611625.4:c.125C>T

ENST00000612417.4:c.125C>T

ENST00000621016.4:c.125C>T

NM_004360.3:c.125C>T

NM_001317184.1:c.125C>T

NM_001317185.1:c.-1491C>T

NM_001317186.1:c.-1695C>T

NM_004360.4:c.125C>T

NM_001317184.2:c.125C>T

NM_001317185.2:c.-1491C>T

NM_001317186.2:c.-1695C>T

Likely Benign

Met criteria codes **1**

BS2

Not Met criteria codes **25**

PS1	PS2	PS3	PS4	BP5	BP7
BP4	BP3	BP1	BP2	PP1	PP2
PP3	PP4	PM6	PM2	PM1	
PM3	PM5	PM4	BA1	BS1	
BS4	BS3	PVS1			

Expert Panel

CDH1 VCEP [↗](#)

Criteria Specification Information **!**

[↗](#) Criteria Specifications for this VCEP

Evidence submitted by expert panel

CDH1 VCEP

The c.125C>T (NM_004360.5) variant in CDH1 is a missense variant predicted to cause substitution of Pro by Leu at amino acid 42 (p.Pro42Leu). This variant has been observed in more than 10 heterozygous individuals with no DGC, SRC tumours and whose families do not suggest HDGC (BS2; GeneDx, Invitae, Color). In summary, the CDH1 VCEP classified the variant as likely benign for DGLBCS based on BS2 alone based on the ACMG/AMP criteria applied, as specified by the ClinGen CDH1 VCEP: BS2. (CDH1 VCEP specifications version 3.1; 04/24/2023)

Met criteria codes

BS2			This variant has been observed in more than 10 heterozygous individuals with no DGC, SRC tumours and whose families do not suggest HDGC (BS2; GeneDx, Invitae, Color).
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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
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			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
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
PP4		✖	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		✖	Variant absent in gnomAD v2.1.1. Variant present in 2 of 143312 alleles in gnomAD v3 (2 of 13658 alleles in Latino population)
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
BA1		✖	Variant absent in gnomAD v2.1.1. Variant present in 2 of 143312 alleles in gnomAD v3 (2 of 13658 alleles in Latino population)
BS1		✖	Variant absent in gnomAD v2.1.1. Variant present in 2 of 143312 alleles in gnomAD v3 (2 of 13658 alleles in Latino population)
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
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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