

Variant: NM_004360.5(CDH1):c.1118C>T (p.Pro373Leu)

CA167953 [↗](#)

142285 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 1aac37aa-cb97-4617-9023-3e7f155f338f

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HGVS expressions

NM_004360.5(CDH1):c.1118C>T (p.Pro373Leu)

NM_004360.5:c.1118C>T

NC_000016.10:g.68812244C>T

CM000678.2:g.68812244C>T

NC_000016.9:g.68846147C>T

CM000678.1:g.68846147C>T

NC_000016.8:g.67403648C>T

NG_008021.1:g.79953C>T

ENST00000261769.10:c.1118C>T

ENST00000261769.9:c.1118C>T

ENST00000422392.6:c.1118C>T

ENST00000562836.5:n.1189C>T

ENST00000565810.1:n.162C>T

ENST00000566510.5:c.962C>T

ENST00000566612.5:c.1118C>T

ENST00000611625.4:c.1118C>T

ENST00000612417.4:c.1118C>T

ENST00000621016.4:c.1118C>T

NM_004360.3:c.1118C>T

NM_001317184.1:c.1118C>T

NM_001317185.1:c.-498C>T

NM_001317186.1:c.-702C>T

NM_004360.4:c.1118C>T

NM_001317184.2:c.1118C>T

NM_001317185.2:c.-498C>T

NM_001317186.2:c.-702C>T

Likely Benign

Met criteria codes 1

BS2

Not Met criteria codes 25

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

Expert Panel

CDH1 VCEP [↗](#)

Criteria Specification Information

[↗](#) **Criteria Specification:** ClinGen CDH1 Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines Version 3.1

[↗](#) **Criteria Specification Approval History**

Evidence submitted by expert panel

CDH1 VCEP

The c.1118C>T (p.Pro373Leu) variant has been observed in >10 individuals 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, the clinical significance of this variant is classified as likely benign based on BS2 alone. ACMG/AMP criteria applied, as specified by the CDH1 Variant Curation Expert Panel (Variant Interpretation Guidelines Version 3.1): BS2.

Met criteria codes

BS2			Observed in 32 non-HDGC patients
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Not Met criteria codes

BS4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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BS3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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BS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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PVS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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BP5			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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BP7			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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BP2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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BP3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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BP1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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BP4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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BA1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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PS2		✗	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
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
PP4		✗	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
PM6		✗	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM2		✗	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
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



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