

Variant: *NM_004360.5(CDH1):c.2253C>T (p.Asn751=)*

Version: 2.0

CA167533 [↗](#)

142145 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: d4fc3027-83dc-4710-bfde-83d2dd1e1f28

Approved on: 2023/8/10

Published on: 2023/8/10

HGVS expressions

NM_004360.5:c.2253C>T

NM_004360.5(CDH1):c.2253C>T (p.Asn751=)

NC_000016.10:g.68828262C>T

CM000678.2:g.68828262C>T

NC_000016.9:g.68862165C>T

CM000678.1:g.68862165C>T

NC_000016.8:g.67419666C>T

NG_008021.1:g.95971C>T

ENST00000261769.10:c.2253C>T

ENST00000261769.9:c.2253C>T

ENST00000422392.6:c.2070C>T

ENST00000562118.1:n.471C>T

ENST00000562836.5:n.2324C>T

ENST00000566510.5:c.*919C>T

ENST00000566612.5:c.*493C>T

ENST00000611625.4:c.2316C>T

ENST00000612417.4:c.1853+1708C>T

ENST00000621016.4:c.1866-5941C>T

NM_004360.3:c.2253C>T

NM_001317184.1:c.2070C>T

NM_001317185.1:c.705C>T

NM_001317186.1:c.288C>T

NM_004360.4:c.2253C>T

NM_001317184.2:c.2070C>T

NM_001317185.2:c.705C>T

NM_001317186.2:c.288C>T

Benign

Met criteria codes **2**

BP2 BA1

Not Met criteria codes **24**

PS1 PS2 PS3 PS4 BP5 BP7
BP4 BP3 BP1 PP1 PP2 PP3
PP4 PM6 PM2 PM1 PM3

Expert Panel

CDH1 VCEP [↗](#)

Criteria Specification Information

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

Evidence submitted by expert panel

CDH1 VCEP

The NM_004360.5(CDH1):c.2253C>T (p.Asn751=) variant has an allele frequency of 0.10006 (10%, 3546/35438 alleles, 185 homozygotes) in the Latino subpopulation of the gnomAD v2.1.1 cohort (BA1; BP2). Therefore, 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): BA1, BP2.

Met criteria codes

BP2			185 Latino homozygotes in the controls cohort in gnomAD v2.1.1
BA1			MAF > 0.2%: Latino AF is 0.10006 (3546/35438 alleles, 185 homozygotes in gnomAD v2.1.1)

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			Not assessed, because BA1 met
BP7			Not assessed, because BA1 met
BP4			Not assessed, because BA1 met
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
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		✖	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
BS1		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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
BS2		✖	Not assessed, because BA1 met
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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