

Variant: *NM_004360.5(CDH1):c.324A>G (p.Arg108=)*

Version: 2.0

CA290000 [↗](#)

136691 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: d6d29fdd-b27d-4917-bbcc-7116b29d2805

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HGVS expressions

NM_004360.5:c.324A>G

NM_004360.5(CDH1):c.324A>G (p.Arg108=)

NC_000016.10:g.68801830A>G

CM000678.2:g.68801830A>G

NC_000016.9:g.68835733A>G

CM000678.1:g.68835733A>G

NC_000016.8:g.67393234A>G

NG_008021.1:g.69539A>G

ENST00000261769.10:c.324A>G

ENST00000261769.9:c.324A>G

ENST00000422392.6:c.324A>G

ENST00000561751.1:c.91A>G

ENST00000562836.5:n.395A>G

ENST00000564676.5:n.606A>G

ENST00000564745.1:n.319A>G

ENST00000566510.5:c.324A>G

ENST00000566612.5:c.324A>G

ENST00000611625.4:c.324A>G

ENST00000612417.4:c.324A>G

ENST00000621016.4:c.324A>G

NM_004360.3:c.324A>G

NM_001317184.1:c.324A>G

NM_001317185.1:c.-1292A>G

NM_001317186.1:c.-1496A>G

NM_004360.4:c.324A>G

NM_001317184.2:c.324A>G

NM_001317185.2:c.-1292A>G

NM_001317186.2:c.-1496A>G

Benign

Met criteria codes **2**

BA1 BP2

Not Met criteria codes **24**

PS1 PS2 PS3 PS4 PP1 PP2
PP3 PP4 PM1 PM3 PM5

Expert Panel

CDH1 VCEP [↗](#)

Criteria Specification Information

Evidence submitted by expert panel

CDH1 VCEP

The NM_004360.5(CDH1):c.324A>G (p.Arg108=) variant has an allele frequency of 0.00769 (0.769%, 192/24966 alleles) in the African subpopulation and one homozygote in the European (Non-Finnish) 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

BA1			MAF > 0.2%: African AF is 0.00769 (192/24966 alleles in gnomAD v2.1.1)
BP2			One European (Non-Finnish) homozygote in the controls cohort 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
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
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
PM2		✖	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
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

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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