

Variant: *NM_004360.5(CDH1):c.269G>A (p.Arg90Gln)*

Version: 2.0

CA169145 [↗](#)

142692 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: d6a83f99-4418-4412-a73a-bedd91dad34c

Approved on: 2023/8/10

Published on: 2023/8/10

HGVS expressions

NM_004360.5:c.269G>A

NM_004360.5(CDH1):c.269G>A (p.Arg90Gln)

NC_000016.10:g.68801775G>A

CM000678.2:g.68801775G>A

NC_000016.9:g.68835678G>A

CM000678.1:g.68835678G>A

NC_000016.8:g.67393179G>A

NG_008021.1:g.69484G>A

ENST00000261769.10:c.269G>A

ENST00000261769.9:c.269G>A

ENST00000422392.6:c.269G>A

ENST00000561751.1:c.36G>A

ENST00000562836.5:n.340G>A

ENST00000564676.5:n.551G>A

ENST00000564745.1:n.264G>A

ENST00000566510.5:c.269G>A

ENST00000566612.5:c.269G>A

ENST00000611625.4:c.269G>A

ENST00000612417.4:c.269G>A

ENST00000621016.4:c.269G>A

NM_004360.3:c.269G>A

NM_001317184.1:c.269G>A

NM_001317185.1:c.-1347G>A

NM_001317186.1:c.-1551G>A

NM_004360.4:c.269G>A

NM_001317184.2:c.269G>A

NM_001317185.2:c.-1347G>A

NM_001317186.2:c.-1551G>A

Likely Benign

Met criteria codes **1**

BS2

Not Met criteria codes **25**

BA1

BP4

BP3

BP1

BP2

BP5

BP7

BS1

BS4

BS3

PP1

PP2

Expert Panel

CDH1 VCEP [↗](#)

Criteria Specification Information

PP3

PP4

PS1

PS2

PS3

PS4

PM6

PM2

PM1

PM3

PM5

PM4

PVS1

Evidence Links 0

Criteria Specification: ClinGen CDH1 Expert Panel

Specifications to the ACMG/AMP Variant Interpretation Guidelines Version 3.1

Criteria Specification Approval History

Criteria Specifications for this VCEP

Evidence submitted by expert panel

CDH1 VCEP

The c.269G>A (p.Arg90Gln) missense variant has a frequency of 0.00001989 (5 of 251,334) in the gnomAD v2.1.1 cohort, with a maximum non-founder allele frequency of 0.00004401 (5 of 113,622) in the European non-Finnish subpopulation (<http://gnomad.broadinstitute.org>). This variant has been observed in ≥10 (55) individuals without DGC, SRC tumours or LBC and whose families do not suggest HDGC (BS2; SCV000567761.3, SCV000254829.7). 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			Variant seen in >10 (55) individuals w/o DCG, SRC tumors, or LBC & whose families do not suggest HDGC (at least 50 cases between Invitae, GeneDx, Ambry)
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Not Met criteria codes

BA1			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
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
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
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
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
PM6		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM2		✖	frequency of 0.00001989 (5 of 251,334) in the gnomAD v2.1.1 cohort, with a maximum non-founder allele frequency of 0.00004401 (5 of 113,622) in the European non-Finnish subpopulation
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
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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