

Variant: NM_004360.4(CDH1):c.188G>A (p.Arg63Gln)

CA288048 [↗](#)

127919 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 4de15575-228f-4350-8bab-2e2dbfd23694

Approved on: 2018-11-21

Published on: 2018-12-07

HGVS expressions

NM_004360.4:c.188G>A

NM_004360.4(CDH1):c.188G>A (p.Arg63Gln)

NC_000016.10:g.68801694G>A

CM000678.2:g.68801694G>A

NC_000016.9:g.68835597G>A

CM000678.1:g.68835597G>A

NC_000016.8:g.67393098G>A

NG_008021.1:g.69403G>A

ENST00000261769.10:c.188G>A

ENST00000261769.9:c.188G>A

ENST00000422392.6:c.188G>A

ENST00000562836.5:n.259G>A

ENST00000564676.5:n.470G>A

ENST00000564745.1:n.183G>A

ENST00000566510.5:c.188G>A

ENST00000566612.5:c.188G>A

ENST00000611625.4:c.188G>A

ENST00000612417.4:c.188G>A

ENST00000621016.4:c.188G>A

NM_004360.3:c.188G>A

NM_001317184.1:c.188G>A

NM_001317185.1:c.-1428G>A

NM_001317186.1:c.-1632G>A

NM_004360.5:c.188G>A

NM_001317184.2:c.188G>A

NM_001317185.2:c.-1428G>A

NM_001317186.2:c.-1632G>A

NM_004360.5(CDH1):c.188G>A (p.Arg63Gln)

Benign

Met criteria codes 2

BS2 BS1

Not Met criteria codes 24

BA1 BP2 BP3 BP1 BP4 BP7
BP5 BS4 BS3 PP4 PP1 PP2
PP3 PS2 PS4 PS3 PS1 PM6

Expert Panel

CDH1 VCEP [↗](#)

Criteria Specification Information

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

Evidence submitted by expert panel

CDH1 VCEP

The c.188G>A (p.Arg63Gln) variant has an allele frequency of 0.00109 (0.11%, 6/5486 alleles) in the “Other” subpopulation of the gnomAD cohort (BS1). This variant has also been observed in >10 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, 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): BS1, BS2.

Met criteria codes

BS2			Observed in 61 individuals w/o dx of HDGC
BS1			The "other" population in gnomAD meets criteria.

Not Met criteria codes

BA1			The "other" population in gnomAD meets criteria.
BP2			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
BP4			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
BP5			It has seen co-occurrence with a mutation in another gene that clearly explains a proband's phenotype.
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
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
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
PM6			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM2			The "other" population in gnomAD meets criteria.
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

PVS1

No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History

Showing 1 to 2 of 2 rows

The information on this website is not intended for direct diagnostic use or medical decision-making without review by a genetics professional. Individuals should not change their health behavior solely on the basis of information contained on this website. If you have questions about the information contained on this website, please see a health care professional.