

Variant: *NM_004360.5(CDH1):c.2076T>C (p.Ala692=)*

Version: 2.0

CA169363 [↗](#)

142770 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 7147ec7b-30b2-419d-9646-4283a89cbe74

Approved on: 2023/8/9

Published on: 2023/8/10

HGVS expressions

NM_004360.5(CDH1):c.2076T>C (p.Ala692=)

NM_004360.5:c.2076T>C

NC_000016.10:g.68823538T>C

CM000678.2:g.68823538T>C

NC_000016.9:g.68857441T>C

CM000678.1:g.68857441T>C

NC_000016.8:g.67414942T>C

NG_008021.1:g.91247T>C

ENST00000261769.10:c.2076T>C

ENST00000261769.9:c.2076T>C

ENST00000422392.6:c.1893T>C

ENST00000562118.1:n.294T>C

ENST00000562836.5:n.2147T>C

ENST00000566510.5:c.*742T>C

ENST00000566612.5:c.*316T>C

ENST00000611625.4:c.2139T>C

ENST00000612417.4:c.1830+1419T>C

ENST00000621016.4:c.1865+1384T>C

NM_004360.3:c.2076T>C

NM_001317184.1:c.1893T>C

NM_001317185.1:c.528T>C

NM_001317186.1:c.111T>C

NM_004360.4:c.2076T>C

NM_001317184.2:c.1893T>C

NM_001317185.2:c.528T>C

NM_001317186.2:c.111T>C

Benign

Met criteria codes 2

BA1 BP2

Not Met criteria codes 24

BS1 BS4 BS3 BS2 BP5 BP7
BP4 BP3 BP1 PVS1 PS1
PS2 PS3 PS4 PP1 PP2 PP3

Expert Panel

CDH1 VCEP [↗](#)

Criteria Specification Information

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

PP4

PM1

PM3

PM5

PM4

PM6

PM2

[Criteria Specification Approval History](#)
[Criteria Specifications for this VCEP](#)

Evidence Links 0

Evidence submitted by expert panel			
<i>CDH1 VCEP</i>			
The NM_004360.5(CDH1):c.2076T>C (p.Ala692=) variant has an allele frequency of 0.88894 (88.89%, 22156/24924 alleles, 9856 homozygotes) in the African subpopulation of the gnomAD v2.1.1 cohort (BA1; BP2_Supporting). 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_Supporting.			
Met criteria codes			
BA1		✓	MAF > 0.2%: African AF is 0.88894 (22156/24924 alleles, 9856 homozygotes in gnomAD v2.1.1)
BP2		✓	9856 Africa homozygotes in the controls cohort in gnomAD v2.1.1
Not Met criteria codes			
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
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
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Curation History ↗		
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