

Variant: *NM_004360.5(CDH1):c.1896C>T (p.His632=)*

Version: 2.0

CA165821 [↗](#)

141570 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 9aea178a-3518-4d66-ae22-80a965e9331b

Approved on: 2023/8/10

Published on: 2023/8/10

HGVS expressions

NM_004360.5(CDH1):c.1896C>T (p.His632=)

NM_004360.5:c.1896C>T

NC_000016.10:g.68822185C>T

CM000678.2:g.68822185C>T

NC_000016.9:g.68856088C>T

CM000678.1:g.68856088C>T

NC_000016.8:g.67413589C>T

NG_008021.1:g.89894C>T

ENST00000261769.10:c.1896C>T

ENST00000261769.9:c.1896C>T

ENST00000422392.6:c.1713C>T

ENST00000562836.5:n.1967C>T

ENST00000566510.5:c.*562C>T

ENST00000566612.5:c.*136C>T

ENST00000611625.4:c.1959C>T

ENST00000612417.4:c.1830+66C>T

ENST00000621016.4:c.1865+31C>T

NM_004360.3:c.1896C>T

NM_001317184.1:c.1713C>T

NM_001317185.1:c.348C>T

NM_001317186.1:c.-70C>T

NM_004360.4:c.1896C>T

NM_001317184.2:c.1713C>T

NM_001317185.2:c.348C>T

NM_001317186.2:c.-70C>T

Benign

Met criteria codes 2

BA1 BP2

Not Met criteria codes 24

BP4 BP3 BP1 BP5 BP7 BS2
BS1 BS4 BS3 PP1 PP2 PP3
PP4 PS1 PS2 PS3 PS4 PM6
PM2 PM1 PM3 PM5 PM4
PVS1

Expert Panel

CDH1 VCEP [↗](#)

Criteria Specification Information

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

[↗](#) **Criteria Specification Approval History**

Evidence submitted by expert panel

CDH1 VCEP

The NM_004360.5(CDH1):c.1896C>T (p.His632=) variant has an allele frequency of 0.05030 (5%, 1255/24950 alleles, 32 homozygotes) in the African 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 0.05030 (1255/24950 alleles, 32 homozygotes in gnomAD v2.1.1)
BP2			Thirty-two Africa homozygotes in the controls cohort in gnomAD v2.1.1

Not Met criteria codes

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
BP5			Not assessed, because BA1 met
BP7			Not assessed, because BA1 met
BS2			Not assessed, because BA1 met
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			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
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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