

Variant: NM_004360.4(CDH1):c.604G>A (p.Val202Ile)

CA163776 [↗](#)

133853 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 4f56b139-4c6b-4107-aebc-4368f6c6f68b

Approved on: 2018-11-21

Published on: 2018-12-07

HGVS expressions

NM_004360.4(CDH1):c.604G>A (p.Val202Ile)

NM_004360.4:c.604G>A

NC_000016.10:g.68808765G>A

CM000678.2:g.68808765G>A

NC_000016.9:g.68842668G>A

CM000678.1:g.68842668G>A

NC_000016.8:g.67400169G>A

NG_008021.1:g.76474G>A

ENST00000261769.10:c.604G>A

ENST00000261769.9:c.604G>A

ENST00000422392.6:c.604G>A

ENST00000561751.1:c.371G>A

ENST00000562836.5:n.675G>A

ENST00000564676.5:n.886G>A

ENST00000566510.5:c.531+198G>A

ENST00000566612.5:c.604G>A

ENST00000567320.1:n.114G>A

ENST00000611625.4:c.604G>A

ENST00000612417.4:c.604G>A

ENST00000621016.4:c.604G>A

NM_004360.3:c.604G>A

NM_001317184.1:c.604G>A

NM_001317185.1:c.-1012G>A

NM_001317186.1:c.-1216G>A

NM_004360.5:c.604G>A

NM_001317184.2:c.604G>A

NM_001317185.2:c.-1012G>A

NM_001317186.2:c.-1216G>A

NM_004360.5(CDH1):c.604G>A (p.Val202Ile)

Benign

Met criteria codes 1

BA1

Not Met criteria codes 25

BS4 BS3 BS1 BS2 BP5 BP7
BP2 BP3 BP1 BP4 PVS1

Expert Panel

CDH1 VCEP [↗](#)

Criteria Specification Information

PS2

PS4

PS3

PS1

PP4

PP1

PP2

PP3

PM3

PM1

PM4

PM5

PM6

PM2

Evidence Links

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- 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.604G>A (p.Val202Ile) variant has an allele frequency of 0.00354 (0.35%, 109/30,772 alleles) in the South Asian subpopulation of the gnomAD cohort (BA1). 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): BA1.

Met criteria codes

BA1			South Asian subpopulation in 1000 genomes has the allele at a frequency of 1% (10 alleles)
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Not Met criteria codes

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
BS1			South Asian subpopulation in 1000 genomes has the allele at a frequency of 1% (10 alleles)
BS2			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
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
PVS1			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
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
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

PM6	 	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM2		South Asian subpopulation in 1000 genomes has the allele at a frequency of 1% (10 alleles)

Curation History 	
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