

Variant: *NM_004360.5(CDH1):c.160A>G (p.Arg54Gly)*

Version: 2.0

[CA293951](#)

[140862 \(ClinVar\)](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 1511cd87-14c9-4efe-a6e9-3a9e43b2a6e2

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HGVS expressions

NM_004360.5:c.160A>G

NM_004360.5(CDH1):c.160A>G (p.Arg54Gly)

NC_000016.10:g.68738408A>G

CM000678.2:g.68738408A>G

NC_000016.9:g.68772311A>G

CM000678.1:g.68772311A>G

NC_000016.8:g.67329812A>G

NG_008021.1:g.6117A>G

ENST00000261769.10:c.160A>G

ENST00000261769.9:c.160A>G

ENST00000422392.6:c.160A>G

ENST00000566510.5:c.160A>G

ENST00000566612.5:c.160A>G

ENST00000611625.4:c.160A>G

ENST00000612417.4:c.160A>G

ENST00000621016.4:c.160A>G

NM_004360.3:c.160A>G

NM_001317184.1:c.160A>G

NM_001317185.1:c.-1456A>G

NM_001317186.1:c.-1660A>G

NM_004360.4:c.160A>G

NM_001317184.2:c.160A>G

NM_001317185.2:c.-1456A>G

NM_001317186.2:c.-1660A>G

Likely Benign

Met criteria codes **1**

BS2

Not Met criteria codes **25**

PVS1 BS1 BS4 BS3 BP4
BP3 BP1 BP2 BP5 BP7 PS1
PS2 PS3 PS4 BA1 PP1 PP2
PP3 PP4 PM1 PM3 PM5
PM4 PM6 PM2

Expert Panel

Criteria Specification Information

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.160A>G (p.Arg54Gly) missense variant has a frequency of 0.00002737 (5 of 182,668) in the gnomAD v2.1.1 cohort, with a maximum non-founder allele frequency of 0.00004080 (3 of 73,538) in the non-Finnish European subpopulation (<http://gnomad.broadinstitute.org>). This variant has been observed in >50 individuals without DGC, SRC tumours or LBC and whose families do not suggest HDGC (BS2; SCV000183777.5, SCV000545364.6, SCV000210888.14). 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 observed in > 50 individuals without DGC, SRC tumors, or LBC & whose families do not suggest HDGC
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Not Met criteria codes


PVS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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BS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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BS4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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BS3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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BP4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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BP3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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BP1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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BP2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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BP5			
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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
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
BA1			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		frequency of 0.00002737 (5 of 182,668) in the gnomAD v2.1.1 cohort, with a maximum non-founder allele frequency of 0.00004080 (3 of 73,538) in the non-Finnish European subpopulation

Curation History

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