

Variant: *NM_004360.5(CDH1):c.940A>T (p.Lys314Ter)*

Version: 2.0

CA396459763 [↗](#)

428632 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 8277d227-4d10-4ebb-bc74-fee3cba23a52

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HGVS expressions

NM_004360.5:c.940A>T

NM_004360.5(CDH1):c.940A>T (p.Lys314Ter)

NC_000016.10:g.68811791A>T

CM000678.2:g.68811791A>T

NC_000016.9:g.68845694A>T

CM000678.1:g.68845694A>T

NC_000016.8:g.67403195A>T

NG_008021.1:g.79500A>T

ENST00000261769.10:c.940A>T

ENST00000261769.9:c.940A>T

ENST00000422392.6:c.940A>T

ENST00000561751.1:c.562A>T

ENST00000562836.5:n.1011A>T

ENST00000566510.5:c.784A>T

ENST00000566612.5:c.940A>T

ENST00000611625.4:c.940A>T

ENST00000612417.4:c.940A>T

ENST00000621016.4:c.940A>T

NM_004360.3:c.940A>T

NM_001317184.1:c.940A>T

NM_001317185.1:c.-676A>T

NM_001317186.1:c.-880A>T

NM_004360.4:c.940A>T

NM_001317184.2:c.940A>T

NM_001317185.2:c.-676A>T

NM_001317186.2:c.-880A>T

Pathogenic

Met criteria codes **4**

PVS1

PM2_Supporting

PS4_Supporting

PM5_Supporting

Not Met criteria codes **22**

BS1

BS4

BS3

BS2

BP5

BP7

BP4

BP3

BP1

BP2

BA1

PS1

Expert Panel

CDH1 VCEP [↗](#)

Criteria Specification Information

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

Evidence submitted by expert panel

CDH1 VCEP

The c.940A>T (p.Lys314Ter) variant is predicted to result in a premature termination codon in exon 7 (PVS1, PM5_Supporting). The variant is absent from the gnomAD population database (PM2_Supporting; <https://gnomad.broadinstitute.org/>). This variant has previously been reported in one individual meeting IGCLC criteria for HDGC (PS4_Supporting; PMID: 26182300). In summary, this variant meets criteria to be classified as pathogenic based on ACMP/AMP criteria as specified by the CDH1 Variant Curation Expert Panel (Variant Interpretation Guidelines Version 3.1): PVS1, PM2_Supporting, PS4_Supporting, PM5_Supporting.

Met criteria codes

PVS1			This variant results in a premature termination codon in exon 7.
PM2_Supporting			This variant is absent from populations in gnomAD, ExAC, 1000 Genomes and ESP.
PS4_Supporting			This variant was reported in one individual meeting IGCLC criteria for HDGC based on a personal and family history of diffuse gastric cancer (PMID: 26182300).
PM5_Supporting			Apply PM5_Supporting to nonsense/frameshift variants that are predicted/proved to undergo NMD.

Not Met criteria codes

BS1			This variant is absent from populations in gnomAD, ExAC, 1000 Genomes and ESP.
BS4			To our knowledge, segregation of this variant has not been reported.
BS3			BS3 does not apply to this variant.
BS2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP5			To our knowledge, this variant has not been reported in a case with an alternate molecular basis for disease.
BP7			BP7 does not apply to this variant.
BP4			BP4 does not apply to this variant.

BP3		✖	BP3 does not apply to CDH1.
BP1		✖	BP1 does not apply to CDH1.
BP2		✖	To our knowledge, this variant has not been observed in cis or trans with a known pathogenic variant.
BA1		✖	This variant is absent from populations in gnomAD, ExAC, 1000 Genomes and ESP.
PS1		✖	PS1 does not apply to this variant.
PS2		✖	To our knowledge, this variant has not been reported as de novo.
PS3		✖	PS3 does not apply to this variant.
PP1		✖	To our knowledge, segregation of this variant has not been reported.
PP2		✖	PP2 does not apply to CDH1.
PP3		✖	PP3 does not apply to this variant.
PP4		✖	PP4 does not apply to CDH1.
PM6		✖	To our knowledge, this variant has not been reported as de novo.
PM1		✖	PM1 does not apply to this variant.
PM3		✖	PM3 does not apply to CDH1.
PM4		✖	PM4 does not apply to this variant.

Curation History 

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▾

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