

Variant: NM_004360.4(CDH1):c.2506G>T (p.Glu836Ter)

CA396472215 [↗](#)

479504 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 1a5e5e0d-db23-4e6c-a7f2-002891504b0f

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HGVS expressions

NM_004360.4:c.2506G>T

NM_004360.4(CDH1):c.2506G>T (p.Glu836Ter)

NC_000016.10:g.68833356G>T

CM000678.2:g.68833356G>T

NC_000016.9:g.68867259G>T

CM000678.1:g.68867259G>T

NC_000016.8:g.67424760G>T

NG_008021.1:g.101065G>T

ENST00000261769.10:c.2506G>T

ENST00000261769.9:c.2506G>T

ENST00000422392.6:c.2323G>T

ENST00000562118.1:n.724G>T

ENST00000562836.5:n.2577G>T

ENST00000566510.5:c.*1172G>T

ENST00000566612.5:c.*746G>T

ENST00000611625.4:c.2569G>T

ENST00000612417.4:c.1854-835G>T

ENST00000621016.4:c.1866-847G>T

NM_004360.3:c.2506G>T

NM_001317184.1:c.2323G>T

NM_001317185.1:c.958G>T

NM_001317186.1:c.541G>T

NM_004360.5:c.2506G>T

NM_001317184.2:c.2323G>T

NM_001317185.2:c.958G>T

NM_001317186.2:c.541G>T

NM_004360.5(CDH1):c.2506G>T (p.Glu836Ter)

Pathogenic

Met criteria codes 3

PS2

PM2_Supporting

PVS1_Strong

Not Met criteria codes 23

BP5

BP7

BP2

BP3

BP1

BP4

PS4

PS3

PS1

PP4

PP1

PP2

PP3

PM3

PM1

PM4

PM5

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.2506G>T (p.Glu836*) variant results in a premature stop codon that leads to a truncated protein. While it is located within the nonsense mediated decay resistance region,it is recognized as the most c-terminal pathogenic variant in CDH1 (PVS1_Strong, PMID: 29798843). This variant is absent in the gnomAD cohort (PM2_Supporting; <http://gnomad.broadinstitute.org>). There is one known de novo observation with parental confirmation in a patient with diffuse gastric cancer and/or lobular breast cancer (PS2; PMID: 29798843). In summary, this variant meets criteria to be classified as likely pathogenic based on the ACMG/AMP criteria applied as specified by the CDH1 Variant Curation Expert Panel: PVS1_Strong, PM2_Supporting, PS2.

Met criteria codes

PS2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM2_Supporting			Absent from controls in gnomAD
PVS1_Strong			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Not Met criteria codes

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			
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
BA1		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS2		✖	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
BS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History 		
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