The ClinGen Evidence Repository is an **FDA-recognized human genetic variant database** containing expert-curated assertions regarding variants' pathogenicity and supporting evidence summaries. [Disclaimer]

Variant: NM_004360.5(CDH1):c.2505T>G (p.Tyr835Ter)

Version: 1.3

CA396472208

921477 (ClinVar)

Gene: CDH1 (HGNC:999)
Condition: CDH1-related diffuse gastric and lobular breast cancer (MONDO:0100488)
Inheritance Mode: Autosomal dominant inheritance
UUID: e4b09407-f4ff-42aa-8b88-e45bbdd1299f
Approved on: 2023/8/4
Published on: 2023/8/4

HGVS expressions

NM_004360.5:c.2505T>G
NM_004360.5(CDH1):c.2505T>G (p.Tyr835Ter)
NC_000016.10:g.68833355T>G
CM000678.2:g.68833355T>G
NC_000016.9:g.68867258T>G
CM000678.1:g.68867258T>G
NC_000016.8:g.67424759T>G
NG_008021.1:g.101064T>G
ENST00000261769.10:c.2505T>G
ENST00000261769.9:c.2505T>G
ENST00000422392.6:c.2322T>G
ENST00000562118.1:n.723T>G
ENST00000562836.5:n.2576T>G
ENST00000566510.5:c.*1171T>G
ENST00000566612.5:c.*745T>G
ENST00000611625.4:c.2568T>G
ENST00000612417.4:c.1854-836T>G
ENST00000621016.4:c.1866-848T>G
NM_004360.3:c.2505T>G
NM_001317184.1:c.2322T>G
NM_001317185.1:c.957T>G
NM_001317186.1:c.540T>G
NM_004360.4:c.2505T>G
NM_001317184.2:c.2322T>G
NM_001317185.2:c.957T>G
NM_001317186.2:c.540T>G

Likely Pathogenic

Met criteria codes 3

PM2_Supporting PVS1_Strong PM5_Supporting

Not Met criteria codes 23

BA1 PP1 PP2 PP3 PP4 PM6 PM1 PM3 PM4 BS1 BS4 BS3 BS2 PS1 PS2 PS3

Expert Panel

CDH1 VCEP

Criteria Specification Information

Criteria Specification: *ClinGen CDH1 Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines Version 3.1*

PS4 BP5 BP7 BP4 BP3 BP1 BP2

Criteria Specification Approval History

Criteria Specifications for this VCEP

Evidence Links 0

Evidence submitted by expert panel

CDH1 VCEP

The c.2505T>G (p.Tyr835Ter) variant results in a premature translational stop signal upstream of the most 3' well-characterized pathogenic variant c.2506G>T (p.Glu836Ter) in a region that is not predicted to undergo nonsense-mediated decay (PVS1_Strong, PM5_Supporting). It is absent in the gnomAD cohort (PM2_Supporting; http://https://gnomad.broadinstitute.org/). 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, PM5_Supporting. (CDH1 VCEP specifications version 3.1; 03/27/2023)

Met criteria codes

PM2_Supporting	✔	Absent in gnomAD v2.1.1, coverage >30X
PVS1_Strong	✔	Not predicted to undergo NMD, nonsense variant between c.2389-c.2508
PM5_Supporting	✔	PM5_Supporting can be applied to truncating variants located upstream of the last known pathogenic truncating variant [c.2506G>T (p.Glu836Ter)].

Not Met criteria codes

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
PM6	✖	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
PM4	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS1	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS4	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS3	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS2	ℹ	✖	Variant not seen at Ambry, Invitae, GeneDx
PS1		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS2	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS3	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS4	ℹ	✖	Variant not seen at Ambry, Invitae, GeneDx
BP5	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP7	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP4	ℹ	✖	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

BP2		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History

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