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_004360.4(CDH1):c.337A>T (p.Lys113Ter)

Version: 3.0

CA10577535

234594 (ClinVar)

Gene: CDH1 (HGNC:999)
Condition: CDH1-related diffuse gastric and lobular breast cancer (MONDO:0100488)
Inheritance Mode: Autosomal dominant inheritance
UUID: 42e9bffc-fe2b-4d76-b132-479a024adf4e
Approved on: 2023/8/4
Published on: 2023/8/4

HGVS expressions

NM_004360.4:c.337A>T
NM_004360.4(CDH1):c.337A>T (p.Lys113Ter)
NC_000016.10:g.68801843A>T
CM000678.2:g.68801843A>T
NC_000016.9:g.68835746A>T
CM000678.1:g.68835746A>T
NC_000016.8:g.67393247A>T
NG_008021.1:g.69552A>T
ENST00000261769.10:c.337A>T
ENST00000261769.9:c.337A>T
ENST00000422392.6:c.337A>T
ENST00000561751.1:c.104A>T
ENST00000562836.5:n.408A>T
ENST00000564676.5:n.619A>T
ENST00000564745.1:n.332A>T
ENST00000566510.5:c.337A>T
ENST00000566612.5:c.337A>T
ENST00000611625.4:c.337A>T
ENST00000612417.4:c.337A>T
ENST00000621016.4:c.337A>T
NM_004360.3:c.337A>T
NM_001317184.1:c.337A>T
NM_001317185.1:c.-1279A>T
NM_001317186.1:c.-1483A>T
NM_004360.5:c.337A>T
NM_001317184.2:c.337A>T
NM_001317185.2:c.-1279A>T
NM_001317186.2:c.-1483A>T

Pathogenic

Met criteria codes 3

PVS1 PM5_Supporting PM2_Supporting

Not Met criteria codes 23

Expert Panel

CDH1 VCEP

Criteria Specification Information

PM1 PM3 PM4 PM6 BA1
BS2 BS1 BS4 BS3 BP5 BP7
BP4 BP3 BP1 BP2 PS1 PS2
PS3 PS4 PP1 PP2 PP3 PP4

Evidence Links 0

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.337A>T (p.Lys113Ter) variant is predicted to result in a premature stop codon that leads to nonsense mediate decay (PVS1 and PM5_supporting). The variant is absent in the gnomAD cohort (PM2_supporting; http://gnomad.broadinstitute.org). Therefore, this variant meets criteria to be classified as pathogenic based on the ACMG/AMP criteria applied as specified by the CDH1 Variant Curation Expert Panel (CDH1 VCEP specifications version 3.1): PVS1, PM2_supporting, PM5_supporting.

Met criteria codes

PVS1	✔	Stop gain variant in exon 3 of 16. Expected to undergo NMD.
PM5_Supporting	✔	Stop gain variant in exon 3 of 16. Expected to undergo NMD.
PM2_Supporting	✔	Not present in gnomAD

Not Met criteria codes

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	✖	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

BS1		✖	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
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
BP4		✖	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
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		✖	The GeneDx proband does not fulfill HDGC clinical criteria (SCV000279542.8)
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

Curation History

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