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.1064delT (p.Leu355Terfs)

Version: 2.0

CA336431

224528 (ClinVar)

Gene: CDH1 (HGNC:999)
Condition: CDH1-related diffuse gastric and lobular breast cancer (MONDO:0100488)
Inheritance Mode: Autosomal dominant inheritance
UUID: 9c7b15f1-c81d-4e2e-afc8-adffdaf11274
Approved on: 2023/8/25
Published on: 2023/8/25

HGVS expressions

NM_004360.4(CDH1):c.1064delT (p.Leu355Terfs)
NM_004360.4:c.1064del
NM_004360.4:c.1064delT
NC_000016.10:g.68812190del
CM000678.2:g.68812190del
NC_000016.9:g.68846093del
CM000678.1:g.68846093del
NC_000016.8:g.67403594del
NG_008021.1:g.79899del
ENST00000261769.10:c.1064del
ENST00000261769.9:c.1064del
ENST00000422392.6:c.1064del
ENST00000561751.1:c.686del
ENST00000562836.5:n.1135del
ENST00000565810.1:n.108del
ENST00000566510.5:c.908del
ENST00000566612.5:c.1064del
ENST00000611625.4:c.1064del
ENST00000612417.4:c.1064del
ENST00000621016.4:c.1064del
NM_004360.3:c.1064del
NM_001317184.1:c.1064del
NM_001317185.1:c.-552del
NM_001317186.1:c.-756del
NM_004360.5:c.1064del
NM_001317184.2:c.1064del
NM_001317185.2:c.-552del
NM_001317186.2:c.-756del

Pathogenic

Met criteria codes 4

PVS1 PM5_Supporting PM2_Supporting PS4_Supporting

Not Met criteria codes 22

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 PP1 PP2 PP3 PP4

Evidence Links 1

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.1064delT p.(Leu355Terfs) variant is predicted to result in a premature stop codon that leads to a truncated or absent protein (PVS1, PM5_Supporting). The variant is absent in the gnomAD cohort (PM2_Supporting; http://gnomad.broadinstitute.org). This variant has been reported in at least one family meeting HDGC clinical criteria (PS4_Supporting; PMID: 16061854). In summary, 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 (Variant Interpretation Guidelines Version 3.1): PVS1, PM2_Supporting, PS4_Supporting, PM5_Supporting.

Met criteria codes

PVS1	✔	Exon 8 of 16. Predicted NMD.
PM5_Supporting	✔	Apply PM5_Supporting to nonsense/frameshift variants that are predicted/proved to undergo NMD.
PM2_Supporting	✔	Absent in population databases.
PS4_Supporting	✔	One individual with DGC at 27 years old (variant published as c.1063del). PMID: 16061854
		One individual with DGC at 27 years old (variant published as c.1063del). PubMed:16061854

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	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
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	ℹ	✖	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	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
PP1	ℹ	✖	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

Showing 1 to 2 of 2 rows

The information on this website is not intended for direct diagnostic use or medical decision-making without review by a genetics professional. Individuals should not change their health behavior solely on the basis of information contained on this website. If you have questions about the information contained on this website, please see a health care professional.

ClinGen Terms of Use.
¤ Powered by BCM's Genboree.