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.1569T>A (p.Tyr523Ter)

Version: 3.0

CA396464802

479518 (ClinVar)

Gene: CDH1 (HGNC:999)
Condition: CDH1-related diffuse gastric and lobular breast cancer (MONDO:0100488)
Inheritance Mode: Autosomal dominant inheritance
UUID: 06d1d804-aff1-42eb-af50-e6526e8ea2d9
Approved on: 2023/8/4
Published on: 2023/8/4

HGVS expressions

NM_004360.5(CDH1):c.1569T>A (p.Tyr523Ter)
NM_004360.5:c.1569T>A
NC_000016.10:g.68819283T>A
CM000678.2:g.68819283T>A
NC_000016.9:g.68853186T>A
CM000678.1:g.68853186T>A
NC_000016.8:g.67410687T>A
NG_008021.1:g.86992T>A
ENST00000261769.10:c.1569T>A
ENST00000261769.9:c.1569T>A
ENST00000422392.6:c.1386T>A
ENST00000562836.5:n.1640T>A
ENST00000566510.5:c.*235T>A
ENST00000566612.5:c.1566-2718T>A
ENST00000611625.4:c.1632T>A
ENST00000612417.4:c.1569T>A
ENST00000621016.4:c.1569T>A
NM_004360.3:c.1569T>A
NM_001317184.1:c.1386T>A
NM_001317185.1:c.21T>A
NM_001317186.1:c.-254-2718T>A
NM_004360.4:c.1569T>A
NM_001317184.2:c.1386T>A
NM_001317185.2:c.21T>A
NM_001317186.2:c.-254-2718T>A

Pathogenic

Met criteria codes 3

PVS1 PM2_Supporting PM5_Supporting

Not Met criteria codes 23

PS1 PS2 PS3 PS4 PP1 PP2 PP3 PP4 PM1 PM3 PM4 PM6 BA1 BS1 BS4 BS3

Expert Panel

CDH1 VCEP

Criteria Specification Information

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

BS2 BP5 BP7 BP4 BP3 BP1 BP2

Criteria Specifications for this VCEP

Evidence Links 0

Evidence submitted by expert panel

CDH1 VCEP

The c.1569T>A p.(Tyr523Ter) 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	✔	Exon 11 of 16, predicted NMD.
PM2_Supporting	✔	Absent in population databases.
PM5_Supporting	✔	Exon 11 of 16, predicted NMD.

Not Met criteria codes

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	✖	SCV000661672.2- breast cancer in 40s (ILC) SCV000830946.1- two members in a family have stomach cancer and one at 30s. Both cases do not meet HDGC phenotype criteria.
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
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
PM6	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BA1	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	✖	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		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History

Showing 1 to 3 of 3 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.