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Variant: NM_004360.4(CDH1):c.1460_1461del (p.Val487Alafs)

Version: 3.0

CA891843864

567085 (ClinVar)

Gene: CDH1 (HGNC:999)
Condition: CDH1-related diffuse gastric and lobular breast cancer (MONDO:0100488)
Inheritance Mode: Autosomal dominant inheritance
UUID: 682240bb-090e-4b45-9fa1-c93c2e646a05
Approved on: 2023/8/4
Published on: 2023/8/4

HGVS expressions

NM_004360.4:c.1460_1461del
NM_004360.4(CDH1):c.1460_1461del (p.Val487Alafs)
NC_000016.10:g.68815654_68815655del
CM000678.2:g.68815654_68815655del
NC_000016.9:g.68849557_68849558del
CM000678.1:g.68849557_68849558del
NC_000016.8:g.67407058_67407059del
NG_008021.1:g.83363_83364del
ENST00000261769.10:c.1460_1461del
ENST00000261769.9:c.1460_1461del
ENST00000422392.6:c.1277_1278del
ENST00000562836.5:n.1531_1532del
ENST00000566510.5:c.*126_*127del
ENST00000566612.5:c.1460_1461del
ENST00000611625.4:c.1523_1524del
ENST00000612417.4:c.1460_1461del
ENST00000621016.4:c.1460_1461del
NM_004360.3:c.1460_1461del
NM_001317184.1:c.1277_1278del
NM_001317185.1:c.-89_-88del
NM_001317186.1:c.-360_-359del
NM_004360.5:c.1460_1461del
NM_001317184.2:c.1277_1278del
NM_001317185.2:c.-89_-88del
NM_001317186.2:c.-360_-359del

Pathogenic

Met criteria codes 3

PM2_Supporting PVS1 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.1460_1461del (p.Val487Alafs*3) 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

PM2_Supporting	✔	Absent in gnomAD cohort.
PVS1	✔	Deletion of 2 bp in exon 10 of 16, predicted to cause a frameshift leading to NMD.
PM5_Supporting	✔	Deletion of 2 bp in exon 10 of 16, predicted to cause a frameshift leading to NMD.

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	✖	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
PS4	ℹ	✖	Proband doesn't meet IGCLC HDGC criteria. (SCV000814618.1).
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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