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Variant: NM_004360.4(CDH1):c.1488_1494delCGAGGAC (p.Glu497Leufs)

Version: 2.0

CA10580117

229907 (ClinVar)

Gene: CDH1 (HGNC:999)
Condition: CDH1-related diffuse gastric and lobular breast cancer (MONDO:0100488)
Inheritance Mode: Autosomal dominant inheritance
UUID: f4cc2a94-3541-4121-abe1-a72e59d7a6bf
Approved on: 2023/8/25
Published on: 2023/8/25

HGVS expressions

NM_004360.4(CDH1):c.1488_1494delCGAGGAC (p.Glu497Leufs)
NM_004360.4:c.1488_1494del
NC_000016.10:g.68815682_68815688del
CM000678.2:g.68815682_68815688del
NC_000016.9:g.68849585_68849591del
CM000678.1:g.68849585_68849591del
NC_000016.8:g.67407086_67407092del
NG_008021.1:g.83391_83397del
ENST00000261769.10:c.1488_1494del
ENST00000261769.9:c.1488_1494del
ENST00000422392.6:c.1305_1311del
ENST00000562836.5:n.1559_1565del
ENST00000566510.5:c.*154_*160del
ENST00000566612.5:c.1488_1494del
ENST00000611625.4:c.1551_1557del
ENST00000612417.4:c.1488_1494del
ENST00000621016.4:c.1488_1494del
NM_004360.3:c.1488_1494del
NM_001317184.1:c.1305_1311del
NM_001317185.1:c.-61_-55del
NM_001317186.1:c.-332_-326del
NM_004360.5:c.1488_1494del
NM_001317184.2:c.1305_1311del
NM_001317185.2:c.-61_-55del
NM_001317186.2:c.-332_-326del

Pathogenic

Met criteria codes 4

PS4_Supporting PVS1 PM5_Supporting PM2_Supporting

Not Met criteria codes 22

PM1 PM3 PM4 PM6 BA1 BS2 BS1 BS4 BS3 BP4 BP3

Expert Panel

CDH1 VCEP

Criteria Specification Information

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

BP1 BP2 BP5 BP7 PS1 PS2
PS3 PP1 PP2 PP3 PP4

Criteria Specification Approval History
Criteria Specifications for this VCEP

Evidence Links 1

Evidence submitted by expert panel

CDH1 VCEP

The c.1488_1494delCGAGGAC p.(Glu497Leufs) variant is predicted to result in a premature stop codon that leads to a truncated or absent protein (PVS1, PM5_Supporting). The variant is present in <1/50,000 alleles 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: 10477433). 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

PS4_Supporting	✔	A single case of poorly differentiated, diffuse gastric cancer in an individual age 31 years (PMID: 10477433). Variant reported as 1487del7. A single case of poorly differentiated, diffuse gastric cancer in an individual age 31 years (PMID: 10477433). Variant reported as 1487del7. PubMed:10477433
PVS1	✔	Exon 10 of 16. Predicted NMD.
PM5_Supporting	✔	Apply PM5_Supporting to nonsense/frameshift variants that are predicted/proved to undergo NMD.
PM2_Supporting	✔	gnomAD v2.1.1 - Found in 2 of 113760 alleles (0.001758%) in the non-Finnish European population. <0.002% if present in 2 or more individuals.

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
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
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
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
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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