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Variant: NM_004360.4(CDH1):c.2293C>T (p.Gln765Ter)

Version: 2.0

CA10580156

230451 (ClinVar)

Gene: CDH1 (HGNC:999)
Condition: CDH1-related diffuse gastric and lobular breast cancer (MONDO:0100488)
Inheritance Mode: Autosomal dominant inheritance
UUID: d4d80a05-4db4-4643-877f-10dbb705c844
Approved on: 2023/8/29
Published on: 2023/8/29

HGVS expressions

NM_004360.4(CDH1):c.2293C>T (p.Gln765Ter)
NM_004360.4:c.2293C>T
NC_000016.10:g.68828302C>T
CM000678.2:g.68828302C>T
NC_000016.9:g.68862205C>T
CM000678.1:g.68862205C>T
NC_000016.8:g.67419706C>T
NG_008021.1:g.96011C>T
ENST00000261769.10:c.2293C>T
ENST00000261769.9:c.2293C>T
ENST00000422392.6:c.2110C>T
ENST00000562118.1:n.511C>T
ENST00000562836.5:n.2364C>T
ENST00000566510.5:c.*959C>T
ENST00000566612.5:c.*533C>T
ENST00000611625.4:c.2356C>T
ENST00000612417.4:c.1853+1748C>T
ENST00000621016.4:c.1866-5901C>T
NM_004360.3:c.2293C>T
NM_001317184.1:c.2110C>T
NM_001317185.1:c.745C>T
NM_001317186.1:c.328C>T
NM_004360.5:c.2293C>T
NM_001317184.2:c.2110C>T
NM_001317185.2:c.745C>T
NM_001317186.2:c.328C>T

Pathogenic

Met criteria codes 4

PVS1 PM2_Supporting PS4_Supporting PM5_Supporting

Not Met criteria codes 22

PS1 PS2 PS3 PP1 PP2 PP3 PP4 PM1 PM3 PM4 PM6

Expert Panel

CDH1 VCEP

Criteria Specification Information

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

BA1 BS1 BS4 BS3 BS2 BP5
BP7 BP4 BP3 BP1 BP2

Criteria Specification Approval History
Criteria Specifications for this VCEP

Evidence Links 1

Evidence submitted by expert panel

CDH1 VCEP

The c.2293C>T (p.Gln765*) variant is predicted to result in a premature stop codon that leads to a truncated or absent protein (PVS1, PM5_Supporting). This variant is absent in the gnomAD cohort (PM2_Supporting; http://gnomad.broadinstitute.org). The variant has been reported in at least one family meeting HDGC clinical criteria (PS4_Supporting; SCV000839094.1). 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	✔	Stop gain introduced at position 765 in exon 14 of 16. Predicted to undergo NMD.
PM2_Supporting	✔	Absent in gnomAD
PS4_Supporting	✔	One family that meets HDGC criteria (SCV000839094.1). Proband with unknown whether family fulfills HDGC criteria (SCV000274004.4). Somatic variant in a breast tumour from a white female **PubMed:29156750**
PM5_Supporting	✔	Apply PM5_Supporting to nonsense/frameshift variants that are predicted/proved to undergo 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
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	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History

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