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Variant: NM_004360.5(CDH1):c.2T>C (p.Met1Thr)

Version: 2.0

CA396451185

486826 (ClinVar)

Gene: CDH1 (HGNC:999)
Condition: CDH1-related diffuse gastric and lobular breast cancer (MONDO:0100488)
Inheritance Mode: Autosomal dominant inheritance
UUID: 3f6e0f9d-ae4a-4d07-a4a1-4f43338d71eb
Approved on: 2023/8/25
Published on: 2023/8/25

HGVS expressions

NM_004360.5(CDH1):c.2T>C (p.Met1Thr)
NM_004360.5:c.2T>C
NC_000016.10:g.68737417T>C
CM000678.2:g.68737417T>C
NC_000016.9:g.68771320T>C
CM000678.1:g.68771320T>C
NC_000016.8:g.67328821T>C
NG_008021.1:g.5126T>C
ENST00000261769.10:c.2T>C
ENST00000261769.9:c.2T>C
ENST00000422392.6:c.2T>C
ENST00000566510.5:c.2T>C
ENST00000566612.5:c.2T>C
ENST00000611625.4:c.2T>C
ENST00000612417.4:c.2T>C
ENST00000621016.4:c.2T>C
NM_004360.3:c.2T>C
NM_001317184.1:c.2T>C
NM_001317185.1:c.-1614T>C
NM_001317186.1:c.-1818T>C
NM_004360.4:c.2T>C
NM_001317184.2:c.2T>C
NM_001317185.2:c.-1614T>C
NM_001317186.2:c.-1818T>C

Pathogenic

Met criteria codes 3

PVS1 PM2_Supporting PS4_Moderate

Not Met criteria codes 23

BS1 BS4 BS3 BS2 BP5 BP7 BP4 BP3 BP1 BP2 PS1 PS2 PS3 PM1 PM3 PM5 PM4 BA1 PM6 PP1 PP2 PP3 PP4

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
Criteria Specifications for this VCEP

Evidence Links 0

Evidence submitted by expert panel

CDH1 VCEP

The c.2T>C (p.Met1Thr) variant alters the start codon of the CDH1 coding sequence and is predicted to lead to an absent protein (PVS1). This variant is absent in the gnomAD cohort (PM2_supporting; http://gnomad.broadinstitute.org). This variant has been reported in at least 2 probands/families meeting HDGC clinical criteria (PS4_moderate; PMID: 20373070, SCV000760804.3). In summary, this variant meets criteria to be classified as pathogenic based on the ACMG/AMP Variant Interpretation Guidelines Version 3.1 as specified by the CDH1 Variant Curation Expert Panel: PVS1, PS4_moderate, PM2_supporting.

Met criteria codes

PVS1	✔	PVS1 applies to initiation codon variants
PM2_Supporting	✔	Absent from all population databases
PS4_Moderate	✔	Two probands/families meet HDGC phenotype criteria.

Not Met criteria codes

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
BS2	✖	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
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	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
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
PM5		✖	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
BA1	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
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	ℹ	✖	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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