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Variant: NM_004360.4(CDH1):c.2324delG (p.Gly775Alafs)

Version: 3.0

CA16615415

406669 (ClinVar)

Gene: CDH1 (HGNC:999)
Condition: CDH1-related diffuse gastric and lobular breast cancer (MONDO:0100488)
Inheritance Mode: Autosomal dominant inheritance
UUID: 07bc6bc1-21bc-476e-bab1-e1fc34001d34
Approved on: 2023/8/4
Published on: 2023/8/4

HGVS expressions

NM_004360.4(CDH1):c.2324delG (p.Gly775Alafs)
NM_004360.4:c.2324del
NC_000016.10:g.68829682del
CM000678.2:g.68829682del
NC_000016.9:g.68863585del
CM000678.1:g.68863585del
NC_000016.8:g.67421086del
NG_008021.1:g.97391del
ENST00000261769.10:c.2324del
ENST00000261769.9:c.2324del
ENST00000422392.6:c.2141del
ENST00000562118.1:n.542del
ENST00000562836.5:n.2395del
ENST00000566510.5:c.*990del
ENST00000566612.5:c.*564del
ENST00000611625.4:c.2387del
ENST00000612417.4:c.1853+3128del
ENST00000621016.4:c.1866-4521del
NM_004360.3:c.2324del
NM_001317184.1:c.2141del
NM_001317185.1:c.776del
NM_001317186.1:c.359del
NM_004360.5:c.2324del
NM_001317184.2:c.2141del
NM_001317185.2:c.776del
NM_001317186.2:c.359del

Pathogenic

Met criteria codes 3

PVS1 PM5_Supporting PM2_Supporting

Not Met criteria codes 23

PM1 PM3 PM4 PM6 BA1 BS2 BS1 BS4 BS3 BP5 BP7

Expert Panel

CDH1 VCEP

Criteria Specification Information

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

BP4 BP3 BP1 BP2 PS1 PS2
PS3 PS4 PP1 PP2 PP3 PP4

Criteria Specification Approval History
Criteria Specifications for this VCEP

Evidence Links 0

Evidence submitted by expert panel

CDH1 VCEP

The c.2324delG (p.Gly775Alafs) 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	✔	A 1 bp deletion in exon 15/16, predicted to result in a premature termination codon that leads to NMD
PM5_Supporting	✔	A 1 bp deletion in exon 15/16, predicted to result in a premature termination codon that leads to NMD
PM2_Supporting	✔	Absent in gnomAD

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	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
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
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 does not fulfil HDGC clinical criteria, because pathology is not available (SCV000545472.2)
PP1	ℹ	✖	Two meioses across one family (SCV000545472.2).
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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