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Variant: NM_004360.4(CDH1):c.1612delG (p.Asp538Thrfs)

Version: 2.0

CA645509536

437928 (ClinVar)

Gene: CDH1 (HGNC:999)
Condition: CDH1-related diffuse gastric and lobular breast cancer (MONDO:0100488)
Inheritance Mode: Autosomal dominant inheritance
UUID: 47f620ff-80ee-453c-9f55-58567c1f14be
Approved on: 2023/8/25
Published on: 2023/8/25

HGVS expressions

NM_004360.4:c.1612delG
NM_004360.4(CDH1):c.1612delG (p.Asp538Thrfs)
NC_000016.10:g.68819326del
CM000678.2:g.68819326del
NC_000016.9:g.68853229del
CM000678.1:g.68853229del
NC_000016.8:g.67410730del
NG_008021.1:g.87035del
ENST00000261769.10:c.1612del
ENST00000261769.9:c.1612del
ENST00000422392.6:c.1429del
ENST00000562836.5:n.1683del
ENST00000566510.5:c.*278del
ENST00000566612.5:c.1566-2675del
ENST00000611625.4:c.1675del
ENST00000612417.4:c.1612del
ENST00000621016.4:c.1612del
NM_004360.3:c.1612del
NM_001317184.1:c.1429del
NM_001317185.1:c.64del
NM_001317186.1:c.-254-2675del
NM_004360.4:c.1612del
NM_004360.5:c.1612del
NM_001317184.2:c.1429del
NM_001317185.2:c.64del
NM_001317186.2:c.-254-2675del

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 2

Evidence submitted by expert panel

CDH1 VCEP

The c.1612delG p.(Asp538Thrfs) variant is predicted to result in a premature stop codon that leads to a truncated or absent protein (PVS1, PM5_Supporting). The variant is absent 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: 29131691). 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	✔	Exon 11 of 16. Predicted NMD.
PM2_Supporting	✔	Absent in population databases.
PS4_Supporting	✔	PMID: 29131691 - 41-year-old with metastatic GC characterized by the presence of signet ring cells. Same patient as in PMID: 29131691 PubMed:29295527 41-year-old with metastatic GC characterized by the presence of signet ring cells PubMed:29131691
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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