

Variant: *NM_004360.5(CDH1):c.793G>T (p.Glu265Ter)*

Version: 3.0

CA16615233 [↗](#)

406633 (ClinVar) [↗](#)

Gene: CDH1 ([HGNC:999](#))

Condition: CDH1-related diffuse gastric and lobular breast cancer ([MONDO:0100488](#))

Inheritance Mode: Autosomal dominant inheritance

UUID: fa795b67-a5d9-42c3-ac78-94f3c3c2bd94

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HGVS expressions

NM_004360.5(CDH1):c.793G>T (p.Glu265Ter)

NM_004360.5:c.793G>T

NC_000016.10:g.68810302G>T

CM000678.2:g.68810302G>T

NC_000016.9:g.68844205G>T

CM000678.1:g.68844205G>T

NC_000016.8:g.67401706G>T

NG_008021.1:g.78011G>T

ENST00000261769.10:c.793G>T

ENST00000261769.9:c.793G>T

ENST00000422392.6:c.793G>T

ENST00000561751.1:c.455-1382G>T

ENST00000562836.5:n.864G>T

ENST00000566510.5:c.637G>T

ENST00000566612.5:c.793G>T

ENST00000611625.4:c.793G>T

ENST00000612417.4:c.793G>T

ENST00000621016.4:c.793G>T

NM_004360.3:c.793G>T

NM_001317184.1:c.793G>T

NM_001317185.1:c.-823G>T

NM_001317186.1:c.-1027G>T

NM_004360.4:c.793G>T

NM_001317184.2:c.793G>T

NM_001317185.2:c.-823G>T

NM_001317186.2:c.-1027G>T

Pathogenic

Met criteria codes 3

PM2_Supporting

PVS1

PM5_Supporting

Not Met criteria codes 23

BS2

BS1

BS4

BS3

BP4

BP3

BP1

BP2

BP5

BP7

PS1

PS2

Expert Panel

CDH1 VCEP [↗](#)

Criteria Specification Information

[↗](#) **Criteria Specification:** ClinGen CDH1 Expert Panel
Specifications to the ACMG/AMP Variant Interpretation
Guidelines Version 3.1

Evidence Links0

Evidence submitted by expert panel

CDH1 VCEP

The c.793G>T (p.Glu265Ter) 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

PM2_Supporting	<div><div>i</div></div>	<div>✓</div>	Absent in gnomAD v2.1.1
PVS1	<div><div>i</div></div>	<div>✓</div>	PTC in exon 6 (of 16), expected to lead to NMD
PM5_Supporting	<div><div>i</div></div>	<div>✓</div>	PTC in exon 6 (of 16), expected to lead to NMD

Not Met criteria codes

BS2	<div><div>i</div></div>	<div>✗</div>	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS1	<div><div>i</div></div>	<div>✗</div>	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS4	<div><div>i</div></div>	<div>✗</div>	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS3	<div><div>i</div></div>	<div>✗</div>	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP4	<div><div>i</div></div>	<div>✗</div>	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP3		<div>✗</div>	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP1		<div>✗</div>	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
PS4			One family does not meet HDGC clinical criteria (SCV000545402.2)
PP1			One unaffected carrier (SCV000545402.2)
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
BA1			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			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

Curation History [↗](#)

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