

Variant: *NM_004360.5(CDH1):c.1613A>T (p.Asp538Val)*

Version: 2.0

CA338255 [↗](#)

216589 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 7a7009f6-789d-458a-a313-c7508dc251da

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HGVS expressions

NM_004360.5(CDH1):c.1613A>T (p.Asp538Val)

NM_004360.5:c.1613A>T

NC_000016.10:g.68819327A>T

CM000678.2:g.68819327A>T

NC_000016.9:g.68853230A>T

CM000678.1:g.68853230A>T

NC_000016.8:g.67410731A>T

NG_008021.1:g.87036A>T

ENST00000261769.10:c.1613A>T

ENST00000261769.9:c.1613A>T

ENST00000422392.6:c.1430A>T

ENST00000562836.5:n.1684A>T

ENST00000566510.5:c.*279A>T

ENST00000566612.5:c.1566-2674A>T

ENST00000611625.4:c.1676A>T

ENST00000612417.4:c.1613A>T

ENST00000621016.4:c.1613A>T

NM_004360.3:c.1613A>T

NM_001317184.1:c.1430A>T

NM_001317185.1:c.65A>T

NM_001317186.1:c.-254-2674A>T

NM_004360.4:c.1613A>T

NM_001317184.2:c.1430A>T

NM_001317185.2:c.65A>T

NM_001317186.2:c.-254-2674A>T

Uncertain Significance

Met criteria codes **2**

PM2_Supporting

PS4_Moderate

Not Met criteria codes **24**

BS1

BS4

BS3

BS2

PVS1

BP5

BP7

BP4

BP3

BP1

BP2

BA1

PS1

PS2

PS3

PP1

PP2

PP3

PP4

PM6

PM1

PM3

PM5

PM4

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**

Evidence submitted by expert panel

CDH1 VCEP

The c.1613A>T (p.Asp538Val) variant is absent in the gnomAD cohort (PM2_Supporting). This variant has been reported in at least two families meeting HDGC clinical criteria (PS4_Moderate; SCV000254814.3). In summary, this variant meets criteria to be classified as a variant of uncertain significance based on the ACMG/AMP criteria applied as specified by the CDH1 Variant Curation Expert Panel (Variant Interpretation Guidelines Version 3.1): PM2_Supporting, PS4_Moderate.

Met criteria codes

PM2_Supporting			Absent in all population databases
PS4_Moderate			This variant has been reported in at least two families meeting HDGC clinical criteria (SCV000254814.3). Internal lab contributor: Gastric cancer dx @ 40s, pathology not specified. No reported FHx of cancer (Not Meet). SCV000254814.3: Proband #1: male with diffuse gastric cancer by 50s. No family history. (Not Meet) Proband #2: male with metastatic gastric cancer at 30s. FHx of gastric cancer. (Meet) Proband #3: female with diffuse gastric cancer at late 30s (Meet).

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
PVS1			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			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
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			2 meioses
PP2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP3			In silico splicing predictors (SSF, MaxEnt, NNSPLICE, GeneSplicer) predict no splicing effect.
PP4			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
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



No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History

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