

## Variant: NM\_004360.4(CDH1):c.2100del (p.Val701Serfs)

Version: 2.0

CA658683961 [↗](#)

492677 (ClinVar) [↗](#)

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

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

**Inheritance Mode:** Autosomal dominant inheritance

**UUID:** 71001505-c352-40be-b876-0a9c4bd2ef36

**Approved on:** 2023-08-29

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### HGVS expressions

#### NM\_004360.4:c.2100del

NM\_004360.4(CDH1):c.2100del (p.Val701Serfs)

NC\_000016.10:g.68823562del

CM000678.2:g.68823562del

NC\_000016.9:g.68857465del

CM000678.1:g.68857465del

NC\_000016.8:g.67414966del

NG\_008021.1:g.91271del

ENST00000261769.10:c.2100del

ENST00000261769.9:c.2100del

ENST00000422392.6:c.1917del

ENST00000562118.1:n.318del

ENST00000562836.5:n.2171del

ENST00000566510.5:c.\*766del

ENST00000566612.5:c.\*340del

ENST00000611625.4:c.2163del

ENST00000612417.4:c.1830+1443del

ENST00000621016.4:c.1865+1408del

NM\_004360.3:c.2100del

NM\_001317184.1:c.1917del

NM\_001317185.1:c.552del

NM\_001317186.1:c.135del

NM\_004360.5:c.2100del

NM\_001317184.2:c.1917del

NM\_001317185.2:c.552del

NM\_001317186.2:c.135del

**Pathogenic**

#### Met criteria codes **4**

PS4\_Supporting PM2\_Supporting

PVS1 PM5\_Supporting

#### Not Met criteria codes **22**

BS2 BS1 BS4 BS3 BP4 BP3

BP1 BP2 BP5 BP7 PS1 PS2

#### Expert Panel

#### Criteria Specification Information

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

[↗](#) **Criteria Specification Approval History**

[PS3](#) [BA1](#) [PP1](#) [PP2](#) [PP3](#) [PP4](#)  
[PM1](#) [PM3](#) [PM4](#) [PM6](#)

[Criteria Specifications for this VCEP](#)










Evidence Links **1**

Evidence submitted by expert panel














## CDH1 VCEP

























The c.2100del p.(Val701Serfs) variant is predicted to result in a premature stop codon that leads to a truncated or absent protein (PVS1, PM5\_Supporting). The variant has been reported in at least one family meeting HDGC clinical criteria (PS4\_Supporting; PMID: 26182300). This variant is absent in the gnomAD cohort (PM2\_Supporting; <http://gnomad.broadinstitute.org>). 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

<b>PS4_Supporting</b>			Identified in one family with diffuse gastric cancer (proband diagnosed at 20 yo, at least 2 DGC cases in the family). Identified in one family with diffuse gastric cancer (proband diagnosed 20 yo, at least 2 DGC cases in the family). <a href="#">PubMed:26182300</a> 
<b>PM2_Supporting</b>			Not observed in population databases.
<b>PVS1</b>			Upstream of codon 795, which is the NMD boundary.
<b>PM5_Supporting</b>			Apply PM5_Supporting to nonsense/frameshift variants that are predicted/proved to undergo NMD.

### Not Met criteria codes

<b>BS2</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>BS1</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>BS4</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>BS3</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>BP4</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>BP3</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>BP1</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>BP2</b>			

			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>BP5</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>BP7</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>PS1</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>PS2</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>PS3</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>BA1</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>PP1</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>PP2</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>PP3</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>PP4</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>PM1</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>PM3</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>PM4</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>PM6</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History [↗](#)

Showing 1 to 2 of 2 rows

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