

Variant: *NM_004360.4(CDH1):c.208dup (p.Ser70Phefs)*

Version: 2.1

CA645596587 [↗](#)

463742 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 529b4404-fa68-47ea-9b98-9114bb2d72ba

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HGVS expressions

NM_004360.4(CDH1):c.208dup (p.Ser70Phefs)

NM_004360.4:c.208dup

NC_000016.10:g.68801714dup

CM000678.2:g.68801714dup

NC_000016.9:g.68835617dup

CM000678.1:g.68835617dup

NC_000016.8:g.67393118dup

NG_008021.1:g.69423dup

ENST00000261769.10:c.208dup

ENST00000261769.9:c.208dup

ENST00000422392.6:c.208dup

ENST00000562836.5:n.279dup

ENST00000564676.5:n.490dup

ENST00000564745.1:n.203dup

ENST00000566510.5:c.208dup

ENST00000566612.5:c.208dup

ENST00000611625.4:c.208dup

ENST00000612417.4:c.208dup

ENST00000621016.4:c.208dup

NM_004360.3:c.208dup

NM_001317184.1:c.208dup

NM_001317185.1:c.-1408dup

NM_001317186.1:c.-1612dup

NM_004360.5:c.208dup

NM_001317184.2:c.208dup

NM_001317185.2:c.-1408dup

NM_001317186.2:c.-1612dup

Pathogenic

Met criteria codes 4

PM2_Supporting

PS4_Moderate

PM5_Supporting

PVS1

Not Met criteria codes 22

BA1

BP4

BP3

BP1

BP2

BP5

BP7

BS2

BS1

BS4

BS3

PP1

Expert Panel

CDH1 VCEP [↗](#)

Criteria Specification Information

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

Evidence submitted by expert panel

CDH1 VCEP

The c.208dup p.(Ser70Phefs) 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 two family meeting HDGC clinical criteria (PS4_Moderate; SCV000637771.2, SCV000779081.2). 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_Moderate, PM5_Supporting.

Met criteria codes

PM2_Supporting			Absent in population database.
PS4_Moderate			SCV000637771.2, SCV000779081.2 - meet IGCLC HDGC criteria.
PM5_Supporting			Apply PM5_Supporting to nonsense/frameshift variants that are predicted/proved to undergo NMD.
PVS1			Predicted NMD.

Not Met criteria codes

BA1			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
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
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			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
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
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
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

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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