

Variant: *NM_004360.5(CDH1):c.1779dup (p.Ile594fs)*

Version: 2.0

CA10577548 [↗](#)

234610 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 3e24a117-e2ed-4f5e-9992-bd7eac3b6454

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HGVS expressions

NM_004360.5(CDH1):c.1779dup (p.Ile594fs)

NM_004360.5:c.1779dup

NC_000016.10:g.68822068dup

CM000678.2:g.68822068dup

NC_000016.9:g.68855971dup

CM000678.1:g.68855971dup

NC_000016.8:g.67413472dup

NG_008021.1:g.89777dup

ENST00000261769.10:c.1779dup

ENST00000261769.9:c.1779dup

ENST00000422392.6:c.1596dup

ENST00000562836.5:n.1850dup

ENST00000566510.5:c.*445dup

ENST00000566612.5:c.*19dup

ENST00000611625.4:c.1842dup

ENST00000612417.4:c.1779dup

ENST00000621016.4:c.1779dup

NM_004360.3:c.1779dup

NM_001317184.1:c.1596dup

NM_001317185.1:c.231dup

NM_001317186.1:c.-187dup

NM_004360.4:c.1779dup

NM_001317184.2:c.1596dup

NM_001317185.2:c.231dup

NM_001317186.2:c.-187dup

Pathogenic

Met criteria codes 4

PM2_Supporting

PVS1

PM5_Supporting

PS4_Supporting

Not Met criteria codes 22

PS1

PS2

PS3

BP5

BP7

BP4

BP3

BP1

BP2

PP1

PP2

PP3

PP4

PM6

PM1

PM3

PM4

BA1

BS1

BS4

BS3

BS2

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.1779dup p.(Ile594fs) 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>). Additionally, this variant has been reported in at least one family meeting HDGC clinical criteria (PS4_Supporting; PMID: 15235021). 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

PM2_Supporting			Absent in population databases.
PVS1			Exon 12 of 16, predicted NMD.
PM5_Supporting			Apply PM5_Supporting to nonsense/frameshift variants that are predicted/proved to undergo NMD.
PS4_Supporting			DGC at 33 years old (variant reported as 1779insC) (PMID: 15235021).

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
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
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
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
BA1		✖	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

BS2

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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