

Variant: *NM_004360.4(CDH1):c.124_126delCCCinsT*
(*p.Pro42Serfs*)

Version: 2.0

CA10580072 [↗](#)

230956 (ClinVar) [↗](#)

Gene: [CDH1](#)

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 88a545eb-341c-416c-944c-9454a71b04e5

Approved on: 2023/8/29

Published on: 2023/8/29

HGVS expressions

NM_004360.4(CDH1):c.124_126delCCCinsT (p.Pro42Serfs)

NM_004360.4:c.124_126delCCCinsT

NC_000016.10:g.68738372_68738374delinsT

CM000678.2:g.68738372_68738374delinsT

NC_000016.9:g.68772275_68772277delinsT

CM000678.1:g.68772275_68772277delinsT

NC_000016.8:g.67329776_67329778delinsT

NG_008021.1:g.6081_6083delinsT

ENST00000261769.10:c.124_126delinsT

ENST00000261769.9:c.124_126delinsT

ENST00000422392.6:c.124_126delinsT

ENST00000566510.5:c.124_126delinsT

ENST00000566612.5:c.124_126delinsT

ENST00000611625.4:c.124_126delinsT

ENST00000612417.4:c.124_126delinsT

ENST00000621016.4:c.124_126delinsT

NM_004360.3:c.124_126delinsT

NM_001317184.1:c.124_126delinsT

NM_001317185.1:c.-1492_-1490delinsT

NM_001317186.1:c.-1696_-1694delinsT

NM_004360.4:c.124_126delinsT

NM_004360.5:c.124_126delinsT

NM_001317184.2:c.124_126delinsT

NM_001317185.2:c.-1492_-1490delinsT

NM_001317186.2:c.-1696_-1694delinsT

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

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 submitted by expert panel

CDH1 VCEP

The c.124_126delCCCinsT (p.Pro42Serfs*16) 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 one family meeting HDGC clinical criteria (PS4_Supporting; PMID 30745422). 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			Allele is absent from populations in gnomAD, ExAC, 1000 Genomes and ESP.
PVS1			This variant is predicted to result in a premature stop codon in exon 2 that leads to a truncated or absent protein.
PM5_Supporting			Apply PM5_Supporting to nonsense/frameshift variants that are predicted/proved to undergo NMD.
PS4_Supporting			This variant was identified in one family meeting IGCLC criteria for HDGC (PMID: 30745422). This variant was identified in a family meeting IGCLC criteria for HDGC. PubMed:30745422

Not Met criteria codes

PS1			Not applicable.
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			Not applicable.
BP4			Not applicable.

BP3		✖	Not applicable.
BP1		✖	Not applicable.
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		✖	Not applicable.
PP3		✖	Not applicable.
PP4		✖	Not applicable.
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		✖	Not applicable.
PM4		✖	Not applicable.
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











Showing 1 to 2 of 2 rows

--

The information on this website is not intended for direct diagnostic use or medical decision-making without review by a genetics professional. Individuals should not change their health behavior solely on the basis of information contained on this website. If you have questions about the information contained on this website, please see a health care professional.