The ClinGen Evidence Repository is an **FDA-recognized human genetic variant database** containing expert-curated assertions regarding variants' pathogenicity and supporting evidence summaries. [Disclaimer]

Variant: NM_004360.5(CDH1):c.480_486delinsAGAATA (p.Ile161fs)

Version: 2.0

CA658658479

479524 (ClinVar)

Gene: CDH1 (HGNC:999)
Condition: CDH1-related diffuse gastric and lobular breast cancer (MONDO:0100488)
Inheritance Mode: Autosomal dominant inheritance
UUID: 9b7f21ff-48e0-4483-9bac-0393271e0fbc
Approved on: 2023/8/25
Published on: 2023/8/25

HGVS expressions

NM_004360.5:c.480_486delinsAGAATA
NM_004360.5(CDH1):c.480_486delinsAGAATA (p.Ile161fs)
NC_000016.10:g.68808516_68808522delinsAGAATA
CM000678.2:g.68808516_68808522delinsAGAATA
NC_000016.9:g.68842419_68842425delinsAGAATA
CM000678.1:g.68842419_68842425delinsAGAATA
NC_000016.8:g.67399920_67399926delinsAGAATA
NG_008021.1:g.76225_76231delinsAGAATA
ENST00000261769.10:c.480_486delinsAGAATA
ENST00000261769.9:c.480_486delinsAGAATA
ENST00000422392.6:c.480_486delinsAGAATA
ENST00000561751.1:c.247_253delinsAGAATA
ENST00000562836.5:n.551_557delinsAGAATA
ENST00000564676.5:n.762_768delinsAGAATA
ENST00000564745.1:n.475_481delinsAGAATA
ENST00000566510.5:c.480_486delinsAGAATA
ENST00000566612.5:c.480_486delinsAGAATA
ENST00000611625.4:c.480_486delinsAGAATA
ENST00000612417.4:c.480_486delinsAGAATA
ENST00000621016.4:c.480_486delinsAGAATA
NM_004360.3:c.480_486delinsAGAATA
NM_001317184.1:c.480_486delinsAGAATA
NM_001317185.1:c.-1136_-1130delinsAGAATA
NM_001317186.1:c.-1340_-1334delinsAGAATA
NM_004360.4:c.480_486delinsAGAATA
NM_001317184.2:c.480_486delinsAGAATA
NM_001317185.2:c.-1136_-1130delinsAGAATA
NM_001317186.2:c.-1340_-1334delinsAGAATA

Pathogenic

Met criteria codes 4

PVS1 PM2_Supporting PM5_Supporting PS4_Supporting

Not Met criteria codes 22

Expert Panel

CDH1 VCEP

Criteria Specification Information

BS1 BS4 BS3 BS2 BP5 BP7
BP4 BP3 BP1 BP2 PS1 PS2
PS3 PM1 PM3 PM4 BA1
PM6 PP1 PP2 PP3 PP4

Evidence Links 0

Criteria Specification: *ClinGen CDH1 Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines Version 3.1*
Criteria Specification Approval History
Criteria Specifications for this VCEP

Evidence submitted by expert panel

CDH1 VCEP

The c.480_486delinsAGAATA p.(Ile161Glufs*54) 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; SCV000661678.2). 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_Supporting, PM5_Supporting.

Met criteria codes

PVS1	✔	Frameshift in exon 4 and PTC created 54 amino acids downstream (Ile161Glufs*54) in exon 5 of 16, expected to undergo NMD.
PM2_Supporting	✔	Absent in gnomAD v2.1.1, in a region of sufficient coverage
PM5_Supporting	✔	Apply PM5_Supporting to nonsense/frameshift variants that are predicted/proved to undergo NMD.
PS4_Supporting	✔	One family that fulfils HDGC clinical criteria: obligate carrier with signet cell type gastric cancer diagnosed in 30s (Ambry - SCV000661678.2)

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
BP5	✖	

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

BP7	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP4	ℹ	✖	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	ℹ	✖	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	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS3	ℹ	✖	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	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BA1	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM6	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP1	ℹ	✖	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

Curation History

Showing 1 to 2 of 2 rows

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