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_004360.4(CDH1):c.2512A>G (p.Ser838Gly)

Version: 3.0

CA121994

12233 (ClinVar)

Gene: CDH1 (HGNC:999)
Condition: CDH1-related diffuse gastric and lobular breast cancer (MONDO:0100488)
Inheritance Mode: Autosomal dominant inheritance
UUID: 85364a5d-816a-4c65-a3f3-29c77698b3b9
Approved on: 2023-08-17
Published on: 2023-08-17

HGVS expressions

NM_004360.4:c.2512A>G
NM_004360.4(CDH1):c.2512A>G (p.Ser838Gly)
NC_000016.10:g.68833362A>G
CM000678.2:g.68833362A>G
NC_000016.9:g.68867265A>G
CM000678.1:g.68867265A>G
NC_000016.8:g.67424766A>G
NG_008021.1:g.101071A>G
ENST00000261769.10:c.2512A>G
ENST00000261769.9:c.2512A>G
ENST00000422392.6:c.2329A>G
ENST00000562118.1:n.730A>G
ENST00000562836.5:n.2583A>G
ENST00000566510.5:c.*1178A>G
ENST00000566612.5:c.*752A>G
ENST00000611625.4:c.2575A>G
ENST00000612417.4:c.1854-829A>G
ENST00000621016.4:c.1866-841A>G
NM_004360.3:c.2512A>G
NM_001317184.1:c.2329A>G
NM_001317185.1:c.964A>G
NM_001317186.1:c.547A>G
NM_004360.5:c.2512A>G
NM_001317184.2:c.2329A>G
NM_001317185.2:c.964A>G
NM_001317186.2:c.547A>G

Likely Benign

Met criteria codes 1

BS2

Not Met criteria codes 25

BA1 PP1 PP2 PP3 PP4 PM1 PM3 PM5 PM4 PM6 PM2 PVS1 BS1 BS4 BS3

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

BP5 BP7 BP4 BP3 BP1 BP2
PS1 PS2 PS3 PS4

Criteria Specifications for this VCEP

Evidence Links 0

Evidence submitted by expert panel

CDH1 VCEP

The c.2512A>G (p.Ser838Gly) variant has been observed in >10 individuals without a diagnosis of diffuse gastric cancer, signet ring tumor or lobular breast cancer and whose family histories do not suggest HDGC (BS2; internal laboratory contributors). In summary, the clinical significance of this variant is classified as likely benign based on BS2 alone. ACMG/AMP criteria applied, as specified by the CDH1 Variant Curation Expert Panel (Variant Interpretation Guidelines Version 3.1): BS2.

Met criteria codes

BS2	✔	Observed in 188 individuals w/o a dx of HDGC.

Not Met criteria codes

BA1	✖	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
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
PM5	✖	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
PM6		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM2		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PVS1		✖	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
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
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
PS4		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History

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