

Variant: NM_000535.7(PMS2):c.241G>T (p.Glu81Ter)

Version: 2.0

CA366744765 [↗](#)

439243 (ClinVar) [↗](#)

Gene: PMS2 ([HGNC:5395](#))

Condition: colorectal cancer, hereditary nonpolyposis, type 4 ([MONDO:0013699](#))

Inheritance Mode: Autosomal dominant inheritance

UUID: 7ac25e37-23ce-459b-a108-d230290a2161

Approved on: 2024/9/18

Published on: 2025/8/7

HGVS expressions

NM_000535.7(PMS2):c.241G>T (p.Glu81Ter)

- NM_000535.7:c.241G>T
- NC_000007.14:g.6003981C>A
- CM000669.2:g.6003981C>A
- NC_000007.13:g.6043612C>A
- CM000669.1:g.6043612C>A
- NC_000007.12:g.6010138C>A
- NG_008466.1:g.10126G>T
- ENST00000699814.2:c.241G>T
- ENST00000699840.2:c.241G>T
- ENST00000699930.2:c.241G>T
- ENST00000406569.8:c.241G>T
- ENST00000415839.3:n.306G>T
- ENST00000644110.2:c.241G>T
- ENST00000699752.1:c.241G>T
- ENST00000699753.1:c.-165G>T
- ENST00000699754.1:c.241G>T
- ENST00000699755.1:c.-165G>T
- ENST00000699756.1:c.241G>T
- ENST00000699757.1:c.26G>T
- ENST00000699758.1:c.241G>T
- ENST00000699759.1:n.313G>T
- ENST00000699760.1:c.26G>T
- ENST00000699761.1:c.-112G>T
- ENST00000699762.1:c.-165G>T
- ENST00000699763.1:c.-165G>T
- ENST00000699764.1:c.241G>T
- ENST00000699765.1:c.26G>T
- ENST00000699766.1:c.241G>T
- ENST00000699767.1:c.241G>T
- ENST00000699768.1:c.241G>T
- ENST00000699769.1:n.321G>T
- ENST00000699770.1:n.297G>T
- ENST00000699811.1:c.-340G>T
- ENST00000699814.1:c.165G>T
- ENST00000699815.1:c.-165G>T
- ENST00000699816.1:c.-165G>T

ENST00000699817.1:c.164-1345G>T
ENST00000699818.1:c.-165G>T
ENST00000699819.1:c.-178G>T
ENST00000699820.1:c.241G>T
ENST00000699821.1:c.-165G>T
ENST00000699822.1:c.-165G>T
ENST00000699823.1:c.-112G>T
ENST00000699824.1:c.241G>T
ENST00000699825.1:c.-165G>T
ENST00000699826.1:c.-165G>T
ENST00000699827.1:c.241G>T
ENST00000699828.1:c.241G>T
ENST00000699829.1:c.-112G>T
ENST00000699830.1:c.241G>T
ENST00000699831.1:n.202G>T
ENST00000699833.1:n.321G>T
ENST00000699834.1:n.447G>T
ENST00000699837.1:c.-62G>T
ENST00000699838.1:c.*141G>T
ENST00000699839.1:c.427G>T
ENST00000699840.1:c.241G>T
ENST00000699916.1:c.-165G>T
ENST00000699917.1:c.241G>T
ENST00000699918.1:c.241G>T
ENST00000699919.1:c.241G>T
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ENST00000699933.1:n.321G>T
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ENST00000699952.1:c.241G>T
ENST00000699953.1:c.241G>T
ENST00000699954.1:c.241G>T
ENST00000703409.1:n.440G>T
ENST00000265849.12:c.241G>T
ENST00000642292.1:c.-165G>T
ENST00000642456.1:c.-165G>T
ENST00000643595.1:c.241G>T
ENST00000644110.1:c.26G>T
ENST00000265849.11:c.241G>T
ENST00000380416.5:n.320G>T
ENST00000382321.5:c.241G>T
ENST00000406569.7:n.241G>T
ENST00000415839.2:n.253G>T
ENST00000441476.6:c.26G>T
ENST00000469652.1:n.62+2012G>T
NM_000535.5:c.241G>T
NR_003085.2:n.323G>T
NM_000535.6:c.241G>T

NM_001322003.1:c.-165G>T
NM_001322004.1:c.-165G>T
NM_001322005.1:c.-165G>T
NM_001322006.1:c.241G>T
NM_001322007.1:c.26G>T
NM_001322008.1:c.26G>T
NM_001322009.1:c.-165G>T
NM_001322010.1:c.-165G>T
NM_001322011.1:c.-644G>T
NM_001322012.1:c.-644G>T
NM_001322013.1:c.-165G>T
NM_001322014.1:c.241G>T
NM_001322015.1:c.-244G>T
NR_136154.1:n.328G>T
NM_001322003.2:c.-165G>T
NM_001322004.2:c.-165G>T
NM_001322005.2:c.-165G>T
NM_001322006.2:c.241G>T
NM_001322008.2:c.26G>T
NM_001322009.2:c.-165G>T
NM_001322010.2:c.-165G>T
NM_001322011.2:c.-644G>T
NM_001322012.2:c.-644G>T
NM_001322013.2:c.-165G>T
NM_001322014.2:c.241G>T
NM_001322015.2:c.-244G>T
NM_001322007.2:c.26G>T

Likely Pathogenic

Met criteria codes 2

PM2_Supporting

PVS1

Evidence Links 0

Expert Panel

InSiGHT Hereditary Colorectal Cancer/Polypsis VCEP

Criteria Specification Information

Criteria Specification: ClinGen InSiGHT Hereditary Colorectal Cancer/Polypsis Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for PMS2 Version 1.0.0

Criteria Specification Approval History

Criteria Specifications for this VCEP

Evidence submitted by expert panel

InSiGHT Hereditary Colorectal Cancer/Polypsis VCEP

The c.241G>T (p.Glu81Ter) (NM_000535.7) variant in PMS2 is a nonsense variant predicted to cause a premature stop codon in exon 3 in a gene in which loss-of-function is an established disease mechanism (PVS1). This variant is extremely rare (1 in 1594104 alleles) in gnomAD v4.1 (PM2_Supporting). In summary, this variant meets the criteria to be classified as likely pathogenic for Lynch-Syndrome based on the

ACMG/AMP criteria applied, as specified by the ClinGen InSiGHT Hereditary Colorectal Cancer/ Polyposis VCEP: PVS1, PM2_SUP (VCEP specifications version 1)

Met criteria codes

PM2_Supporting			This variant is extremely rare (1 in 1594104 alleles) in gnomAD v4.1 dataset (PM2_Supporting).
PVS1			The c.241G>T (p.Glu81Ter) (NM_000535.7) variant in PMS2 is a nonsense variant predicted to cause a premature stop codon in exon 3 in a gene in which loss-of-function is an established disease mechanism (PVS1).

Curation History

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