

Variant: NM_000038.6(APC):c.7803_7807del (p.Ser2601fs)

Version: 1.0

CA915943525 [↗](#)

648862 (ClinVar) [↗](#)

Gene: APC ([HGNC:324](#))

Condition: familial adenomatous polyposis 1 ([MONDO:0021056](#))

Inheritance Mode: Autosomal dominant inheritance

UUID: bff93256-6b9b-4bf6-ad8c-3124aeddac0b

Approved on: 2025/5/15

Published on: 2025/5/19

HGVS expressions

NM_000038.6(APC):c.7803_7807del (p.Ser2601fs)

NM_000038.6:c.7803_7807del

NC_000005.10:g.112843397_112843401del

CM000667.2:g.112843397_112843401del

NC_000005.9:g.112179094_112179098del

CM000667.1:g.112179094_112179098del

NC_000005.8:g.112206993_112206997del

NG_008481.4:g.155877_155881del

ENST00000504915.3:c.7857_7861del

ENST00000505350.2:c.*7809_*7813del

ENST00000507379.6:c.7749_7753del

ENST00000509732.6:c.7803_7807del

ENST00000512211.7:c.7803_7807del

ENST00000257430.9:c.7803_7807del

ENST00000257430.8:c.7803_7807del

ENST00000508376.6:c.7803_7807del

ENST00000520401.1:c.231-13252_231-13248del

NM_000038.5:c.7803_7807del

NM_001127510.2:c.7803_7807del

NM_001127511.2:c.7749_7753del

NM_001354895.1:c.7803_7807del

NM_001354896.1:c.7857_7861del

NM_001354897.1:c.7833_7837del

NM_001354898.1:c.7728_7732del

NM_001354899.1:c.7719_7723del

NM_001354900.1:c.7680_7684del

NM_001354901.1:c.7626_7630del

NM_001354902.1:c.7530_7534del

NM_001354903.1:c.7500_7504del

NM_001354904.1:c.7425_7429del

NM_001354905.1:c.7323_7327del

NM_001354906.1:c.6954_6958del

NM_001127510.3:c.7803_7807del

NM_001127511.3:c.7749_7753del

NM_001354895.2:c.7803_7807del

NM_001354896.2:c.7857_7861del

NM_001354897.2:c.7833_7837del

NM_001354898.2:c.7728_7732del
NM_001354899.2:c.7719_7723del
NM_001354900.2:c.7680_7684del
NM_001354901.2:c.7626_7630del
NM_001354902.2:c.7530_7534del
NM_001354903.2:c.7500_7504del
NM_001354904.2:c.7425_7429del
NM_001354905.2:c.7323_7327del
NM_001354906.2:c.6954_6958del

Likely Pathogenic

Met criteria codes 2

PM2_Supporting PVS1

Not Met criteria codes 1

PS4

Evidence Links 0

Expert Panel

InSiGHT Hereditary Colorectal Cancer/Polyposis VCEP

Criteria Specification Information

Criteria Specification: ClinGen InSiGHT Hereditary Colorectal Cancer/Polyposis Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for APC Version 2.1.0

Criteria Specification Approval History

Criteria Specifications for this VCEP

Evidence submitted by expert panel

InSiGHT Hereditary Colorectal Cancer/Polyposis VCEP

The NM_000038.6(APC):c.7803_7807del (p.Ser2601ArgfsTer17) variant in APC is a frameshift variant located between codon 49 and 2645 and predicted to cause a premature stop codon in exon 16 in a gene in which loss-of-function is an established disease mechanism (PVS1). This variant is absent from gnomAD v2.1.1 (PM2_Supporting). This variant has been reported in 1 proband with a colorectal cancer/polyposis associated phenotype not meeting phenotypic criteria (PS4 not met; internal data Labcorp Genetics [formerly Invitae]). In summary, this variant meets the criteria to be classified as Likely Pathogenic for autosomal-dominant inherited FAP based on the ACMG/AMP criteria applied, as specified by the ClinGen InSiGHT Hereditary Colorectal Cancer/Polyposis VCEP: PVS1 and PM2_Supporting (VCEP specifications version v2.1.0; date of approval 11/24/2023).

Met criteria codes

PM2_Supporting			This variant is absent from gnomAD v2.1.1 (PM2_Supporting).
PVS1			The NM_000038.6(APC):c.7803_7807del (p.Ser2601ArgfsTer17) variant in APC is a frameshift variant located between codon 49 and 2645 and predicted to cause a premature stop codon in exon 16 in a gene in which loss-of-function is an established disease mechanism (PVS1).

Not Met criteria codes

PS4			This variant has been reported in 1 proband with a colorectal cancer/polyposis associated phenotype not meeting phenotypic criteria (PS4 not met; internal data Labcorp Genetics [formerly Invitae]).
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Curation History



 

 

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