

Variant: NM_000038.6(APC):c.7964_7965del (p.Glu2655fs)

Version: 1.0

CA2543984220 [↗](#)

1361956 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 50834394-dfbd-48f7-a86f-b49477a6b963

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HGVS expressions

NM_000038.6(APC):c.7964_7965del (p.Glu2655fs)

NM_000038.6:c.7964_7965del

NC_000005.10:g.112843558_112843559del

CM000667.2:g.112843558_112843559del

NC_000005.9:g.112179255_112179256del

CM000667.1:g.112179255_112179256del

NC_000005.8:g.112207154_112207155del

NG_008481.4:g.156038_156039del

ENST00000504915.3:c.8018_8019del

ENST00000505350.2:c.*7970_*7971del

ENST00000507379.6:c.7910_7911del

ENST00000509732.6:c.7964_7965del

ENST00000512211.7:c.7964_7965del

ENST00000257430.9:c.7964_7965del

ENST00000257430.8:c.7964_7965del

ENST00000508376.6:c.7964_7965del

ENST00000520401.1:c.231-13091_231-13090del

NM_000038.5:c.7964_7965del

NM_001127510.2:c.7964_7965del

NM_001127511.2:c.7910_7911del

NM_001354895.1:c.7964_7965del

NM_001354896.1:c.8018_8019del

NM_001354897.1:c.7994_7995del

NM_001354898.1:c.7889_7890del

NM_001354899.1:c.7880_7881del

NM_001354900.1:c.7841_7842del

NM_001354901.1:c.7787_7788del

NM_001354902.1:c.7691_7692del

NM_001354903.1:c.7661_7662del

NM_001354904.1:c.7586_7587del

NM_001354905.1:c.7484_7485del

NM_001354906.1:c.7115_7116del

NM_001127510.3:c.7964_7965del

NM_001127511.3:c.7910_7911del

NM_001354895.2:c.7964_7965del

NM_001354896.2:c.8018_8019del

NM_001354897.2:c.7994_7995del

NM_001354898.2:c.7889_7890del
NM_001354899.2:c.7880_7881del
NM_001354900.2:c.7841_7842del
NM_001354901.2:c.7787_7788del
NM_001354902.2:c.7691_7692del
NM_001354903.2:c.7661_7662del
NM_001354904.2:c.7586_7587del
NM_001354905.2:c.7484_7485del
NM_001354906.2:c.7115_7116del

Uncertain Significance

Met criteria codes 1

PM2_Supporting

Not Met criteria codes 3

BS2

PVS1

BP2

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.7964_7965del (p.Glu2655fs) variant in APC is a frameshift variant located downstream of codon 2645, therefore PVS1 is not applicable based on the ACMG/AMP criteria specified by the ClinGen InSiGHT Hereditary Colorectal Cancer/Polyposis VCEP (HCCP VCEP). This variant is absent from gnomAD v2.1.1 (PM2_Supporting). The variant was observed in a 30 years old patient with 3 to 5 histologically confirmed adenomas, a desmoid and a positive FAP family history, who also carried, in unknown phase, NM_000038.6(APC):c.426_427del (p.Leu143fs) (classified as Pathogenic in ClinVar - Variation ID: 142574) (BP2 not met; internal data Labcorp Genetics (formerly Invitae)). The variant has been reported in 4 additional probands without a colorectal cancer/polyposis associated phenotype worth 0.5 healthy individual points in total (BS2 not met, internal data Ambry and Labcorp Genetics (formerly Invitae)). In summary, this variant is a variant of uncertain significance (VUS) for autosomal-dominant inherited FAP based on the ACMG/AMP criteria applied, as specified by the HCCP VCEP: criteria PM2_Supporting applied (VCEP specifications 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).

Not Met criteria codes

BS2



The variant has been reported in 4 additional probands without a colorectal cancer/polyposis associated phenotype worth 0.5 healthy individual points in total (BS2 not met, internal data Ambry and Labcorp Genetics (formerly Invitae)).

PVS1			The NM_000038.6(APC):c.7964_7965del (p.Glu2655fs) variant in APC is a frameshift variant located downstream of codon 2645, therefore PVS1 is not applicable based on the ACMG/AMP criteria specified by the ClinGen InSiGHT Hereditary Colorectal Cancer/Polyposis VCEP (HCCP VCEP).
BP2			The variant was observed in a 30 years old patient with 3 to 5 histologically confirmed adenomas, a desmoid and a positive FAP family history, who also carried, in unknown phase, NM_000038.6(APC):c.426_427del (p.Leu143fs) (classified as Pathogenic in ClinVar - Variation ID: 142574) (BP2 not met; internal data Labcorp Genetics (formerly Invitae)).

Curation History 		
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