

Variant: *NM_000038.6(APC):c.123A>G (p.Ala41=)*

Version: 1.0

CA445752659 [↗](#)

631238 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: e979db2a-db6d-41d1-8719-f439c8a6f20c

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HGVS expressions

NM_000038.6:c.123A>G

NM_000038.6(APC):c.123A>G (p.Ala41=)

NC_000005.10:g.112755013A>G

CM000667.2:g.112755013A>G

NC_000005.9:g.112090710A>G

CM000667.1:g.112090710A>G

NC_000005.8:g.112118609A>G

NG_008481.4:g.67493A>G

ENST00000502371.3:c.123A>G

ENST00000504915.3:c.123A>G

ENST00000505084.2:n.179A>G

ENST00000505350.2:c.*129A>G

ENST00000507379.6:c.166-11313A>G

ENST00000509732.6:c.123A>G

ENST00000512211.7:c.123A>G

ENST00000257430.9:c.123A>G

ENST00000257430.8:c.123A>G

ENST00000505350.1:c.*129A>G

ENST00000507379.5:c.166-11313A>G

ENST00000508376.6:c.123A>G

ENST00000508624.5:c.123A>G

ENST00000509732.5:c.123A>G

ENST00000512211.6:c.123A>G

NM_000038.5:c.123A>G

NM_001127510.2:c.123A>G

NM_001127511.2:c.166-11313A>G

NM_001354895.1:c.123A>G

NM_001354896.1:c.123A>G

NM_001354897.1:c.166-11313A>G

NM_001354898.1:c.61-11313A>G

NM_001354899.1:c.123A>G

NM_001354900.1:c.-42-11313A>G

NM_001354901.1:c.-42-11313A>G

NM_001354902.1:c.166-11313A>G

NM_001354903.1:c.123A>G

NM_001354904.1:c.61-11313A>G

NM_001354905.1:c.-42-11313A>G

NM_001354906.1:c.-913A>G
NM_001127510.3:c.123A>G
NM_001127511.3:c.166-11313A>G
NM_001354895.2:c.123A>G
NM_001354896.2:c.123A>G
NM_001354897.2:c.166-11313A>G
NM_001354898.2:c.61-11313A>G
NM_001354899.2:c.123A>G
NM_001354900.2:c.-42-11313A>G
NM_001354901.2:c.-42-11313A>G
NM_001354902.2:c.166-11313A>G
NM_001354903.2:c.123A>G
NM_001354904.2:c.61-11313A>G
NM_001354905.2:c.-42-11313A>G
NM_001354906.2:c.-913A>G

Likely Benign

Met criteria codes 3

BP7 BP4 PM2_Supporting

Not Met criteria codes 1

BS2

Evidence Links 0

Expert Panel

InSiGHT Hereditary Colorectal Cancer/Polypsis VCEP

Criteria Specification Information

Criteria Specifications for this VCEP

Evidence submitted by expert panel

InSiGHT Hereditary Colorectal Cancer/Polypsis VCEP

The c.123A>G (p.Ala41=) variant in APC is synonymous (silent) variant that is not predicted by SpliceAI and VarSEAK to impact splicing (BP4 and BP7 met). This variant is absent from gnomAD v2.1.1 (PM2_supporting). This variant has been observed in heterozygous state in 1 healthy adult individual worth 0.5 healthy individual points in total (BS2_supporting not met; Invitae internal data). In summary, this variant meets the criteria to be classified as Likely Benign for FAP based on the ACMG/AMP criteria applied, as specified by the ClinGen InSiGHT Hereditary Colorectal Cancer/Polypsis Variant Curation Expert Panel: BP4, BP7 (VCEP specifications version 1; date of approval 12/12/2022).

Met criteria codes

BP7



The c.123A>G (p.Ala41=) variant in APC is synonymous (silent) variant that is not predicted by SpliceAI and VarSEAK to impact splicing (BP4 and BP7 met).

BP4



The c.123A>G (p.Ala41=) variant in APC is synonymous (silent) variant that is not predicted by SpliceAI and VarSEAK to impact splicing (BP4 and BP7 met).

PM2_Supporting



This variant is absent from gnomAD v2.1.1 (PM2_supporting).

Not Met criteria codes

BS2



This variant has been observed in heterozygous state in 1 healthy unrelated adult individuals worth 0.5 healthy individual points in total (BS2_supporting not met; Invitae internal data).

Curation History [↗](#)

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