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Variant: NM_000038.6(APC):c.420G>C (p.Glu140Asp)

Version: 1.0

CA008951

41529 (ClinVar)

Gene: APC (HGNC:324)
Condition: familial adenomatous polyposis 1 (MONDO:0021056)
Inheritance Mode: Autosomal dominant inheritance
UUID: 2e794a2d-90d8-45db-99ed-2ae3f3865213
Approved on: 2023/2/18
Published on: 2023/3/14

HGVS expressions

NM_000038.6:c.420G>C
NM_000038.6(APC):c.420G>C (p.Glu140Asp)
NC_000005.10:g.112767388G>C
CM000667.2:g.112767388G>C
NC_000005.9:g.112103085G>C
CM000667.1:g.112103085G>C
NC_000005.8:g.112130984G>C
NG_008481.4:g.79868G>C
ENST00000502371.3:c.420G>C
ENST00000504915.3:c.420G>C
ENST00000505084.2:n.476G>C
ENST00000505350.2:c.*426G>C
ENST00000507379.6:c.450G>C
ENST00000509732.6:c.420G>C
ENST00000512211.7:c.420G>C
ENST00000257430.9:c.420G>C
ENST00000257430.8:c.420G>C
ENST00000507379.5:c.450G>C
ENST00000508376.6:c.420G>C
ENST00000508624.5:c.420G>C
ENST00000512211.6:c.420G>C
NM_000038.5:c.420G>C
NM_001127510.2:c.420G>C
NM_001127511.2:c.450G>C
NM_001354895.1:c.420G>C
NM_001354896.1:c.420G>C
NM_001354897.1:c.450G>C
NM_001354898.1:c.345G>C
NM_001354899.1:c.420G>C
NM_001354900.1:c.243G>C
NM_001354901.1:c.243G>C
NM_001354902.1:c.450G>C
NM_001354903.1:c.420G>C
NM_001354904.1:c.345G>C
NM_001354905.1:c.243G>C
NM_001354906.1:c.-616G>C
NM_001127510.3:c.420G>C

NM_001127511.3:c.450G>C
NM_001354895.2:c.420G>C
NM_001354896.2:c.420G>C
NM_001354897.2:c.450G>C
NM_001354898.2:c.345G>C
NM_001354899.2:c.420G>C
NM_001354900.2:c.243G>C
NM_001354901.2:c.243G>C
NM_001354902.2:c.450G>C
NM_001354903.2:c.420G>C
NM_001354904.2:c.345G>C
NM_001354905.2:c.243G>C
NM_001354906.2:c.-616G>C

Likely Benign

Met criteria codes 4

PM2_Supporting BS2 BP1 BP2

Evidence Links 0

Expert Panel

InSiGHT Hereditary Colorectal Cancer/Polyposis VCEP

Criteria Specification Information

Criteria Specifications for this VCEP

Evidence submitted by expert panel

InSiGHT Hereditary Colorectal Cancer/Polyposis VCEP

The c.420G>C variant in APC is a missense variant predicted to cause substitution of glutamic acid by aspartic acid at amino acid 140 (p.Glu140Asp). APCis defined by the ClinGen InSiGHT Hereditary Colorectal Cancer/Polyposis Variant Curation Expert Panel (HCCP VCEP) as a gene for which primarily truncating variants are known to cause disease (BP1). This variant has been observed in heterozygous state in 67 healthy unrelated adult individuals worth ≥ 10 healthy individual points in total (BS2; Ambry Genetics, Invitae, GeneDX internal data). This variant has also been observed in trans with a variant classified as (likely) pathogenic by the HCCP VCEP in an individual with FAP (BP2; Invitae Internal Data). Finally, this variant is absent from gnomAD v2.1.1 (PM2_Supporting). 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 HCCP VCEP: BS2, BP1, BP2 (VCEP specifications version 1; date of approval 12/12/2022).

Met criteria codes

PM2_Supporting	✔	This variant is absent from gnomAD v2.1.1 (PM2_Supporting).
BS2	✔	This variant has been observed in heterozygous state in 67 healthy unrelated adult individuals worth ≥ 10 healthy individual points in total (BS2; Ambry Genetics, Invitae, GeneDX internal data).
BP1	✔	APC, in which the variant was identified, is defined by the ClinGen InSiGHT Hereditary Colorectal Cancer/Polyposis VCEP as a gene for which primarily truncating variants are known to cause disease (BP1).
BP2	✔	This variant has been observed in trans with a variant classified as (likely) pathogenic by the ClinGen InSiGHT Hereditary Colorectal Cancer/ Polyposis VCEP in an individual with FAP (BP2; Invitae Internal Data).

Curation History

Showing 1 to 1 of 1 rows

See Report	Preferred Variant Title	Classification	Condition	Published Date	Version	Criteria Specification	Gene
View	NM_000038.6(APC):c.420G>C (p.Glu14…	Likely Benign	Familial Adenomatous Polyposis 1	2023-03-14	1.0	-	APC

Showing 1 to 1 of 1 rows

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