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Variant: NM_000038.6(APC):c.235A>G (p.Ser79Gly)

Version: 1.0

CA16021856

628873 (ClinVar)

Gene: APC (HGNC:324)
Condition: familial adenomatous polyposis 1 (MONDO:0021056)
Inheritance Mode: Autosomal dominant inheritance
UUID: ee94dd49-57a8-410f-8320-64bc1bd28f01
Approved on: 2023/2/18
Published on: 2023/3/14

HGVS expressions

NM_000038.6:c.235A>G
NM_000038.6(APC):c.235A>G (p.Ser79Gly)
NC_000005.10:g.112767203A>G
CM000667.2:g.112767203A>G
NC_000005.9:g.112102900A>G
CM000667.1:g.112102900A>G
NC_000005.8:g.112130799A>G
NG_008481.4:g.79683A>G
ENST00000502371.3:c.235A>G
ENST00000504915.3:c.235A>G
ENST00000505084.2:n.291A>G
ENST00000505350.2:c.*241A>G
ENST00000507379.6:c.265A>G
ENST00000509732.6:c.235A>G
ENST00000512211.7:c.235A>G
ENST00000257430.9:c.235A>G
ENST00000257430.8:c.235A>G
ENST00000507379.5:c.265A>G
ENST00000508376.6:c.235A>G
ENST00000508624.5:c.235A>G
ENST00000509732.5:c.235A>G
ENST00000512211.6:c.235A>G
NM_000038.5:c.235A>G
NM_001127510.2:c.235A>G
NM_001127511.2:c.265A>G
NM_001354895.1:c.235A>G
NM_001354896.1:c.235A>G
NM_001354897.1:c.265A>G
NM_001354898.1:c.160A>G
NM_001354899.1:c.235A>G
NM_001354900.1:c.58A>G
NM_001354901.1:c.58A>G
NM_001354902.1:c.265A>G
NM_001354903.1:c.235A>G
NM_001354904.1:c.160A>G
NM_001354905.1:c.58A>G
NM_001354906.1:c.-801A>G

NM_001127510.3:c.235A>G
NM_001127511.3:c.265A>G
NM_001354895.2:c.235A>G
NM_001354896.2:c.235A>G
NM_001354897.2:c.265A>G
NM_001354898.2:c.160A>G
NM_001354899.2:c.235A>G
NM_001354900.2:c.58A>G
NM_001354901.2:c.58A>G
NM_001354902.2:c.265A>G
NM_001354903.2:c.235A>G
NM_001354904.2:c.160A>G
NM_001354905.2:c.58A>G
NM_001354906.2:c.-801A>G

Likely Benign

Met criteria codes 3

BP1 PM2_Supporting BS2_Supporting

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.235A>G variant in APC is a missense variant predicted to cause substitution of serine by glycine at amino acid 79 (p.Ser79Gly). This variant is absent from gnomAD v2.1.1 (PM2_Supporting). APC is 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 6 heterozygous individuals with no features of FAP, worth 3 healthy individual points (BS2_Supporting; Ambry Genetics Internal Data; 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 HCCP VCEP: BS2_supporting, BP1. (VCEP specifications version 1; date of approval: 12/12/2022).

Met criteria codes

BP1	✔	APC, in which the variant was identified, is defined by the InSiGHT ClinGen Hereditary Colorectal Cancer/Polyposis VCEP as a gene for which primarily truncating variants are known to cause disease (BP1).
PM2_Supporting	✔	This variant is absent from gnomAD v2.1.1 (PM2_Supporting).
BS2_Supporting	✔	This variant has been observed in 6 heterozygous individuals worth ≥ 3 healthy individual points in total (BS2_Supporting met; Ambry Internal Data; GeneDx 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.235A>G (p.Ser79...	Likely Benign	Familial Adenomatous Polyposis 1	2023-03-14	1.0	-	APC

Showing 1 to 1 of 1 rows

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