

Variant: NM_000038.6(APC):c.5465T>A (p.Val1822Asp)

Version: 1.0

CA010422 [↗](#)

21030 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 382336bf-1845-4ef0-9f6d-e061437c0adb

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HGVS expressions

NM_000038.6:c.5465T>A

NM_000038.6(APC):c.5465T>A (p.Val1822Asp)

NC_000005.10:g.112841059T>A

CM000667.2:g.112841059T>A

NC_000005.9:g.112176756T>A

CM000667.1:g.112176756T>A

NC_000005.8:g.112204655T>A

NG_008481.4:g.153539T>A

ENST00000504915.3:c.5519T>A

ENST00000505350.2:c.*5471T>A

ENST00000507379.6:c.5411T>A

ENST00000509732.6:c.5465T>A

ENST00000512211.7:c.5465T>A

ENST00000257430.9:c.5465T>A

ENST00000257430.8:c.5465T>A

ENST00000508376.6:c.5465T>A

ENST00000508624.5:c.*4787T>A

ENST00000520401.1:c.230+12087T>A

NM_000038.5:c.5465T>A

NM_001127510.2:c.5465T>A

NM_001127511.2:c.5411T>A

NM_001354895.1:c.5465T>A

NM_001354896.1:c.5519T>A

NM_001354897.1:c.5495T>A

NM_001354898.1:c.5390T>A

NM_001354899.1:c.5381T>A

NM_001354900.1:c.5342T>A

NM_001354901.1:c.5288T>A

NM_001354902.1:c.5192T>A

NM_001354903.1:c.5162T>A

NM_001354904.1:c.5087T>A

NM_001354905.1:c.4985T>A

NM_001354906.1:c.4616T>A

NM_001127510.3:c.5465T>A

NM_001127511.3:c.5411T>A

NM_001354895.2:c.5465T>A

NM_001354896.2:c.5519T>A

NM_001354897.2:c.5495T>A
NM_001354898.2:c.5390T>A
NM_001354899.2:c.5381T>A
NM_001354900.2:c.5342T>A
NM_001354901.2:c.5288T>A
NM_001354902.2:c.5192T>A
NM_001354903.2:c.5162T>A
NM_001354904.2:c.5087T>A
NM_001354905.2:c.4985T>A
NM_001354906.2:c.4616T>A

Benign

Met criteria codes 2

BA1 BP1

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.5465T>A variant in APC is a missense variant predicted to cause the substitution of Valine by Asparagine at amino acid position 1822 (p.Val1822Asp). 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). The highest population minor allele frequency in gnomAD v2.1.1 is 0.9598 (23897/24898 alleles) in the African/African American population, which is higher than the ClinGen APC VCEP threshold (>0.1%) for BA1, and therefore meets this criterion (BA1). In summary, this variant meets the criteria to be classified as Benign for FAP based on the ACMG/AMP criteria applied, as specified by the HCCP VCEP: BA1 and BP1 (VCEP specifications version 1; date of approval: 12/12/2022).

Met criteria codes

BA1



The highest population minor allele frequency in gnomAD v2.1.1 is 0.9598 (23897/24898 alleles) in the African/African American population, which is higher than the ClinGen APC VCEP threshold (>0.1%) for BA1, and therefore meets this criterion (BA1).

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).

Curation History

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