

Variant: *NM_000038.6(APC):c.1956C>T (p.His652=)*

Version: 1.0

CA16618077 [↗](#)

419202 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 92b30bd7-d4e0-4d93-98c4-99e7e30f70c2

Approved on: 2023/2/25

Published on: 2023/3/14

HGVS expressions

NM_000038.6:c.1956C>T

NM_000038.6(APC):c.1956C>T (p.His652=)

NC_000005.10:g.112835163C>T

CM000667.2:g.112835163C>T

NC_000005.9:g.112170860C>T

CM000667.1:g.112170860C>T

NC_000005.8:g.112198759C>T

NG_008481.4:g.147643C>T

ENST00000502371.3:c.1621C>T

ENST00000504915.3:c.2010C>T

ENST00000505350.2:c.*1962C>T

ENST00000507379.6:c.1902C>T

ENST00000509732.6:c.1956C>T

ENST00000512211.7:c.1956C>T

ENST00000257430.9:c.1956C>T

ENST00000257430.8:c.1956C>T

ENST00000502371.2:c.309C>T

ENST00000504915.2:c.645C>T

ENST00000507379.5:c.1902C>T

ENST00000508376.6:c.1956C>T

ENST00000508624.5:c.*1278C>T

ENST00000512211.6:c.1956C>T

ENST00000520401.1:c.230+6191C>T

NM_000038.5:c.1956C>T

NM_001127510.2:c.1956C>T

NM_001127511.2:c.1902C>T

NM_001354895.1:c.1956C>T

NM_001354896.1:c.2010C>T

NM_001354897.1:c.1986C>T

NM_001354898.1:c.1881C>T

NM_001354899.1:c.1872C>T

NM_001354900.1:c.1833C>T

NM_001354901.1:c.1779C>T

NM_001354902.1:c.1683C>T

NM_001354903.1:c.1653C>T

NM_001354904.1:c.1578C>T

NM_001354905.1:c.1476C>T

NM_001354906.1:c.1107C>T
NM_001127510.3:c.1956C>T
NM_001127511.3:c.1902C>T
NM_001354895.2:c.1956C>T
NM_001354896.2:c.2010C>T
NM_001354897.2:c.1986C>T
NM_001354898.2:c.1881C>T
NM_001354899.2:c.1872C>T
NM_001354900.2:c.1833C>T
NM_001354901.2:c.1779C>T
NM_001354902.2:c.1683C>T
NM_001354903.2:c.1653C>T
NM_001354904.2:c.1578C>T
NM_001354905.2:c.1476C>T
NM_001354906.2:c.1107C>T

Likely Pathogenic

Met criteria codes 3

PM2_Supporting

PS4_Supporting

PS3

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.1956C>T variant in APC is predicted to be a synonymous (silent) variant (p.His652=). It is located at the third-last nucleotide of exon 15. Two RNA based RT-PCR assays demonstrate out-of-frame skipping of exon 15, with evidence that the variant allele produces <10% of the full-length transcript (PS3; PMID: 15459959, 22987206). This variant has been reported in 3 probands meeting least 1.5 phenotype points (PS4_Supporting; PMID: 15459959, 22987206). The variant is not reported in gnomAD (PM2_supporting). In summary, this variant meets the criteria to be classified as Likely Pathogenic for FAP based on the ACMG/AMP criteria applied, as specified by the ClinGen InSiGHT Hereditary Colorectal Cancer/Polyposis Variant Curation Expert Panel: PS3, PS4_Supporting and PM2_Supporting (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).

PS4_Supporting



1,5 phenotype points (PS4_Supporting): PMID: 15459959: index and father with $\geq$ 20 colorectal adenomas a with 20 to 70 yr PMID: 22987206: index with $\geq$ 20 colorectal adenomas a with 20 to 70 yr, mother affected CRC but no phenotypic information

PS3



PS3 according to the APC-VCEP modification of PS3: two RNA based RT-PCR assays demonstrate out-of-frame skipping of exon 15, with evidence that the variant allele produces < 10% of the full-length transcript.

Curation History

Showing 1 to 1 of 1 rows

ClinGen Terms of Use.
 ✕ Powered by BCM's Genboree.