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Variant: NM_000038.6(APC):c.694C>T (p.Arg232Ter)

Version: 1.0

CA012693

42248 (ClinVar)

Gene: APC (HGNC:324)
Condition: familial adenomatous polyposis 1 (MONDO:0021056)
Inheritance Mode: Autosomal dominant inheritance
UUID: 939c0109-5c27-4f3a-ad04-3e81276ff0ad
Approved on: 2025/5/19
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HGVS expressions

NM_000038.6(APC):c.694C>T (p.Arg232Ter)
NM_000038.6:c.694C>T
NC_000005.10:g.112792494C>T
CM000667.2:g.112792494C>T
NC_000005.9:g.112128191C>T
CM000667.1:g.112128191C>T
NC_000005.8:g.112156090C>T
NG_008481.4:g.104974C>T
ENST00000502371.3:c.694C>T
ENST00000504915.3:c.694C>T
ENST00000505084.2:n.750C>T
ENST00000505350.2:c.*700C>T
ENST00000507379.6:c.676-8785C>T
ENST00000509732.6:c.694C>T
ENST00000512211.7:c.694C>T
ENST00000257430.9:c.694C>T
ENST00000257430.8:c.694C>T
ENST00000507379.5:c.676-8785C>T
ENST00000508376.6:c.694C>T
ENST00000508624.5:c.694C>T
ENST00000512211.6:c.694C>T
NM_000038.5:c.694C>T
NM_001127510.2:c.694C>T
NM_001127511.2:c.676-8785C>T
NM_001354895.1:c.694C>T
NM_001354896.1:c.694C>T
NM_001354897.1:c.724C>T
NM_001354898.1:c.619C>T
NM_001354899.1:c.646-8785C>T
NM_001354900.1:c.517C>T
NM_001354901.1:c.517C>T
NM_001354902.1:c.724C>T
NM_001354903.1:c.694C>T
NM_001354904.1:c.619C>T
NM_001354905.1:c.517C>T
NM_001354906.1:c.-342C>T
NM_001127510.3:c.694C>T

NM_001127511.3:c.676-8785C>T
NM_001354895.2:c.694C>T
NM_001354896.2:c.694C>T
NM_001354897.2:c.724C>T
NM_001354898.2:c.619C>T
NM_001354899.2:c.646-8785C>T
NM_001354900.2:c.517C>T
NM_001354901.2:c.517C>T
NM_001354902.2:c.724C>T
NM_001354903.2:c.694C>T
NM_001354904.2:c.619C>T
NM_001354905.2:c.517C>T
NM_001354906.2:c.-342C>T

Pathogenic

Met criteria codes 3

PM2_Supporting PVS1 PS4

Evidence Links 0

Expert Panel

InSiGHT Hereditary Colorectal Cancer/Polyposis VCEP

Criteria Specification Information

Criteria Specification: *ClinGen InSiGHT Hereditary Colorectal Cancer/Polyposis Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for APC Version 2.1.0*

Criteria Specification Approval History

Criteria Specifications for this VCEP

Evidence submitted by expert panel

InSiGHT Hereditary Colorectal Cancer/Polyposis VCEP

The NM_000038.6:c.694C>T p.(Arg232Ter) variant in APC is a nonsense variant located between codon 49 and 2645 and predicted to cause a premature stop codon in exon 7 in a gene in which loss-of-function is an established disease mechanism (PVS1). This variant has been reported in at least 10 probands meeting phenotypic criteria, resulting in a total phenotype score of > 4 (PS4, [PMID: 1316610, 20223039, 7524601, 21110124, 19279422 and several more]). This variant is absent from gnomAD v2.1.1 (PM2_Supporting). In summary, this variant meets the criteria to be classified as Pathogenic for autosomal-dominant inherited FAP based on the ACMG/AMP criteria applied, as specified by the ClinGen InSiGHT Hereditary Colorectal Cancer/Polyposis VCEP: criteria PVS1, PS4 and PM2_Supporting applied (VCEP specifications version v2.1.0; date of approval 11/24/2023).

Met criteria codes

PM2_Supporting	✔	This variant is absent from gnomAD v2.1.1 (PM2_Supporting).
PVS1	✔	The NM_000038.6:c.694C>T p.(Arg232Ter) variant in APC is a nonsense variant located between codon 49 and 2645 and predicted to cause a premature stop codon in exon 7 in a gene in which loss-of-function is an established disease mechanism (PVS1).
PS4	✔	

This variant has been reported in at least 10 probands meeting phenotypic criteria, resulting in a total phenotype score of > 4 (PS4, [PMID: 1316610, 20223039, 7524601, 21110124, 19279422 and several more]).

Curation History

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