The ClinGen Evidence Repository is an **FDA-recognized human genetic variant database** containing expert-curated assertions regarding variants' pathogenicity and supporting evidence summaries. [Disclaimer]

Variant: NM_000038.6(APC):c.8446C>T (p.Arg2816Ter)

Version: 1.0

CA16039654

2562354 (ClinVar)

Gene: APC (HGNC:324)
Condition: classic or attenuated familial adenomatous polyposis (MONDO:0021057)
Inheritance Mode: Autosomal dominant inheritance
UUID: 49b80a12-8bbe-4852-8587-ace119076134
Approved on: 2025/5/19
Published on: 2025/5/19

HGVS expressions

NM_000038.6:c.8446C>T
NM_000038.6(APC):c.8446C>T (p.Arg2816Ter)
NC_000005.10:g.112844040C>T
CM000667.2:g.112844040C>T
NC_000005.9:g.112179737C>T
CM000667.1:g.112179737C>T
NC_000005.8:g.112207636C>T
NG_008481.4:g.156520C>T
ENST00000504915.3:c.8500C>T
ENST00000505350.2:c.*8452C>T
ENST00000507379.6:c.8392C>T
ENST00000509732.6:c.8446C>T
ENST00000512211.7:c.8446C>T
ENST00000257430.9:c.8446C>T
ENST00000257430.8:c.8446C>T
ENST00000508376.6:c.8446C>T
ENST00000520401.1:c.231-12609C>T
NM_000038.5:c.8446C>T
NM_001127510.2:c.8446C>T
NM_001127511.2:c.8392C>T
NM_001354895.1:c.8446C>T
NM_001354896.1:c.8500C>T
NM_001354897.1:c.8476C>T
NM_001354898.1:c.8371C>T
NM_001354899.1:c.8362C>T
NM_001354900.1:c.8323C>T
NM_001354901.1:c.8269C>T
NM_001354902.1:c.8173C>T
NM_001354903.1:c.8143C>T
NM_001354904.1:c.8068C>T
NM_001354905.1:c.7966C>T
NM_001354906.1:c.7597C>T
NM_001127510.3:c.8446C>T
NM_001127511.3:c.8392C>T
NM_001354895.2:c.8446C>T
NM_001354896.2:c.8500C>T
NM_001354897.2:c.8476C>T

NM_001354898.2:c.8371C>T
NM_001354899.2:c.8362C>T
NM_001354900.2:c.8323C>T
NM_001354901.2:c.8269C>T
NM_001354902.2:c.8173C>T
NM_001354903.2:c.8143C>T
NM_001354904.2:c.8068C>T
NM_001354905.2:c.7966C>T
NM_001354906.2:c.7597C>T

Uncertain Significance

Met criteria codes 1

PM2_Supporting

Not Met criteria codes 2

BS2 PVS1

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(APC):c.8446C>T (p.Arg2816Ter) variant in APC is a nonsense variant located downstream of codon 2645, therefore PVS1 is not applicable based on the ACMG/AMP criteria specified by the ClinGen InSiGHT Hereditary Colorectal Cancer/Polyposis VCEP (HCCP VCEP). This variant is absent from gnomAD v2.1.1 (PM2_Supporting). This variant has been reported in two individuals without a colorectal cancer/polyposis associated phenotype worth 1 healthy individual point in total (BS2 not met; internal data Labcorp Genetics (formerly Invitae) and Ambry). In summary, this variant is a variant of uncertain significance (VUS) for autosomal-dominant inherited FAP based on the ACMG/AMP criteria applied, as specified by the HCCP VCEP: criteria 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).

Not Met criteria codes

BS2	✖	This variant has been reported in two individuals without a colorectal cancer/polyposis associated phenotype worth 1 healthy individual point in total (BS2 not met; internal data Labcorp Genetics (formerly Invitae) and Ambry).
PVS1	✖	The NM_000038.6(APC):c.8446C>T (p.Arg2816Ter) variant in APC is a nonsense variant located downstream of codon 2645, therefore PVS1 is not applicable based on the ACMG/AMP criteria specified by the ClinGen InSiGHT Hereditary Colorectal Cancer/Polyposis VCEP (HCCP VCEP).

Curation History

Showing 1 to 1 of 1 rows

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