

Variant: NM_000038.6(APC):c.8488dup (p.Ser2830fs)

Version: 1.0

CA2697546419 [↗](#)

2773776 (ClinVar) [↗](#)

Gene: APC ([HGNC:324](#))
Condition: classic or attenuated familial adenomatous polyposis ([MONDO:0021057](#))
Inheritance Mode: Autosomal dominant inheritance
UUID: 82e1ee8f-0d68-4d27-9112-f8370590e4ea
Approved on: 2025/5/19
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HGVS expressions

NM_000038.6:c.8488dup
NM_000038.6(APC):c.8488dup (p.Ser2830fs)
NC_000005.10:g.112844082dup
CM000667.2:g.112844082dup
NC_000005.9:g.112179779dup
CM000667.1:g.112179779dup
NC_000005.8:g.112207678dup
NG_008481.4:g.156562dup
ENST00000504915.3:c.8542dup
ENST00000505350.2:c.*8494dup
ENST00000507379.6:c.8434dup
ENST00000509732.6:c.8488dup
ENST00000512211.7:c.8488dup
ENST00000257430.9:c.8488dup
ENST00000257430.8:c.8488dup
ENST00000508376.6:c.8488dup
ENST00000520401.1:c.231-12567dup
NM_000038.5:c.8488dup
NM_001127510.2:c.8488dup
NM_001127511.2:c.8434dup
NM_001354895.1:c.8488dup
NM_001354896.1:c.8542dup
NM_001354897.1:c.8518dup
NM_001354898.1:c.8413dup
NM_001354899.1:c.8404dup
NM_001354900.1:c.8365dup
NM_001354901.1:c.8311dup
NM_001354902.1:c.8215dup
NM_001354903.1:c.8185dup
NM_001354904.1:c.8110dup
NM_001354905.1:c.8008dup
NM_001354906.1:c.7639dup
NM_001127510.3:c.8488dup
NM_001127511.3:c.8434dup
NM_001354895.2:c.8488dup
NM_001354896.2:c.8542dup
NM_001354897.2:c.8518dup

NM_001354898.2:c.8413dup
NM_001354899.2:c.8404dup
NM_001354900.2:c.8365dup
NM_001354901.2:c.8311dup
NM_001354902.2:c.8215dup
NM_001354903.2:c.8185dup
NM_001354904.2:c.8110dup
NM_001354905.2:c.8008dup
NM_001354906.2:c.7639dup

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/Polypsis VCEP

The NM_000038.6(APC):c.8488dup (p.Ser2830fs*3) variant in APC is a frameshift 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/Polypsis VCEP (HCCP VCEP). This variant is absent from gnomAD v2.1.1 (PM2_Supporting). The variant has been reported in one individual without a colorectal cancer/polypsis associated phenotype not meeting criteria for BS2 (BS2 not met; Ambry internal data). 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   The variant has been reported in one individual without a colorectal cancer/polypsis associated phenotype not meeting criteria for BS2 (BS2 not met; Ambry internal data).

PVS1   The NM_000038.6(APC):c.8488dup (p.Ser2830fs*3) variant in APC is a frameshift 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/Polypsis VCEP (HCCP VCEP).

Curation History



 

 

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