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.5038C>T (p.Gln1680Ter)

Version: 1.0

CA10578390

230520 (ClinVar)

Gene: APC (HGNC:324)
Condition: familial adenomatous polyposis 1 (MONDO:0021056)
Inheritance Mode: Autosomal dominant inheritance
UUID: 05f45393-7afc-483e-bc11-5723e575bad1
Approved on: 2025/5/19
Published on: 2025/5/19

HGVS expressions

NM_000038.6(APC):c.5038C>T (p.Gln1680Ter)
NM_000038.6:c.5038C>T
NC_000005.10:g.112840632C>T
CM000667.2:g.112840632C>T
NC_000005.9:g.112176329C>T
CM000667.1:g.112176329C>T
NC_000005.8:g.112204228C>T
NG_008481.4:g.153112C>T
ENST00000504915.3:c.5092C>T
ENST00000505350.2:c.*5044C>T
ENST00000507379.6:c.4984C>T
ENST00000509732.6:c.5038C>T
ENST00000512211.7:c.5038C>T
ENST00000257430.9:c.5038C>T
ENST00000257430.8:c.5038C>T
ENST00000508376.6:c.5038C>T
ENST00000508624.5:c.*4360C>T
ENST00000520401.1:c.230+11660C>T
NM_000038.5:c.5038C>T
NM_001127510.2:c.5038C>T
NM_001127511.2:c.4984C>T
NM_001354895.1:c.5038C>T
NM_001354896.1:c.5092C>T
NM_001354897.1:c.5068C>T
NM_001354898.1:c.4963C>T
NM_001354899.1:c.4954C>T
NM_001354900.1:c.4915C>T
NM_001354901.1:c.4861C>T
NM_001354902.1:c.4765C>T
NM_001354903.1:c.4735C>T
NM_001354904.1:c.4660C>T
NM_001354905.1:c.4558C>T
NM_001354906.1:c.4189C>T
NM_001127510.3:c.5038C>T
NM_001127511.3:c.4984C>T
NM_001354895.2:c.5038C>T
NM_001354896.2:c.5092C>T

NM_001354897.2:c.5068C>T
NM_001354898.2:c.4963C>T
NM_001354899.2:c.4954C>T
NM_001354900.2:c.4915C>T
NM_001354901.2:c.4861C>T
NM_001354902.2:c.4765C>T
NM_001354903.2:c.4735C>T
NM_001354904.2:c.4660C>T
NM_001354905.2:c.4558C>T
NM_001354906.2:c.4189C>T

Likely Pathogenic

Met criteria codes 2

PVS1 PM2_Supporting

Not Met criteria codes 1

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(APC):c.5038C>T (p.Gln1680Ter) variant in APC is a nonsense variant located between codon 49 and 2645 and predicted to cause a premature stop codon in exon 16 in a gene in which loss-of-function is an established disease mechanism (PVS1). The variant has been reported in 1 proband with a colorectal cancer/polyposis associated phenotype not meeting phenotypic criteria (PS4 not met; Ambry Genetics). This variant is absent from gnomAD v2.1.1 (PM2_Supporting). In summary, this variant meets the criteria to be classified as Likely 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 and PM2_Supporting applied (VCEP specifications version v2.1.0; date of approval 11/24/2023).

Met criteria codes

PVS1	✔	The NM_000038.6(APC):c.5038C>T (p.Gln1680Ter) variant in APC is a nonsense variant located between codon 49 and 2645 and predicted to cause a premature stop codon in exon 16 in a gene in which loss-of-function is an established disease mechanism (PVS1).
PM2_Supporting	✔	This variant is absent from gnomAD v2.1.1 (PM2_Supporting).

Not Met criteria codes

PS4	✖	

The variant has been reported in 1 proband with a colorectal cancer/polyposis associated phenotype not meeting phenotypic criteria (PS4 not met; Ambry Genetics).

Curation History

Showing 1 to 1 of 1 rows

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