

Variant: *NM_000038.6(APC):c.203del (p.Leu68fs)*

Version: 1.0

CA1139658985 [↗](#)

934724 (ClinVar) [↗](#)

Gene: APC ([HGNC:324](#))
Condition: familial adenomatous polyposis 1 ([MONDO:0021056](#))
Inheritance Mode: Autosomal dominant inheritance
UUID: 11e3fbb8-fe19-45e6-a2dc-435180a69a70
Approved on: 2025/5/19
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HGVS expressions

NM_000038.6(APC):c.203del (p.Leu68fs)
NM_000038.6:c.203del
NC_000005.10:g.112766393del
CM000667.2:g.112766393del
NC_000005.9:g.112102090del
CM000667.1:g.112102090del
NC_000005.8:g.112129989del
NG_008481.4:g.78873del
ENST00000502371.3:c.203del
ENST00000504915.3:c.203del
ENST00000505084.2:n.259del
ENST00000505350.2:c.*209del
ENST00000507379.6:c.233del
ENST00000509732.6:c.203del
ENST00000512211.7:c.203del
ENST00000257430.9:c.203del
ENST00000257430.8:c.203del
ENST00000507379.5:c.233del
ENST00000508376.6:c.203del
ENST00000508624.5:c.203del
ENST00000509732.5:c.203del
ENST00000512211.6:c.203del
NM_000038.5:c.203del
NM_001127510.2:c.203del
NM_001127511.2:c.233del
NM_001354895.1:c.203del
NM_001354896.1:c.203del
NM_001354897.1:c.233del
NM_001354898.1:c.128del
NM_001354899.1:c.203del
NM_001354900.1:c.26del
NM_001354901.1:c.26del
NM_001354902.1:c.233del
NM_001354903.1:c.203del
NM_001354904.1:c.128del
NM_001354905.1:c.26del
NM_001354906.1:c.-833del

NM_001127510.3:c.203del
NM_001127511.3:c.233del
NM_001354895.2:c.203del
NM_001354896.2:c.203del
NM_001354897.2:c.233del
NM_001354898.2:c.128del
NM_001354899.2:c.203del
NM_001354900.2:c.26del
NM_001354901.2:c.26del
NM_001354902.2:c.233del
NM_001354903.2:c.203del
NM_001354904.2:c.128del
NM_001354905.2:c.26del
NM_001354906.2:c.-833del

Pathogenic

Met criteria codes 3

PM2_Supporting

PVS1

PS4_Supporting

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.203del (p.Leu68TyrfsTer2) variant in APC is a nonsense variant located between codon 49 and 2645 and predicted to cause a premature stop codon in exon 3 in a gene in which loss-of-function is an established disease mechanism (PVS1). This variant has been reported in 2 probands meeting phenotypic criteria, resulting in a total phenotype score of 1 (PS4_Supporting, internal data Invitae, MGZ Medical Genetics Center). 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: criteria PVS1, PS4_Supporting and PM2_Supporting applied (VCEP specifications v2.0.3; date of approval 7/24/2023).

Met criteria codes

PM2_Supporting



This variant is absent from gnomAD v2.1.1 (PM2_Supporting).

PVS1



The NM_000038.6(APC):c.203del (p.Leu68TyrfsTer2) variant in APC is a nonsense variant located between codon 49 and 2645 and predicted to cause a premature stop codon in exon 3 in a gene in which loss-of-function is an established disease mechanism (PVS1).

PS4_Supporting

This variant has been reported in 2 probands meeting phenotypic criteria, resulting in a total phenotype score of 1 (PS4_Supporting, internal data Invitae, MGZ Medical Genetics Center).

Curation History

Showing 1 to 1 of 1 rows

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