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A Descriptive Molecular Case Series of Novel Variants in APC-associated Polyposis

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ABSTRACT

Aim: Familial adenomatous polyposis (FAP) is the most common hereditary adenomatous polyposis syndrome caused by pathogenic variants in the *APC* gene. This study aimed to describe the clinical and molecular characteristics of four unrelated patients harboring previously unreported APC variants.

Methods: Four unrelated patients referred for suspected hereditary cancer predisposition were included in the study. Genomic deoxyribonucleic acid extracted from peripheral blood samples was analyzed using a hereditary cancer syndrome multigene panel via next-generation sequencing.

Results: Four previously unreported heterozygous APC variants were identified: NM_000038.6:c.1621del (p.Gln541SerfsTer8), c.2194_2195del (p.Asn732Ter), c.3804_3808del (p.Ile1269PhefsTer5), and c.4066_4067dup (p.Gly1357GlnfsTer59). All variants were predicted to be loss-of-function and were expected to result in premature protein truncation. Only one variant was located within the mutation cluster region (MCR), whereas the remaining three were identified outside the MCR in patients with classical FAP.

Conclusion: This case series expands the molecular spectrum of APC-associated polyposis by describing four previously unreported protein-truncating variants. Our findings further illustrate the clinical heterogeneity associated with APC-related disorders and underscore the value of comprehensive molecular characterization in individuals with suspected hereditary cancer predisposition.

Keywords: APC, FAP, protein-truncating variants, hereditary cancer predisposition

Introduction

Adenomatous polyposis (AP) syndromes are hereditary cancer-predisposition disorders characterized by the development of multiple colorectal adenomatous polyps and a markedly increased lifetime risk of colorectal cancer (CRC). In addition to colorectal involvement, affected individuals may develop a broad spectrum of extraintestinal manifestations, including both benign and malign lesions involving the upper gastrointestinal tract and other organs, based on the underlying genetic defect [1].

Familial AP (FAP) is the most common and best-characterized form of AP and results from heterozygous germline pathogenic variants in the *APC* gene. APC functions as a key tumor suppressor and a negative regulator of the Wnt/β-catenin signaling pathway, thereby playing a critical role in the regulation of cellular proliferation and differentiation [2]. In accordance with Knudson's two-hit hypothesis, tumor initiation occurs following somatic inactivation of the remaining wild-type APC allele (the "second hit"), leading to disruption of the β-catenin destruction complex and constitutive activation of Wnt signaling, a hallmark of early colorectal tumorigenesis [3]. Consistent with this central role, somatic APC alterations are detected in approximately 80% of sporadic CRCs [4,5].

FAP exhibits considerable clinical heterogeneity. The classical phenotype is characterized by the development of hundreds to thousands of colorectal adenomas, beginning in childhood or adolescence, and, if left untreated, confers an almost

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inevitable risk of CRC. In contrast, attenuated FAP is typically associated with a lower adenoma burden and a later age of disease onset. In addition to colorectal polyposis, affected individuals may present with a variety of extraintestinal manifestations, including duodenal adenomas, desmoid tumors, osteomas, congenital hypertrophy of the retinal pigment epithelium, dental abnormalities, and epidermoid cysts. Among these manifestations, upper gastrointestinal neoplasms and desmoid disease represent major contributors to disease-related morbidity and mortality [6].

Accumulating evidence has demonstrated that the location of pathogenic variants within the *APC* gene influences disease severity and specific clinical manifestations. Variants located within the mutation cluster region (MCR) in exon 15 are generally associated with a more severe polyposis phenotype, whereas variants toward the 5' and 3' ends of the gene tend to be associated with attenuated disease. Nevertheless, substantial phenotypic variability has been observed even among individuals harboring identical pathogenic variants, suggesting that additional genetic, epigenetic, and environmental factors contribute to disease expression beyond variant location alone [1].

The widespread implementation of next-generation sequencing (NGS) has substantially expanded the mutational spectrum of APC, leading to the identification of numerous previously unreported variants. Defining the clinical significance of these variants is essential for accurate molecular diagnosis, genetic counseling, risk assessment, and clinical management. In the present study, we describe the clinical and molecular characteristics of four unrelated individuals carrying previously unreported APC variants and thereby contribute to the expanding molecular spectrum of APC-associated polyposis.

Methods

Genetic Analysis

Four unrelated patients who were referred to University of Health Science Türkiye, Ankara Etlik City Hospital between 2022 and 2025 for the evaluation of suspected hereditary cancer predisposition were included in this retrospective, descriptive molecular case series. Patients with molecularly confirmed APC-associated polyposis and sufficient clinical and molecular data for retrospective review were included in the study. The study was conducted in accordance with the Declaration of Helsinki. This has been approved by the Ethics Committee of the University of Health Science Türkiye, Ankara Etlik City Hospital (approval no: AEŞH-BADEK-2024-845, date: 25.09.2024). Written informed consent was obtained from all participants or their legal guardians before genetic testing.

All patients underwent hereditary cancer syndrome (HCS) multigene panel testing based on their personal and/or family histories suggestive of an inherited cancer predisposition syndrome. Genomic deoxyribonucleic acid was extracted from peripheral blood samples and analyzed by NGS using the SOPHiA GENETICS™ Custom Hereditary Cancer Solution (CHCS_C_v2) panel on the Illumina NextSeq 2000 platform. The panel comprised the following 60 cancer-predisposition genes: *APC, ATM, AXIN2, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDK4, CDKN2A, CHEK2, DDB2, EPCAM, ERCC2, ERCC3, ERCC4, ERCC5, FANCA, FANCC, FH, FLCN, GALNT12, HDAC2, HOXB13, MEN1, MET, MITF, MLH1, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NF2, NTHL1, PALB2, PMS2, PMS2CL, POLD1, POLE, POLH, PTCH1, PTEN, RAD51C, RAD51D, RB1, RET, SMAD4, STK11, TP53, TSC1, TSC2, VHL, WT1, XPA,* and *XPC*. Sequence data were aligned to the *GRCh38* human reference genome and analyzed using the SOPHiA DDM™ platform.

Variants were filtered according to the validated quality control criteria of the SOPHiA DDM™ pipeline, including read depth, variant allele fraction, mapping quality, and coverage metrics, and were subsequently reviewed manually using the Integrative Genomics Viewer (IGV). Across all samples, the average sequencing depth in the targeted regions was approximately 500×, and approximately 100% of the target bases achieved a sequencing depth of at least 25×. Orthogonal confirmation by Sanger sequencing was not routinely performed. However, all reported variants were supported by high sequencing depth, appropriate variant allele fractions, and manual review.

Variant interpretation was performed according to the American College of Medical Genetics and Genomics/ Association for Molecular Pathology (ACMG/AMP) guidelines using the APC-specific recommendations provided by the ClinGen InSiGHT Hereditary CRC/Polyposis Variant Curation Expert Panel (VCEP; version 1.0). Variant novelty was assessed by systematically reviewing ClinVar, LOVD, gnomAD (v4.1.1), Franklin, and PubMed. In addition to exact Human Genome Variation Society (HGVS) matches, alternative HGVS representations were also evaluated to minimize the possibility of overlooking previously reported variants. Variants were considered novel when no exact match was identified in any of the queried resources at the time of analysis. All databases were accessed in July 2026.

The corresponding Binary Alignment/Map (BAM) file alignments, visualized with IGV, are provided in Figure 1.

Statistical Analysis

This study is descriptive; no statistical analysis was performed because the dataset did not require comparative or inferential evaluation.

Results

Patients

Case 1: A 51-year-old man was evaluated for FAP. He had undergone total proctocolectomy with permanent ileostomy in 2008 because of extensive colorectal polyposis. His family history was remarkable for FAP, affecting his father, siblings, and nephews. During follow-up, he was diagnosed with primary lung adenocarcinoma. Although definitive concurrent chemoradiotherapy was recommended, he declined this treatment and discontinued therapy after receiving five cycles

of cisplatin plus pemetrexed. Approximately two months after discontinuation of treatment, he presented with a brain metastasis and underwent metastasectomy, after which second-line immunotherapy was planned.

Molecular genetic analysis identified a heterozygous APC variant, NM_000038.6: c.1621del (p.Gln541SerfsTer8) in exon 13 (385x, 45% variant allele fraction). This frameshift variant introduces a premature termination codon, resulting in truncation of the APC protein, and has not been previously reported in the literature. According to the ACMG/AMP variant interpretation guidelines, this variant fulfilled the PVS1, PM2, and PP4 criteria and was therefore classified as likely pathogenic. The patient's phenotype, including extensive colorectal polyposis requiring proctocolectomy and a strong family history, was highly consistent with APC-associated FAP.

Case 2: A 44-year-old patient was referred for genetic evaluation after diagnosis of CRC at age 42. Histopathological examination revealed a well-differentiated adenocarcinoma (pT3N0M0) together with multiple tubular adenomas in the setting of colorectal polyposis. The patient subsequently underwent panproctocolectomy with permanent ileostomy.

The family history was notable for colorectal polyposis in the patient's mother, who was undergoing regular surveillance and had not required colectomy because of a relatively limited polyp burden. In addition, the patient's maternal aunt had a history of colorectal polyposis and had undergone a partial colectomy. Molecular confirmation was not available for either relative.

Follow-up abdominal imaging demonstrated hepatomegaly, a left adrenal nodular lesion, mesenteric panniculitis, parastomal herniation, and right renal malrotation. A soft-tissue lesion measuring 24×46 mm was identified within the left rectus abdominis muscle and initially considered suggestive of a desmoid tumour. However, histopathological examination revealed chronic inflammatory changes with fat necrosis, foreign body-type granulomatous inflammation, haemorrhage, congestion, and fibromuscular and adipose tissue fragments, with no evidence of a desmoid tumour.

Molecular genetic analysis identified a heterozygous novel APC variant, NM_000038.6:c.3804_3808del (p.Ile1269PhefsTer5), in exon 16 (916× depth, 46% variant allele fraction). This deletion results in a frameshift and introduces a premature termination codon, which is predicted to result in loss of normal APC protein function. According to the ACMG/AMP variant interpretation guidelines, the variant fulfilled the PVS1_Strong, PM2, and PP4 criteria and was therefore classified as likely pathogenic.

Case 3: A 39-year-old woman with a clinical diagnosis of FAP was referred for genetic evaluation. She was initially diagnosed at age 25 and subsequently underwent total colectomy because of extensive colorectal polyposis. During follow-up, numerous gastric and duodenal polyps were identified, and further surgical intervention was planned because of recurrent polyposis.

Her family history was strongly suggestive of hereditary polyposis. Her father died of CRC at the age of 45 years. One of her sisters died of CRC at 25 years of age. Another sister was diagnosed with FAP at age 25 and underwent total colectomy with ileostomy. However, molecular genetic data were not available for any of the affected family members.

Molecular genetic analysis revealed a heterozygous novel APC variant, NM_000038.6:c.2194_2195del (p.Asn732Ter), in exon 16 (471× depth, 43% variant allele fraction). This deletion introduces a premature termination codon and is predicted to result in the loss of normal APC protein function. According to the ACMG/AMP variant interpretation guidelines, the variant fulfilled the PVS1_Strong, PM2, and PP4 criteria and was therefore classified as likely pathogenic.

Case 4. A 27-year-old woman was referred for HCS testing because of a remarkable family history of early-onset CRC. She had a personal history of endometrioid intraepithelial neoplasia (EIN).

A colonoscopic examination performed at 27 years of age revealed dozens of polyps measuring 3-5 mm throughout the colon. Three polyps from the ascending colon, three from the transverse colon, three from the descending colon, two from the sigmoid colon, and one from the rectum were removed endoscopically. Histopathological examination demonstrated that these lesions were tubular adenomas. First-degree internal haemorrhoids were also identified.

Upper gastrointestinal endoscopy revealed findings consistent with pangastritis and duodenitis. Biopsy specimens were obtained from the antrum and duodenum, and Helicobacter pylori infection was detected. Thyroid ultrasonography was unremarkable. Magnetic resonance imaging of the brain demonstrated findings suggestive of idiopathic intracranial hypertension, a probable arachnoid cyst, and a small vascular lesion.

Her family history was notable for early-onset CRC. Her mother died of rectal cancer at age 34, and her maternal grandfather died of CRC at approximately age 50. In addition, her maternal aunt had undergone a colectomy for polyposis, and her maternal great-aunt had a history of breast cancer. There was no known consanguinity between her parents.

Molecular genetic analysis identified a heterozygous APC variant, NM_000038.6:c.4066_4067dup (p.Gly1357GlnfsTer59), in exon 16 (565× depth, 45% variant allele fraction). This duplication results in a frameshift and introduces a premature termination codon, leading to loss of normal APC protein function. According to the ACMG/AMP variant interpretation guidelines, the variant fulfilled the PVS1_Strong, PM2, and PP4 criteria and was therefore classified as likely pathogenic.

Based on the clinical findings, family histories, and molecular results, the identified variants were responsible for the observed disease phenotypes. Genetic counselling and cascade testing were recommended for at-risk family members.

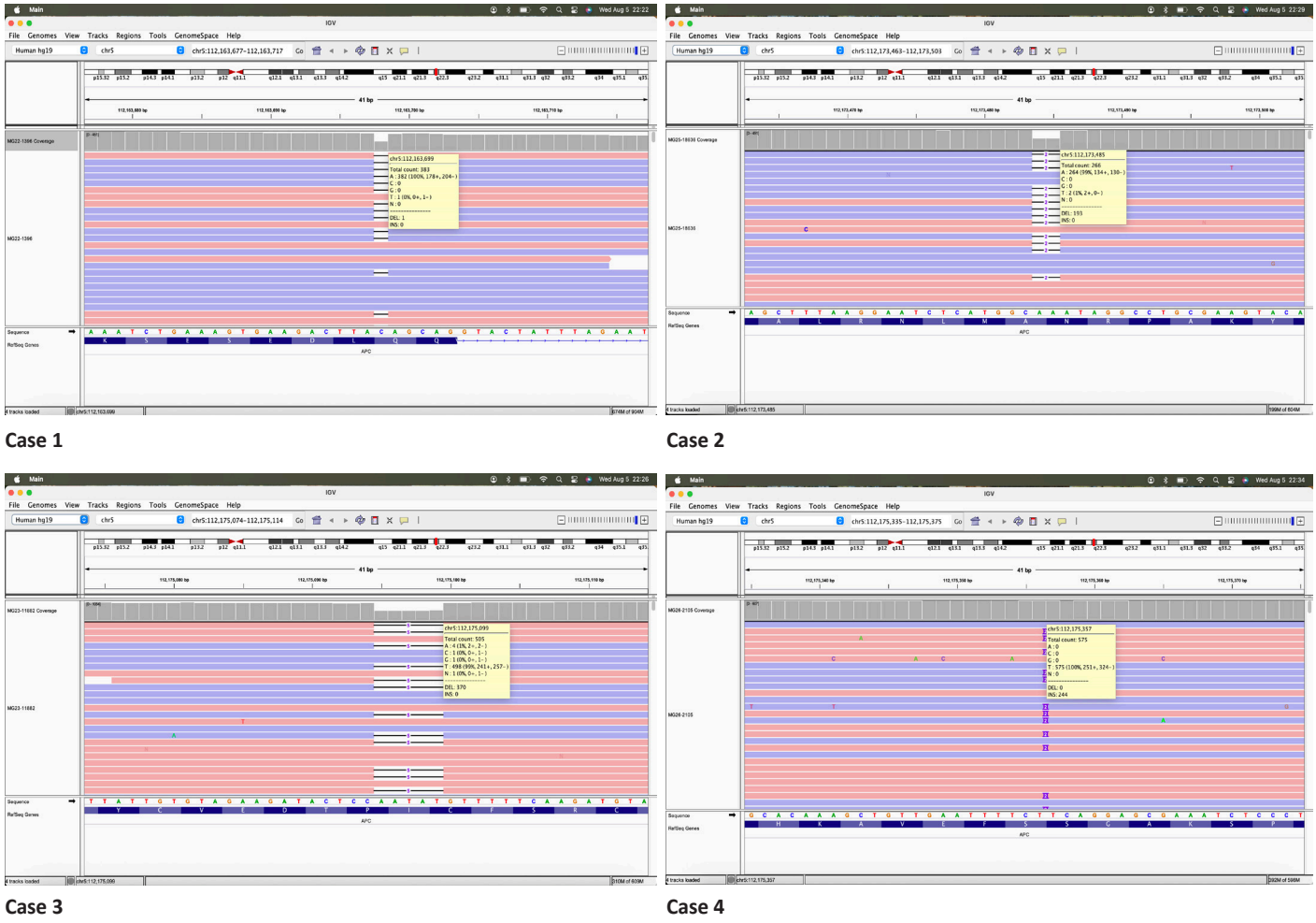

Figure 1. IGV alignments of the identified APC variants. The panels show the genomic location of each variant together with the corresponding sequencing coverage and VAF. The position of the variant is indicated by the blue arrow
IGV: Integrative genomics viewer, VAF: Variant allele fraction

The clinical characteristics, family histories, and molecular findings of the four unrelated patients carrying novel APC variants are summarized in Table 1.

Discussion

FAP is the most common hereditary adenomatous polyposis syndrome and is predominantly caused by germline loss-of-function variants in the *APC* gene. Although the mutational spectrum of APC continues to expand with the identification of previously unreported variants, characterization of these novel variants remains essential for accurate variant interpretation, genetic counseling, risk assessment, and clinical management. Pathogenic APC variants are predominantly protein-truncating alterations, with frameshift and nonsense variants accounting for the majority of reported disease-causing variants, whereas missense variants are encountered less frequently [7]. According to the Leiden Open Variation Database, the most frequently reported APC variants worldwide, c.3927_3931del (p.Glu1309Aspfs) and c.3183_3187del (p.Gln1062Ter), are also truncating variants, further emphasizing that loss of APC function represents the principal molecular mechanism underlying FAP (LOVD, https://www.lovd.nl/). Consistent with this distribution, all four APC variants identified in our cohort were predicted to result in loss of function. However, their molecular consequences are not expected to be identical. The c.1621del (p.Gln541SerfsTer8) variant is predicted to undergo nonsense-mediated mRNA decay (NMD), whereas the remaining three variants, located within the terminal coding exon, are predicted to escape NMD and give rise to truncated protein products. Accordingly, the variants fulfilled either the PVS1 or PVS1_Strong criteria and were classified as likely pathogenic according to the ACMG/AMP guidelines.

Previous studies have consistently associated pathogenic variants located within the APC MCR (MCR; codons 1286-1513) with a more severe polyposis phenotype and an earlier age at disease onset [8]. Among the four novel variants identified in our cohort, only p.Gly1357GlnfsTer59 was located within the MCR (Figure 2). Although this patient has not yet developed a classical FAP phenotype, her relatively young age may preclude full phenotypic expression. Therefore, close

Table 1. Clinical characteristics and molecular findings of the four patients carrying novel APC variants

Case	Age/ sex	Clinical features	Age at diagnosis	Extracolonic manifestations associated with FAP	Family history	APC variant (NM_000038.6)	Variant type	Exon	ACMG evidence	Classification
1	51/M	Classical FAP, extensive colorectal polyposis, total proctocolectomy, lung adenocarcinoma	~30y	-	Father, siblings, and nephews with FAP	c.1621del (p.Gln541SerfsTer8)	PTV	13	**PVS1, PM2, PP4**	Likely pathogenic
2	44/F	FAP, total colectomy	24y	-	Non-extensive colonic polyps in the mother and colectomy for polyposis in the maternal aunt	c.3804_3808del (p.Ile1269PhefsTer5)	PTV	16	**PVS1_ Strong, PM2, PP4**	Likely pathogenic
3	39/F	FAP, prophylactic colorectal colectomy, recurrent polyposis	25y	-	Father and two sisters with FAP; father and one sister died of colorectal cancer	c.2194_2195del (p.Asn732Ter)	PTV	16	**PVS1_ Strong, PM2, PP4**	Likely pathogenic
4	27/F	FAP, colonic tubuler adenoma, EIN, family history of early-onset rectal cancer	27y	-	Mother died of rectal cancer at 34 years; maternal great-aunt with breast cancer	c.4066_4067dup (p.Gly1357GlnfsTer59)	PTV	16	**PVS1_ Strong, PM2, PP4**	Likely pathogenic

The table summarizes the demographic characteristics, major clinical manifestations, family history, molecular findings, variant type, exon location according to the RefSeq transcript NM_000038.6, and ACMG/AMP classification of the novel APC variants identified in this study
FAP: Familial adenomatous polyposis; PTV: Predicted truncating variant, EIN: Endometrioid intraepithelial neoplasia, ACMG: American College of Medical Genetics and Genomics, AMP: Association for Molecular Pathology

endoscopic surveillance and long-term clinical follow-up have been recommended. In contrast, three of the four protein-truncating variants identified in our cohort were located outside the MCR.

Data on the molecular spectrum of APC variants in the Turkish population remain limited. Erdem and Bahsi [9]. reported three missense APC variants, all of which were classified as variants of uncertain significance. In contrast, Ceylan and Ceylan [10]. identified seven pathogenic protein-truncating APC variants, including three nonsense and four frameshift alterations, the majority of which were located in exon 16. Consistent with these observations, three of the four novel variants identified in our cohort were in exon 16. However, given the limited sample size, these findings should not be interpreted as representative of the variant distribution in the Turkish population.

Case 4 presented with EIN. Current evidence does not support a definitive association between APC-associated polyposis and gynecological malignancies, although rare cases have been reported in the literature [11,12]. Therefore, in our patient, the coexistence of EIN and an APC pathogenic variant was considered an uncertain finding rather than an established disease manifestation. Although Case 1 developed lung adenocarcinoma during follow-up, lung cancer is not currently regarded as part of the established extracolonic tumor spectrum of APC-associated polyposis. Given the patient's substantial smoking history, the finding was considered more likely to represent an independent, coexisting malignancy.

Taken together, these findings expand the molecular spectrum of APC-associated polyposis and further emphasize the importance of integrating molecular findings with clinical evaluation and longitudinal follow-up. Larger multicenter studies will be necessary to improve our understanding of genotype-phenotype relationships in APC-associated disease.

Study Limitations

The present study has several limitations. First, this was a small, single-center case series comprising only four unrelated patients; therefore, our findings should not be considered representative of the Turkish population. In addition, functional studies and comprehensive segregation analyses were not available for all cases. In some families, segregation analyses could not be completed because potentially informative relatives were deceased, declined genetic testing, or were unavailable for follow-up. Orthogonal validation by Sanger sequencing was not routinely performed, although all variants were supported by high sequencing depth, appropriate variant allele fractions, and manual IGV review. Larger multicenter studies are needed to further define the molecular spectrum of APC-associated polyposis and to clarify genotype-phenotype relationships.

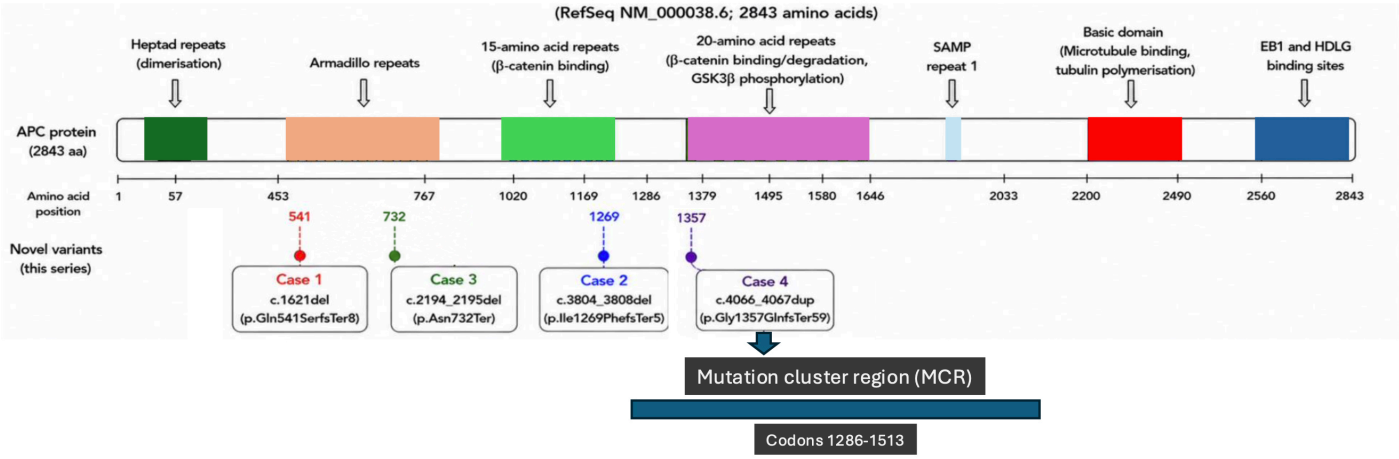

Figure 2. Schematic representation of the APC protein domains and the localization of the novel variants identified in this study. The protein structure is shown according to the RefSeq transcript NM_000038.6 and the RefSeq protein NP_000029.2. The APC protein consists of 2,843 amino acids and contains multiple functional domains, including the N-terminal heptad repeats, armadillo repeats, 15-amino acid β-catenin-binding repeats, 20-amino acid β-catenin-binding/degradation repeats, the SAMP repeat, the basic microtubule-binding domain, and the EB1/HDLG-binding region. The positions of the four novel APC variants identified in this study are indicated according to their amino acid coordinates. The MCR (codons 1286-1513), which has been associated with a more severe polyposis phenotype, is highlighted. The domain organization shown in this figure was inspired by the schematic representation reported by Zhang et al. [13]
Aa: Amino acid, MCR: Mutation cluster region, SAMP: Serine-alanine-methionine-proline repeat, GSK3β: Glycogen synthase kinase 3 beta, EB1: End-binding protein 1, HDLG: Human discs large homolog

Conclusions

This study describes four previously unreported APC variants identified in Turkish patients with APC-associated polyposis. All variants were predicted to result in loss of APC function and classified as likely pathogenic according to the ACMG/AMP guidelines, thereby expanding the mutational spectrum of APC. Overall, our findings provide additional data that may contribute to future variant interpretation studies. Further multicenter studies are needed to improve our understanding of the molecular spectrum of APC-associated polyposis.

Ethics

Ethics Committee Approval: This has been approved by the Ethics Committee of the University of Health Science Türkiye, Ankara Etlik City Hospital (approval no: AEŞH-BADEK-2024-845, date: 25.09.2024).

Informed Consent: Written informed consent was obtained from all participants or their legal guardians before genetic testing.

Footnotes

Authorship Contributions

Surgical and Medical Practices: L.D., Concept: E.T., L.D., Design: E.T., Data Collection or Processing: E.T., Analysis or Interpretation: E.T., L.D., Literature Search: E.T., Writing: E.T.

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Declaration Regarding the Use of AI and AI-Assisted Technologies: During manuscript preparation, the authors used OpenAI's ChatGPT (GPT-5, San Francisco, CA, USA) as a language-support tool to improve the clarity, grammar, and fluency of the text. The tool was not used for data analysis, figure preparation, or the generation of scientific content. All outputs produced by the AI tool were carefully reviewed, edited, and verified by the authors. The authors take full responsibility for the integrity and accuracy of the entire manuscript.

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