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CASE REPORT

Familial adenomatous polyposis: Case report in two sisters at a university hospital in Rio de Janeiro

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Abstract

Familial Adenomatous Polyposis (FAP) is an autosomal dominant disease caused by a mutation in the Adenomatous Polyposis Coli (APC) gene. Its main symptoms include hematochezia, altered bowel habits, and abdominal pain. Diagnosis is based on the presence of multiple colonic adenomas (typically >100 in the classic form), a suggestive family history, and genetic confirmation. Proctocolectomy is the treatment of choice for patients with extensive involvement to reduce the risk of colorectal cancer, but the timing of the procedure should be individualized. We report the case of two sisters, aged 8 and 11, presenting with epigastric pain but no bleeding. Their maternal family history was positive for FAP, with multiple cases across three generations (great-grandmother, grandmother, and mother). A hereditary colorectal cancer panel was performed, identifying a heterozygous variant in the APC gene. Colonoscopy revealed macroscopic findings of sessile colonic polyps (0-Is), and polypectomy was performed. Histopathological examination revealed tubular adenoma with low-grade dysplasia, except for one hyperplastic polyp in the transverse colon of the second sister. FAP is a familial disease with specific symptoms, and family history is crucial for diagnostic screening. Colonoscopy is mandatory, with multiple biopsies following polypectomy. Genetic testing should also be performed for diagnosis. Most patients with FAP require prophylactic colectomy to prevent progression to colorectal cancer.

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INTRODUCTION

Familial adenomatous polyposis (FAP), previously known as Gardner syndrome, is part of a large group of hereditary polyposis syndromes[1]. It is a hereditary condition characterized by the early onset of hundreds to thousands of adenomas distributed throughout the entire large intestine[2], usually beginning in the second decade of life[3].

Considered the most common genetic syndrome associated with colorectal cancer during childhood, FAP plays a highly significant role in the pediatric setting[4]. Among the common pediatric polyposis syndromes, it carries the highest risk of colorectal cancer (CRC)[1], accounting for approximately 0.5% to 1% of all cases[5]. The relevance of FAP is evident, as colorectal cancer is responsible for approximately 50,000 deaths annually in the United States alone[4] and is considered the third leading cause of cancer in the country[6]. Early detection of FAP, particularly during childhood and adolescence, is crucial for preventing this type of cancer.

This autosomal dominant disease is caused by a germline mutation in the adenomatous polyposis coli (APC) gene, located on chromosome 5q21. APC is a tumor suppressor gene whose primary function is to encode a structural protein involved in regulating cell adhesion and migration. Inactivation of APC results in loss of control over cell growth and represents the first step in the development of CRC[2].

The incidence of FAP ranges approximately from 1 in 7,000 to 1 in 30,000 births[7]. Based on recent available data, its prevalence ranges from 2.29 to 3.2 cases per 100,000 individuals[5]. It affects both sexes equally, with a higher incidence between late adolescence and the early twenties[3], and exhibits variable expression[7].

Regarding clinical manifestations, patients with a large number of polyps may be asymptomatic[4]. However, symptoms such as bleeding, diarrhea, and abdominal pain are common[8], making clinical surveillance essential for early detection of complications.

The diagnosis is confirmed by the presence of a pathogenic heterozygous variant in the APC gene, along with one of the following criteria: (a) ≥100 adenomatous polyps in the colon or rectum (which may be fewer in younger patients or those who have undergone colectomy); or (b) <100 adenomatous polyps associated with a confirmed family history of FAP[5].

Our objective is to report the case of two sisters diagnosed with FAP who had a family history of polyposis, illustrating the importance of histopathological evaluation of polyps and the hereditary colorectal cancer panel.

CASE REPORT

Patient 1, aged 8 years, and Patient 2, aged 11 years, both with a significant family history of intestinal polyposis and colorectal cancer, were referred to the pediatric gastroenterology outpatient clinic due to gastrointestinal complaints, including epigastric pain and urgency to defecate associated with straining and anal pain. Both reported daily bowel movements, three times per day, Bristol type 4, without blood or mucus. Neither patient presented with intestinal bleeding or other suggestive clinical signs.

Both patients had a positive family history of familial adenomatous polyposis, with maternal great-grandmother, grandmother, and mother affected by the condition, all of whom died from colorectal cancer. Both denied continuous medication use, previous hospitalizations, allergies, or comorbidities, and their vaccination schedules were up to date. Physical examination of both sisters was unremarkable.

For diagnostic clarification, colonoscopy was initially performed, revealing sessile colonic polyps, Paris classification 0-Is (Figures 1 and 2). Polypectomy and biopsy were



Figure 1. Colonoscopy showing sessile polyps, Paris classification 0-Is.

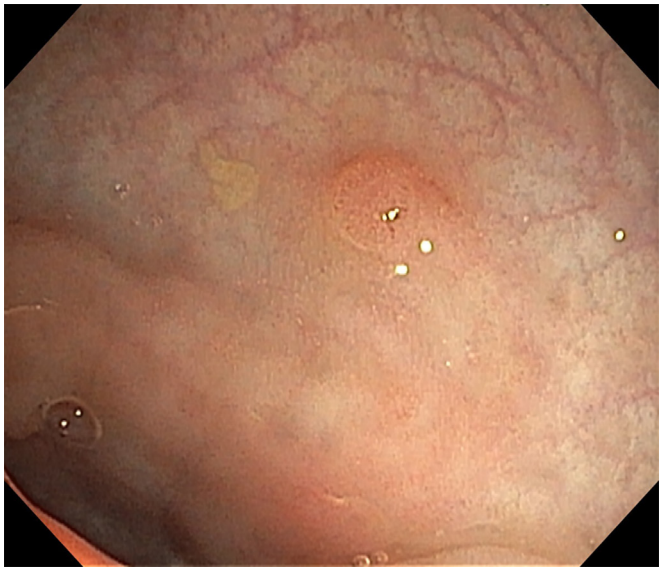


Figure 2. Colonoscopy showing sessile polyps, Paris classification 0-Is.

subsequently performed. Histopathological examination demonstrated tubular adenomas with low-grade dysplasia, except for one hyperplastic polyp in the transverse colon of Patient 2. The hereditary colorectal cancer panel in both patients identified the heterozygous variant chr5:112,838,220C>T in the APC gene, resulting in substitution of arginine at position 876 by a premature stop codon.

During follow-up, colonoscopy in both patients continued to demonstrate sessile colonic polyps covered by erythematous mucosa. Histopathological analysis revealed tubular adenomas with low-grade dysplasia. Continuous clinical and endoscopic surveillance was recommended due to the high risk of progression to colorectal cancer.

DISCUSSION

Familial adenomatous polyposis (FAP) is a disease characterized by a mutation in the APC gene, resulting in decreased intracellular degradation of beta-catenin and activation of a pathway that regulates cell proliferation[6]. This syndrome requires a comprehensive approach, particularly in pediatric patients, as illustrated by the cases described.

Disease severity is predicted by the location of the mutation within the APC gene[9]. Mutations in codons 1250 to 1464, particularly at codon 1309, are associated with more severe gastrointestinal disease; mutations in the C-terminal domain may result in a milder form of the disease[1]. For example, mutations at the 3′ and 5′ ends of the APC gene are associated with a milder polyp phenotype and later progression to colorectal cancer (CRC)[4]. In this particular case, we describe a chr5:112,838,220C>T mutation in the APC gene, resulting in the substitution of arginine at position 876 by a premature stop codon. This position (876) is not directly within the codon ranges described above. Nevertheless, the mutation identified in the patients is considered pathogenic and carries a high risk for CRC.

It is important to note that most individuals affected by FAP have a family history of the syndrome; however, a significant subset (approximately 20% to 30%) of cases results from de novo mutations[7]. In this case, the fact that close family members presented with colorectal cancer associated with FAP reinforces the need for surveillance in patients with a positive family history.

The clinical manifestations of FAP depend on age, genotype, and sex, and it is noteworthy that patients with a large number of polyps may be asymptomatic[4]. Classic symptoms include bleeding, diarrhea, and abdominal pain, usually diagnosed between 20 and 25 years of age[8]. Painless bright red blood per rectum is commonly observed[4]. It is noteworthy that neither patient presented with intestinal bleeding as a clinical manifestation; however, both reported epigastric pain, which is consistent with clinical manifestations described in the literature.

In addition to numerous adenomatous polyps in the gastrointestinal mucosa, FAP also presents extraintestinal manifestations involving multiple organs[7]. During outpatient follow-up, neither patient developed any of these manifestations, which include mandibular and cranial osteomas, supernumerary and impacted teeth, retinal pigment epithelium hamartomas, cutaneous fibromas, epidermoid cysts, and lipomas[1]. Moreover, there is an increased lifetime risk of various tumors that may occur in the duodenum, pancreas, thyroid, and brain[9].

Surveillance of FAP with colonoscopy should begin between 10 and 12 years of age[1]. However, in the presence of symptoms such as hematochezia, a family history of aggressive disease, or an aggressive genetic mutation, earlier screening may be required[4]. Currently, most patients are identified through genetic testing and detection of the specific familial variant at a young age due to a positive family history, allowing diagnosis between 10 and 20 years of age[10]. The importance of surveillance adopted in the patients described, at 8 and 11 years of age, reinforces the most relevant measure to prevent the development of colorectal cancer. In addition, the presence of any extracolonic findings should indicate the need for genetic counseling and genetic testing[1].

On the other hand, it is important to emphasize that the Paris classification is a polyp classification system that allows evaluation of lesion location, size, morphology, granularity, and surface crypt or microvascular patterns. It is recommended to help stratify which lesions are more likely to contain advanced pathology, that is, to differentiate between cancers and premalignant polyps, and to guide treatment strategy[11].

According to this system, lesions are initially divided into polypoid (0-I) or non-polypoid (0-II and 0-III) subtypes. Type 0-I is subdivided into 0-Ip (pedunculated), 0-Is (sessile), and 0-Isp (subpedunculated). Type 0-II is subdivided into 0-IIa (slightly elevated), 0-IIb (flat), and 0-IIc (slightly depressed). Excavated or ulcerated lesions constitute a third category, designated as type 0-III[11]. In this case report, the colonoscopic findings described a sessile polyp. Therefore, the next essential step is to review the histopathological report, which is necessary to define risk and guide follow-up planning.

FAP requires aggressive management to prevent cancer and other complications[1]. Currently, there are no standardized guidelines regarding the type and exact timing of surgery, which should consider genotyping results in conjunction with clinical data[8,5]. Due to the near 100% risk of developing CRC without early treatment, prophylactic colorectal surgery is indicated at a young age (around 15 years)[4,10]. When the polyp burden exceeds the capacity for endoscopic removal, when polyp size is greater than 10 mm, or when polyps with high-grade dysplasia are present, total colectomy is recommended[1,5].

REFERENCES

1. MacFarland SP, Zelley K, Katona BW, Wilkins BJ, Brodeur GM, Mamula P. Gastrointestinal polyposis in pediatric patients. J Pediatr Gastroenterol Nutr. 2019;69(3):273-80. DOI: https://doi.org/10.1097/MPG.0000000000002421.
2. Galiatsatos P, Foulkes WD. Familial adenomatous polyposis. Am J Gastroenterol. 2006;101(2):385-98. DOI: https://doi.org/10.1111/j.1572-0241.2006.00375.x.
3. Half E, Bercovich D, Rozen P. Familial adenomatous polyposis. Orphanet J Rare Dis. 2009;4(22):1-22. DOI: https://doi.org/10.1186/1750-1172-4-22.
4. Septer S, Lawson CE, Anant S, Attard T. Familial adenomatous polyposis in pediatrics: natural history, emerging surveillance and management protocols, chemopreventive strategies, and areas of ongoing debate. Familial Cancer. 2016;15(4):549-59. DOI: https://doi.org/10.1007/s10689-016-9905-5.
5. Kyriakidis F, Kogias D, Venou TM, Karlafti E, Paramythiotis D. Updated perspectives on the diagnosis and management of familial adenomatous polyposis. Appl Clin Genet. 2023;16:139-53. DOI: https://doi.org/10.2147/TACG.S372241.
6. Attard TM, Septer S, Lawson CE, Attard MI, Lee STM, Umar S. Microbiome insights into pediatric familial adenomatous polyposis. Orphanet J Rare Dis. 2022;17(1):416. DOI: https://doi.org/10.1186/s13023-022-02569-2.
7. Dinarvand P, Davaro EP, Doan JV, Ising ME, Evans NR, Phillips NJ, et al. Familial Adenomatous Polyposis Syndrome: An Update and Review of Extraintestinal Manifestations. Arch Pathol Lab Med. 2019;143(11):1382-98. DOI: https://doi.org/10.5858/arpa.2018-0570-RA.
8. Cerasuolo A, Miele E, Russo M, Aversano A, Cammarota F, Duraturo F, et al. Sporadic pediatric severe familial adenomatous polyposis: A case report. Mol Clin Oncol. 2020;13(1):20. DOI: https://doi.org/10.3892/mco.2020.2090.
9. Herzig D, Hardimann K, Weiser M, Yu N, Paquette I, Feingold DL, et al. Management of inherited polyposis syndromes. Dis Colon Rectum. 2017;60(9):881-94. DOI: https://doi.org/10.1097/DCR.0000000000000912.
10. Aelvoet AS, Buttitta F, Ricciardiello L, Dekker E. Management of familial adenomatous polyposis and MUTYH-associated polyposis; new insights. Best Pract Res Clin Gastroenterol. 2022;58-59:101793. DOI: https://doi.org/10.1016/j.bpg.2022.101793.
11. Johnson GGRJ, Helewa R, Moffatt DC, Coneys JG, Park J, Hyun E. Colorectal polyp classification and management of complex polyps for surgeon endoscopists. Can J Surg. 2023;66(5):491-8. DOI: https://doi.org/10.1503/cjs.011422.