Pediatric rectosigmoid atypical juvenile polyps presenting with anal prolapse and acute bleeding: a case report and a comprehensive review of the literature.

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Case Report

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Abstract

Rectosigmoid solitary juvenile polyps are benign lesions, relatively frequent in childhood. The clinical debut of a pediatric polyp with bleeding is relatively frequent, but there are very few reports of anal prolapse of polyps. We present the case of a 7-year-old female patient with no previous history who presented with anal prolapse of a polyp with acute bleeding. An urgent endoscopic examination was performed and two rectosigmoid polypoid lesions were found and resected. The anatomopathological study showed that these were two hamartomatous polyps with mild dysplasia. The patient is asymptomatic and is being followed up. The existing reports in the medical literature of this entity are scarce. In a pediatric patient with an anal prolapse, this entity should be considered in the differential diagnosis.

Introduction

The presence of colonic polyps in the pediatric age group is relatively frequent. Although the population incidence is unknown, recent reports establish the presence of polyps in up to 6% of pediatric colonoscopies (12% in the case of colonoscopies performed for lower gastrointestinal bleeding) [1]. However, there is significant variability between series, with other authors reporting up to 75% incidence [2]. Most polyps are rectosigmoid in location and are usually asymptomatic or present with lower gastrointestinal bleeding (acute or chronic) that can lead to anemia and hemodynamic instability [3]. There are, however, other atypical and infrequent clinical presentations, such as anal prolapse of the polyp, which can be confused with a mucosal rectal prolapse and can lead to self-amputation of the polyp. Although accepted as one of the classic presentations of pediatric rectosigmoid polyp, the literature regarding pediatric polyp anal prolapse is very limited [4-11]. From the histopathological point of view, the vast majority of them correspond to juvenile polyps (also known as retention polyps or juvenile hamartomatous polyps). Nevertheless, sometimes other histological subtypes such as hamartomatous polyps or tubular adenomas are identified. In this last case, low degree dysplasia has been described, although not neoplasms as such [12]. The most frequent presentation is a solitary polyp, although sometimes they may occur in the context of polyposis syndromes, as in the case of juvenile polyposis syndrome (SPJ), PTEN hamartoma tumor syndrome, Peutz-Jeghers syndrome, or familial adenomatous polyposis [13,14].

Material and methods

Clinical data, sociodemographic information, and histological samples of a patient with a confirmed diagnosis of two rectosigmoid atypical juvenile polyps treated at our center in 2023 were collected. All the information was anonymized following current legislation. Informed written and verbal consent for publication was obtained from the patient’s parents. A search was performed in the main medical bibliographic databases with the following Booleans: "(pediatric OR children) AND (polyp OR mass OR tumor) AND (rectal OR anal) AND prolapse OR bleeding)". The search included the following databases: OVID, Web of Science, Pubmed, Medline, and Scopus. The search was last executed in August 2023.

Results

We present the case of a 7-year-old female patient with no medical history of relevance, who came to the Emergency Department for anal prolapse of a painful bleeding mass. As the only relevant event, the parents reported a self-limited episode of frank lower gastrointestinal bleeding in the month before the episode. The patient was clinically and hemodynamically stable, and the analytical study showed a discrete leukocytosis without elevation of acute phase reactants and without anemization. Physical examination revealed a polypoid violaceous mucosal mass prolapsing through the anal canal (Figure 1, above left). Suspecting a prolapsed polyp through the anus, it was decided to perform a surgical exploration under general anesthesia. During the anesthetic induction maneuvers the tumor presented a spontaneous reduction. A rectal examination was performed, with multiple clots and traces of fresh blood. No lesion was palpated. An incomplete colonoscopy was then performed, showing two polypoid pedunculated lesions at approximately 25 cm from the external anal margin. The first one (which presented active bleeding) was resected with a polypectomy loop and diathermy without
incident. The second one, measuring 3 cm (major axis) (Figure 1, above right), could not be resected by this technique and it was decided to externalize it through the anal canal and ligate it with a transfixion at its base (Figure 1, bottom). The transverse colon and ascending colon could not be explored because of the hematic and fecal debris present. The patient had a favorable postoperative course, with no rebleeding, and was discharged 12 hours after the procedure. Histological study of the specimen showed fragments of lobular morphology with glandular epithelial proliferation of irregular contours, partially serrated and with superficial pseudostratification (Figure 2, left). The immunohistochemical study showed nuclear negativity for PTEN in the intestinal epithelium, with diffuse cytoplasmic staining (a pathologic finding) (Figure 2, above right). The immunohistochemical study for DPC4 (SMAD4) (Figure 2, bottom right), MLH-1, MSH2, MSH6 and PMS2 was normal. These findings were compatible with the diagnosis of hamartomatous polyps with mild dysplasia. The patient is currently asymptomatic and under study.

**Discussion**

The literature concerning anal prolapse of polyps in pediatric population is scarce. First, we found 8 adequately documented clinical reports [4-11]. Reported ages of presentation ranged from 2 days to 17 years. Four patients were male and four were female. All patients presented with lower gastrointestinal bleeding and a prolapsed anal mass. Additionally, one presented with spontaneous amputation of the polyp, and two presented with anaemia. All polyps were located in the rectosigmoid region. Four were reported as juvenile polyps, one as an inflammatory polyp, one as an inflammatory pseudopolyp, one as hyperplastic crypts covered with calcified granulation tissue in the context of cap polyposis, and in one case two adenomatous polyps were found. The clinical evolution was favorable, and in only 1 case there was a small recurrence that was resolved with medical management. One case did not report the clinical evolution of the patient. Finally, and trying to be rigorous and exhaustive, we have included the case of a 15-year-old patient with Marshall-Smith syndrome who presented with a polypoid lesion covered by rectal mucosa at the level of the anal margin and who was diagnosed as a cloacogenic polyp. However, we believe that this lesion does not strictly belong to the pathophysiologic spectrum presented here. The main characteristics of these studies are shown in Table 1.

Second, there are some published series of patients in the literature in which explicit reference is made to patients with colonic polyps who have debuted with this clinical presentation [15-21]. Regarding these series it should be considered the literature is mostly based on clinical descriptions provided by the patients, and there is greater heterogeneity. Poddar et al. (1998) reported a series of 236 pediatric patients with polyps and documented 30 cases of anal prolapse. Ukarapol et al. (2007) reported a series of 32 patients with 15 of them (47%) presenting anal prolapse, with these patients also presenting a greater association with the presence of polyposis coli. Haghi Ashtiani et al. (2008) reported a 563 pediatric patients series with colorectal polyps with 7 cases of polyp anal prolapse (1.3%). Akkoyun et al. (2011) conducted a pilot study to validate photography and video as diagnostic tools in the context of pediatric anal lesions and in their series reported anal prolapse of 3 polyps that were subsequently confirmed endoscopically although authors did not report histomorphological findings. In 2014, Lei et al. Reported a series of 322 patients with 8 cases of polyp rectal prolapse (2.5%). In 2015, Rathi et al. published a series of 120 pediatric patients with colorectal polyps and reported 14 cases of rectal prolapse (1 in the polyposis syndrome group and 13 in the non-polyposis group). Lastly, Tripathi et al. (2020) [22] presented a series of 240 pediatric patients affected by gastrointestinal polyps with a small percentage debuting with a mucosal rectal prolapse, although they did not specify whether this prolapse corresponded to the polyp per se or not. Interestingly, all reported series are Asian. We hypothesize that this could be due to a higher prevalence of gastrointestinal parasitosis in the reported countries, which is a known risk factor for mucosal rectal prolapse and which following the same pathophysiological principle could lead to the anal prolapse of the polyp. The main characteristics of these studies are shown in Table 2.

In relation to the histologic findings of our case, it is an uncommon polyp because it presents low-grade dysplasia and because it presents a pathologic immunohistochemical study for PTEN and normal for SMAD4. From the histological point of view, we believe that this is a juvenile polyp (also called retention polyps or formerly inflammatory polyps) with atypical features. The presence of a greater epithelial (glandular) than stromal content, as in our patient, has been correlated with a
potential association with juvenile polyposis syndromes. Hamartomatous polyps characteristic of Peutz-Jeghers syndrome, on the other hand, usually present a greater smooth muscle component and greater arborization than in our case. In our case, the immunohistochemical study for desmin showed isolated scarce traces of smooth muscle. Also, we consider it difficult to establish the diagnosis of hamartomatous polyp (Peutz-Jeghers) with a large bowel polyp exclusively. On the other hand, we believe that other entities such as CAP polyposis syndrome, cloacogenic polyps or solitary rectal ulcer are framed within the pathology of the anal canal and should not be included in the differential diagnosis of the lesion presented here [23,24].

As a reflection, we believe that this clinical entity (polypoid anal prolapse) is probably underdiagnosed, probably due to the intermittent nature of the prolapse and the indolent course that most patients usually present. Therefore, we subscribe to the report by Akkoyun et al. and believe that the use of photographs and videos to characterize the lesion by parents is a simple and inexpensive way to improve the diagnostic yield in this pathology.

The main strength of this work is the rigorous and adequately characterized description of the case, accompanied by an extensive and reasoned analysis of the previously existing literature. As a limitation, it should be considered that the literature review has not been systematic and therefore there may be papers that have not been included in it.

In conclusion, although the clinical presentation of a polyp as an anal prolapse is uncommon, it should be considered in the differential diagnosis of anal tumors. This presentation may be somewhat more prevalent in cases of hamartomatous polyps, like ours [25]. Other entities should also be considered in the diagnosis of an anal mucosal mass, such as mucosal rectal prolapse, haemorrhoids or infrequent lesions in this location such as lipomas [26]. Although most lesions correspond to juvenile or retentive polyps, histopathologic confirmation is essential to adequately characterize the lesion and to rule out the presence of dysplasia. Based on current evidence, there does not appear to be an association between this clinical presentation and polyposis syndromes.

Declarations

Conflicts of interest: There is no conflict of interest or external funding to declare. None of the authors have anything to disclose

Contributor declaration page:
Dr. Javier Arredondo Montero and Dr. Socorro Razquin Lizarraga conceptualized and designed the study, collected and analysed the data, drafted the initial manuscript, and revised the manuscript.

Dr. Elena Carracedo Vega, Dr. Mónica Bronte Anaut, Dr. Gina de Lima Piña and Dr. Rosa Guarch Troyas collected and analysed the data and revised the manuscript.

Original work:
All authors of the manuscript declare that this is an original contribution, not previously published.

Data availability:
Upon reasonable request, all data pertaining to this study are available through the author in correspondence.

Ethics statement:
Ethical approval was not requested due to the retrospective and observational nature of this isolated case report and the absence of research human or animal samples. The principles of the Helsinki Declaration (2013) were followed.

Informed consent:
Prior to the submission of this article, verbal and written informed consent was obtained from the legal guardians of the patient reported in this publication.

References


Tables

Table 1. Previous case reports identified in the medical literature.
<table>
<thead>
<tr>
<th>Authors</th>
<th>Patient’s age</th>
<th>Patient’s sex</th>
<th>Patient’s medical history</th>
<th>Clinical presentation</th>
<th>Endoscopic findings</th>
<th>Histology</th>
<th>Clinical outcome</th>
</tr>
</thead>
<tbody>
<tr>
<td>Arredondo et al. (2023) [11]</td>
<td>2 years</td>
<td>Female</td>
<td>-</td>
<td>Lower GI bleeding. Prolapsed anal mass. Self-amputation of the polyp</td>
<td>-</td>
<td>Juvenile polyp</td>
<td>Favorable (no complications)</td>
</tr>
<tr>
<td>Kakiuchi et al. (2023) [10]</td>
<td>5 years</td>
<td>Female</td>
<td>-</td>
<td>Lower GI bleeding. Prolapsed anal mass. Single rectal pedunculated polyp (1 cm)</td>
<td>Juvenile polyp</td>
<td>Favorable (no complications)</td>
<td></td>
</tr>
<tr>
<td>McClanahan et al. (2021) [9]</td>
<td>13 years</td>
<td>Male</td>
<td>-</td>
<td>Lower GI bleeding. Mild microcytic anemia. Prolapsed anal mass. Single sigmoid semi-pedunculated polyp (3.3 cm)</td>
<td>Juvenile polyp</td>
<td>-</td>
<td></td>
</tr>
<tr>
<td>Kim et al. (2017) [8]</td>
<td>11 months</td>
<td>Male</td>
<td>Sacral epidermal nevus</td>
<td>Lower GI bleeding. Prolapsed anal mass. Multiple rectal multilobulated polypoid groups without a stalk (4-6 cm)</td>
<td>Hyperplastic crypts covered with calcified granulation tissue</td>
<td>Hyperplastic polyp (no dysplasia)</td>
<td>Small recurrence after 2 months treated with mesalazine enema</td>
</tr>
<tr>
<td>Kakembo et al. (2016) [7]</td>
<td>15 years</td>
<td>Male</td>
<td>-</td>
<td>Lower GI bleeding. Prolapsed anal mass.</td>
<td>-</td>
<td>Inflammatory polyp (no dysplasia)</td>
<td>3-stage proctocolectomy with J-pouch reconstruction. Favorable</td>
</tr>
<tr>
<td>Washington et al. (1993) [5]</td>
<td>15 years</td>
<td>Male</td>
<td>Marshall-Smith syndrome</td>
<td>1.5 cm polypoid perianal mass, covered with rectal mucosa</td>
<td>-</td>
<td>Inflammatory cloacogenic polyp</td>
<td>Favorable (no complications)</td>
</tr>
</tbody>
</table>

Table 2. Previous case series identified in the medical literature.
<table>
<thead>
<tr>
<th>Authors</th>
<th>Country</th>
<th>Number of patients with polyps in the series</th>
<th>Age of the patients included in the series</th>
<th>Number of patients which reported anal prolapse of a polyp</th>
<th>Other findings</th>
</tr>
</thead>
<tbody>
<tr>
<td>Rathi et al. (2015)[21]</td>
<td>India</td>
<td>120</td>
<td>7.31 (2-19)</td>
<td>14</td>
<td>No significant association between polypoid anal prolapse and presence of polyposis coli (p=0.99).</td>
</tr>
<tr>
<td>Lei et al. (2014)[20]</td>
<td>China</td>
<td>322</td>
<td>8 (9-14)</td>
<td>8 (2.5%)</td>
<td>-</td>
</tr>
<tr>
<td>Akkoyun et al. (2011) [19]</td>
<td>Turkey</td>
<td>23</td>
<td>5.3 (1-11)**</td>
<td>3</td>
<td>Photographic documentation of the lesion allows for an accurate diagnosis by the pediatric surgeon.</td>
</tr>
<tr>
<td>Haghi Ashtiani et al. (2008)</td>
<td>Iran</td>
<td>563</td>
<td>5.66 (2-17)</td>
<td>7 (1.3%)</td>
<td>-</td>
</tr>
<tr>
<td>Ukarapol et al. (2007)[17]</td>
<td>Thailand</td>
<td>32</td>
<td>6.5*</td>
<td>15 (47%)</td>
<td>Significant association between polypoid anal prolapse and presence of polyposis coli (p=0.006).</td>
</tr>
<tr>
<td>Waitayakul et al. (2004)[16]</td>
<td>Thailand</td>
<td>93</td>
<td>5.1*</td>
<td>39.8%</td>
<td>-</td>
</tr>
<tr>
<td>Poddar et al. (1998) [15]</td>
<td>India</td>
<td>236</td>
<td>6.12 (2-12)</td>
<td>30</td>
<td>-</td>
</tr>
</tbody>
</table>

*: mean age; **: mean age (range)

**Figures**
Figure 1

Clinical and endoscopic findings. **Above, left**: the appearance of the lesion during the initial assessment of the patient. There is a nodular, violaceous tumor prolapsing through the anus. Traces of fresh blood are seen in the most caudal segment of the lesion. **Above, right**: Endoscopic view of the lesion. Large friable polypoid friable tumor, located at the level of the rectosigmoid junction. **Below**: Externalization of the lesion through the anus. A thick vascular pedicle and a friable polypoid lesion are seen.
Figure 2

Photomicrographs. **Left**: (H&E). Polypoid lesions of lobular morphology with glandular epithelial proliferation of irregular contours, partially serrated and with superficial pseudostratification. Mild dysplasia is seen. **Above, right**: Immunohistochemical study for PTEN. Nuclear negativity for PTEN in the intestinal epithelium, with diffuse cytoplasmic staining. **Above, bottom**: Immunohistochemical study for SMAD4 (DPC4). Normal immunohistochemical study for SMAD4.