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Degenerated Juvenile Idiopathic Polyposis: A Case Report

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Abstract **Case Report**

Juvenile polyposis syndrome (JPS) is a rare autosomal dominant condition characterized by the development of hamartomatous polyps in the gastrointestinal tract and is associated with a risk of malignant transformation. Therefore, regular clinical, biological, and endoscopic surveillance is crucial. We report a case of rectal cancer (middle and upper rectum) arising from juvenile idiopathic polyposis.

Keywords: Juvenile idiopathic polyposis, colorectal cancer, colonoscopy, surveillance, case report.

INTRODUCTION

Juvenile polyposis syndrome (JPS) is a rare digestive polyposis classified among hamartomatous familial polyposis syndromes with autosomal dominant inheritance. It is characterized by the occurrence of polyps throughout the gastrointestinal tract [1]. Diagnosis relies on personal and family history, the number of polyps, and their histological characteristics. Given the high risk of digestive and extra-digestive malignancies associated with these genetic conditions, multidisciplinary care involving oncogenetics, gastroenterology, pathology, and surgery is required. Endoscopy is essential for assessing lesions, performing polypectomy, and providing histological evaluation, as degeneration, although rare, has been described [2]. We report a case of degenerated juvenile idiopathic polyposis confirmed by endoscopy, histology, and imaging, aiming to describe the clinical, radiological, and therapeutic features to enhance understanding of JPS.

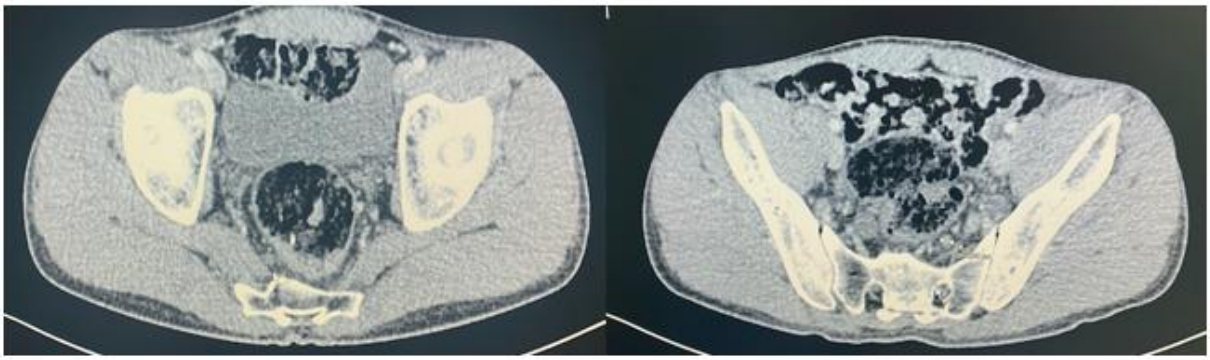
CASE PRESENTATION

A 36-year-old male with a family history of juvenile idiopathic polyposis (his mother, two brothers, and one sister died following malignant transformation of the disease), chronic smoking, and a history of occasional alcohol use (ceased 10 years prior) was referred for evaluation of intermittent atypical periumbilical abdominal pain that had persisted for one year. On examination, the patient appeared pale with general fatigue and unquantified weight loss. He exhibited diffuse periumbilical tenderness, no palpable lymphadenopathy, and a fixed rectal mass located 2 cm from the anal verge. Routine blood tests revealed WBC at 7,060/µL, hemoglobin at 14.3 g/dL, and platelets at 338,000/mm³. Tumor markers were positive, with carcinoembryonic antigen (CEA) 11 times the upper limit of normal and negative CA19-9. Colonoscopy revealed multiple polyps, both sessile and pedunculated, ranging from 1 to 5 cm, along with a tumoral lesion at the anal verge and in the rectum. Biopsy indicated a signet ring cell adenocarcinoma without vascular invasion. Pelvic MRI showed circumferential wall thickening of the lower rectum extending over 39 mm with a wall thickness of 8 mm. Another lesion was observed at the recto-sigmoid junction, 28 mm in height, with a wall thickness of 5 to 9 mm, staged as T2N2Mx. Thoraco-abdominopelvic CT revealed a right posterolateral semi-circumferential mass of the lower and middle rectum, measuring 25 mm in thickness and 77 mm in length, showing heterogeneous enhancement with discrete perirectal fat infiltration. Four mesorectal lymph nodes were detected, the largest measuring 5×8 mm.

(Figure n°1). Two left common iliac nodes measured 25×15 mm and 14×15 mm, and two internal iliac nodes (right and left) measured 24×19 mm and 16×22 mm respectively

(Figure n°2). Multiple polypoid lesions were seen in the left colon, both sessile and pedunculated, with contrast enhancement, staged T3N2M0. During hospitalization, an echocardiography was performed as part of the workup for congenital manifestations and

revealed a partial atrioventricular septal defect with moderate mitral regurgitation due to a mitral cleft. The patient was referred for congenital cardiology follow-up. Concomitant neoadjuvant chemoradiotherapy was proposed.



a **b**

Figure n°1 : Axial abdominopelvic CT scan showing (a) subtle infiltration of the perirectal fat and (b) four mesorectal lymph nodes, the largest measuring 5×8 mm

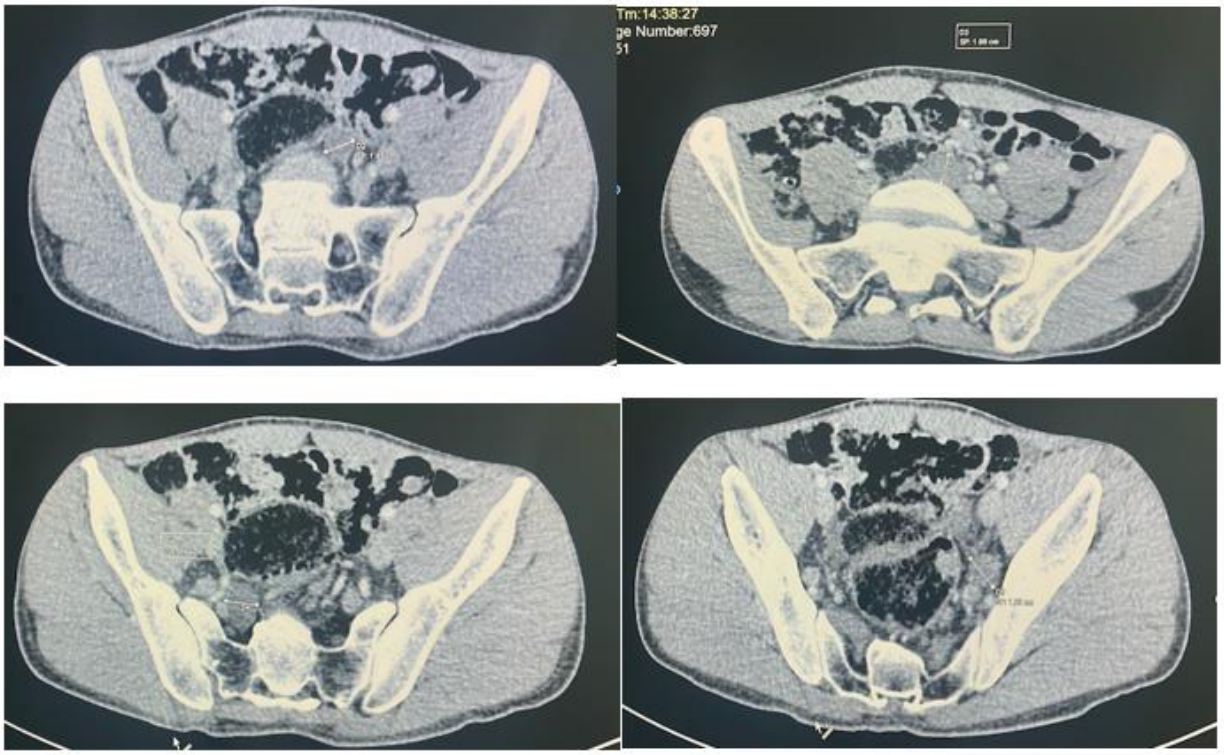


Figure n°2 : Caption: Axial abdominopelvic CT scan showing two left common iliac lymph nodes (25×15 mm and 14×15 mm), and two internal iliac lymph nodes on the right and left (24×19 mm and 16×22 mm, respectively)

DISCUSSION

Juvenile polyposis syndrome is characterized by multiple juvenile polyps in the gastrointestinal tract. The estimated incidence is between 1 in 100,000 and 1 in 160,000 [3]. Polyps may number from one to over 100 and occur throughout the digestive tract, most commonly in the colon and rectum (98%), followed by the stomach (14%), duodenum (7%), jejunum, and ileum (7%) [4]. JPS is inherited in an autosomal dominant fashion, and mutations in SMAD4 and BMPR1A have been identified [5]. Age at diagnosis varies, but symptoms often begin in the first or second decade [6]. Patients are at increased risk for colorectal cancer (17–22%) [7,8], gastric cancer (20–30%) [8,9], and rarely pancreatic or small bowel cancer [10,11]. SMAD4 mutation carriers have a significantly higher incidence of gastric polyposis and gastric cancer than those with BMPR1A mutations [12,13].

The diagnostic criteria for JPS, as revised by Jass *et al*. [14], require at least one of the following: more than five juvenile polyps in the colorectum, multiple juvenile polyps throughout the GI tract, or any number of juvenile polyps with a family history of JPS. Digestive

manifestations include rectal bleeding, melena, atypical abdominal pain, and diarrhea [15]. Extra-digestive anomalies include cardiovascular malformations (telangiectasias, arteriovenous malformations, aneurysms) and neurological anomalies (macrocephaly, hydrocephalus) [16]. Surveillance should begin with annual colonoscopy at age 12, or earlier if symptomatic. Upper endoscopy is recommended every 1–3 years starting at age 12, and the small intestine should be monitored with capsule endoscopy or enteroscopy as needed. Polypectomy is generally sufficient in limited disease. Colectomy with ileorectal anastomosis is indicated in cases of malignancy, high-grade dysplasia, or uncontrolled polyp burden. Rectal surveillance is necessary, and proctocolectomy with ileoanal anastomosis may be required. Gastrectomy may be indicated in cases of advanced dysplasia or massive gastric polyposis. SMAD4 mutation carriers should also undergo annual blood tests and cardiovascular screening as part of screening for hereditary hemorrhagic telangiectasia [17].

CONCLUSION

Juvenile idiopathic polyposis is a rare disease. Clinical and histological evaluation is crucial for diagnosis. Given the significant risk of digestive and extra-digestive cancers associated with the genetic background, multidisciplinary care is necessary. Thorough exploration of the digestive tract enables precise polyp analysis and early intervention, potentially avoiding surgery.

Conflicts of Interest: The authors declare no conflicts of interest.

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