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An Uncommon association of Adenocarcinoma with Chronic Perianal Fistula: A Case Report and Literature Review

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Abstract **Case Report**

Perianal fistula is a common clinical condition, typically resulting from abscess formation of the anal glands or as a complication of surgical intervention. Malignant transformation of chronic fistulas in ano is rare, accounting for 3% to 11% of all anal canal malignancies. Mucinous adenocarcinoma, a subtype of adenocarcinoma, is especially rare within perianal fistula tracts. Here, we report the case of a 42-year-old male with perianal pain and a discharging fistula in ano of 3 years' duration, who underwent fistula surgery but experienced recurrence. Adenocarcinoma was diagnosed through histopathological examination of a biopsy taken from the fistulous tract. While anal fistulas are generally considered benign, chronic perianal fistulas may indicate a more serious underlying condition, such as perianal adenocarcinoma. This case highlights the importance of early referral and a multidisciplinary approach for accurate diagnosis and prompt treatment.

Keywords: Perianal Disease, Perianal Fistula, Mucinous Adenocarcinoma.

INTRODUCTION

Perianal fistula is a common proctological condition. In many cases, prolonged chronic inflammation is a potential cause of cancer associated with fistulas [1]. A chronic, non-healing fistula can be indicative of a serious underlying condition, such as malignancy [2]. Adenocarcinoma arising in the context of chronic fistulas is uncommon, representing only 3% to 11% of all malignancies of the anal canal [3]. The literature contains only a limited number of case reports and case series on this topic. The onset of adenocarcinoma is frequently attributed to dysplastic changes within a chronic or recurrent fistula, or to the infiltration of the fistula by malignant cells derived from the colorectal epithelium. [4]. These tumors are often locally aggressive, and most patients require extensive surgery, which frequently involves flap closure. However, due to the inflammatory nature of the condition, accurately assessing the local extent of the tumor is challenging, leading to a relatively high risk of local recurrence.[5] Some series have reported favorable outcomes with neoadjuvant treatment followed by radical surgery [6,7].

Here, we report a case of mucinous adenocarcinoma arising from a recurrent perianal fistula, along with a review of the literature on its management.

CASE REPORT

A 42-year-old male presented to the digestive department with recurrent perianal pain and a discharging fistula in ano that had persisted for 3 years. He also reported weight loss, anorexia, and generalized weakness. The patient had previously undergone anal fistulotomy at another hospital, and a biopsy of the excised tissue revealed mucinous adenocarcinoma. During the examination, an external opening of the perianal fistula was noted at the 5-o'clock position, with active discharge. An indurated, non-tender mass was palpable around the external opening. No internal opening was visible. The patient's body mass index was 24.8 kg/m², and laboratory investigations were normal. Complete colonoscopy showed normal colonic and rectal mucosa.

Three-dimensional computed tomography (CT) imaging of the abdomen and pelvis revealed a fistulous tract extending from the anal verge to the lower rectum. Subcentimetric mesorectal nodes were observed. Postoperative histopathological analysis revealed

discrete tumor deposits (N1c) with no evidence of dissemination. The case was reviewed at a multidisciplinary tumor board meeting. Based on previous case reports and institutional experience with adenocarcinoma in the lower rectum, the patient received neoadjuvant concurrent chemoradiotherapy, followed by chemotherapy.

After the neoadjuvant treatment, the patient underwent evaluation with CT of the chest and abdomen, and pelvic MRI. The MRI showed asymmetric circumferential mural thickening, extending from 18 mm above the anal verge to the lower rectum. No adjacent organ involvement was found.

The patient then underwent abdominoperineal excision with wide local excision of the fistulous tract and primary wound closure. Histopathological examination of the resected specimens revealed R0 resection with mucinous adenocarcinoma (ypT3N1c). The patient's postoperative recovery was uneventful, and he received adjuvant chemotherapy.

DISCUSSION

Mucinous adenocarcinoma is a rare malignancy, representing only 3% to 11% of anal cancers. Furthermore, its association with perianal fistula is uncommon, with only a limited number of cases reported in the literature [3].

Determining whether fistula in ano developed first or if malignancy led to fistulization presents a significant challenge for clinicians. Mucinous adenocarcinoma is a distinct subtype of adenocarcinoma, characterized by a high incidence of lymph node infiltration and peritoneal implantation. It typically occurs in the proximal colon [8]. Histopathological analysis remains the gold standard for diagnosis. Mucinous adenocarcinoma is often associated with benign inflammatory conditions such as chronic anal fistula, perianal abscess, diabetes, and Crohn's disease [7]. Perianal mucinous adenocarcinoma in fistula tracts is thought to result from dysplastic changes induced by the continuous regeneration of the mucosal lining within the fistula [9]. Consequently, chronic inflammation due to perianal fistulas should not be overlooked, and early medical evaluation is warranted, especially for patients with atypical clinical features or recurrent anal fistulas. These patients should undergo prompt biopsy to exclude the possibility of malignancy.

In 1934, Rosser [10], was the first to report perianal mucinous adenocarcinoma arising from an anorectal fistula. He outlined the following diagnostic criteria:

1. The fistula should antedate the carcinoma by at least 10 years.
2. The tumor present in the rectum or anal canal should be due to direct extension from carcinoma in the fistula.
3. The internal opening of the fistula should be into the anal canal and not into the tumor.

Perianal discharge and discomfort are common clinical symptoms that are nonspecific and may be easily misdiagnosed as benign conditions, such as perianal fistula or abscess [2-11]. In our patient, the fistula had been present for 3 years, and no tumor was detected in the rectal mucosa. Other studies in the literature have reported a wide range of time intervals between the onset of a fistula and its progression to a malignant lesion, varying from 6 months to 20 years [5-16]. The risk of fistula-associated cancer is often correlated with the duration of the disease [17].

Histologic evaluation remains the gold standard for confirming the diagnosis, revealing extracellular mucinous lakes surrounded by well-differentiated, dilated, and tortuous glands, nerves, and vessels. Additionally, the use of Magnetic Resonance Imaging (MRI) and Ultrasonography (US) aids in defining the anatomy of the fistula tract, characterizing the mass, and identifying associated tumors in extramural locations [5-19]. Pai *et al*., [12], reported that malignancy was detected in the surgical specimens of fistulas in 20% of the patients in their series.

Conversely, colonoscopy may have limited significance, as most reported cases have shown unremarkable findings. However, it remains important for ruling out the presence of primary or synchronous tumors, which is crucial for management planning [12-20]. In our patient, colonoscopy showed normal mucosa in both the rectum and colon.

In summary, clinicians should maintain a high index of suspicion for malignancy in cases of chronic, non-healing fistula in ano, and biopsy of the lesion is crucial for an accurate diagnosis.

Due to the absence of a consensus, the optimal management strategy remains controversial. Abdominoperineal resection (APR) is generally considered the first-line treatment. The primary goal of surgery is to achieve R0 resection; however, this is challenging as the resection margins may be obscured by inflammation and edema. The risk of recurrence, both local and distant, is relatively high, as the associated inflammation can complicate margin assessment. The two largest series report improved survival outcomes with surgery alone, with the majority of patients in these series undergoing abdominoperineal excision [13, 14]. Neoadjuvant treatment is thought to play a crucial role in achieving R0 resection and preventing local recurrence; however, this potential has not been definitively established. Several studies have reported improved outcomes with neoadjuvant treatment followed by surgery [6-12]. For instance, Hongo *et al*., [21], observed a complete pathological response in 85% of cases after neoadjuvant treatment, with better outcomes in patients

who received neoadjuvant therapy, abdominoperineal excision, and adjuvant treatment. In an earlier study, Gaertner *et al*., [22], found that all patients who received neoadjuvant treatment were alive at follow-up (7 out of 7). They concluded that better outcomes could be achieved through neoadjuvant treatment combined with extralevator abdominoperineal excision. However, the type of neoadjuvant treatment varies across studies.

Malignancy associated with chronic fistula in ano is typically locally advanced, and the inflammation present may obscure the resection margins. As a result, neoadjuvant treatment can aid in sterilizing the resection margins, reducing the extent of resection, and preventing local recurrence. However, there is no consensus on the use of chemotherapy or chemoradiotherapy. In our patient, neoadjuvant chemoradiotherapy was administered based on the decision of the multidisciplinary tumor board, which was informed by the outcomes of lower rectal adenocarcinoma.

CONCLUSION

The association of anal fistula with perianal mucinous adenocarcinoma is an exceptionally rare clinical entity. Detecting malignancy in chronic, non-healing fistulas necessitates a high level of clinical suspicion, particularly in the presence of characteristic clinical features. Most patients with this condition require abdominoperineal excision or more radical surgical approaches to achieve R0 resection. Neoadjuvant therapy plays a crucial role in enhancing recurrence-free survival outcomes. Further research is needed to establish standardized treatment protocols for this rare and complex condition. This case highlights the critical importance of early diagnosis in improving overall patient outcomes.

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