International Surgery Journal
Roche SN et al. Int Surg J. 2026 Aug;13(8):1480-1482
https://www.ijsurgery.com
pISSN 2349-3305 | eISSN 2349-2902

DOI: https://dx.doi.org/10.18203/2349-2902.isj20262474

Case Report

Redefining the spectrum of fistula-associated malignancy in Crohn's disease: a rare case of colonic adenocarcinoma arising from a perianal fistula

Shruti N. Roche[1]*, Peter J. Hendy[1], Ekta Bhutada[2], Alexis Ford[3]

[1]Department of Gastroenterology, Mater Hospital, South Brisbane, QLD, Australia
[2]Department of Pathology, Mater Hospital, South Brisbane, QLD, Australia
[3]Queensland X-Ray Radiology Department, Mater Hospital, South Brisbane, QLD, Australia

Received: 01 June 2026
Revised: 08 July 2026
Accepted: 18 July 2026

***Correspondence:**
Dr. Shruti N. Roche,
E-mail: shruti.roche@mater.org.au

ABSTRACT

Crohn's disease (CD) is a chronic, relapsing, and progressive disease characterised by transmural inflammation affecting any segment of the gastrointestinal tract. Perianal fistulising CD (PFCD) affects approximately 20% of CD patients and is associated with a more aggressive disease course and reduced quality of life than those without fistulising disease. While the association between luminal colorectal cancer and CD has been well-established, PFCD-associated fistula cancers is a rarer entity. Existing literature is comprised mostly of case reports and case series, thus guidelines for detection and management of these cancers are scarce. Current diagnostic techniques display major shortfalls, necessitating the adoption of a multimodal, multidisciplinary approach. Further research is needed into its true incidence, pathophysiology and demographics, as well as the efficacy of and enhancing current diagnostic techniques. We present a case of a young female who developed an adenocarcinoma within a chronic perianal fistula.

Keywords: Perianal fistula, Adenocarcinoma, Crohn's disease

INTRODUCTION

Crohn's disease (CD) is a chronic, relapsing, and progressive disease characterised by transmural inflammation affecting any segment of the gastrointestinal tract. Perianal fistulising CD (PFCD) affects approximately 20% of CD patients and is associated with a more aggressive disease course and reduced quality of life.[1] While the association between luminal colorectal cancer and CD has been well-established, PFCD-associated fistula cancers are extremely rare. Existing literature is comprised mostly of case reports and case series, posing a diagnostic and therapeutic challenge.

CASE REPORT

Here we describe a case of a 38-year-old female with a 17-year history of complex PFCD, on infliximab and adjunct 6-mercaptopurine, who presented with symptoms of perianal fullness and pain. Initial evaluation with magnetic resonance imaging (MRI) demonstrated complex fistulising disease with interval improvement in her known perianal collections. Due to persistence of her perianal symptoms, she proceeded to an examination under anaesthesia (EUA) which revealed a thickened cutaneous lesion in the perineum, corresponding to a region of non-specific oedema and soft tissue thickening on her MRI (Figure 1). Histology of the cutaneous lesion confirmed colorectal adenocarcinoma (Figure 2). Immunohistochemistry demonstrated strongly positive

staining for CK20 and CDX2 (Figure 3), and proficiency across all four mismatch repair proteins (MLH1, PMS2, MSH2 and MSH6). Staging imaging concluded locally invasive T4 disease with no evidence of metastases. No luminal lesion was identified on colonoscopy. The consensus to treat with total neoadjuvant chemotherapy using folinic acid, fluoruracil and oxaliplatin, followed by pelvic exenteration surgery, was made by the multidisciplinary team. She is currently receiving her fifth cycle of chemotherapy.

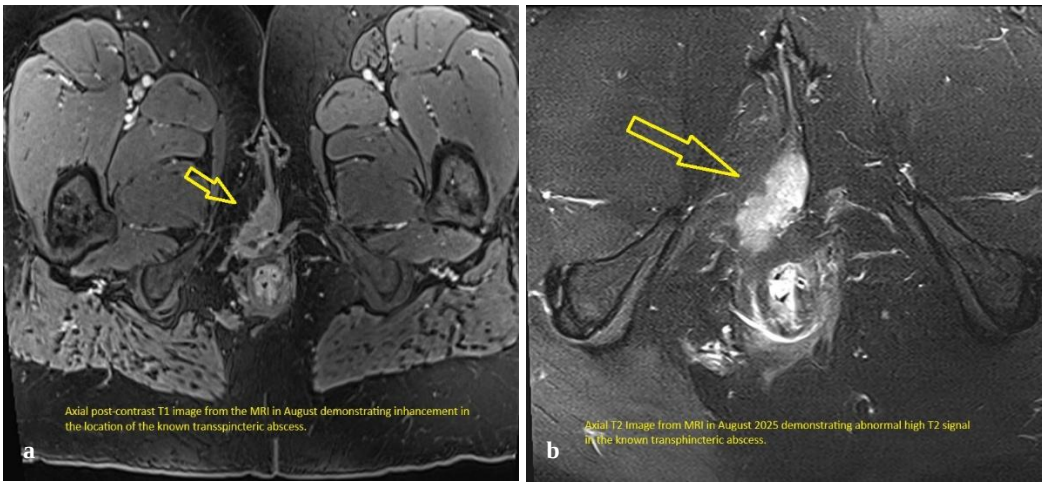


Figure 1: (a) Axial post-contrast T1 image demonstrating enhancement in the location of the known transphincteric abscess and (b) axial T2 image demonstrating abnormal high T2 signal in the known transphincteric abscess.

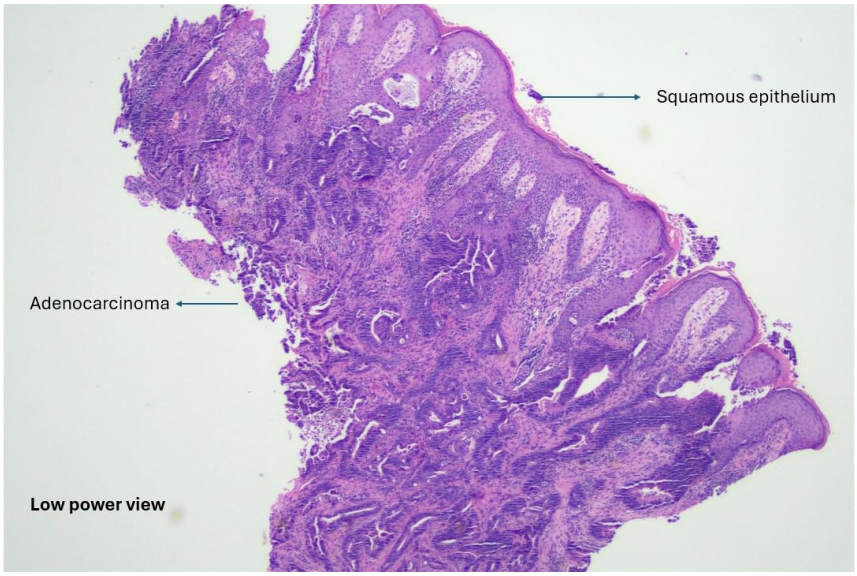
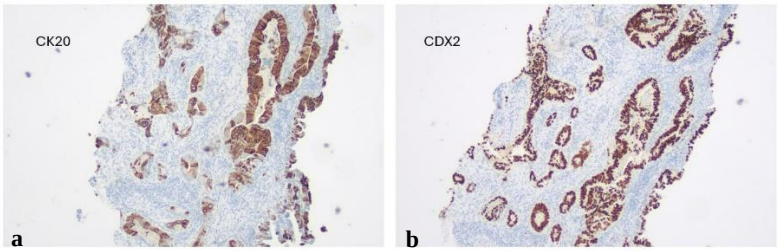


Figure 2: Biopsy of the cutaneous lesion consistent with adenocarcinoma.



Immunostains to confirm colorectal adenocarcinoma

Figure 3 (a and b): Biopsy of the cutaneous lesion demonstrating positive immunohistochemical staining for CK20 and CDX2.

DISCUSSION

Malignancy arising from a chronic perianal fistula was first described in 1975 and is a rare complication of CD, with a 2011 meta-analysis by Laukoetter et al. estimating an incidence of 0.2/1000 patient-years.[2,3] A consistently reported risk factor is duration of PFCD, with an estimated median time of 12 years – though this data is sparse.[4] A retrospective cohort study found an equal distribution of squamous cell carcinoma (SCC) and adenocarcinoma histology among fistula-associated malignancies.[5] Though nomenclature of perianal cancers in PFCD is poorly established, the literature refers to predominantly anal adenocarcinoma, with no previously described cases of ectopic colorectal adenocarcinoma arising from fistulae.

Distinguishing between benign fistulae and PFCD-associated fistula cancer is challenging owing to significant overlap in symptoms. MRI is typically used to evaluate suspected malignancy, however discriminating between active inflammation and malignant transformation is difficult.[6] Treatment depends on the histology, with SCC subtypes more responsive to radiotherapy. Certainly, overall prognosis remains poor, particularly in adenocarcinoma subtypes, with even less is known about the ectopic colorectal adenocarcinomas.[4]

CONCLUSION

In summary, we report a unique case of ectopic colorectal adenocarcinoma arising from a chronic perianal fistula in a patient with CD. Clinicians should maintain an index of suspicion in PFCD of long duration presenting with new or persisting perianal symptoms. A combined MRI/EUA approach should be pursued, with repeat tissue sampling performed where clinical concern remains high. Further research is needed, specifically into the ectopic colorectal adenocarcinoma subtype, in order to improve screening and management.

Verbal consent has been obtained from the patient for publication of this case report.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

REFERENCES

1. Panés J, Rimola J. Perianal fistulizing Crohn's disease: pathogenesis, diagnosis and therapy. Nat Rev Gastroenterol Hepatol. 2017;14(11):652-64.
2. Lightdale CJ, Sternberg SS, Posner G, Sherlock P. Carcinoma complicating Crohn's disease. Report of seven cases and review of the literature. Am J Med. 1975;59(2):262-8.
3. Laukoetter MG, Mennigen R, Hannig CM, Osada N, Rijcken E, Vowinkel T, et al. Intestinal cancer risk in Crohn's disease: a meta-analysis. J Gastrointest Surg. 2011;15(4):576-83.
4. Wong SY, Rowan C, Brockmans ED, Law CCY, Giselbrecht E, Ang C, et al. Perianal Fistulizing Crohn's Disease-Associated Anorectal and Fistula Cancers: Systematic Review and Expert Consensus. Clin Gastroenterol Hepatol. 2025;23(6):927-45.
5. Shwaartz C, Munger JA, Deliz JR, Bornstein JE, Gorfine SR, Chessin DB, et al. Fistula-associated anorectal cancer in the setting of Crohn's disease. Dis Colon Rectum. 2016;59(12):1168-73.
6. Shyn PB. 18F-FDG positron emission tomography: potential utility in the assessment of Crohn's disease. Abdom Imaging. 2012;37(3):377-86.

Cite this article as: Roche SN, Hendy PJ, Bhutada E, Ford A. Redefining the spectrum of fistula-associated malignancy in Crohn’s disease: a rare case of colonic adenocarcinoma arising from a perianal fistula. Int Surg J 2026;13:1480-2.