DOI: https://doi.org/10.4038/sljd.v25i1.230 SLJD Open Access

CASE REPORT

Primary extramammary Paget's disease of the perianal region: a case report and review of the literature

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Abstract

Extramammary Paget's disease (EMPD) is a rare cutaneous adenocarcinoma that arises in apocrine gland–bearing regions, most commonly the vulva, scrotum, or perianal skin. Perianal Paget's disease (PPD) represents around one-fifth of EMPD cases and is often associated with underlying malignancy. We report a 63-year-old male with hypertension and dyslipidaemia who presented with a three-year history of an itchy erythematous plaque around the perianal region. Punch biopsy confirmed EMPD. Systemic evaluation, including contrast-enhanced CT of the chest, abdomen, and pelvis, tumour markers (AFP, CEA, PSA), and colonoscopy, excluded synchronous malignancy.

Following multidisciplinary team (MDT) discussion, the patient underwent wide local excision (WLE) of the lesion with protective sigmoid loop colostomy. Histology demonstrated intraepidermal proliferation of Paget cells without dermal invasion. The postoperative course was uneventful, and the patient was referred for adjuvant radiotherapy. This case highlights the importance of early biopsy, complete staging, and WLE for localized perianal EMPD. Long-term post-therapeutic surveillance remains imperative due to the high propensity for local recurrence characteristically observed in these primary variants.

Keywords: extramammary Paget disease; perianal Paget disease; adenocarcinoma; wide local excision; CK7; immunohistochemistry

Conflicts of Interest: None declared

Funding: None

Received: 27 January 2026 Revised: 09 May 2026 Accepted: 12 May 2026

How to cite this article: Chandrasegara L, Mendis WN. Primary extramammary Paget's disease of the perianal region: a case report and review of the Literature. Sri Lanka J Dermatol. 2026;25(1):150-153. doi: https://doi.org/10.4038/sljd.v25i1.230

Introduction

Extramammary Paget's disease is an uncommon intraepithelial adenocarcinoma that may occur as a primary lesion confined to the epidermis or as a secondary manifestation of an underlying visceral carcinoma. Primary EMPD arises from apocrine or eccrine duct epithelium, whereas secondary EMPD represents epidermotropic spread from adjacent gastrointestinal or genitourinary malignancies.[1,2] The perianal region accounts for approximately 20% of all EMPD cases.[3,4] Because early lesions often mimic chronic eczema or fungal infection, diagnosis is frequently delayed. Histologically, EMPD is characterised by large, pale-staining Paget cells with intracytoplasmic mucin. Immunohistochemistry using cytokeratin 7 and 20 helps to distinguish between primary (CK7+/CK20−) and secondary (CK7+/CK20+) disease.[1,2] Surgical excision with histologically clear margins remains the standard of care for non-invasive lesions.[4,5]

Case Presentation

A 63-year-old male with well-controlled hypertension and dyslipidaemia presented with a three-year history of an itchy erythematous lesion around the anus that had progressively enlarged. There was no pain, bleeding, or alteration in bowel habit. Examination revealed a large, sharply demarcated, erythematous, eczematous plaque with a moist, eroded, verrucous, and hyperkeratotic surface with focal crusting, encircling the anal verge and extending onto the perianal and perineal skin. Accurate measurement was limited by the anatomical location and photographic angle; however, the lesion appeared to involve a substantial circumferential area of the perianal region. The clinical differential diagnosis at presentation included EMPD, squamous cell carcinoma (SCC), and SCC in situ (Bowen's disease/perianal intraepithelial neoplasia) (Figure 1).

Diagnosis and Management: A punch biopsy showed findings consistent with primary extramammary Paget's disease (Figure 2A). Comprehensive staging was performed. Contrast-enhanced CT of the chest, abdomen, and pelvis revealed no nodal or visceral disease. Serum AFP, CEA, and PSA were within normal limits. Colonoscopy revealed normal mucosa without intraluminal lesions or evidence of extension beyond the anal verge (Figure 2B).

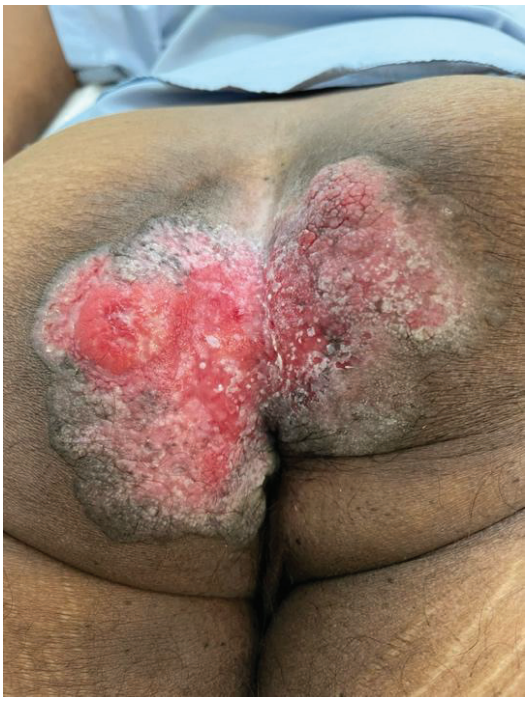
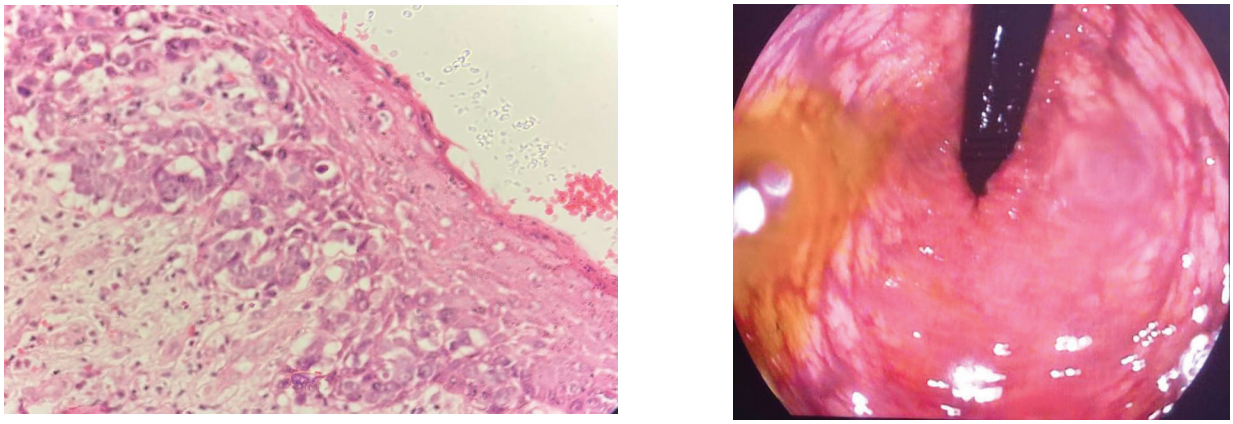


Figure 1. Preoperative clinical photograph showing a sharply demarcated erythematous perianal plaque with erosions, crusting, and hyperkeratotic surface changes.



A B

Figure 2. (A) Histopathology of the punch biopsy (H&E, ×100) showing large pale-staining Paget cells with abundant vacuolated cytoplasm within the epidermis. (B) Colonoscopic view demonstrating normal anal mucosa without intraluminal lesions.

After MDT discussion, a wide local excision with a 2 cm circumferential margin and temporary sigmoid loop colostomy were planned. At surgery, a superficial perianal plaque was excised circumferentially while preserving the anal sphincter complex. The wound was closed primarily, and a protective colostomy was matured in the left iliac fossa.

Histopathological examination confirmed CK7+/CK20-/CDX2-/ Gross Cystic Disease Fluid Protein-15 (GCDFP15+), suggestive of primary Paget's disease without invasion. There was no lymphovascular spread. Focal involvement of resection margins was noted. The postoperative course was uneventful. Because the resection margins were focally involved, the patient received adjuvant external-beam radiotherapy to reduce the risk of local recurrence.

Discussion

Perianal EMPD is a rare cutaneous neoplasm with an indolent but recurrent course. Perianal extramammary Paget's disease remains a diagnostic challenge because early lesions often resemble chronic inflammatory dermatoses such as eczema, contact dermatitis, or fungal infection, leading to delays in biopsy and definitive diagnosis.[2,4] In the present case, the patient had a three-year history of symptoms before histological confirmation. Early biopsy of persistent or atypical perianal dermatoses is therefore crucial to avoid diagnostic delay. Immunophenotypic analysis helps differentiate primary cutaneous disease from secondary involvement by underlying anorectal or genitourinary malignancy. Primary EMPD typically demonstrates CK7+/CK20−/GCDFP-15+ expression, while secondary EMPD shows CK7+/CK20+/CDX2+.[1,2,5] In our patient, the CK7+/CK20− profile, normal colonoscopy, and absence of internal malignancy confirmed a primary lesion.

One study classified perianal EMPD into four stages: I, disease confined to epidermis and adnexa; II, with associated adnexal or visceral carcinoma; III, nodal metastasis; and IV, distant metastasis. Prognosis is favourable for stage I disease, but worsens with invasion or associated carcinoma.[1] For non-invasive lesions, wide local excision with at least 2 cm margins provides optimal local control. Frozen-section margin assessment may assist intra-operatively, though skip lesions can result in false negatives. When circumferential excision produces a large defect, temporary colostomy facilitates healing and prevents infection.[4]

Although wide local excision remains the mainstay of treatment for perianal extramammary Paget's disease, several non-surgical and systemic therapeutic options have been described for recurrent or unresectable disease. Photodynamic therapy utilising 5-aminolaevulinic acid has demonstrated complete response rates of 40–70% in reported series, offering a tissue-preserving alternative. Intralesional interleukin-2 has been employed as a local immunotherapeutic strategy with documented tumour regression. Regarding targeted systemic

therapies, HER2-targeted agents, including trastuzumab, have shown clinical responses in HER2-overexpressing tumours, which account for approximately 15–50% of cases; tyrosine kinase inhibitors such as lapatinib have demonstrated activity in small cohorts; and PIK3CA inhibitors represent an emerging avenue given the frequency of PI3K pathway activation in EMPD tumours.[5]

Adjuvant radiotherapy or topical therapies such as imiquimod can be considered in patients with positive margins or those unfit for further surgery.[2,4] Reported a 5-year overall survival of 90% for perianal EMPD, though recurrence occurred in 50% of cases, highlighting the need for prolonged surveillance.6Follow-up should include regular clinical examination, tumour markers, and colonoscopy.

Conclusion

Primary perianal extramammary Paget's disease is a rare malignancy that may clinically mimic benign inflammatory dermatoses, resulting in delayed diagnosis. Early biopsy of persistent perianal lesions is essential for timely recognition. A comprehensive evaluation is necessary to exclude associated internal malignancies and to differentiate primary from secondary disease. Wide local excision remains the cornerstone of treatment for localized disease, although recurrence is common and long-term surveillance is required.

Ethics statement

Written informed consent was obtained from the patient for participation and publication of clinical details and images.

Acknowledgements

The authors wish to thank the Dermatology and histopathology teams involved in the diagnosis and management of this patient for their support and assistance.

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