SKIN MANIFESTATIONS OF INTERNAL DISEASE

# PERIANAL EXTRAMAMMARY PAGET DISEASE : INTERNAL MALIGNANCY OR PRIMARY ADENOCARCINOMA OF THE SKIN?

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Background: Extramammary Paget's disease (EMPD) can either be a primary adenocarcinoma
of the skin or represent epidermotropic spread from an internal malignancy. Clinical presentation, patient history, and histology can be used to guide work-up, but no diagnostic test can definitively distinguish between the two.

Observation: A 70-year-old female presented for evaluation of a 5-cm itchy, verrucous, circumferential, well defined, pink plaque in the perianal area. The lesion initially presented a year prior and was treated as contact dermatitis with minimal response to topical corticosteroid. The lesion was biopsied, showing in situ EMPD associated with verrucous/papillary epidermal hyperplasia. Immunohistochemical staining was positive for CK7, CK20, and CDX2, indicating potential underlying gastrointestinal malignancy. Confocal microscopy depicted the presence of Paget's cells appearing as dark holes in the epidermis with pagetoid spread. In 2013, the patient was diagnosed with borderline ovarian tumor for which she had an hystero-salpingo-oophorectomy. Her father, as well as maternal uncle, aunt, and grandmother all suffered from GI cancers. Due to strong family history and CK20/CDX2 positive immunohistochemistry, a thorough cancer work-up was conducted. Colonoscopy confirmed disease involvement of the distal anal canal, but found no primary GI malignancy. Enlarged left inguinal lymph nodes were observed on PET. Lymph node FNA, gynecological exam, gastroscopy, pap-smear, mammography, as well as serum CA125 and CEA were unremarkable. Urological evaluation, targetable action gene sequencing, as well as genetic testing are pending. Despite a strong family history of cancer and CK20/CDX2 positive histology, no evidence of internal malignancy has been found. While awaiting final results, surgical treatment options are being weighed against radiotherapy and imiquimod treatment.

Key message: This case highlights classical clinical, confocal, and pathological findings of EMPD, underscoring the diagnostic and therapeutic challenges that present with EMPD.