

Unusual Presentation of Cancer of Unknown Primary Origin

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Background: Colorectal cancer (CRC) is the fourth most diagnosed cancer in the world (1) with an overall lifetime risk of 4.3% for men and 4% for women (2). Adenocarcinomas account for 96% of all CRC (3), while Perianal adenocarcinomas account for 2-3% of all gastrointestinal malignancies (4), usually originating from perianal abscesses and Fistula in Ano. Here we present an unusual case of a 74-year-old man who presented with a right gluteal lesion and was found to have moderately differentiated adenocarcinoma on punch biopsy with immunohistochemistry (IHC) suggestive of colorectal origin. However, lesion in the colon was not identified.

Case Description: The patient is a 74-year-old man with a past medical history of Crohn's disease with recurrent perianal fistulas and abscesses who presented with a progressively increasing right gluteal lesion for the past two years. Physical examination revealed a large fungating friable mass extending over the entire right buttock area, with yellow foul-smelling slough and serosanguineous discharge. Laboratory workup revealed leucocytosis, microcytic anaemia, and elevated carcinoembryonic antigen (CEA) levels. An MRI of the pelvis revealed a 9.8 x 16.1 x 7.7 cm heterogeneous mass inseparable from the posterior aspect of the anus and along the course of a previously seen perianal fistula. Histopathology of a punch biopsy of the mass was consistent with moderately differentiated adenocarcinoma, and IHC was suggestive of colorectal origin. Astonishingly, a luminal tumor was not identified on colonoscopy.

Conclusion: Cancer of unknown primary (CUP) is a relatively rare clinical entity and their ability to metastasize prior to formation of a clinically identifiable primary is a diagnostic challenge. Although imaging such as CT and PET scans are helpful, light microscopic examination of initial biopsy is the most important clue towards the organ/site of origin. Euclid findings should prompt IHC as highlighted in this unusual case.