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A case of colorectal malignancy with clinical features of Birt-Hogg-Dubé syndrome

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Description

A 67 year old male patient presented with bleeding per rectum of 2 months duration. On colonoscopy and biopsy, he was diagnosed to have recto sigmoid malignancy with imaging of the abdomen and thorax showing an irregular asymmetric circumferential thickening in the recto sigmoid region of maximum thickness of 28 mm, hypo dense lesions (largest 2.2 x 1.8 cm) in right lobe of liver and bi-lobar simple liver cysts. Exophytic lesions in bilateral kidneys (largest 7.8 x 7.2 cm) were observed. Pulmonary cysts were seen in bilateral lung parenchyma, suggesting a clinical picture of Birt-Hogg-Dubé syndrome. Patient underwent anterior resection and tumour removal with descending end colostomy as he had luminal narrowing and large bowel obstruction followed by systemic chemotherapy with Capecitabine and Oxaliplatin.

Birt-Hogg-Dubé Syndrome (BHDS) is a relatively rare, inherited genetic disorder characterized by the development of benign skin tumours, lung cysts, and an increased risk of kidney cancer [1]. It is caused by mutations in the FLCN gene leading to impaired production of folliculin protein [2]. Individuals with BHDS often present with fibrofolliculomas (small, skin-coloured bumps on the face and neck), and pulmonary cysts that may lead to spontaneous pneumothorax (lung collapse) [3]. Renal cell carcinoma is the most dreaded outcome, with most common subtypes being chromophobe and oncolytic variant [4].

Management of BHDS is mainly preventive and symptom-focused, including routine skin examinations, lung function monitoring, and imaging of the kidneys. Genetic counselling is recommended for affected families to assess and manage risks. Given the syndrome's complexity and the variability in expression among individuals, ongoing research aims to clarify the role of folliculin and improve clinical guidelines for managing BHDS.

BHDS co-existing with colorectal malignancy has been reported in literature previously [1,5]. Thus, it necessitates the need for screening activities directed to colorectal malignancies and exploring the molecular pathogenesis of this entity in future research.

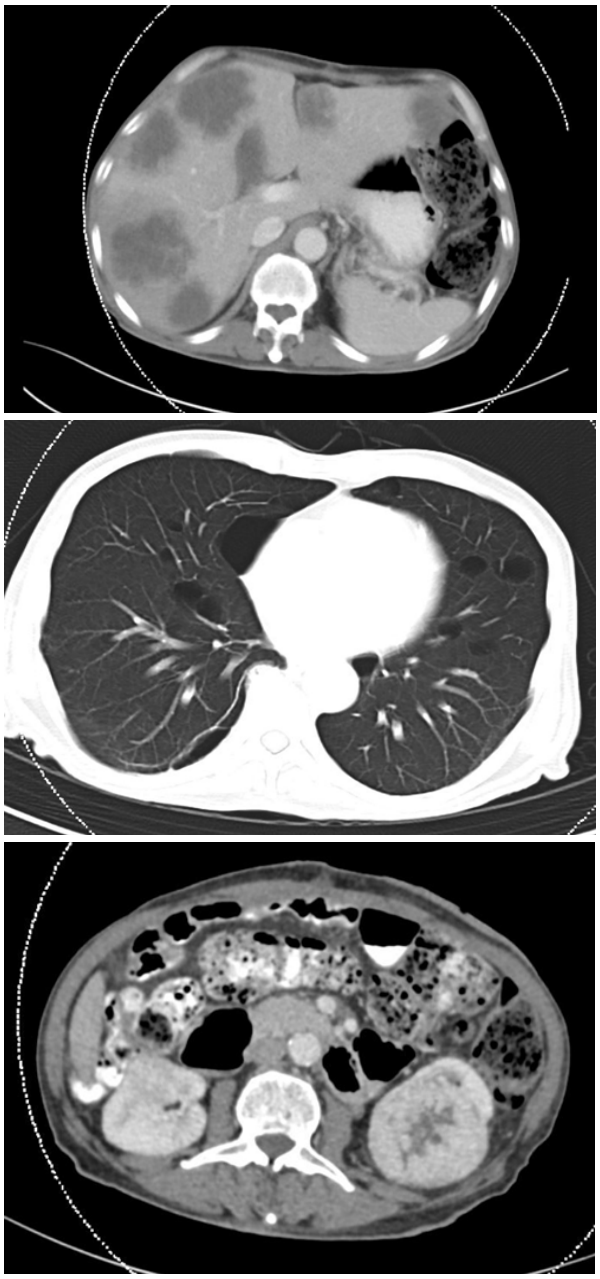

Figure 1: Clinical image.

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