



CLINICAL IMAGE IN GASTROENTEROLOGY

Birt-Hogg-Dubé syndrome and colon polyps[☆]



Síndrome de Birt-Hogg-Dubé y pólipos en el colon

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The Birt-Hogg-Dubé syndrome (BHD) is an inherited autosomal dominant genodermatosis caused by mutations of the folliculin (FLCN) gene. Patients present with fibrofolliculomas (5 are pathognomonic), pulmonary cysts, and renal cancer. The relation of BHD to polyps and colorectal cancer was initially described, but this association is not currently recognized. Nevertheless, FLCN gene mutations have recently been implicated in the development of colon cancer, questioning this viewpoint. We present herein the case of a 55-year-old man that was asymptomatic and evaluated for multiple facial lesions histologically compatible with fibrofolliculomas located predominantly on the nose and cheek (Fig. 1). After BHD diagnosis, an MRI of the kidney and a chest x-ray were ordered and did not identify any pathologic findings. A fecal occult blood test was positive. Colonoscopy revealed a 3 cm cecal sessile polyp (Fig. 2), and biopsy reported a tubulovillous adenoma with high-grade dysplasia that was extirpated through a right



Figure 1 Clinical image showing multiple, smooth, firm, cupuliform normal skin-colored papules on the nasal dorsum (a) and on the right cheek (b), indicative of fibrofolliculomas.

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hemicolectomy. The histologic study (Fig. 3) confirmed the initial findings and the resection margins were disease-free. Despite the fact that malignant colorectal pathology screening after BHD diagnosis is recommended only if there is a family history of colorectal cancer, which was not the case with our patient, a fecal occult blood test enabled the early diagnosis of colon cancer.



Figure 2 Endoscopic image showing an ulcerated sessile polyp located in the cecum.

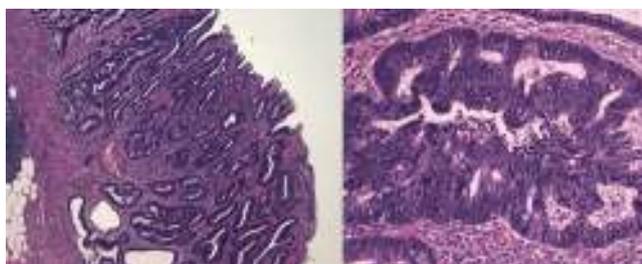


Figure 3 Photomicrograph with hematoxylin-eosin stain showing a tubulovillous adenoma with foci of high-grade dysplasia.

Ethical responsibilities

Protection of persons and animals. The authors declare that no experiments were performed on humans or animals for this study.

Data confidentiality. The authors declare that no patient data appear in this article.

Right to privacy and informed consent. The authors declare that no patient data appear in this article.

Financial disclosure

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Conflict of interest

The authors declare that there is no conflict of interest.