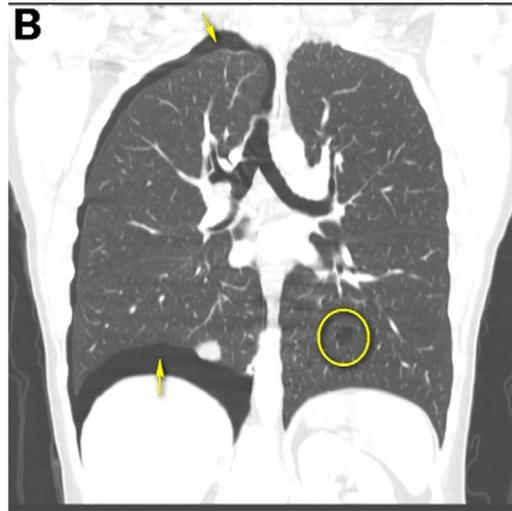
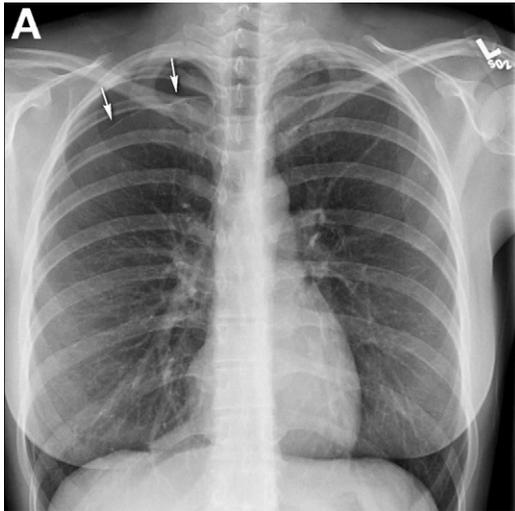


## Recurrent Pneumothoraces After Diagnostic Colonoscopy

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**Question:** A 37-year-old woman with history of stage IIA colon adenocarcinoma 4 years prior, underwent an unremarkable colonoscopy for intermittent abdominal pain and for surveillance. The next day, she experienced pleuritic right-sided chest pain; 3 days after colonoscopy, she presented to the emergency department with progressive right-sided chest pain and decreased breath sounds

on examination. Chest radiography revealed a 10%–15% right-sided pneumothorax (Figure A), which was managed with Thoravent chest tube placement. Computed tomography (CT) of the abdomen and pelvis did not reveal pneumoperitoneum. Subsequently, the patient had 2 spontaneous pneumothoraces, occurring at 1 and 4 months after the initial episode. Chest CT revealed recurrent pneumothoraces and lung blebs (Figure B). After mechanical pleurodesis and stapling of lung bleb failed, right lung wedge resection and talc pleurodesis was successful.

Interestingly, the patient's father also had history of spontaneous pneumothoraces, and recently had a partial nephrectomy for renal cell carcinoma. He had no prior history of colorectal carcinoma.

What is the diagnosis, and what is the likelihood of there being a genetic component?

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**Conflicts of interest:** The authors disclose no conflicts.

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## Answer to the Clinical Challenges and Images in GI: Image 5: Birt–Hogg–Dubé Syndrome

Complications associated with diagnostic colonoscopy are rare.<sup>1</sup> Pneumothorax is especially uncommon after diagnostic colonoscopy. Because of our patient's history of recurrent spontaneous pneumothoraces, lung blebs, and history of colon cancer and polyps (she previously had negative genetic testing for common familial colon cancer syndromes), and recent diagnosis of renal cell carcinoma in patient's father, clinical suspicion for Birt–Hogg–Dubé syndrome (BHD) was high. Subsequent genetic testing of the patient and her father confirmed BHD in both. The patient had a germline mutation in the folliculin gene associated with BHD.

BHD is an autosomal-dominant genetic condition commonly characterized by skin fibrofolliculomas, pulmonary cysts, spontaneous pneumothorax, and renal cell carcinoma.<sup>1</sup> Spontaneous pneumothorax may occur in up to one-quarter of affected patients. Although current data are inconclusive, BHD may carry an increased risk of colonic neoplasia, and this may depend on FLCN allelic heterogeneity that varies between families.<sup>2</sup> A germline mutation in the FLCN gene (which encodes folliculin) has been identified in 90% of affected BHD families. This mutation is localized to the short arm of chromosome 17. DNA sequencing of renal tumors from patients with germline FLCN mutations has identified somatic mutations in the wild-type copy of the gene, suggesting that FLCN is a loss-of-function, tumor suppressor gene.

There have been several published cases of pneumothorax after diagnostic colonoscopy.<sup>3</sup> The majority of these cases report colonic perforation at the time of pneumothorax diagnosis. Postulated causes of pneumothorax include air leakage from either overt colonic perforation or from dissection of air through the colonic wall (pneumatosis coli), which may pass along the fascial planes to the mediastinum causing rupture of pleura. Alternatively, air may pass through small diaphragmatic fenestrations and enter the pleural space by means of a pressure gradient.

In our patient, without imaging evidence of pneumoperitoneum, BHD likely increases her risk of pneumothorax after colonoscopy. This presents a challenge in her future management because she will need further colonoscopies for colon cancer surveillance. In summary, awareness of BHD is important to allow recognition of increased risk of pneumothorax in affected patients who may also require colonoscopies for polyp and cancer surveillance.

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