Colorectal Adenocarcinoma in a Patient with Tuberous Sclerosis

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Abstract

Tuberous sclerosis (TS) is a neurocutaneous disorder that presents with the classic triad of seizure, mental retardation, and facial angiofibroma. In most cases there is a constellation of manifestations, including tumors in the brain, heart, kidneys, lungs, skin, and eyes. Two-thirds of the cases result from a de novo mutation, and one third from autosomal dominant inheritance. The TSC1 and TSC2 genes encode for the hamartin and tuberin proteins, respectively. TSC1 and TSC2 are tumor suppressor genes. We present the case of a 63 year-old white female with a history of nephrolithiasis, melanoma and a two-year history of daily hematochezia which had worsened. During her hospitalization, the patient was diagnosed with TS based on one major criteria: a renal angiomyolipoma, and several minor criteria: the presence of bone cysts, cerebral white-matter “migration” tracts, and a hamartoma. At colonoscopy, a 5 mm polyp in the ascending colon, a 25 mm polyp at the rectosigmoid junction, and a rectal polyp were found. Biopsy of the rectosigmoid polyp revealed a low-grade adenocarcinoma arising from a tubular adenoma. Twenty-six lymph nodes were negative for metastatic disease. A biopsy of the rectal mass revealed a tubulovillous adenoma. Although gastrointestinal malignancies are rare in patients with TS, there are some previous reports suggesting an association. TS is well associated with rectal hamartomas, but the association with colon adenocarcinoma has not been established. Our report of a patient with TS diagnosed with rectosigmoidal adenocarcinoma and rectal adenoma adds to previous reports suggesting an association between TS and gastrointestinal tumor.