

A patient referred for positive fecal occult blood test finally diagnosed with anorectal melanoma

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Rectal melanoma is a rare disease, infamous for its aggressive nature and poor prognosis. It constitutes only 0.5% of rectal malignancies and it is often misdiagnosed as hemorrhoids, adenomatous polyps or rectal cancer. Patients present with nonspecific complaints such as rectal bleeding or anal pain. We report a case of primary rectal melanoma initially diagnosed with lung metastasis. A 67-year-old female with a history of fatty liver and mediastinal thymoma on a regular medical follow-up, was referred to our gastroenterology department with positive fecal occult blood test on the national health medical check-up. On patient examination, she was suffering from constipation and anal pain for the last few months. Esophagoduodenoscopy performed a year ago displayed chronic atrophic gastritis and the patient had never underwent colonoscopic evaluation. Colonoscopy revealed a 3cm sized hard fungating mass with black pigmentation, directly involving and above the anal verge (Figure 1). Prominent nucleoli were present in tumor cells and were positive for melan-A, supporting the diagnosis of melanoma (Figure 1). PET-CT showed multiple well-defined hypermetabolic nodules in both lungs, consistent with rectal melanoma with pulmonary metastasis (Figure 1). As she was on a regular follow-up screening for mediastinal thymoma, chest CT performed 9 months prior to the current visit exhibited no pathologic finding in the lung parenchyme. Furthermore, 2 months prior abdomen ultrasonography revealed no remarkable finding. She was then transferred to the department of oncology for palliative chemotherapy. Three cycles of pembrolizumab unfortunately disclosed disease progression - increased in size and number of metastatic lung nodules and rectal mass from 2.5cm to 3.6cm. Without clinical evidence of gastrointestinal obstructive symptoms, the patient is under the second line palliative chemotherapy with cisplatin, dacabazine and tamoxifen. Treatment of rectum melanoma is controversial due to absence of randomized trials - surgery, radiotherapy, chemotherapy yield uncertain results. New-onset alarm symptoms in aged patients should be managed with meticulous examination.

