Malignant melanomas in the gastrointestinal tract are usually metastases from cutaneous melanomas [1], and primary melanomas in the esophagus or anorectal lesions rarely occur. Primary colonic malignant melanoma is extremely rare, with only eight cases previously reported [2]. Although melanomas usually exhibit macroscopic pigmentation, 30% are amelanotic [3]. Diagnosis of amelanotic melanoma of the gastrointestinal tract by endoscopic examination is difficult owing to its resemblance to gastrointestinal stromal tumor (GIST). We report the case of a patient with amelanotic melanoma of the cecum presenting as a submucosal tumor (SMT), detected by colonoscopy. To our knowledge, this is the first report of primary amelanotic melanoma of the colon.

A 39-year-old woman was referred to our hospital for lower abdominal pain. Colonoscopy revealed an SMT (diameter 20 mm) in the cecum (Fig. 1). Endoscopic ultrasonography revealed a hypoechoic SMT derived from the muscularis propria, which was suspected to be a GIST (Fig. 2). The patient was offered two possible options: conservative follow-up and surgery. The patient gave informed consent for surgery and laparoscopic ileocaecal resection was carried out. The resected tumor measured 20 × 15 × 10 mm and its cut surface was milky white in color (Fig. 3). On histological examination, the tumor cells were spindle shaped with abundant cytoplasm (Fig. 4a). In addition, these cells showed strong positive immunohistochemical staining for HMB-45 (Fig. 4b) but weak positive staining for both smooth muscle antigen and c-kit. These characteristics were consistent with malignant melanoma and the tumor was identified as an amelanotic melanoma. Whole-body computed tomography and positron emission tomography did not reveal any other primary tumors, and thus a diagnosis of primary melanoma of the cecum was established.

Competing interests: None

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Fig. 4 a Histological section showing spindle-shaped tumor cells with abundant cytoplasm (hematoxylin and eosin; magnification × 400). b Strongly positive immunohistochemical staining for HMB-45 (magnification × 400).