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Pigmented Neurofibroma in the Appendix Diagnosed with Colonoscopy

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Pigmented (melanotic) neurofibromas in the appendix are extremely rare. To our knowledge, this is the first case of appendiceal pigmented neurofibroma in a patient without neurofibromatosis 1, which was colonoscopically diagnosed. Histologically, the tumor cells were spindle shaped with indistinct cell borders. The tumor cells had cytoplasms with many gray-brown pigments. Immunohistochemical examinations showed that the tumor cells were strongly positive for S-100 protein, and were focally positive for Melan-A. They were also diffusely positive for Leu7 (CD57), or neuron-specific enolase (NSE). No positive expression of tumor cells was observed for α-smooth muscle actin, desmin, CD34, c-kit, HMB45. No mitotic figures were observed. The Ki-67 labeling index was less than 1%. The tumor was thus diagnosed as primary pigmented neurofibroma in the appendix. In the present case, tumor cells showed no mitotic activity and a Ki-67 index of less than 1%, suggesting a benign tumor.

Key words: appendiceal tumor; pigmented neurofibroma

Pigmented (melanotic) neurofibromas are rare (Fetsch et al., 2000). According to the Armed Forces Institute of Pathology, pigmented neurofibromas constitute less than 1% of all neurofibromas (Sharon et al., 2001). One case of diffuse neurofibroma in the appendix associated with neurofibromatosis 1 (von Recklinghausen's disease) was reported (Miettinen et al., 2001). The present article reports a colonoscopically diagnosed pigmented neurofibroma in the appendix. To our knowledge, this is the first case of appendiceal pigmented neurofibroma in a patient without neurofibromatosis 1.

Patient Report

Clinical summary

A 59-year-old Japanese man, who had stools with occasional signs of blood on occasion, underwent colonoscopic examination. A doughnut-like protruding tumor, slightly whitish in color and about 1.5 cm in diameter, was found in the appendiceal orifice (Fig. 1). However, there were no remarkable abnormalities upon physical or laboratory examinations.

Pathological findings

Histopathological evaluation of the biopsy specimen revealed pigmented neurofibroma in the

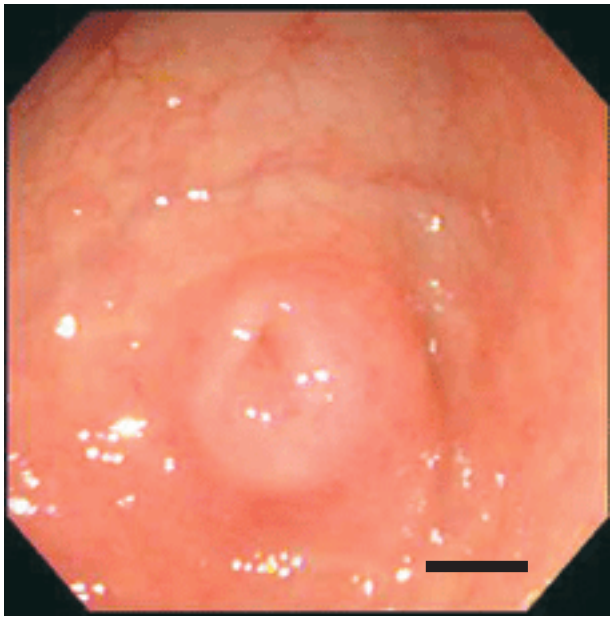

Fig. 1. Endoscopic finding. There is a doughnut-like protruding tumor at the appendiceal orifice. The tumor is slightly whitish in color and 1.5 cm at its greatest dimension. Macroscopically, the surface of the tumor is smooth. Bar = 1 cm.

appendix. Histologically, the tumor cells proliferated in the lamina muscularis mucosae and submucosal tissues (Fig. 2). The tumor was composed of spindle shaped cells with indistinct cell borders. No mitotic figures were observed (Fig. 2). Most of the tumor cells had many gray-brown pigments within cytoplasms, which stained black with Fontana-Masson stain for melanin (Fig. 3). Immunohistochemical studies of the tumor cells were performed for S-100 protein, Melan-A, Leu7 (CD57), neuron-specific enolase, α-smooth muscle actin, desmin, CD34, c-kit and HMB45. The Ki-67 labeling index was less than 1%. The results of the immunohistochemical studies are summarized in Table 1.

Discussion

Sometimes appendiceal tumors are found when the patient has symptoms in the appendix or through intussusception (Abramson et al., 1997). In our case, there were no appendix symptoms

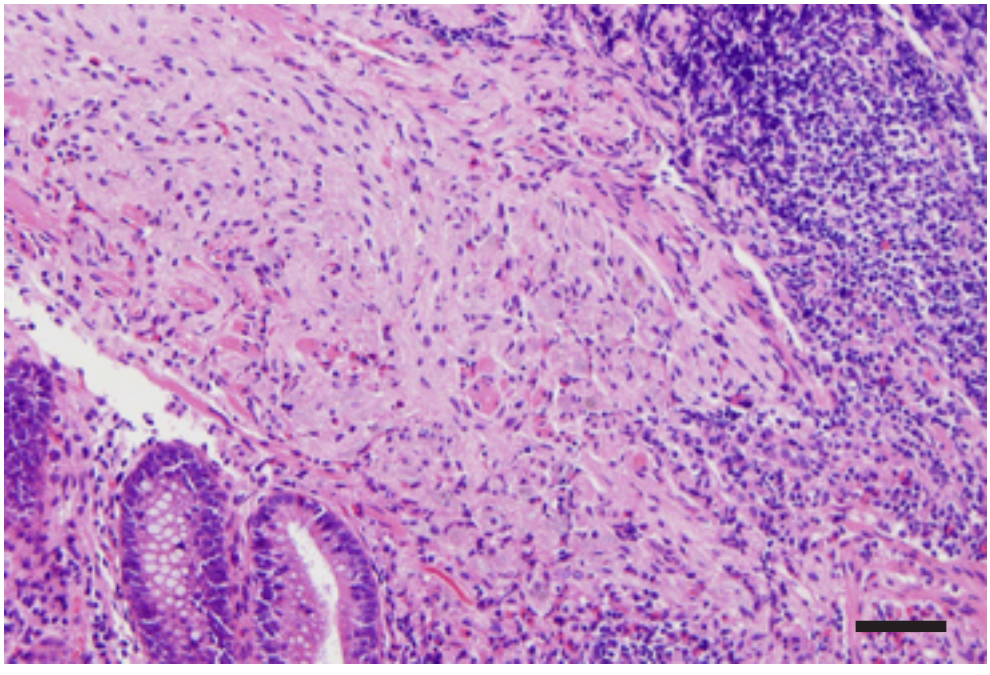

Fig. 2. Tumor cells proliferate in the lamina muscularis mucosa. The tumor is composed of spindle-shaped cells and round cells with indistinct cell borders. The cells have clear eosinophilic cytoplasms. The nuclei of the tumor cells are characteristically elongated and wavy (hematoxylin and eosin stain). Bar = 20 μm.