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MUCOSAL SCHWANN CELL HAMARTOMA ON SCREENING COLONOSCOPY: AN UNUSUAL FINDING

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ABSTRACT

Schwann cells are found in the peripheral nervous system and can sometimes appear as benign hamartoma lesions in various parts of the body. Although rare in the gastrointestinal (GI) tract, they have been observed in the colon.

Recently, mucosal Schwann cell hamartomas of the GI tract have been studied, and it was discovered that they had yet to be investigated up to 2009. In this context, we present the case of a 60-year-old man who was found to have lesions in the transverse colon during a routine colonoscopy. No further investigations were conducted since these lesions have not been associated with any risk of malignancy transformation and have not been linked to any inherited syndromes.

KEYWORDS

Hamartomas, Schwann cell, colonoscopy, polyps

LEARNING POINTS

- Mucosal Schwann cell hamartomas are rare types of polyps that can be found anywhere in the gastrointestinal tract.
- They are benign lesions not usually associated with any inherited syndrome and they are usually found incidentally by endoscopy.
- These polyps are benign and might not require further follow-up once diagnosed.

INTRODUCTION

A mucosal Schwann cell hamartoma is a rare neurogenic lesion first discovered in 2009 by Gibson and Hornik[1]. This condition is not associated with inherited syndromes, and it was named hamartomas to distinguish it from other histologically similar neurogenic fibromas associated with neurofibromatosis and other inherited lesions[1]. These lesions are frequently identified during routine colonoscopy screenings and are often mistaken for regular gastrointestinal polyps and adenoma[2].

In this case, we present a 60-year-old man who underwent a screening colonoscopy and was found to have transverse colon polyps, which were later confirmed to be mucosal Schwann cell hamartomas.

CASE DESCRIPTION

We report on a 60-year old man who is known to have diabetes mellitus type II, hypertension and hypothyroidism

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due to total thyroidectomy (previous retrosternal goitre). He was on oral medications for his high blood pressure and diabetes, plus thyroid hormone replacement. He denied any abdominal pain, vomiting or nausea, blood in stool or abdominal distension. He denied any personal or family history of gastrointestinal malignancy and no family history of any inherited medical condition. His physical examination was unremarkable, with no abdominal distension, tenderness, or lymphadenopathy. Digital per rectal exam showed no evidence of melena or mass lesion. His primary laboratory investigations included a complete blood count and a complete metabolic panel that was unremarkable, and his tumour markers were not elevated. A colonoscopy was undertaken as part of age-appropriate cancer screening. He was found to have a 9 mm tubular polyp in the rectum, three sessile sigmoid polyps (2–3 mm in size), two 2 mm sessile polyps in the transverse colon and one sessile polyp, 3 mm in size, in the ascending colon. All of the polyps had no ulcerated surface, and none of them was bleeding. They were removed via cold snare and then sent for histopathological examination. Upon histopathological exam, the rectal polyp was confirmed to be a tubular adenoma, and the sigmoid and the ascending colon polyps were hyperplastic. A biopsy of the transverse colonic polyps revealed overall histopathological findings consistent with mucosal Schwann cell hamartoma. In particular, the biopsy revealed an ill-defined proliferation of uniform bland spindle cells in the lamina propria with eosinophilic cytoplasm *(Fig. 1A and B)*. The colonic crypts were preserved. Mild chronic inflammatory cell infiltrate was observed; no ganglion cells were identified. The spindle cells showed diffuse and strong positivity for S100 *(Fig. 1C)*, positive SOX10 *(Fig. 1D)*. DOG1 and CD117 were negative, by that we excluded gastrointestinal stromal tumour (GIST). CD68 was negative so granuloma was ruled out, and smooth muscle antibody (SMA) was negative to rule out leiomyoma.

DISCUSSION

Colonic polyps are abnormal growths that protrude into the colon's lumen. They are found in approximately one-third of patients who undergo screening colonoscopy. Removing these growths has been shown to reduce the incidence of colorectal cancer[3]. There are three types of colonic polyps: (1) adenomatous polyps, which are premalignant lesions and account for 50%–65% of all colonic polyps; (2) hyperplastic polyps, which are small lesions of less than 5 mm in size and account for 10%–30% of all polyps but have no clinical significance; and (3) mucosal tags, lipomas and hamartomas, which have no known clinical implications[4].

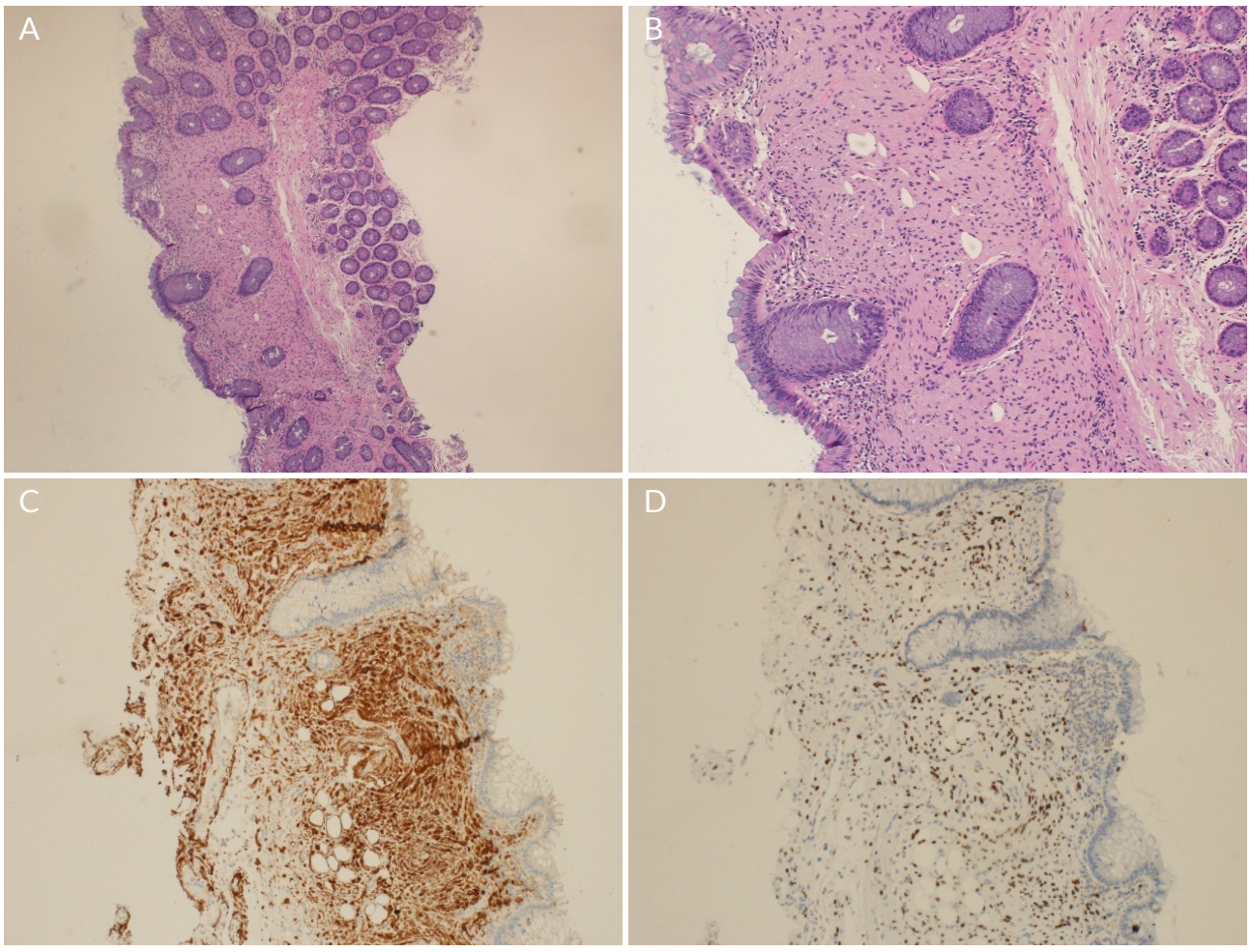

Figure 1. Mucosal Schwann cell hamartoma of the colon. (A) Proliferation of bland spindle cells in the lamina propria (H&E, ×40). (B) Higher magnification (H&E, ×100). (C) Strong and diffuse positive immunoreactivity for S100 (×100). (D) Strong positive immunoreactivity for SOX10 (×100).

The risk of malignant transformation is influenced by the patient's age, size, number and histological features of the polyp[5]. Therefore, the surveillance colonoscopy interval differs from one patient to another. Mucosal Schwann cell hamartomas are rare forms of colonic outgrowths. They were first identified in 2009 by Gibson and Hornik when they found 26 patients with sessile polyps of mesenchymal origin, ranging in size from 1–6 mm[1]. These polyps are histologically composed of uniform spindle cells within the lamina propria and strongly stain positive for S-100 but negative for other stains such as smooth muscle actin[6].

They are unusual polyps usually detected incidentally, and patients typically do not have any specific complaints. These lesions are similar to neurofibromas but contain some intralesional heterogeneity, are less uniformly cellular and stain less with S-100 protein. These polyps are primarily found in the colon but can also appear in different parts of the gastrointestinal system, such as the gallbladder[7]. Since their discovery in 2009, several cases of mucosal Schwann cell hamartomas have been reported, and they are all histologically similar[8–13]. These polyps are not associated with inherited syndrome and pose no risk of malignancy. Therefore, a follow-up colonoscopy is unnecessary[1,12]. After finding two small mucosal Schwann cell hamartoma polyps in the transverse colon, our 60-year-old patient will undergo a follow-up colonoscopy in five years due to other low-risk colonic polyps.

CONCLUSION

Mucosal Schwann cell hamartomas are a type of polyp that can occur anywhere in the gastrointestinal tract. These lesions are scarce and have no risk of turning into cancer. They are not associated with inherited disorders and have unique histological features that set them apart from other polyps. Once diagnosed, there is no need for further investigations or follow-up, as these polyps do not require any treatment.

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