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Rectal Mucosal Schwann-Cell Hamartoma: A Case Report and Literature Review

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Abstract

Background: Mucosal Schwann cell hamartoma is a newly recognized disease entity that describes lesions which share some features but are distinct from schwannomas and neurofibromas. This mesenchymal lesion, consisting of a proliferation of Schwann cells in the lamina propria and a strong positivity for the S-100 protein, should be differentiated from other similar lesions because it exists solely in the intestines as a polypoid lesion. Here, we report on a case of Schwann cell hamartoma diagnosed on pathology of rectal polyp removed during colonoscopy

Case presentation: Our patient is a 60 y/o male who presented for an outpatient colonoscopy to evaluate weight loss and intermittent hematochezia. Colonoscopy revealed sigmoid diverticulosis and a small 5 mm rectal polyp. The polyp was sessile and removed with a cold biopsy forceps. On pathology, the rectal polyp showed S-100 positivity and had benign bland spindle cell proliferation in the lamina propria, findings that were consistent with a diagnosis of Mucosal Schwann cell hamartoma. A follow up colonoscopy was recommended in 3 years.

Conclusion: Mucosal Schwann cell hamartoma is considered a benign lesion and no reports of malignant transformation have been described. However, further follow up data is needed before making final recommendations. Our case report emphasizes this emerging disease and we propose close follow ups for possible malignant transformation.

Key-words: Schwann Cell; Neuroma; Polyp; Hamartoma; Neurofibroma

Introduction

Mucosal Schwann cell hamartomas that are mesenchymal in origin were long considered to be schwannomas or neurofibromas based on their common immunohistochemical staining features. The increase in routine colonoscopies has resulted in the identification of more colorectal polyps that have been evaluated in greater detail, resulting in an increased incidence of Schwann cell hamartomas as a separate entity [1]. The pathological identification of these lesions with histomorphology has characterized them as a unique entity, different from other benign nerve sheath tumors of mesenchymal origin [1]. As compared to neurofibromas and schwannomas, these lesions are not associated with any inherited syndromes such as neurofibromatosis type 1 or schwannomatosis [1, 2]. Mucosal Schwann cell hamartomas should be considered in the differential diagnosis of intestinal mesenchymal tumors, which include neurofibromas, schwannomas, mucosal neuromas, smooth muscle tumors, gastrointestinal stromal tumors, and ganglioneuromas [1, 3].

Histologically, these mucosal tumors exhibit a diffuse proliferation of uniform bland spindle cells with elongated, tapering nuclei, in addition to abundant, dense eosinophilic cytoplasm with no nuclear atypia, pleomorphism, mitosis, or ganglionic cells and with indistinct cell borders in the lamina propria [1, 2]. S-100 positivity is seen in mucosal Schwann cell hamartomas just like in neurofibromas and schwannomas but with more diffuse and extensive positivity [1]. Although there is a proliferation of Schwann cells and S-100 positivity, mucosal Schwann cell hamartomas are different from schwannomas in that they lack Verocay bodies, Antoni A and Antoni B areas, lymphoid infiltrations, and lymphoid cuffs. Neurofibromas and ganglioneuromas can also be distinguished from Mucosal Schwann cell hamartomas on the basis of the presence of fibroblasts with axons and ganglion cells, respectively [1, 4, 5].

Case Report

A 55-year-old male presented to us complaining of unintentional weight loss and intermittent hematochezia. His past medical history was significant for constipation, diverticulosis, and diverticulitis. The abdomen revealed slight tenderness in the patients' right upper quadrant. On the skin exam, there was no pigmentation or findings suggestive of neurofibromas. The patient's laboratory parameters were normal. Colonoscopy was performed that, in addition to multiple diverticula, showed a single polyp in the rectum respectively (Figure 1). On pathological report, the rectal polyp showed benign bland spindle cell proliferation in the lamina propria and

S-100 positivity (Figure 2 and 3). On the basis of the pathological report and immunohistochemical staining, we diagnosed the polyp as Mucosal Schwann cell hamartoma. We planned a three-year follow-up colonoscopy for the patient to check for any recurrences.

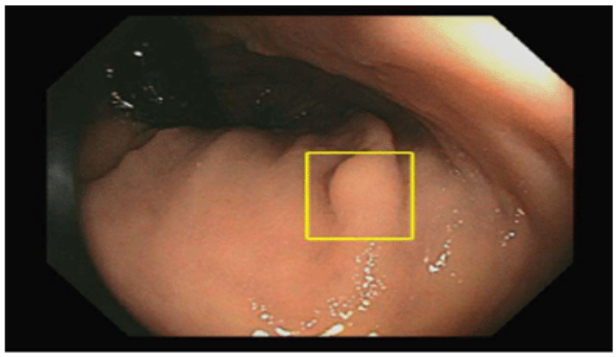

Figure 1: Rectal polyp, 5 mm in size, found on colonoscopy

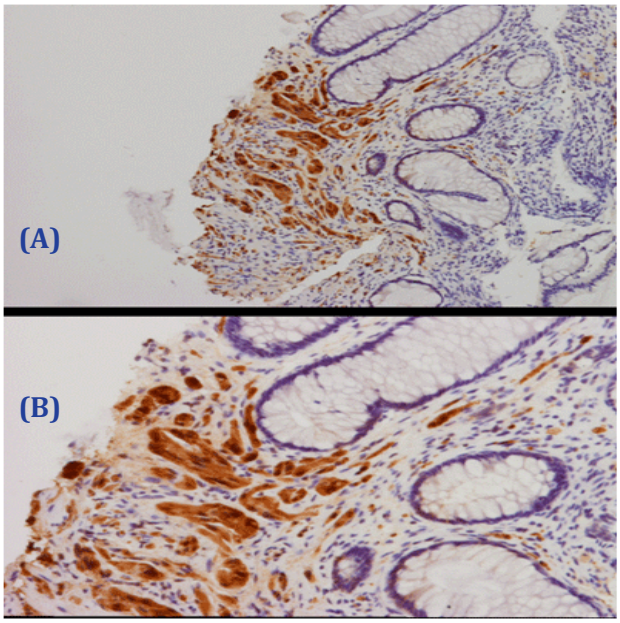

Figure 2: Immunochemistry Findings **(A)** Diffuse positivity for the S-100 protein (Schwann-cell immunophenotype) **(B)** A 20X view of the same findings of immunoreactivity for S-100 protein

Discussion

Colorectal polyps have long been a topic of discussion. With early screening colonoscopies, polyps are currently being studied more on the basis of their histological origin, morphological appearance, and immunohistochemistry staining, as some polyps are associated with certain hereditary syndromes with poorer prognosis while some follow a benign course. Among these polyps, neurofibromas are the most studied, which in addition to having S100 positive neural proliferations also lack ganglion cells [1]. There are different growth patterns for neurofibromas. The

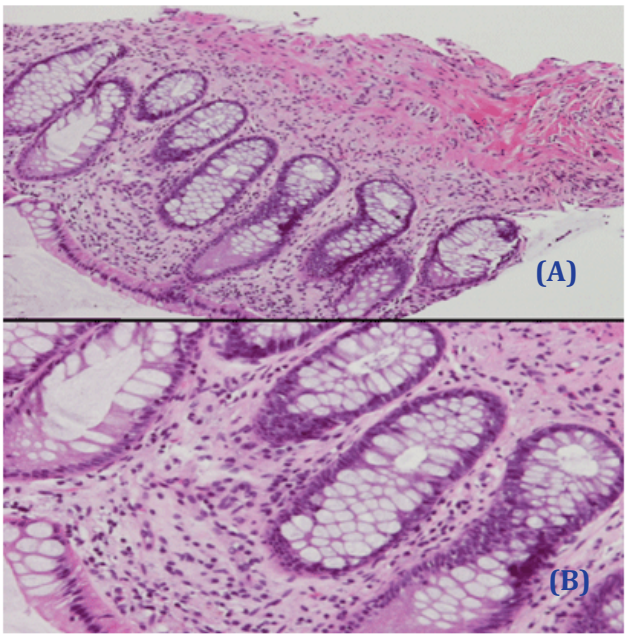

Figure 3: Histological Slide: **(A)** Diffuse proliferation of spindle cells with elongated, tapering nuclei in the lamina propria of the colonic mucosa **(B)** A 20X view of the same findings

one associated with neurofibromatosis type 1 is the plexiform growth pattern [6, 7]. Neurofibromas are mostly found in the stomach or small intestine, with colonic involvement very rare [1]. The isolated neurofibroma of the gastrointestinal tract described in 1937 is a rare entity [6, 8, 9] and a solitary neurfibromal colonic polyp without NF1 is also rare, with only 12 cases seen from 1937 to 2000 [10, 11] and less than 20 such cases reported in the literature until now [12]. The less studied Schwann cell hamartoma has been identified in the last few years. In 2009, Gibson and Hornick analyzed 26 cases of solitary colorectal polyps with Schwann cell proliferation, and the results were compared with five submucosal neurofibromas from neurofibromatosis type 1 patients [1]. This disease entity was proposed as mucosal Schwann cell hamartoma based on its unique histopathological appearance and that it is not associated with any hereditary disorders like neurofibromas seen in neurofibromatosis type 1.

The different types of colorectal polyps consist of neurofibromas, mucosal neuromas, ganglioneuromas, gastrointestinal stromal tumors (GIST's), mucosal schwannomas, and the newly proposed entity, Mucosal Schwann cell hamartoma [1, 6]. A brief literature review of few of these types is important for understanding the new disease entity. A neurofibroma, upon pathological examination, presents as a tumor consisting of a mixture of spindle cells with wavy nuclei and strands of collagen, as well as Schwann cells, perineural fibroblasts, endothelial cells, axons and mast cells [6, 13]. Mucosal neuromas are one of the clinical features of multiple endocrine neoplasia- type 2B. They are found on the lips and rarely seen in the gastrointestinal tract. They are composed of a hyperblastic bundle of nerve fibers and

axons, thus differentiating it from Schwann cell hamartoma [1]. Ganglioneuromas have gangliocytes and therefore can be distinguished from Schwann cell hamartomas, which lack ganglion cells [1].

Conclusion

Mucosal Schwann cell hamartoma is a very rare diagnosis. Though it is not associated with any of the hereditary syndromes, its malignant potential and local recurrence is yet to be studied. Further follow up data is needed before making final recommendations. Our case report adds further insight into this rare disease entity.

Consent

Informed patient consent was obtained for case publication.

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