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Case Report
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A Rare Case of Mucosal Schwann Cell Hamartoma of Colonic Mucosa

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Abstract

Mucosal Schwann Cell Hamartoma (MSCH) is one of the spindle cell neoplasms, which is relatively uncommon in Lower Gastrointestinal Tract (GIT). It is a newly recognized disease entity with no hereditary association, usually composed of bland spindle cells in lamina propria, showing S-100 positivity on immunohistochemical analysis. We present an unusual case of colonic mucosal Schwann cell hamartoma, in an asymptomatic elderly gentleman, detected incidentally at screening colonoscopy, along with a brief literature review. He underwent a thorough evaluation with contrast enhanced CT scan and Multidisciplinary Discussion (MDT) at our institution, following endoscopic polypectomy. Histopathology with immunohistochemistry confirmed the diagnosis of mucosal Schwann cell hamartoma. As MSCH is uncommon in colorectum & non-invasive in behavior, management and long-term follow-up is still under study. We aim to emphasize the significance of increased awareness required among the colorectal surgeons and pathologists, in order to differentiate it from other benign neural lesions of mesenchymal origin.

Keywords: Mucosal Schwann Cell Hamartoma (MSCH); Spindle cell neoplasm; Neurofibromatosis; Polyp

Introduction

The term Mucosal Schwann Cell Hamartomas (MSCH's) was first proposed by Gibson and Hornick in 2009 [1]. To describe a group of neuronal polyps purely composed of S-100 - positive Schwann cells, in an attempt to distinguish them from true "neuromas" and "neurofibromas."

Benign neural tumors are relatively rare in GIT. In lower GIT, they present as polyps and are mostly detected incidentally during routine/screening colonoscopy. Significant association is found between certain types of neural lesions (neurofibromas/mucosal neuromas) and specific inherited syndromes such as Neurofibromatosis type 1 (NF-1), multiple endocrine neoplasia type 2B (MEN-2B) etc.

Incidence of mucosal Schwann cell hamartomas is increasing now-a-days due to detailed evaluation of colorectal polyps, when patients undergo examination as a part of screening and for symptoms such as occult rectal bleeding, abdominal pain or altered bowel habits. Clinically, these lesions occur as polypoid lesions. Histologically, they are characterized by presence of uniform, bland spindle cell proliferation, confined to lamina propria with entrapment of colonic crypts. Spindle cells are distinct as they have elongated, tapering nuclei and abundant dense eosinophilic cytoplasm with indistinct cell borders. Cells lack atypia, pleomorphism, mitoses, axons & gangliocytes. They show strong reactivity to S-100 and negative for CD117, CD34, EMA, SMA and other markers on immunohistochemistry, which is a marking feature that characterizes MSCH's [1] and helps to differentiate from schwannomas and neurofibromas.

Case Presentation

A 31-year-old gentleman, with a background of neurofibromatosis, was admitted under urology for evaluation of recurrent Urinary Tract Infection (UTI) and underwent Computed Tomography of Kidneys & Urinary Bladder (CT-KUB), to rule out hydronephrosis. He had presented with a 1-day history of lower abdominal pain with no dysuria, altered bowel habits, bleeding per rectum or any weight loss. He underwent left ureteric pyeloplasty at 14 years of age and suffered recurrent episodes of UTI's following this. He had no significant family history. On examination, his abdomen was soft with mild tenderness in hypogastric region. Per rectal examination was unremarkable. Laboratory investigations revealed slightly raised inflammatory markers (WBC-17.3, CRP-161).

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Table 1: Literature review of cases.

Review	Country	Age (years)	Gender Male (M) Female (F)	Presentation	Size of lesion	Site of lesion
1. Gibson & Hornick [1]	Boston, USA -2009	62	M:F=10:16	Asymptomatic followed by diarrhoea, lower GI bleeding	Sessile, 1-6 mm (Mean 2.5)	Distal colon (rectosigmoid predominantly)
	No. of cases= 26	(Mean)				
2. Pasquini et al. [2]	Rome, Italy -2009	60	F	Occult blood in stool	Sessile, 5 mm	Rectosigmoid
3. Rocco et al. [3]	Milan, Italy -2011	67	F	Screening	Sessile, 3 mm	Sigmoid colon
4. Sagami et al. [4]	Japan -2012	40	M	Occult blood in stool	Many small whitish nodules	Sigmoid colon
5. Bae et al. [5]	Seoul, Korea -2013	41	F	Screening	8 mm	Descending colon
6. Neis et al. [6]	Rochester, Minnesota -2013	59	M	Underlying Ulcerative colitis	3 mm	Sigmoid colon
7. Ferro de Beca et al. [7]	Porto, Portugal -2014	72	M	Screening	5 mm	Sigmoid colon
8. Klair et al. [8]	Arkansas, USA -2014	78	F	Abdominal pain, intermittent tenesmus	7 mm with rectal erythema & inflammation	Rectum
9. Kanar et al. [9]	Florida, USA -2015	67	M	Screening	6 mm	Sigmoid colon
10. Bae et al. [10]	Seoul, Korea -2015	20	M	Abdominal discomfort, loose stools	4 mm with scattered tiny polyp-like mucosal elevation	Rectum
11. Han et al. [11]	Seoul, Korea -2015	49	M	Screening	2 mm, tiny polyp-like mucosal elevation	Rectum
12. Syed et al. [12]	Oklahoma, USA -2017	60	M	Weight loss, intermittent hematochezia	5 mm	Rectum
13. Chintanaboina et al. [13]	Pennsylvania, USA -2018	55	F	Screening	5 mm	Ascending colon
14. Chen et al. [14]	Chiayi, Taiwan -2019	31	M	Intermittent abdominal discomfort	Polypoid lesion, 4 mm	Sigmoid colon
15. Feng et al. [15]	LA, USA -2019	60	F	Screening	Sessile, 4 mm	Sigmoid colon
16 Present case	Luton, UK -2020	31	M	Screening	Sub-pedunculated, 6 mm	Splenic flexure

CT KUB showed fat stranding in suprapubic region, around small bowel loop and diverticular sigmoid colon, suggestive of appearances secondary to acute diverticulitis/small bowel inflammation. It also confirmed normal right kidney and atrophic left kidney with scarring. Subsequently, in view of CT findings, he was referred to the colorectal team, who advised conservative management with IV antibiotics and analgesics. Following uneventful recovery with conservative treatment, he underwent colonoscopy in 6 weeks' time on an outpatient basis, which showed 2 sub-pedunculated polyps-measuring 5 mm and 6 mm, in ascending colon and splenic flexure (Figure 1) respectively, both excised by hot snare polypectomy.

Histology confirmed sessile serrated polyp in ascending colon. However, microscopy of polyp from splenic flexure showed adipose tissue in submucosa with fibrous septae, raising the possibility of a submucosal lipoma. In addition, mucosa also showed an irregular area with fibrosis and bland spindle cells extending into underlying adipose tissue with no evidence of dysplasia or malignancy, as seen in (Figure 2a and 2b) and the excision was complete. Immunohistochemistry revealed bland spindle cells expressing S-100 as seen in (Figure 2c and 2d), surrounded by CD-34 positive spindle cells, with low proliferative index (Ki-67). All other markers including SMA, desmin and CD117 were negative. These features favored a diagnosis of benign neural lesion like a mucosal Schwann cell hamartoma in colonic mucosa, along with a possible submucosal lipoma. Hence, a second pathological opinion was sought from expert team that confirmed above findings. Differential diagnosis of perineurioma and benign fibroblastic polyp were also considered, based on the histological appearance, however; phenotypic appearance resembled and confirmed mucosal Schwann cell hamartoma.

He was clinically well and was seen in clinic, 4 weeks after the procedure. Following MDT discussion, since expert histology confirmed mucosal Schwann cell hamartoma with no features of malignancy, he was offered surveillance in the form of flexible sigmoidoscopy in 3 months' time for review.

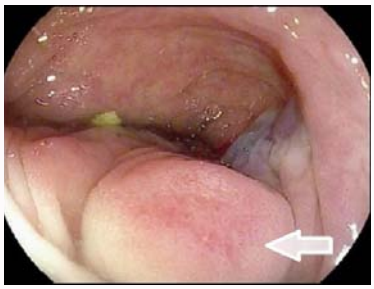

Figure 1: Colonoscopy showing sub-pedunculated polyp in splenic flexure, measuring 6 mm in size (white arrow).

Discussion

Mucosal Schwann cell hamartoma is a newly recognized, unique clinical entity comprising of benign mucosal proliferation with Schwann cells. This was initially proposed by Gibson and Hornick, who studied clinicopathological and immunohistochemical features in 26 colorectal polyps of neural origin, in an attempt to discriminate them from neuromas/neurofibromas. Table 1 illustrates only case reports of MSCH's existing in literature.

Colonoscopy is the gold standard technique for screening of colorectal polyps [12]. Differential diagnosis of spindle cell tumors is broad and includes neuromas, neurofibromas, benign fibroblastic polyps, schwannomas, ganglioneuromas, Gastrointestinal Stromal Tumors (GIST's), intestinal perineuriomas and mucosal benign epithelioid nerve sheath tumor. Histopathology combined with

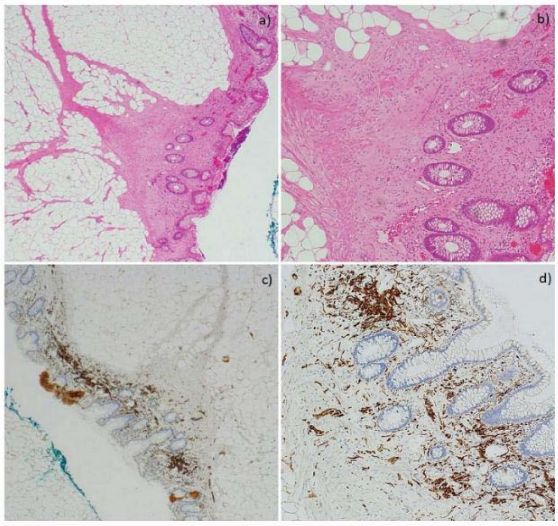

Figure 2: Microscopic view showing bland spindle cells & fibrosis, with no evidence of dysplasia or malignancy: a) low power magnification (x4), b) high power magnification (x10); Cells staining strongly positive for S-100: c) low power magnification (x4), d) high power magnification (x10).

immunohistochemistry helps us to distinguish MSCH's from above mentioned pathologies and clinch the definitive diagnosis [16,17].

The distinctive feature of MSCH's is diffuse proliferation of lamina propria with benign, bland spindle cells along with separation of adjacent colonic crypts, with no whorling/palisading/fasciculation. These spindle cells stain strongly positive for S-100 and have no association with any inherited syndromes. Although, neurofibromas have similar histologic features, however they are strongly associated with NF-1 and are less uniformly cellular with some intralesional heterogeneity, containing Schwann cells, fibroblasts, perineural-like cells, scattered Neurofilament Protein (NFP) positive axons and show less staining for S-100 protein, compared to MSCH's. Benign fibroblastic polyps (Vanek's tumor) are rare benign tumors, arising from submucosa, consisting of spindle cells expressing vimentin but negative for S-100. Mucosal schwannomas are often detected in stomach, uncommon in colon, typically show verocay bodies, Antoni A and Antoni B regions, peripheral lymphoid cuffs and lymphoid infiltrations. Ganglioneuromas exhibit fibroblasts with ganglion cells, which are absent in MSCH's. GIST's are another distinct set of mesenchymal tumors, submucosal in origin, rarely found in colon and are CD117/CD34 positive. Mucosal neuromas, composed of hyperplastic bundles of nerve fibres and axons, occur commonly on lips and tongue, very infrequently in GIT and are highly associated with MEN-2B. Though, mucosal perineuriomas display similar morphological features, they stain positive for EMA and negative for S-100, which aids in diagnosis. Careful morphological and immunohistochemical evaluation is important in differentiating MSCH's from other neural/stromal lesions of GIT [16,5].

Local excision of MSCH's is usually curative. However, currently there is no specific recommendation regarding management and plan on follow-up of individuals, diagnosed with these lesions in colorectum.

Conclusion

Mucosal Schwann cell hamartomas of colorectum is a group of rare, benign mesenchymal lesions, having no link with inherited syndromes. All lesions published in literature underwent complete excision. Currently, we do not know about its growth potential, when left untreated. There isn't enough information or evidence available regarding its invasive behavior/recurrence. Also, there is no absolute benchmark to plan the surveillance and follow-up of individuals diagnosed with these lesions. Additional research is required to elucidate its etiopathogenesis and formulate surveillance guidelines. Hence, we hereby suggest further data collection and maintaining a record in dedicated registry, in order to understand its clinical significance, avoid diagnostic dilemma and increase our knowledge.

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