A Rare Case of Spindle Cell Neoplasm of Rectum

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Abstract
Spindle cell neoplasms usually occur in head, neck, orbit, soft tissues of scalp and along the upper aerodigestive tract. They are relatively uncommon in lower gastrointestinal tract and represent a distinct clinical entity. Increased awareness is required among colorectal surgeons and pathologists due to their benign nature & uncertain etiology, to avoid misdiagnosis of rectal cancer. Definitive diagnosis necessitates immunohistochemical analysis. We present an unusual case of spindle cell neoplasm of rectum in an asymptomatic elderly gentleman, detected on screening colonoscopy. Following thorough evaluation with MRI pelvis, CT scan thorax, abdomen, pelvis with contrast and multidisciplinary meeting discussion (MDT) at our institution, he was successfully treated with a specialized minimally invasive approach (TAMIS). Histopathology with immunohistochemistry confirmed the diagnosis of spindle cell neoplasm. As they are uncommon in colorectum & non-invasive, management and long-term follow-up is still under study. These lesions should be differentiated from other stromal tumours in GIT.

Keywords: Spindle cell neoplasms; Trans-anal minimally invasive surgery (TAMIS); EMA (epithelial membrane antigen); Intestinal perineuriomas.

1. Introduction
Spindle cell neoplasms are rare benign mesenchymal lesions that affect the gastrointestinal tract. They are rarely found in colon and rectum (estimated incidence of 0.1% to 1.46% of all colonic polyps), while stomach and small intestine being the most common site of occurrence. Recent molecular studies have identified activating mutations in PDGFRα (Platelet derived growth factor alpha) in the pathogenesis of IFP’s. Huss, et al. [1]. Schildhaus, et al. [2].

Colonic spindle tumours with fibroblastic proliferations, also called as benign fibroblastic polyps (BFP’s), were first described by Eslami-Varzaneh, et al. [3]. They are usually detected incidentally on routine screening colonoscopy in asymptomatic individuals. Predominantly, they are found in women, in middle age group (60 years). They appear as single, sessile, well-circumscribed submucoosal lesions on scopy but are most often pedunculated, intraluminal polypoid lesions and may occur as large mural masses. The epicentre of tumor is usually in the submucosa but it can infiltrate into the overlying mucosa, muscularis propria or serosa [3].

Histologically, they are characterized by bland spindle cell proliferation within lamina propria that separates and distorts colonic crypt architecture. The epithelial surface is typically intact but may show superficial erosion. Cytologically, the spindle cells are bland with pale, eosinophilic cytoplasm, having indistinct cell borders, oval to fusiform nuclei, with inconspicuous nucleoli. Cells lack pleomorphism, hyperchromasia and significant mitotic activity. On immunohistochemistry, spindle cells stain variably positive for vimentin, with rare focal positivity for CD34 and smooth muscle actin but negative for c-kit, CD117, S-100 and other muscle markers. Ultrastructurally, they have features of fibroblastic differentiation. These observations are consistent with a histopathological diagnosis of spindle cell neoplasm and aids in differentiating them from gastrointestinal stromal tumours (GIST’s) [3, 4].

2. Case Report
A 68-year old gentleman with a background of diet-controlled diabetes, hypertensive on medications, underwent colonoscopy as a part of Bowel Cancer Screening Programme (BCSP) and was detected to have polyps in right colon and rectum. He had no history of altered bowel habits, abdominal pain, bleeding per rectum or any weight loss. He had past history of prostate cancer, treated with surgery (Robotic assisted laparoscopic prostatectomy) 2 years ago. He had no significant family history. His abdominal examination was unremarkable. Rectal examination revealed small polypoid lesion in lower half. Laboratory investigations were unremarkable. Colonoscopy showed a 2 cm polypoid lesion in lower rectum on a wide stalk, 1-2 cms above anorectal junction with suspicious pit pattern which was biopsied (Fig. 1).

Subsequently, he underwent MRI scan of the rectum to evaluate further characteristics of the lesion and CT scan of thorax, abdomen and pelvis with contrast, in view of suspicious findings on colonoscopy. MRI rectum (Fig. 2) showed a 2.4 x 1.9 cm polypoid lesion, arising from the posterior wall of lower rectum, 6 cm from anal verge, well confined to rectal wall, without any extension into mesorectal fat. However, CT scan of thorax, abdomen, pelvis was normal.

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Following discussion in Multidisciplinary meeting (MDT) at our centre, he underwent complete excision of rectal polyp by Trans-anal Minimally Invasive Surgery (TAMIS) under general anaesthesia. Operative findings confirmed a single, pedunculated rectal polyp measuring 2.5 cms in size, located posterolateral- at 5 o’clock position, 5 cms from anal verge and hard in consistency. He was discharged the next day, following surgical procedure.

Histology revealed a 23 mm partially ulcerated polyp, covered mostly by squamous epithelium and focally by colonic mucosa, with clear margins of excision. Polyp composed of haphazard intersecting fascicles of bland spindle cells with pale eosinophilic cytoplasm as seen in Fig. 3a & b). Stroma was variably edematous with prominent capillaries, fibrous and focally myxoid. Ulcerated surface contained granulation tissue while core of the polyp consisted of mixed inflammatory infiltrate including plasma cells, lymphocytes and scattered eosinophils. Mitotic count: 1/5 mm square (20 HPFs) and Ki-67 proliferative index: <10%. Necrosis or atypia was absent. Overlying squamous and colorectal mucosa was normal.

Immunohistochemistry revealed CD34 diffusely highlighting the spindle cells while SMA was very focal. All other markers including CD117, DOG 1, desmin, S100 and EMA were negative as seen in Fig. 3c) & d). Based on above findings, our histologist reported it as spindle cell neoplasm of rectal polyp. In view of negative CD117 & DOG1, there was a suspicion about gastrointestinal stromal tumour (GIST), hence second pathological opinion was sought from expert team, that confirmed above findings.

He was seen in clinic at 4 weeks post-surgery. Following MDT discussion, since expert histology confirmed spindle cell neoplasm with no features of malignancy and patient was clinically well, he was booked for flexible sigmoidoscopy at 3 months’ time for review.

3. Discussion

Spindle cell neoplasms are defined as neoplasms comprising of spindle-shaped cells. It is very difficult to diagnose these tumours with only simple routine haematoxylin and eosin staining on histopathology. Hence, immunohistochemical staining is imperative to reach to correct diagnosis.

Inflammatory fibroid polyps (IFP) was first described as “polypoid fibroma” by Konjetzny [5]. Vanek described it as eosinophilic submucosal granuloma in 1949 [6]. Helwing and Rainer coined the term “inflammatory fibroid polyp” in 1953 [7].

Spindle cell tumours with fibroblastic proliferation, also called as benign fibroblastic polyps, are rare benign tumours originating from the submucosa of the gastrointestinal tract. They are a new described entity, initially thought to be the result of an inflammatory response to underlying submucosal granuloma secondary to an irritating stimulus (e.g. trauma, tuberculosis, helicobacter pylori, Crohn disease, sarcoidosis). However, its true etiology and pathogenesis is still poorly understood [8]. In gastrointestinal tract, stomach (70%) is the most common site of occurrence, followed by small intestine (20%). It is rarely found in esophagus and colorectum [9].

Few case reports of benign fibroblastic polyps exist in literature (Table 1).

| Table-1. Literature review of cases |
|-----------------------------------|
| **Country** | **Age (years)** | **Gender** | **Presentation** | **Size of lesion** | **Site of lesion** | **Surgical procedure** |
|----------------|----------------|------------|-----------------|-------------------|-------------------|-----------------------|
| 1. [10] Bari, Italy (1984) | 8 | M | Rectal bleeding | 3 cms | Rectum | Surgical resection |
| 2. [11] Nagoya, Japan (2008) | 66 | M | Asymptomatic (routine screening) | 0.3 cm | Rectum | Endoscopic submucosal dissection (ESD) |
| 3. [12] Taichung, Taiwan (2012) | 65 | F | Haematochezia & weight loss (Mimicking rectal cancer with LN enlargement & sacral attachment) | 3 x 2.7 cms | Rectum | Total mesorectal excision of rectal and sacral tumour (TME) |
| 4. [13] New York, USA (2012) | 74 | F | Asymptomatic (routine screening) | 1.2 cms | Hepatic flexure | Endoclipping + Argon plasma coagulation + Hot snare polypectomy |
| 5. [14] Wakayama, Japan (2013) | 65 | M | Asymptomatic (routine screening) | 0.5 cm | Sigmoid colon | Endoscopic mucosal resection (EMR) |
6. [15] Toledo, Ohio (2013) 83 M Haematochezia & abdominal pain 7 x 4 x 3 cms Descending colon Endoscopic mucosal resection (EMR-piecemeal excision)

7. [16] Telangana, India (2013) 26 F Intestinal obstruction 5 x 5 cms Caecum Surgical resection

8. [17] Lagos, Nigeria (2016) 59 M Asymptomatic (routine screening) 0.3 cm Rectum Endoscopic cold snare biopsy

9. [18] Hartford, USA (2019) 53 M Asymptomatic (routine screening) 4.2 cms Proximal rectum Trans-anal excision

10. Our case Luton, UK (2019) 68 M Asymptomatic (routine screening) 2.4 x 1.9 cms Distal rectum Trans-anal excision

One series in Baltimore USA, analysed 83 cases of IFP’s in gastrointestinal tract, between 1999-2012 and found large intestine (37%) to be the most commonly involved site, followed by the gastric antrum (23%) and small intestine (20%). This study also concluded that IFPs represent a heterogenous set of submucosal lesions, with varied morphologic and histologic features, amenable to endoscopic biopsy [19].

Spindle cell tumours of gastrointestinal tract, usually arise from submucosa with overlying mucosal ulceration. A characteristic cell type is stellate or spindle-shaped bland stromal cell, that tends to be arranged in an onion-skin pattern, in a vascularized and hyalinized stroma with abundant inflammatory cells, containing large number of eosinophils. These tumors are thought to be derived from fibroblasts or dendritic cells. Stromal cells are reactive for vimentin, CD34 (82%-100%), fascin, calponin and CD35 but, in contrast to GISTs, they are negative for CD117. One fourth of cases show positive staining for SMA [20, 21].

IFP’s of colon and rectum are mostly discovered incidentally on routine screening colonoscopy. However, symptoms depend on location and size of lesions. Colonic IFP’s can present with abdominal pain (54%), bloody stools (33%), weight loss (21%), diarrhoea, and anaemia (17%) [22]. A familial occurrence of IFP’s has been reported (Devon polyposis syndrome) [23].

Colonoscopy is the gold standard procedure for colonic screening of colorectal polyps [24]. Histopathology and immunohistochemical analysis points to definitive diagnosis of spindle cell tumours. Differential diagnosis includes smooth muscle tumors, leiomyoma, neurofibroma, schwannoma, ganglioneuroma, prolapsing mucosal polyp, gastrointestinal stromal tumors (GIST’s), and intestinal perineuriomas. It is quite difficult to differentiate spindle cell neoplasms from intestinal perineuriomas as both have identical clinical and histological features. Spindle cell tumours stain uniformly negative for CD-117 (positive in GIST’s) and EMA (positive in intestinal perineuriomas), which aid in diagnosis [25, 26].

Local excision of spindle cell neoplasms is usually curative. Presently, there is no standard criteria available regarding management and follow-up of individuals diagnosed with these lesions in colorectum.

4. Conclusion

Spindle cell neoplasm is a group of rare benign mesenchymal lesions of gastrointestinal tract, having indolent clinical course. Clinically there is no evidence of its invasive behaviour, metastasis and recurrence. Complete resection is usually curative. However, there is no absolute norm or recommendations for management and surveillance. Whether routine endoscopic evaluation following complete resection is indicated or not, is a matter of debate. Further detailed research is required to explicate etiopathogenesis, treatment guidelines, recurrence rate and follow-up in patients with colonic and rectal spindle cell tumours. We hereby, suggest introduction of registry to track these lesions, to understand pathology and potential complications.

Compliance with ethical standards:
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Fig 1. Colonoscopy revealed a low rectal polyp with wide stalk, measuring 2 cms in size.

Fig 2. MRI images (sagittal & coronal) showed a 2.4 x 1.9 cm polypoid lesion, arising from the posterior wall of lower rectum, 6 cm from anal verge, well confined to rectal wall.

Fig 3. Microscopic view showing bland spindle cells with pale eosinophilic cytoplasm and fibrous & myxoid stroma with mixed inflammatory cells a) low power magnification (x 4), b) high power magnification (x 200), c) CD-117 staining- Negative (x 200), d) DOG-1 staining- Negative (x 400)
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